CENTOGENE
THE RARE DISEASE COMPANY

Test Catalogue

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CENTOGENE CLINICAL DIAGNOSTIC PRODUCTS AND SERVICES

› Metabolic Disorders - Panels

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoDx®	Targets exonic regions of >6500 genes associated with known clinical phenotypes		50158	5
CentoLCV	Genome wide analysis of structural variants and large copy number changes through sWGS		50118	5
CentoLSD Enzyme Panel	Acid lipase (Wolman), Acidic alpha-glucosidase (Pompe), Acidic sphingomyelinase (Niemann-Pick Type A and Type B), Alpha-fucosidase (Alpha-fucosidase deficiency), Alpha-galactosidase (Fabry), Alpha-L-iduronidase (MPS I), Alpha-mannosidase (Alpha-mannosidase deficiency,), Alpha-N-acetylgalactosaminidase (Schindler/Kanzaki), Arylsulfatase B (MPS VI), Beta-galactosidase (MPS IVB), Beta-glucocerebrosidase and Chitotriosidase (Gaucher), Beta-glucuronidase (MPS VII), Beta-hexosaminidase (Tay-Sachs), Beta-mannosidase (Beta-mannosidase deficiency), Hexosaminidase AB (Sandhoff), Iduronate-2-sulfatase (MPS II), N-acetyl-alpha-glucosaminidase (MPS IIIB), N-acetylgalatosamine-6-sulfate-sulfatase (MPS IVA), Palmitoyl-protein thioesterase (Neuronal ceroid lipofuscinosis type 1, NCL1, Infantile NCL, Santavuori-Haltia disease), Tripeptidyl peptidase (Neuronal ceroid lipofuscinosis type2, NCL2, Late infantile NCL, Jansky-Bielschowsky disease)		25000	14
CentoLSD Enzyme Panel X-TRA	Acid lipase (Wolman, LIPA), Acidic alpha-glucosidase (Pompe, GAA), Acidic sphingomyelinase (Niemann-Pick Type A and Type B, SMPD1), Alpha-fucosidase (Alpha-fucosidase deficiency, FUCA1), Alpha-galactosidase (Fabry, GLA), Alpha-L-iduronidase (MPS I, IDUA), Alpha-mannosidase (Alpha-mannosidase deficiency, MAN2B1), Alpha-N-acetylgalactosaminidase (Schindler/Kanzaki, NAGA), Arylsulfatase B (MPS VI, ARSB), Beta-galactosidase (MPS IVB, GLB1), Beta-glucocerebrosidase and Chitotriosidase (Gaucher, GBA), Beta-glucuronidase (MPS VII, GUSB), Beta-hexosaminidase (Tay-Sachs, HEXA), Beta-mannosidase (Beta-mannosidase deficiency, MANBA), Hexosaminidase AB (Sandhoff, HEXA/HEXB), Iduronate-2-sulfatase (MPS II, IDS), N-acetyl-alpha-glucosaminidase (MPS IIIB, NAGLU), N-acetylgalatosamine-6-sulfate-sulfatase (MPS IVA, GALNS), Palmitoyl-protein thioesterase (Neuronal ceroid lipofuscinosis type 1, NCL1, Infantile NCL, Santavuori-Haltia disease, PPT1), Tripeptidyl peptidase (Neuronal ceroid lipofuscinosis type2, NCL2, Late infantile NCL, Jansky-Bielschowsky disease, TPP1)		25001	15

1: Hotspot Testing
2: Carrier Testing (point mutation)
3: Carrier Testing (del/dup)
4: Single Gene Sequencing (Sanger)
5: NGS Panel
6: NGS Panel + CNV
7: NGS Panel Genomic
8: Deletion/Duplication with qPCR/MLPA
9: Sequencing + Deletion/Duplication
10: Repeat Expansions
11: Somatic Mutation Analysis
12: Biochemical Enzyme Analysis
13: Biomarker Analysis
14: Biochemical Enzyme Panel
15: Biochemical Genetics Panel Plus
16: Single Gene Sequencing (NGS)
17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoMetabolic®	ABCA1, ABCB4, ABCC2, ABCD1, ABCD4, ABCG5, ABCG8, ACAT1, ADA, AGA, AGL, AGPS, AGXT, ALAD, ALAS2, ALDH4A1, ALDOA, ALDOB, ALG3, ALPL, ANTXR2, APOA2, APOA5, APOB, APOC2, APOE, ARG1, ARSA, ARSB, ASAH1, ASL, ASS1, ATP7A, ATP7B, BCKDHA, BCKDHB, BTD, CBS, CD320, CETP, CLN3, CLN5, CLN6, CLN8, CPOX, CPS1, CPT1A, CTNS, CTSA, CTSD, CTSK, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DBT, DDC, DHCR7, DIABLO, DLX4, DNAJC5, DPYD, ENO3, ENPP1, EPHX2, ETHE1, FAH, FBP1, FECH, FGF23, FUCA1, G6PC, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GATM, GBA, GBE1, GHR, GK, GLA, GLB1, GM2A, GNPAT, GNPTAB, GNPTG, GNS, GUSB, GYG1, GYS1, GYS2, HCFC1, HEXA, HEXB, HFE, HJV, HGD, HGSNAT, HLCS, HMBS, HPD, HPRT1, HSD3B2, HYAL1, IDS, IDUA, ITIH4, IVD, KHK, LAMP2, LCAT, LDHA, LDLR, LDLRAP1, LIPA, LIPC, LIPI, LMBRD1, LPA, LPL, MAN2B1, MANBA, MCOLN1, MFSD8, MMAA, MMAB, MMACHC, MMADHC, MMUT, NAGA, NAGLU, NAGS, NEU1, NPC1, NPC2, OTC, PAH, PCSK9, PDHB, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PKLR, PNPO, POR, PPOX, PPP1R17, PPT1, PRKAG2, PSAP, PYGL, PYGM, RBCK1, SGSH, SI, SLC17A5, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A36, SLC2A1, SLC2A2, SLC2A3, SLC37A4, SLC3A1, SLC3A2, SLC40A1, SLC6A19, SLC6A8, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SUMF1, TAT, TFR2, TPP1, UGT1A1, UMPS, UROD, UROS		5446	6
CentoMPS Enzyme Panel	Alpha-L-iduronidase (MPS I), Iduronate-2-sulfatase (MPS II), N-acetyl-alpha-glucosaminidase (MPS IIIB), N-acetylgalatosamine-6-sulfate-sulfatase (MPS IVA), Beta-galactosidase (MPS IVB), Arylsulfatase B (MPS VI), Beta-glucuronidase (MPS VII), Alpha-mannosidase (Alpha-mannosidase deficiency)		25004	14
CentoMPS Enzyme Panel X-TRA	Alpha-L-iduronidase (Hurler disease, MPS I, IDUA), Iduronate-2-sulfatase (Hunter disease, MPS II, IDS), N-acetyl-alpha-glucosaminidase (Sanfilippo syndrome, MPS IIIb, NAGLU), N-acetylgalatosamine-6-sulfate-sulfatase (Morquio disease, MPS IVA, GALNS), Beta-galactosidase (Morquio disease, MPS IVB, GLB1), Arylsulfatase B (Maroteaux-Lamy syndrome, MPS VI, ARSB), Beta-glucuronidase (Sly syndrome, MPS VII, GUSB), Alpha-mannosidase (Alpha-mannosidase deficiency, MAN2B1)		25005	15
CentoNCL Enzyme Panel	Palmitoyl-protein thioesterase (Neuronal ceroid lipofuscinosis type 1, NCL1, Infantile NCL, Santavuori-Haltia disease) Tripeptidyl peptidase (Neuronal ceroid lipofuscinosis type2, NCL2, Late infantile NCL, Jansky-Bielschowsky disease)		25006	14
CentoNCL Enzyme Panel X-TRA	Palmitoyl-protein thioesterase (Neuronal ceroid lipofuscinosis type 1, NCL1, Infantile NCL, Santavuori-Haltia disease, PPT1) Tripeptidyl peptidase (Neuronal ceroid lipofuscinosis type2, NCL2, Late infantile NCL, Jansky-Bielschowsky disease, TPP1)		25007	15
CentoSphingo Enzyme Panel	Acidic sphingomyelinase (Niemann-Pick Type A and Type B), Beta-glucocerebrosidase and Chitotriosidase (Gaucher), Alpha-galactosidase (Fabry), Acidic alpha-glucosidase (Pompe), Beta-hexosaminidase (Tay-Sachs), Hexosaminidase AB (Sandhoff), Alpha-N-acetylgalactosaminidase (Schindler/Kanzaki), Acid lipase (Wolman) , Alpha-mannosidase (Alpha-mannosidase deficiency), Beta-mannosidase (Beta-mannosidase deficiency), Alpha-fucosidase (Alpha-fucosidase deficiency)		25002	14

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Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoSphingo Enzyme Panel X-TRA	Acidic sphingomyelinase (Niemann-Pick Type A and Type B, SMPD1), Beta-glucocerebrosidase and Chitotriosidase (Gaucher, GBA), Alpha-galactosidase (Fabry, GLA), Acidic alpha-glucosidase (Pompe, GAA), Beta-hexosaminidase (Tay-Sachs, HEXA), Hexosaminidase AB (Sandhoff, HEXA/HEXB), Alpha-N-acetylgalactosaminidase (Schindler/Kanzaki, NAGA), Acid lipase (Wolman, LIPA) , Alpha-mannosidase (Alpha-mannosidase deficiency, MAN2B1), Beta-mannosidase (Beta-mannosidase deficiency, MANBA), Alpha-fucosidase (Alpha-fucosidase deficiency, FUCA1)		25003	15
Diabetes and obesity panel	ABCA1, ABCC8, ACAT1, ACSF3, AFF4, AGL, AKT2, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, APOA1, APOA5, APOB, APOC2, APOE, ARL13B, ARL6, ATP6V0A2, B4GALT1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BDNF, BLK, C8ORF37, CC2D2A, CCDC28B, CEL, CEP164, CEP290, CHD2, CISD2, COG1, COG4, COG5, COG6, COG7, COG8, CP, CREBBP, CUL4B, CYP27A1, DDOST, DHDDS, DOLK, DPM1, DPM2, DPM3, DYRK1B, EIF2AK3, ENO3, ENPP1, EPM2A, FBP1, FOXP3, G6PC, GAA, GATA6, GBE1, GCK, GHR, GLI3, GLIS3, GLUD1, GMPPA, GNAS, GNE, GPIHBP1, GYG1, GYS1, GYS2, HADH, HEXA, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, IER3IP1, IFT172, IFT27, INPP5E, INS, INSR, KCNJ11, KIF7, KLF11, LAMP2, LARGE1, LDHA, LDLR, LDLRAP1, LEP, LEPR, LIPA, LIPE, LPL, LZTFL1, MAGEL2, MAN1B1, MC4R, MGAT2, MKKS, MKS1, MOGS, MPDU1, MPI, MPV17, MTTP, MYO5A, MYO7A, NDN, NEUROD1, NEUROG3, NGLY1, NHLRC1, NPHP1, NPHP3, NR0B2, NTRK2, OFD1, OXCT1, PAX4, PC, PCK1, PCNT, PCSK1, PCSK9, PDX1, PFKM, PGAM2, PGK1, PGM1, PHF6, PHKA1, PHKA2, PHKB, PHKG2, PMM2, PNPLA6, POMC, PPARG, PRKAG2, PROM1, PRPH2, PTF1A, PYGL, PYGM, RAB23, RAI1, RBCK1, RDH5, RFT1, RFX6, RHO, RLBP1, RPGRIP1L, RPS6KA3, SDCCAG8, SIM1, SLC16A1, SLC19A2, SLC2A2, SLC35A1, SLC35A2, SLC35C1, SPG11, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM67, TRIM32, TTC21B, TTC8, TUSC3, UCP2, UCP3, VPS13B, WDPCP, WFS1, ZFP57		5449	6

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Panel name	Genes	Test code	Available test methods
CentoICU®	AARS1, AARS2, AASS, ABAT, ABCA12, ABCA3, ABCB11, ABCC8, ABCD1, ABCD3, ABCD4, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACTA1, ACY1, ADA, ADAMTS13, ADAMTSL2, ADAR, ADK, ADNP, ADSL, AGA, AGK, AGL, AGPAT2, AGPS, AGRN, AGXT, AHCY, AICDA, AIFM1, AIMP1, AKAP9, AKR1D1, AKT2, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX12B, ALOXE3, ALPL, ALS2, AMACR, AMN, AMPD1, AMT, ANK1, ANKRD26, ANKS6, ANTXR1, ANTXR2, AP2S1, AP4B1, AP4E1, AP4M1, AP4S1, APOB, APTX, ARG1, ARL6, ARSA, ARSB, ARX, ASAH1, ASL, ASNS, ASPA, ASPM, ASS1, ATIC, ATP1A3, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATPAF2, ATR, ATRX, AUH, B3GLCT, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCKDK, BCS1L, BDNF, BICD2, BIN1, BLNK, BOLA3, BRAF, BRAT1, BRCA2, BSCL2, BSND, BTD, BTK, C12orf65, CA12, CACNA1C, CACNB2, CALM1, CAMTA1, CASK, CASR, CAST, CAV1, CAV3, CAVIN1, CBS, CCDC103, CCDC114, CCDC78, CD19, CD247, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD59, CD79A, CD79B, CD81, CD96, CDAN1, CDK5RAP2, CDKL5, CDKN1C, CENPJ, CEP152, CEP290, CERS3, CFAP298, CFH, CFHR3, CFL2, CFTR, CHAT, CHD7, CHKB, CHM, CHRNA1, CHRNB1, CHRND, CHRNE, CIDEC, CLCN1, CLCNKA, CLCNKB, CLDN16, CLN3, CLN5, CLN6, CLN8, CLPB, CNTN1, COA5, COG1, COG6, COG7, COL11A1, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A2, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COMP, COQ2, COQ8A, COQ9, CORO1A, COX10, COX15, COX20, COX6B1, CPS1, CPT1A, CPT2, CR2, CRPPA, CRTAP, CTNS, CTPS1, CTSA, CTSD, CUL4B, CXCR4, CYP11B1, CYP11B2, CYP17A1, CYP4F22, CYP7B1, D2HGDH, DBT, DCLRE1C, DDC, DDOST, DDR2, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DIAPH1, DLAT, DLD, DMD, DNA2, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC19, DNM2, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPM2, DPYD, DRC1, DSP, DST, DUOX2, DUOXA2, DYSF, EDN3, EEF1A2, EGR2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELANE, ENPP1, EPB42, EPCAM, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYA1, EYA4, F10, F11, F13A1, F2, F5, F7, F8, F9, FADD, FAH, FANCA, FANCB, FANCC, FANCD2, FANCL, FARS2, FASTKD2, FBN1, FBP1, FBXL4, FGA, FGB, FGFR2, FGFR3, FGG, FH, FIG4, FKBP14, FKRP, FKTN, FOXC1, FOXG1, FOXP3, FOXRED1, FRAS1, FUCA1, G6PC, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GARS1, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GFAP, GFM1, GFPT1, GJA1, GJB2, GJB4, GK, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLUD1, GLYCTK, GMPPB, GNAS, GNE, GNMT, GNPAT, GNPTAB, GP1BA, GP1BB, GP9, GPC3, GPHN, GPSM2, GSS, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBA1, HBA2, HBB, HCFC1, HESX1, HEXA, HEXB, HGD, HGF, HIBCH, HLCS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPGD, HRAS, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, ICOS, IDUA, IER3IP1, IFIH1, IFT172, IGF1, IGF1R, IGHMBP2, IGLL1, IKBKB, IL12RB1, IL2RA, IL2RG, IL7R, INS, INSR, INVS, IRF8, ITGA2B, ITGA6, ITGA7, ITGB3, ITGB4, IVD, JAG1, JAGN1, JAK3, JAM3, KAT6A, KAT6B, KBTBD13, KCNE1, KCNH1, KCNH2, KCNJ10, KCNJ11, KCNQ1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIF1B, KLF1, KLHL40, KLHL41, KLHL7, KRAS, KRT5, LAMA2, LAMA3, LAMB3, LAMC2, LAMP2, LAMTOR2, LARS2, LAS1L, LCT, LHX3, LHX4, LIAS, LIG4, LIPA, LIPN, LIPT1, LMBRD1, LMNA, LPIN1, LRBA, LRPPRC, LRRC8A, MAGEL2, MAGT1, MALT1, MAN2B1, MANBA, MAP2K1, MAP2K2, MAT1A, MCCC1, MCCC2, MCEE, MCM4, MCPH1, MECP2, MED12, MEF2C, MEGF10, MFN2, MFSD8, MITF, MKKS, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MPC1, MPI, MPL, MPV17, MPZ, MRPL3, MRPL44, MSMO1, MTHFR, MTM1, MTMR14, MTO1, MTR, MTRR, MUSK, MVK, MYCN, MYH9, NAA10, NAGA, NAGS, NALCN, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NDUFV2, NEB, NEU1, NEUROG3, NEXN, NFKB2, NFU1, NGF, NGLY1, NHEJ1, NIPAL4, NIPBL, NKX2-1, NKX2-5, NLRC4, NLRP3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NR0B1, NR3C2, NRAS, NSD1, NSDHL, NUBPL, OAT, OCLN, OCRL, OPA3, OPHN1, ORC1, ORC4, OTC, OXCT1, PAFAH1B1, PAH, PAX2, PAX3, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH19, PCNT, PDCD10, PDE10A, PDHA1, PDHB, PDHX, PDP1, PDSS2, PDX1, PEPD, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGM1, PHGDH, PHKG2, PHOX2B, PIGA, PIGN, PIGT, PIGV, PIK3CD, PKD2, PKHD1, PKLR, PLCB4, PLEC, PLOD1, PLP1, PMM2, PMP22, PNKP, PNP, PNPLA1, PNPO, PNPT1, POGZ, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU1F1, PPT1, PRDM16, PRKAG2, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PSAP, PSAT1, PSPH, PTPN11, PTPRC, PTRH2, PTS, PURA, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RAC2, RAF1, RAG1, RAG2, RANBP2, RAPSN, RARS2, RB1, RBBP8, RBM8A, RET, RFT1, RFX5, RFX6, RIT1, RMND1, RNASEH2C, RNASET2, RORC, RPS19, RRM2B, RXYLT1, RYR1, SALL1, SATB2, SBDS, SCN1A, SCN2A, SCN4A, SCN5A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SELENON, SERAC1, SERPINC1, SERPING1, SFTPB, SFTPC, SHOC2, SIL1, SIX3, SIX5, SKI, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC26A2, SLC26A3, SLC2A1, SLC30A2, SLC33A1, SLC3A1, SLC4A1, SLC52A1, SLC52A3, SLC5A1, SLC5A5, SLC6A1, SLC6A3, SLC6A5, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNAI2, SNX10, SOS1, SOX10, SOX2, SOX9, SPAST, SPEG, SPINK5, SPINT2, SPR, SPRED1, SPTA1, SPTAN1, SPTB, SRD5A3, ST3GAL3, ST3GAL5, STAR, STAT1, STAT3, STIL, STIM1, STS, STT3B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SYNE1, TACO1, TAT, TAZ, TBC1D24, TBCE, TBX19, TBX5, TCAP, TCN2, TFR2, TG, TGM1, TH, THRA, TJP2, TMCO1, TMEM165, STING1, TMEM70, TNFRSF13B, TNFRSF13C, TNFSF4, TNNT1, TP63, TPM2, TPM3, TPO, TPP1, TRIP11, TRMU, TRPV4, TSC1, TSC2, TSFM, TSHB, TSHR, TSPYL1, TTC7A, TTN, TUBA8, TUBB2A, TWNK, UBA1, UGT1A1, UMPS, UNG, UPB1, UQCRC2, UROD, UROS, WAS, WDPCP, WDR62, WDR73, WFS1, WNK1, WT1, ZAP70, ZEB2, ZFP57, ZNF423	5445	5

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13: Biomarker Analysis
14: Biochemical Enzyme Panel
15: Biochemical Genetics Panel Plus
16: Single Gene Sequencing (NGS)
17: Single Gene Sequencing + CNV (NGS)

Disease	Gene	Test code	OMIM Gene	Available test methods
2-aminoadipic 2-oxoadipic aciduria	DHTKD1	2317	614984	2, 4, 17
2-methylbutyrylglycinuria	ACADSB	2778	600301	2, 4, 17
2,4-dienoyl-CoA reductase 1	DECR1	365	222745	2, 4, 17
2,4-dienoyl-CoA reductase deficiency	NADK2	3791	615787	17
3-beta-hydroxysteroid dehydrogenase deficiency type 2	HSD3B2	74	613890	2, 3, 4, 8, 9
3-hydroxy-3-methylglutaryl-CoA lyase deficiency	HMGCL	366	613898	2, 3, 4, 8, 9, 17
3-hydroxy-3-methylglutaryl-CoA synthase 2 deficiency	HMGCS2	2840	600234	2, 4, 8, 9, 17
3-hydroxyacyl-CoA dehydrogenase deficiency	HADH	367	601609	2, 4, 17
3-hydroxyisobutryl-CoA hydrolase deficiency	HIBCH	2839	610690	2, 4, 17
3-methylcrontonyl-CoA carboxylase 1 deficiency	MCCC1	368	609010	2, 4, 17
3-methylcrontonyl-CoA carboxylase 2 deficiency	MCCC2	369	609014	2, 4, 17
3-methylglutaconic aciduria type 1	AUH	1350	600529	2, 3, 4, 8, 9, 17
3-methylglutaconic aciduria type 3	OPA3	1402	606580	2, 4, 8, 9
3-methylglutaconic aciduria type 5	DNAJC19	2298	608977	2, 4
3-methylglutaconic aciduria type 7, with cataracts, neurologic involvement and neutropenia	CLPB	3099	616254	3, 8, 17
3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	SERAC1	2299	614725	2, 3, 4, 8, 9, 17
6q24-related transient neonatal diabetes mellitus type 1	UPD chr. 6	3097		8
17-hydroxylation activity deficiency	CYP17A1	73	609300	2, 4, 8, 9, 17
Abetalipoproteinemia	MTTP	2763	157147	2, 4, 17
Acatalasemia	CAT	3770	115500	17
Acetycholinesterase deficiency	ACHE	2786	100740	2, 4
Acetyl-CoA carboxylase deficiency	ACACA	3046	200350	2, 17
Acute Alcohol sensitivity	ALDH2	3764	100650	17
Acute hepatic porphyria	ALAD	3126	125270	17
Acyl-CoA medium-chain dehydrogenase deficiency	ACADM	330	607008	2, 3, 4, 8, 9, 17
Acyl-CoA multiple dehydrogenase deficiency	ETFA	332	608053	2, 3, 4, 8, 9, 17
Acyl-CoA multiple dehydrogenase deficiency	ETFB	333	130410	2, 3, 4, 8, 9, 17
Acyl-CoA short-chain dehydrogenase deficiency	ACADS	334	606885	2, 3, 4, 8, 9, 17
Acyl-CoA very long-chain dehydrogenase deficiency	ACADVL	335	609575	2, 3, 4, 8, 9, 17
Adenine phosphoribosyltransferase deficiency	APRT	2761	102600	2, 4
Adenylosuccinase deficiency	ADSL	1571	608222	2, 4, 17
Adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2	336	613815	2, 3, 4, 8, 9
Adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	POR	75	124015	2, 4, 8, 9, 17
Adrenal hyperplasia due to steroid 11-beta-hydroxylase deficiency	CYP11B1	337	610613	2, 3, 4, 8, 9, 17
Adrenal hypoplasia	NR0B1	338	300473	2, 4, 8, 9
AICA-ribosiduria due to ATIC deficiency	ATIC	3766	601731	17
Alkaptonuria	HGD	16	607474	2, 3, 4, 8, 9, 17
Alpha-2-macroglobulin deficiency	A2M	1825	103950	2, 4, 17
Alpha-ketoglutarate dehydrogenase deficiency	OGDH	2854	613022	2, 4, 17
Alpha-methylacyl CoA racemase deficiency	AMACR	339	604489	2, 3, 4, 8, 9
Aminoacylase deficiency	ACY1	1572	104620	2, 4, 17
AMP deaminase deficiency, erythrocytic	AMPD3	2254	102772	2, 4, 17
Amyloidosis, familial visceral	APOA1	2106	107680	2, 3, 4, 8, 9
Andersen disease	GBE1	340	607839	2, 3, 4, 8, 9, 17
Anemia dyserythropoietic type 1A	CDAN1	341	607465	2, 3, 4, 8, 9, 17
Anemia dyserythropoietic type 2	SEC23B	342	610512	2, 3, 4, 8, 9, 17
Antitrypsin-alpha-1 deficiency	SERPINA1	345	107400	2, 3, 4, 8, 9
Aplastic anemia	PRF1	225	170280	2, 4, 8, 9
Aplastic anemia, SBDS related	SBDS	252	607444	2, 3, 4, 8, 9
Apolipoprotein A-II deficiency	APOA2	3820	107670	17
Apolipoprotein C-II deficiency	APOC2	346	608083	2, 3, 4, 8, 9
Apparent mineralocorticoid excess	HSD11B2	76	614232	2, 3, 4, 8, 9
Arginase deficiency	ARG1	347	608313	2, 4
Arginine-glycine amidinotransferase deficiency	GATM	348	602360	2, 3, 4, 8, 9, 17
Argininosuccinic aciduria	ASL	349	608310	2, 3, 4, 8, 9, 17
Aromatic L-amino acid decarboxylase deficiency (AADC)	DDC	2835	107930	2, 4, 13, 17
Asparaginesynthetase deficiency	ASNS	2013	108370	2, 4, 17
Aspartylglucosaminuria	AGA	2244	613228	2, 4, 17
Beta-Galactosamide alpha-2,6-Sialyltransferase 2 deficiency	ST6GAL2	2006	608472	2, 4
Beta-ureidopropionase deficiency	UPB1	2466	606673	2, 3, 4, 8, 9, 17
Bile acid malabsorption, primary	SLC10A2	3044	601295	2, 4
Bile acid synthesis defect type 1, congenital	HSD3B7	3244	607764	17

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Bile acid synthesis defect type 2, congenital	AKR1D1	2779	604741	2, 4, 17
Bile acid synthesis defect type 3, congenital	CYP7B1	350	603711	2, 3, 4, 8, 9
Bile acid synthesis defect type 4, congenital	AMACR	339	604489	2, 3, 4, 8, 9
Bile acid synthesis defect type 5, congenital	ABCD3	3761	170995	17
Biotinidase deficiency	BTD	351	609019	2, 3, 4, 8, 9, 17
Bloom syndrome	BLM	352	604610	2, 3, 4, 8, 9, 17
Branched-chain aminotransferase 1 deficiency	BCAT1	2656	113520	2, 4, 17
Branched-chain aminotransferase 2 deficiency	BCAT2	2657	113530	2, 4
Branched-chain ketoacid dehydrogenase kinase deficiency	BCKDK	2795	614901	2, 4, 17
Bronchiectasis with or without elevated sweat chloride type 2	SCNN1A	214	600228	2, 3, 4, 8, 9, 17
Butyrylcholinesterase deficiency	BCHE	2741	177400	2, 3, 4, 8, 9
Carbamoylphosphate synthetase I deficiency	CPS1	353	608307	2, 3, 4, 8, 9, 17
Carnitine deficiency	SLC22A5	1463	603377	2, 4, 8, 9, 17
Carnitine palmitoyltransferase 1A deficiency	CPT1A	1779	600528	2, 3, 4, 8, 9, 17
Carnitine palmitoyltransferase 1B deficiency	CPT1B	1362	601987	2, 4, 17
Carnitine palmitoyltransferase 2 deficiency, infantile	CPT2	354	600650	2, 3, 4, 8, 9, 17
Carnitine palmitoyltransferase 2 deficiency, lethal neonatal	CPT2	354	600650	2, 3, 4, 8, 9, 17
Carnitine-acylcarnitine translocase deficiency	SLC25A20	1573	613698	2, 3, 4, 8, 9, 17
Catechol-o-methyltransferase deficiency	COMT	2768	116790	2, 4
Ceroid lipofuscinosis neuronal type 1	PPT1	355	600722	2, 3, 4, 8, 9, 12
Ceroid lipofuscinosis neuronal type 2	TPP1	1613	607998	2, 3, 4, 8, 9, 12, 17
Ceroid lipofuscinosis neuronal type 3	CLN3	1273	607042	2, 3, 4, 8, 9
Ceroid lipofuscinosis neuronal type 4	DNAJC5	1569	611203	2, 4
Ceroid lipofuscinosis neuronal type 5	CLN5	358	608102	2, 3, 4, 8, 9
Ceroid lipofuscinosis neuronal type 6	CLN6	359	606725	2, 3, 4, 8, 9
Ceroid lipofuscinosis neuronal type 7	MFSD8	360	611124	2, 3, 4, 8, 9, 17
Ceroid lipofuscinosis neuronal type 8	CLN8	361	607837	2, 3, 4, 8, 9
Ceroid lipofuscinosis neuronal type 10	CTSD	356	116840	2, 3, 4, 8, 9, 17
Ceroid lipofuscinosis neuronal type 11	GRN	762	138945	2, 4, 8, 9, 17
Chanarin-Dorfman syndrome	ABHD5	362	604780	2, 3, 4, 8, 9
Chloramphenicol resistance, MT-RNR2 related	MT-RNR2	2556	561010	4
Cholestasis benign recurrent intrahepatic type 2	ABCB11	177	603201	2, 3, 4, 8, 9, 17
Cholestasis intrahepatic, of pregnancy, type 3	ABCB4	1858	171060	2, 4, 8, 9, 17
Cholestasis progressive intrahepatic type 1	ATP8B1	114	602397	2, 3, 4, 8, 9, 17
Cholestasis progressive intrahepatic type 2	ABCB11	177	603201	2, 3, 4, 8, 9, 17
Cholestasis progressive intrahepatic type 3	ABCB4	1858	171060	2, 4, 8, 9, 17
Cholestasis, benign recurrent intrahepatic	ATP8B1	114	602397	2, 3, 4, 8, 9, 17
Cholestasis, intrahepatic, of pregnancy, type 1	ATP8B1	114	602397	2, 3, 4, 8, 9, 17
Cholesteryl ester storage disease	LIPA	489	613497	2, 3, 4, 8, 9, 12
Chylomicron retention disease	SAR1B	3188	607690	2, 4
Citrin deficiency	SLC25A13	363	603859	2, 3, 4, 8, 9, 17
Citrullinemia	ASS1	364	603470	2, 3, 4, 8, 9, 17
Coenzyme Q10 deficiency type 6	COQ6	3773	614647	17
Coenzyme Q10 deficiency type 7	COQ4	3772	612898	17
Colchicine resistance	ABCB1	1790	171050	2, 4, 17
Combined D-2- and L-2-hydroxyglutaric aciduria	SLC25A1	2235	190315	2, 3, 4, 8, 9, 17
Combined malonic and methylmalonic aciduria	ACSF3	1454	614245	2, 4, 17
Combined oxidative phosphorylation deficiency type 1	GFM1	1406	606639	2, 4, 17
Combined oxidative phosphorylation deficiency type 2	MRPS16	981	609204	2, 4
Combined oxidative phosphorylation deficiency type 3	TSFM	1410	604723	2, 4, 17
Combined oxidative phosphorylation deficiency type 4	TUFM	1412	602389	2, 4, 17
Combined oxidative phosphorylation deficiency type 5	MRPS22	2500	605810	2, 4, 17
Combined oxidative phosphorylation deficiency type 6	AIFM1	1353	300169	2, 3, 4, 8, 9, 17
Combined oxidative phosphorylation deficiency type 7	C12ORF65	1365	613541	2, 4
Combined oxidative phosphorylation deficiency type 8	AARS2	2607	612035	2, 4, 17
Combined oxidative phosphorylation deficiency type 9	MRPL3	2184	607118	2, 4, 17
Combined oxidative phosphorylation deficiency type 10	MTO1	2316	614667	2, 3, 4, 8, 9, 17
Combined oxidative phosphorylation deficiency type 11	RMND1	2366	614917	2, 4, 17
Combined oxidative phosphorylation deficiency type 12	EARS2	2658	612799	2, 3, 4, 8, 9, 17
Combined oxidative phosphorylation deficiency type 13	PNPT1	2686	610316	2, 4, 17
Combined oxidative phosphorylation deficiency type 14	FARS2	2685	611592	2, 4, 17
Combined oxidative phosphorylation deficiency type 15	MTFMT	2025	611766	2, 3, 4, 8, 9, 17
Combined oxidative phosphorylation deficiency type 16	MRPL44	2684	611849	2, 4
Combined oxidative phosphorylation deficiency type 17	ELAC2	1197	605367	2, 4, 17

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Combined oxidative phosphorylation deficiency type 18	SFXN4	2345	615564	2, 4, 17
Combined oxidative phosphorylation deficiency type 19	LYRM4	2683	613311	2, 4
Combined oxidative phosphorylation deficiency type 20	VARS2	2507	612802	2, 4, 17
Combined oxidative phosphorylation deficiency type 21	TARS2	2682	612805	2, 4, 17
Combined oxidative phosphorylation deficiency type 22	ATP5F1A	2681	164360	2, 4, 17
Combined oxidative phosphorylation deficiency type 23	GTPBP3	2680	608536	2, 4, 17
Combined oxidative phosphorylation deficiency type 24	NARS2	2679	612803	2, 3, 4, 8, 9, 17
Combined oxidative phosphorylation deficiency type 25	MARS2	2678	609728	2, 4
Combined oxidative phosphorylation deficiency type 26	TRMT5	2677	611023	2, 4
Congenital disorder of glycosylation, type Ip	ALG11	2393	613666	2, 4
Congenital disorder of glycosylation, type Iq	SRD5A3	2394	611715	2, 4
Congenital disorder of glycosylation, type Iw	STT3A	2932	601134	2, 4, 17
Coproporphyria	CPOX	2223	612732	2, 4, 8, 9, 17
Coronary artery disease, susceptibility to	LPA	3823	152200	17
Coumarin resistance	VKORC1	2960	608547	2, 4
Coumarin/Warfarin resistance due to CYP2C9 variants	CYP2C9	2880	601130	2, 4, 8, 9
CR1 deficiency	CR1	3774	120620	17
Creatine deficiency syndrome X-linked	SLC6A8	276	300036	2, 4, 8, 9
CYP2C19 related poor drug metabolism	CYP2C19	1949	124020	2, 4, 8, 9, 17
Cystathioninuria	CTH	2845	219500	2, 4, 17
Cystic fibrosis	CFTR	370	602421	2, 4, 8
Cystic fibrosis, SLC6A14 related	SLC6A14	2700	300444	2, 3, 4, 8, 9, 17
Cystinosis, nephropathic	CTNS	371	606272	2, 3, 4, 8, 9, 17
Cytochrome P450 deficiency	CYP1A2	1988	124060	2, 4, 8, 9
D-2-hydroxyglutaric aciduria type 1	D2HGDH	1515	609186	2, 4, 8, 9, 17
D-2-hydroxyglutaric aciduria type 2	IDH2	2143	147650	2, 3, 4, 8, 9
D-bifunctional protein deficiency	HSD17B4	1095	601860	2, 3, 4, 8, 9, 17
D-glyceric aciduria	GLYCTK	2838	610516	2, 4
Diabetes insipidus, nephrogenic, X-linked	AVPR2	28	300538	2, 3, 4, 8, 9
Diabetes insipidus, neurohypophyseal	AVP	2785	192340	2, 3, 4, 8, 9
Diabetes mellitus type 1	INS	90	176730	2, 4, 8, 9
Diabetes mellitus, insulin-dependent type 20	HNF1A	450	142410	2, 4, 8, 9, 17
Diabetes mellitus, insulin-resistant with acanthosis nigricans	INSR	372	147670	2, 3, 4, 8, 9, 17
Diabetes mellitus, neonatal	GLIS3	1458	610192	2, 4, 17
Diabetes mellitus, noninsulin-dependent	ABCC8	429	600509	2, 4, 8, 9, 17
Diabetes mellitus, noninsulin-dependent	AKT2	2782	164731	2, 4, 17
Diabetes mellitus, noninsulin-dependent	KCNJ11	146	600937	2, 4, 8, 9
Diabetes mellitus, permanent neonatal	ABCC8	429	600509	2, 4, 8, 9, 17
Diabetes mellitus, transient neonatal type 2	ABCC8	429	600509	2, 4, 8, 9, 17
Diabetes, IGF2 related	IGF2	1751	147470	2, 4, 8, 9
Diarrhea type 1, secretory chloride, congenital	SLC26A3	2909	126650	2, 4, 17
Diarrhea type 3, secretory sodium, congenital, syndromic	SPINT2	3807	605124	17
Diarrhea type 4, malabsorptive, congenital	NEUROG3	1504	604882	2, 4, 8, 9
Dihydropyrimidine dehydrogenase deficiency	DPYD	2257	612779	2, 4, 8, 9, 17
Dihydropyrimidinuria	DPYS	2970	613326	2, 4
Dimethylglycine dehydrogenase deficiency	DMGDH	2470	605849	2, 4, 17
Dyggve-Melchior-Clausen disease	DYM	3043	607461	2, 4, 17
Efavirenz, poor metabolism of	CYP2B6	2502	123930	2, 4, 8, 9
Enterokinase deficiency	TMPRSS15	2516	606635	2, 4, 17
Erythrocyte lactate transporter defect	SLC16A1	2643	600682	2, 3, 4, 8, 9
Fabry disease	GLA	373	300644	2, 4, 8, 9, 12, 13
Factor II deficiency	F2	374	176930	2, 4, 17
Factor V deficiency	F5	375	612309	2, 4, 17
Factor XIIIB deficiency	F13B	2328	134580	2, 4, 17
Fanconi anemia type A	FANCA	376	607139	2, 4, 8, 9, 17
Fanconi anemia type B	FANCB	195	300515	2, 4, 8, 9, 17
Fanconi anemia type C	FANCC	378	613899	2, 4
Fanconi anemia type D1	BRCA2	379	600185	2, 4, 8, 9, 16
Fanconi anemia type D2	FANCD2	380	613984	2, 4, 8, 9, 17
Fanconi anemia type E	FANCE	381	613976	2, 4, 17
Fanconi anemia type F	FANCF	382	613897	2, 4
Fanconi anemia type G	FANCG	383	602956	2, 3, 4, 8, 9, 17
Fanconi anemia type I	FANCI	384	611360	2, 4, 17
Fanconi anemia type J	BRIP1	385	605882	2, 4, 8, 9, 17

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Fanconi anemia type L	FANCL	386	608111	2, 4, 17
Fanconi anemia type M	FANCM	387	609644	2, 4, 17
Fanconi anemia type N	PALB2	388	610355	2, 4, 8, 9, 17
Fanconi anemia type P	SLX4	389	613278	2, 4, 17
Fanconi anemia, XRCCR2 related	XRCC2	297	600375	2, 3, 4, 8, 9
Fanconi-Bickel syndrome	SLC2A2	390	138160	2, 3, 4, 8, 9, 17
Farber disease	ASAH1	391	613468	2, 3, 4, 8, 9, 13, 17
Favism, susceptibility to	G6PD	400	305900	2, 3, 4, 8, 9, 17
Fish eye disease	LCAT	1962	606967	2, 3, 4, 8, 9
Fructose-1,6-bisphosphatase deficiency	FBP1	392	611570	2, 3, 4, 8, 9
Fructose intolerance	ALDOB	393	612724	2, 3, 4, 8, 9, 17
Fructose uptake deficiency, SLC2A5 related	SLC2A5	2737	138230	2, 4
Fructosuria essential	KHK	394	614058	2, 4
Fucosidosis	FUCA1	197	612280	2, 3, 4, 8, 9, 12
Fumarase deficiency	FH	1335	136850	2, 4, 8, 9, 17
GABA-transaminase deficiency	ABAT	2776	137150	2, 4, 17
Galactokinase deficiency	GALK1	396	604313	2, 3, 4, 8, 9
Galactose epimerase deficiency	GALE	397	606953	2, 3, 4, 8, 9, 17
Galactosemia	GALT	398	606999	2, 4, 8, 9, 17
Galactosialidosis	CTSA	1433	613111	2, 3, 4, 8, 9, 17
Gallbladder disease type 1	ABCB4	1858	171060	2, 4, 8, 9, 17
Gaucher disease type 1	GBA	399	606463	2, 3, 4, 8, 9, 12, 13
Gaucher disease type 2	GBA	399	606463	2, 3, 4, 8, 9, 12, 13
Gaucher disease type 3C	GBA	399	606463	2, 3, 4, 8, 9, 12, 13
Gaucher disease type 3	GBA	399	606463	2, 3, 4, 8, 9, 12, 13
Gaucher disease, atypical	PSAP	232	176801	2, 3, 4, 8, 9, 17
Gaucher disease, perinatal lethal	GBA	399	606463	2, 3, 4, 8, 9
Glucocorticoid deficiency type 1	MC2R	1521	607397	2, 4
Glucocorticoid deficiency type 2	MRAP	1887	609196	2, 4
Glucocorticoid deficiency type 4, with or without mineralocorticoid deficiency	NNT	3148	607878	17
Glucose/Galactose malabsorption	SLC5A1	1977	182380	2, 3, 4, 8, 9, 17
Glutamate formiminotransferase deficiency	FTCD	2241	606806	2, 4, 17
Glutamine deficiency, congenital	GLUL	2616	138290	2, 4, 17
Glutaric acidemia type 1	GCDH	401	608801	2, 3, 4, 8, 9, 17
Glutaric acidemia type 2C	ETFDH	331	231675	2, 3, 4, 8, 9, 17
Glutaric aciduria type 3	SUGCT	2869	609187	2, 4, 17
Glutathione S-transferase theta-1 defficiency	GSTT1	1915	600436	2, 4, 8, 9
Glutathione synthetase deficiency	GSS	1570	601002	2, 3, 4, 8, 9, 17
Glycerol kinase deficiency	GK	402	300474	2, 3, 4, 8, 9, 17
Glycine encephalopathy with normal serum glycine	SLC6A9	3369	601019	17
Glycogen storage disease of heart (lethal)	PRKAG2	226	602743	2, 3, 4, 8, 9, 17
Glycogen storage disease type 0 muscle	GYS1	403	138570	2, 3, 4, 8, 9, 17
Glycogen storage disease type 0	GYS2	1875	138571	2, 3, 4, 8, 9, 17
Glycogen storage disease type 1A	G6PC	404	613742	2, 3, 4, 8, 9
Glycogen storage disease type 2	GAA	405	606800	2, 3, 4, 8, 9, 17
Glycogen storage disease type 3	AGL	9	610860	2, 3, 4, 8, 9, 17
Glycogen storage disease type 4	GBE1	340	607839	2, 3, 4, 8, 9, 17
Glycogen storage disease type 5	PYGM	235	608455	2, 3, 4, 8, 9, 17
Glycogen storage disease type 6B	PYGL	407	613741	2, 3, 4, 8, 9, 17
Glycogen storage disease type 7	PFKM	213	610681	2, 3, 4, 8, 9, 17
Glycogen storage disease type 9A	PHKA2	408	300798	2, 3, 4, 8, 9, 17
Glycogen storage disease type 9B	PHKB	409	172490	2, 3, 4, 8, 9, 17
Glycogen storage disease type 9C	PHKG2	1873	172471	2, 4, 17
Glycogen storage disease type 10	PGAM2	1392	612931	2, 3, 4, 8, 9
Glycogen storage disease type 11	LDHA	410	150000	2, 3, 4, 8, 9, 17
Glycogen storage disease type 12	ALDOA	1874	103850	2, 4, 17
Glycogen storage disease type 13	ENO3	411	131370	2, 4, 17
Glycogen storage disease type 14	PGM1	1393	171900	2, 3, 4, 8, 9, 17
Glycogen storage disease type 15	GYG1	1574	603942	2, 4, 17
Glycosylation disorder type 1A	PMM2	218	601785	2, 3, 4, 8, 9, 17
Glycosylation disorder type 1B	MPI	168	154550	2, 4, 17
Glycosylation disorder type 1C	ALG6	15	604566	2, 4, 17
Glycosylation disorder type 1D	ALG3	14	608750	2, 4, 17
Glycosylation disorder type 1E	DPM1	412	603503	2, 4, 17

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Glycosylation disorder type 1F	MPDU1	167	604041	2, 4
Glycosylation disorder type 1G	ALG12	12	607144	2, 4, 17
Glycosylation disorder type 1H	ALG8	37	608103	2, 4, 17
Glycosylation disorder type 1I	ALG2	13	607905	2, 4
Glycosylation disorder type 1J	DPAGT1	634	191350	2, 4, 17
Glycosylation disorder type 1K	ALG1	11	605907	2, 3, 4, 8, 9, 17
Glycosylation disorder type 1L	ALG9	17	606941	2, 4, 17
Glycosylation disorder type 1M	DOLK	414	610746	2, 4
Glycosylation disorder type 1N	RFT1	241	611908	2, 4, 17
Glycosylation disorder type 1O	DPM3	1575	605951	2, 4
Glycosylation disorder type 1S	ALG13	1726	300776	2, 4, 17
Glycosylation disorder type 1U	DPM2	2619	603564	2, 4
Glycosylation disorder type 2A	MGAT2	165	602616	2, 4
Glycosylation disorder type 2B	MOGS	1576	601336	2, 3, 4, 8, 9, 17
Glycosylation disorder type 2C	SLC35C1	274	605881	2, 4
Glycosylation disorder type 2D	B4GALT1	316	137060	2, 4
Glycosylation disorder type 2E	COG7	198	606978	2, 4, 17
Glycosylation disorder type 2F	SLC35A1	273	605634	2, 4, 17
Glycosylation disorder type 2G	COG1	416	606973	2, 3, 4, 8, 9, 17
Glycosylation disorder type 2H	COG8	417	606979	2, 4, 17
Glycosylation disorder type 2I	COG5	1578	606821	2, 4, 17
Glycosylation disorder type 2J	COG4	1577	606976	2, 4, 17
Glycosylation disorder type 2K	TMEM165	1196	614726	2, 3, 4, 8, 9
Glycosylation disorder type 2M	SLC35A2	2045	314375	2, 4
Glycosylation disorder type 3	COG6	1579	606977	2, 3, 4, 8, 9, 17
Glycosylation disorder type IR	DDOST	1107	602202	2, 4, 17
Glycosylation disorder x-linked	SSR4	1963	300090	2, 4, 17
GM1-gangliosidosis type 1	GLB1	127	611458	2, 3, 4, 8, 9, 17
GM1-gangliosidosis type 2	GLB1	127	611458	2, 3, 4, 8, 9, 17
GM2-gangliosidosis type 2	HEXB	133	606873	2, 3, 4, 8, 9, 17
Guanidinoacetate methyltransferase deficiency	GAMT	418	601240	2, 3, 4, 8, 9
Hartnup disorder	SLC6A19	419	608893	2, 3, 4, 8, 9
Hawkinsinuria	HPD	2841	609695	2, 4, 17
HDL deficiency, type 2	ABCA1	2098	600046	2, 4, 17
Hemochromatosis classical	HFE	420	613609	2, 4, 8, 9
Hemochromatosis type 2A	HJV	421	608374	2, 4, 8, 9
Hemochromatosis type 2B	HAMP	422	606464	2, 4, 8, 9
Hemochromatosis type 3	TFR2	423	604720	2, 4, 8, 9, 17
Hemochromatosis type 4	SLC40A1	424	604653	2, 4, 8, 9, 17
Hemolytic anemia due to G6PD deficiency	G6PD	400	305900	2, 3, 4, 8, 9, 17
Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency	GPI	3628	172400	17
Hemophilia A	F8	1879	300841	2, 4, 8, 9, 17
High density lipoprotein cholesterol level QTL 10	CETP	3821	118470	17
Holocarboxylase synthetase deficiency	HLCS	1459	609018	2, 3, 4, 8, 9, 17
Hurler syndrome	IDUA	141	252800	2, 3, 4, 8, 9, 12, 17
Hurler-Scheie syndrome	IDUA	141	252800	2, 3, 4, 8, 9, 12, 17
Hydrops, lactic acidosis, and sideroblastic anemia	LARS2	3633	604544	17
Hyperalphalipoproteinemia	CETP	3821	118470	17
Hyperammonemia due to carbonic anhydrase VA deficiency	CA5A	3287	114761	17
Hypercalcemia infantile type	CYP24A1	1888	126065	2, 3, 4, 8, 9, 17
Hyperchlorhidrosis, isolated	CA12	3769	603263	17
Hypercholanemia	BAAT	2186	602938	2, 4
Hypercholanemia	TJP2	1580	607709	2, 4, 17
Hypercholesterolemia autosomal dominant type 3	PCSK9	425	607786	2, 3, 4, 8, 9, 17
Hypercholesterolemia autosomal recessive	LDLRAP1	426	605747	2, 4, 17
Hypercholesterolemia due to LDL-receptor-disorder autosomal dominant	LDLR	427	606945	2, 4, 8, 9, 17
Hypercholesterolemia type B autosomanl dominant	APOB	428	107730	2, 3, 4, 8, 9, 17
Hypercholesterolemia, familial, due to LDLR defect, modifier of	EPHX2	3778	132811	17
Hypercholesterolemia, familial, modifier of	APOA2	3820	107670	17
Hypercholesterolemia, susceptibility to	ITIH4	3822	600564	17
Hypercholesterolemia, susceptibility to	PPP1R17	3824	604088	17
Hyperchylomicronemia type 5	APOA5	2406	606368	2, 4, 17
Hyperinsulinaemia, association with, G6PC2 related	G6PC2	1503	608058	2, 4
Hyperinsulinemic hypoglycemia type 1	ABCC8	429	600509	2, 4, 8, 9, 17

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Hyperinsulinemic hypoglycemia type 2	KCNJ11	146	600937	2, 4, 8, 9
Hyperinsulinemic hypoglycemia type 3	GCK	430	138079	2, 4, 8, 9, 17
Hyperinsulinemic hypoglycemia type 6	GLUD1	431	138130	2, 4, 17
Hyperinsulinemic hypoglycemia type 7	SLC16A1	2643	600682	2, 3, 4, 8, 9
Hyperinsulinism, UCP2 related	UCP2	2872	601693	2, 4, 17
Hyperlipidemia, familial combined, susceptibility to	USF1	2408	191523	2, 4
Hyperlipoproteinemia type 1	LPL	432	609708	2, 4, 8, 9, 17
Hyperlysinemia type 1	AASS	2775	605113	17
Hypermanganesemia with dystonia, polycythemia and cirrhosis	SLC30A10	745	611146	2, 3, 4, 8, 9
Hypermethioninemia due to adenosine kinase deficiency	ADK	2602	102750	2, 4, 17
Hyperornithinemia- Hyperammonemia - Homocitrullinuria syndrome	SLC25A15	2015	603861	2, 3, 4, 8, 9
Hyperoxaluria type 1	AGXT	433	604285	2, 4, 8, 9, 17
Hyperoxaluria type 2	GRHPR	434	604296	2, 4, 8, 9, 17
Hyperoxaluria type 3	HOGA1	1865	613597	2, 3, 4, 8, 9
Hyperoxaluria, SLC26A6 related	SLC26A6	2267	610068	2, 4, 17
Hyperphenylalaninemia, BH4 deficient, type C	QDPR	435	612676	2, 4, 17
Hyperphenylalaninemia, BH4 deficient, type D	PCBD1	2861	126090	2, 4
Hyperphenylalaninemia, BH4-deficient, type A	PTS	2501	612719	2, 3, 4, 8, 9, 17
Hyperprolinemia type 1	PRODH	2864	606810	2, 4, 17
Hyperprolinemia type 2	ALDH4A1	3765	606811	17
Hypertriglyceridemia, susceptibility to	LIPI	2407	609252	2, 4
Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis	SARS2	3802	612804	17
Hypoaldosteronism congenital due to CMO I deficiency	CYP11B2	79	124080	2, 3, 4, 8, 9, 17
Hypoaldosteronism, congenital, due to CMO II deficiency	CYP11B2	79	124080	2, 3, 4, 8, 9, 17
Hypoalphalipoproteinemia	APOA1	2106	107680	2, 3, 4, 8, 9
Hypobetalipoproteinemia type 1	APOB	428	107730	2, 3, 4, 8, 9, 17
Hypocalcemia, autosomal dominant 2	GNA11	2074	139313	2, 4, 17
Hypocalciuric hypercalcemia, familial type 3	AP2S1	2073	602242	2, 4
Hypoglycemia of infancy, leucine-sensitive	ABCC8	429	600509	2, 4, 8, 9, 17
Hypoinsulinemic hypoglycemia with hemihypertrophy	AKT2	2782	164731	2, 4, 17
Hypomagnesemia type 1	TRPM6	2035	607009	2, 3, 4, 8, 9, 17
Hypomagnesemia type 2	FXYD2	1581	601814	2, 4
Hypomagnesemia type 3	CLDN16	1582	603959	2, 3, 4, 8, 9
Hypomagnesemia type 4	EGF	1946	131530	2, 3, 4, 8, 9, 17
Hypomagnesemia type 5	CLDN19	1583	610036	2, 4
Hypomagnesemia type 6	CNNM2	1890	607803	2, 3, 4, 8, 9, 17
Hypophosphatasia, adult	ALPL	2237	171760	2, 3, 4, 8, 9, 17
Hypophosphatasia, childhood	ALPL	2237	171760	2, 3, 4, 8, 9, 17
Hypophosphatasia, infantile	ALPL	2237	171760	2, 3, 4, 8, 9, 17
Hypophosphatemic rickets with hypercalciuria	SLC34A3	2649	609826	2, 3, 4, 8, 9, 17
Hypophosphatemic rickets, autosomal dominant	FGF23	1133	605380	2, 4, 8, 9
Hypouricemia, renal type 1	SLC22A12	2295	607096	2, 4
Hypouricemia, renal type 2	SLC2A9	2296	606142	2, 4, 17
Insulin-like growth factor resistance	IGF1R	1725	147370	2, 4, 8, 9, 17
Isobutyryl-CoA dehydrogenase deficiency	ACAD8	2777	604773	2, 4, 17
Isovaleric acidemia	IVD	436	607036	2, 3, 4, 8, 9, 17
Krabbe disease	GALC	437	606890	2, 4, 8, 9, 12, 17
Krabbe disease, atypical	PSAP	232	176801	2, 3, 4, 8, 9, 17
L-2-hydroxyglutaric aciduria	L2HGDH	438	609584	2, 4, 8, 9, 17
Lactase deficiency, congenital	LCT	2852	603202	2, 4, 17
Lactate dehydrogenase-B deficiency	LDHB	3154	150100	2, 4
Lacticacidemia due to PDX1 deficiency	PDHX	1396	608769	2, 3, 4, 8, 9, 17
Lactose intolerance, adult type	MCM6	1930	601806	2, 4, 17
LCAD deficiency	ACADL	1348	609576	2, 4, 17
LCAT DEFICIENCY	LCAT	1962	606967	2, 3, 4, 8, 9
Leukocyte adhesion deficiency	ITGB1	2220	135630	2, 4, 17
Leukocyte adhesion deficiency	ITGB2	2221	600065	2, 4, 17
Lipodystrophy generalized type 1	AGPAT2	439	603100	2, 3, 4, 8, 9
Lipodystrophy generalized type 2	BSCL2	440	606158	2, 3, 4, 8, 9, 17
Lipodystrophy generalized type 4	CAVIN1	2207	603198	2, 4, 8, 9
Lipodystrophy type 2, familial partial	LMNA	158	150330	2, 4, 8, 9, 17
Lipodystrophy, familial partial, type 3	PPARG	2566	601487	2, 4, 8, 9, 17
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA	132	600890	2, 3, 4, 8, 9, 17
LPA deficiency, congenital	LPA	3823	152200	17

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Lung alpha-beta hydrolase deficiency type 1	ABHD1	1828	612195	2, 4
Lysosomal acid phosphatase deficiency	ACP2	1883	171650	2, 4, 17
Malonyl-CoA decarboxylase deficiency	MLYCD	2183	606761	2, 4, 8, 9
Mannose-binding protein deficiency	MBL2	441	154545	2, 3, 4, 8, 9
Mannosidosis, beta A, lysosomal-like	MANBAL	2414		2, 4
Mannosidosis-alpha	MAN2B1	442	609458	2, 3, 4, 8, 9, 12, 17
Mannosidosis-beta	MANBA	443	609489	2, 3, 4, 8, 9, 12, 17
Maple syrup urine disease type 1a	BCKDHA	444	608348	2, 3, 4, 8, 9, 17
Maple syrup urine disease type 1b	BCKDHB	445	248611	2, 3, 4, 8, 9, 17
Maple syrup urine disease type 2	DBT	446	248610	2, 3, 4, 8, 9, 17
Maple syrup urine disease type 3	DLD	447	238331	2, 3, 4, 8, 9, 17
Maple syrup urine disease, mild variant	PPM1K	2413	611065	2, 4
Maturity-onset diabetes of the young type 1	HNF4A	448	600281	2, 4, 8, 9, 17
Maturity-onset diabetes of the young type 2	GCK	430	138079	2, 4, 8, 9, 17
Maturity-onset diabetes of the young type 3	HNF1A	450	142410	2, 4, 8, 9, 17
Maturity-onset diabetes of the young type 4	PDX1	451	600733	2, 4, 8, 9
Maturity-onset diabetes of the young type 5	HNF1B	452	189907	2, 4, 8, 9, 17
Maturity-onset diabetes of the young type 6	NEUROD1	126	601724	2, 4, 8, 9
Maturity-onset diabetes of the young type 7	KLF11	106	603301	2, 4, 8, 9
Maturity-onset diabetes of the young type 8	CEL	107	114840	2, 4, 8, 9, 17
Maturity-onset diabetes of the young type 9	PAX4	108	167413	2, 4, 8, 9, 17
Maturity-onset diabetes of the young type 10	INS	90	176730	2, 4, 8, 9
Maturity-onset diabetes of the young type 11	BLK	1502	191305	2, 4, 17
Maturity-onset diabetes of the young, NKX2-2 related	NKX2-2	1505	604612	2, 4
Maturity-onset diabetes of the young, RFX6 related	RFX6	1506	612659	2, 3, 4, 8, 9, 17
Maturity-onset diabetes of the young, ZFP57 related	ZFP57	1507	612192	2, 4, 8, 9
Mediterranean fever	MEFV	786	608107	2, 4, 8, 9, 17
MELAS syndrome, MT-TL1 related	MT-TL1	1317	590050	4
Metachromatic Leukodystrophy	ARSA	23	607574	2, 3, 4, 8, 9, 12
Methylacetoacetic aciduria	ACAT1	453	607809	2, 3, 4, 8, 9, 17
Methylcobalamin deficiency CblG type	MTR	173	156570	2, 4, 17
Methylmalonate semialdehyde dehydrogenase deficiency	ALDH6A1	2784	603178	2, 4, 17
Methylmalonic aciduria CblA type	MMAA	456	607481	2, 4
Methylmalonic aciduria CblB type	MMAB	457	607568	2, 4, 17
Methylmalonic aciduria CblC type	MMACHC	458	609831	2, 3, 4, 8, 9
Methylmalonic aciduria CblD type	MMADHC	454	611935	2, 3, 4, 8, 9
Methylmalonic aciduria CblF type	LMBRD1	729	612625	2, 4, 17
Methylmalonic aciduria CblJ type	ABCD4	1727	603214	2, 4, 17
Methylmalonic aciduria CblR type	CD320	1456	606475	2, 3, 4, 8, 9
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	MMUT	455	609058	2, 3, 4, 8, 9, 17
Methylmalonyl-CoA epimerase deficiency	MCEE	2181	608419	2, 4
Mevalonic aciduria	MVK	1741	251170	2, 3, 4, 8, 9, 17
Microvascular complications of diabetes type 1	VEGFA	1584	192240	2, 4, 17
Microvascular complications of diabetes type 6, susceptibility to	SOD2	2416	147460	2, 4
Mirage syndrome	SAMD9	3621	610456	17
Mitchell-Riley syndrome	RFX6	1506	612659	2, 3, 4, 8, 9, 17
Mitochondrial complex III deficiency, nuclear type 7	UQCC2	2873	614461	2, 4
Mitochondrial phosphate carrier deficiency	SLC25A3	3803	600370	17
Mitochondrial pyruvate carrier deficiency	MPC1	2849	614738	2, 4
Molybdenum cofactor deficiency type A	MOCS1	1127	603707	2, 3, 4, 8, 9, 17
Molybdenum cofactor deficiency type B	MOCS2	949	603708	2, 3, 4, 8, 9
Molybdenum cofactor deficiency type C	GPHN	1567	603930	2, 3, 4, 8, 9, 17
Monocarboxylate transporter 1 deficiency	SLC16A1	2643	600682	2, 3, 4, 8, 9
Mucolipidosis type 2 alpha/beta	GNPTAB	130	607840	2, 3, 4, 8, 9, 17
Mucolipidosis type 3 gamma	GNPTG	1585	607838	2, 3, 4, 8, 9, 17
Mucolipidosis type 3	GNPTAB	130	607840	2, 3, 4, 8, 9, 17
Mucolipidosis type 4	MCOLN1	459	605248	2, 3, 4, 8, 9, 17
Mucopolysaccharidosis type 1H	IDUA	141	252800	2, 3, 4, 8, 9, 12, 17
Mucopolysaccharidosis type 2	IDS	140	300823	2, 4, 8, 9, 12, 17
Mucopolysaccharidosis type 3A	SGSH	460	605270	2, 3, 4, 8, 9
Mucopolysaccharidosis type 3B	NAGLU	461	609701	2, 3, 4, 8, 9, 12
Mucopolysaccharidosis type 3C	HGSNAT	462	610453	2, 3, 4, 8, 9, 17
Mucopolysaccharidosis type 3D	GNS	96	607664	2, 3, 4, 8, 9, 17
Mucopolysaccharidosis type 4A	GALNS	463	612222	2, 3, 4, 8, 9, 12, 17

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Mucopolysaccharidosis type 4B	GLB1	127	611458	2, 3, 4, 8, 9, 12, 17
Mucopolysaccharidosis type 6	ARSB	464	611542	2, 3, 4, 8, 9, 12
Mucopolysaccharidosis type 7	GUSB	465	611499	2, 3, 4, 8, 9, 12, 17
Mucopolysaccharidosis type 9	HYAL1	466	607071	2, 4
Muscle glycogenosis	PHKA1	1394	311870	2, 17
Myopathy due to myoadenylate deaminase deficiency	AMPD1	467	102770	2, 3, 4, 8, 9, 17
N-acetylglutamate synthase deficiency	NAGS	468	608300	2, 3, 4, 8, 9, 17
Neuraminidase deficiency	NEU1	181	608272	2, 3, 4, 8, 9
Niemann-Pick disease type A/B	SMPD1	469	607608	2, 4, 8, 9, 12, 13
Niemann-Pick disease type C1	NPC1	189	607623	2, 4, 8, 9, 13, 17
Niemann-Pick disease type C2	NPC2	470	601015	2, 4, 8, 9, 13
Odontohypophosphatasia	ALPL	2237	171760	2, 3, 4, 8, 9, 17
Ornithine transcarbamoylase deficiency	OTC	206	300461	2, 4, 8, 9, 17
Orotic aciduria	UMPS	471	613891	2, 3, 4, 8, 9
Pancreatic agenesis type 2	PTF1A	2865	607194	2, 4, 8, 9
Pancreatic and cerebellar agenesis	PTF1A	2865	607194	2, 4, 8, 9
Pentosuria	DCXR	1234	608347	2, 4
Periodic fever autosomal dominant	TNFRSF1A	476	191190	2, 3, 4, 8, 9, 17
Phenylketonuria	PAH	1461	612349	2, 4, 8, 9, 17
Phenylketonuria modifier, SLC7A5 related	SLC7A5	2648	600182	2, 4, 17
Phosphoenolpyruvate carboxykinase deficiency, cytosolic	PCK1	2670	614168	2, 4, 17
Phosphoenolpyruvate carboxykinase deficiency, mitochondrial	PCK2	2655	614095	2, 4, 17
Phosphoglycerate dehydrogenase deficiency	PHGDH	669	606879	2, 3, 4, 8, 9, 17
Phosphoribosylpyrophosphate synthetase superactivity	PRPS1	231	311850	2, 3, 4, 8, 9
Phosphoserine aminotransferase deficiency	PSAT1	1993	610936	2, 4, 17
Phosphoserine phosphatase deficiency	PSPH	1994	172480	2, 4
Pituitary stalk interruption syndrome, GPR161 related	GPR161	2503		2, 4
Pompe disease	GAA	405	606800	2, 4, 8, 9, 12, 17
Porphyria acute intermittent	HMBS	478	609806	2, 4, 8, 9, 17
Porphyria congenital erythropoietic	UROS	1980	606938	2, 4, 8, 9, 17
Porphyria variegata	PPOX	2666	600923	2, 4, 8, 9, 17
Prolidase deficiency	PEPD	2206	613230	2, 4, 17
Propionic acidemia	PCCA	479	232000	2, 4, 8, 9, 17
Propionic acidemia	PCCB	480	232050	2, 3, 4, 8, 9, 17
Prosaposin deficiency	PSAP	232	176801	2, 3, 4, 8, 9, 17
Protoporphyria, erythropoietic, X-linked	ALAS2	2783	301300	2, 4, 17
Pseudohermaphroditism with gynecomastia	HSD17B3	2088	605573	2, 4
Pyridoxamine 5'-phosphate oxidase deficiency	PNPO	1586	603287	2, 3, 4, 8, 9
Pyruvate carboxylase deficiency	PC	519	608786	2, 3, 4, 8, 9, 17
Pyruvate dehydrogenase E1-alpha deficiency	PDHA1	211	300502	2, 4, 8, 9, 17
Pyruvate dehydrogenase E1-beta deficiency	PDHB	1395	179060	2, 3, 4, 8, 9, 17
Pyruvate dehydrogenase E2 deficiency	DLAT	1438	608770	2, 4, 17
Pyruvate dehydrogenase lipoic acid synthetase deficiency	LIAS	2218	607031	2, 4, 17
Pyruvate dehydrogenase phosphatase deficiency	PDP1	743	605993	2, 4, 17
Pyruvate kinase deficiency with hemolytic anemia	PKLR	481	609712	2, 4, 8, 9
Refsum disease	PEX7	212	601757	2, 3, 4, 8, 9, 17
Refsum disease	PHYH	215	602026	2, 3, 4, 8, 9, 17
Riboflavin deficiency	SLC52A1	2867	607883	2, 4
Ribose 5-phosphate isomerase deficiency	RPIA	3801	180430	17
Rickets, vitamin D 25-hydroxylation-deficient, type 1B	CYP2R1	2279	608713	2, 4
Rickets, vitamin D dependent, type 1	CYP27B1	2959	609506	2, 4, 17
Saccharopinuria	AASS	2775	605113	17
Sandhoff disease	HEXB	133	606873	2, 3, 4, 8, 9, 12, 17
Sarcosinemia	SARDH	2617	604455	2, 4, 17
Scheie syndrome	IDUA	141	252800	2, 3, 4, 8, 9, 12, 17
Schindler disease	NAGA	483	104170	2, 4, 12
Serine hydrolase deficiency, SERHL2 related	SERHL2	2308		2, 4
Succinic semialdehyde dehydrogenase deficiency	ALDH5A1	2266	610045	2, 4, 17
Succinyl CoA:3-oxoacid CoA transferase deficiency	OXCT1	1762	601424	2, 3, 4, 8, 9, 17
Sucrase-isomaltase deficiency	SI	1808	609845	2, 17
Sulfatase deficiency	SUMF1	285	607939	2, 3, 4, 8, 9, 17
Sulfite oxidase deficiency	SUOX	1821	606887	2, 3, 4, 8, 9
Surfactant metabolism dysfunction	SFTPD	2056	178635	2, 4, 17
Surfactant metabolism dysfunction type 1	SFTPB	1810	178640	2, 3, 4, 8, 9, 17

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Surfactant metabolism dysfunction type 2	SFTPC	1811	178620	2, 3, 4, 8, 9
Surfactant metabolism dysfunction type 3	ABCA3	1812	601615	2, 3, 4, 8, 9, 17
Surfactant metabolism dysfunction type 4	CSF2RA	1813	306250	2, 4, 8, 9, 17
Surfactant metabolism dysfunction type 5	CSF2RB	1814	138981	2, 4, 17
Tangier disease	ABCA1	2098	600046	2, 4, 17
Tay-Sachs disease	HEXA	484	606869	2, 4, 8, 9, 12, 17
Tay-Sachs disease AB variant	GM2A	485	613109	2, 3, 4, 8, 9
Thiamine metabolism dysfunction syndrome type 5	TPK1	1792	606370	2, 4, 17
TJP1 deficiency	TJP1	2113	601009	2, 4
TPMT deficiency	TPMT	1475	187680	2, 4, 8, 9
Transaldolase deficiency	TALDO1	1937	602063	2, 3, 4, 8, 9
Transcobalamin II deficiency	TCN2	45	613441	2, 4, 17
Trifunctional protein deficiency	HADHA	132	600890	2, 3, 4, 8, 9, 17
Trimethylaminuria	FMO3	1763	136132	2, 3, 4, 8, 9
Triosephosphate isomerase deficiency	TPI1	2902	190450	2, 4, 17
Tumoral calcinosis, hyperphosphatemic, familial, type 1	GALNT3	3557	601756	17
Tyrosine kinase 2 deficiency	TYK2	2079	176941	2, 3, 4, 8, 9, 17
Tyrosinemia type 1B	GSTZ1	1876	603758	2, 3, 4, 8, 9
Tyrosinemia type 1	FAH	486	613871	2, 3, 4, 8, 9, 17
Tyrosinemia type 2	TAT	2230	613018	2, 3, 4, 8, 9, 17
Tyrosinemia type 3	HPD	2841	609695	2, 4, 17
Urbach-Wiethe disease	ECM1	1896	602201	2, 4
Von-Gierke disease	G6PC	404	613742	2, 3, 4, 8, 9
Wilson disease	ATP7B	26	606882	2, 4, 8, 9
Wolman disease	LIPA	489	613497	2, 3, 4, 8, 9, 12
Xanthinuria type 1	XDH	1981	607633	2, 4, 17
Xanthinuria type 2	MOCOS	3789	613274	17

1: Hotspot Testing
2: Carrier Testing (point mutation)
3: Carrier Testing (del/dup)
4: Single Gene Sequencing (Sanger)
5: NGS Panel
6: NGS Panel + CNV
7: NGS Panel Genomic
8: Deletion/Duplication with qPCR/MLPA
9: Sequencing + Deletion/Duplication
10: Repeat Expansions
11: Somatic Mutation Analysis
12: Biochemical Enzyme Analysis
13: Biomarker Analysis
14: Biochemical Enzyme Panel
15: Biochemical Genetics Panel Plus
16: Single Gene Sequencing (NGS)
17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
Amyotrophic lateral sclerosis (ALS) panel	ALS2, ANG, C9orf72, CHCHD10, CHMP2B, CSF1R, DCTN1, ERBB4, FIG4, FUS, GRN, HNRNPA1, ITM2B, KIF5A, MAPT, MATR3, NEFH, OPTN, PFN1, PRNP, PRPH, PSEN1, PSEN2, SETX, SIGMAR1, SLC52A3, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TREM2, TUBA4A, UBQLN2, VAPB, VCP		5467	6
Amyotrophic lateral sclerosis (ALS) panel	ALS2, ANG, C9orf72, CHCHD10, CHMP2B, CSF1R, DCTN1, ERBB4, FIG4, FUS, GRN, HNRNPA1, ITM2B, KIF5A, MAPT, MATR3, NEFH, OPTN, PFN1, PRNP, PRPH, PSEN1, PSEN2, SETX, SIGMAR1, SLC52A3, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TREM2, TUBA4A, UBQLN2, VAPB, VCP		5467	6
Ataxia comprehensive panel	ABCB7, ABHD12, ABHD5, ACADVL, ACO2, AFG3L2, AHI1, ALDH5A1, AMACR, ANO10, AP1S2, APTX, ARL13B, ARL6, ARSA, ATCAY, ATM, ATN1, ATP13A2, ATP1A3, ATP2B3, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, B9D1, BBS1, BBS12, BEAN1, BSCL2, BTD, C12ORF65, C19orf12, CA8, CACNA1A, CACNB4, CAMTA1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CHMP1A, CLCN2, CLN5, CLN6, CLPP, COASY, COQ2, COQ8A, COQ9, COX20, CP, CPLANE1, CSPP1, CWF19L1, CYP27A1, DARS2, DLAT, DNAJC19, DNAJC5, DNMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EXOSC3, FA2H, FBXL4, FGF14, FLVCR1, FTL, FXN, GALC, GBA, GBA2, GFAP, GJB1, GJC2, GOSR2, GRID2, GRM1, GSS, HEPACAM, HEXB, HIBCH, INPP5E, ITM2B, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1A, KIF1C, KIF5A, KIF7, LAMA1, LMNB1, LRPPRC, MARS2, MKS1, MLC1, MRE11, MTFMT, MTPAP, MTTP, NDUFAF6, NDUFS1, NDUFS2, NDUFS4, NDUFS7, NDUFV1, NOP56, NPC1, NPC2, NPHP1, NUBPL, OFD1, OPA1, OPA3, OPHN1, PANK2, PAX6, PDHX, PDSS1, PDSS2, PDYN, PEX10, PEX2, PEX7, PHYH, PLA2G6, PLP1, PNKD, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PPP2R2B, PRICKLE1, PRKCG, PRRT2, RARS2, RPGRIP1L, RRM2B, RUBCN, SACS, SCN2A, SETX, SIL1, SLC16A2, SLC17A5, SLC1A3, SLC20A2, SLC25A46, SLC2A1, SLC52A3, SLC9A6, SPG11, SPG7, SPR, SPTBN2, STUB1, SYNE1, TBP, TCTN2, TGM6, TMEM216, TMEM237, TMEM240, TMEM67, TPP1, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TTPA, TUBB4A, TWNK, UBA5, VAMP1, VLDLR, VRK1, WDR81, WFS1, WWOX, ZFYVE26		5459	6

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17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
Ataxia comprehensive panel	ABCB7, ABHD12, ABHD5, ACADVL, ACO2, AFG3L2, AHI1, ALDH5A1, AMACR, ANO10, AP1S2, APTX, ARL13B, ARL6, ARSA, ATCAY, ATM, ATN1, ATP13A2, ATP1A3, ATP2B3, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, B9D1, BBS1, BBS12, BEAN1, BSCL2, BTD, C12ORF65, C19orf12, CA8, CACNA1A, CACNB4, CAMTA1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CHMP1A, CLCN2, CLN5, CLN6, CLPP, COASY, COQ2, COQ8A, COQ9, COX20, CP, CPLANE1, CSPP1, CWF19L1, CYP27A1, DARS2, DLAT, DNAJC19, DNAJC5, DNMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EXOSC3, FA2H, FBXL4, FGF14, FLVCR1, FTL, FXN, GALC, GBA, GBA2, GFAP, GJB1, GJC2, GOSR2, GRID2, GRM1, GSS, HEPACAM, HEXB, HIBCH, INPP5E, ITM2B, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1A, KIF1C, KIF5A, KIF7, LAMA1, LMNB1, LRPPRC, MARS2, MKS1, MLC1, MRE11, MTFMT, MTPAP, MTTP, NDUFAF6, NDUFS1, NDUFS2, NDUFS4, NDUFS7, NDUFV1, NOP56, NPC1, NPC2, NPHP1, NUBPL, OFD1, OPA1, OPA3, OPHN1, PANK2, PAX6, PDHX, PDSS1, PDSS2, PDYN, PEX10, PEX2, PEX7, PHYH, PLA2G6, PLP1, PNKD, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PPP2R2B, PRICKLE1, PRKCG, PRRT2, RARS2, RPGRIP1L, RRM2B, RUBCN, SACS, SCN2A, SETX, SIL1, SLC16A2, SLC17A5, SLC1A3, SLC20A2, SLC25A46, SLC2A1, SLC52A3, SLC9A6, SPG11, SPG7, SPR, SPTBN2, STUB1, SYNE1, TBP, TCTN2, TGM6, TMEM216, TMEM237, TMEM240, TMEM67, TPP1, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TTPA, TUBB4A, TWNK, UBA5, VAMP1, VLDLR, VRK1, WDR81, WFS1, WWOX, ZFYVE26		5459	6
Ataxia panel	ABCB7, ABHD12, ABHD5, ACADVL, ACO2, AFG3L2, AHI1, ALDH5A1, AMACR, ANO10, AP1S2, APTX, ARL13B, ARL6, ARSA, ATCAY, ATM, ATN1, ATP13A2, ATP1A3, ATP2B3, ATP8A2, B9D1, BBS1, BBS12, BSCL2, BTD, C12orf65, C19orf12, CA8, CACNA1A, CACNB4, CAMTA1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CHMP1A, CLCN2, CLN5, CLN6, CLPP, COASY, COQ2, COQ8A, COQ9, COX20, CP, CPLANE1, CSPP1, CWF19L1, CYP27A1, DARS2, DLAT, DNAJC19, DNAJC5, DNMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EXOSC3, FA2H, FBXL4, FGF14, FLVCR1, FTL, FXN, GALC, GBA, GBA2, GFAP, GJB1, GJC2, GOSR2, GRID2, GRM1, GSS, HEPACAM, HEXB, HIBCH, INPP5E, ITM2B, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1A, KIF1C, KIF5A, KIF7, LAMA1, LMNB1, LRPPRC, MARS2, MKS1, MLC1, MRE11, MTFMT, MTPAP, MTTP, NDUFAF6, NDUFS1, NDUFS2, NDUFS4, NDUFS7, NDUFV1, NPC1, NPC2, NPHP1, NUBPL, OFD1, OPA1, OPA3, OPHN1, PANK2, PAX6, PDHX, PDSS1, PDSS2, PDYN, PEX10, PEX2, PEX7, PHYH, PLA2G6, PLP1, PNKD, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PRICKLE1, PRKCG, PRRT2, RARS2, RPGRIP1L, RRM2B, RUBCN, SACS, SCN2A, SETX, SIL1, SLC16A2, SLC17A5, SLC1A3, SLC20A2, SLC25A46, SLC2A1, SLC52A3, SLC9A6, SPG11, SPG7, SPR, SPTBN2, STUB1, SYNE1, TCTN2, TGM6, TMEM216, TMEM237, TMEM240, TMEM67, TPP1, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TTPA, TUBB4A, TWNK, UBA5, VAMP1, VLDLR, VRK1, WDR81, WFS1, WWOX, ZFYVE26		5438	6

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17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
Ataxia panel	ABCB7, ABHD12, ABHD5, ACADVL, ACO2, AFG3L2, AHI1, ALDH5A1, AMACR, ANO10, AP1S2, APTX, ARL13B, ARL6, ARSA, ATCAY, ATM, ATN1, ATP13A2, ATP1A3, ATP2B3, ATP8A2, B9D1, BBS1, BBS12, BSCL2, BTD, C12orf65, C19orf12, CA8, CACNA1A, CACNB4, CAMTA1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CHMP1A, CLCN2, CLN5, CLN6, CLPP, COASY, COQ2, COQ8A, COQ9, COX20, CP, CPLANE1, CSPP1, CWF19L1, CYP27A1, DARS2, DLAT, DNAJC19, DNAJC5, DNMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EXOSC3, FA2H, FBXL4, FGF14, FLVCR1, FTL, FXN, GALC, GBA, GBA2, GFAP, GJB1, GJC2, GOSR2, GRID2, GRM1, GSS, HEPACAM, HEXB, HIBCH, INPP5E, ITM2B, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1A, KIF1C, KIF5A, KIF7, LAMA1, LMNB1, LRPPRC, MARS2, MKS1, MLC1, MRE11, MTFMT, MTPAP, MTTP, NDUFAF6, NDUFS1, NDUFS2, NDUFS4, NDUFS7, NDUFV1, NPC1, NPC2, NPHP1, NUBPL, OFD1, OPA1, OPA3, OPHN1, PANK2, PAX6, PDHX, PDSS1, PDSS2, PDYN, PEX10, PEX2, PEX7, PHYH, PLA2G6, PLP1, PNKD, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PRICKLE1, PRKCG, PRRT2, RARS2, RPGRIP1L, RRM2B, RUBCN, SACS, SCN2A, SETX, SIL1, SLC16A2, SLC17A5, SLC1A3, SLC20A2, SLC25A46, SLC2A1, SLC52A3, SLC9A6, SPG11, SPG7, SPR, SPTBN2, STUB1, SYNE1, TCTN2, TGM6, TMEM216, TMEM237, TMEM240, TMEM67, TPP1, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TTPA, TUBB4A, TWNK, UBA5, VAMP1, VLDLR, VRK1, WDR81, WFS1, WWOX, ZFYVE26		5438	6
Ataxia repeat expansion panel	ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BEAN1, CACNA1A, FXN, NOP56, PPP2R2B, TBP	CACNA1A, FXN	5466	
Ataxia repeat expansion panel	ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BEAN1, CACNA1A, FXN, NOP56, PPP2R2B, TBP	CACNA1A, FXN	5466	
CentoArrayCyto® 750K	Genome-wide CMA with 750,000 markers		50068	5
CentoArrayCyto® HD	Genome-wide CMA with 2.6 million markers		50001	5
CentoDx®	Targets exonic regions of >6500 genes associated with known clinical phenotypes		50158	5

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16: Single Gene Sequencing (NGS)
17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoIEM	AARS2, ABCA1, ABCB4, ABCC2, ABCC8, ABCD1, ABCD3, ABCD4, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACOX1, ACSF3, ACY1, ADA, ADAMTSL2, ADAR, ADGRG1, ADK, ADSL, AFG3L2, AGA, AGL, AGPAT2, AGPS, AGXT, AHCY, AIFM1, AIMP1, AKT2, ALAD, ALAS2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALPL, AMN, AMT, ANK1, ANTXR2, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, APOA5, APOB, APOC2, APOE, APP, APTX, ARG1, ARSA, ARSB, ASAH1, ASL, ASPA, ASS1, ATM, ATP13A2, ATP6V0A2, ATP7A, ATP7B, ATPAF2, AUH, B3GALNT2, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCS1L, BEST1, BLK, BOLA3, BRAT1, BSCL2, BTD, C12orf65, C19orf12, CA5A, CASP10, CASP8, CAV1, CAVIN1, CBLIF, CBS, CD320, CEL, CERS1, CIDEC, CISD2, CLCN2, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COL4A1, COL4A2, COQ2, COQ8A, COQ9, COX10, COX15, COX20, COX6B1, CP, CPOX, CPS1, CPT1A, CPT2, CSF1R, CTC1, CTH, CTLA4, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, CUBN, CYP11B1, CYP17A1, CYP19A1, CYP21A2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS2, DBT, DCAF17, DDC, DDOST, DGUOK, DHCR7, DHDDS, DIABLO, DKC1, DLAT, DLD, DNAJC5, DNM1L, DOLK, DPM1, DPM2, DPM3, DPYD, DYM, EARS2, ECHS1, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ENO3, ENPP1, EPB42, EPHX2, EPM2A, ERCC6, ETFA, ETFB, ETFDH, ETHE1, F2, F5, FA2H, FADD, FAH, FAM126A, FARS2, FAS, FASLG, FASTKD2, FBN1, FBP1, FBXL4, FECH, FGF23, FH, FHL1, FOLR1, FOXP3, FOXRED1, FTL, FUCA1, G6PC, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GATA6, GATM, GBA, GBE1, GCDH, GCK, GCSH, GFAP, GFER, GFM1, GFM2, GFPT1, GHR, GJA1, GJB1, GJC2, GK, GLA, GLB1, GLDC, GLIS3, GLRX5, GLUD1, GLUL, GM2A, GMPPA, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GPC3, GRN, GTPBP3, GUSB, GYG1, GYS1, GYS2, HADH, HADHA, HADHB, HAMP, HCFC1, HEPACAM, HEXA, HEXB, HFE, HGD, HGSNAT, HIBCH, HJV, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPRT1, HRAS, HSD17B10, HSD17B4, HSD3B2, HSPD1, HTRA1, HYAL1, IARS2, IBA57, IDS, IDUA, IER3IP1, IFIH1, INS, INSR, ISCA2, ITK, IVD, JAG1, JAM3, KCNC1, KCNJ10, KCNJ11, KCNT1, KCTD7, KIF5A, KLF11, KRAS, L2HGDH, LAMA2, LAMB1, LAMP2, LARGE1, LDB3, LDHA, LDLR, LDLRAP1, LIAS, LIPA, LIPE, LIPT1, LMBRD1, LMNA, LMNB1, LPIN1, LPL, LRPPRC, LYRM7, LYST, MAGT1, MAN1B1, MAN2B1, MANBA, MARS2, MCCC1, MCCC2, MCEE, MCOLN1, MFSD8, MGAT2, MGME1, MLC1, MLPH, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MRPL44, MRPS22, MTFMT, MTHFR, MTR, MTRR, MYO5A, MYOT, NAGA, NAGLU, NAGS, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF4, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEU1, NEUROD1, NEUROG3, NFU1, NGLY1, NHLRC1, NOTCH3, NPC1, NPC2, NRAS, NUBPL, OAT, OCLN, OCRL, OTC, OXCT1, PAH, PANK2, PAX4, PC, PCCA, PCCB, PCK1, PCSK9, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PIK3R1, PKLR, PLA2G6, PLCG2, PLIN1, PLP1, PMM2, PNPO, PNPT1, POLG, POLR3A, POLR3B, POR, PPARG, PPOX, PPT1, PRF1, PRICKLE1, PRKAG2, PRKCD, PRODH, PSAP, PSEN1, PTF1A, PTS, PYCR2, PYGL, PYGM, QDPR, RAB27A, RAI1, RARS1, RARS2, RBCK1, RFT1, RFX6, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPIA, RRM2B, SAMHD1, SCARB2, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SERAC1, SERPINI1, SGSH, SLC16A1, SLC16A2, SLC17A5, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A20, SLC25A4, SLC2A1, SLC2A2, SLC35A1, SLC35A2, SLC35C1, SLC3A1, SLC40A1, SLC4A1, SLC6A8, SLC6A9, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNTA1, SOX10, SPART, SPG11, SPG7, SPTA1, SPTB, SRD5A3, SSR4, STAT1, STT3A, STT3B, STX11, STXBP2, SUCLA2, SUCLG1, SUGCT, SUMF1, SUOX, SURF1, SYNE1, TACO1, TAT, TAZ, TBC1D24, TCF4, TCN2, TFR2, TGFB1, TINF2, TK2, TMEM165, TMEM70, TPK1, TPP1, TREM2, TREX1, TRPV4, TSFM, TTC19, TUBB4A, TUFM, TUSC3, TWNK, TYMP, TYROBP, UCP2, UGT1A1, UMPS, UNC13D, UQCRQ, UROD, UROS, WDR45, WFS1, ZFP57, ZFYVE26		5427	6
CentoLCV	Genome wide analysis of structural variants and large copy number changes through sWGS		50118	5

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17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoMito® Comprehensive	AARS2, AASS, ABAT, ABCB6, ABCB7, ABCD1, ABCD3, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACSL4, AFG3L2, AGK, AGXT, AIFM1, AK2, ALAS2, ALDH18A1, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, AMACR, AMT, APTX, ATIC, ATP5F1A, ATP5F1E, ATP7B, ATPAF2, ATXN2, AUH, BAX, BCKDHA, BCKDHB, BCKDK, BCL2, BCS1L, BOLA3, BRIP1, BTD, C12orf65, CA5A, CASP8, CAT, CAVIN1, CEL, CHCHD10, CISD2, CLPB, CLPP, COA5, COA6, COA8, COASY, COMT, COQ2, COQ4, COQ6, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX6A1, COX6B1, COX7B, CPOX, CPS1, CPT1A, CPT1C, CPT2, CRBN, CYB5A, CYB5R3, CYC1, CYCS, CYP11A1, CYP11B1, CYP11B2, CYP24A1, CYP27A1, CYP27B1, D2HGDH, DARS2, DBT, DGUOK, DHCR24, DHODH, DHTKD1, DIABLO, DLAT, DLD, DMGDH, DMPK, DNA2, DNAJC19, DNAJC3, DNM1L, EARS2, ECHS1, ELAC2, EPHX2, ETFA, ETFB, ETFDH, ETHE1, FAH, FARS2, FASTKD2, FBXL4, FECH, FH, FKBP10, FOXRED1, FTH1, FXN, GARS1, GATM, GCDH, GCSH, GDAP1, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLUD1, GLYCTK, GPI, GPT2, GPX1, GRHPR, GSR, GTPBP3, HADH, HADHA, HADHB, HAMP, HARS2, HAX1, HCCS, HIBCH, HINT1, HK1, HLCS, HMBS, HMGCL, HMGCS2, HOGA1, HSD17B10, HSD17B4, HSD3B2, HSPA9, HSPD1, HTRA2, IARS2, IBA57, IDH2, IDH3B, ISCA2, ISCU, IVD, KARS1, KIF1B, KRT5, L2HGDH, LARS2, LIAS, LIPT1, LONP1, LRPPRC, LYRM7, MAOA, MARS2, MCCC1, MCCC2, MCEE, MFN2, MGME1, MICU1, MIP, MLH1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS22, MSRB3, MTFMT, MTO1, MTPAP, MTRR, MUTYH, NADK2, NAGS, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NNT, NTHL1, NUBPL, OAT, OGDH, OGG1, OPA1, OPA3, OTC, OXCT1, P4HB, PAM16, PANK2, PARK7, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PDX1, PET100, PEX11B, PHYH, PINK1, PKLR, PNPLA8, PNPO, PNPT1, POLG, POLG2, PPOX, PRODH, PTRH2, PTS, PUS1, PYCR1, PYCR2, QDPR, RARS1, RARS2, RDH11, RMND1, RNASEH1, RNASEL, RPIA, RPL35A, RPS14, RRM2B, SARS2, SBDS, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SECISBP2, SERAC1, SFXN4, SLC16A1, SLC19A3, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC9A6, SNAP29, SOD1, SOD2, SPG7, SPR, SPTLC2, STAR, SUCLA2, SUCLG1, SUGCT, SUOX, SURF1, TACO1, TARS2, TCIRG1, TFR2, TIMM8A, TK2, TMEM126A, TMEM70, TMLHE, TPI1, TPK1, TRMU, TRNT1, TSFM, TTC19, TUBB3, TUFM, TWNK, TYMP, UNG, UQCC2, UQCRB, UQCRC2, UQCRQ, VARS2, WDR81, WFS1, XPNPEP3, YARS2		5447	6

1: Hotspot Testing
2: Carrier Testing (point mutation)
3: Carrier Testing (del/dup)
4: Single Gene Sequencing (Sanger)
5: NGS Panel
6: NGS Panel + CNV
7: NGS Panel Genomic
8: Deletion/Duplication with qPCR/MLPA
9: Sequencing + Deletion/Duplication
10: Repeat Expansions
11: Somatic Mutation Analysis
12: Biochemical Enzyme Analysis
13: Biomarker Analysis
14: Biochemical Enzyme Panel
15: Biochemical Genetics Panel Plus
16: Single Gene Sequencing (NGS)
17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoMito® Comprehensive	AARS2, AASS, ABAT, ABCB6, ABCB7, ABCD1, ABCD3, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACSL4, AFG3L2, AGK, AGXT, AIFM1, AK2, ALAS2, ALDH18A1, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, AMACR, AMT, APTX, ATIC, ATP5F1A, ATP5F1E, ATP7B, ATPAF2, ATXN2, AUH, BAX, BCKDHA, BCKDHB, BCKDK, BCL2, BCS1L, BOLA3, BRIP1, BTD, C12orf65, CA5A, CASP8, CAT, CAVIN1, CEL, CHCHD10, CISD2, CLPB, CLPP, COA5, COA6, COA8, COASY, COMT, COQ2, COQ4, COQ6, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX6A1, COX6B1, COX7B, CPOX, CPS1, CPT1A, CPT1C, CPT2, CRBN, CYB5A, CYB5R3, CYC1, CYCS, CYP11A1, CYP11B1, CYP11B2, CYP24A1, CYP27A1, CYP27B1, D2HGDH, DARS2, DBT, DGUOK, DHCR24, DHODH, DHTKD1, DIABLO, DLAT, DLD, DMGDH, DMPK, DNA2, DNAJC19, DNAJC3, DNM1L, EARS2, ECHS1, ELAC2, EPHX2, ETFA, ETFB, ETFDH, ETHE1, FAH, FARS2, FASTKD2, FBXL4, FECH, FH, FKBP10, FOXRED1, FTH1, FXN, GARS1, GATM, GCDH, GCSH, GDAP1, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLUD1, GLYCTK, GPI, GPT2, GPX1, GRHPR, GSR, GTPBP3, HADH, HADHA, HADHB, HAMP, HARS2, HAX1, HCCS, HIBCH, HINT1, HK1, HLCS, HMBS, HMGCL, HMGCS2, HOGA1, HSD17B10, HSD17B4, HSD3B2, HSPA9, HSPD1, HTRA2, IARS2, IBA57, IDH2, IDH3B, ISCA2, ISCU, IVD, KARS1, KIF1B, KRT5, L2HGDH, LARS2, LIAS, LIPT1, LONP1, LRPPRC, LYRM7, MAOA, MARS2, MCCC1, MCCC2, MCEE, MFN2, MGME1, MICU1, MIP, MLH1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS22, MSRB3, MTFMT, MTO1, MTPAP, MTRR, MUTYH, NADK2, NAGS, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NNT, NTHL1, NUBPL, OAT, OGDH, OGG1, OPA1, OPA3, OTC, OXCT1, P4HB, PAM16, PANK2, PARK7, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PDX1, PET100, PEX11B, PHYH, PINK1, PKLR, PNPLA8, PNPO, PNPT1, POLG, POLG2, PPOX, PRODH, PTRH2, PTS, PUS1, PYCR1, PYCR2, QDPR, RARS1, RARS2, RDH11, RMND1, RNASEH1, RNASEL, RPIA, RPL35A, RPS14, RRM2B, SARS2, SBDS, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SECISBP2, SERAC1, SFXN4, SLC16A1, SLC19A3, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC9A6, SNAP29, SOD1, SOD2, SPG7, SPR, SPTLC2, STAR, SUCLA2, SUCLG1, SUGCT, SUOX, SURF1, TACO1, TARS2, TCIRG1, TFR2, TIMM8A, TK2, TMEM126A, TMEM70, TMLHE, TPI1, TPK1, TRMU, TRNT1, TSFM, TTC19, TUBB3, TUFM, TWNK, TYMP, UNG, UQCC2, UQCRB, UQCRC2, UQCRQ, VARS2, WDR81, WFS1, XPNPEP3, YARS2		5447	6
CentoMito® Genome	MT-ND1, MT-ND2, MT-CO1, MT-CO2, MT-ATP8, MT-ATP6, MT-CO3, MT-ND3, MT-ND4L, MT-ND4, MT-ND5, MT-ND6, MT-CYB, MT-TF, MT-RNR1, MT-TV, MT-RNR2, MT-TL1, MT-TI, MT-TQ, MT-TM, MT-TW, MT-TA, MT-TN, MT-TC, MT-TY, MT-TS1, MT-TD, MT-TK, MT-TG, MT-TR, MT-TH, MT-TS2, MT-TL2, MT-TE, MT-TT, MT-TP		50153	6
CentoMito® Genome	MT-ND1, MT-ND2, MT-CO1, MT-CO2, MT-ATP8, MT-ATP6, MT-CO3, MT-ND3, MT-ND4L, MT-ND4, MT-ND5, MT-ND6, MT-CYB, MT-TF, MT-RNR1, MT-TV, MT-RNR2, MT-TL1, MT-TI, MT-TQ, MT-TM, MT-TW, MT-TA, MT-TN, MT-TC, MT-TY, MT-TS1, MT-TD, MT-TK, MT-TG, MT-TR, MT-TH, MT-TS2, MT-TL2, MT-TE, MT-TT, MT-TP		50153	6

1: Hotspot Testing
2: Carrier Testing (point mutation)
3: Carrier Testing (del/dup)
4: Single Gene Sequencing (Sanger)
5: NGS Panel
6: NGS Panel + CNV
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8: Deletion/Duplication with qPCR/MLPA
9: Sequencing + Deletion/Duplication
10: Repeat Expansions
11: Somatic Mutation Analysis
12: Biochemical Enzyme Analysis
13: Biomarker Analysis
14: Biochemical Enzyme Panel
15: Biochemical Genetics Panel Plus
16: Single Gene Sequencing (NGS)
17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoNeuro™	A2M, AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCB7, ABCC6, ABCC8, ABCD1, ABCD3, ABCD4, ABHD12, ABHD5, ACACA, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACKR1, ACO2, ACOX1, ACSF3, ACSL4, ACTA1, ACTA2, ACTB, ACTG1, ACTN4, ACVRL1, ACY1, ADA, ADAM10, ADAMTSL2, ADAR, ADCY5, ADGRG1, ADGRV1, ADK, ADNP, ADSL, AFF2, AFG3L2, AGA, AGK, AGL, AGPS, AGRN, AGXT, AHCY, AHI1, AIFM1, AIMP1, AKAP9, AKT1, AKT3, ALAD, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX5AP, ALS2, ALX1, ALX3, ALX4, AMACR, AMN, AMPD1, AMPD2, AMT, ANG, ANK2, ANK3, ANKRD11, ANO10, ANO3, ANO5, ANOS1, ANTXR2, AP1S1, AP1S2, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APOE, APP, APTX, AR, ARFGEF2, ARG1, ARHGAP31, ARHGEF10, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARL13B, ARL6, ARL6IP1, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASCL1, ASL, ASNS, ASPA, ASPM, ASS1, ASXL1, ASXL3, ATCAY, ATIC, ATL1, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2A1, ATP2A2, ATP2B3, ATP5F1E, ATP6AP2, ATP6V0A2, ATP7A, ATP7B, ATP8A2, ATPAF2, ATR, ATRX, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, AUH, AUTS2, B3GALNT2, B3GLCT, B4GALNT1, B4GALT1, B4GAT1, B9D1, B9D2, BAG3, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCOR, BCS1L, BDNF, BEAN1, BEST1, BICD2, BIN1, BLOC1S3, BLOC1S6, BOLA3, BRAF, BRAT1, BRCA2, BRWD3, BSCL2, BSND, BTD, C12orf57, C12orf65, C19orf12, C8orf37, C9ORF72, CA2, CA5A, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1F, CACNA1H, CACNA1S, CACNB2, CACNB4, CAMTA1, CAPN3, CARD11, CASK, CASR, CAV1, CAV3, CBL, CBS, CC2D1A, CC2D2A, CCDC28B, CCDC40, CCDC78, CCDC88C, CCM2, CCT5, CD320, CD36, CD59, CD96, CDH15, CDK5RAP2, CDKL5, CDON, CEL, CENPF, CENPJ, CEP135, CEP152, CEP164, CEP290, CEP41, CEP63, CERS1, CFL2, CHAT, CHCHD10, CHD2, CHD7, CHD8, CHKB, CHMP1A, CHMP2B, CHRM3, CHRNA1, CHRNA2, CHRNA4, CHRNB1, CHRNB2, CHRND, CHRNE, CHRNG, CHST14, CHSY1, CIB2, CILK1, CISD2, CISH, CLCN1, CLCN2, CLCN4, CLCNKA, CLCNKB, CLDN16, CLDN19, CLIC2, CLN3, CLN5, CLN6, CLN8, CLPP, CNBP, CNGB3, CNNM2, CNTN1, CNTNAP2, COA5, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL12A1, COL2A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COQ2, COQ4, COQ8A, COQ9, COX10, COX15, COX20, COX6A1, COX6B1, CP, CPA6, CPLANE1, CPS1, CPT1A, CPT2, CR1, CRADD, CRBN, CREBBP, CRIPT, CRPPA, CRYAB, CSF1R, CSF2RB, CSPP1, CSRP3, CST3, CSTB, CTC1, CTDP1, CTNNA3, CTNNB1, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, CUL3, CUL4B, CUL7, CWF19L1, CYB5R3, CYP11B1, CYP11B2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS2, DBT, DCAF17, DCTN1, DCX, DDC, DDHD1, DDHD2, DDOST, DDX3X, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHH, DHTKD1, DIAPH3, DKC1, DLAT, DLD, DLG3, DMD, DMPK, DNA2, DNAH9, DNAJB2, DNAJB6, DNAJC19, DNAJC5, DNAJC6, DNM1, DNM1L, DNM2, DNMT1, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DRD3, DSC3, DST, DTNBP1, DUSP6, DYM, DYNC1H1, DYNC2H1, DYRK1A, DYSF, EARS2, EBP, ECEL1, ECHS1, EDN3, EDNRB, EEF1A2, EFHC1, EFTUD2, EGF, EGR2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4E, EIF4G1, ELOVL4, ELOVL5, ELP1, EMD, EMX2, ENO3, ENPP1, ENTPD1, EP300, EPB41L1, EPM2A, ERBB4, ERCC1, ERCC2, ERCC5, ERCC6, ERLIN1, ERLIN2, ESCO2, ESRRB, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, EXOSC8, F2, F5, FA2H, FADD, FAH, FAM126A, FANCB, FARS2, FASTKD2, FBLN5, FBN1, FBN2, FBXL4, FBXO38, FBXO7, FCGR2B, FEZF1, FGA, FGD1, FGD4, FGF10, FGF12, FGF14, FGF17, FGF8, FGFR1, FGFR2, FGFR3, FH, FHL1, FIG4, FKBP10, FKRP, FKTN, FLNA, FLNC, FLRT3, FLVCR1, FLVCR2, FMR1, FOLR1, FOXC1, FOXG1, FOXL2, FOXP1, FOXP2, FOXP3, FOXRED1, FREM1, FRMD7, FRRS1L, FSHB, FTL, FTO, FTSJ1, FUCA1, FUS, FXN, FXYD2, G6PD, GAA, GABRA1, GABRB3, GABRD, GABRG2, GAD1, GALC, GALNS, GALT, GAMT, GAN, GARS1, GATM, GBA, GBA2, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GDI1, GDNF, GFAP, GFER, GFM1, GFM2, GFPT1, GIGYF2, GJA1, GJB1, GJB3, GJC2, GK, GLA, GLB1, GLDC, GLE1, GLI2, GLI3, GLRA1, GLRB, GLUD1, GLUL, GM2A, GMPPA, GMPPB, GNAL, GNAO1, GNAS, GNB4, GNE, GNPAT, GNPTAB, GNPTG, GNRH1, GNRHR, GNS, GOSR2, GP1BA, GPC3, GPHN, GPR143, GPT2, GPX1, GRIA3, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIP1, GRM1, GRN, GSN, GSS, GTPBP3, GUF1, GUSB, GYG1, GYS1, HADHA, HADHB, HAMP, HBB, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HEPACAM, HERC2, HESX1, HEXA, HEXB, HFE, HGSNAT, HIBCH, HINT1, HK1, HLCS, HMGCL, HMGCS2, HNRNPA1, HNRNPDL, HNRNPU, HOXA1, HOXD10, HPCA, HPD, HPRT1, HPS1, HPS3, HPS4, HPS5, HPS6, HPSE2, HRAS, HS6ST1, HSD17B10, HSD17B4, HSPB1, HSPB3, HSPB8, HSPD1, HSPG2, HTRA1, HTRA2, HUWE1, HYAL1, HYDIN, IARS2,		5424	6

1: Hotspot Testing
2: Carrier Testing (point mutation)
3: Carrier Testing (del/dup)
4: Single Gene Sequencing (Sanger)
5: NGS Panel
6: NGS Panel + CNV
7: NGS Panel Genomic
8: Deletion/Duplication with qPCR/MLPA
9: Sequencing + Deletion/Duplication
10: Repeat Expansions
11: Somatic Mutation Analysis
12: Biochemical Enzyme Analysis
13: Biomarker Analysis
14: Biochemical Enzyme Panel
15: Biochemical Genetics Panel Plus
16: Single Gene Sequencing (NGS)
17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
Dementia panel	ALS2, ANG, APOE, APP, ARSA, ATL1, ATP7B, ATXN2, BSCL2, C9orf72, CHCHD10, CHMP2B, CP, CSF1R, DCTN1, ERBB4, FIG4, FTL, FUS, GRN, HEXA, HNRNPA1, HSPD1, ITM2B, KIF5A, MAPT, MATR3, NEFH, NOTCH3, NPC1, OPTN, PANK2, PFN1, PRNP, PRPH, PSEN1, PSEN2, REEP1, SETX, SIGMAR1, SLC52A3, SNCA, SOD1, SORL1, SPAST, SPG11, SQSTM1, TARDBP, TBK1, TREM2, TUBA4A, TYROBP, UBE3A, UBQLN2, VAPB, VCP, WASHC5		5448	6
Dementia panel	ALS2, ANG, APOE, APP, ARSA, ATL1, ATP7B, ATXN2, BSCL2, C9orf72, CHCHD10, CHMP2B, CP, CSF1R, DCTN1, ERBB4, FIG4, FTL, FUS, GRN, HEXA, HNRNPA1, HSPD1, ITM2B, KIF5A, MAPT, MATR3, NEFH, NOTCH3, NPC1, OPTN, PANK2, PFN1, PRNP, PRPH, PSEN1, PSEN2, REEP1, SETX, SIGMAR1, SLC52A3, SNCA, SOD1, SORL1, SPAST, SPG11, SQSTM1, TARDBP, TBK1, TREM2, TUBA4A, TYROBP, UBE3A, UBQLN2, VAPB, VCP, WASHC5		5448	6
Dystonia panel	ADAR, ADCY5, ANO3, ARSA, ATM, ATP13A2, ATP1A3, ATP7B, BCAP31, BTD, C19orf12, CACNA1B, CBS, CLN3, CLN5, CLN6, CLN8, COASY, COL6A3, CP, CTSD, CTSF, D2HGDH, DCAF17, DDC, DNAJC5, ECHS1, FA2H, FTL, FUCA1, GALC, GALT, GAMT, GATM, GCDH, GCH1, GLB1, GM2A, GNAL, GRN, HPCA, HPRT1, KCNMA1, KCTD17, KCTD7, MCEE, MFSD8, MMAA, MMAB, MMADHC, MMUT, NPC1, NPC2, PAH, PANK2, PLA2G6, PLP1, PNKD, POLG, PPT1, PRKN, PRKRA, PRRT2, RELN, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SERAC1, SGCE, SLC19A3, SLC20A2, SLC2A1, SLC30A10, SLC6A3, SLC6A8, SPR, SUCLA2, TH, THAP1, TIMM8A, TOR1A, TPP1, TREX1, TUBB4A, VPS13A, WDR45, XK		5450	6
Dystonia panel	ADAR, ADCY5, ANO3, ARSA, ATM, ATP13A2, ATP1A3, ATP7B, BCAP31, BTD, C19orf12, CACNA1B, CBS, CLN3, CLN5, CLN6, CLN8, COASY, COL6A3, CP, CTSD, CTSF, D2HGDH, DCAF17, DDC, DNAJC5, ECHS1, FA2H, FTL, FUCA1, GALC, GALT, GAMT, GATM, GCDH, GCH1, GLB1, GM2A, GNAL, GRN, HPCA, HPRT1, KCNMA1, KCTD17, KCTD7, MCEE, MFSD8, MMAA, MMAB, MMADHC, MMUT, NPC1, NPC2, PAH, PANK2, PLA2G6, PLP1, PNKD, POLG, PPT1, PRKN, PRKRA, PRRT2, RELN, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SERAC1, SGCE, SLC19A3, SLC20A2, SLC2A1, SLC30A10, SLC6A3, SLC6A8, SPR, SUCLA2, TH, THAP1, TIMM8A, TOR1A, TPP1, TREX1, TUBB4A, VPS13A, WDR45, XK		5450	6
Dystonia panel	ADAR, ADCY5, ANO3, ARSA, ATM, ATP13A2, ATP1A3, ATP7B, BCAP31, BTD, C19orf12, CACNA1B, CBS, CLN3, CLN5, CLN6, CLN8, COASY, COL6A3, CP, CTSD, CTSF, D2HGDH, DCAF17, DDC, DNAJC5, ECHS1, FA2H, FTL, FUCA1, GALC, GALT, GAMT, GATM, GCDH, GCH1, GLB1, GM2A, GNAL, GRN, HPCA, HPRT1, KCNMA1, KCTD17, KCTD7, MCEE, MFSD8, MMAA, MMAB, MMADHC, MMUT, NPC1, NPC2, PAH, PANK2, PLA2G6, PLP1, PNKD, POLG, PPT1, PRKN, PRKRA, PRRT2, RELN, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SERAC1, SGCE, SLC19A3, SLC20A2, SLC2A1, SLC30A10, SLC6A3, SLC6A8, SPR, SUCLA2, TH, THAP1, TIMM8A, TOR1A, TPP1, TREX1, TUBB4A, VPS13A, WDR45, XK		5450	6

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13: Biomarker Analysis
14: Biochemical Enzyme Panel
15: Biochemical Genetics Panel Plus
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17: Single Gene Sequencing + CNV (NGS)

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Epilepsy panel	AARS1, AARS2, ABCC8, ABCD1, ABCD3, ACAD9, ACADM, ACADS, ACADVL, ACOX1, ACY1, ADA, ADAMTSL2, ADAR, ADGRG1, ADSL, AFG3L2, AGA, AGK, AGPS, AIFM1, AIMP1, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMT, ANTXR2, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, APP, APTX, ARG1, ARHGEF9, ARSA, ARSB, ARV1, ARX, ASAH1, ASL, ASPA, ASS1, ATM, ATP13A2, ATP1A2, ATP6V0A2, ATP7A, ATP7B, ATPAF2, AUH, B3GALNT2, B3GLCT, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCS1L, BEST1, BOLA3, BRAT1, BTD, C12orf65, C19orf12, CA5A, CACNA1A, CACNA1H, CACNB4, CASK, CAV1, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLCN4, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPP, CNNM2, CNTNAP2, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COL4A1, COL4A2, COQ2, COQ8A, COQ9, COX10, COX15, COX20, COX6B1, CP, CPA6, CPS1, CPT1A, CPT2, CSF1R, CSTB, CTC1, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS2, DBT, DCAF17, DDOST, DEPDC5, DGUOK, DHCR7, DHDDS, DKC1, DLAT, DLD, DNAJC5, DNM1, DNM1L, DOCK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DYM, DYRK1A, EARS2, ECHS1, EEF1A2, EFHC1, EGF, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPM2A, ERCC6, ETFA, ETFB, ETFDH, ETHE1, F2, F5, FA2H, FAH, FAM126A, FARS2, FASTKD2, FBN1, FBXL4, FGF12, FH, FHL1, FOLR1, FOXG1, FOXRED1, FRRS1L, FTL, FUCA1, FXYD2, GAA, GABRA1, GABRB3, GABRD, GABRG2, GALC, GALNS, GALT, GAMT, GAN, GBA, GBE1, GCDH, GCSH, GFAP, GFER, GFM1, GFM2, GFPT1, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLUD1, GLUL, GM2A, GMPPA, GNAO1, GNE, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GPC3, GRIN1, GRIN2A, GRIN2B, GRN, GTPBP3, GUF1, GUSB, HADHA, HADHB, HCFC1, HCN1, HEPACAM, HEXA, HEXB, HGSNAT, HIBCH, HLCS, HMGCL, HMGCS2, HNRNPU, HRAS, HSD17B4, HSPD1, HTRA1, HYAL1, IARS2, IBA57, IDS, IDUA, IFIH1, IQSEC2, ISCA2, ITPA, IVD, JAG1, JAM3, KCNA1, KCNA2, KCNB1, KCNC1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIF5A, L2HGDH, LAMA2, LAMB1, LAMP2, LARGE1, LDB3, LGI1, LIAS, LIPA, LIPT1, LMNB1, LRPPRC, LYRM7, LYST, MAGT1, MAN1B1, MAN2B1, MANBA, MARS2, MBD5, MCCC1, MCCC2, MCOLN1, MECP2, MED17, MEF2C, MFN2, MFSD8, MGAT2, MGME1, MLC1, MLPH, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MRPL44, MRPS22, MTFMT, MTHFR, MTOR, MTR, MYO5A, MYOT, NAGA, NAGLU, NAGS, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF4, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NECAP1, NEDD4L, NEU1, NFU1, NGLY1, NHLRC1, NOTCH3, NPC1, NPC2, NRXN1, NUBPL, OAT, OCLN, OCRL, OPA1, OPA3, OTC, PAH, PANK2, PC, PCCA, PCCB, PCDH19, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGK1, PGM1, PHYH, PIGA, PIGO, PIGV, PLA2G6, PLCB1, PLCG2, PLP1, PMM2, PNKP, PNPO, PNPT1, POLG, POLG2, POLR3A, POLR3B, PPT1, PRICKLE1, PRODH, PRRT2, PSAP, PSEN1, PTS, PURA, PYCR2, QARS1, QDPR, RAB27A, RAI1, RARS1, RARS2, RBFOX1, RELN, RFT1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, ROGDI, RPIA, RRM2B, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEC23B, SERAC1, SERPINI1, SGCE, SGSH, SIK1, SLC12A3, SLC12A5, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A20, SLC25A22, SLC25A3, SLC25A4, SLC2A1, SLC35A1, SLC35A2, SLC35C1, SLC6A1, SLC6A8, SLC7A7, SLC9A6, SMC1A, SMPD1, SNTA1, SOX10, SPART, SPG11, SPG7, SPTAN1, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STAT1, STT3A, STT3B, STX1B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYN1, SYNE1, SYNGAP1, SYNJ1, SZT2, TACO1, TBC1D24, TBCE, TCF4, TGFB1, TIMM8A, TINF2, TK2, TMEM126A, TMEM165, TMEM70, TPK1, TPP1, TREM2, TREX1, TRPM6, TRPV4, TSC1, TSC2, TSFM, TTC19, TUBB4A, TUFM, TUSC3, TWNK, TYMP, TYROBP, UBE3A, UMPS, UQCRQ, WDR45, WFS1, WWOX, ZEB2, ZFYVE26		5451	6

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17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
Intellectual disability panel	ABAT, ABCD1, ACE, ACSL4, ACTB, ACTG1, ACTN4, ADA, ADAR, ADCY5, ADGRG1, ADK, ADNP, ADSL, AFF2, AHI1, AIMP1, AKAP9, AKT3, ALDH18A1, ALDH5A1, ALDH7A1, ALG11, ALG13, ALX4, AMPD1, AMPD2, AMT, ANK2, ANK3, ANKRD11, AP1S1, AP1S2, AP3B1, AP4B1, AP4M1, ARFGEF2, ARHGEF10, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARL13B, ARX, ASAH1, ASCL1, ASPM, ASXL1, ASXL3, ATP13A2, ATP1A3, ATP6AP2, ATP6V0A2, ATP7A, ATP8A2, ATR, ATRX, AUTS2, B3GALNT2, B4GAT1, B9D1, B9D2, BBS4, BCAP31, BCKDK, BCOR, BCS1L, BDNF, BLOC1S3, BLOC1S6, BRAF, BRCA2, BRWD3, C12orf57, C12orf65, C19orf12, CA2, CA8, CACNA1A, CACNA1C, CACNA1D, CACNA1F, CACNA1H, CACNB2, CAMTA1, CARD11, CASK, CBS, CC2D1A, CC2D2A, CCDC40, CCDC88C, CDH15, CDK5RAP2, CDKL5, CDON, CENPF, CENPJ, CEP135, CEP152, CEP290, CEP41, CEP63, CHD2, CHD7, CHD8, CHMP1A, CHRM3, CIB2, CLCN4, CLIC2, CLN8, CNGB3, CNTNAP2, COASY, COL4A1, CPLANE1, CPT2, CRADD, CRBN, CREBBP, CRIPT, CRPPA, CSPP1, CTC1, CTNNA3, CTNNB1, CUL3, CUL4B, CUL7, CYB5R3, CYP11B1, CYP27A1, DARS2, DCAF17, DCX, DDC, DDX3X, DHCR7, DKC1, DLG3, DNM1, DOCK8, DPYD, DRD3, DST, DTNBP1, DYM, DYNC1H1, DYRK1A, EDN3, EDNRB, EEF1A2, EFTUD2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4E, ELOVL4, EMX2, EP300, EPB41L1, ESRRB, ETFB, EXOSC3, FAM126A, FANCB, FBN1, FGA, FGD1, FGFR2, FGFR3, FH, FKRP, FKTN, FLNA, FLVCR1, FMR1, FOLR1, FOXC1, FOXG1, FOXL2, FOXP1, FOXP2, FTL, FTO, FTSJ1, G6PD, GABRB3, GAD1, GAMT, GATM, GCK, GDI1, GDNF, GFAP, GIGYF2, GJC2, GK, GLI2, GLRB, GMPPA, GMPPB, GNAO1, GNAS, GPC3, GPHN, GPT2, GPX1, GRIA3, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIP1, HBB, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HEPACAM, HERC2, HNRNPU, HOXA1, HPRT1, HPS1, HPS4, HPS5, HPS6, HRAS, HSD17B10, HSPD1, HUWE1, HYDIN, IDS, IER3IP1, IFIH1, IGBP1, IGF1, IGF1R, IL1RAPL1, INPP5E, INVS, IQSEC2, IRX5, ITGA7, ITGB3, ITPR1, JAG1, KANK1, KAT6A, KAT6B, KATNB1, KCNB1, KCNC1, KCNC3, KCND3, KCNJ10, KCNK9, KCNMA1, KCNQ2, KCNQ3, KDM5C, KDM6A, KIF11, KIF1A, KIF5C, KIF7, KIRREL3, KMT2A, KMT2C, KMT2D, KNL1, KRAS, L1CAM, LAMA1, LAMA2, LAMB1, LAMC3, LAMP2, LARGE1, LEP, LINS1, LMX1B, LRBA, LRP2, LZTR1, MAGEL2, MAGT1, MAN1B1, MAOA, MBD5, MBTPS2, MCCC2, MCM4, MCPH1, MECP2, MED12, MED13L, MED17, MED23, MEF2C, MEGF10, MEIS2, MET, MFRP, MFSD2A, MGAT2, MIB1, MID1, MITF, MKKS, MKS1, MLC1, MPDZ, MSMO1, MTHFR, MTM1, MTOR, MTR, MYCN, MYO5A, NAA10, NAGA, NALCN, NDE1, NDP, NDST1, NDUFA1, NECTIN1, NEXMIF, NF1, NFIX, NHEJ1, NHS, NIPA1, NIPBL, NLGN3, NLGN4X, NOTCH2, NPHP1, NPHP3, NR2F1, NR3C2, NRXN1, NSD1, NSDHL, NSUN2, NTRK1, NXF5, OCLN, OCRL, OFD1, OPHN1, ORC1, OTC, PAFAH1B1, PAH, PAK3, PANK2, PAX3, PAX6, PCDH15, PCDH19, PCNT, PDE6D, PDHA1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PHC1, PHF6, PHF8, PIGA, PIGL, PIGN, PIGO, PIGV, PIK3R2, PITX2, PLA2G6, PLCB1, PLK4, PLN, PLP1, PNKP, POGZ, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PORCN, POT1, PPOX, PPT1, PQBP1, PRICKLE1, PRKN, PRODH, PRPS1, PRSS12, PTCH1, PTEN, PTPN11, PTPRC, PTS, PURA, PYCR1, PYCR2, QARS1, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RAD21, RAI1, RARS2, RBBP8, RBFOX1, RBM10, RELN, RET, RIMS1, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, ROR2, RPGRIP1L, RPL10, RPS6KA3, RXYLT1, SALL1, SAMHD1, SASS6, SATB2, SBF1, SCN1A, SCN2A, SCN4A, SCN8A, SCN9A, SCO2, SDCCAG8, SDHA, SEPSECS, SETBP1, SETD2, SGCA, SHANK2, SHH, SHROOM4, SIL1, SIX3, SLC12A5, SLC16A2, SLC25A12, SLC25A15, SLC25A19, SLC27A4, SLC2A1, SLC35A2, SLC35A3, SLC4A4, SLC6A1, SLC6A3, SLC6A8, SLC7A7, SLC9A6, SLC9A9, SLCO1B3, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMPD1, SMS, SNAI2, SNAP29, SNIP1, SOBP, SOD1, SOX10, SOX2, SOX3, SPAST, SRD5A3, ST3GAL3, STAMBP, STIL, STRA6, STXBP1, SYN1, SYNE1, SYNGAP1, SYNJ1, SYP, TAF2, TAF6, TBC1D20, TBC1D24, TBCE, TBL1XR1, TBX1, TCF4, TCTN1, TCTN2, TCTN3, TECR, TECTA, TFAP2A, TGIF1, THRA, TIMM8A, TINF2, TMCO1, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMLHE, TPO, TRAPPC9, TREX1, TRMT10A, TRPC6, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TSPAN7, TTC21B, TTI2, TTN, TUBA1A, TUBA8, TUBB2B, TUBB3, TUBB4A, TUBGCP4, TUBGCP6, TUSC3, TYR, UBE2A, UBE3A, UBR1, UPF3B, USH2A, USP9X, VDR, VLDLR, VPS13B, VPS53, VRK1, WAC, WDR45, WDR62, WDR81, WNT1, WNT5A, WWOX, YWHAE, ZBTB18, ZDHHC9, ZEB2, ZFYVE26, ZIC2, ZIC3, ZNF335, ZNF41, ZNF423, ZNF711, ZNF81		5425	6

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Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
Neuromuscular panel	AARS1, ABHD5, ACAD9, ACADM, ACADVL, ACTA1, AGL, AGRN, AHCY, AIFM1, ALDOA, ALG14, ALG2, AMPD1, ANO5, ARHGEF10, ARHGEF9, ASAH1, ATL1, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BSCL2, CACNA1S, CAPN3, CASK, CAV1, CAV3, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCN1, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, COQ2, COX6A1, CPT2, CRPPA, CRYAB, CSRP3, CTDP1, DAG1, DCTN1, DES, DGUOK, DHCR24, DHTKD1, DMD, DMPK, DNA2, DNAJB2, DNAJB6, DNM2, DNMT1, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYNC1H1, DYSF, ECEL1, EGR2, ELP1, EMD, ENO3, ERCC5, ERCC6, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FBLN5, FBN2, FBXO38, FGD4, FHL1, FIG4, FKBP10, FKRP, FKTN, FLNC, GAA, GAN, GARS1, GBA, GBE1, GDAP1, GFPT1, GJB1, GLE1, GLRA1, GLRB, GMPPB, GNB4, GNE, GPHN, GYG1, GYS1, HADHA, HADHB, HINT1, HK1, HNRNPDL, HOXD10, HRAS, HSPB1, HSPB3, HSPB8, HSPG2, IGHMBP2, INF2, ISCU, ITGA7, KARS1, KAT6B, KBTBD13, KCNA1, KCNE3, KIF1A, KIF1B, KIF5A, KLHL40, KLHL41, LAMA2, LAMB2, LAMP2, LARGE1, LDB3, LDHA, LIMS2, LITAF, LMNA, LMOD3, LPIN1, LRSAM1, MAGEL2, MAMLD1, MARS1, MATR3, MED25, MEGF10, MFN2, MICU1, MPV17, MPZ, MTM1, MTMR14, MTMR2, MUSK, MYBPC1, MYBPC3, MYH2, MYH3, MYH7, MYH8, MYO18B, MYOT, MYPN, NALCN, NDRG1, NEB, NTRK1, OPA1, OPA3, PDK3, PFKM, PGAM2, PGK1, PGM1, PHKA1, PIEZO2, PLEC, PLEKHG5, PLOD2, PMM2, PMP22, PNPLA2, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, PRKAG2, PRPS1, PRX, PYGM, QARS1, RAB7A, RAPSN, RBCK1, REEP1, RETREG1, RRM2B, RXYLT1, RYR1, SBF1, SBF2, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCE, SGCG, SH3TC2, SIL1, SLC12A6, SLC16A1, SLC22A5, SLC25A1, SLC25A20, SLC25A46, SLC5A7, SLC6A5, SMCHD1, SMN1, SMN2, SPEG, SPG11, SPTLC1, SPTLC2, SUCLA2, SYNE1, SYNE2, TAZ, TCAP, TFG, TGFB3, TK2, TMEM43, TNNI2, TNNT1, TNNT3, TNPO3, TOR1A, TPM2, TPM3, TRAPPC11, TRIM2, TRIM32, TRPV4, TSEN2, TSFM, TTN, TWNK, TYMP, UBA1, VAMP1, VAPB, VCP, VIPAS39, VRK1, WNK1, XK, YARS1		5426	6

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Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
Neuromuscular panel	AARS1, ABHD5, ACAD9, ACADM, ACADVL, ACTA1, AGL, AGRN, AHCY, AIFM1, ALDOA, ALG14, ALG2, AMPD1, ANO5, ARHGEF10, ARHGEF9, ASAH1, ATL1, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BSCL2, CACNA1S, CAPN3, CASK, CAV1, CAV3, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCN1, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, COQ2, COX6A1, CPT2, CRPPA, CRYAB, CSRP3, CTDP1, DAG1, DCTN1, DES, DGUOK, DHCR24, DHTKD1, DMD, DMPK, DNA2, DNAJB2, DNAJB6, DNM2, DNMT1, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYNC1H1, DYSF, ECEL1, EGR2, ELP1, EMD, ENO3, ERCC5, ERCC6, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FBLN5, FBN2, FBXO38, FGD4, FHL1, FIG4, FKBP10, FKRP, FKTN, FLNC, GAA, GAN, GARS1, GBA, GBE1, GDAP1, GFPT1, GJB1, GLE1, GLRA1, GLRB, GMPPB, GNB4, GNE, GPHN, GYG1, GYS1, HADHA, HADHB, HINT1, HK1, HNRNPDL, HOXD10, HRAS, HSPB1, HSPB3, HSPB8, HSPG2, IGHMBP2, INF2, ISCU, ITGA7, KARS1, KAT6B, KBTBD13, KCNA1, KCNE3, KIF1A, KIF1B, KIF5A, KLHL40, KLHL41, LAMA2, LAMB2, LAMP2, LARGE1, LDB3, LDHA, LIMS2, LITAF, LMNA, LMOD3, LPIN1, LRSAM1, MAGEL2, MAMLD1, MARS1, MATR3, MED25, MEGF10, MFN2, MICU1, MPV17, MPZ, MTM1, MTMR14, MTMR2, MUSK, MYBPC1, MYBPC3, MYH2, MYH3, MYH7, MYH8, MYO18B, MYOT, MYPN, NALCN, NDRG1, NEB, NTRK1, OPA1, OPA3, PDK3, PFKM, PGAM2, PGK1, PGM1, PHKA1, PIEZO2, PLEC, PLEKHG5, PLOD2, PMM2, PMP22, PNPLA2, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, PRKAG2, PRPS1, PRX, PYGM, QARS1, RAB7A, RAPSN, RBCK1, REEP1, RETREG1, RRM2B, RXYLT1, RYR1, SBF1, SBF2, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCE, SGCG, SH3TC2, SIL1, SLC12A6, SLC16A1, SLC22A5, SLC25A1, SLC25A20, SLC25A46, SLC5A7, SLC6A5, SMCHD1, SMN1, SMN2, SPEG, SPG11, SPTLC1, SPTLC2, SUCLA2, SYNE1, SYNE2, TAZ, TCAP, TFG, TGFB3, TK2, TMEM43, TNNI2, TNNT1, TNNT3, TNPO3, TOR1A, TPM2, TPM3, TRAPPC11, TRIM2, TRIM32, TRPV4, TSEN2, TSFM, TTN, TWNK, TYMP, UBA1, VAMP1, VAPB, VCP, VIPAS39, VRK1, WNK1, XK, YARS1		5426	6
Parkinson disease panel	ATP13A2, ATP1A3, ATP6AP2, ATP7B, C19orf12, DCTN1, DNAJC6, FBXO7, FTL, FUS, GBA, GCH1, GRN, HTRA2, LRRK2, MAPT, PANK2, PARK7, PINK1, PLA2G6, PRKN, PRKRA, RAB39B, SLC20A2, SLC30A10, SLC6A3, SNCA, SNCB, SPR, SYNJ1, TH, TMEM230, UCHL1, VPS13A, VPS35, WDR45		5455	6
Parkinson disease panel	ATP13A2, ATP1A3, ATP6AP2, ATP7B, C19orf12, DCTN1, DNAJC6, FBXO7, FTL, FUS, GBA, GCH1, GRN, HTRA2, LRRK2, MAPT, PANK2, PARK7, PINK1, PLA2G6, PRKN, PRKRA, RAB39B, SLC20A2, SLC30A10, SLC6A3, SNCA, SNCB, SPR, SYNJ1, TH, TMEM230, UCHL1, VPS13A, VPS35, WDR45		5455	6
Parkinson disease panel	ATP13A2, ATP1A3, ATP6AP2, ATP7B, C19orf12, DCTN1, DNAJC6, FBXO7, FTL, FUS, GBA, GCH1, GRN, HTRA2, LRRK2, MAPT, PANK2, PARK7, PINK1, PLA2G6, PRKN, PRKRA, RAB39B, SLC20A2, SLC30A10, SLC6A3, SNCA, SNCB, SPR, SYNJ1, TH, TMEM230, UCHL1, VPS13A, VPS35, WDR45		5455	6
Spastic paraplegia panel	ABCD1, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BICD2, BSCL2, C12ORF65, C19orf12, CCT5, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FARS2, GBA2, GJC2, HSPD1, IBA57, KDM5C, KIF1A, KIF1C, KIF5A, L1CAM, MAG, MARS1, NIPA1, NT5C2, PGAP1, PLP1, PNPLA6, RAB3GAP2, REEP1, REEP2, RTN2, SACS, SLC16A2, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, TTR, USP8, VAMP1, VPS37A, WASHC5, ZFY-VE26, ZFYVE27		5458	6

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Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
Spastic paraplegia panel	ABCD1, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BICD2, BSCL2, C12ORF65, C19orf12, CCT5, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FARS2, GBA2, GJC2, HSPD1, IBA57, KDM5C, KIF1A, KIF1C, KIF5A, L1CAM, MAG, MARS1, NIPA1, NT5C2, PGAP1, PLP1, PNPLA6, RAB3GAP2, REEP1, REEP2, RTN2, SACS, SLC16A2, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, TTR, USP8, VAMP1, VPS37A, WASHC5, ZFY-VE26, ZFYVE27		5458	6
Spastic paraplegia panel	ABCD1, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BICD2, BSCL2, C12ORF65, C19orf12, CCT5, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FARS2, GBA2, GJC2, HSPD1, IBA57, KDM5C, KIF1A, KIF1C, KIF5A, L1CAM, MAG, MARS1, NIPA1, NT5C2, PGAP1, PLP1, PNPLA6, RAB3GAP2, REEP1, REEP2, RTN2, SACS, SLC16A2, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, TTR, USP8, VAMP1, VPS37A, WASHC5, ZFY-VE26, ZFYVE27		5458	6
Spastic paraplegia panel	ABCD1, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BICD2, BSCL2, C12ORF65, C19orf12, CCT5, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FARS2, GBA2, GJC2, HSPD1, IBA57, KDM5C, KIF1A, KIF1C, KIF5A, L1CAM, MAG, MARS1, NIPA1, NT5C2, PGAP1, PLP1, PNPLA6, RAB3GAP2, REEP1, REEP2, RTN2, SACS, SLC16A2, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, TTR, USP8, VAMP1, VPS37A, WASHC5, ZFY-VE26, ZFYVE27		5458	6

› Neurology

Disease	Gene	Test code	OMIM Gene	Available test methods
46,XY gonadal dysgenesis, partial, with minifascicular neuropathy	DHH	531	605423	2, 3, 4, 8, 9
Achalasia addisonianism alacrimia syndrome	AAAS	121	605378	2, 3, 4, 8, 9, 17
Acrocallosal syndrome	KIF7	151	611254	2, 3, 4, 8, 9, 17
Acyl-CoA peroxisomal oxidase deficiency	ACOX1	615	609751	2, 4, 17
Adrenoleukodystrophy, x-linked	ABCD1	4	300371	2, 4, 8, 9, 17
Adrenoleukodystrophy, x-linked	PLXNB3	2214	300214	2, 4
Adrenoleukodystrophy/Adrenomyeloneuropathy	ABCD1	4	300371	2, 4, 8, 9, 17
Agenesis of the corpus callosum with peripheral neuropathy	SLC12A6	267	604878	2, 3, 4, 8, 9, 17
Aicardi-Goutieres syndrome type 1	TREX1	616	606609	2, 4, 8, 9
Aicardi-Goutieres syndrome type 2	RNASEH2B	617	610326	2, 4, 8, 9, 17
Aicardi-Goutieres syndrome type 3	RNASEH2C	618	610330	2, 4, 8, 9
Aicardi-Goutieres syndrome type 4	RNASEH2A	619	606034	2, 4, 8, 9
Aicardi-Goutieres syndrome type 5	SAMHD1	620	606754	2, 4, 8, 9, 17
Aicardi-Goutieres syndrome type 6	ADAR	86	146920	2, 4, 17
Aicardi-Goutieres syndrome type 7	IFIH1	2188	606951	2, 4, 17
Alexander disease	GFAP	621	137780	2, 3, 4, 8, 9, 17
Allan-Herndon-Dudley syndrome	SLC16A2	268	300095	2, 3, 4, 8, 9
Allan-Herndon-Dudley syndrome	SLC16A2	268	300095	2, 3, 4, 8, 9
Alpha-thalassemia/mental retardation syndrome	ATRX	27	300032	2, 4, 8, 9, 17
Al-Raqad syndrome	DCPS	2742	610534	2, 4
Alternating hemiplegia of childhood type 1	ATP1A2	587	182340	2, 4, 8, 9, 17
Alternating hemiplegia of childhood type 2	ATP1A3	560	182350	2, 4, 8, 9, 17
Alzheimer disease type 1	APP	761	104760	2, 4, 8, 9, 17
Alzheimer disease type 1	NOS3	3793	163729	17
Alzheimer disease type 2	APOE	18	107741	2, 3, 4, 8, 9
Alzheimer disease type 3	PSEN1	233	104311	2, 4, 8, 9, 17
Alzheimer disease type 4	PSEN2	234	600759	2, 3, 4, 8, 9, 17
Alzheimer disease type 18, susceptibility to	ADAM10	3763	602192	17
Alzheimers disease, early onset, autosomal dominant	SORL1	1286	602005	2, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Alzheimers disease, RTN3 related	RTN3	2354	604249	2, 4
Amish infantile epilepsy syndrome	ST3GAL5	764	604402	2, 4, 17
Amyloidosis	TTR	542	176300	2, 3, 4, 8, 9
Amyloidosis, finnish type	GSN	1958	137350	2, 4, 17
Amyotrophic lateral sclerosis risk factor	CHGB	645	118920	2, 4, 17
Amyotrophic lateral sclerosis type 1	SOD1	691	147450	2, 3, 4, 8, 9
Amyotrophic lateral sclerosis type 2, juvenile	ALS2	692	606352	2, 3, 4, 8, 9, 17
Amyotrophic lateral sclerosis type 4	SETX	264	608465	2, 4, 8, 9, 17
Amyotrophic lateral sclerosis type 6	FUS	38	137070	2, 3, 4, 8, 9, 17
Amyotrophic lateral sclerosis type 8	VAPB	693	605704	2, 4
Amyotrophic lateral sclerosis type 9	ANG	694	105850	2, 4
Amyotrophic lateral sclerosis type 10	TARDBP	287	605078	2, 3, 4, 8, 9, 17
Amyotrophic lateral sclerosis type 11	FIG4	530	609390	2, 3, 4, 8, 9, 17
Amyotrophic lateral sclerosis type 12	OPTN	205	602432	2, 3, 4, 8, 9, 17
Amyotrophic lateral sclerosis type 14	VCP	1594	601023	2, 4, 17
Amyotrophic lateral sclerosis type 16	SIGMAR1	1624	601978	2, 4
Amyotrophic lateral sclerosis type 17	CHMP2B	1623	609512	2, 4
Amyotrophic lateral sclerosis type 18	PFN1	93	176610	2, 4
Amyotrophic lateral sclerosis type 21	MATR3	737	164015	2, 3, 4, 8, 9, 17
Amyotrophic lateral sclerosis with frontotemporal dementia	C9orf72	1241	614260	10
Amyotrophic lateral sclerosis, CREST related	SS18L1	1614	606472	2, 4
Amyotrophic lateral sclerosis, susceptibility to	NEFH	1595	162230	2, 3, 4, 8, 9, 17
Amyotrophic lateral sclerosis, VPS54 related	VPS54	1625	614633	2, 4, 17
Amyotrophic lateral sclerosis, x-linked juvenile and adult-onset ALS	UBQLN2	696	300264	2, 4
Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to	TRPM7	3814	605692	17
Amyotrophy hereditary neuralgic	SEPT9	543	604061	2, 4, 8, 9, 17
Angelman syndrome	chr. 15q11	91		8
Angelman syndrome	UBE3A	312	601623	2, 4, 8, 9, 17
Angelman-like syndrome	CDKL5	770	300203	2, 4, 8, 9, 17
Angelman-like syndrome	MECP2	163	300005	2, 4, 8, 9
Arts syndrome	PRPS1	231	311850	2, 3, 4, 8, 9
Asperger syndrome susceptibility X-linked type 2	NLGN3	186	300336	2, 3, 4, 8, 9, 17
Ataxia and muscle hypotonia	COX20	1794	614698	2, 4
Ataxia telangiectasia like disorder	MRE11	1151	600814	2, 3, 4, 8, 9, 17
Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus	DNAJC3	3479	601184	17
Ataxia, posterior column, with retinitis pigmentosa	FLVCR1	1590	609144	2, 3, 4, 8, 9, 17
Ataxia, progressive seizures, mental deterioration, and hearing loss, MT-TV related	MT-TV	2555	590105	4
Ataxia, sensory type 1, autosomal dominant	RNF170	3799	614649	17
Ataxia-oculomotor apraxia type 1	APTX	573	606350	2, 4, 8, 9, 17
Ataxia-oculomotor apraxia type 2	SETX	264	608465	2, 4, 8, 9, 17
Ataxia-oculomotor apraxia type 3	PIK3R5	725	611317	2, 3, 4, 8, 9, 17
Ataxia-oculomotor apraxia type 4	PNKP	774	605610	2, 3, 4, 8, 9, 17
Ataxia-telangiectasia	ATM	1859	607585	2, 8, 17
Attention deficit-hyperactivity disorder	DRD4	2101	126452	2, 4
Attention deficit-hyperactivity disorder	DRD5	2102	126453	2, 4
Autism spectrum disorder	AHNAK2	2212	608570	2, 4, 17
Autism spectrum disorder	ANKS3	2253		2, 4
Autism spectrum disorder	BPIFA3	2251		2, 4
Autism spectrum disorder	EN2	1796	131310	2, 4
Autism spectrum disorder	RABGGTA	2252	601905	2, 4, 17
Autism spectrum disorder, MYO16 related	MYO16	2958	615479	2, 17
Autism spectrum, MXRA5 related	MXRA5	2213		2, 4, 17
Autism spectrum/ hyperactivity/ bipolar disorder, GRM7 related	GRM7	2754	604101	2, 3, 4, 8, 9
Autism susceptibility, type 16	SLC9A9	3640	608396	17
Autism susceptibility, type 18	CHD8	3330	601683	17
Autism susceptibility, type 19	EIF4E	3776	133440	17
Autism susceptibility, X-linked type 1	NLGN3	186	300336	2, 3, 4, 8, 9, 17
Autism susceptibility, X-linked type 2	NLGN4X	622	300427	2, 4, 8, 9, 17
Autism susceptibility, X-linked type 3	MECP2	163	300005	2, 4, 8, 9
Autism susceptibility, X-linked type 4	PTCHD1	2703	300828	2, 4
Autism susceptibility, X-linked type 5	RPL10	798	312173	2, 4
Autism susceptibility, X-linked type 6	TMLHE	3812	300777	17
Autism susceptibility, X-linked type 17	SHANK2	2034	603290	2, 4, 8, 9, 17
Autism, ATP1B4 related	ATP1B4	2242		2, 4

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Disease	Gene	Test code	OMIM Gene	Available test methods
Autism, AVPR1A related	AVPR1A	2368	600821	2, 4
Autism, C7orf43 related	C7orf43	2579		2, 4
Autism, CELF6 related	CELF6	2404	612681	2, 4
Autism, EFCAB13 related	EFCAB13	2475		2, 4
Autism, FAAH2 related	FAAH2	2403	300654	2, 4, 17
Autism, FCRL6 related	FCRL6	2285	613562	2, 4
Autism, GYG2 related	GYG2	2654	300198	2, 4
Autism, IQCE related	IQCE	2644		2, 4
Autism, MBD1 related	MBD1	2031	156535	2, 4, 17
Autism, NTNG1 related	NTNG1	2477	608818	2, 4, 8, 9
Autism, OR13H1 related	OR13H1	2032		2, 4
Autism, OXTR related	OXTR	2367	167055	2, 4
Autism, PKHD1L1 related	PKHD1L1	2027	607843	2, 17
Autism, RNF128 related	RNF128	2306	300439	2, 4
Autism, RRM1 related	RRM1	2508		2, 4, 17
Autism, SETD2 related	SETD2	2383	612778	2, 4, 17
Autism, SLC22A9 related	SLC22A9	2646	607579	2, 4
Autism, UNC13B related	UNC13B	2535	605836	2, 4
Autism, ZNF778 related	ZNF778	2369		2, 4
Autism/Mental retardation/Angelman syndrome, susceptibility to, ATP10A related	ATP10A	2645	605855	2, 4, 17
Bethlem myopathy	COL6A1	697	120220	2, 3, 4, 8, 9, 17
Bethlem myopathy	COL6A2	698	120240	2, 3, 4, 8, 9, 17
Bethlem myopathy type 1	COL6A3	699	120250	2, 3, 4, 8, 9, 17
Bethlem myopathy type 2	COL12A1	1757	120320	2, 17
Borjeson-Forssman-Lehmann syndrome	PHF6	799	300414	2, 3, 4, 8, 9, 17
Brody myopathy	ATP2A1	700	108730	2, 3, 4, 8, 9, 17
Brunner syndrome	MAOA	800	309850	2, 4, 17
Budd-Chiari syndrome	F5	375	612309	2, 4
Canavan disease	ASPA	626	608034	2, 4, 8, 9
CAPOS syndrome	ATP1A3	560	182350	2, 4, 8, 9, 17
Cardiomyopathy, hypertrophic, type 24	LDB3	157	605906	2, 4, 17
Cataracts with facial dysmorphism and neuropathy	CTDP1	544	604927	2, 3, 4, 8, 9, 17
Central core disease	RYR1	249	180901	2, 17
Central hypoventilation syndrome with or without Hirschsprung disease	PHOX2B	740	603851	2, 3, 4, 8, 9, 10
Central hypoventilation syndrome, congenital	ASCL1	712	100790	2, 4, 8, 9
Centronuclear myopathy type 1	DNM2	546	602378	2, 3, 4, 8, 9, 17
Centronuclear myopathy type 1	MTMR14	496	611089	2, 4, 17
Centronuclear myopathy type 2	BIN1	735	601248	2, 4, 17
Centronuclear myopathy type 3	MYF6	613	159991	2, 4
Centronuclear myopathy type 4	CCDC78	1108	614666	2, 4, 17
Centronuclear myopathy type 5	SPEG	3378	615950	17
Cerebellar ataxia	CP	1591	117700	2, 4, 17
Cerebellar ataxia and mental retardation with or without quadrupedal locomotion type 3	CA8	2330	114815	2, 4, 17
Cerebellar ataxia with deafness and narcolepsy, autosomal recessive	DNMT1	878	126375	2, 3, 4, 8, 9, 17
Cerebellar ataxia with mental retardation and dysequilibrium syndrome type 2	WDR81	2195	614218	2, 4, 17
Cerebellar ataxia with spasticity	GBA2	1523	609471	2, 3, 4, 8, 9, 17
Cerebellar ataxia, mental retardation, and dysequilibrium syndrome type 4	ATP8A2	1530	605870	2, 17
Cerebellar ataxia, nonprogressive, with mental retardation	CAMTA1	2515	611501	2, 3, 4, 8, 9, 17
Cerebellar ataxia, SNX14 related	SNX14	2304	616105	2, 4, 17
Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion type 1	VLDLR	94	192977	2, 4, 8, 9, 17
Cerebral amyloid angiopathy	CST3	1871	604312	2, 3, 4, 8, 9
Cerebral amyloid angiopathy, APP related	APP	761	104760	2, 4, 8, 9, 17
Cerebral infarction, susceptibility to	PRKCH	3794	605437	17
Cerebral palsy type 1, spastic quadriplegic	GAD1	2211	605363	2, 4, 17
Cerebral palsy type 2, spastic quadriplegic	KANK1	2294	607704	2, 3, 4, 8, 9, 17
Cerebrotendinous xanthomatosis	CYP27A1	627	606530	2, 3, 4, 8, 9, 17
Cervical dystonia	CIZ1	757	611420	2, 3, 4, 8, 9, 17
Charcot-Marie-Tooth disease, axonal type 20	DYNC1H1	1279	600112	2, 17
CHILD syndrome	NSDHL	202	300275	2, 3, 4, 8, 9, 17
Chorea, hereditary benign	NKX2-1	551	600635	2, 3, 4, 8, 9
Choreoacanthocytosis	VPS13A	1445	605978	2, 3, 8, 17
CK syndrome	NSDHL	202	300275	2, 3, 4, 8, 9, 17
CMT1A	PMP22	219	601097	2, 4, 8, 9
CMT1B	MPZ	169	159440	2, 4, 8, 9

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Disease	Gene	Test code	OMIM Gene	Available test methods
CMT1C	LITAF	510	603795	2, 4
CMT1D	EGR2	511	129010	2, 4, 8, 9
CMT1E	PMP22	219	601097	2, 4, 8, 9
CMT2A1	KIF1B	513	605995	2, 3, 4, 8, 9
CMT2A2	MFN2	514	608507	2, 4, 8, 9, 17
CMT2B1	LMNA	158	150330	2, 4, 8, 9, 17
CMT2B2	MED25	516	610197	2, 3, 4, 8, 9, 17
CMT2B	RAB7A	515	602298	2, 4, 8, 9
CMT2C	TRPV4	517	605427	2, 3, 4, 8, 9, 17
CMT2D	GARS1	518	600287	2, 4, 8, 9, 17
CMT2F	HSPB1	135	602195	2, 4, 8, 9
CMT2I	MPZ	169	159440	2, 4, 8, 9
CMT2J	MPZ	169	159440	2, 4, 8, 9
CMT2K	GDAP1	520	606598	2, 4, 8, 9
CMT2L	HSPB8	136	608014	2, 4, 8, 9
CMT2N	AARS1	512	601065	2, 4, 17
CMT2P	LRSAM1	2674	610933	2, 4, 17
CMT4, CTDP1 related	CTDP1	544	604927	2, 3, 4, 8, 9
CMT4A	GDAP1	520	606598	2, 4, 8, 9
CMT4B1	MTMR2	523	603557	2, 4, 8, 9, 17
CMT4B2	SBF2	524	607697	2, 4, 8, 9, 17
CMT4C	SH3TC2	525	608206	2, 4, 8, 9, 17
CMT4D	NDRG1	526	605262	2, 4, 17
CMT4E	EGR2	511	129010	2, 4, 8, 9
CMT4E	MPZ	169	159440	2, 4, 8, 9
CMT4F	PRX	528	605725	2, 4, 8, 9, 17
CMT4H	FGD4	529	611104	2, 3, 4, 8, 9, 17
CMT4J	FIG4	530	609390	2, 3, 4, 8, 9, 17
CMTDIF	GNB4	1539	610863	2, 4, 17
CMTRIB	KARS1	2806	601421	2, 4, 17
CMTRIB	KARS1	2806	601421	2, 4, 17
CMTRIB	KARS1	2806	601421	2, 4, 17
CMTRID	COX6A1	2653	602072	2, 4
CMTX1	GJB1	545	304040	2, 4, 8, 9
CMTX4	AIFM1	1353	300169	2, 3, 4, 8, 9, 17
CMTX5	PRPS1	231	311850	2, 3, 4, 8, 9
COACH syndrome	CC2D2A	628	612013	2, 3, 4, 8, 9, 17
COACH syndrome	RPGRIP1L	1138	610937	2, 4, 17
COACH syndrome	TMEM67	301	609884	2, 4, 17
Coenzyme Q10 deficiency type 1	COQ2	577	609825	2, 4
Coenzyme Q10 deficiency type 2	PDSS1	578	607429	2, 3, 4, 8, 9, 17
Coenzyme Q10 deficiency type 3	PDSS2	579	610564	2, 3, 4, 8, 9, 17
Coenzyme Q10 deficiency type 5	COQ9	580	612837	2, 3, 4, 8, 9, 17
Coffin-Lowry syndrome	RPS6KA3	247	300075	2, 4, 8, 9, 17
Cohen syndrome	VPS13B	323	607817	2, 8, 17
Compton-North congenital myopathy	CNTN1	1237	600016	2, 4, 17
Congenital muscular dystrophy and hypoglycosylation of α-dystroglycan	B3GALNT2	1536	610194	2, 4, 17
Convulsions, benign familial infantile, 3	SCN2A	255	182390	2, 3, 4, 8, 9, 17
Convulsions, familial infantile, with paroxysmal choreoathetosis	PRRT2	558	614386	2, 3, 4, 8, 9
Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia	IGBP1	629	300139	2, 4, 17
Cortical dysplasia, complex, with other brain malformations, type 1	TUBB3	310	602661	2, 4
Cortical dysplasia, complex, with other brain malformations, type 5	TUBB2A	2067	615101	2, 3, 4, 8, 9
Cortical dysplasia-focal epilepsy syndrome	CNTNAP2	768	604569	2, 3, 4, 8, 9, 17
Corticobasal Degeneration, CFL1 related	CFL1	2286	601442	2, 4
CR1 deficiency	CR1	3774	120620	17
Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome	TMCO1	3811	614123	17
Creatine deficiency syndrome X-linked	SLC6A8	276	300036	2, 4, 8, 9
Creatine phosphokinase, elevated serum	CAV3	701	601253	2, 4, 8, 9
Creutzfeldt-Jakob disease	PRNP	227	176640	2, 4, 8, 9, 10
Cytochrome c oxidase 1 deficiency	MT-CO1	1738	516030	4
Cytochrome c oxidase 2 deficiency	MT-CO2	2357	516040	4
Cytochrome c oxidase 3 deficiency	MT-CO3	1739	516050	4
Danon disease	LAMP2	155	309060	2, 3, 4, 8, 9, 17
Dejerine-Sottas disease	EGR2	511	129010	2, 4, 8, 9

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Disease	Gene	Test code	OMIM Gene	Available test methods
Dejerine-Sottas disease	GJB1	545	304040	2, 4, 8, 9
Dejerine-Sottas disease	MPZ	169	159440	2, 4, 8, 9
Dejerine-Sottas disease	PMP22	219	601097	2, 4, 8, 9
Dejerine-Sottas disease	PRX	528	605725	2, 4, 8, 9, 17
Dementia, familial, British type	ITM2B	2623	603904	2, 4
Dementia, familial, Danish type	ITM2B	2623	603904	2, 4
Dementia, frontotemporal	GRN	762	138945	2, 4, 8, 9, 17
Dementia, frontotemporal	MAPT	763	157140	2, 4, 8, 9, 17
Dementia, frontotemporal	PSEN1	233	104311	2, 4, 8, 9, 17
Dementia, frontotemporal	TARDBP	287	605078	2, 3, 4, 8, 9, 17
Dementia, Lewy body	SNCA	563	163890	2, 4, 8, 9
Dent disease type 2	OCRL	203	300535	2, 3, 4, 8, 9, 17
Dentatorubral-pallidoluysian atrophy	ATN1	581	607462	2, 4, 10, 17
Developmental delay and microcephaly, SLC1A4 related	SLC1A4	2689	600229	2, 4
Developmental delay, GNAQ related	GNAQ	2140	600998	2, 3, 4, 8, 9
Developmental delay, KMT2C related	KMT2C	2499	606833	2, 17
DI-CMTB	DNM2	546	602378	2, 3, 4, 8, 9, 17
DI-CMTC	YARS1	547	603623	2, 3, 4, 8, 9, 17
DI-CMTD	MPZ	169	159440	2, 4, 8, 9
Dravet syndrome	GABRG2	769	137164	2, 3, 4, 8, 9, 17
Dravet syndrome	SCN2A	255	182390	2, 3, 4, 8, 9, 17
Dravet syndrome, modifier of	SCN9A	258	603415	2, 3, 4, 8, 9, 17
Dysautonomia, FRRS1L-related	FRRS1L	2305	604574	2, 4
Dyskinesia, familial, with facial myokymia	ADCY5	2125	600293	2, 3, 4, 8, 9, 17
Dyskinesia, limb and orofacial, infantile-onset	PDE10A	3514	610652	17
Dyslexia	PCDH11X	2033	300246	2, 4, 17
Dyssegmental dysplasia, Silverman-Handmaker type	HSPG2	137	142461	2, 17
Dystonia juvenile-onset	ACTB	779	102630	2, 3, 4, 8, 9
Dystonia, DOPA-responsive, autosomanl recessive	SPR	552	182125	2, 3, 4, 8, 9
Dystonia-deafness syndrome	TIMM8A	298	300356	2, 4, 8, 9
DYT1	TOR1A	553	605204	2, 4, 8, 9
DYT2	HPCA	2884	142622	2, 4
DYT3	TAF1	554	313650	2, 3, 4, 8, 9, 17
DYT4	TUBB4A	1359	602662	2, 3, 4, 8, 9
DYT5A	GCH1	555	600225	2, 4, 8, 9, 17
DYT6	THAP1	556	609520	2, 4, 8, 9
DYT8	SLC2A1	193	138140	2, 4, 8, 9, 17
DYT10	PRRT2	558	614386	2, 3, 4, 8, 9
DYT11, DRD2 related	DRD2	2276	126450	2, 3, 4, 8, 9
DYT11	SGCE	559	604149	2, 4, 8, 9, 17
DYT12	ATP1A3	560	182350	2, 4, 8, 9, 17
DYT16	PRKRA	561	603424	2, 4, 8, 9
DYT18	SLC2A1	193	138140	2, 4, 8, 9, 17
DYT23	CACNA1B	2965	601012	2, 4, 17
DYT24	ANO3	85	610110	2, 3, 4, 8, 9, 17
DYT25	GNAL	84	139312	2, 3, 4, 8, 9, 17
DYT26, myoclonic	KCTD17	3030	616386	2, 4, 17
DYT27	COL6A3	699	120250	2, 3, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 1	ARX	24	300382	2, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 2	CDKL5	770	300203	2, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 3	SLC25A22	771	609302	2, 4
Early infantile epileptic encephalopathy type 4	STXBP1	772	602926	2, 3, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 5	SPTAN1	773	182810	2, 4, 17
Early infantile epileptic encephalopathy type 6	SCN1A	253	182389	2, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 7	KCNQ2	149	602235	2, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 8	ARHGEF9	21	300429	2, 4, 17
Early infantile epileptic encephalopathy type 9	PCDH19	208	300460	2, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 10	PNKP	774	605610	2, 3, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 11	SCN2A	255	182390	2, 3, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 12	PLCB1	775	607120	2, 3, 8, 17
Early infantile epileptic encephalopathy type 13	SCN8A	624	600702	2, 4, 17
Early infantile epileptic encephalopathy type 14	KCNT1	89	608167	2, 3, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 15	ST3GAL3	2676	606494	2, 4, 17
Early infantile epileptic encephalopathy type 16	TBC1D24	783	613577	2, 3, 4, 8, 9, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Early infantile epileptic encephalopathy type 17	GNAO1	3045	139311	2, 4, 17
Early infantile epileptic encephalopathy type 19	GABRA1	673	137160	2, 4, 17
Early infantile epileptic encephalopathy type 20	PIGA	2238	311770	2, 3, 4, 8, 9
Early infantile epileptic encephalopathy type 21	NECAP1	2673	611623	2, 4, 17
Early infantile epileptic encephalopathy type 23	DOCK7	2672	615730	2, 17
Early infantile epileptic encephalopathy type 24	HCN1	2671	602780	2, 3, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 25	SLC13A5	2270	608305	2, 3, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 26	KCNB1	2494	600397	2, 4
Early infantile epileptic encephalopathy type 27	GRIN2B	1600	138252	2, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 28	WWOX	2472	605131	2, 4, 8, 9, 17
Early infantile epileptic encephalopathy type 29	AARS1	512	601065	2, 4, 17
Early infantile epileptic encephalopathy type 30	SIK1	2669	605705	2, 4, 17
Early infantile epileptic encephalopathy type 31	DNM1	2667	602377	2, 4, 17
Early infantile epileptic encephalopathy type 32	KCNA2	2651	176262	2, 4
Early infantile epileptic encephalopathy type 33	EEF1A2	2514	602959	2, 4, 17
Early infantile epileptic encephalopathy type 40	GUF1	3231	617064	2, 4, 17
Early infantile epileptic encephalopathy type 45	GABRB1	3229	137190	2, 4, 17
Early infantile epileptic encephalopathy type 46	GRIN2D	3230	602717	2, 4, 17
Early infantile epileptic encephalopathy type 47	FGF12	3234	601513	2, 4
Early infantile epileptic encephalopathy type 67	CUX2	1917	610648	2, 4
Emery-Dreifuss muscular dystrophy type 1	EMD	702	300384	2, 3, 4, 8, 9
Emery-Dreifuss muscular dystrophy type 2	LMNA	158	150330	2, 4, 8, 9, 17
Emery-Dreifuss muscular dystrophy type 4	SYNE1	286	608441	2, 3, 8, 17
Emery-Dreifuss muscular dystrophy type 5	SYNE2	703	608442	2, 17
Emery-Dreifuss muscular dystrophy type 6	FHL1	1249	300163	2, 3, 4, 8, 9, 17
Encephalomyopathy, mitochondrial, MT-TL2 related	MT-TL2	1313	590055	4
Encephalomyopathy, mitochondrial, MT-TR related	MT-TR	2563	590005	4
Encephalopathy lethal, due to defective mitochondrial peroxisomal fission	DNM1L	1439	603850	2, 4, 17
Encephalopathy mitochondrial	VDAC1	1596	604492	2, 4
Encephalopathy mitochondrial with proximal renal tubulopathy due to cytochrome c oxidase deficiency	COX10	1367	602125	2, 4, 8, 9, 17
Encephalopathy mitochondrial, MT-TW related	MT-TW	2557	590095	4
Encephalopathy neonatal severe	MECP2	163	300005	2, 4, 8, 9
Encephalopathy thiamine-responsive	SLC19A3	270	606152	2, 3, 4, 8, 9
Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, type 5	TRAF3	3813	601896	17
Encephalopathy, acute, necrotizing, type 1	RANBP2	630	601181	2, 3, 4, 8, 9, 17
Encephalopathy, familial, with neuroserpin inclusion bodies	SERPINI1	2063	602445	2, 4, 17
Encephalopathy, progressive, with or without lipodystrophy	BSCL2	440	606158	2, 3, 4, 8, 9, 17
Endplate acetylcholinesterase deficiency	COLQ	704	603033	2, 4, 17
Epidermolysis bullosa simplex with muscular dystrophy	PLEC	705	601282	2, 17
Epidermolysis bullosa simplex, autosomal recessive type 2	DST	1263	113810	2, 17
Epilepsy with neurodevelopmental defects	GRIN2A	1599	138253	2, 4, 8, 9, 17
Epilepsy, childhood absence type 2	GABRG2	769	137164	2, 3, 4, 8, 9, 17
Epilepsy, childhood absence type 4, susceptibility to	GABRA1	673	137160	2, 4, 17
Epilepsy, childhood absence type 5	GABRB3	796	137192	2, 4, 8, 9, 17
Epilepsy, childhood absence type 6, susceptibility to	CACNA1H	662	607904	2, 4, 17
Epilepsy, familial focal with variable foci	DEPDC5	1617	614191	2, 3, 4, 8, 9, 17
Epilepsy, familial temporal lobe type 1	LGI1	765	604619	2, 4, 8, 9, 17
Epilepsy, familial temporal lobe type 5	CPA6	879	609562	2, 4, 17
Epilepsy, familial temporal lobe type 7	RELN	63	600514	2, 17
Epilepsy, focal, SCN3A related	SCN3A	2399	182391	2, 4, 17
Epilepsy, HCN2 related	HCN2	2495	602781	2, 4
Epilepsy, hearing loss, and mental retardation syndrome	SPATA5	2807	613940	2, 4, 17
Epilepsy, idiopathic generalized type 10	GABRD	778	137163	2, 3, 4, 8, 9, 17
Epilepsy, idiopathic generalized type 11	CLCN2	1094	600570	2, 3, 4, 8, 9, 17
Epilepsy, idiopathic generalized type 12	SLC2A1	193	138140	2, 4, 8, 9, 17
Epilepsy, juvenile absence type 1	EFHC1	897	608815	2, 3, 4, 8, 9, 17
Epilepsy, nocturnal frontal lobe	KCNT1	89	608167	2, 3, 4, 8, 9, 17
Epilepsy, nocturnal frontal lobe type 1	CHRNA4	766	118504	2, 4, 8, 9, 17
Epilepsy, nocturnal frontal lobe type 3	CHRNB2	780	118507	2, 4, 8, 9
Epilepsy, nocturnal frontal lobe type 4	CHRNA2	767	118502	2, 3, 4, 8, 9, 17
Epilepsy, progressive myoclonic type 4, with or without renal failure	SCARB2	1976	602257	2, 3, 4, 8, 9, 17
Epilepsy, progressive myoclonic type 5	PRICKLE2	2549	608501	2, 4, 17
Epilepsy, progressive myoclonic type 7	KCNC1	3586	176258	17
Epilepsy, X-linked, with learning disabilities and behavior disorders	SYN1	801	313440	2, 3, 4, 8, 9, 17

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Epileptic encephalopathy, childhood-onset	CHD2	1902	602119	2, 17
Epileptic encephalopathy, Lennox-Gastaut type	MAPK10	1592	602897	2, 4, 17
Episodic ataxia type 1	KCNA1	582	176260	2, 4, 8, 9
Episodic ataxia type 2	CACNA1A	583	601011	2, 4, 8, 9, 10, 17
Episodic ataxia type 5	CACNB4	584	601949	2, 3, 4, 8, 9, 17
Episodic ataxia type 6	SLC1A3	585	600111	2, 3, 4, 8, 9, 17
Episodic pain syndrome type 2, familial	SCN10A	2264	604427	2, 4, 17
Episodic pain syndrome type 3, familial	SCN11A	2265	604385	2, 4, 17
Erythermalgia, primary	SCN9A	258	603415	2, 3, 4, 8, 9, 17
Ethylmalonic encephalopathy	ETHE1	967	608451	2, 3, 4, 8, 9
Facial paresis type 3	HOXB1	1781	142968	2, 4
Facioscapulohumeral dystrophy-like phenotype, FAT1 related	FAT1	2578	600976	2, 4, 17
Familial hemiplegic migraine type 1	CACNA1A	583	601011	2, 4, 8, 9
Familial hemiplegic migraine type 2	ATP1A2	587	182340	2, 4, 8, 9, 17
Familial hemiplegic migraine type 3	SCN1A	253	182389	2, 4, 8, 9, 17
Familial infantile myoclonic epilepsy	TBC1D24	783	613577	2, 3, 4, 8, 9, 17
Fatal familial imsomnia	PRNP	227	176640	2, 4, 8, 9, 10
Febrile seizures, familial, type 4	ADGRV1	2191	602851	2, 17
FG syndrome type 1	MED12	164	300188	2, 3, 4, 8, 9, 17
FG syndrome type 2	FLNA	803	300017	2, 3, 4, 8, 9, 17
FG syndrome type 4	CASK	804	300172	2, 4, 8, 9, 17
Filaminopathy	FLNC	708	102565	2, 4, 17
Fragile X syndrome	FMR1	588	309550	2, 4, 8, 9, 10
Fragile X tremor/ataxia syndrome	FMR1	588	309550	2, 4, 8, 9, 10
Friedreich ataxia	FXN	589	606829	2, 4, 8, 9, 10
Frontometaphyseal dysplasia	FLNA	803	300017	2, 3, 4, 8, 9, 17
Fucosidosis	FUCA1	197	612280	2, 3, 4, 8, 9, 12
Fukuyama congenital muscular dystrophy	FKTN	709	607440	2, 4, 8, 9, 17
Gaze palsy, horizontal, with progressive scoliosis	ROBO3	2231	608630	2, 4, 17
Generalized epilepsy and paroxysmal dyskinesia	KCNMA1	784	600150	2, 4, 17
Generalized epilepsy with febrile seizures plus type 1	SCN1B	254	600235	2, 3, 4, 8, 9
Generalized epilepsy with febrile seizures plus type 2	SCN1A	253	182389	2, 4, 8, 9, 17
Generalized epilepsy with febrile seizures plus type 3	GABRG2	769	137164	2, 3, 4, 8, 9, 17
Generalized epilepsy with febrile seizures plus type 7	SCN9A	258	603415	2, 3, 4, 8, 9, 17
Generalized epilepsy with febrile seizures plus type 9	STX1B	3129	601485	17
Gerstmann-Straussler disease	PRNP	227	176640	2, 4, 8, 9, 10
Giant axonal neuropathy type 1	GAN	548	605379	2, 3, 4, 8, 9, 17
Gillespie syndrome	ITPR1	607	147265	2, 17
GLUT1 deficiency syndrome type 1	SLC2A1	193	138140	2, 4, 8, 9, 17
Glycine encephalopathy	AMT	1455	238310	2, 4, 8, 9, 17
Glycine encephalopathy	GCSH	632	238330	2, 4, 8, 9
Glycosylation disorde type 2A	MGAT2	165	602616	2, 4
Glycosylation disorde type 2C	SLC35C1	274	605881	2, 4
Glycosylation disorder type 1C	ALG6	15	604566	2, 4, 17
Glycosylation disorder type 1E	DPM1	412	603503	2, 4, 17
Glycosylation disorder type 1J	DPAGT1	634	191350	2, 4, 17
Glycosylation disorder type 1M	DOLK	414	610746	2, 4
Glycosylation disorder type 2D	B4GALT1	316	137060	2, 4
Glycosylation disorder type 2E	COG7	198	606978	2, 4, 17
Glycosylation disorder type 2F	SLC35A1	273	605634	2, 4, 17
Glycosylation disorder type 2G	COG1	416	606973	2, 3, 4, 8, 9, 17
Glycosylation disorder type 2H	COG8	417	606979	2, 4, 17
GM1-gangliosidosis	GLB1	127	611458	2, 3, 4, 8, 9, 17
Gordon Holmes Syndrome	RNF216	3602	609948	2, 4, 17
Griscelli syndrome type 2	RAB27A	236	603868	2, 3, 4, 8, 9
Hereditary motor and sensory neuropathy, Okinawa type	TFG	2593	602498	2, 4, 17
Hereditary myopathy with early respiratory failure	TTN	309	188840	2, 17
Heterotopia, periventricular, ED variant	FLNA	803	300017	2, 3, 4, 8, 9, 17
Heterotopia, periventricular, X-linked dominant	FLNA	803	300017	2, 3, 4, 8, 9, 17
Hippocampal longterm potentiation, RFN39 related	RNF39	2082	607524	2, 4
Hoyeraal-Hreidarsson syndrome	DKC1	810	300126	2, 4, 8, 9, 17
HSAN1	SPTLC1	532	605712	2, 4, 8, 9, 17
HSAN2A	WNK1	533	605232	2, 3, 4, 8, 9, 17
HSAN2B	RETREG1	534	613114	2, 3, 4, 8, 9, 17

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HSAN3	ELP1	535	603722	2, 3, 4, 8, 9, 17
HSAN4	NTRK1	536	191315	2, 3, 4, 8, 9, 17
HSAN5	NGF	537	162030	2, 3, 4, 8, 9
HSAN8	PRDM12	2851	616458	2, 4
HSN2C	KIF1A	150	601255	2, 3, 4, 8, 9, 17
Huntington disease	HTT	591	613004	10
Huntington disease, ZDHHC17 related	ZDHHC17	2293	607799	2, 4
Huntington disease-like type 1	PRNP	227	176640	2, 4, 8, 9, 10
Huntington disease-like type 2	JPH3	592	605268	10
Hydranencephaly with abnormal genitalia/Lissencephaly X-linked 2	ARX	24	300382	2, 4, 8, 9, 17
Hydrocephalus with aqueductal stenosis and congenital intestinal pseudoobstraction	L1CAM	153	308840	2, 3, 4, 8, 9
Hydrocephalus, nonsyndromic, autosomal recessive type 1	CCDC88C	2052	611204	2, 4, 17
Hydrocephalus, nonsyndromic, autosomal recessive type 2	MPDZ	2051	603785	2, 17
Hyperekplexia	GLRA1	787	138491	2, 4, 8, 9, 17
Hyperekplexia	GLRB	788	138492	2, 4, 8, 9, 17
Hyperekplexia	SLC6A5	789	604159	2, 4, 8, 9, 17
Hyperekplexia, EIEE8 related	ARHGEF9	21	300429	2, 4, 17
Hyperkalemic periodic paralysis	SCN4A	256	603967	2, 4, 8, 9, 17
Hyperphenylalaninemia, BH4-deficient, B	GCH1	555	600225	2, 4, 8, 9, 17
Hypokalemic periodic paralysis type 1	CACNA1S	710	114208	2, 4, 8, 9, 17
Hypomyelination with brainstem and spinal cord involvement and leg spasticity	DARS	2766	603084	2, 4, 17
Inclusion body myopathy	GNE	129	603824	2, 3, 4, 8, 9, 17
Inclusion body myopathy	MYH2	2275	160740	2, 4, 17
Infantile neuroaxonal dystrophy type 1	PLA2G6	570	603604	2, 3, 4, 8, 9, 17
Insensitivity to pain, channelopathy-associated	SCN9A	258	603415	2, 3, 4, 8, 9, 17
Intellectual disability nonsyndromic, CIC related	CIC	2120	612082	2, 4
Intellectual disability nonsyndromic, CNKSR2 related	CNKSR2	2449	300724	2, 4, 17
Intellectual disability, TBR1 related	TBR1	2822	604616	2, 4
Intestinal pseudoobstraction, neuronal	FLNA	803	300017	2, 3, 4, 8, 9, 17
Jensen syndrome	TIMM8A	298	300356	2, 4, 8, 9
Joubert syndrome type 1	INPP5E	639	613037	2, 4, 17
Joubert syndrome type 2	TMEM216	640	613277	2, 4
Joubert syndrome type 3	AHI1	641	608894	2, 4, 17
Joubert syndrome type 4	NPHP1	190	607100	2, 4, 8, 9, 17
Joubert syndrome type 5	CEP290	642	610142	2, 3, 4, 8, 9
Joubert syndrome type 6	TMEM67	301	609884	2, 4, 17
Joubert syndrome type 7	RPGRIP1L	1138	610937	2, 4, 17
Joubert syndrome type 8	ARL13B	643	608922	2, 4, 17
Joubert syndrome type 9	CC2D2A	628	612013	2, 3, 4, 8, 9, 17
Joubert syndrome type 10	OFD1	204	300170	2, 3, 4, 8, 9, 17
Joubert syndrome type 13	TCTN1	1877	609863	2, 4, 17
Joubert syndrome type 14	TMEM237	1867	614423	2, 4, 17
Joubert syndrome type 15	CEP41	636	610523	2, 4, 17
Joubert syndrome type 16	TMEM138	1282	614459	2, 4
Joubert syndrome type 17	CPLANE1	637	614571	2, 17
Joubert syndrome type 18	TCTN3	2200	613847	2, 3, 4, 8, 9, 17
Joubert syndrome type 20	TMEM231	1529	614949	2, 4, 17
Joubert syndrome type 21	CSPP1	2131	611654	2, 4, 17
Joubert syndrome type 22	PDE6D	2957	602676	2, 4
Joubert syndrome type 23	KIAA0586	2801	610178	2, 4
Joubert syndrome type 24	TCTN2	1407	613846	2, 4, 17
Joubert syndrome, EXOC8 related	EXOC8	1955	615283	2, 4
Joubert syndrome, EXOSC8 related	EXOSC8	1219	606019	2, 4, 17
Kabuki syndrome type 2	KDM6A	720	300128	2, 3, 4, 8, 9, 17
Kenny-Caffey syndrome type 2	FAM111A	2046	615292	2, 4, 17
King-Denborough syndrome	RYR1	249	180901	2, 17
Kohlschutter Tonz syndrome	ROGDI	638	614574	2, 3, 4, 8, 9
Krabbe disease	GALC	437	606890	2, 4, 8, 9, 12, 17
Leigh syndrome	BCS1L	1423	603647	2, 3, 4, 8, 9, 17
Leigh syndrome	COX15	1368	603646	2, 3, 4, 8, 9, 17
Leigh syndrome	FOXRED1	1430	613622	2, 4, 17
Leigh syndrome	NDUFA2	1378	602137	2, 4
Leigh syndrome	NDUFA9	1964	603834	2, 4, 17
Leigh syndrome	NDUFA10	1376	603835	2, 4, 17

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Leigh syndrome	NDUFAF1	41	606934	2, 4
Leigh syndrome	NDUFAF2	1379	609653	2, 3, 4, 8, 9
Leigh syndrome	NDUFAF3	42	612911	2, 4
Leigh syndrome	NDUFAF6	1366	612392	2, 3, 4, 8, 9, 17
Leigh syndrome	NDUFS3	1383	603846	2, 4
Leigh syndrome	NDUFS4	1384	602694	2, 4
Leigh syndrome	NDUFS7	1387	601825	2, 3, 4, 8, 9, 17
Leigh syndrome	NDUFS8	1388	602141	2, 4
Leigh syndrome	NUBPL	43	613621	2, 4, 17
Leigh syndrome	SDHA	1188	600857	2, 3, 4, 8, 9, 17
Leigh syndrome and mitochondrial encephalopathy	ACAD9	1352	611103	2, 4, 17
Leigh syndrome due to COX deficiency	SURF1	1428	185620	2, 3, 4, 8, 9
Leigh syndrome due to mitochondrial complex I deficiency	MT-ND3	646	516002	4
Leigh syndrome due to mitochondrial complex I deficiency	MT-ND5	1315	516005	4
Leigh syndrome due to mitochondrial complex I deficiency	MT-ND6	1316	516006	4
Leigh syndrome due to mitochondrial complex I deficiency	NDUFA12	2282	614530	2, 4
Leigh syndrome due to pyruvate and alpha-ketoglutarate dehydrogenase deficiencies, LIPT1 related	LIPT1	2364	610284	2, 4
Leigh syndrome due to pyruvate carboxylase deficiency	PC	519	608786	2, 3, 4, 8, 9, 17
Leigh syndrome due to the mitochondrial complex IV deficiency	TACO1	1408	612958	2, 4
Leigh syndrome, French-Canadian type	LRPPRC	1441	607544	2, 4, 17
Leigh syndrome, X-linked	PDHA1	211	300502	2, 4, 8, 9, 17
Lesch-Nyham syndrome	HPRT1	812	308000	2, 4, 8, 9, 17
Leukodystrophy demyelinating adult-onset, autosomal dominant	LMNB1	647	150340	2, 4, 8, 9, 17
Leukodystrophy hypomyelinating	GJC2	508	608803	2, 4, 8, 9
Leukodystrophy hypomyelinating type 3	AIMP1	752	603605	2, 3, 4, 8, 9
Leukodystrophy hypomyelinating type 4	HSPD1	495	118190	2, 3, 4, 8, 9, 17
Leukodystrophy hypomyelinating type 5	FAM126A	1602	610531	2, 3, 4, 8, 9, 17
Leukodystrophy hypomyelinating type 6	TUBB4A	1359	602662	2, 3, 4, 8, 9
Leukodystrophy hypomyelinating type 7	POLR3A	644	614258	2, 3, 4, 8, 9, 17
Leukodystrophy hypomyelinating type 8	POLR3B	782	614366	2, 3, 4, 8, 9, 17
Leukodystrophy hypomyelinating type 9	RARS	2811	107820	2, 4, 17
Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation	DARS2	649	610956	2, 3, 4, 8, 9, 17
Leukoencephalopathy with dystonia and motor neuropathy	SCP2	1346	184755	2, 4, 17
Leukoencephalopathy with vanishing white matter	EIF2B1	650	606686	2, 4, 17
Leukoencephalopathy with vanishing white matter	EIF2B2	651	606454	2, 4, 17
Leukoencephalopathy with vanishing white matter	EIF2B3	652	606273	2, 4, 17
Leukoencephalopathy with vanishing white matter	EIF2B4	653	606687	2, 4, 17
Leukoencephalopathy with vanishing white matter	EIF2B5	654	603945	2, 3, 4, 8, 9, 17
Leukoencephalopathy, cystic without megalencephaly	RNASET2	797	612944	2, 3, 4, 8, 9, 17
Leukoencephalopathy, diffuse hereditary, with spheroids	CSF1R	1284	164770	2, 3, 4, 8, 9, 17
Leukoencephalopathy, progressive, with ovarian failure	AARS2	2607	612035	2, 4, 17
Lewy body dementia, susceptibility to	GBA	399	606463	2, 3, 4, 8, 9
Limb-girdle muscular dystrophy, autosomal dominant type 1A	MYOT	711	604103	2, 4, 8, 9, 17
Limb-girdle muscular dystrophy, autosomal dominant type 1B	LMNA	158	150330	2, 4, 8, 9, 17
Limb-girdle muscular dystrophy, autosomal dominant type 1C	CAV3	701	601253	2, 4, 8, 9
Limb-girdle muscular dystrophy, autosomal dominant type 1E	DNAJB6	1285	611332	2, 4, 17
Limb-girdle muscular dystrophy, autosomal recessice type 2F	SGCD	265	601411	2, 4, 8, 9, 17
Limb-girdle muscular dystrophy, autosomal recessive type 2A	CAPN3	713	114240	2, 4, 8, 9, 17
Limb-girdle muscular dystrophy, autosomal recessive type 2B	DYSF	714	603009	2, 4, 8, 9, 17
Limb-girdle muscular dystrophy, autosomal recessive type 2C	SGCG	715	608896	2, 4, 8, 9
Limb-girdle muscular dystrophy, autosomal recessive type 2D	SGCA	716	600119	2, 4, 8, 9, 17
Limb-girdle muscular dystrophy, autosomal recessive type 2E	SGCB	717	600900	2, 4, 8, 9
Limb-Girdle Muscular Dystrophy, autosomal recessive type 2G	TCAP	289	604488	2, 4, 8, 9
Limb-girdle muscular dystrophy, autosomal recessive type 2H	TRIM32	307	602290	2, 4, 17
Limb-girdle muscular dystrophy, autosomal recessive type 2I	FKRP	718	606596	2, 4, 8, 9
Limb-girdle muscular dystrophy, autosomal recessive type 2J	TTN	309	188840	2, 17
Limb-girdle muscular dystrophy, autosomal recessive type 2K	POMT1	223	607423	2, 4, 8, 9, 17
Limb-girdle muscular dystrophy, autosomal recessive type 2L	ANO5	719	608662	2, 4, 8, 9, 17
Limb-girdle muscular dystrophy, autosomal recessive type 2M	FKTN	709	607440	2, 4, 8, 9, 17
Limb-girdle muscular dystrophy, autosomal recessive type 2N	POMT1	223	607423	2, 4, 8, 9, 17
Limb-girdle muscular dystrophy, autosomal recessive type 2S	TRAPPC11	2505	614138	2, 4, 17
Limb-girdle muscular dystrophy, autosomal recessive type 12C	POMK	2639	615247	2, 4
Lissencephaly type 4 with microcephaly	NDE1	1117	609449	2, 4, 17
Lissencephaly/Subcortical laminal heteropia, X-linked	DCX	813	300121	2, 4, 8, 9, 17

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Lowe oculocerebrorenal syndrome	OCRL	203	300535	2, 3, 4, 8, 9, 17
Major affective disorder 7	XBP1	3817	194355	17
Malignant hyperthermia type 5	CACNA1S	710	114208	2, 4, 8, 9, 17
Mandibulofacial dysostosis with microcephaly	EFTUD2	1103	603892	2, 3, 4, 8, 9, 17
Marden-Walker syndrome	PIEZO2	2397	613629	2, 3, 4, 8, 9, 17
MASA syndrome	L1CAM	153	308840	2, 3, 4, 8, 9
McLeod syndrome with or without chronic granulomatous disease	XK	2208	314850	2, 3, 4, 8, 9
Megalencephalic leukoencephalopathy with subcortical cysts	MLC1	655	605908	2, 4, 8, 9, 17
Megalencephalic leukoencephalopathy with subcortical cysts 2A	HEPACAM	1089	611642	2, 3, 4, 8, 9, 17
MELAS syndrome, MT-TL1 related	MT-TL1	1317	590050	4
Melnick-Needles syndrome	FLNA	803	300017	2, 3, 4, 8, 9, 17
Mental retardation and microcephaly with pontine and cerebellar hypoplasia	CASK	804	300172	2, 4, 8, 9, 17
Mental retardation non-syndromic	ELK1	833	311040	2, 4, 17
Mental retardation non-syndromic	KLF8	834	300286	2, 4, 17
Mental retardation non-syndromic	NXF5	835	300319	2, 4, 17
Mental retardation non-syndromic	ZCCHC12	836	300701	2, 4
Mental retardation with Cerebellar ataxia and dysequilibrium syndrome type 2	WDR81	2195	614218	2, 4, 17
Mental retardation with hypotonic facies syndrome, X-linked	ATRX	27	300032	2, 4, 8, 9, 17
Mental retardation with language impairment and autistic features	FOXP1	656	605515	17
Mental retardation X-linked, SMARCA1 related	SMARCA1	2259	300012	2, 4, 17
Mental retardation X-linked, syndromic, Claes-Jensen type	KDM5C	852	314690	2, 3, 4, 8, 9, 17
Mental retardation X-linked, syndromic, Lubs type	MECP2	163	300005	2, 4, 8, 9
Mental retardation, anterior maxillary protrusion, and strabismus	SOBP	3806	613667	17
Mental retardation, autosomal dominant type 1	MBD5	2204	611472	2, 4
Mental retardation, autosomal dominant type 2	DOCK8	1782	611432	2, 4, 8, 9, 17
Mental retardation, autosomal dominant type 3	CDH15	3283	114019	17
Mental retardation, autosomal dominant type 5	SYNGAP1	1932	603384	2, 4, 17
Mental retardation, autosomal dominant type 6	GRIN2B	1600	138252	2, 4, 8, 9, 17
Mental retardation, autosomal dominant type 7	DYRK1A	2634	600855	2, 3, 4, 8, 9, 17
Mental retardation, autosomal dominant type 8	GRIN1	2365	138249	2, 4, 17
Mental retardation, autosomal dominant type 9	KIF1A	150	601255	2, 3, 4, 8, 9, 17
Mental retardation, autosomal dominant type 11	EPB41L1	3777	602879	17
Mental retardation, autosomal dominant type 12	ARID1B	1554	614556	2, 4, 8, 9, 17
Mental retardation, autosomal dominant type 13	DYNC1H1	1279	600112	2, 17
Mental retardation, autosomal dominant type 13	TRAPPC9	1918	611966	2, 4, 17
Mental retardation, autosomal dominant type 14	ARID1A	1508	603024	2, 4, 8, 9, 17
Mental retardation, autosomal dominant type 15	SMARCB1	1555	601607	2, 4, 8, 9, 17
Mental retardation, autosomal dominant type 16	SMARCA4	1556	603254	2, 3, 4, 8, 9, 17
Mental retardation, autosomal dominant type 17	PACS1	2879	607492	2, 4
Mental retardation, autosomal dominant type 18	GATAD2B	2479	614998	2, 4
Mental retardation, autosomal dominant type 19	CTNNB1	1826	116806	2, 3, 4, 8, 9, 17
Mental retardation, autosomal dominant type 20	MEF2C	1603	600662	2, 4, 8, 9, 17
Mental retardation, autosomal dominant type 23	SETD5	2903	615743	2, 4, 17
Mental retardation, autosomal dominant type 24	DEAF1	2759	602635	2, 4
Mental retardation, autosomal dominant type 25	AHDC1	2342	615790	2, 4
Mental retardation, autosomal dominant type 27	SOX11	2823	600898	2, 4
Mental retardation, autosomal dominant type 28	ADNP	2400	611386	2, 4
Mental retardation, autosomal dominant type 31	PURA	2415	600473	2, 4
Mental retardation, autosomal dominant type 32	KAT6A	2813	601408	2, 4, 17
Mental retardation, autosomal dominant type 37	POGZ	2791	614787	2, 4, 17
Mental retardation, autosomal dominant type 38	EEF1A2	2514	602959	2, 4, 17
Mental retardation, autosomal recessive type 1	PRSS12	3220	606709	17
Mental retardation, autosomal recessive type 2	CRBN	2609	609262	2, 4, 17
Mental retardation, autosomal recessive type 3	CC2D1A	1914	610055	2, 4, 17
Mental retardation, autosomal recessive type 5	NSUN2	1119	610916	2, 3, 4, 8, 9, 17
Mental retardation, autosomal recessive type 7	TUSC3	2395	601385	2, 3, 4, 8, 9, 17
Mental retardation, autosomal recessive type 12	ST3GAL3	2676	606494	2, 4, 17
Mental retardation, autosomal recessive type 14	TECR	2243	610057	2, 4, 17
Mental retardation, autosomal recessive type 15	MAN1B1	2192	604346	2, 4, 17
Mental retardation, autosomal recessive type 18	MED23	1919	605042	2, 4, 17
Mental retardation, autosomal recessive type 27	LINS1	2825	610350	2, 4, 17
Mental retardation, autosomal recessive type 36	ADAT3	2600	615302	2, 4
Mental retardation, autosomal recessive type 37	ANK3	2339	600465	2, 17
Mental retardation, autosomal recessive type 38	HERC2	2758	605837	2, 17

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Mental retardation, autosomal recessive type 39	TTI2	1940	614426	2, 4, 17
Mental retardation, autosomal recessive type 40	TAF2	3449	604912	17
Mental retardation, autosomal recessive type 41	KPTN	2341	615620	2, 4
Mental retardation, autosomal recessive type 42	PGAP1	2362	611655	2, 4, 17
Mental retardation, autosomal recessive type 46	NDST1	2289	600853	2, 4, 17
Mental retardation, autosomal recessive type 49	GPT2	2734	138210	2, 4, 17
Mental retardation, autosomal recessive type 55	PUS3	3300	616283	2, 4
Mental retardation, X-linked	RAB40AL	1240	300405	2, 4
Mental retardation, X-linked syndromic, Christianson type	SLC9A6	825	300231	2, 4, 17
Mental retardation, X-linked syndromic, Nascimento-type	UBE2A	826	312180	2, 4
Mental retardation, X-linked syndromic, Raymond type	ZDHHC9	827	300646	2, 3, 4, 8, 9, 17
Mental retardation, X-linked syndromic, Turner type	HUWE1	853	300697	2, 17
Mental retardation, X-linked type 1	IQSEC2	2604	300522	2, 3, 4, 8, 9, 17
Mental retardation, X-linked type 3	HCFC1	2168	300019	2, 4, 17
Mental retardation, X-Linked type 13	MECP2	163	300005	2, 4, 8, 9
Mental retardation, X-linked type 14	UPF3B	314	300298	2, 3, 4, 8, 9, 17
Mental retardation, X-linked type 15	CUL4B	838	300304	2, 4, 17
Mental retardation, X-linked type 16	FGD1	315	300546	2, 4, 8, 9, 17
Mental retardation, X-linked type 17	HSD17B10	134	300256	2, 4
Mental retardation, X-linked type 19	RPS6KA3	247	300075	2, 4, 8, 9, 17
Mental retardation, X-linked type 21	IL1RAPL1	837	300206	2, 3, 4, 8, 9
Mental retardation, X-linked type 29	ARX	24	300382	2, 4, 8, 9, 17
Mental retardation, X-linked type 30	PAK3	816	300142	2, 4, 8, 9, 17
Mental retardation, X-linked type 32	CLIC2	1618	300138	2, 4
Mental retardation, X-linked type 41	GDI1	840	300104	2, 4, 17
Mental retardation, X-linked type 44	FTSJ1	841	300499	2, 4, 17
Mental retardation, X-linked type 45	ZNF81	817	314998	2, 4, 17
Mental retardation, X-linked type 46	ARHGEF6	842	300267	2, 4
Mental retardation, X-linked type 58	TSPAN7	818	300096	2, 4, 8, 9
Mental retardation, X-linked type 59	AP1S2	843	300629	2, 4
Mental retardation, X-linked type 63	ACSL4	844	300157	2, 4, 17
Mental retardation, X-linked type 72	RAB39B	819	300774	2, 4
Mental retardation, X-linked type 88, AGTR2 related	AGTR2	845	300034	2, 4
Mental retardation, X-linked type 89	ZNF41	820	314995	2, 4, 17
Mental retardation, X-linked type 90	DLG3	846	300189	2, 3, 4, 8, 9, 17
Mental retardation, X-linked type 91	ZDHHC15	821	300576	2, 3, 4, 8, 9, 17
Mental retardation, X-linked type 92	ZNF674	822	300573	2, 4, 17
Mental retardation, X-linked type 93	BRWD3	847	300553	2, 3, 4, 8, 9, 17
Mental retardation, X-linked type 94	GRIA3	848	305915	2, 4, 17
Mental retardation, X-linked type 95	MAGT1	160	300715	2, 4, 17
Mental retardation, X-linked type 96	SYP	823	313475	2, 4
Mental retardation, X-linked type 97	ZNF711	824	314990	2, 4, 17
Mental retardation, X-linked type 99	USP9X	2808	300072	2, 17
Mental retardation, X-linked type 101	MID2	2215	300204	2, 3, 4, 8, 9
Mental retardation, X-linked type 102	DDX3X	2940	300160	2, 4, 17
Mental retardation, X-linked with epilepsy	ATP6AP2	849	300556	2, 4, 17
Mental retardation, X-linked, associated with fragile site FRAXE	AFF2	850	300806	2, 4, 8, 9, 17
Mental retardation, x-linked, EFHC2 related	EFHC2	2284	300817	2, 4, 17
Mental retardation, X-linked, nonsyndromic	NEXMIF	851	300524	2, 4, 17
Mental retardation, X-linked, Siderius type	PHF8	815	300560	2, 4, 17
Mental retardation, X-linked, Snyder-Robinson type	SMS	2300	300105	2, 4, 17
Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	OPHN1	828	300127	2, 3, 4, 8, 9
Mental retardation, X-linked, with isolated growth hormone deficiency	SOX3	280	313430	2, 4, 8, 9
MERRF syndrome, MT-TK related	MT-TK	657	590060	4
MERRF syndrome, MT-TK related	MT-TK	657	590060	4
MERRF syndrome, MT-TK related	MT-TK	657	590060	4
MERRF syndrome, MT-TP related	MT-TP	658	590075	4
MERRF syndrome, MT-TP related	MT-TP	658	590075	4
MERRF syndrome, MT-TP related	MT-TP	658	590075	4
MERRF/MELAS overlap syndrome, MT-TS1 related	MT-TS1	1319	590080	4
MERRF/MELAS overlap syndrome, MT-TS2 related	MT-TS2	1320	590085	4
Metachromatic leukodystrophy due to Saposin B deficiency	PSAP	232	176801	2, 3, 4, 8, 9, 17
Methionine adenosyltransferase deficiency, autosomal recessive	MAT1A	2486	610550	2, 4, 17
Microcephaly-capillary malformation syndrome	STAMBP	1630	606247	2, 4, 17

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Minicore myopathy with external ophthalmoplegia	RYR1	249	180901	2, 17
Mirror movements type 1	DCC	2611	120470	2, 3, 4, 8, 9, 17
Mirror movements type 2	RAD51	1280	179617	2, 3, 4, 8, 9
Mirror movements type 3	DNAL4	2610	610565	2, 3, 4, 8, 9
Mitochondrial complex I deficiency	FOXRED1	1430	613622	2, 4, 17
Mitochondrial complex I deficiency	MT-ND1	1314	516000	4
Mitochondrial complex I deficiency	MT-ND2	1735	516001	4
Mitochondrial complex I deficiency	MT-ND3	646	516002	4
Mitochondrial complex I deficiency	MT-ND4L	1737	516004	4
Mitochondrial complex I deficiency	MT-ND4	1736	516003	4
Mitochondrial complex I deficiency	MT-ND5	1315	516005	4
Mitochondrial complex I deficiency	MT-ND6	1316	516006	4
Mitochondrial complex I deficiency	NDUFA1	831	300078	2, 4
Mitochondrial complex I deficiency	NDUFA11	1377	612638	2, 4
Mitochondrial complex I deficiency	NDUFAF1	41	606934	2, 4
Mitochondrial complex I deficiency	NDUFAF3	42	612911	2, 4
Mitochondrial complex I deficiency	NDUFAF4	1380	611776	2, 4
Mitochondrial complex I deficiency	NDUFAF5	979	612360	2, 3, 4, 8, 9, 17
Mitochondrial complex I deficiency	NDUFB3	1080	603839	2, 4
Mitochondrial complex I deficiency	NDUFB3	1080	603839	2, 4
Mitochondrial complex I deficiency	NDUFS1	1381	157655	2, 3, 4, 8, 9, 17
Mitochondrial complex I deficiency	NDUFS2	1382	602985	2, 4, 17
Mitochondrial complex I deficiency	NDUFS4	1384	602694	2, 4
Mitochondrial complex I deficiency	NDUFS6	1386	603848	2, 4
Mitochondrial complex I deficiency	NDUFV1	1389	161015	2, 4, 17
Mitochondrial complex I deficiency	NDUFV2	1390	600532	2, 3, 4, 8, 9
Mitochondrial complex I deficiency, MT-TN related	MT-TN	2559	590010	4
Mitochondrial complex II deficiency	SDHAF1	1426	612848	2, 4, 8, 9
Mitochondrial complex II deficiency	SDHD	1190	602690	2, 4, 8, 9
Mitochondrial complex III deficiency	BCS1L	1423	603647	2, 3, 4, 8, 9, 17
Mitochondrial complex III deficiency	UQCRB	47	191330	2, 4
Mitochondrial complex III deficiency	UQCRC2	1531	191329	2, 3, 4, 8, 9, 17
Mitochondrial complex III deficiency	UQCRQ	48	612080	2, 4
Mitochondrial complex III deficiency, nuclear type 2	TTC19	1411	613814	2, 3, 4, 8, 9, 17
Mitochondrial complex III deficiency, nuclear type 7	UQCC2	2873	614461	2, 4
Mitochondrial complex IV deficiency	COA8	3152	616003	2, 4
Mitochondrial complex IV deficiency	COX6B1	1369	124089	2, 4
Mitochondrial complex IV deficiency	FASTKD2	1372	612322	2, 4, 17
Mitochondrial complex IV deficiency	MT-CO3	1739	516050	4
Mitochondrial complex IV deficiency	PET100	2862	614770	2, 4
Mitochondrial complex V (ATP synthase) deficiency	MT-ATP6	1472	516060	4
Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1	ATPAF2	1354	608918	2, 4, 17
Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	TMEM70	1409	612418	2, 3, 4, 8, 9
Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3	ATP5F1E	980	606153	2, 4
Mitochondrial complex V deficiency, nuclear type 4	ATP5F1A	2681	164360	2, 4, 17
Mitochondrial Disorders, AKAP1 related	AKAP1	2476	602449	2, 3, 4, 8, 9, 17
Mitochondrial DNA depletion syndrome	DGUOK	721	601465	2, 3, 4, 8, 9
Mitochondrial DNA depletion syndrome	SUCLA2	284	603921	2, 4, 8, 9, 17
Mitochondrial DNA depletion syndrome	TK2	299	188250	2, 4, 8, 9, 17
Mitochondrial DNA depletion syndrome	TK2	299	188250	2, 4, 8, 9, 17
Mitochondrial DNA depletion syndrome 8B, MNGIE type	RRM2B	248	604712	2, 4, 8, 9, 17
Mitochondrial DNA depletion syndrome type 4A	POLG	220	174763	2, 4, 8, 9, 17
Mitochondrial DNA depletion syndrome type 4B	POLG	220	174763	2, 4, 8, 9
Mitochondrial DNA depletion syndrome type 6	MPV17	794	137960	2, 4, 8, 9
Mitochondrial DNA depletion syndrome type 7	TWNK	549	606075	2, 4, 8, 9, 17
Mitochondrial DNA depletion syndrome type 11	MGME1	2234	615076	2, 4
Mitochondrial DNA depletion syndrome type 13	FBXL4	2233	605654	2, 4, 17
Mitochondrial DNA depletion syndrome, encephalomyopathic type with methylmalonic aciduria	SUCLG1	1427	611224	2, 4, 8, 9, 17
Mitochondrial encephalomyopathy	MFF	1966	614785	2, 4
Mitochondrial encephalomyopathy	MT-CYB	1522	516020	4
Mitochondrial myopathy and sideroblastic anemia type 1	PUS1	44	608109	2, 4
Mitochondrial myopathy and sideroblastic anemia type 1	PUS1	44	608109	2, 4
Mitochondrial neurogastrointestinal encephalopathy syndrome without leukoencephalopathy	POLG	220	174763	2, 4, 8, 9

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Mitochondrial neurogastrointestinal encephalopathy syndrome without leukoencephalopathy	TYMP	795	131222	2, 3, 4, 8, 9, 17
Mitochondrial respiratory chain complex II deficiency	SDHA	1188	600857	2, 3, 4, 8, 9, 17
Mitochondrial respiratory chain disease, TIMM21 related	TIMM21	2028	615180	2, 4
Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	ECHS1	2624	602292	2, 3, 4, 8, 9, 17
Miyoshi muscular dystrophy type 3	ANO5	719	608662	2, 4, 8, 9, 17
Miyoshi myopathy	DYSF	714	603009	2, 4, 8, 9, 17
Multiple congenital anomalies-hypotonia-seizures syndrome type 3	PIGT	2338	610272	2, 4, 17
Multiple mitochondrial dysfunctions syndrome type 1	NFU1	2312	608100	2, 4, 17
Multiple mitochondrial dysfunctions syndrome type 2	BOLA3	2313	613183	2, 4
Multiple mitochondrial dysfunctions syndrome type 3	IBA57	2314	615316	2, 4
Multiple mitochondrial dysfunctions syndrome type 4	ISCA2	2794	615317	2, 4
Multisystemic smooth muscle dysfunction syndrome	ACTA2	6	102620	2, 3, 4, 8, 9, 17
Muscle hypertrophy	MSTN	723	601788	2, 4
Muscle-eye-brain disease, POMK related	POMK	2639	615247	2, 4
Muscular dystrophy type 1A	LAMA2	724	156225	2, 4, 8, 9, 17
Muscular dystrophy type 1C	FKRP	718	606596	2, 4, 8, 9
Muscular dystrophy type 1D	LARGE1	156	603590	2, 4, 8, 9, 17
Muscular dystrophy, Becker type	DMD	726	300377	2, 4, 8
Muscular dystrophy, congenital, LMNA related	LMNA	158	150330	2, 4, 8, 9, 17
Muscular dystrophy, congenital, megaconial type	CHKB	2668	612395	2, 4, 17
Muscular dystrophy, Duchenne type	DMD	726	300377	2, 4, 8
Muscular dystrophy, limb-girdle type 2A	CAPN3	713	114240	2, 4, 8, 9, 17
Muscular dystrophy, limb-girdle, type 2Q	PLEC	705	601282	2, 17
Muscular dystrophy, oculopharyngeal	PABPN1	728	602279	2, 3, 4, 8, 9, 10
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A2	POMT2	760	607439	2, 4, 8, 9, 17
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A3	POMGNT1	1598	606822	2, 4, 8, 9, 17
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A8	POMGNT2	2799	614828	2, 4
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A10	RXYLT1	2716	605862	2, 4
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A13	B4GAT1	2201	605517	2, 4
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B1	POMT1	223	607423	2, 4, 8, 9, 17
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B2	POMT2	760	607439	2, 4, 8, 9, 17
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B3	POMGNT1	1598	606822	2, 4, 8, 9, 17
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C2	POMT2	760	607439	2, 4, 8, 9, 17
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C3	POMGNT1	1598	606822	2, 4, 8, 9, 17
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C9	DAG1	1597	128239	2, 4, 17
Muscular-skeletal disorder, CAPN1 related	CAPN1	2358	114220	2, 4
Myasthenia congenital with tubular aggregates 1	GFPT1	1547	138292	2, 4, 17
Myasthenic syndrome associated with acetylcholine receptor deficiency	MUSK	62	601296	2, 3, 4, 8, 9, 17
Myasthenic syndrome due to mutation in SCN4A	SCN4A	256	603967	2, 4, 8, 9, 17
Myasthenic syndrome, congenital	AGRN	1546	103320	2, 3, 4, 8, 9, 17
Myasthenic syndrome, congenital	CHAT	731	118490	2, 3, 4, 8, 9, 17
Myasthenic syndrome, congenital	CHRNB1	732	100710	2, 3, 4, 8, 9, 17
Myasthenic syndrome, congenital	CHRNE	730	100725	2, 3, 4, 8, 9, 17
Myasthenic syndrome, congenital, fast channel	CHRNA1	60	100690	2, 3, 4, 8, 9, 17
Myasthenic syndrome, congenital, slow-channel	CHRNA1	60	100690	2, 3, 4, 8, 9, 17
Myasthenic syndrome, congenital, type 3A, slow channel	CHRND	61	100720	2, 3, 4, 8, 9, 17
Myasthenic syndrome, congenital, type 3B, fast-channel	CHRND	61	100720	2, 3, 4, 8, 9, 17
Myasthenic syndrome, congenital, type 3C, associated with acetylcholine receptor deficiency	CHRND	61	100720	2, 3, 4, 8, 9, 17
Myasthenic syndrome, congenital, type 10	DOK7	1227	610285	2, 3, 4, 8, 9, 17
Myasthenic syndrome, congenital, type 11, associated with acetylcholine receptor deficiency	RAPSN	1228	601592	2, 3, 4, 8, 9
Myasthenic syndrome, congenital, type 20, presynaptic	SLC5A7	3406	608761	17
Myoclonic dystonia, DRD2 related	DRD2	2276	126450	2, 3, 4, 8, 9
Myoclonic epilepsy of Lafora	EPM2A	663	607566	2, 4, 8, 9
Myoclonic epilepsy of Lafora	NHLRC1	184	608072	2, 4, 8, 9
Myoclonus, familial cortical	NOL3	2548	605235	2, 4
Myoglobinuria acute recurrent	LPIN1	562	605518	2, 3, 4, 8, 9, 17
Myopathy due to Integrin 7A deficiency	ITGA7	733	600536	2, 4, 17
Myopathy due to myoadenylate deaminase deficiency	AMPD1	467	102770	2, 3, 4, 8, 9, 17
Myopathy with extrapyramidal signs	MICU1	3280	605084	17
Myopathy with fiber-type disproportion	ACTA1	5	102610	2, 3, 4, 8, 9
Myopathy with fiber-type disproportion	SELENON	263	606210	2, 3, 4, 8, 9, 17
Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	MEGF10	2744	612453	2, 3, 4, 8, 9, 17
Myopathy, COL6A6 related	COL6A6	2380		2, 4, 17
Myopathy, desmin related, associated with mutation in the CRYAB gene	CRYAB	690	123590	2, 4

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Myopathy, distal type 1	MYH7	176	160760	2, 4, 8, 9, 17
Myopathy, distal type 4	FLNC	708	102565	2, 4, 17
Myopathy, distal with anterior tibial onset	DYSF	714	603009	2, 4, 8, 9, 17
Myopathy, distal, Tateyama type	CAV3	701	601253	2, 4, 8, 9
Myopathy, early-onset with fatal cardiomyopathy	TTN	309	188840	2, 17
Myopathy, lactic acidosis, and sideroblastic anemia type 2	YARS2	49	610957	2, 4, 17
Myopathy, limb girdle with bone fragility	MTAP	975	156540	2, 4, 8, 9, 17
Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	GFER	1405	600924	2, 4
Myopathy, MT-TQ related	MT-TQ	1322	590030	4
Myopathy, myofibrillar type 6	BAG3	30	603883	2, 4, 8, 9, 17
Myopathy, myofibrillar, Desmin related	DES	741	125660	2, 3, 4, 8, 9, 17
Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related	CRYAB	690	123590	2, 4
Myopathy, myofibrillar, ZASP related	LDB3	157	605906	2, 4, 17
Myopathy, scapulohumeroperoneal	ACTA1	5	102610	2, 3, 4, 8, 9
Myopathy, tubular aggregate, type 1	STIM1	2391	605921	2, 3, 4, 8, 9, 17
Myosclerosis, autosomal recessive	COL6A2	698	120240	2, 3, 4, 8, 9, 17
Myosin storage myopathy	MYH7	176	160760	2, 4, 8, 9, 17
Myotilinopathy	MYOT	711	604103	2, 4, 8, 9, 17
Myotonia congenita	CLCN1	744	118425	2, 4, 8, 9, 17
Myotonic dystrophy type 1	DMPK	593	605377	10, 17
Myotonic dystrophy type 1	DMPK	593	605377	10, 17
Myotonic dystrophy type 2	CNBP	329	116955	10
Myotonic dystrophy type 2	CNBP	329	116955	10
Myotubular myopathy X-linked	MTM1	172	300415	2, 4, 8, 9, 17
Narcolepsy	HCRT	1894	602358	2, 4
Nemaline myopathy type 1	TPM3	746	191030	2, 3, 4, 8, 9, 17
Nemaline myopathy type 2, autosomal recessive	NEB	2574	161650	2, 3, 8, 17
Nemaline myopathy type 3	ACTA1	5	102610	2, 3, 4, 8, 9
Nemaline myopathy type 4	TPM2	306	190990	2, 3, 4, 8, 9, 17
Nemaline myopathy type 5	TNNT1	747	191041	2, 3, 4, 8, 9, 17
Nemaline myopathy type 6	KBTBD13	1166	613727	2, 4
Nemaline myopathy type 7	CFL2	748	601443	2, 4
Neonatal death due Leigh syndrome, MT-TV related	MT-TV	2555	590105	4
Neurodegeneration due to cerebral folate transport deficiency	FOLR1	1589	136430	2, 3, 4, 8, 9
Neurodegeneration with brain iron accululation type 5	WDR45	1889	300526	2, 3, 4, 8, 9, 17
Neurodegeneration with brain iron accumulation type 4	C19orf12	1593	614297	2, 3, 4, 8, 9
Neurodegeneration with brain iron accumulation type 6	COASY	2325	609855	2, 4, 17
Neurodegeneration with brain iron accumulation, GTPBP2 related	GTPBP2	2964	607434	2, 4
Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	BRAT1	3307	614506	17
Neurodevelopmental disorder, ADAM22 related	ADAM22	2059	603709	2, 4, 8, 9
Neurodevelopmental disorder, APC2-related	APC2	2010	612034	2, 3, 4, 8, 9
Neurodevelopmental disorder, CNTNAP4 related	CNTNAP4	2022	610518	2, 4, 17
Neurodevelopmental disorder, CROCC related	CROCC	1922	615776	2, 4
Neurodevelopmental disorder, FRMPD4 related	FRMPD4	2402	300838	2, 4, 17
Neurodevelopmental disorder, KCTD3 related	KCTD3	2344	613272	2, 4
Neurodevelopmental disorder, MACF1 related	MACF1	2023	608271	2, 17
Neurodevelopmental disorder, MTOR related	MTOR	2153	601231	2, 17
Neurodevelopmental disorder, NGEF related	NGEF	2136	605991	2, 4
Neurodevelopmental disorder, PIGQ related	PIGQ	2620	605754	2, 4
Neurodevelopmental disorder, TUBB related	TUBB	2068	191130	2, 4
Neurodevelopmental disorder, ZNF311 related	ZNF311	2057		2, 4
Neurodevelopmental malformation and microcephaly	KIF2A	1628	602591	2, 4, 17
Neurodevelopmental malformation and microcephaly	KIF5C	1629	604593	2, 4, 17
Neurodevelopmental malformation and microcephaly	TUBG1	1627	191135	2, 4, 17
Neurogenic scapuloperoneal syndrome, Kaeser type	DES	741	125660	2, 3, 4, 8, 9, 17
Neuromyotonia and axonal neuropathy, autosomal recessive	HINT1	1615	601314	2, 3, 4, 8, 9
Neuronal migration disorder	CTNNA2	1610	114025	2, 4, 17
Neuronal migration disorder	EOMES	1587	604615	2, 4, 17
Neuronal migration disorder	SPTBN5	2072	605916	2, 17
Neuronal migration disorder	SRGAP2	1588	606524	2, 4
Neuronopathy distal hereditary motor type 2A	HSPB8	136	608014	2, 4, 8, 9
Neuronopathy distal hereditary motor type 2B	HSPB1	135	602195	2, 4, 8, 9
Neuronopathy distal hereditary motor type 5	GARS1	518	600287	2, 4, 8, 9, 17
Neuronopathy distal hereditary motor type 6	IGHMBP2	540	600502	2, 4, 8, 9, 17

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Neuronopathy distal hereditary motor type 7B	DCTN1	541	601143	2, 3, 4, 8, 9, 17
Neuropathy sensor type 1E	DNMT1	878	126375	2, 3, 4, 8, 9, 17
Neuropathy with liability to pressure palsies [HNPP]	PMP22	219	601097	2, 4, 8, 9
Neuropathy with sensory ataxic, dysarthria, and ophthalmoparesis	POLG	220	174763	2, 4, 8, 9
Neuropathy, distal hereditary motor, type 5A	BSCL2	440	606158	2, 3, 4, 8, 9, 17
Neuropathy, hereditary sensory and autonomic type 1C	SPTLC2	3451	605713	17
Neuropathy, hereditary sensory and autonomic type 2	SCN9A	258	603415	2, 3, 4, 8, 9, 17
Neuropathy, hereditary sensory and autonomic type 6	DST	1263	113810	2, 17
Neuropathy, hereditary sensory, type 1D	ATL1	490	606439	2, 4, 8, 9, 17
Neuropathy, hereditary sensory, type 1F	ATL3	3767	609369	17
Neuropathy, hereditary sensory, with spastic paraplegia	CCT5	2588	610150	2, 4, 17
Neuropathy, hereditary sensory, with spastic paraplegia	CCT5	2588	610150	2, 4, 17
Neutral lipid storage disease with myopathy	PNPLA2	750	609059	2, 4, 17
Niemann-Pick disease type C1	NPC1	189	607623	2, 4, 8, 9, 13, 17
Nonaka myopathy	GNE	129	603824	2, 3, 4, 8, 9
Norrie disease	NDP	319	300658	2, 3, 4, 8, 9
Occipital horn syndrome	ATP7A	25	300011	2, 4, 8, 9, 17
Opitz G syndrome	MID1	854	300552	2, 3, 4, 8, 9, 17
Oral-facial-digital syndrome type 1	OFD1	204	300170	2, 3, 4, 8, 9, 17
Pantothenate kinase-associated neurodegeneration	PANK2	665	606157	2, 4, 8, 9, 17
Paramyotonia congenita of von Eulenburg	SCN4A	256	603967	2, 4, 8, 9, 17
Parietal foramina type 2	ALX4	666	605420	2, 4, 8, 9
PARK1 Parkinson	SNCA	563	163890	2, 4, 8, 9
PARK2 Parkinson	PRKN	564	602544	2, 4, 8, 9, 17
PARK4 Parkinson	SNCA	563	163890	2, 4, 8, 9
PARK5 Parkinson	UCHL1	1797	191342	2, 4, 8, 9
PARK6 Parkinson	PINK1	565	608309	2, 4, 8, 9, 17
PARK7 Parkinson	PARK7	566	602533	2, 4, 8, 9
PARK8 Parkinson	LRRK2	567	609007	2, 4, 8, 9, 17
PARK9 Parkinson	ATP13A2	568	610513	2, 3, 4, 8, 9
PARK13 Parkinson	HTRA2	569	606441	2, 3, 4, 8, 9, 17
PARK14 Parkinson	PLA2G6	570	603604	2, 3, 4, 8, 9, 17
PARK15 Parkinson	FBXO7	571	605648	2, 3, 4, 8, 9, 17
PARK17 Parkinson	VPS35	572	601501	2, 3, 4, 8, 9, 17
PARK19 Parkinson, juvenile-onset	DNAJC6	1912	608375	2, 3, 4, 8, 9, 17
PARK20 Parkinson	SYNJ1	1795	604297	2, 3, 4, 8, 9, 17
PARK21 Parkinson	DNAJC13	2622	614334	2, 4, 17
Parkinson disease, late-onset, susceptibility to	GBA	399	606463	2, 3, 4, 8, 9
Parkinson disease, susceptibility to, MT-TT related	MT-TT	2565	590090	4
Parkinsonism with spasticity, X-linked	ATP6AP2	849	300556	2, 4, 17
Parkinsonism-Dystonia, infantile	SLC6A3	1281	126455	2, 3, 4, 8, 9, 17
Paroxysmal exercise-induced dyskinesia with epilepsy and/or hemolytic anemia	SLC2A1	193	138140	2, 4, 8, 9, 17
Paroxysmal nonkinesigenic dyskinesia	PNKD	557	609023	2, 3, 4, 8, 9, 17
Partington syndrome	ARX	24	300382	2, 4, 8, 9, 17
Pelizaeus-Merzbacher disease	PLP1	217	300401	2, 4, 8, 9, 17
Pelizaeus-Merzbacher disease	SLC16A2	268	300095	2, 3, 4, 8, 9
Peripheral demyelinating neuropathy Waardenburg syndrome and Hirschsprung disease	SOX10	279	602229	2, 3, 4, 8, 9
Periventricular heterotopia with microcephaly	ARFGEF2	1283	605371	2, 3, 4, 8, 9, 17
Peroxisome biogenesis disorder 14B	PEX11B	939	603867	2, 4, 8, 9
Peroxisome biogenesis disorder type 1B	PEX1	678	602136	2, 3, 4, 8, 9, 17
Peroxisome biogenesis disorder type 2A	PEX5	681	600414	2, 3, 4, 8, 9, 17
Peroxisome biogenesis disorder type 2B	PEX5	681	600414	2, 3, 4, 8, 9, 17
Peroxisome biogenesis disorder type 10A	PEX3	680	603164	2, 4, 17
Perrault syndrome	HSD17B4	1095	601860	2, 3, 4, 8, 9, 17
Perrault syndrome type 2	HARS2	3782	600783	17
Perrault syndrome type 4	LARS2	3633	604544	17
Perrault syndrome type 5	TWNK	549	606075	2, 4, 8, 9
Phosphoglycerate kinase 1 deficiency	PGK1	855	311800	2, 4, 17
Pick disease	PSEN1	233	104311	2, 4, 8, 9, 17
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	TREM2	671	605086	2, 3, 4, 8, 9
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	TYROBP	670	604142	2, 3, 4, 8, 9
Polyglucosan body myopathy type 1 with or without immunodeficiency	RBCK1	2329	610924	2, 4, 17
Polymicrogyria bilateral occipital	NR2E1	631	603849	2, 4
Pompe disease	GAA	405	606800	2, 4, 8, 9, 12, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Pontocerebellar hypoplasia type 1A	VRK1	672	602168	2, 3, 4, 8, 9, 17
Pontocerebellar hypoplasia type 1B	EXOSC3	1006	606489	2, 3, 4, 8, 9
Pontocerebellar hypoplasia type 2A	TSEN54	1734	608755	2, 3, 4, 8, 9, 17
Pontocerebellar hypoplasia type 2B	TSEN2	1990	608753	2, 4, 17
Pontocerebellar hypoplasia type 2C	TSEN34	1991	608754	2, 4
Pontocerebellar hypoplasia type 2D	SEPSECS	2630	613009	2, 4, 17
Pontocerebellar hypoplasia type 2E	VPS53	2722	615850	2, 4, 17
Pontocerebellar hypoplasia type 4	TSEN54	1734	608755	2, 3, 4, 8, 9, 17
Pontocerebellar hypoplasia type 5	TSEN54	1734	608755	2, 3, 4, 8, 9, 17
Pontocerebellar hypoplasia type 6	RARS2	1436	611524	2, 4, 17
Pontocerebellar hypoplasia type 8	CHMP1A	1992	164010	2, 4
Pontocerebellar hypoplasia, type 9	AMPD2	2584	102771	2, 4, 17
Pontocerebellar hypoplasia, type 10	CLP1	2631	608757	2, 4
Porencephaly type 2	COL4A2	1431	120090	2, 3, 4, 8, 9, 17
Potassium-aggravated myotonia	SCN4A	256	603967	2, 4, 8, 9, 17
Prader-Willi syndrome	chr. 15q11	91		8
Prader-Willi syndrome	NDN	1752	602117	2, 4, 8, 9
Prader-Willi syndrome	SNRPN	1753	182279	2, 4, 8, 9
Primary lateral sclerosis, juvenile	ALS2	692	606352	2, 3, 4, 8, 9, 17
Progressive external ophthalmoplegia with mitochondrial deletions type 1, autosomal dominant	POLG	220	174763	2, 4, 8, 9
Progressive external ophthalmoplegia with mitochondrial deletions type 2, autosomal dominant	SLC25A4	271	103220	2, 4, 8, 9
Progressive external ophthalmoplegia with mitochondrial deletions type 2, autosomal recessive	RNASEH1	3798	604123	17
Progressive external ophthalmoplegia with mitochondrial deletions type 2, autosomal recessive	RNASEH1	3798	604123	17
Progressive external ophthalmoplegia with mitochondrial deletions type 3, autosomal dominant	TWNK	549	606075	2, 4, 8, 9
Progressive external ophthalmoplegia with mitochondrial deletions type 3, autosomal dominant	TWNK	549	606075	2, 4, 8, 9
Progressive external ophthalmoplegia with mitochondrial deletions type 4, autosomal dominant	POLG2	221	604983	2, 4, 8, 9, 17
Progressive external ophthalmoplegia with mitochondrial deletions type 5, autosomal dominant	RRM2B	248	604712	2, 4, 8, 9, 17
Progressive external ophthalmoplegia with mitochondrial deletions type 5, autosomal dominant	RRM2B	248	604712	2, 4, 8, 9, 17
Progressive external ophthalmoplegia with mitochondrial deletions type 5, autosomal dominant	RRM2B	248	604712	2, 4, 8, 9, 17
Progressive myoclonus epilepsy type 1A	PRICKLE1	792	608500	2, 3, 4, 8, 9, 17
Progressive myoclonus epilepsy type 3	KCTD7	791	611725	2, 3, 4, 8, 9
Progressive myoclonus epilepsy type 6	GOSR2	2547	604027	2, 4, 17
Progressive myoclonus epilepsy type 8	CERS1	2546	606919	2, 4, 17
Psychomotor retardation	TANC1	1909	611397	2, 4, 17
Ptosis, congenital	ZFHX4	2804	606940	2, 4, 17
Pyridoxine-dependent epilepsy	ALDH7A1	10	107323	2, 3, 4, 8, 9, 17
Pyruvate carboxylase deficiency	PC	519	608786	2, 3, 4, 8, 9, 17
Raynaud-Claes syndrome	CLCN4	3390	302910	17
Renpenning syndrome	PQBP1	856	300463	2, 4, 8, 9
Rett syndrome preserved speech variant	MECP2	163	300005	2, 4, 8, 9
Rigid spine muscular dystrophy	SELENON	263	606210	2, 3, 4, 8, 9, 17
Rippling muscle disease	CAV3	701	601253	2, 4, 8, 9
Rolandic epilepsy, mental retardation, and speech dyspraxia	SRPX2	857	300642	2, 4, 17
Roussy-Levy syndrome	PMP22	219	601097	2, 4, 8, 9
Salih ataxia	RUBCN	3025	613516	2, 4, 17
Scapuloperoneal myopathy, MYH7 related	MYH7	176	160760	2, 4, 8, 9, 17
Schizophrenia, CALR related	CALR	2205	109091	2, 4
Schizophrenia, CELSR2 related	CELSR2	2002	604265	2, 4
Schizophrenia, GRID2 related	GRID2	2302	602368	2, 3, 4, 8, 9, 17
Schizophrenia, NOTCH4 related	NOTCH4	2121	164951	2, 4, 17
Schwartz-Jampel syndrome type 1	HSPG2	137	142461	2, 17
Segawa syndrome, autosomal recessive	TH	3	191290	2, 3, 4, 8, 9
Seizures, benign familial infantile, type 2	PRRT2	558	614386	2, 3, 4, 8, 9
Seizures, benign neonatal, type 1	KCNQ2	149	602235	2, 4, 8, 9, 17
Seizures, benign neonatal, type 2	KCNQ3	199	602232	2, 4, 8, 9, 17
SESAME syndrome	KCNJ10	832	602208	2, 4, 8, 9
Sialuria, finish type	SLC17A5	269	604322	2, 3, 4, 8, 9, 17

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Simpson-Golabi-Behmel syndrome type 1	GPC3	858	300037	2, 4, 8, 9, 17
Sjogren-Larsson syndrome	ALDH3A2	674	609523	2, 3, 4, 8, 9, 17
Slowed nerve conduction velocity, autosomanal dominant	ARHGEF10	550	608136	2, 4, 17
Smith-Magenis syndrome	RAI1	859	607642	2, 4, 8, 9, 17
Smith-Magenis syndrome, ULK2 related	ULK2	2320	608650	2, 4, 17
Spastic ataxia Charlevoix-Saguenay type	SACS	594	604490	2, 4, 8, 9, 17
Spastic ataxia type 1, autosomal dominant	VAMP1	1729	185880	2, 3, 4, 8, 9
Spastic ataxia type 2, autosomal recessive	KIF1C	2379	603060	2, 4, 17
Spastic ataxia type 3, autosomal recessive	MARS2	2678	609728	2, 4
Spastic ataxia type 4, autosomal recessive	MTPAP	3790	613669	17
Spastic ataxia type 5, autosomal recessive	AFG3L2	611	604581	2, 3, 4, 8, 9, 17
Spastic paralysis, infantile onset ascending	ALS2	692	606352	2, 3, 4, 8, 9, 17
Spastic paraplegia type 74, autosomal recessive	IBA57	2314	615316	2, 4
Spastic paraplegia type 74, autosomal recessive	IBA57	2314	615316	2, 4
SPG1	L1CAM	153	308840	2, 3, 4, 8, 9
SPG2	PLP1	217	300401	2, 4, 8, 9, 17
SPG3A	ATL1	490	606439	2, 4, 8, 9, 17
SPG4	SPAST	491	604277	2, 4, 8, 9, 17
SPG5A	CYP7B1	350	603711	2, 3, 4, 8, 9
SPG6	NIPA1	492	608145	2, 4, 8, 9
SPG7	SPG7	501	602783	2, 3, 4, 8, 9, 17
SPG8	WASHC5	493	610657	2, 3, 4, 8, 9, 17
SPG10	KIF5A	494	602821	2, 3, 4, 8, 9, 17
SPG11	SPG11	502	610844	2, 4, 8, 9, 17
SPG12	RTN2	1278	603183	2, 3, 4, 8, 9, 17
SPG13	HSPD1	495	118190	2, 3, 4, 8, 9, 17
SPG15	ZFYVE26	503	612012	2, 3, 4, 8, 9, 17
SPG17	BSCL2	440	606158	2, 3, 4, 8, 9, 17
SPG18	ERLIN2	2590	611605	2, 4, 17
SPG20	SPART	504	607111	2, 3, 4, 8, 9, 17
SPG21	SPG21	505	608181	2, 3, 4, 8, 9, 17
SPG26	B4GALNT1	2587	601873	2, 4, 17
SPG28	DDHD1	2151	614603	2, 4, 17
SPG30	KIF1A	150	601255	2, 3, 4, 8, 9, 17
SPG31	REEP1	497	609139	2, 4, 8, 9, 17
SPG33	ZFYVE27	498	610243	2, 3, 4, 8, 9, 17
SPG35	FA2H	196	611026	2, 3, 4, 8, 9
SPG39	PNPLA6	507	603197	2, 3, 4, 8, 9, 17
SPG42	SLC33A1	499	603690	2, 3, 4, 8, 9
SPG43	C19orf12	1593	614297	2, 3, 4, 8, 9
SPG44	GJC2	508	608803	2, 4, 8, 9
SPG45	NT5C2	2573	600417	2, 4, 17
SPG47	AP4B1	1803	607245	2, 4, 17
SPG48	AP5Z1	2322	613653	2, 4, 17
SPG49	TECPR2	2592	615000	2, 4, 17
SPG50	AP4M1	1069	602296	2, 4, 17
SPG51	AP4E1	2585	607244	2, 4, 17
SPG52	AP4S1	2586	607243	2, 4
SPG53	VPS37A	2018	609927	2, 4, 17
SPG54	DDHD2	2589	615003	2, 4, 17
SPG55	C12ORF65	1365	613541	2, 4
SPG56	CYP2U1	1728	610670	2, 3, 4, 8, 9, 17
SPG57	TFG	2593	602498	2, 4, 17
SPG59, USP8 related	USP8	2597	603158	2, 4, 17
SPG60, WDR48 related	WDR48	2598	612167	2, 4, 17
SPG61	ARL6IP1	2572	607669	2, 4
SPG62, ERLIN1 related	ERLIN1	2595	611604	2, 4, 17
SPG63	AMPD2	2584	102771	2, 4, 17
SPG64	ENTPD1	1908	601752	2, 4, 17
SPG66, ARSI related	ARSI	2594	610009	2, 4, 17
SPG68, FLRT1 related	FLRT1	2596	604806	2, 4, 17
SPG71, ZFR related	ZFR	2599	615635	2, 4, 17
SPG72	REEP2	2591	609347	2, 4, 17
SPG72	REEP2	2591	609347	2, 4, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
SPG73	CPT1C	2780	608846	2, 4, 17
Spheroid body myopathy	MYOT	711	604103	2, 4, 8, 9, 17
Spinal and bulbar muscular atrophy X-linked	AR	20	313700	2, 4, 8, 9, 10, 17
Spinal muscular atrophy distal, autosomal recessive type 4	PLEKHG5	538	611101	2, 3, 4, 8, 9, 17
Spinal muscular atrophy type 1	SMN1	754	600354	2, 4, 8, 9
Spinal muscular atrophy type 2	SMN1	754	600354	2, 4, 8, 9
Spinal muscular atrophy type 3	SMN1	754	600354	2, 4, 8, 9
Spinal muscular atrophy type 4	SMN1	754	600354	2, 4, 8, 9
Spinal muscular atrophy type 5	DNAJB2	736	604139	2, 3, 4, 8, 9, 17
Spinal muscular atrophy with progressive myoclonic epilepsy	ASAH1	391	613468	2, 3, 4, 8, 9, 17
Spinal muscular atrophy, distal, X-linked	ATP7A	25	300011	2, 4, 8, 9, 17
Spinal muscular atrophy, lower extremity, autosomal dominant, type 2	BICD2	2014	609797	2, 4, 17
Spinal muscular atrophy, lower extremity-predominant type 1, autosomal dominant	DYNC1H1	1279	600112	2, 17
Spinocerebellar ataxia type 1, autosomal dominant	ATXN1	595	601556	10
Spinocerebellar ataxia type 1, X-linked	ATP2B3	2652	300014	2, 4, 17
Spinocerebellar ataxia type 2, autosomal dominant	ATXN2	596	601517	10
Spinocerebellar ataxia type 3, autosomal dominant	ATXN3	597	607047	10
Spinocerebellar ataxia type 4, autosomal dominant	PLEKHG4	1804	609526	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia type 5, autosomal dominant	SPTBN2	598	604985	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia type 6, autosomal dominant	CACNA1A	583	601011	2, 4, 8, 9, 10, 17
Spinocerebellar ataxia type 7, autosomal dominant	ATXN7	600	607640	10
Spinocerebellar ataxia type 7, autosomal recessive	TPP1	1613	607998	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia type 8, autosomal dominant	ATXN8OS	601	613289	10
Spinocerebellar ataxia type 8, autosomal recessive	SYNE1	286	608441	2, 17
Spinocerebellar ataxia type 9, autosomal rececssive	COQ8A	575	606980	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia type 10, autosomal dominant	ATXN10	602	611150	10
Spinocerebellar ataxia type 10, autosomal recessive	ANO10	1806	613726	2, 4, 17
Spinocerebellar ataxia type 11, autosomal dominant	TTBK2	603	611695	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia type 12, autosomal dominant	PPP2R2B	604	604325	10
Spinocerebellar ataxia type 12, autosomal recessive	WWOX	2472	605131	2, 4, 8, 9, 17
Spinocerebellar ataxia type 13, autosomal dominant	KCNC3	605	176264	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia type 13, autosomal recessive	GRM1	1612	604473	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia type 14, autosomal dominant	PRKCG	606	176980	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia type 15	ITPR1	607	147265	2, 17
Spinocerebellar ataxia type 17, autosomal dominant	TBP	288	600075	10
Spinocerebellar ataxia type 17, autosomal recessive	CWF19L1	2892	616120	2, 4, 17
Spinocerebellar ataxia type 18, autosomal dominant	IFRD1	608	603502	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia type 18, autosomal recessive	GRID2	2302	602368	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia type 21, autosomal dominant	TMEM240	2533	616101	2, 4
Spinocerebellar ataxia type 22, autosomal dominant	KCND3	1611	605411	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia type 23, autosomal dominant	PDYN	609	131340	2, 4, 8, 9
Spinocerebellar ataxia type 26, autosomal dominant	EEF2	2297	130610	2, 4, 17
Spinocerebellar ataxia type 27, autosomal dominant	FGF14	610	601515	2, 3, 4, 8, 9
Spinocerebellar ataxia type 28, autosomal dominant	AFG3L2	611	604581	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia type 29, congenital nonprogressive	ITPR1	607	147265	2, 17
Spinocerebellar ataxia type 31, autosomal dominant	BEAN1	612	612051	10
Spinocerebellar ataxia type 35, autosomal dominant	TGM6	1857	613900	2, 4, 17
Spinocerebellar ataxia type 36, autosomal dominant	NOP56	1856	614154	10
Spinocerebellar ataxia with axonal neuropathy, autosomal recessive	TDP1	290	607198	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia with axonal neuropathy, autosomal recessive	TDP1	290	607198	2, 3, 4, 8, 9, 17
Spinocerebellar ataxia, infantile-onset	TWNK	549	606075	2, 4, 8, 9, 17
Stocco dos Santos X-linked mental retardation syndrome	SHROOM4	860	300579	2, 4, 17
Striatal degeneration	PDE8B	623	603390	2, 4, 17
Thiamine metabolism dysfunction syndrome 4 progressive polyneuropathy type	SLC25A19	487	606521	2, 4
Thyrotoxic periodic paralysis type 1	CACNA1S	710	114208	2, 4, 8, 9, 17
Thyrotoxic periodic paralysis type 2	KCNJ18	2089	613236	2, 3, 4, 8, 9
Tibial muscular dystrophy, tardive	TTN	309	188840	2, 17
Tourette syndrome	SLITRK1	675	609678	2, 4, 8, 9
Tremor essential type 4	FUS	38	137070	2, 3, 4, 8, 9, 17
Tremor, sssential type 1, hereditary	DRD3	3094	126451	17
Tuberous sclerosis	TSC1	676	605284	2, 4, 8, 9, 17
Tuberous sclerosis type 2	TSC2	677	191092	2, 4, 8, 9, 17
Ullrich congenital muscular dystrophy	COL6A1	697	120220	2, 3, 4, 8, 9, 17
Ullrich congenital muscular dystrophy	COL6A2	698	120240	2, 3, 4, 8, 9, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Ullrich congenital muscular dystrophy type 1	COL6A3	699	120250	2, 3, 4, 8, 9, 17
Ullrich congenital muscular dystrophy type 2	COL12A1	1757	120320	2, 17
Unverricht-Lundborg disease	CSTB	793	601145	2, 3, 4, 8, 9, 10
Urocanase deficiency	UROC1	2343	613012	2, 4, 17
Ventriculomegaly with cystic kidney disease	CRB2	2727	609720	2, 4
Vitamin E familial deficiency	TTPA	614	600415	2, 3, 4, 8, 9
Waardenburg syndrome type 2D	SNAI2	2103	602150	2, 4, 8, 9
Waardenburg syndrome/Hirschsprung disease	EDNRB	194	131244	2, 4, 8, 9, 17
Walker-Warburg syndrome	CRPPA	521	614631	2, 3, 4, 8, 9, 17
Walker-Warburg syndrome	FKTN	709	607440	2, 4, 8, 9, 17
Walker-Warburg syndrome or muscle-eye-brain disease, FKRP related	FKRP	718	606596	2, 4, 8, 9
Warburg micro syndrome type 1	RAB3GAP1	237	602536	2, 3, 4, 8, 9, 17
WDR27-related brain disorders	WDR27	2166		2, 4
Wieacker-Wolff syndrome	ZC4H2	2735	300897	2, 4
Wilson-Turner syndrome	LAS1L	2752	300964	17
Zellweger syndrome	PEX1	678	602136	2, 3, 4, 8, 9, 17
Zellweger syndrome	PEX2	679	170993	2, 4
Zellweger syndrome	PEX6	682	601498	2, 3, 4, 8, 9, 17
Zellweger syndrome	PEX10	683	602859	2, 4
Zellweger syndrome	PEX12	684	601758	2, 3, 4, 8, 9
Zellweger syndrome	PEX13	685	601789	2, 3, 4, 8, 9
Zellweger syndrome	PEX14	686	601791	2, 4, 17
Zellweger syndrome	PEX16	687	603360	2, 3, 4, 8, 9, 17
Zellweger syndrome	PEX19	688	600279	2, 3, 4, 8, 9
Zellweger syndrome	PEX26	689	608666	2, 4

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› Ophthalmology - Panels

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoDx®	Targets exonic regions of >6500 genes associated with known clinical phenotypes		50158	5
CentoLCV	Genome wide analysis of structural variants and large copy number changes through sWGS		50118	5
CentoVision	ABCA4, ABCB6, ABHD12, ACO2, ACVR1, ADAM9, ADGRV1, AFG3L2, AGK, AHI1, AIPL1, ALDH18A1, ALDH1A3, AP3B1, APTX, ARL13B, ARL6, ASB10, ATF6, AUH, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BFSP1, BFSP2, BLOC1S3, BLOC1S6, BMP4, C12orf65, C1QTNF5, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CANT1, CC2D2A, CCDC28B, CDH23, CDH3, CDHR1, CEP164, CEP290, CEP41, CERKL, CHD7, CHM, CHMP4B, CIB2, CISD2, CLCN7, CLN3, CLN5, CLN6, CLN8, CLPB, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL2A1, COL4A1, COL9A1, COL9A2, COL9A3, COX7B, CPLANE1, CRB1, CRX, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CSPP1, CTC1, CTDP1, CTNNB1, CTSD, CYP1B1, CYP27A1, DGUOK, DHDDS, DKC1, DNA2, DNAJC19, DNM1L, DTNBP1, EDN3, EDNRB, EFEMP1, ELOVL4, ENPP1, EPHA2, ERCC1, ERCC2, ERCC5, ERCC6, EYA1, EYS, FAM126A, FAM161A, FLVCR1, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FSCN2, FTL, FYCO1, FZD4, GALE, GALK1, GALT, GBA, GCNT2, GDF3, GDF6, GJA1, GJA3, GJA8, GNAT2, GNPTG, GPR143, GRIP1, GRN, GUCA1A, GUCA1B, GUCY2D, HCCS, HESX1, HEXA, HGSNAT, HK1, HMX1, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, HTRA2, IDH3B, IFT140, IFT172, IFT27, IMPDH1, IMPG2, INPP5E, IQCB1, KCNJ13, KCNV2, KIF11, KIF7, KIT, KLHL7, LCA5, LEMD2, LEP, LEPR, LIM2, LMX1B, LOXL1, LRAT, LRMDA, LRP2, LRP5, LSS, LTBP2, LYST, LZTFL1, MAB21L2, MAF, MAK, MC1R, MERTK, MFN2, MFRP, MFSD8, MIP, MITF, MKKS, MKS1, MLPH, MYH9, MYO5A, MYO7A, MYOC, NAA10, NDP, NF2, NHS, NMNAT1, NPHP1, NPHP3, NPHP4, NR0B2, NR2F1, NRL, NTF4, OCA2, OCRL, OFD1, OPA1, OPA3, OPN1LW, OPN1MW, OPTN, OSTM1, OTX2, P3H2, PAX2, PAX3, PAX6, PCARE, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX2, PEX7, PHF6, PHYH, PIK3R5, PITPNM3, PITX2, PITX3, PLA2G5, PNKP, PNPLA6, POLG, POLG2, POMC, POMGNT1, PPARG, PPT1, PQBP1, PRCD, PRKCG, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, PRPS1, PRSS56, RAB18, RAB27A, RAB3GAP1, RAB3GAP2, RARB, RAX, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RRM2B, RS1, RTN4IP1, SAG, SBF2, SDCCAG8, SEMA4A, SERAC1, SETX, SHH, SIL1, SIX3, SIX6, SLC16A12, SLC24A5, SLC25A4, SLC25A46, SLC33A1, SLC45A2, SLC9A6, SMCHD1, SMOC1, SNAI2, SNRNP200, SNX10, SOX10, SOX2, SPATA7, SPG7, STRA6, TBC1D20, TBK1, TCIRG1, TCTN1, TCTN2, TCTN3, TDRD7, TENM3, TFAP2A, TIMM8A, TIMP3, TK2, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TNFRSF11A, TNFSF11, TOPORS, TPP1, TRIM32, TRNT1, TSPAN12, TTC21B, TTC8, TTPA, TULP1, TWNK, TYMP, TYR, TYRP1, UNC45B, USH1C, USH1G, USH2A, VCAN, VIM, VPS13B, VSX2, WDPCP, WDR19, WDR36, WFS1, WHRN, ZIC2, ZNF423, ZNF513		5432	6

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Disease	Gene	Test code	OMIM Gene	Available test methods
Achromatopsia type 2	CNGA3	50	600053	2, 4, 17
Achromatopsia type 3	CNGB3	940	605080	2, 3, 4, 8, 9, 17
Achromatopsia type 4	GNAT2	51	139340	2, 4, 17
Achromatopsia type 6	PDE6H	1604	601190	2, 4
Aland Island eye disease	CACNA1F	861	300110	2, 4, 17
Albinism, ocular type I, Nettleship-Falls type	GPR143	131	300808	2, 4, 8, 9, 17
Albinism, oculocutaneous nonsyndromic	SLC24A5	1631	609802	2, 4, 17
Albinism, oculocutaneous type 1A	TYR	311	606933	2, 4, 8, 9
Albinism, oculocutaneous type 1B	TYR	311	606933	2, 4, 8, 9
Albinism, oculocutaneous type 2	OCA2	991	611409	2, 4, 8, 9, 17
Albinism, oculocutaneous type 3	TYRP1	862	115501	2, 3, 4, 8, 9
Albinism, oculocutaneous type 4	SLC45A2	863	606202	2, 3, 4, 8, 9
Albinism, oculocutaneous type 5	LRMDA	1538	614537	2, 4
Aniridia	PAX6	207	607108	2, 4, 8, 9, 17
Anterior segment mesenchymal dysgenesis	PITX3	2571	602669	2, 4
Bardet-Biedl syndrome type 14	CEP290	642	610142	2, 3, 4, 8, 9
Bestrophinopathy	BEST1	865	607854	2, 4, 8, 9
Bietti crystalline corneoretinal dystrophy	CYP4V2	2901	608614	2, 4, 8, 9
Blepharophimosis, epicanthus inversus, and ptosis	FOXL2	1516	605597	2, 4, 8, 9
Blepharophimosis-ptosis-intellectual disability syndrome	UBE3B	1907	608047	2, 4, 17
Bothnia retinal dystrophy	RLBP1	866	180090	2, 3, 4, 8, 9
Bradyopsia	RGS9BP	1606	607814	2, 4
Bradyopsia	RGS9	1605	604067	2, 4, 17
Branchiootorenal syndrome type 1	EYA1	125	601653	2, 4, 8, 9, 17
Branchiootorenal syndrome type 2	SIX5	2060	600963	2, 3, 4, 8, 9, 17
Brittle cornea syndrome	ZNF469	867	612078	2, 4, 17
Cataract 11, multiple types	PITX3	2571	602669	2, 4
Cataract type 17, multiple types	CRYBB1	873	600929	2, 4
Cataract type 23	CRYBA4	2000	123631	2, 4
Cataract type 41	WFS1	325	606201	2, 4, 8, 9, 17
Cataract type 43	UNC45B	2774	611220	2, 4, 17
Cataract, autosomal dominant	GCNT2	868	600429	2, 4, 17
Cataract, autosomal recessive congenital nuclear type 2	CRYBB3	872	123630	2, 4
Cataract, autosomal recessive congenital type 1	CRYAA	869	123580	2, 4
Cataract, autosomal recessive congenital type 2	FYCO1	870	607182	2, 4, 17
Cataract, autosomal recessive congenital type 4	TDRD7	871	611258	2, 4, 17
Cataract, autosomal recessive type 38	AGK	1092	610345	2, 3, 4, 8, 9, 17
Cataract, congenital	SORD	2526	182500	2, 4
Cataract, congenital, associated with Marinesco-Sjogren Syndrome	SIL1	874	608005	2, 3, 4, 8, 9, 17
Cataract, cortical pulverulent, late-onset	LIM2	875	154045	2, 4
Cataract, lamellar	HSF4	876	602438	2, 4, 17
Cataract, posterior polar type 2	CRYAB	690	123590	2, 4
Cataract, pulverulent or cerulean, with or without microcornea	MAF	2222	177075	2, 4, 8, 9
Cataract, X-linked	NHS	185	300457	2, 3, 4, 8, 9, 17
Cataract-microcornea syndrome	GJA8	877	600897	2, 4, 8, 9
Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia	IARS2	3785	612801	17
Cerebellar-retinal degeneration, infantile	ACO2	2898	100850	2, 4, 17
Choroidal dystrophy, central areolar type 2	PRPH2	230	179605	2, 4, 8, 9
Coat plus syndrome	CTC1	1288	613129	2, 3, 4, 8, 9, 17
Coloboma of optic nerve	PAX6	207	607108	2, 4, 8, 9, 17
Coloboma, ocular, autosomal dominant	PAX6	207	607108	2, 4, 8, 9, 17
Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation	YAP1	3328	606608	17
Colobomatous microphthalmia	TENM1	522	300588	2, 17
Cone-rod dystrophy	AIPL1	1349	604392	2, 4, 8, 9
Cone-rod dystrophy	UNC119	1442	604011	2, 4
Cone-rod dystrophy type 2	CRX	1364	602225	2, 4, 8, 9
Cone-rod dystrophy type 3	ABCA4	2	601691	2, 3, 4, 8, 9, 17
Cone-rod dystrophy type 4	PDE6C	1398	600827	2, 4, 17
Cone-rod dystrophy type 5	PITPNM3	1400	608921	2, 4, 17
Cone-rod dystrophy type 7	RIMS1	1435	606629	2, 17

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Cone-rod dystrophy type 9	ADAM9	1351	602713	2, 4, 17
Cone-rod dystrophy type 11	RAX2	1419	610362	2, 4
Cone-rod dystrophy type 12	PROM1	926	604365	2, 3, 4, 8, 9, 17
Cone-rod dystrophy type 13	RPGRIP1	246	605446	2, 4, 8, 9, 17
Cone-rod dystrophy type 14	GUCA1A	192	600364	2, 4
Cone-rod dystrophy type 15	CDHR1	1371	609502	2, 4, 17
Cone-rod dystrophy type 17	CD3G	2818	186740	2, 4, 17
Cone-rod dystrophy type 20	POC1B	2730	614784	2, 4, 17
Cone-rod dystrophy, C21orf2 related	C21orf2	2480	603191	2, 4
Cone-rod dystrophy, X-linked type 3	CACNA1F	861	300110	2, 4, 17
Corneal dystrophy, epithelial basement membrane	TGFBI	1893	601692	2, 4, 17
Corneal dystrophy, posterior polymorphous, type 1	VSX1	2900	605020	2, 4
Corneal endothelial dystrophy type 2	SLC4A11	880	610206	2, 4, 17
Corneal intraepithelial dyskeratosis and ectodermal dysplasia	NLRP1	2116	606636	2, 3, 4, 8, 9, 17
Corneal opacification and other ocular anomalies	PXDN	2910	605158	2, 4, 17
Doyne honeycob retinal dystrophy	EFEMP1	881	601548	2, 3, 4, 8, 9, 17
Duane Retraction syndrome	CHN1	882	118423	2, 4, 17
Duane Retraction syndrome	SALL4	251	607343	2, 3, 4, 8, 9, 17
Dyschromatosis symmetrica hereditaria	ADAR	86	146920	2, 4, 17
Early onset glaucoma, phenotype modifier of, COL15A1 related	COL15A1	2650	120325	2, 17
Ectopia lentis et pupillae	ADAMTSL4	2163	610113	2, 3, 4, 8, 9, 17
Ectopia lentis, familial	FBN1	950	134797	2, 8, 17
Ectopia lentis, isolated, autosomal recessive	ADAMTSL4	2163	610113	2, 3, 4, 8, 9, 17
Exfoliation syndrome, susceptibility to	LOXL1	2569	153456	2, 4, 17
Exudative vitreoretinopathy	FZD4	1607	604579	2, 4, 8, 9
Exudative vitreoretinopathy type 2	NDP	319	300658	2, 3, 4, 8, 9
Exudative vitreoretinopathy type 5	TSPAN12	1608	613138	2, 4, 17
Fibrosis of extraocular muscles, congenital type 1	KIF21A	883	608283	2, 4, 17
Fibrosis of extraocular muscles, congenital type 3a	TUBB3	310	602661	2, 4
Fleck retina, familial benign	PLA2G5	1609	601192	2, 4
Foveal hypoplasia type 1	PAX6	207	607108	2, 4, 8, 9, 17
Fundus albipunctatus	PRPH2	230	179605	2, 4, 8, 9
Fundus albipunctatus	RDH5	1434	601617	2, 4
Gaze palsy, horizontal, with progressive scoliosis	ROBO3	2231	608630	2, 4, 17
Gillespie syndrome	ITPR1	607	147265	2, 17
Glaucoma, open angle type 1A	MYOC	884	601652	2, 4
Glaucoma, open angle type 1E	OPTN	205	602432	2, 3, 4, 8, 9, 17
Glaucoma, open angle type 1F	ASB10	2567	615054	2, 4, 17
Glaucoma, open angle type 1F	NTF4	2570	162662	2, 4
Glaucoma, open angle type 1G	WDR36	885	609669	2, 4, 17
Glaucoma, primary type 3A	CYP1B1	886	601771	2, 4, 8, 9, 17
Glaucoma, primary type 3D	LTBP2	887	602091	2, 4, 17
Gyrate atrophy of choroid and retina with or without ornithinemia	OAT	1986	613349	2, 3, 4, 8, 9, 17
Hereditary Retinoblastoma	RB1	1643	614041	2, 4, 8, 9, 17
Hermansky Pudlak syndrome type 4	HPS4	1416	606682	2, 4, 17
Hermansky-Pudlak syndrome type 1	HPS1	1414	604982	2, 4, 17
Hermansky-Pudlak syndrome type 2	AP3B1	1399	603401	2, 4, 17
Hermansky-Pudlak syndrome type 3	HPS3	1415	606118	2, 4, 17
Hermansky-Pudlak syndrome type 5	HPS5	1417	607521	2, 4, 17
Hermansky-Pudlak syndrome type 6	HPS6	1418	607522	2, 4, 17
Hermansky-Pudlak syndrome type 7	DTNBP1	1437	607145	2, 4, 17
Hermansky-Pudlak syndrome type 8	BLOC1S3	1357	609762	2, 4
Hyperferritinemia-cataract syndrome	FTL	1633	134790	2, 3, 4, 8, 9
Iridogoniodysgenesis type 1	FOXC1	888	601090	2, 4, 8, 9
Jalili syndrome	CNNM4	1634	607805	2, 4, 17
Keratoconus type 1	VSX1	2900	605020	2, 4
Leber congenital amaurosis type 1	GUCY2D	902	600179	2, 4, 8, 9, 17
Leber congenital amaurosis type 3	SPATA7	1422	609868	2, 4, 17
Leber congenital amaurosis type 5	LCA5	1440	611408	2, 4, 8, 9, 17
Leber congenital amaurosis type 6	RPGRIP1	246	605446	2, 4, 8, 9, 17
Leber congenital amaurosis type 7	CRX	1364	602225	2, 4, 8, 9
Leber congenital amaurosis type 8	CRB1	919	604210	2, 3, 4, 8, 9, 17
Leber congenital amaurosis type 9	NMNAT1	1731	608700	2, 3, 4, 8, 9
Leber congenital amaurosis type 10	CEP290	642	610142	2, 3, 4, 8, 9

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Leber congenital amaurosis type 11	IMPDH1	906	146690	2, 4, 8, 9, 17
Leber congenital amaurosis type 12	RD3	1429	180040	2, 4
Leber congenital amaurosis type 16	KCNJ13	1732	603208	2, 4
Leber congenital amaurosis type 17	GDF6	1997	601147	2, 4, 8, 9
Leber congenital amaurosis with myopathy	DTHD1	1954		2, 4
Leber optic atrophy	MT-ATP6	1472	516060	4
Leber optic atrophy	MT-CO1	1738	516030	4
Leber optic atrophy	MT-CO3	1739	516050	4
Leber optic atrophy	MT-CYB	1522	516020	4
Leber optic atrophy	MT-ND1	1314	516000	4
Leber optic atrophy	MT-ND2	1735	516001	4
Leber optic atrophy	MT-ND4L	1737	516004	4
Leber optic atrophy	MT-ND4	1736	516003	4
Leber optic atrophy	MT-ND5	1315	516005	4
Leber optic atrophy	MT-ND6	1316	516006	4
Macular degeneration, age-related type 3	FBLN5	2491	604580	2, 4, 17
Macular degeneration, age-related type 6	RAX2	1419	610362	2, 4
Macular degeneration, age-related type 8, association with	ARMS2	2907	611313	2, 4, 8, 9
Macular degeneration, age-related type 11	CST3	1871	604312	2, 3, 4, 8, 9
Macular degeneration, early-onset	FBN2	590	612570	2, 3, 8, 17
Macular dystrophy retinal type 2	PROM1	926	604365	2, 3, 4, 8, 9, 17
Macular dystrophy with central cone involvement	MFSD8	360	611124	2, 3, 4, 8, 9, 17
Macular dystrophy, BEST2-related	BEST2	2520	607335	2, 4
Macular dystrophy, BEST3-related	BEST3	2521	607337	2, 4
Macular dystrophy, BEST4-related	BEST4	2522	607336	2, 4
Macular dystrophy, vitelliform	BEST1	865	607854	2, 4, 8, 9, 17
Macular dystrophy, vitelliform	PRPH2	230	179605	2, 4, 8, 9
Mainzer Saldino syndrome	IFT140	1118	614620	2, 4, 17
Marshall syndrome	COL11A1	944	120280	2, 8, 17
Megalocornea, X-linked	CHRDL1	1289	300350	2, 3, 4, 8, 9
MELAS syndrome, MT-TL1 related	MT-TL1	1317	590050	4
Microphthalmia syndromic type 2	BCOR	829	300485	2, 3, 4, 8, 9, 17
Microphthalmia syndromic type 3	SOX2	893	184429	2, 4, 8, 9
Microphthalmia syndromic type 5	OTX2	894	600037	2, 3, 4, 8, 9
Microphthalmia syndromic type 6	BMP4	891	112262	2, 4, 8, 9
Microphthalmia syndromic type 6	SIX6	892	606326	2, 4
Microphthalmia syndromic type 7	HCCS	830	300056	2, 3, 4, 8, 9
Microphthalmia syndromic type 8	ALDH1A3	1525	600463	2, 3, 4, 8, 9, 17
Microphthalmia syndromic type 9	STRA6	758	610745	2, 3, 4, 8, 9, 17
Microphthalmia, isolated type 2	VSX2	324	142993	2, 3, 4, 8, 9
Microphthalmia, isolated type 3	RAX	890	601881	2, 4
Microphthalmia, isolated type 4	GDF6	1997	601147	2, 4, 8, 9
Microphthalmia, isolated type 5	MFRP	2350	606227	2, 4, 17
Microphthalmia, isolated type 6	PRSS56	2353	613858	2, 4, 17
Microphthalmia, isolated type 9	GDF3	1948	606522	2, 4, 8, 9
Microphthalmia, isolated with coloboma type 3	VSX2	324	142993	2, 3, 4, 8, 9
Microphthalmia, isolated with coloboma type 6, digenic	GDF3	1948	606522	2, 4, 8, 9
Microphthalmia, isolated with coloboma type 6, digenic	GDF6	1997	601147	2, 4, 8, 9
Microphthalmia, isolated with coloboma type 9	TENM3	1845	610083	2, 4, 17
Microphthalmia, syndromic type 1	NAA10	2351	300013	2, 4, 17
Microphthalmia, syndromic type 11	VAX1	2355	604294	2, 4
Microphthalmia, VAX2 related	VAX2	2356	604295	2, 4
Microspherophakia and/or megalocornea	LTBP2	887	602091	2, 4, 17
Nance-Horan syndrome	NHS	185	300457	2, 3, 4, 8, 9, 17
Nanophthalmia type 2	MFRP	2350	606227	2, 4, 17
Night blindness type 1, congenital stationary, autosomal dominant	RHO	243	180380	2, 4, 8, 9
Night blindness, congenital stationar type 1B	GRM6	896	604096	2, 4, 17
Night blindness, congenital stationar type 1C	TRPM1	2012	603576	2, 3, 4, 8, 9
Night blindness, congenital stationary type 1A	NYX	895	300278	2, 4
Night blindness, congenital stationary type 2A	CACNA1F	861	300110	2, 4, 17
Night blindness, congenital stationary type 2B	CABP4	898	608965	2, 3, 4, 8, 9
Night blindness, congenital stationary type 3	GNAT1	899	139330	2, 4
Night blindness, congenital stationary, autosomal dominant type 2	PDE6B	209	180072	2, 4, 17
Night blindness, congenital stationary, type 1E	GPR179	1290	614515	2, 4, 17

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Nonarteritic anterior ischemic optic neuropathy	GP1BA	67	606672	2, 4
Nystagmus type 1	FRMD7	83	300628	2, 4, 8, 9, 17
Nystagmus type 6	GPR143	131	300808	2, 4, 8, 9, 17
Occult macular dystrophy	RP1L1	1532	608581	2, 3, 4, 8, 9, 17
Oguchi disease	GRK1	900	180381	2, 4
Oguchi disease	SAG	250	181031	2, 4, 17
Ophthalmoplegia, isolated, MT-TN related	MT-TN	2559	590010	4
Optic atrophy type 1	OPA1	1401	605290	2, 4, 8, 9, 17
Optic atrophy type 3	OPA3	1402	606580	2, 4, 8, 9
Optic atrophy type 7	TMEM126A	901	612988	2, 4
Optic atrophy type 9	ACO2	2898	100850	2, 4, 17
Optic atrophy with or without deafness, ophthalmoplegia, myopathy, ataxia, and neuropathy	OPA1	1401	605290	2, 4, 8, 9, 17
Patterned dystrophy of retinal pigment epithelium	PRPH2	230	179605	2, 4, 8, 9
Peters Anomaly	CYP1B1	886	601771	2, 4, 8, 9
Peters anomaly	PAX6	207	607108	2, 4, 8, 9, 17
Peters Anomaly	PITX2	216	601542	2, 4, 8, 9
Peters-Plus syndrome	B3GLCT	29	610308	2, 3, 4, 8, 9, 17
Pigmented paravenous chorioretinal atrophy	CRB1	919	604210	2, 3, 4, 8, 9, 17
Plasminogen deficiency type 1	PLG	2335	173350	2, 4, 17
Progressive external ophthalmoplegia with mitochondrial deletions type 1, autosomal dominant	POLG	220	174763	2, 4, 8, 9
Progressive external ophthalmoplegia with mitochondrial deletions type 1, autosomal recessive	POLG	220	174763	2, 4, 8, 9
Progressive external ophthalmoplegia with mitochondrial deletions type 2, autosomal recessive	RNASEH1	3798	604123	17
Progressive external ophthalmoplegia with mitochondrial deletions type 3, autosomal dominant	TWNK	549	606075	2, 4, 8, 9
Progressive external ophthalmoplegia with mitochondrial deletions type 4, autosomal dominant	POLG2	221	604983	2, 4, 8, 9, 17
Progressive external ophthalmoplegia with mitochondrial deletions type 6, autosomal dominant	DNA2	2209	601810	2, 4, 17
Retinal cone dystrophy type 3B	KCNV2	905	607604	2, 4, 8, 9
Retinal cone dystrophy type 4	CACNA2D4	1370	608171	2, 4, 17
Retinal degeneration, late-onset, autosomal dominant	C1QTNF5	1637	608752	2, 4
Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities	ITM2B	2623	603904	2, 4
Retinal dystrophy, juvenile cataracts, and short stature syndrome	RDH11	3797	607849	17
Retinal nonattachment nonsyndromic congenital	ATOH7	968	609875	2, 4
Retinitis pigmentosa juvenile	LRAT	95	604863	2, 4
Retinitis pigmentosa SEMA4C related	SEMA4C	2301	604462	2, 4, 17
Retinitis pigmentosa type 1, autosomal dominant	RP1	245	603937	2, 4, 8, 9, 17
Retinitis pigmentosa type 2 X-linked	RP2	117	300757	2, 4, 8, 9
Retinitis pigmentosa type 3 X-linked	RPGR	1291	312610	2, 4, 8, 9, 17
Retinitis pigmentosa type 4, autosomal dominant/recessive	RHO	243	180380	2, 4, 8, 9
Retinitis pigmentosa type 7, autosomal dominant	PRPH2	230	179605	2, 4, 8, 9
Retinitis pigmentosa type 7	ROM1	118	180721	2, 4
Retinitis pigmentosa type 9, autosomal dominant	RP9	918	607331	2, 4
Retinitis pigmentosa type 10, autosomal dominant	IMPDH1	906	146690	2, 4, 8, 9, 17
Retinitis pigmentosa type 11, autosomal dominant	PRPF31	907	606419	2, 4, 8, 9, 17
Retinitis pigmentosa type 12, autosomal recessive	CRB1	919	604210	2, 3, 4, 8, 9, 17
Retinitis pigmentosa type 13, autosomal dominant	PRPF8	908	607300	2, 17
Retinitis pigmentosa type 14, autosomal recessive	TULP1	920	602280	2, 3, 4, 8, 9, 17
Retinitis pigmentosa type 17, autosomal dominant	CA4	909	114760	2, 4, 17
Retinitis pigmentosa type 18, autosomal dominant	PRPF3	910	607301	2, 4, 17
Retinitis pigmentosa type 19, autosomal recessive	ABCA4	2	601691	2, 3, 4, 8, 9, 17
Retinitis pigmentosa type 20, autosomal recessive	RPE65	921	180069	2, 3, 4, 8, 9, 17
Retinitis pigmentosa type 23 X-linked	OFD1	204	300170	2, 3, 4, 8, 9, 17
Retinitis pigmentosa type 25	EYS	922	612424	2, 8, 17
Retinitis pigmentosa type 26, autosomal recessive	CERKL	923	608381	2, 3, 4, 8, 9, 17
Retinitis pigmentosa type 27, autosomal dominant	NRL	911	162080	2, 4
Retinitis pigmentosa type 28, autosomal recessive	FAM161A	111	613596	2, 4, 17
Retinitis pigmentosa type 30, autosomal dominant	FSCN2	912	607643	2, 4, 17
Retinitis pigmentosa type 31, autosomal dominant	TOPORS	913	609507	2, 4, 17
Retinitis pigmentosa type 33, autosomal dominant	SNRNP200	914	601664	2, 4, 17
Retinitis pigmentosa type 35, autosomal dominant/recessive	SEMA4A	262	607292	2, 4, 17
Retinitis pigmentosa type 36, autosomal recessive	PRCD	924	610598	2, 4
Retinitis pigmentosa type 38, autosomal recessive	MERTK	925	604705	2, 3, 4, 8, 9, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Retinitis pigmentosa type 39	USH2A	321	608400	2, 8, 17
Retinitis pigmentosa type 40, autosomal recessive	PDE6B	209	180072	2, 4, 17
Retinitis pigmentosa type 41, autosomal recessive	PROM1	926	604365	2, 3, 4, 8, 9, 17
Retinitis pigmentosa type 42, autosomal dominant	KLHL7	915	611119	2, 4, 17
Retinitis pigmentosa type 43, autosomal recessive	PDE6A	927	180071	2, 4, 17
Retinitis pigmentosa type 44, autosomal dominant/recessive	RGR	242	600342	2, 4
Retinitis pigmentosa type 45, autosomal recessive	CNGB1	928	600724	2, 4, 17
Retinitis pigmentosa type 46, autosomal recessive	IDH3B	929	604526	2, 4, 17
Retinitis pigmentosa type 47, autosomal recessive	SAG	250	181031	2, 4, 17
Retinitis pigmentosa type 48, autosomal dominant	GUCA1B	916	602275	2, 4
Retinitis pigmentosa type 49, autosomal recessive	CNGA1	930	123825	2, 4, 17
Retinitis pigmentosa type 50, autosomal dominant	BEST1	865	607854	2, 4, 8, 9
Retinitis pigmentosa type 51, autosomal recessive	TTC8	308	608132	2, 4, 17
Retinitis pigmentosa type 53, autosomal recessive	RDH12	931	608830	2, 4, 8, 9
Retinitis pigmentosa type 54, autosomal recessive	PCARE	932	613425	2, 4, 17
Retinitis pigmentosa type 55, autosomal recessive	ARL6	22	608845	2, 4
Retinitis pigmentosa type 56, autosomal recessive	IMPG2	933	607056	2, 3, 4, 8, 9, 17
Retinitis pigmentosa type 57, autosomal recessive	PDE6G	934	180073	2, 4
Retinitis pigmentosa type 58, autosomal recessive	ZNF513	935	613598	2, 4
Retinitis pigmentosa type 59, autosomal recessive	DHDDS	1639	608172	2, 4, 17
Retinitis pigmentosa type 60	PRPF6	1638	613979	2, 4, 17
Retinitis pigmentosa type 61, autosomal recessive	CLRN1	936	606397	2, 3, 4, 8, 9
Retinitis pigmentosa type 62, autosomal recessive	MAK	1640	154235	2, 4, 17
Retinitis pigmentosa type 64, autosomal recessive	C8ORF37	1641	614477	2, 4
Retinitis pigmentosa type 66, autosomal recessive	RBP3	1642	180290	2, 4, 17
Retinitis pigmentosa type 74, autosomal recessive	BBS2	35	606151	2, 3, 4, 8, 9
Retinitis pigmentosa, juvenile, autosomal recessive	SPATA7	1422	609868	2, 4, 17
Retinitis punctata albescens	RHO	243	180380	2, 4, 8, 9
Retinoschisis	RS1	1644	300839	2, 3, 4, 8, 9
Revesz syndrome	TINF2	937	604319	2, 4, 17
Ring dermoid of cornea	PITX2	216	601542	2, 4, 8, 9
Senior-Loken syndrome type 5	IQCB1	1421	609237	2, 4, 17
Sorsby fundus dystrophy	TIMP3	1645	188826	2, 4
Stargardt Disease type 1	ABCA4	2	601691	2, 3, 4, 8, 9, 17
Stargardt Disease type 1	CNGB3	940	605080	2, 3, 4, 8, 9
Stargardt Disease type 3	ELOVL4	941	605512	2, 4
Stargardt Disease type 4	PROM1	926	604365	2, 3, 4, 8, 9, 17
Stickler syndrome type 1, nonsyndromic ocular	COL2A1	943	120140	2, 4, 8, 9, 17
Stickler syndrome type 1	COL2A1	943	120140	2, 4, 8, 9, 17
Stickler syndrome type 2	COL11A1	944	120280	2, 8, 17
Stickler syndrome type 3	COL11A2	945	120290	2, 4, 17
Stickler syndrome type 5	COL9A2	97	120260	2, 4, 17
Stickler syndrome, autosomal recessive	COL9A1	942	120210	2, 4, 17
Sveinsson choreoretinal atrophy	TEAD1	2530	189967	2, 4
Usher syndrome type 1D	CDH23	1646	605516	2, 17
Usher syndrome type 1D/F	PCDH15	110	605514	2, 4, 8, 9, 17
Usher syndrome type 1G	USH1G	2189	607696	2, 4
Usher syndrome type 1J	CIB2	87	605564	2, 4
Usher syndrome type 2C	ADGRV1	2191	602851	2, 17
Usher syndrome type 2C	PDZD7	2190	612971	2, 4, 17
Usher syndrome type 2D	WHRN	1654	607928	2, 4, 17
Usher syndrome type 3A	CLRN1	936	606397	2, 3, 4, 8, 9
Vitreoretinochoroidopathy	BEST1	865	607854	2, 4, 8, 9
Waardenburg syndrome/albinism	MITF	777	156845	2, 4, 8, 9, 17
Waardenburg syndrome/albinism	TYR	311	606933	2, 4, 8, 9
Wagner syndrome	VCAN	948	118661	2, 4, 17
Warburg micro syndrome 3	RAB18	1647	602207	2, 4
Warburg micro syndrome type 1	RAB3GAP1	237	602536	2, 3, 4, 8, 9, 17
Weill-Marchesani syndrome - AR	ADAMTS10	951	608990	2, 3, 4, 8, 9, 17
Weill-Marchesani syndrome, dominant type 2	FBN1	950	134797	2, 8, 17
Wolfram syndrome type 1	WFS1	325	606201	2, 4, 8, 9, 17
Wolfram syndrome type 2	CISD2	952	611507	2, 3, 4, 8, 9
Wolfram-like syndrome, autosomal dominant	WFS1	325	606201	2, 4, 8, 9, 17

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Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoDx®	Targets exonic regions of >6500 genes associated with known clinical phenotypes		50158	5
CentoHear	ABHD12, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ANKH, ATP2B2, ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BSND, BTD, CABP2, CACNA1D, CCDC50, CD151, CD164, CDC14A, CDH23, CDKN1C, CEACAM16, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCAF17, DCDC2, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNMT1, DSPP, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA1, EYA4, FDXR, FGF3, FGFR1, FGFR2, FGFR3, FOXI1, GAB1, GATA3, GIPC3, GJA1, GJB2, GJB3, GJB6, GPRASP2, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HARS1, HARS2, HGF, HOMER2, HOXB1, HSD17B4, ILDR1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIT, KITLG, LARS2, LHFPL5, LOXHD1, LRP2, LRTOMT, MAN2B1, MANBA, MARVELD2, MCM2, MET, MGP, MITF, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PEX1, PEX26, PEX6, PJVK, PMP22, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, RDX, RMND1, ROR1, RPS6KA3, S1PR2, SALL1, SALL4, SEMA3E, SERPINB6, SIX1, SIX5, SLC12A1, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLC44A4, SLC52A2, SLC52A3, SLITRK6, SMAD4, SMPX, SNAI2, SOX10, SOX2, SPATA5, STRC, SUCLA2, SUCLG1, SYNE4, TBC1D24, TBX1, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRMU, TSPEAR, TWNK, TYR, USH1C, USH1G, USH2A, VCAN, WBP2, WFS1, WHRN		5444	6
CentoHear	ABHD12, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ANKH, ATP2B2, ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BSND, BTD, CABP2, CACNA1D, CCDC50, CD151, CD164, CDC14A, CDH23, CDKN1C, CEACAM16, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCAF17, DCDC2, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNMT1, DSPP, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA1, EYA4, FDXR, FGF3, FGFR1, FGFR2, FGFR3, FOXI1, GAB1, GATA3, GIPC3, GJA1, GJB2, GJB3, GJB6, GPRASP2, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HARS1, HARS2, HGF, HOMER2, HOXB1, HSD17B4, ILDR1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIT, KITLG, LARS2, LHFPL5, LOXHD1, LRP2, LRTOMT, MAN2B1, MANBA, MARVELD2, MCM2, MET, MGP, MITF, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PEX1, PEX26, PEX6, PJVK, PMP22, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, RDX, RMND1, ROR1, RPS6KA3, S1PR2, SALL1, SALL4, SEMA3E, SERPINB6, SIX1, SIX5, SLC12A1, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLC44A4, SLC52A2, SLC52A3, SLITRK6, SMAD4, SMPX, SNAI2, SOX10, SOX2, SPATA5, STRC, SUCLA2, SUCLG1, SYNE4, TBC1D24, TBX1, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRMU, TSPEAR, TWNK, TYR, USH1C, USH1G, USH2A, VCAN, WBP2, WFS1, WHRN		5444	6

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Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoHear	ABHD12, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ANKH, ATP2B2, ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BSND, BTD, CABP2, CACNA1D, CCDC50, CD151, CD164, CDC14A, CDH23, CDKN1C, CEACAM16, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, CO-L4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCAF17, DCDC2, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNMT1, DSPP, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA1, EYA4, FDXR, FGF3, FGFR1, FGFR2, FGFR3, FOXI1, GAB1, GATA3, GIPC3, GJA1, GJB2, GJB3, GJB6, GPRASP2, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HARS1, HARS2, HGF, HOMER2, HOXB1, HSD17B4, ILDR1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIT, KITLG, LARS2, LHFPL5, LOXHD1, LRP2, LRTOMT, MAN2B1, MANBA, MARVELD2, MCM2, MET, MGP, MITF, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PEX1, PEX26, PEX6, PJVK, PMP22, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, RDX, RMND1, ROR1, RPS6KA3, S1PR2, SALL1, SALL4, SEMA3E, SERPINB6, SIX1, SIX5, SLC12A1, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLC44A4, SLC52A2, SLC52A3, SLITRK6, SMAD4, SMPX, SNAI2, SOX10, SOX2, SPATA5, STRC, SUCLA2, SUCLG1, SYNE4, TBC1D24, TBX1, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRMU, TSPEAR, TWNK, TYR, USH1C, USH1G, USH2A, VCAN, WBP2, WFS1, WHRN		5444	6
CentoHear	ABHD12, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ANKH, ATP2B2, ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BSND, BTD, CABP2, CACNA1D, CCDC50, CD151, CD164, CDC14A, CDH23, CDKN1C, CEACAM16, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, CO-L4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCAF17, DCDC2, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNMT1, DSPP, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA1, EYA4, FDXR, FGF3, FGFR1, FGFR2, FGFR3, FOXI1, GAB1, GATA3, GIPC3, GJA1, GJB2, GJB3, GJB6, GPRASP2, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HARS1, HARS2, HGF, HOMER2, HOXB1, HSD17B4, ILDR1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIT, KITLG, LARS2, LHFPL5, LOXHD1, LRP2, LRTOMT, MAN2B1, MANBA, MARVELD2, MCM2, MET, MGP, MITF, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PEX1, PEX26, PEX6, PJVK, PMP22, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, RDX, RMND1, ROR1, RPS6KA3, S1PR2, SALL1, SALL4, SEMA3E, SERPINB6, SIX1, SIX5, SLC12A1, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLC44A4, SLC52A2, SLC52A3, SLITRK6, SMAD4, SMPX, SNAI2, SOX10, SOX2, SPATA5, STRC, SUCLA2, SUCLG1, SYNE4, TBC1D24, TBX1, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRMU, TSPEAR, TWNK, TYR, USH1C, USH1G, USH2A, VCAN, WBP2, WFS1, WHRN		5444	6

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CentoLCV	Genome wide analysis of structural variants and large copy number changes through sWGS		50118	5

› Ear, Nose and Throat

Disease	Gene	Test code	OMIM Gene	Available test methods
Alport syndrome, autosomal recessive	COL4A3	983	120070	2, 3, 4, 8, 9, 17
Alport syndrome, autosomal recessive	COL4A4	984	120131	2, 3, 4, 8, 9, 17
Alport syndrome, X-Linked	COL4A5	985	303630	2, 4, 8, 9, 17
Auditory neuropathy, autosomal dominant	DIAPH3	1674	614567	2, 4, 17
Auriculocondylar syndrome type 2	PLCB4	1015	600810	2, 3, 4, 8, 9, 17
Branchiootic syndrome type 1	EYA1	125	601653	2, 4, 8, 9, 17
Brown-Vialetto-Van Laere syndrome 1	SLC52A3	1941	613350	2, 3, 4, 8, 9
Brown-Vialetto-Van Laere syndrome type 2	SLC52A2	3021	607882	2, 4
Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia	IARS2	3785	612801	17
Ciliogenesis related disorder	PTPN23	1973	606584	2, 4
Deafness and male infertility, CATSPER2 related	CATSPER2	1233	607249	2, 4
Deafness with keratopachydermia and constrictions of fingers and toes	GJB2	953	121011	2, 4, 8, 9
Deafness, autosomal dominant type 1	DIAPH1	965	602121	2, 4, 17
Deafness, autosomal dominant type 2A	KCNQ4	966	603537	2, 3, 4, 8, 9, 17
Deafness, autosomal dominant type 2B	GJB3	964	603324	2, 4, 8, 9
Deafness, autosomal dominant type 3A	GJB2	953	121011	2, 4, 8, 9
Deafness, autosomal dominant type 3B	GJB6	954	604418	2, 4, 8, 9
Deafness, autosomal dominant type 4B	CEACAM16	2443	614591	2, 4
Deafness, autosomal dominant type 4	MYH14	970	608568	2, 4, 17
Deafness, autosomal dominant type 5	GSDME	971	608798	2, 4, 17
Deafness, autosomal dominant type 6	WFS1	325	606201	2, 4, 8, 9, 17
Deafness, autosomal dominant type 9	COCH	972	603196	2, 3, 4, 8, 9, 17
Deafness, autosomal dominant type 10	EYA4	973	603550	2, 3, 4, 8, 9, 17
Deafness, autosomal dominant type 11	MYO7A	178	276903	2, 3, 4, 8, 9, 17
Deafness, autosomal dominant type 12	TECTA	974	602574	2, 4, 17
Deafness, autosomal dominant type 13	COL11A2	945	120290	2, 4, 17
Deafness, autosomal dominant type 15	POU4F3	224	602460	2, 4
Deafness, autosomal dominant type 17	MYH9	179	160775	2, 4, 8, 9, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Deafness, autosomal dominant type 20	ACTG1	989	102560	2, 3, 4, 8, 9
Deafness, autosomal dominant type 22	MYO6	976	600970	2, 4, 17
Deafness, autosomal dominant type 23	SIX1	977	601205	2, 4, 8, 9
Deafness, autosomal dominant type 25	SLC17A8	1665	607557	2, 4, 17
Deafness, autosomal dominant type 28	GRHL2	1666	608576	2, 4, 17
Deafness, autosomal dominant type 36	TMC1	300	606706	2, 3, 4, 8, 9, 17
Deafness, autosomal dominant type 39, with dentinogenesis type 1	DSPP	1667	125485	2, 4
Deafness, autosomal dominant type 40	CRYM	1668	123740	2, 4, 17
Deafness, autosomal dominant type 44	CCDC50	978	611051	2, 4, 17
Deafness, autosomal dominant type 48	MYO1A	1670	601478	2, 4, 17
Deafness, autosomal dominant type 52	POU4F3	224	602460	2, 4
Deafness, autosomal dominant type 64	DIABLO	1671	605219	2, 4
Deafness, autosomal dominant type 65	TBC1D24	783	613577	2, 3, 4, 8, 9, 17
Deafness, autosomal recessive	GJB3	964	603324	2, 4, 8, 9
Deafness, autosomal recessive	SUN1	1528	607723	2, 4, 17
Deafness, autosomal recessive type 1A	GJB2	953	121011	2, 4, 8, 9
Deafness, autosomal recessive type 1B	GJB6	954	604418	2, 4, 8, 9
Deafness, autosomal recessive type 2	MYO7A	178	276903	2, 3, 4, 8, 9, 17
Deafness, autosomal recessive type 3	MYO15A	955	602666	2, 17
Deafness, autosomal recessive type 4	FOXI1	2109	601093	2, 4, 8, 9
Deafness, autosomal recessive type 6	TMIE	956	607237	2, 4
Deafness, autosomal recessive type 7	TMC1	300	606706	2, 3, 4, 8, 9, 17
Deafness, autosomal recessive type 8/10	TMPRSS3	1648	605511	2, 4, 17
Deafness, autosomal recessive type 9	OTOF	957	603681	2, 3, 4, 8, 9, 17
Deafness, autosomal recessive type 12	CDH23	1646	605516	2, 17
Deafness, autosomal recessive type 15	GIPC3	1649	608792	2, 4
Deafness, autosomal recessive type 16	STRC	958	606440	2, 3, 4, 8, 9
Deafness, autosomal recessive type 18	USH1C	959	605242	2, 3, 4, 8, 9, 17
Deafness, autosomal recessive type 22	OTOA	1650	607038	2, 4, 17
Deafness, autosomal recessive type 23	PCDH15	110	605514	2, 4, 8, 9, 17
Deafness, autosomal recessive type 24	RDX	1651	179410	2, 4, 17
Deafness, autosomal recessive type 25	GRXCR1	1652	613283	2, 4
Deafness, autosomal recessive type 28	TRIOBP	960	609761	2, 4, 17
Deafness, autosomal recessive type 29	CLDN14	1653	605608	2, 4
Deafness, autosomal recessive type 30	MYO3A	961	606808	2, 3, 4, 8, 9, 17
Deafness, autosomal recessive type 31	WHRN	1654	607928	2, 4, 17
Deafness, autosomal recessive type 35	ESRRB	1655	602167	2, 3, 4, 8, 9, 17
Deafness, autosomal recessive type 36	ESPN	1656	606351	2, 4, 17
Deafness, autosomal recessive type 39	HGF	962	142409	2, 4, 17
Deafness, autosomal recessive type 42	ILDR1	1657	609739	2, 4, 17
Deafness, autosomal recessive type 48	CIB2	87	605564	2, 4
Deafness, autosomal recessive type 49	MARVELD2	1658	610572	2, 4, 17
Deafness, autosomal recessive type 53	COL11A2	945	120290	2, 4, 17
Deafness, autosomal recessive type 59	PJVK	1659	610219	2, 4
Deafness, autosomal recessive type 61	SLC26A5	1660	604943	2, 4, 17
Deafness, autosomal recessive type 63	LRTOMT	1662	612414	2, 4, 17
Deafness, autosomal recessive type 66	DCDC2	2647	605755	2, 4, 17
Deafness, autosomal recessive type 67	LHFPL5	1663	609427	2, 4
Deafness, autosomal recessive type 70	PNPT1	2686	610316	2, 4, 17
Deafness, autosomal recessive type 74	MSRB3	1661	613719	2, 4
Deafness, autosomal recessive type 76	SYNE4	1527	615535	2, 4
Deafness, autosomal recessive type 77	LOXHD1	722	613072	2, 4, 17
Deafness, autosomal recessive type 79	TPRN	1664	613354	2, 4, 17
Deafness, autosomal recessive type 86	TBC1D24	783	613577	2, 3, 4, 8, 9, 17
Deafness, autosomal recessive type 89	KARS1	2806	601421	2, 4, 17
Deafness, autosomal recessive type 91	SERPINB6	1878	173321	2, 4, 17
Deafness, autosomal recessive type 93	CABP2	2307	614899	2, 4
Deafness, congenital with inner ear agenesis, microtia, and microdontia	FGF3	2625	164950	2, 3, 4, 8, 9
Deafness, dystonia, and cerebral hypomyelination, X-linked	BCAP31	2311	300398	2, 4, 17
Deafness, nonsyndromic, sensorineural, mitochondrial	MT-RNR1	1827	561000	4
Deafness, X-linked type 1	PRPS1	231	311850	2, 3, 4, 8, 9
Deafness, X-linked type 2	POU3F4	1672	300039	2, 4, 8, 9
Deafness, X-linked type 4	SMPX	1673	300226	2, 4
Deafness, X-linked type 5	AIFM1	1353	300169	2, 3, 4, 8, 9, 17

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Deafness, X-linked type 6	COL4A6	2753	303631	2, 17
Fazio-Londe disease	SLC52A3	1941	613350	2, 3, 4, 8, 9
Hearing loss, MAP1A related	MAP1A	2003	600178	2, 4
Hearing loss, MYH7B related	MYH7B	2615	609928	2, 4
Heimler syndrome type 1	PEX1	678	602136	2, 3, 4, 8, 9, 17
Keratitis ichthyosis deafness syndrome autosomal dominant	GJB2	953	121011	2, 4, 8, 9
Keratoderma, palmoplantar, with deafness	GJB2	953	121011	2, 4, 8, 9
Knuckle pads and leukonychia sensorineural deafness	GJB2	953	121011	2, 4, 8, 9
Marshall syndrome	COL11A1	944	120280	2, 8, 17
Mitochondrial modifier of deafness	TRMU	46	610230	2, 4, 17
Opticoacoustic nerve atrophy with dementia	TIMM8A	298	300356	2, 4, 8, 9
Otopaladigital syndrome type 1	FLNA	803	300017	2, 3, 4, 8, 9, 17
Otopaladigital syndrome type 2	FLNA	803	300017	2, 3, 4, 8, 9, 17
Pendred syndrome	SLC26A4	988	605646	2, 4, 8, 9, 17
Pneumothorax, primary spontaneous	FLCN	55	607273	2, 4, 8, 9
Primary ciliary dyskinesia type 1	DNAI1	1476	604366	2, 4, 8, 9, 17
Primary ciliary dyskinesia type 2	DNAAF3	1292	614566	2, 4, 17
Primary ciliary dyskinesia type 3	DNAH5	1477	603335	2, 8, 17
Primary ciliary dyskinesia type 5	HYDIN	1478	610812	2, 17
Primary ciliary dyskinesia type 6	NME8	1479	607421	2, 4, 17
Primary ciliary dyskinesia type 7	DNAH11	1480	603339	2, 17
Primary ciliary dyskinesia type 9	DNAI2	1481	605483	2, 4, 17
Primary ciliary dyskinesia type 10	DNAAF2	2245	612517	2, 4, 17
Primary ciliary dyskinesia type 11	RSPH4A	1482	612647	2, 4, 17
Primary ciliary dyskinesia type 12	RSPH9	1483	612648	2, 4
Primary ciliary dyskinesia type 13	DNAAF1	1484	613190	2, 4, 17
Primary ciliary dyskinesia type 14	CCDC39	1485	613798	2, 4, 17
Primary ciliary dyskinesia type 15	CCDC40	1486	613799	2, 4, 17
Primary ciliary dyskinesia type 16	DNAL1	1487	610062	2, 4, 17
Primary ciliary dyskinesia type 17	CCDC103	749	614677	2, 4
Primary ciliary dyskinesia type 18	DNAAF5	1488	614864	2, 3, 4, 8, 9, 17
Primary ciliary dyskinesia type 19	LRRC6	1489	614930	2, 4, 17
Primary ciliary dyskinesia type 20	CCDC114	1490	615038	2, 4, 17
Primary ciliary dyskinesia type 23	ARMC4	2951	615408	2, 4, 17
Primary ciliary dyskinesia type 24	RSPH1	2952	609314	2, 4, 17
Primary ciliary dyskinesia type 25	DYX1C1	2953	608706	2, 4
Primary ciliary dyskinesia type 26	CFAP298	2954	615494	2, 4
Primary ciliary dyskinesia type 27	CCDC65	2955	611088	2, 4, 17
Primary ciliary dyskinesia type 28	SPAG1	2956	603395	2, 4, 17
Primary ciliary dyskinesia type 29	CCNO	2772	607752	2, 4
Primary ciliary dyskinesia, DNAH9 related	DNAH9	2007	603330	2, 17
Progressive hearing loss	P2RX2	1540	600844	2, 4
Pulmonary fibrosis, idiopathic	SFTPA2	2511	178642	2, 4
Sinoatrial node dysfunction and deafness	CACNA1D	2796	114206	17
Tietz albinism-deafness syndrome	MITF	777	156845	2, 4, 8, 9, 17
Wolfram syndrome type 1	WFS1	325	606201	2, 4, 8, 9, 17
Wolfram syndrome type 2	CISD2	952	611507	2, 3, 4, 8, 9
Wolfram-like syndrome, autosomal dominant	WFS1	325	606201	2, 4, 8, 9, 17

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Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
Abnormal mineralization panel	ABCC6, ALPL, ANKH, ANO5, AP2S1, BMP1, CA2, CASR, CLCN5, CLCN7, COL1A1, COL1A2, CREB3L1, CRTAP, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FGF23, FGFR1, FGFR3, FKBP10, GALNT3, GJA1, GNA11, GNAS, GORAB, HPGD, HRAS, IFITM5, KRAS, LRP5, MBTPS2, MTAP, NOTCH2, NRAS, OCRL, OSTM1, P3H1, P4HB, PHEX, PLEKHM1, PLOD2, PLS3, PPIB, PTDSS1, PTH1R, SERPINF1, SERPINH1, SLC26A2, SLC34A1, SLC34A3, SLC9A3R1, SLCO2A1, SNX10, SOST, SOX9, SP7, TBXAS1, TCIRG1, TGFB1, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TYROBP, VDR, WNT1		5437	6
CentoDx®	Targets exonic regions of >6500 genes associated with known clinical phenotypes		50158	5
CentoImmuno	ACTB, ADA, ADAR, AICDA, AIRE, AK2, AP3B1, ARMC4, ATM, BLM, BLNK, BLOC1S3, BTK, C3, CARD11, CASP10, CASP8, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CD19, CD247, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD59, CD79A, CD79B, CD81, CFAP298, CFH, CFI, CFTR, CHD7, CLCN7, CLPB, CORO1A, CR2, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DCLRE1C, DGKE, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DOCK8, DRC1, DTNBP1, ELANE, FADD, FAS, FASLG, FGA, FGB, FGG, FOXN1, FOXP3, G6PC3, G6PD, GFI1, HAX1, HPS1, HPS3, HPS4, HPS5, HPS6, HYDIN, ICOS, IFIH1, IFNGR1, IFNGR2, IGLL1, IKBKB, IKBKG, IKZF1, IL12B, IL12RB1, IL12RB2, IL1RN, IL21R, IL2RA, IL2RG, IL7R, IRF8, ISG15, ITK, JAGN1, JAK3, KRAS, LAMTOR2, LIG4, LPIN2, LRBA, LRRC6, LRRC8A, LYST, MAGT1, MALT1, MCM4, MEFV, MOGS, MS4A1, MVK, NBN, NCF1, NCF2, NCF4, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP12, NLRP3, NME8, NOD2, NOP10, NRAS, OFD1, PARN, PEPD, PIK3CD, PIK3R1, PLCG2, PNP, POLE, PRF1, PRKCD, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RBCK1, RFX5, RNASEH2A, RNASEH2B, RNASEH2C, RORC, RSPH1, RSPH4A, RSPH9, RTEL1, SAMD9, SAMHD1, SBDS, SERPING1, SH2D1A, SLC35C1, SLC7A7, SPAG1, SPINK5, SRP72, STAT1, STAT3, STAT5B, STIM1, STING1, STX11, STXBP2, TAZ, TBX1, TCN2, TERT, THBD, TICAM1, TINF2, TLR3, TNFRSF13B, TNFRSF13C, TNFRSF1A, TREX1, TRNT1, TTC7A, TYK2, UNC13D, UNG, VPS13B, VPS45, WAS, WRAP53, XIAP, ZAP70, ZMYND10		5428	6
CentoLCV	Genome wide analysis of structural variants and large copy number changes through sWGS		50118	5
CentoSkin	ABCA12, ALDH18A1, ALOX12B, ALOXE3, AP1S1, APCDD1, ATP6V0A2, ATP7A, CDSN, CERS3, CHST8, CLDN1, COL17A1, COL7A1, CSTA, CYP4F22, DSG1, DSG4, DSP, DST, EBP, EFEMP2, ELN, ERCC2, ERCC3, EXPH5, FBLN5, FERMT1, FLG, GJB2, GJB3, GJB4, GTF2H5, HR, ITGA3, ITGA6, ITGB4, JUP, KRT1, KRT10, KRT14, KRT2, KRT5, KRT71, KRT74, LAMA3, LAMB3, LAMC2, LIPH, LIPN, LORICRIN, LPAR6, MMP1, MPLKIP, NIPAL4, PEX7, PHYH, PKP1, PLEC, PNPLA1, POMP, PYCR1, RPL21, SLC27A4, SNAP29, SNRPE, SPINK5, ST14, STS, SUMF1, TGM1, TGM5		5430	6
Connective tissue and related disorders panel	ABCC6, ACTA2, ADAMTS2, ADAMTSL2, ALDH18A1, ATP6V0A2, ATP7A, B3GALT6, B3GAT3, B4GALT7, BGN, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, CREB3L1, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLCN, FLNA, FOXE3, GORAB, ITGB4, KCNJ13, LAMA3, LAMB3, LAMC2, LOX, LRP2, MAT2A, MBTPS2, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, PYCR1, RIN2, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SP7, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, VCAN, WNT1, ZNF469		5436	6

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Panel name	Genes	Test code	Available test methods
CentoICU®	AARS1, AARS2, AASS, ABAT, ABCA12, ABCA3, ABCB11, ABCC8, ABCD1, ABCD3, ABCD4, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACTA1, ACY1, ADA, ADAMTS13, ADAMTSL2, ADAR, ADK, ADNP, ADSL, AGA, AGK, AGL, AGPAT2, AGPS, AGRN, AGXT, AHCY, AICDA, AIFM1, AIMP1, AKAP9, AKR1D1, AKT2, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX12B, ALOXE3, ALPL, ALS2, AMACR, AMN, AMPD1, AMT, ANK1, ANKRD26, ANKS6, ANTXR1, ANTXR2, AP2S1, AP4B1, AP4E1, AP4M1, AP4S1, APOB, APTX, ARG1, ARL6, ARSA, ARSB, ARX, ASAH1, ASL, ASNS, ASPA, ASPM, ASS1, ATIC, ATP1A3, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATPAF2, ATR, ATRX, AUH, B3GLCT, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCKDK, BCS1L, BDNF, BICD2, BIN1, BLNK, BOLA3, BRAF, BRAT1, BRCA2, BSCL2, BSND, BTD, BTK, C12orf65, CA12, CACNA1C, CACNB2, CALM1, CAMTA1, CASK, CASR, CAST, CAV1, CAV3, CAVIN1, CBS, CCDC103, CCDC114, CCDC78, CD19, CD247, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD59, CD79A, CD79B, CD81, CD96, CDAN1, CDK5RAP2, CDKL5, CDKN1C, CENPJ, CEP152, CEP290, CERS3, CFAP298, CFH, CFHR3, CFL2, CFTR, CHAT, CHD7, CHKB, CHM, CHRNA1, CHRNB1, CHRND, CHRNE, CIDEC, CLCN1, CLCNKA, CLCNKB, CLDN16, CLN3, CLN5, CLN6, CLN8, CLPB, CNTN1, COA5, COG1, COG6, COG7, COL11A1, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A2, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COMP, COQ2, COQ8A, COQ9, CORO1A, COX10, COX15, COX20, COX6B1, CPS1, CPT1A, CPT2, CR2, CRPPA, CRTAP, CTNS, CTPS1, CTSA, CTSD, CUL4B, CXCR4, CYP11B1, CYP11B2, CYP17A1, CYP4F22, CYP7B1, D2HGDH, DBT, DCLRE1C, DDC, DDOST, DDR2, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DIAPH1, DLAT, DLD, DMD, DNA2, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC19, DNM2, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPM2, DPYD, DRC1, DSP, DST, DUOX2, DUOXA2, DYSF, EDN3, EEF1A2, EGR2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELANE, ENPP1, EPB42, EPCAM, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYA1, EYA4, F10, F11, F13A1, F2, F5, F7, F8, F9, FADD, FAH, FANCA, FANCB, FANCC, FANCD2, FANCL, FARS2, FASTKD2, FBN1, FBP1, FBXL4, FGA, FGB, FGFR2, FGFR3, FGG, FH, FIG4, FKBP14, FKRP, FKTN, FOXC1, FOXG1, FOXP3, FOXRED1, FRAS1, FUCA1, G6PC, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GARS1, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GFAP, GFM1, GFPT1, GJA1, GJB2, GJB4, GK, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLUD1, GLYCTK, GMPPB, GNAS, GNE, GNMT, GNPAT, GNPTAB, GP1BA, GP1BB, GP9, GPC3, GPHN, GPSM2, GSS, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBA1, HBA2, HBB, HCFC1, HESX1, HEXA, HEXB, HGD, HGF, HIBCH, HLCS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPGD, HRAS, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, ICOS, IDUA, IER3IP1, IFIH1, IFT172, IGF1, IGF1R, IGHMBP2, IGLL1, IKBKB, IL12RB1, IL2RA, IL2RG, IL7R, INS, INSR, INVS, IRF8, ITGA2B, ITGA6, ITGA7, ITGB3, ITGB4, IVD, JAG1, JAGN1, JAK3, JAM3, KAT6A, KAT6B, KBTBD13, KCNE1, KCNH1, KCNH2, KCNJ10, KCNJ11, KCNQ1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIF1B, KLF1, KLHL40, KLHL41, KLHL7, KRAS, KRT5, LAMA2, LAMA3, LAMB3, LAMC2, LAMP2, LAMTOR2, LARS2, LAS1L, LCT, LHX3, LHX4, LIAS, LIG4, LIPA, LIPN, LIPT1, LMBRD1, LMNA, LPIN1, LRBA, LRPPRC, LRRC8A, MAGEL2, MAGT1, MALT1, MAN2B1, MANBA, MAP2K1, MAP2K2, MAT1A, MCCC1, MCCC2, MCEE, MCM4, MCPH1, MECP2, MED12, MEF2C, MEGF10, MFN2, MFSD8, MITF, MKKS, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MPC1, MPI, MPL, MPV17, MPZ, MRPL3, MRPL44, MSMO1, MTHFR, MTM1, MTMR14, MTO1, MTR, MTRR, MUSK, MVK, MYCN, MYH9, NAA10, NAGA, NAGS, NALCN, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NDUFV2, NEB, NEU1, NEUROG3, NEXN, NFKB2, NFU1, NGF, NGLY1, NHEJ1, NIPAL4, NIPBL, NKX2-1, NKX2-5, NLRC4, NLRP3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NR0B1, NR3C2, NRAS, NSD1, NSDHL, NUBPL, OAT, OCLN, OCRL, OPA3, OPHN1, ORC1, ORC4, OTC, OXCT1, PAFAH1B1, PAH, PAX2, PAX3, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH19, PCNT, PDCD10, PDE10A, PDHA1, PDHB, PDHX, PDP1, PDSS2, PDX1, PEPD, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGM1, PHGDH, PHKG2, PHOX2B, PIGA, PIGN, PIGT, PIGV, PIK3CD, PKD2, PKHD1, PKLR, PLCB4, PLEC, PLOD1, PLP1, PMM2, PMP22, PNKP, PNP, PNPLA1, PNPO, PNPT1, POGZ, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU1F1, PPT1, PRDM16, PRKAG2, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PSAP, PSAT1, PSPH, PTPN11, PTPRC, PTRH2, PTS, PURA, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RAC2, RAF1, RAG1, RAG2, RANBP2, RAPSN, RARS2, RB1, RBBP8, RBM8A, RET, RFT1, RFX5, RFX6, RIT1, RMND1, RNASEH2C, RNASET2, RORC, RPS19, RRM2B, RXYLT1, RYR1, SALL1, SATB2, SBDS, SCN1A, SCN2A, SCN4A, SCN5A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SELENON, SERAC1, SERPINC1, SERPING1, SFTPB, SFTPC, SHOC2, SIL1, SIX3, SIX5, SKI, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC26A2, SLC26A3, SLC2A1, SLC30A2, SLC33A1, SLC3A1, SLC4A1, SLC52A1, SLC52A3, SLC5A1, SLC5A5, SLC6A1, SLC6A3, SLC6A5, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNAI2, SNX10, SOS1, SOX10, SOX2, SOX9, SPAST, SPEG, SPINK5, SPINT2, SPR, SPRED1, SPTA1, SPTAN1, SPTB, SRD5A3, ST3GAL3, ST3GAL5, STAR, STAT1, STAT3, STIL, STIM1, STS, STT3B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SYNE1, TACO1, TAT, TAZ, TBC1D24, TBCE, TBX19, TBX5, TCAP, TCN2, TFR2, TG, TGM1, TH, THRA, TJP2, TMCO1, TMEM165, STING1, TMEM70, TNFRSF13B, TNFRSF13C, TNFSF4, TNNT1, TP63, TPM2, TPM3, TPO, TPP1, TRIP11, TRMU, TRPV4, TSC1, TSC2, TSFM, TSHB, TSHR, TSPYL1, TTC7A, TTN, TUBA8, TUBB2A, TWNK, UBA1, UGT1A1, UMPS, UNG, UPB1, UQCRC2, UROD, UROS, WAS, WDPCP, WDR62, WDR73, WFS1, WNK1, WT1, ZAP70, ZEB2, ZFP57, ZNF423	5445	5

› Osteology, Dermatology and Immunology

Disease	Gene	Test code	OMIM Gene	Available test methods
3MC syndrome type 1	MASP1	1293	600521	2, 4, 17
3MC syndrome type 2	COLEC11	1294	612502	2, 4
Achondrogenesis type 1A	TRIP11	1928	604505	2, 3, 4, 8, 9, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Achondrogenesis type 1B	SLC26A2	1933	606718	2, 3, 4, 8, 9
Achondrogenesis type 2	COL2A1	943	120140	2, 4, 8, 9, 17
Achondroplasia	FGFR3	1452	134934	2, 3, 4, 8, 9
Acne inversa familial type 3	PSEN1	233	104311	2, 4, 8, 9, 17
Acrodermatitis enteropathica	SLC39A4	1358	607059	2, 3, 4, 8, 9, 17
Acrodysostosis 2	PDE4D	1053	600129	2, 3, 4, 8, 9, 17
Acrofacial dysostosis 1, Nager type	SF3B4	742	605593	2, 3, 4, 8, 9
Acrokeratosis verruciformis	ATP2A2	3623	108740	17
Adams-Oliver syndrome type 1	ARHGAP31	990	610911	2, 3, 4, 8, 9, 17
Adams-Oliver syndrome type 2	DOCK6	1275	614194	2, 3, 4, 8, 9, 17
Adams-Oliver syndrome type 3	RBPJ	1799	147183	2, 4
Adams-Oliver syndrome type 4	EOGT	2633	614789	2, 3, 4, 8, 9, 17
Adermatoglyphia	SMARCAD1	3804	612761	17
Albinism, oculocutaneous nonsyndromic	SLC24A5	1631	609802	2, 4, 17
Albinism, oculocutaneous type 1A	TYR	311	606933	2, 4, 8, 9
Albinism, oculocutaneous type 1B	TYR	311	606933	2, 4, 8, 9
Albinism, oculocutaneous type 2	OCA2	991	611409	2, 4, 8, 9, 17
Albinism, oculocutaneous type 3	TYRP1	862	115501	2, 3, 4, 8, 9
Albinism, oculocutaneous type 4	SLC45A2	863	606202	2, 3, 4, 8, 9
Albinism, oculocutaneous type 5	LRMDA	1538	614537	2, 4
Alopecia universalis	HR	2692	602302	2, 3, 4, 8, 9, 17
Amelogenesis imperfecta type 1A	LAMB3	1025	150310	2, 3, 4, 8, 9
Amelogenesis imperfecta type 1B	ENAM	2720	606585	2, 4, 17
Amelogenesis imperfecta type 1C	ENAM	2720	606585	2, 4, 17
Amelogenesis imperfecta type 1E	AMELX	2717	300391	2, 4
Amelogenesis imperfecta type 1F	AMBN	2711	601259	2, 4
Amelogenesis imperfecta type 1G	FAM20A	2719	611062	2, 4, 17
Amelogenesis imperfecta type 1H	ITGB6	2712	147558	2, 4, 17
Amelogenesis imperfecta type 2A1	KLK4	2709	603767	2, 4
Amelogenesis imperfecta type 2A2	MMP20	2710	604629	2, 4
Amelogenesis imperfecta type 2A3	WDR72	2715	613214	2, 4, 17
Amelogenesis imperfecta type 2A4	C4orf26	2713	614829	2, 4
Amelogenesis imperfecta type 2A5	SLC24A4	1526	609840	2, 4, 17
Amelogenesis imperfecta type 3	FAM83H	2718	611927	2, 4
Amelogenesis imperfecta type 4	DLX3	1748	600525	2, 3, 4, 8, 9
Amelotin deficiency	AMTN	2714	610912	2, 4
Amyloidosis, primary localized cutaneous, type 1	OSMR	2348	601743	2, 4, 17
Amyloidosis, primary localized cutaneous, type 2	IL31RA	2349	609510	2, 4
Arthrogryposis, distal, type 1A	TPM2	306	190990	2, 3, 4, 8, 9, 17
Arthrogryposis, distal, type 1B	MYBPC1	992	160794	2, 3, 4, 8, 9, 17
Arthrogryposis, distal, type 2A	MYH3	175	160720	2, 3, 4, 8, 9, 17
Arthrogryposis, distal, type 2B	MYH3	175	160720	2, 3, 4, 8, 9, 17
Arthrogryposis, distal, type 2B	TNNI2	994	191043	2, 4
Arthrogryposis, distal, type 2B	TNNT3	993	600692	2, 4, 17
Arthrogryposis, distal, type 3	PIEZO2	2397	613629	2, 3, 4, 8, 9, 17
Arthrogryposis, distal, type 5D	ECEL1	2536	605896	2, 3, 4, 8, 9, 17
Arthrogryposis, distal, type 5	PIEZO2	2397	613629	2, 3, 4, 8, 9, 17
Arthrogryposis, distal, type 7	MYH8	995	160741	2, 3, 4, 8, 9, 17
Arthrogryposis, mental retardation, and seizures	SLC35A3	1931	605632	2, 4, 17
Arthrogryposis, renal dysfunction, and cholestasis type 1	VPS33B	1760	608552	2, 4, 17
Arthrogryposis, renal dysfunction, and cholestasis type 2	VIPAS39	2467	613401	2, 3, 4, 8, 9, 17
Arthropathy, progressive pseudorheumatoid, of childhood	WISP3	326	603400	2, 4
Atelosteogenesis type 1	FLNB	1884	603381	2, 4, 17
Atelosteogenesis type 3	FLNB	1884	603381	2, 4, 17
Atrichia with papular lesions	HR	2692	602302	2, 3, 4, 8, 9, 17
Atypical Mycobacterial infection	IFNGR2	1987	147569	2, 4
Atypical Mycobacterial infection	IKBKG	1740	300248	2, 4
Atypical Mycobacterial infection	IL12RB1	1999	601604	2, 4, 17
Atypical Mycobacterial infection	STAT1	2076	600555	2, 4, 17
Atypical Mycobacterial infection, IL12RB2 related	IL12RB2	2105	601642	2, 4, 17
Autoimmune lymphoproliferative syndrome type 1A	FAS	1109	134637	2, 3, 4, 8, 9, 17
Autoimmune lymphoproliferative syndrome type 1B	FASLG	1113	134638	2, 3, 4, 8, 9
Autoimmune lymphoproliferative syndrome type 2A	CASP10	1156	601762	2, 4, 17
Autoimmune lymphoproliferative syndrome type 2B	CASP8	1155	601763	2, 4, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Autoimmune lymphoproliferative syndrome type 3	PRKCD	2462	176977	2, 4, 17
Autoimmune polyendocrinopathy syndrome type 1	AIRE	1831	607358	2, 3, 4, 8, 9, 17
Autoinflammation, lipodystroph and dermatosis syndrome	PSMB8	1295	177046	2, 3, 4, 8, 9
Avascular necrosis of the femoral head, primary	COL2A1	943	120140	2, 4, 8, 9, 17
Bare lymphocyte syndrome, type 2, complementation group A	CIITA	2979	600005	2, 4, 17
Bare lymphocyte syndrome, type 2	RFXANK	1899	603200	2, 3, 4, 8, 9
B-cell expansion with NFKB and T-cell anergy	CARD11	2797	607210	2, 4, 17
Beare-Stevenson cutis gyrata syndrome	FGFR2	1730	176943	2, 4, 8, 9, 17
Bent bone dysplasia syndrome	FGFR2	1730	176943	2, 4, 8, 9, 17
Bone mineral density QTL18, osteoporosis	PLS3	2435	300131	2, 4, 8, 9, 17
Buschke-Ollendorff syndrome	LEMD3	996	607844	2, 3, 4, 8, 9, 17
C1q deficiency	C1QA	2883	120550	2, 4
C2 deficiency	C2	1837	613927	2, 4, 17
C3 deficiency	C3	1838	120700	2, 3, 4, 8, 9, 17
C5 deficiency	C5	2465	120900	2, 17
C7 deficiency	C7	2196	217070	2, 4, 17
Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia	IARS2	3785	612801	17
Chediak-Higashi syndrome	LYST	997	606897	2, 17
Choanal atresia and lymphedema	PTPN14	2739	603155	2, 4, 17
Chondrocalcinosis type 2	ANKH	2084	605145	2, 4, 17
Chondrodysplasia punctata, X-linked dominant	EBP	2445	300205	2, 4
Chondrodysplasia punctata, X-linked recessive	ARSE	1864	300180	2, 4, 17
Chondrosarcoma, familial	EXT1	1733	608177	2, 4, 8, 9, 17
Cleidocranial dysplasia	RUNX2	999	600211	2, 4, 8, 9, 17
Cold autoinflammatory syndrome type 2	NLRP12	2248	609648	2, 4, 17
Cold autoinflammatory syndrome type 4, familial	NLRC4	3659	606831	17
Cole disease	ENPP1	2236	173335	2, 3, 4, 8, 9, 17
Cole-Carpenter syndrome type 1	P4HB	3325	176790	17
Combined cellular and humoral immune defects with granulomas	RAG2	1970	179616	2, 4, 8, 9
Combined immunodeficiency, B cell-negative, T cell-negative, NK cell positive	RAG2	1970	179616	2, 4, 8, 9
Combined immunodeficiency, X-linked, moderate	IL2RG	1820	308380	2, 4, 17
Contractural arachnodactyly, congenital	FBN2	590	612570	2, 3, 8, 17
Cornelia de Lange syndrome type 1	NIPBL	1000	608667	2, 4, 8, 9, 17
Cornelia de Lange syndrome type 2	SMC1A	278	300040	2, 3, 4, 8, 9, 17
Cornelia de Lange syndrome type 3	SMC3	1001	606062	2, 3, 4, 8, 9, 17
Cornelia de Lange syndrome type 4	RAD21	1446	606462	2, 3, 4, 8, 9, 17
Cornelia de Lange syndrome type 5	HDAC8	1375	300269	2, 3, 4, 8, 9, 17
Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome	TMCO1	3811	614123	17
Craniofacial-skeletal-dermatologic dysplasia	FGFR2	1730	176943	2, 4, 8, 9, 17
Crouzon syndrome with acanthosis nigricans	FGFR3	1452	134934	2, 3, 4, 8, 9
Cutaneous telangiectasia and cancer syndrome, familial	ATR	1851	601215	2, 17
Cutis laxa type 1A, autosomal recessive	FBLN5	2491	604580	2, 4, 17
Cutis laxa type 1B, autosomal recessive	EFEMP2	77	604633	2, 3, 4, 8, 9, 17
Cutis laxa type 2, autosomal dominant	FBLN5	2491	604580	2, 4, 17
Cutis laxa type 2A, autosomal recessive	ATP6V0A2	2490	611716	2, 4, 17
Cutis laxa type 2B, autosomal recessive	PYCR1	78	179035	2, 4, 17
Cutis laxa type 3A, autosomal recessive	ALDH18A1	2492	138250	2, 4, 17
Cutis laxa type 3B, autosomal recessive	PYCR1	78	179035	2, 4, 17
Cutis laxa, autosomal dominant	ELN	2271	130160	2, 4, 8, 9, 17
Czech dysplasia	COL2A1	943	120140	2, 4, 8, 9, 17
Dentin dysplasia, type 2	DSPP	1667	125485	2, 4
Dentinogenesis imperfecta, Shields type 2	DSPP	1667	125485	2, 4
Dentinogenesis imperfecta, Shields type 3	DSPP	1667	125485	2, 4
Dermatitis, atopic type 2	FLG	1002	135940	2, 3, 4, 8, 9
Dermatopathia pigmentosa reticularis	KRT14	1023	148066	2, 3, 4, 8, 9
Diaphyseal medullary stenosis with malignant fibrous histiocytoma	MTAP	975	156540	2, 4, 8, 9, 17
Diarrhea type 2 with microvillus atrophy	MYO5B	1895	606540	2, 4, 8, 9, 17
Diarrhea type 6	GUCY2C	2830	601330	2, 4, 17
Dyschromatosis universalis hereditaria type 3	ABCB6	3131	605452	2, 4, 17
Dyskeratosis congenita, autosomal recessive type 1	NOP10	1003	606471	2, 4
Dyskeratosis congenita, autosomal recessive type 2	NHP2	1004	606470	2, 4
Dyskeratosis congenita, autosomal recessive type 4/ autosomal dominant type 2	TERT	292	187270	2, 4, 8, 9, 17
Dyskeratosis congenita, autosomal recessive type 5	RTEL1	2064	608833	2, 4, 17
Dyskeratosis congenita, autosomal recessive type 6	PARN	2705	604212	2, 4, 17

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Dyskeratosis congenita, autosomal recessive type 7	ACD	3270	609377	2, 4
Dyskeratosis congenita, X-linked	DKC1	810	300126	2, 4, 8, 9, 17
Dyssegmental dysplasia, Silverman-Handmaker type	HSPG2	137	142461	2, 17
Ectodermal dysplasia type 4, hair/nail type	KRT85	2361	602767	2, 4
Ectodermal dysplasia, ectrodactyly, and macular dystrophy	CDH3	1678	114021	2, 4, 17
Ectodermal dysplasia, hidrotic	GJB6	954	604418	2, 4, 8, 9
Ectodermal dysplasia, hypohidrotic, autosomal recessive	EDAR	1007	604095	2, 4, 8, 9, 17
Ectodermal dysplasia, hypohidrotic, autosomal recessive	EDARADD	1008	606603	2, 4, 8, 9
Ectodermal dysplasia, hypohidrotic, with immune deficiency	IKBKG	1740	300248	2, 4
Ectodermal dysplasia, hypohidrotic, X-linked	EDA	1009	300451	2, 4, 8, 9, 17
Ectodermal dysplasia/skin fragility syndrome	PKP1	2433	601975	2, 4, 17
Ehlers-Danlos syndrome type 1/2	COL5A1	1010	120215	2, 4, 8, 9, 17
Ehlers-Danlos syndrome type 1/2	COL5A2	1011	120190	2, 3, 4, 8, 9, 17
Ehlers-Danlos syndrome type 3	COL3A1	1013	120180	2, 4, 8, 9, 17
Ehlers-Danlos syndrome type 3	TNXB	1012	600985	2, 3, 4, 8, 9, 17
Ehlers-Danlos syndrome type 4	COL3A1	1013	120180	2, 4, 8, 9, 17
Ehlers-Danlos syndrome type 4	COL5A1	1010	120215	2, 4, 8, 9, 17
Ehlers-Danlos syndrome type 6	PLOD1	1016	153454	2, 4, 8, 9, 17
Ehlers-Danlos syndrome type 7A	COL1A1	1017	120150	2, 4, 8, 9, 17
Ehlers-Danlos syndrome type 7B	COL1A2	1054	120160	2, 4, 8, 9, 17
Ehlers-Danlos syndrome type 7C	ADAMTS2	1361	604539	2, 3, 4, 8, 9, 17
Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss	FKBP14	1296	614505	2, 4
Ehlers-Danlos syndrome, musculocontractural type 1	CHST14	2429	608429	2, 4, 8, 9
Ehlers-Danlos syndrome, musculocontractural type 2	DSE	2489	605942	2, 3, 4, 8, 9, 17
Ehlers-Danlos syndrome, progeroid type 1	B4GALT7	2005	604327	2, 3, 4, 8, 9
Ehlers-Danlos syndrome, progeroid type, type 2	B3GALT6	3056	615291	2, 4
Emberger syndrome	GATA2	2544	137295	2, 4, 8, 9
Epidermolysis bullosa dystrophica	COL7A1	1019	120120	2, 3, 4, 8, 9, 17
Epidermolysis bullosa dystrophica, autosomal recessive, modifier of	MMP1	3159	120353	2, 4, 17
Epidermolysis bullosa junctionalis with pyloric atresia	ITGA6	1020	147556	2, 3, 4, 8, 9, 17
Epidermolysis bullosa junctionalis with pyloric atresia	ITGB4	1021	147557	2, 3, 4, 8, 9, 17
Epidermolysis bullosa simplex	KRT5	1022	148040	2, 3, 4, 8, 9, 17
Epidermolysis bullosa simplex with pyloric atresia	PLEC	705	601282	2, 17
Epidermolysis bullosa simplex, autosomal recessive type 1	KRT14	1023	148066	2, 3, 4, 8, 9
Epidermolysis bullosa simplex, autosomal recessive type 2	DST	1263	113810	2, 17
Epidermolysis bullosa simplex, Dowling-Meara type	KRT14	1023	148066	2, 3, 4, 8, 9
Epidermolysis bullosa simplex, Koebner type	KRT14	1023	148066	2, 3, 4, 8, 9
Epidermolysis bullosa simplex, Ogna type	PLEC	705	601282	2, 17
Epidermolysis bullosa simplex, Weber-Cockayne type	KRT14	1023	148066	2, 3, 4, 8, 9
Epidermolysis bullosa, generalized atrophic benign	LAMA3	154	600805	2, 17
Epidermolysis bullosa, junctional	COL17A1	1024	113811	2, 3, 4, 8, 9, 17
Epidermolysis bullosa, junctional	LAMC2	1026	150292	2, 4, 17
Epidermolysis bullosa, junctional, Herlitz type	LAMA3	154	600805	2, 17
Epidermolysis bullosa, junctional, Herlitz type	LAMB3	1025	150310	2, 3, 4, 8, 9, 17
Epidermolysis bullosa, junctional, non-Herlitz type	LAMB3	1025	150310	2, 3, 4, 8, 9
Epidermolysis bullosa, lethal acantholytic	DSP	1297	125647	2, 4, 8, 9, 17
Epidermolysis bullosa, nonspecific, autosomal recessive	EXPH5	2431	612878	2, 4, 17
Epidermolytic hyperkeratosis	KRT1	1027	139350	2, 4
Epidermolytic hyperkeratosis	KRT10	1028	148080	2, 3, 4, 8, 9, 17
Epidermolytic palmoplantar keratoderma	KRT9	1029	607606	2, 3, 4, 8, 9
Epiphyseal dysplasia, multiple, type 1	COMP	1451	600310	2, 3, 4, 8, 9, 17
Epiphyseal dysplasia, multiple, type 3	COL9A3	2164	120270	2, 4, 17
Epiphyseal dysplasia, multiple, type 5	MATN3	2165	602109	2, 4, 17
Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis and hyper IgE	DSG1	2169	125670	2, 4, 17
Erythrokeratodermia variabilis et progressive	GJB3	964	603324	2, 4, 8, 9
Erythrokeratodermia variabilis et progressive	GJB4	1031	605425	2, 4
Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis	COX4I2	1030	607976	2, 4
Exostoses, multiple, type 1	EXT1	1733	608177	2, 4, 8, 9, 17
Exostoses, multiple, type 2	EXT2	2375	608210	2, 4, 8, 9, 17
Fanconi anemia, complementation group Q	ERCC4	2543	133520	2, 4, 17
Feingold syndrome type 2	MIR17HG	1032	609415	2, 4
Fibrochondrogenesis 2	COL11A2	945	120290	2, 4, 17
Fibrochondrogenesis type 1	COL11A1	944	120280	2, 8, 17
Fibrodysplasia ossificans progressiva	ACVR1	707	102576	2, 3, 4, 8, 9, 17

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Fibrosis of extraocular muscles, congenital type 2	PHOX2A	1033	602753	2, 3, 4, 8, 9
Floating-Harbor syndrome	SRCAP	1034	611421	2, 3, 4, 8, 9, 17
Focal dermal hypoplasia	PORCN	805	300651	2, 3, 4, 8, 9, 17
Frank-ter Haar syndrome	SH3PXD2B	266	613293	2, 4, 17
Geleophysic dysplasia type 1	ADAMTSL2	1035	612277	2, 3, 4, 8, 9, 17
Geroderma osteodysplasticum	GORAB	3057	607983	2, 4
Ghosal hematodiaphyseal syndrome	TBXAS1	2428	274180	2, 4, 17
Gnathodiaphyseal dysplasia	ANO5	719	608662	2, 4, 8, 9
Gracile bone dysplasia	FAM111A	2046	615292	2, 4, 17
Granulomatous disease, chronic, autosomal recessive, cytochrome b- positive, type 1	NCF1	1968	608512	2, 3, 4, 8, 9, 17
Granulomatous disease, chronic, autosomal recessive, cytochrome b-negative	CYBA	2172	608508	2, 4
Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 2	NCF2	2173	608515	2, 4, 17
Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 3	NCF4	2174	601488	2, 4, 17
Granulomatous disease, chronic, X-linked	CYBB	998	300481	2, 3, 4, 8, 9, 17
Greenberg skeletal dysplasia	LBR	2446	600024	2, 4, 17
Griscelli syndrome type 1	MYO5A	785	160777	2, 3, 4, 8, 9, 17
Griscelli syndrome type 3	MLPH	1967	606526	2, 4, 17
Haim-Munk syndrome	CTSC	488	602365	2, 3, 4, 8, 9, 17
Heimler syndrome type 1	PEX1	678	602136	2, 3, 4, 8, 9, 17
Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	CD59	2663	107271	2, 4, 17
Hemophagocytic lymphohistiocytosis type 2	PRF1	225	170280	2, 4, 8, 9
Hemophagocytic lymphohistiocytosis type 3	UNC13D	313	608897	2, 4, 8, 9, 17
Hemophagocytic lymphohistiocytosis type 4	STX11	282	605014	2, 4, 8, 9
Hemophagocytic lymphohistiocytosis type 5	STXBP2	283	601717	2, 4, 17
Hennekam lymphangiectasia-lymphedema syndrome type 1	CCBE1	2642	612753	2, 4
Hepatic venoocclusive disease with immunodeficiency	SP110	2398	604457	2, 4, 17
Herpes simplex encephalitis type 2, susceptibility to	TLR3	2219	603029	2, 4, 17
Histiocytosis-lymphadenopathy plus syndrome	SLC29A3	2451	612373	2, 4
Hyaline fibromatosis syndrome	ANTXR2	1724	608041	2, 3, 4, 8, 9, 17
Hyper-IgE recurrent infection syndrome	STAT3	1679	102582	2, 4, 8, 9, 17
Hyper-IgE recurrent infection syndrome, autosomal recessive	DOCK8	1782	611432	2, 4, 8, 9, 17
Hypertrophic osteoarthropathy type 1	HPGD	2268	601688	2, 4, 17
Hypertrophic osteoarthropathy type 2	SLCO2A1	759	601460	2, 4, 17
Hypochondroplasia	FGFR3	1452	134934	2, 3, 4, 8, 9
Hypophosphatemic rickets with hypercalciuria	SLC34A3	2649	609826	2, 3, 4, 8, 9, 17
Hypophosphatemic rickets, autosomal dominant	FGF23	1133	605380	2, 4, 8, 9
Hypophosphatemic rickets, autosomal recessive type 1	DMP1	1134	600980	2, 4
Hypophosphatemic rickets, autosomal recessive type 2	ENPP1	2236	173335	2, 3, 4, 8, 9, 17
Hypophosphatemic rickets, X-linked	PHEX	1135	300550	2, 4, 8, 9, 17
Hypotrichosis and recurrent skin vesicles	DSC3	3523	600271	17
Hypotrichosis type 1	APCDD1	2691	607479	2, 4
Hypotrichosis type 2	CDSN	1443	602593	2, 4
Hypotrichosis type 3	KRT74	1676	608248	2, 4, 17
Hypotrichosis type 4	HR	2692	602302	2, 3, 4, 8, 9, 17
Hypotrichosis type 6	DSG4	2664	607892	2, 4, 17
Hypotrichosis type 7	LIPH	2269	607365	2, 4, 17
Hypotrichosis type 8	LPAR6	2693	609239	2, 4
Hypotrichosis type 11	SNRPE	2694	128260	2, 4
Hypotrichosis type 12	RPL21	2695	603636	2, 4
Hypotrichosis type 13	KRT71	1675	608245	2, 4, 17
Hypotrichosis-lymphedema-telangiectasia syndrome	SOX18	2452	601618	2, 4
Ichthyosiform erythroderma, congenital, nonbullous type 1	ALOXE3	1038	607206	2, 3, 4, 8, 9, 17
Ichthyosiform erythroderma, congenital, nonbullous type 1	NIPAL4	1039	609383	2, 4, 17
Ichthyosis congenital, autosomal recessive, PNPLA1 related	PNPLA1	1298	612121	2, 4, 17
Ichthyosis congenital, Harlequin fetus type	ABCA12	1	607800	2, 3, 4, 8, 9, 17
Ichthyosis follicularis, atricia, and photophobia syndrome	MBTPS2	318	300294	2, 4, 17
Ichthyosis prematurity syndrome	SLC27A4	1802	604194	2, 4, 17
Ichthyosis vulgaris	FLG	1002	135940	2, 3, 4, 8, 9
Ichthyosis, bullous type	KRT2	1041	600194	2, 3, 4, 8, 9, 17
Ichthyosis, congenital, autosomal recessive type 1	TGM1	296	190195	2, 3, 4, 8, 9, 17
Ichthyosis, congenital, autosomal recessive, type 2	ALOX12B	1037	603741	2, 3, 4, 8, 9, 17
Ichthyosis, congenital, autosomal recessive, type 9	CERS3	2180	615276	2, 4, 17
Ichthyosis, congenital, autosomal recessive, type 11	ST14	2583	606797	2, 4, 17
Ichthyosis, lamellar type 2	ABCA12	1	607800	2, 3, 4, 8, 9, 17

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Ichthyosis, lamellar type 3	CYP4F22	1042	611495	2, 4, 17
Ichthyosis, lamellar type 4	LIPN	1043	613924	2, 3, 4, 8, 9, 17
Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	CLDN1	2582	603718	2, 4
Ichthyosis, spastic quadriplegia, and mental retardation	ELOVL4	941	605512	2, 4
Ichthyosis, X-linked	STS	1044	300747	2, 4, 8, 9, 17
Immunodeficiency common variable type 1	ICOS	648	604558	2, 3, 4, 8, 9
Immunodeficiency common variable type 2	TNFRSF13B	2386	604907	2, 3, 4, 8, 9
Immunodeficiency common variable type 3	CD19	2481	107265	2, 4, 17
Immunodeficiency common variable type 4	TNFRSF13C	2411	606269	2, 4
Immunodeficiency common variable type 6	CD81	2832	186845	2, 4, 17
Immunodeficiency common variable type 8	LRBA	2409	606453	2, 17
Immunodeficiency common variable type 10	NFKB2	2410	164012	2, 4, 17
Immunodeficiency due to defect in MAPBP-interacting protein	LAMTOR2	2847	610389	2, 4
Immunodeficiency due to purine nucleoside phosphorylase deficiency	PNP	2463	164050	2, 4
Immunodeficiency type 2, with hyper-IgM	AICDA	2239	605257	2, 4
Immunodeficiency type 3, with hyper-IgM	CD40	2819	109535	2, 4, 17
Immunodeficiency type 5, with hyper IgM	UNG	2240	191525	2, 4, 17
Immunodeficiency type 8	CORO1A	2461	605000	2, 4, 17
Immunodeficiency type 10	STIM1	2391	605921	2, 3, 4, 8, 9, 17
Immunodeficiency type 11	CARD11	2797	607210	2, 4, 17
Immunodeficiency type 12	MALT1	2817	604860	2, 4, 17
Immunodeficiency type 14	PIK3CD	2071	602839	2, 4, 17
Immunodeficiency type 15	IKBKB	2842	603258	2, 4, 17
Immunodeficiency type 18	CD3E	2460	186830	2, 4, 17
Immunodeficiency type 19	CD3D	2459	186790	2, 4
Immunodeficiency type 21	GATA2	2544	137295	2, 4, 8, 9
Immunodeficiency type 22	LCK	2848	153390	2, 4, 17
Immunodeficiency type 24	CTPS1	2834	123860	2, 4, 17
Immunodeficiency type 25	CD247	2537	186780	2, 4, 17
Immunodeficiency type 32A, mycobacteriosis, autosomal dominant	IRF8	2576	601565	2, 4, 17
Immunodeficiency type 32B, monocyte and dendritic cell deficiency, autosomal recessive	IRF8	2576	601565	2, 4, 17
Immunodeficiency type 34	CYBB	998	300481	2, 3, 4, 8, 9, 17
Immunodeficiency type 35	TYK2	2079	176941	2, 3, 4, 8, 9, 17
Immunodeficiency type 36	PIK3R1	2523	171833	2, 4, 17
Immunodeficiency type 38	ISG15	2577	147571	2, 4, 8, 9
Immunodeficiency type 42	RORC	3800	602943	17
Immunodeficiency, isolated	IKBKG	1740	300248	2, 4
Immunodeficiency, primary, autosomal recessive, IL21R-related	IL21R	2844	605383	2, 4, 17
Immunodeficiency, X-linked with hyper-IgM	CD40LG	1742	300386	2, 3, 4, 8, 9
Immunodeficiency-centromeric instability-facial anomalies syndrome type 1	DNMT3B	2529	602900	2, 3, 4, 8, 9, 17
Immunodeficiency-centromeric instability-facial anomalies syndrome type 2	ZBTB24	2464	614064	2, 4
Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	FOXP3	1045	300292	2, 3, 4, 8, 9, 17
Immunological disorder, PECAM1 related	PECAM1	2058	173445	2, 4
Immunological disorder, PICALM related	PICALM	1783	603025	2, 4, 17
Incontinentia pigmenti type 2	IKBKG	1740	300248	2, 4
Inflammatory bowel disease type 13	ABCB1	1790	171050	2, 4, 17
Inflammatory skin and bowel disease, neonatal, type 1	ADAM17	2738	603639	2, 4, 17
Interleukin 2 receptor deficiency	IL2RA	1959	147730	2, 4, 17
Interleukin 12A deficiency	IL12A	2575	161560	2, 4, 17
Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	ITGA3	2432	605025	2, 4, 17
Invasive pneumococcal disease, recurrent isolated type 1	IRAK4	2937	606883	2, 4, 17
Invasive pneumococcal disease, recurrent isolated type 2	IKBKG	1740	300248	2, 4
IRAK4 deficiency	IRAK4	2937	606883	2, 4, 17
Keratoderma, palmoplantar, punctate type 1A	AAGAB	2382	614888	2, 4
Keratosis follicularis spinulosa declavans, X-linked	MBTPS2	318	300294	2, 4, 17
Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	POMP	2434	613386	2, 4
Keratosis palmoplantaris striata type 1	DSG1	2169	125670	2, 4, 17
Keratosis palmoplantaris striata type 2	DSP	1297	125647	2, 4, 8, 9, 17
Kindler syndrome	FBLIM1	2229	607747	2, 4
Kindler syndrome	FERMT1	1046	607900	2, 4, 17
Laryngoonychocutaneous syndrome	LAMA3	154	600805	2, 3, 8, 17
Legg-Calve-Perthes disease	COL2A1	943	120140	2, 4, 8, 9, 17
Legius syndrome	SPRED1	281	609291	2, 4, 8, 9, 17
Leukocyte adhesion deficiency type 3	FERMT3	2917	607901	2, 4

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LIG4 syndrome	LIG4	1543	601837	2, 4, 17
Loeys-Dietz syndrome type 1A	TGFBR1	294	190181	2, 4, 8, 9, 17
Loeys-Dietz syndrome type 1B	TGFBR2	295	190182	2, 3, 4, 8, 9, 17
Loeys-Dietz syndrome type 1C	SMAD3	1047	603109	2, 4, 17
Loeys-Dietz syndrome type 2A	TGFBR1	294	190181	2, 4, 8, 9, 17
Loeys-Dietz syndrome type 2B	TGFBR2	295	190182	2, 3, 4, 8, 9, 17
Lymphedema, hereditary, type 1A	FLT4	2906	136352	2, 4, 17
Lymphedema, hereditary, type IC	GJC2	508	608803	2, 4, 8, 9
Lymphoproliferative syndrome type 2	CD27	3127	186711	2, 4
Majeed syndrome	LPIN2	1965	605519	2, 4, 17
Major histocompatibility comples 1 deficiency	MR1	1780	600764	2, 3, 4, 8, 9
Mal de Meleda	SLURP1	1048	606119	2, 4
Malaria, cerebral, susceptibility to	ICAM1	3786	147840	17
Malaria, resistance to	FCGR2B	3779	604590	17
Mandibuloacral dysplasia with type B lipodystrophy	ZMPSTE24	328	606480	2, 4, 8, 9
Marfan syndrome, TGFBR1 related	TGFBR1	294	190181	2, 4, 8, 9, 17
Marfan syndrome, TGFBR2 related	TGFBR2	295	190182	2, 3, 4, 8, 9, 17
McKusick-Kaufman syndrome	MKKS	166	604896	2, 3, 4, 8, 9
Meconium ileus	GUCY2C	2830	601330	2, 4, 17
MEDNIK syndrome	AP1S1	2581	603531	2, 4
MERRF syndrome, MT-TK related	MT-TK	657	590060	4
MERRF syndrome, MT-TP related	MT-TP	658	590075	4
Metaphyseal chondrodysplasia, Schmid type	COL10A1	2412	120110	2, 3, 4, 8, 9
Metaphyseal chondromatosis with increased urinary excretion of D-2-hydroxyglutarate	IDH1	2142	147700	2, 4
Muckle-wells syndrome	NLRP3	1050	606416	2, 3, 4, 8, 9, 17
Multicentric carpotarsal osteolysis syndrome	MAFB	633	608968	2, 4
Multicentric osteolysis, nodulosis, and arthropathy	MMP2	2225	120360	2, 3, 4, 8, 9, 17
Multiple pterygium syndrome lethal type	CHRNA1	60	100690	2, 3, 4, 8, 9, 17
Multiple pterygium syndrome lethal type	CHRND	61	100720	2, 3, 4, 8, 9, 17
Mycobacterial infection, atypical, familial disseminated	IFNGR1	1886	107470	2, 4, 17
Naegeli-Franceschetti-Jadassohn syndrome	KRT14	1023	148066	2, 3, 4, 8, 9
Nail-Patella syndrome	LMX1B	159	602575	2, 4, 8, 9
Nephrolithiasis/osteoporosis, hypophosphatemic, type 2	SLC9A3R1	2966	604990	2, 4
Netherton syndrome	SPINK5	1051	605010	2, 3, 4, 8, 9, 17
Neurofibromatosis type 1 -like syndrome	SPRED1	281	609291	2, 4, 8, 9, 17
Neurofibromatosis type 1	NF1	182	613113	2, 8, 17
Neurofibromatosis type 2	NF2	183	607379	2, 4, 8, 9, 17
Neutropenia, nonimmune chronic idiopathic, of adults	GFI1	2455	600871	2, 3, 4, 8, 9
Neutropenia, severe congenital type 2, autosomal dominant	GFI1	2455	600871	2, 3, 4, 8, 9
Neutropenia, severe congenital type 4, autosomal recessive	G6PC3	2456	611045	2, 3, 4, 8, 9
Neutropenia, severe congenital type 5, autosomal recessive	VPS45	2810	610035	2, 4, 17
Neutropenia, severe congenital type 6, autosomal recessive	JAGN1	2846	616012	2, 4
Neutrophil immunodeficiency syndrome	RAC2	2539	602049	2, 4, 17
Odontoonychodermal dysplasia	WNT10A	1052	606268	2, 3, 4, 8, 9
Olmsted syndrome	TRPV3	739	607066	2, 4, 17
Omenn syndrome	DCLRE1C	2081	605988	2, 4, 8, 9, 17
Omenn syndrome	RAG2	1970	179616	2, 4, 8, 9
Omodysplasia type 1	GPC6	2065	604404	2, 3, 4, 8, 9
Opsismodysplasia	INPPL1	1868	600829	2, 4, 17
Osseous heteroplasia, progressive	GNAS	1345	139320	2, 4, 8, 9, 17
Osteoarthritis with mild chondrodysplasia	COL2A1	943	120140	2, 4, 8, 9, 17
Osteogenesis and dental anomalies, CSF1 related	CSF1	1989	120420	2, 3, 4, 8, 9
Osteogenesis disorders, CREB3L1 related	CREB3L1	2387	258480	2, 4, 17
Osteogenesis imperfecta	COL1A1	1017	120150	2, 4, 8, 9, 17
Osteogenesis imperfecta	COL1A2	1054	120160	2, 4, 8, 9, 17
Osteogenesis imperfecta type 5	IFITM5	664	614757	2, 4, 8, 9
Osteogenesis imperfecta type 6	SERPINF1	2436	172860	2, 4, 17
Osteogenesis imperfecta type 7	CRTAP	1057	605497	2, 4, 17
Osteogenesis imperfecta type 8	P3H1	1058	610339	2, 4, 17
Osteogenesis imperfecta type 9	PPIB	1055	123841	2, 4
Osteogenesis imperfecta type 10	SERPINH1	1824	613848	2, 4
Osteogenesis imperfecta type 11	FKBP10	1056	607063	2, 4, 17
Osteogenesis imperfecta type 12	SP7	113	606633	2, 4
Osteogenesis imperfecta type 13	BMP1	98	112264	2, 3, 4, 8, 9, 17

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15: Biochemical Genetics Panel Plus
16: Single Gene Sequencing (NGS)
17: Single Gene Sequencing + CNV (NGS)

Disease	Gene	Test code	OMIM Gene	Available test methods
Osteogenesis imperfecta type 14	TMEM38B	2210	611236	2, 3, 4, 8, 9
Osteogenesis imperfecta type 15	WNT1	2437	164820	2, 4
Osteogenesis imperfecta with congenital joint contractures	PLOD2	1217	601865	2, 4, 17
Osteolysis, familial expansile	TNFRSF11A	1086	603499	2, 4, 17
Osteomyelitis, sterile multifocal, with periostitis and pustulosis	IL1RN	1777	147679	2, 3, 4, 8, 9
Osteopathia striata with cranial sclerosis	AMER1	1059	300647	2, 4, 8, 9
Osteopetrosis of infancy, malignant	SNX10	1299	614780	2, 3, 4, 8, 9
Osteopetrosis, autosomal dominant type 1	CLCN7	706	602727	2, 3, 4, 8, 9, 17
Osteopetrosis, autosomal recessive type 1	TCIRG1	807	604592	2, 4, 17
Osteopetrosis, autosomal recessive type 2	TNFSF11	808	602642	2, 4
Osteopetrosis, autosomal recessive type 3	CA2	668	611492	2, 3, 4, 8, 9
Osteopetrosis, autosomal recessive type 4	CLCN7	706	602727	2, 3, 4, 8, 9, 17
Osteopetrosis, autosomal recessive type 5	OSTM1	811	607649	2, 4, 17
Osteopetrosis, autosomal recessive type 6	PLEKHM1	814	611466	2, 4, 17
Osteopetrosis, autosomal recessive type 7	TNFRSF11A	1086	603499	2, 4, 17
Osteoporosis pseudoglioma syndrome	LRP5	124	603506	2, 4, 8, 9, 17
Pachyonychia congenita type 1	KRT16	2326	148067	2, 4
Pachyonychia congenita type 2	KRT17	2318	148069	2, 4
Pachyonychia congenita type 3	KRT6A	1961	148041	2, 4
Pachyonychia congenita type 4	KRT6B	2327	148042	2, 4
Paget disease of bone	SQSTM1	2337	601530	2, 4, 17
Paget disease, juvenile	TNFRSF11B	2336	602643	2, 4
Palmoplantar keratoderma, nonepidermolytic, focal	KRT16	2326	148067	2, 4
Papillon-Lefevre syndrome	CTSC	488	602365	2, 3, 4, 8, 9, 17
Peeling skin syndrome type 1	CDSN	1443	602593	2, 4
Peeling skin syndrome type 2	TGM5	1061	603805	2, 3, 4, 8, 9, 17
Peeling skin syndrome type 3	CHST8	1444	610190	2, 4
Peeling skin syndrome type 4	CSTA	2911	184600	2, 4
Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads	CAST	3620	114090	17
Piebaldism	KIT	1178	164920	2, 4, 8, 9, 17
Piebaldism	SNAI2	2103	602150	2, 4, 8, 9
Pityriasis rubra pilaris	CARD14	2640	607211	2, 4, 17
Poikiloderma with neutropenia	USB1	2618	613276	2, 3, 4, 8, 9
Porokeratosis type 3, disseminated superficial actinic	MVK	1741	251170	2, 3, 4, 8, 9, 17
Porphyria cutanea tarda	UROD	320	613521	2, 4, 8, 9, 17
Protoporphyria, erythropoietic type 1	FECH	3464	612368	17
Pseudoachondroplasia	COMP	1451	600310	2, 3, 4, 8, 9, 17
Pseudoxanthoma elasticum	ABCC6	2334	603234	2, 4, 8, 9, 17
Pseudoxanthoma elasticum, forme fruste	ABCC6	2334	603234	2, 4, 8, 9, 17
Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency	GGCX	3177	137167	2, 4, 17
Psoriasis susceptibility type 11	IL12B	1911	161561	2, 4, 17
Psoriasis type 2	CARD14	2640	607211	2, 4, 17
Psoriasis, generalized pustular	IL36RN	1823	605507	2, 3, 4, 8, 9
Pterygium syndrome	CHRNG	1300	100730	2, 3, 4, 8, 9, 17
Pulmonary fibrosis and/or bone marrow failure, telomere-related, type 1	TERT	292	187270	2, 4, 8, 9, 17
Pulmonary fibrosis and/or bone marrow failure, telomere-related, type 4	PARN	2705	604212	2, 4, 17
Pycnodysostosis	CTSK	1063	601105	2, 3, 4, 8, 9, 17
Pyogenic bacterial infections, recurrent, due to MYD88 deficiency	MYD88	2938	602170	2, 4
Pyogenic sterile arthritis, pyoderma gangrenosum, and acne	PSTPIP1	2255	606347	2, 4, 17
Radioulnar synostosis, FGFRL1 related	FGFRL1	2288	605830	2, 4
Raine syndrome	FAM20C	2496	611061	2, 4
Restrictive dermopathy, lethal	LMNA	158	150330	2, 4, 8, 9, 17
Restrictive dermopathy, lethal	ZMPSTE24	328	606480	2, 4, 8, 9
Reticular dysgenesis	AK2	2061	103020	2, 4, 17
Reticulate acropigmentation of Kitamura	ADAM10	3763	602192	17
Rheumatoid arthritis, susceptibility to	AFF3	2118	601464	2, 4, 17
Rheumatoid arthritis, TNFAIP3 related	TNFAIP3	2122	191163	2, 4
Rickets, vitamin D 25-hydroxylation-deficient, type 1B	CYP2R1	2279	608713	2, 4
Rickets, vitamin D dependent, type 1	CYP27B1	2959	609506	2, 4, 17
Rickets, vitamin D-resistant, type 2A	VDR	1276	601769	2, 3, 4, 8, 9, 17
Sarcoidosis, early-onset	NOD2	2310	605956	2, 3, 4, 8, 9, 17
Selective T-cell defect	ZAP70	2291	176947	2, 4, 17
Severe combined immunodeficiency due to ADA deficiency	ADA	1682	608958	2, 4, 17
Severe combined immunodeficiency due to IL2 deficiency	IL2	2843	147680	2, 4

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Disease	Gene	Test code	OMIM Gene	Available test methods
Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	NHEJ1	1211	611290	2, 3, 4, 8, 9
Severe combined immunodeficiency, Athabascan type	DCLRE1C	2081	605988	2, 4, 8, 9, 17
Severe combined immunodeficiency, B cell-negative	RAG1	1807	179615	2, 4, 17
Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	PTPRC	2458	151460	2, 3, 4, 8, 9, 17
Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	IL7R	2457	146661	2, 4, 17
Skin fragility-woolly hair syndrome	DSP	1297	125647	2, 4, 8, 9, 17
Skin hair eye pigmentation type 6	SLC24A4	1526	609840	2, 4, 17
Smith-McCort dysplasia	DYM	3043	607461	2, 4, 17
Spondylocarpotarsal synostosis syndrome	FLNB	1884	603381	2, 4, 17
Spondylocheirodysplasia, Ehlers-Danlos syndrome-like	SLC39A13	2430	608735	2, 4, 17
Spondylocostal dysostosis, autosomal recessive type 1	DLL3	2202	602768	2, 3, 4, 8, 9
Spondylocostal dysostosis, autosomal recessive type 2	MESP2	1062	605195	2, 4, 8, 9
Spondylocostal dysostosis, autosomal recessive type 3	LFNG	1064	602576	2, 3, 4, 8, 9
Spondyloenchondrodysplasia with immune dysregulation	ACP5	1723	171640	2, 4
Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	B3GALT6	3056	615291	2, 4
Spondyloepimetaphyseal dysplasia, MATN3 related	MATN3	2165	602109	2, 4, 17
Spondyloepiphyseal dysplasia with congenital joint dislocations	CHST3	109	603799	2, 3, 4, 8, 9
Spondylometaepiphyseal dysplasia, short limb-hand type	DDR2	1065	191311	2, 4, 17
Steatocystoma multiplex	KRT17	2318	148069	2, 4
Stiff skin syndrome	FBN1	950	134797	2, 8, 17
Systemic lupus erythematosus	DNASE1	1066	125505	2, 4
Systemic lupus erythematosus type 16	DNASE1L3	2975	602244	2, 4
Systemic lupus erythematosus, susceptibility to	ITGAM	2277	120980	2, 4, 17
T-cell immunodeficiency, congenital alopecia, and nail dystrophy	FOXN1	2538	600838	2, 4, 17
Telangiectasia hereditary hemorrhagic type 5	GDF2	2261	605120	2, 4, 8, 9
Telangiectasia, hereditary hemorrhagic, of Rendu, Osler and Weber type 1	ENG	1068	131195	2, 4, 8, 9, 17
Telangiectasia, hereditary hemorrhagic, type 2	ACVRL1	1067	601284	2, 4, 8, 9, 17
Terminal osseous dysplasis	FLNA	803	300017	2, 3, 4, 8, 9, 17
Thrombospondin Type 1 domain-containing protein 1	THSD1	2894	616821	2, 4
Trichodontoosseous syndrome	DLX3	1748	600525	2, 3, 4, 8, 9
Trichohepatoenteric syndrome type 1	TTC37	1513	614589	2, 3, 4, 8, 9, 17
Trichohepatoenteric syndrome type 2	SKIV2L	317	600478	2, 3, 4, 8, 9, 17
Trichorhinophalangeal syndrome type 1	TRPS1	120	604386	2, 4, 8, 9, 17
Trichothiodystrophy	ERCC2	1552	126340	2, 4, 17
Trichothiodystrophy	ERCC3	1861	133510	2, 4, 17
Trichothiodystrophy	GTF2H5	1755	608780	2, 4
Trichothiodystrophy, nonphotosensitive type 1	MPLKIP	1070	609188	2, 4, 8, 9
Tuftelin deficiency	TUFT1	2721	600087	2, 4
Tylosis with esophageal cancer	RHBDF2	1049	614404	2, 3, 4, 8, 9, 17
UV-sensitive syndrome type 1	ERCC6	1822	609413	2, 3, 4, 8, 9, 17
UV-sensitive syndrome type 3	UVSSA	1036	614632	2, 4, 17
Van Buchem disease	SOST	2427	605740	2, 4
Vasculopathy, infantile-onset, TMEM173/STING related	TMEM173	2723	612374	2, 3, 4, 8, 9
Vitiligo-associated multiple autoimmune disease	NLRP1	2116	606636	2, 3, 4, 8, 9, 17
Vohwinkel syndrome with ichthyosis	LORICRIN	1778	152445	2, 4, 8, 9
Waardenburg syndrome type 1	PAX3	1071	606597	2, 4, 8, 9, 17
Waardenburg syndrome type 2E	SOX10	279	602229	2, 3, 4, 8, 9
Waardenburg syndrome type 4C	SOX10	279	602229	2, 3, 4, 8, 9
WHIM syndrome	CXCR4	2931	162643	2, 4, 8, 9
Winchester Syndrome	MMP14	1935	600754	2, 3, 4, 8, 9
Wolcott-Rallison syndrome	EIF2AK3	1072	604032	2, 4, 17
Wrinkly skin syndrome	ATP6V0A2	2490	611716	2, 4, 17
Xeroderma pigmentosum, group A	XPA	1550	611153	2, 4
Xeroderma pigmentosum, group C	XPC	1551	613208	2, 4, 17
Xeroderma pigmentosum, group D	ERCC2	1552	126340	2, 4, 17
Xeroderma pigmentosum, group E, DDB-negative subtype	DDB2	2542	600811	2, 4
Xeroderma pigmentosum, group F	ERCC4	2543	133520	2, 4, 17
Xeroderma pigmentosum, group G	ERCC5	2167	133530	2, 4, 17
Xeroderma pigmentosum, variant type	POLH	1553	603968	2, 4, 17
XFE progeroid syndrome	ERCC4	2543	133520	2, 4, 17

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› Cardiovascular and Pneumology - Panels

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoCardio™	ABCC9, ACTA2, ACTC1, ACTN2, ACVR2B, ACVRL1, AKAP9, ANK2, ANKRD1, ARH-GAP31, ATM, B3GAT3, BAG3, BCOR, BMPR2, BRAF, CACNA1C, CACNB2, CALM1, CALM2, CASQ2, CAV3, CAVIN4, CBL, CDH2, CFAP53, CFC1, CHD7, CITED2, CLDN16, CLDN19, CNNM2, COL1A1, COL1A2, COL3A1, CO-L4A1, COL4A2, COL5A1, COL5A2, CREBBP, CRELD1, CRYAB, CSRP3, CTNNA3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, EFEMP2, EGF, EHMT1, ELN, EMD, ENG, EP300, EVC, EVC2, EYA4, FBN1, FBN2, FHL1, FKTN, FLNA, FLNC, FOXC1, FOXF1, FOXH1, FXYD2, GAA, GATA4, GATA5, GATA6, GDF1, GDF2, GJA1, GJA5, GLA, GPC3, GPD1L, HCCS, HCN4, HFE, HRAS, HTRA1, ILK, JAG1, JPH2, JUP, KCNA1, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNK3, KCNQ1, KDM6A, KMT2D, KRAS, LAMA4, LAMP2, LDB3, LMNA, LZTR1, MAP2K2, MED12, MED13L, MEIS2, MFAP5, MIB1, MMP21, MMP3, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYO6, MYOZ2, MYPN, NEBL, NEXN, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NOTCH1, NOTCH2, NOTCH3, NPPA, NR2F2, NRAS, NSD1, PDLIM3, PKD1L1, PKD2, PKP2, PLN, PRDM16, PRKAG2, PRKG1, PSEN1, PSEN2, PTPN11, RAF1, RASA1, RBM10, RBM20, RIT1, RYR2, SALL1, SALL4, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SEMA3A, SGCD, SHOC2, SKI, SLC12A3, SLC22A5, SLC25A4, SLC2A10, SLMAP, SMAD3, SMAD4, SMAD6, SMC3, SNTA1, SOS1, SOS2, SOX2, STRA6, SYNE1, SYNE2, TAB2, TAZ, TBX1, TBX20, TBX5, TCAP, TFAP2B, TGFB2, TGFB3, TGFBR1, TGFBR2, TLL1, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TREX1, TRIM63, TRPM4, TRPM6, TTN, TTR, VCL, ZEB2, ZFPM2, ZIC3		5423	6
CentoDx®	Targets exonic regions of >6500 genes asso-ciated with known clinical phenotypes		50158	5
CentoLCV	Genome wide analysis of structural variants and large copy number changes through sWGS		50118	5
Pulmonary panel	ABCA3, ABCC8, ACVRL1, AP3B1, ASCL1, BLOC1S3, BLOC1S6, BMPR1B, BMPR2, CAV1, CCDC39, CCDC40, CFTR, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, CSF2RA, CSF2RB, DKC1, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAL1, DOCK8, DTNBP1, ECE1, EDN3, EFEMP2, EI-F2AK4, ELN, ENG, FBLN5, FBN1, FLCN, FOXF1, GDF2, GDNF, GLRA1, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA3, KCNA5, KCNK3, MECP2, NF1, NFU1, NKX2-1, NME8, NOP10, NOTCH3, PARN, PHOX2A, PHOX2B, POLD1, RAPSN, RASA1, RET, RSPH1, RSPH4A, RSPH9, RTEL1, SARS2, SCN4A, SCNN1A, SCNN1B, SCNN1G, SERPINA1, SFTPA2, SFTPB, SFTPC, SLC6A5, SLC7A7, SMAD4, SMAD9, SMPD1, STAT3, STRA6, TERT, TINF2, TSC1, TSC2, ZEB2		5456	6

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Panel name	Genes	Test code	Available test methods
CentoICU®	AARS1, AARS2, AASS, ABAT, ABCA12, ABCA3, ABCB11, ABCC8, ABCD1, ABCD3, ABCD4, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACTA1, ACY1, ADA, ADAMTS13, ADAMTSL2, ADAR, ADK, ADNP, ADSL, AGA, AGK, AGL, AGPAT2, AGPS, AGRN, AGXT, AHCY, AICDA, AIFM1, AIMP1, AKAP9, AKR1D1, AKT2, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX12B, ALOXE3, ALPL, ALS2, AMACR, AMN, AMPD1, AMT, ANK1, ANKRD26, ANKS6, ANTXR1, ANTXR2, AP2S1, AP4B1, AP4E1, AP4M1, AP4S1, APOB, APTX, ARG1, ARL6, ARSA, ARSB, ARX, ASAH1, ASL, ASNS, ASPA, ASPM, ASS1, ATIC, ATP1A3, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATPAF2, ATR, ATRX, AUH, B3GLCT, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCKDK, BCS1L, BDNF, BICD2, BIN1, BLNK, BOLA3, BRAF, BRAT1, BRCA2, BSCL2, BSND, BTD, BTK, C12orf65, CA12, CACNA1C, CACNB2, CALM1, CAMTA1, CASK, CASR, CAST, CAV1, CAV3, CAVIN1, CBS, CCDC103, CCDC114, CCDC78, CD19, CD247, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD59, CD79A, CD79B, CD81, CD96, CDAN1, CDK5RAP2, CDKL5, CDKN1C, CENPJ, CEP152, CEP290, CERS3, CFAP298, CFH, CFHR3, CFL2, CFTR, CHAT, CHD7, CHKB, CHM, CHRNA1, CHRNB1, CHRND, CHRNE, CIDEC, CLCN1, CLCNKA, CLCNKB, CLDN16, CLN3, CLN5, CLN6, CLN8, CLPB, CNTN1, COA5, COG1, COG6, COG7, COL11A1, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A2, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COMP, COQ2, COQ8A, COQ9, CORO1A, COX10, COX15, COX20, COX6B1, CPS1, CPT1A, CPT2, CR2, CRPPA, CRTAP, CTNS, CTPS1, CTSA, CTSD, CUL4B, CXCR4, CYP11B1, CYP11B2, CYP17A1, CYP4F22, CYP7B1, D2HGDH, DBT, DCLRE1C, DDC, DDOST, DDR2, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DIAPH1, DLAT, DLD, DMD, DNA2, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC19, DNM2, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPM2, DPYD, DRC1, DSP, DST, DUOX2, DUOXA2, DYSF, EDN3, EEF1A2, EGR2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELANE, ENPP1, EPB42, EPCAM, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYA1, EYA4, F10, F11, F13A1, F2, F5, F7, F8, F9, FADD, FAH, FANCA, FANCB, FANCC, FANCD2, FANCL, FARS2, FASTKD2, FBN1, FBP1, FBXL4, FGA, FGB, FGFR2, FGFR3, FGG, FH, FIG4, FKBP14, FKRP, FKTN, FOXC1, FOXG1, FOXP3, FOXRED1, FRAS1, FUCA1, G6PC, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GARS1, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GFAP, GFM1, GFPT1, GJA1, GJB2, GJB4, GK, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLUD1, GLYCTK, GMPPB, GNAS, GNE, GNMT, GNPAT, GNPTAB, GP1BA, GP1BB, GP9, GPC3, GPHN, GPSM2, GSS, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBA1, HBA2, HBB, HCFC1, HESX1, HEXA, HEXB, HGD, HGF, HIBCH, HLCS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPGD, HRAS, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, ICOS, IDUA, IER3IP1, IFIH1, IFT172, IGF1, IGF1R, IGHMBP2, IGLL1, IKBKB, IL12RB1, IL2RA, IL2RG, IL7R, INS, INSR, INVS, IRF8, ITGA2B, ITGA6, ITGA7, ITGB3, ITGB4, IVD, JAG1, JAGN1, JAK3, JAM3, KAT6A, KAT6B, KBTBD13, KCNE1, KCNH1, KCNH2, KCNJ10, KCNJ11, KCNQ1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIF1B, KLF1, KLHL40, KLHL41, KLHL7, KRAS, KRT5, LAMA2, LAMA3, LAMB3, LAMC2, LAMP2, LAMTOR2, LARS2, LAS1L, LCT, LHX3, LHX4, LIAS, LIG4, LIPA, LIPN, LIPT1, LMBRD1, LMNA, LPIN1, LRBA, LRPPRC, LRRC8A, MAGEL2, MAGT1, MALT1, MAN2B1, MANBA, MAP2K1, MAP2K2, MAT1A, MCCC1, MCCC2, MCEE, MCM4, MCPH1, MECP2, MED12, MEF2C, MEGF10, MFN2, MFSD8, MITF, MKKS, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MPC1, MPI, MPL, MPV17, MPZ, MRPL3, MRPL44, MSMO1, MTHFR, MTM1, MTMR14, MTO1, MTR, MTRR, MUSK, MVK, MYCN, MYH9, NAA10, NAGA, NAGS, NALCN, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NDUFV2, NEB, NEU1, NEUROG3, NEXN, NFKB2, NFU1, NGF, NGLY1, NHEJ1, NIPAL4, NIPBL, NKX2-1, NKX2-5, NLRC4, NLRP3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NR0B1, NR3C2, NRAS, NSD1, NSDHL, NUBPL, OAT, OCLN, OCRL, OPA3, OPHN1, ORC1, ORC4, OTC, OXCT1, PAFAH1B1, PAH, PAX2, PAX3, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH19, PCNT, PDCD10, PDE10A, PDHA1, PDHB, PDHX, PDP1, PDSS2, PDX1, PEPD, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGM1, PHGDH, PHKG2, PHOX2B, PIGA, PIGN, PIGT, PIGV, PIK3CD, PKD2, PKHD1, PKLR, PLCB4, PLEC, PLOD1, PLP1, PMM2, PMP22, PNKP, PNP, PNPLA1, PNPO, PNPT1, POGZ, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU1F1, PPT1, PRDM16, PRKAG2, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PSAP, PSAT1, PSPH, PTPN11, PTPRC, PTRH2, PTS, PURA, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RAC2, RAF1, RAG1, RAG2, RANBP2, RAPSN, RARS2, RB1, RBBP8, RBM8A, RET, RFT1, RFX5, RFX6, RIT1, RMND1, RNASEH2C, RNASET2, RORC, RPS19, RRM2B, RXYLT1, RYR1, SALL1, SATB2, SBDS, SCN1A, SCN2A, SCN4A, SCN5A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SELENON, SERAC1, SERPINC1, SERPING1, SFTPB, SFTPC, SHOC2, SIL1, SIX3, SIX5, SKI, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC26A2, SLC26A3, SLC2A1, SLC30A2, SLC33A1, SLC3A1, SLC4A1, SLC52A1, SLC52A3, SLC5A1, SLC5A5, SLC6A1, SLC6A3, SLC6A5, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNAI2, SNX10, SOS1, SOX10, SOX2, SOX9, SPAST, SPEG, SPINK5, SPINT2, SPR, SPRED1, SPTA1, SPTAN1, SPTB, SRD5A3, ST3GAL3, ST3GAL5, STAR, STAT1, STAT3, STIL, STIM1, STS, STT3B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SYNE1, TACO1, TAT, TAZ, TBC1D24, TBCE, TBX19, TBX5, TCAP, TCN2, TFR2, TG, TGM1, TH, THRA, TJP2, TMCO1, TMEM165, STING1, TMEM70, TNFRSF13B, TNFRSF13C, TNFSF4, TNNT1, TP63, TPM2, TPM3, TPO, TPP1, TRIP11, TRMU, TRPV4, TSC1, TSC2, TSFM, TSHB, TSHR, TSPYL1, TTC7A, TTN, TUBA8, TUBB2A, TWNK, UBA1, UGT1A1, UMPS, UNG, UPB1, UQCRC2, UROD, UROS, WAS, WDPCP, WDR62, WDR73, WFS1, WNK1, WT1, ZAP70, ZEB2, ZFP57, ZNF423	5445	5

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Alveolar capillary dysplasia with misalignment of pulmonary veins	FOXF1	2048	601089	2, 4, 8, 9
Arrhythmogenic right ventricular cardiomyopathy type 1	TGFB3	1301	190230	2, 4, 8, 9, 17
Arrhythmogenic right ventricular cardiomyopathy type 5	TMEM43	1302	612048	2, 4, 17
Arrhythmogenic right ventricular cardiomyopathy type 8	DSP	1297	125647	2, 4, 8, 9, 17
Arrhythmogenic right ventricular cardiomyopathy type 9	PKP2	1303	602861	2, 4, 8, 9, 17
Arrhythmogenic right ventricular cardiomyopathy type 10	DSG2	1304	125671	2, 4, 8, 9, 17
Arrhythmogenic right ventricular cardiomyopathy type 11	DSC2	1305	125645	2, 4, 8, 9, 17
Arrhythmogenic right ventricular cardiomyopathy type 12	JUP	1306	173325	2, 4
Arrhythmogenic right ventricular dysplasia type 2	RYR2	1100	180902	2, 17
Atrial fibrillation type 3	KCNQ1	148	607542	2, 4, 8, 9, 17
Atrial fibrillation type 4	KCNE2	144	603796	2, 4, 8, 9
Atrial fibrillation type 6	NPPA	1308	108780	2, 4
Atrial fibrillation type 7	KCNA5	1307	176267	2, 4
Atrial fibrillation type 10	SCN5A	257	600163	2, 4, 8, 9, 17
Atrial fibrillation type 11	GJA5	59	121013	2, 4
Atrial fibrillation type 12	ABCC9	1082	601439	2, 17
Atrial septal defect type 3	MYH6	39	160710	2, 4, 17
Atrial septal defect type 4	TBX20	2891	606061	2, 3, 4, 8, 9, 17
Atrial septal defect type 5	ACTC1	7	102540	2, 4
Atrial septal defect type 6	TLL1	3348	606742	17
Atrial septal defect type 8	CITED2	2890	602937	2, 4
Atrial septal defect type 9	GATA6	1040	601656	2, 3, 4, 8, 9, 17
Atrioventricular septal defect type 4	GATA4	1209	600576	2, 4, 8, 9, 17
Atrioventricular septal defect type 5	GATA6	1040	601656	2, 3, 4, 8, 9, 17
Barth syndrome	TAZ	1309	300394	2, 3, 4, 8, 9
Bicuspid aortic valve	TIMP1	2224	305370	2, 4
Brugada syndrome type 1	SCN5A	257	600163	2, 4, 8, 9, 17
Brugada syndrome type 2	GPD1L	1073	611778	2, 4, 17
Brugada syndrome type 3	CACNA1C	1074	114205	2, 3, 8, 17
Brugada syndrome type 4	CACNB2	1075	600003	2, 4, 17
Brugada syndrome type 5	SCN1B	254	600235	2, 3, 4, 8, 9
Brugada syndrome type 6	KCNE3	1076	604433	2, 4
Brugada syndrome type 7	SCN3B	1310	608214	2, 4
Brugada syndrome type 8	HCN4	116	605206	2, 4, 17
Brugada syndrome type 9	SLMAP	1545	602701	2, 4, 17
Cardiac defects, CNOT3 related	CNOT3	2287	604910	2, 4, 17
Cardiac defects, PPP1R8 related	PPP1R8	2352	602636	2, 4
Cardiac valvular dysplesia, X-linked	FLNA	803	300017	2, 3, 4, 8, 9, 17
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency	SCO2	1425	604272	2, 4, 8, 9
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency type 2	COX15	1368	603646	2, 3, 4, 8, 9, 17
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency type 3	COA5	2833	613920	2, 4
Cardiofaciocutaneous syndrome	BRAF	1078	164757	2, 4, 17
Cardiofaciocutaneous syndrome	KRAS	152	190070	2, 3, 4, 8, 9
Cardiofaciocutaneous syndrome type 3	MAP2K1	161	176872	2, 4, 17
Cardiofaciocutaneous syndrome type 4	MAP2K2	162	601263	2, 3, 4, 8, 9, 17
Cardiomyopathy, apical hypertrophic, and neuropathy, MT-ATP8 related	MT-ATP8	2554	516070	4
Cardiomyopathy, dilated	MYBPC3	174	600958	2, 4, 8, 9, 17
Cardiomyopathy, dilated type 1A	LMNA	158	150330	2, 4, 8, 9, 17
Cardiomyopathy, dilated type 1AA	ACTN2	1683	102573	2, 4, 17
Cardiomyopathy, dilated type 1BB	DSG2	1304	125671	2, 4, 8, 9, 17
Cardiomyopathy, dilated type 1C	LDB3	157	605906	2, 4, 17
Cardiomyopathy, dilated type 1CC	NEXN	1684	613121	2, 4, 17
Cardiomyopathy, dilated type 1D	TNNT2	303	191045	2, 4, 8, 9, 17
Cardiomyopathy, dilated type 1DD	RBM20	1685	613171	2, 4, 17
Cardiomyopathy, dilated type 1E	SCN5A	257	600163	2, 4, 8, 9, 17
Cardiomyopathy, dilated type 1EE	MYH6	39	160710	2, 4, 17
Cardiomyopathy, dilated type 1G	TTN	309	188840	2, 17
Cardiomyopathy, dilated type 1GG	SDHA	1188	600857	2, 3, 4, 8, 9, 17
Cardiomyopathy, dilated type 1HH	BAG3	30	603883	2, 4, 8, 9, 17
Cardiomyopathy, dilated type 1I	DES	741	125660	2, 3, 4, 8, 9, 17
Cardiomyopathy, dilated type 1J	EYA4	973	603550	2, 3, 4, 8, 9, 17
Cardiomyopathy, dilated type 1KK	MYPN	2803	608517	2, 4, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Cardiomyopathy, dilated type 1L	SGCD	265	601411	2, 4, 8, 9, 17
Cardiomyopathy, dilated type 1LL	PRDM16	2258	605557	2, 4, 17
Cardiomyopathy, dilated type 1M	CSRP3	1081	600824	2, 4
Cardiomyopathy, dilated type 1N	TCAP	289	604488	2, 4, 8, 9
Cardiomyopathy, dilated type 1O	ABCC9	1082	601439	2, 17
Cardiomyopathy, dilated type 1P	PLN	1083	172405	2, 4
Cardiomyopathy, dilated type 1R	ACTC1	7	102540	2, 4
Cardiomyopathy, dilated type 1S	MYH7	176	160760	2, 4, 8, 9, 17
Cardiomyopathy, dilated type 1T	TMPO	1084	188380	2, 4, 17
Cardiomyopathy, dilated type 1U	PSEN1	233	104311	2, 4, 8, 9, 17
Cardiomyopathy, dilated type 1V	PSEN2	234	600759	2, 3, 4, 8, 9, 17
Cardiomyopathy, dilated type 1W	VCL	1085	193065	2, 4, 17
Cardiomyopathy, dilated type 1X	FKTN	709	607440	2, 4, 8, 9, 17
Cardiomyopathy, dilated type 1Y	TPM1	305	191010	2, 4, 17
Cardiomyopathy, dilated type 1Z	TNNC1	1087	191040	2, 4
Cardiomyopathy, dilated type 1	CRYAB	690	123590	2, 4
Cardiomyopathy, dilated type 2A	TNNI3	302	191044	2, 4
Cardiomyopathy, dilated type 2B	GATAD1	1686	614518	2, 4
Cardiomyopathy, dilated type 3B	DMD	726	300377	2, 4, 8
Cardiomyopathy, dilated with ataxia	DNAJC19	2298	608977	2, 4
Cardiomyopathy, dilated with hypergonadotropic hypogonadism	LMNA	158	150330	2, 4, 8, 9, 17
Cardiomyopathy, dilated with woolly hair and keratoderma	DSP	1297	125647	2, 4, 8, 9, 17
Cardiomyopathy, familial hypertrophic	CAV3	701	601253	2, 4, 8, 9
Cardiomyopathy, familial hypertrophic type 1	MYH7	176	160760	2, 4, 8, 9, 17
Cardiomyopathy, familial hypertrophic type 2	TNNT2	303	191045	2, 4, 8, 9, 17
Cardiomyopathy, familial hypertrophic type 3	TPM1	305	191010	2, 4, 17
Cardiomyopathy, familial hypertrophic type 4	MYBPC3	174	600958	2, 4, 8, 9, 17
Cardiomyopathy, familial hypertrophic type 6	PRKAG2	226	602743	2, 3, 4, 8, 9, 17
Cardiomyopathy, familial hypertrophic type 7	TNNI3	302	191044	2, 4
Cardiomyopathy, familial hypertrophic type 8	MYL3	1090	160790	2, 4
Cardiomyopathy, familial hypertrophic type 9	TTN	309	188840	2, 17
Cardiomyopathy, familial hypertrophic type 10	MYL2	1091	160781	2, 4
Cardiomyopathy, familial hypertrophic type 11	ACTC1	7	102540	2, 4
Cardiomyopathy, familial hypertrophic type 12	CSRP3	1081	600824	2, 4
Cardiomyopathy, familial hypertrophic type 16	MYOZ2	1688	605602	2, 4
Cardiomyopathy, familial hypertrophic type 17	JPH2	1687	605267	2, 4, 17
Cardiomyopathy, familial hypertrophic type 19	CALR3	1689	611414	2, 4, 17
Cardiomyopathy, familial restrictive type 1	TNNI3	302	191044	2, 4
Cardiomyopathy, fatal, MT-TI related	MT-TI	1312	590045	4
Cardiomyopathy, hypertrophic, midventricular, digenic	MYLK2	1690	606566	2, 4, 17
Cardiomyopathy, hypertrophic, MT-TG related	MT-TG	2562	590035	4
Cardiomyopathy, hypertrophic, type 18	PLN	1083	172405	2, 4
Cardiomyopathy, idiopathic dilated, mitochondrial, MT-TH related	MT-TH	1311	590040	4
Cardiomyopathy, infantile hypertrophic, MT-ATP8 related	MT-ATP8	2554	516070	4
Cardiomyopathy, left ventricular noncompaction, MYH7B related	MYH7B	2615	609928	2, 4
Central hypoventilation syndrome with or without Hirschsprung disease	PHOX2B	740	603851	2, 3, 4, 8, 9, 10
Central hypoventilation syndrome, congenital	ASCL1	712	100790	2, 4, 8, 9
Congenital heart defects and ectodermal dysplasia	PRKD1	3795	605435	17
Congenital heart defects multiple types	GATA5	3781	611496	17
Congenital heart defects multiple types	TAB2	2021	605101	2, 4, 17
Coronary heart disease, susceptibility to, type 6	MMP3	2469	185250	2, 4, 17
CR1 deficiency	CR1	3774	120620	17
Danon disease	LAMP2	155	309060	2, 3, 4, 8, 9, 17
Diaphragmatic hernia type 3	ZFPM2	2283	603693	2, 4, 17
Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis	DSP	1297	125647	2, 4, 8, 9, 17
Dopamine beta-hydroxylase (DBH) deficiency	DBH	1509	609312	2, 4, 17
Familial atrial fibrillation type 13	SCN1B	254	600235	2, 3, 4, 8, 9
Heart block, progressive, familial, type 1A	SCN5A	257	600163	2, 4, 8, 9, 17
Heart-hand syndrome, Slovenian type	LMNA	158	150330	2, 4, 8, 9, 17
Jervell and Lange-Nielsen syndrome type 1	KCNQ1	148	607542	2, 4, 8, 9, 17
Jervell and Lange-Nielsen syndrome type 2	KCNE1	143	176261	2, 4, 8, 9
Left ventricular noncompaction 1, with or without congenital heart defects	DTNA	3775	601239	17
Left ventricular noncompaction 7	MIB1	3788	608677	17
Legionnaire disease, susceptibility to	TLR5	3810	603031	17

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Long QT syndrome type 1	KCNQ1	148	607542	2, 4, 8, 9, 17
Long QT syndrome type 2	KCNH2	145	152427	2, 4, 8, 9, 17
Long QT syndrome type 3	SCN5A	257	600163	2, 4, 8, 9, 17
Long QT syndrome type 4	ANK2	1093	106410	2, 3, 4, 8, 9, 17
Long QT syndrome type 5	KCNE1	143	176261	2, 4, 8, 9
Long QT syndrome type 6	KCNE2	144	603796	2, 4, 8, 9
Long QT syndrome type 8	CACNA1C	1074	114205	2, 3, 8, 17
Long QT syndrome type 9	CAV3	701	601253	2, 4, 8, 9
Long QT syndrome type 10	SCN4B	1096	608256	2, 4
Long QT syndrome type 11	AKAP9	1097	604001	2, 17
Long QT syndrome type 12	SNTA1	1098	601017	2, 4, 17
Long QT syndrome type 13	KCNJ5	1099	600734	2, 4
Long QT syndrome type 15	CALM2	1549	114182	2, 3, 4, 8, 9
Marfan syndrome	FBN1	950	134797	2, 8, 17
MASS syndrome	FBN1	950	134797	2, 8, 17
McKusick-Kaufman syndrome	MKKS	166	604896	2, 3, 4, 8, 9
MELAS syndrome	MT-TC	2560	590020	4
MELAS syndrome	MT-TF	987	590070	4
MELAS syndrome, MT-TL1 related	MT-TL1	1317	590050	4
MERRF/MELAS overlap syndrome, MT-TS1 related	MT-TS1	1319	590080	4
MERRF/MELAS overlap syndrome, MT-TS2 related	MT-TS2	1320	590085	4
Mitochondrial myopathy and sideroblastic anemia type 1	PUS1	44	608109	2, 4
Mitochondrial myopathy, infantile, transient, MT-TE related	MT-TE	2564	590025	4
Mitochondrial myopathy, isolated	MT-TD	1321	590015	4
Mitochondrial myopathy, MT-TA related	MT-TA	2558	590000	4
Mitochondrial myopathy, MT-TM related	MT-TM	1323	590065	4
Mitochondrial phosphate carrier deficiency	SLC25A3	3803	600370	17
Moyamoya disease type 5	ACTA2	6	102620	2, 3, 4, 8, 9, 17
Multisystemic smooth muscle dysfunction syndrome	ACTA2	6	102620	2, 3, 4, 8, 9, 17
Myopathy, MT-TQ related	MT-TQ	1322	590030	4
Pancreatic agenesis and congenital heart defects	GATA6	1040	601656	2, 3, 4, 8, 9, 17
Progressive familial heart block	TRPM4	1544	606936	2, 4, 17
Pulmonary fibrosis, idiopathic	SFTPA1	2055	178630	2, 4
Pulmonary newborn hypertension	CRHR1	1325	122561	2, 4, 8, 9, 17
Pulmonary venoocclusive disease type 2	EIF2AK4	2942	609280	2, 4, 17
Sengers syndrome	AGK	1092	610345	2, 3, 4, 8, 9, 17
Short QT syndrome type 1	KCNH2	145	152427	2, 4, 8, 9, 17
Short QT syndrome type 2	KCNQ1	148	607542	2, 4, 8, 9, 17
Short QT syndrome type 3	KCNJ2	147	600681	2, 4, 8, 9
Sick sinus syndrome type 1	SCN5A	257	600163	2, 4, 8, 9, 17
Sick sinus syndrome type 3	MYH6	39	160710	2, 4, 17
Sinoatrial node dysfunction and deafness	CACNA1D	2796	114206	17
Sudden infant death syndrome, susceptibility to	SCN5A	257	600163	2, 4, 8, 9, 17
Sudden infant death with dysgenesis of the testes syndrome	TSPYL1	2871	604714	2, 4
Testicular anomalies with or without congenital heart disease	GATA4	1209	600576	2, 4, 8, 9, 17
Tetralogy of Fallot	ALDH1A2	2135	603687	2, 4, 17
Tetralogy of Fallot	GATA4	1209	600576	2, 4, 8, 9, 17
Tetralogy of Fallot	GATA6	1040	601656	2, 3, 4, 8, 9, 17
Tetralogy of Fallot	ZFPM2	2283	603693	2, 4, 17
Thoracic aortic aneurysm dissection	SMAD2	3178	601366	2, 4, 17
Transposition of the great arteries, dextro-looped 1	MED13L	2912	608771	2, 4, 17
Ventricular fibrillation, paroxysmal familial type 1	SCN5A	257	600163	2, 4, 8, 9, 17
Ventricular septal defect type 1	GATA4	1209	600576	2, 4, 8, 9, 17
Ventricular septal defect type 2	CITED2	2890	602937	2, 4
Ventricular tachycardia, catecholaminergic polymorphic type 1	RYR2	1100	180902	2, 17
Ventricular tachycardia, catecholaminergic polymorphic type 2	CASQ2	1101	114251	2, 4, 17
Ventricular tachycardia, catecholaminergic polymorphic type 4	CALM1	1548	114180	2, 3, 4, 8, 9, 17
Ventricular tachycardia, catecholaminergic polymorphic type 5	TRDN	2381	603283	2, 17
Wolff -Parkinson-White syndrome	PRKAG2	226	602743	2, 3, 4, 8, 9, 17

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17: Single Gene Sequencing + CNV (NGS)

› Vascular Diseases - Panels

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoDx®	Targets exonic regions of >6500 genes associated with known clinical phenotypes		50158	5
CentoLCV	Genome wide analysis of structural variants and large copy number changes through sWGS		50118	5

› Vascular Diseases

Disease	Gene	Test code	OMIM Gene	Available test methods
Angioedema, hereditary	SERPING1	1105	606860	2, 4, 8, 9, 13
Antithrombin III deficiency	SERPINC1	1860	107300	2, 4, 8, 9, 17
Aortic aneurysm, familial thoracic type 3	TGFBR2	295	190182	2, 3, 4, 8, 9, 17
Aortic aneurysm, familial thoracic type 4	MYH11	1102	160745	2, 17
Aortic aneurysm, familial thoracic type 5	TGFBR1	294	190181	2, 4, 8, 9, 17
Aortic aneurysm, familial thoracic type 6	ACTA2	6	102620	2, 3, 4, 8, 9, 17
Aortic aneurysm, familial thoracic type 7	MYLK	1104	600922	2, 4, 17
Aortic aneurysm, familial thoracic type 8	PRKG1	2688	176894	2, 4, 17
Aortic aneurysm, familial thoracic, MAT2A related	MAT2A	2487	601468	2, 4, 17
Aortic valve disease type 1	NOTCH1	1449	190198	2, 3, 4, 8, 9, 17
Aortic valve disease type 2	SMAD6	1450	602931	2, 3, 4, 8, 9
Arterial calcification type 1, generalized, infantile	ENPP1	2236	173335	2, 3, 4, 8, 9, 17
Arterial calcification type 2, generalized, infantile	ABCC6	2334	603234	2, 4, 8, 9, 17
Arterial Tortuosity Syndrome	SLC2A10	272	606145	2, 3, 4, 8, 9, 17
Atherosclerosis, SOAT1 related	SOAT1	2323	102642	2, 4
Bernard Soulier syndrome type A1	GP1BA	67	606672	2, 4
Bernard Soulier syndrome type A2	GP1BA	67	606672	2, 4
Bernard Soulier syndrome type B	GP1BB	68	138720	2, 4
Bernard Soulier syndrome type C	GP9	69	173515	2, 4
CADASIL	NOTCH3	188	600276	2, 4, 8, 9, 17
Capillary malformation-arteriovenous malformation	RASA1	2232	139150	2, 4, 8, 9, 17
CARASIL	HTRA1	138	602194	2, 3, 4, 8, 9, 17
Carotid intimal medial thickness type 1	PPARG	2566	601487	2, 4, 8, 9, 17
Cerebral cavernous malformations type 1	KRIT1	52	604214	2, 4, 8, 9, 17
Cerebral cavernous malformations type 2	CCM2	53	607929	2, 4, 8, 9, 17
Cerebral cavernous malformations type 3	PDCD10	54	609118	2, 4, 8, 9
Coarctation of the aorta	MCTP2	2246		2, 4, 17
Coronary artery disease in familial hypercholesterolemia, protection against	ABCA1	2098	600046	2, 4, 17
Fabry disease	GLA	373	300644	2, 4, 8, 9, 12, 13
Factor II deficiency	F2	374	176930	2, 4, 17
Factor VII deficiency	F7	2385	613878	2, 4, 8, 9, 17
Glycoprotein Ia C807T polymorphism	ITGA2	1110	192974	2, 4, 17
Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	JAM3	2771	606871	2, 4, 17
Homocystinuria	MTHFR	1112	607093	2, 4, 8, 9, 17
Homocystinuria due to cystathionine beta-synthase deficiency	CBS	1111	613381	2, 3, 4, 8, 9, 17
Homocystinuria-megaloblastic anemia, cbl E type	MTRR	982	602568	2, 4, 17
Hypertension early onset	NR3C2	82	600983	2, 3, 4, 8, 9, 17
Hypertension, ADD2 related	ADD2	2107	102681	2, 4
Hypertension, salt-sensitive essential, susceptibility to	CYP3A5	2518	605325	2, 4, 8, 9, 17
Hypertension, susceptibility to	NOS2	3792	163730	17
Loeys-Dietz syndrome type 4	TGFB2	889	190220	2, 3, 4, 8, 9, 17
Moyamoya disease type 2, susceptibility to	RNF213	119	613768	2, 3, 8, 17
Moyamoya type 6 with achalasia	GUCY1A3	2809	139396	2, 4
Myoglobinuria acute recurrent	LPIN1	562	605518	2, 3, 4, 8, 9, 17
Myopathy with lactic acidosis hereditary	ISCU	667	611911	2, 4
Orthostatic intolerance	SLC6A2	2075	163970	2, 4, 17
Parkes Weber syndrome	RASA1	2232	139150	2, 4, 8, 9, 17
Plasminogen activator inhibitor type 1	SERPINE1	1114	173360	2, 4
Polyarteritis nodosa, childhood-onset	ADA2	2256	607575	2, 3, 4, 8, 9
Protein C Deficiency, AD	PROC	1115	612283	2, 4, 8, 9, 17
Protein S Deficiency, autosomal dominant	PROS1	1116	176880	2, 4, 8, 9, 17
Pseudohypoaldosteronism type 2D	KLHL3	377	605775	2, 4, 17

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8: Deletion/Duplication with qPCR/MLPA
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Disease	Gene	Test code	OMIM Gene	Available test methods
Pseudohypoaldosteronism type 2E	CUL3	802	603136	2, 4, 17
Pulmonary hypertension, primary type	BMPR2	2290	600799	2, 4, 8, 9, 17
Pulmonary venoocclusive disease type 1	BMPR2	2290	600799	2, 4, 8, 9, 17
Sneddon syndrome	ADA2	2256	607575	2, 3, 4, 8, 9
Stormorken syndrome	STIM1	2391	605921	2, 3, 4, 8, 9, 17
Supravalvar aortic stenosis	ELN	2271	130160	2, 4, 8, 9, 17
Thrombophilia due to thrombin defect	F2	374	176930	2, 4, 17
Transposition of the great arteries, dextro-looped 1	MED13L	2912	608771	2, 4, 17
Vascular system defects due to CALCRL deficiency	CALCRL	2612	114190	2, 4
Vascular system defects due to GNA13 deficiency	GNA13	755	604406	2, 4

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Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
Atypical hemolytic uremic syndrome panel	ADAMTS13, C3, CD46, CD59, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, CR1, CR2, DGKE, INF2, MMACHC, MMUT, PIGA, PLG, THBD		5439	6
CentoDx®	Targets exonic regions of >6500 genes associated with known clinical phenotypes		50158	5
CentoLCV	Genome wide analysis of structural variants and large copy number changes through sWGS		50118	5
CentoNephro	ABCB11, ABCB4, ABCC2, ACTN4, ACVR2B, AGPS, AHI1, AIPL1, AKR1D1, ALDOB, ALG8, ALPL, ANKH, ANKS6, ANO5, ANOS1, AP2S1, ARHGAP31, ARHGDIA, ARL13B, ARL6, ARMC4, ARSL, ATP6V0A4, ATP6V1B1, ATP8B1, ATR, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BICC1, BMP1, BMPR1B, BSND, C8ORF37, CA2, CABP4, CANT1, CASP10, CASR, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC28B, CCDC39, CCDC40, CCDC65, CCNO, CD2AP, CDKN1C, CENPF, CENPJ, CEP152, CEP164, CEP290, CEP41, CEP63, CFAP298, CFAP53, CFC1, CFTR, CHD7, CHSY1, CILK1, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CNGA3, COL10A1, COL4A1, COL4A3, COL4A4, COL4A5, COL9A3, COMP, COQ2, COQ6, COQ8B, COQ9, CPLANE1, CRB1, CRELD1, CRTAP, CRX, CSPP1, CTNS, CUBN, CUL3, CYP7B1, DDR2, DGKE, DGUOK, DHCR7, DMP1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DUSP6, DYM, DYNC2H1, EBP, EIF2AK3, ENPP1, ESCO2, EVC, EVC2, FAH, FAS, FASLG, FEZF1, FGF17, FGF23, FGF8, FGFR1, FKBP10, FLNB, FLRT3, FSHB, FXYD2, GDF1, GDF5, GFM1, GHR, GLA, GLI2, GLI3, GLIS2, GNA11, GNAS, GNPAT, GNRH1, GNRHR, GUCY2D, HAMP, HESX1, HEXA, HFE, HNF1B, HNF4A, HS6ST1, HSD11B2, HSD3B7, HSPG2, HYDIN, HYLS1, IFITM5, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IL17RD, IMPDH1, INF2, INPP5E, INPPL1, INVS, IQCB1, ITGA3, JAG1, KCNJ1, KCNJ10, KCNJ13, KCNJ5, KIF7, KISS1, KISS1R, KLHL3, LAMB2, LBR, LCA5, LCT, LEFTY2, LEP, LEPR, LHB, LHX3, LHX4, LIFR, LMX1B, LRAT, LRP5, LRRC6, LZTFL1, MAGI2, MATN3, MCEE, MERTK, MKKS, MKS1, MMAA, MMAB, MMADHC, MMP13, MMP21, MMP9, MMUT, MPV17, MYH9, MYO7A, NBAS, NEK1, NEK8, NEUROG3, NKX2-5, NKX3-2, NME8, NMNAT1, NODAL, NOG, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR0B1, NR0B2, NR3C2, NSDHL, NSMF, OCRL, OFD1, OTX2, P3H1, PAX2, PCSK1, PDE6D, PDSS2, PEX1, PEX10, PEX12, PEX2, PEX26, PEX5, PEX6, PEX7, PHEX, PHF6, PKD1L1, PKD2, PKHD1, PLCE1, PLOD2, PMM2, PNPLA6, POLG, POLR3B, POMC, POU1F1, PPARG, PPIB, PRKAR1A, PROK2, PROKR2, PROM1, PROP1, PRPH2, PTH1R, RBBP8, RD3, RDH12, RDH5, RHO, RLBP1, RMND1, ROR2, RPE65, RPGRIP1, RPGRIP1L, RRM2B, RSPH1, RSPH4A, RSPH9, RUNX2, SALL1, SALL4, SBDS, SCARB2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEMA3A, SERPINA1, SERPINF1, SERPINH1, SLC12A1, SLC12A3, SLC25A13, SLC25A15, SLC26A2, SLC26A3, SLC2A2, SLC34A1, SLC34A3, SLC35D1, SLC4A1, SLC4A4, SLCO1B1, SLCO1B3, SMPD1, SOX10, SOX2, SOX3, SOX9, SPAG1, SPATA7, SPINT2, SPRY4, TAC3, TACR3, TBX5, TCTN1, TCTN2, TCTN3, TFR2, TJP2, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNFRSF11B, TRIM32, TRIP11, TRMU, TRPC6, TRPS1, TRPV4, TSC1, TSC2, TTC21B, TTC8, TULP1, UGT1A1, VHL, VIPAS39, WDPCP, WDR11, WDR19, WDR34, WDR35, WDR60, WDR73, WNK1, WNK4, WNT5A, WNT7A, WT1, ZIC3, ZMYND10, ZNF423		5433	6

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CentoNephro Plus	ABCB11, ABCB4, ABCC2, ACTN4, ACVR2B, AGPS, AHI1, AIPL1, AKR1D1, ALDOB, ALG8, ALPL, ANKH, ANKS6, ANO5, ANOS1, AP2S1, ARHGAP31, ARHGDIA, ARL13B, ARL6, ARMC4, ARSL, ATP6V0A4, ATP6V1B1, ATP8B1, ATR, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BICC1, BMP1, BMPR1B, BSND, C8orf37, CA2, CABP4, CANT1, CASP10, CASR, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC28B, CCDC39, CCDC40, CCDC65, CCNO, CD2AP, CDKN1C, CENPF, CENPJ, CEP152, CEP164, CEP290, CEP41, CEP63, CFAP298, CFAP53, CFC1, CFTR, CHD7, CHSY1, CILK1, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CNGA3, COL10A1, COL4A1, COL4A3, COL4A4, COL4A5, COL9A3, COMP, COQ2, COQ6, COQ8B, COQ9, CPLANE1, CRB1, CRELD1, CRTAP, CRX, CSPP1, CTNS, CUBN, CUL3, CYP7B1, DDR2, DGKE, DGUOK, DHCR7, DMP1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DUSP6, DYM, DYNC2H1, EBP, EIF2AK3, ENPP1, ESCO2, EVC, EVC2, FAH, FAS, FASLG, FEZF1, FGF17, FGF23, FGF8, FGFR1, FKBP10, FLNB, FLRT3, FSHB, FXYD2, GDF1, GDF5, GFM1, GHR, GLA, GLI2, GLI3, GLIS2, GNA11, GNAS, GNPAT, GNRH1, GNRHR, GUCY2D, HAMP, HESX1, HEXA, HFE, HNF1B, HNF4A, HS6ST1, HSD11B2, HSD3B7, HSPG2, HYDIN, HYLS1, IFITM5, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IL17RD, IMPDH1, INF2, INPP5E, INPPL1, INVS, IQCB1, ITGA3, JAG1, KCNJ1, KCNJ10, KCNJ13, KCNJ5, KIF7, KISS1, KISS1R, KLHL3, LAMB2, LBR, LCA5, LCT, LEFTY2, LEP, LEPR, LHB, LHX3, LHX4, LIFR, LMX1B, LRAT, LRP5, LRRC6, LZTFL1, MAGI2, MATN3, MCEE, MERTK, MKKS, MKS1, MMAA, MMAB, MMADHC, MMP13, MMP21, MMP9, MMUT, MPV17, MYH9, MYO7A, NBAS, NEK1, NEK8, NEUROG3, NKX2-5, NKX3-2, NME8, NMNAT1, NODAL, NOG, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR0B1, NR0B2, NR3C2, NSDHL, NSMF, OCRL, OFD1, OTX2, P3H1, PAX2, PCSK1, PDE6D, PDSS2, PEX1, PEX10, PEX12, PEX2, PEX26, PEX5, PEX6, PEX7, PHEX, PHF6, PKD1, PKD1L1, PKD2, PKHD1, PLCE1, PLOD2, PMM2, PNPLA6, POLG, POLR3B, POMC, POU1F1, PPARG, PPIB, PRKAR1A, PROK2, PROKR2, PROM1, PROP1, PRPH2, PTH1R, RBBP8, RD3, RDH12, RDH5, RHO, RLBP1, RMND1, ROR2, RPE65, RPGRIP1, RPGRIP1L, RRM2B, RSPH1, RSPH4A, RSPH9, RUNX2, SALL1, SALL4, SBDS, SCARB2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEMA3A, SERPINA1, SERPINF1, SERPINH1, SLC12A1, SLC12A3, SLC25A13, SLC25A15, SLC26A2, SLC26A3, SLC2A2, SLC34A1, SLC34A3, SLC35D1, SLC4A1, SLC4A4, SLCO1B1, SLCO1B3, SMPD1, SOX10, SOX2, SOX3, SOX9, SPAG1, SPATA7, SPINT2, SPRY4, TAC3, TACR3, TBX5, TCTN1, TCTN2, TCTN3, TFR2, TJP2, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNFRSF11B, TRIM32, TRIP11, TRMU, TRPC6, TRPS1, TRPV4, TSC1, TSC2, TTC21B, TTC8, TULP1, UGT1A1, VHL, VIPAS39, WDPCP, WDR11, WDR19, WDR34, WDR35, WDR60, WDR73, WNK1, WNK4, WNT5A, WNT7A, WT1, ZIC3, ZMYND10, ZNF423		5463	6
Congenital adrenal hyperplasia panel	CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, PRKAR1A, STAR	CYP21A2	5460	6
Pancreatitis panel	ABCB4, APC, APOA5, APOC2, ATM, BMPR1A, BRCA1, BRCA2, CASR, CDKN2A, CFTR, CPA1, CTRC, EPCAM, GPIHBP1, LPL, MEN1, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SMAD4, SPINK1, STK11, TP53, UBR1, VHL		5454	6

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Panel name	Genes	Test code	Available test methods
CentoICU®	AARS1, AARS2, AASS, ABAT, ABCA12, ABCA3, ABCB11, ABCC8, ABCD1, ABCD3, ABCD4, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACTA1, ACY1, ADA, ADAMTS13, ADAMTSL2, ADAR, ADK, ADNP, ADSL, AGA, AGK, AGL, AGPAT2, AGPS, AGRN, AGXT, AHCY, AICDA, AIFM1, AIMP1, AKAP9, AKR1D1, AKT2, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX12B, ALOXE3, ALPL, ALS2, AMACR, AMN, AMPD1, AMT, ANK1, ANKRD26, ANKS6, ANTXR1, ANTXR2, AP2S1, AP4B1, AP4E1, AP4M1, AP4S1, APOB, APTX, ARG1, ARL6, ARSA, ARSB, ARX, ASAH1, ASL, ASNS, ASPA, ASPM, ASS1, ATIC, ATP1A3, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATPAF2, ATR, ATRX, AUH, B3GLCT, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCKDK, BCS1L, BDNF, BICD2, BIN1, BLNK, BOLA3, BRAF, BRAT1, BRCA2, BSCL2, BSND, BTD, BTK, C12orf65, CA12, CACNA1C, CACNB2, CALM1, CAMTA1, CASK, CASR, CAST, CAV1, CAV3, CAVIN1, CBS, CCDC103, CCDC114, CCDC78, CD19, CD247, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD59, CD79A, CD79B, CD81, CD96, CDAN1, CDK5RAP2, CDKL5, CDKN1C, CENPJ, CEP152, CEP290, CERS3, CFAP298, CFH, CFHR3, CFL2, CFTR, CHAT, CHD7, CHKB, CHM, CHRNA1, CHRNB1, CHRND, CHRNE, CIDEC, CLCN1, CLCNKA, CLCNKB, CLDN16, CLN3, CLN5, CLN6, CLN8, CLPB, CNTN1, COA5, COG1, COG6, COG7, COL11A1, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A2, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COMP, COQ2, COQ8A, COQ9, CORO1A, COX10, COX15, COX20, COX6B1, CPS1, CPT1A, CPT2, CR2, CRPPA, CRTAP, CTNS, CTPS1, CTSA, CTSD, CUL4B, CXCR4, CYP11B1, CYP11B2, CYP17A1, CYP4F22, CYP7B1, D2HGDH, DBT, DCLRE1C, DDC, DDOST, DDR2, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DIAPH1, DLAT, DLD, DMD, DNA2, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC19, DNM2, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPM2, DPYD, DRC1, DSP, DST, DUOX2, DUOXA2, DYSF, EDN3, EEF1A2, EGR2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELANE, ENPP1, EPB42, EPCAM, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYA1, EYA4, F10, F11, F13A1, F2, F5, F7, F8, F9, FADD, FAH, FANCA, FANCB, FANCC, FANCD2, FANCL, FARS2, FASTKD2, FBN1, FBP1, FBXL4, FGA, FGB, FGFR2, FGFR3, FGG, FH, FIG4, FKBP14, FKRP, FKTN, FOXC1, FOXG1, FOXP3, FOXRED1, FRAS1, FUCA1, G6PC, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GARS1, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GFAP, GFM1, GFPT1, GJA1, GJB2, GJB4, GK, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLUD1, GLYCTK, GMPPB, GNAS, GNE, GNMT, GNPAT, GNPTAB, GP1BA, GP1BB, GP9, GPC3, GPHN, GPSM2, GSS, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBA1, HBA2, HBB, HCFC1, HESX1, HEXA, HEXB, HGD, HGF, HIBCH, HLCS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPGD, HRAS, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, ICOS, IDUA, IER3IP1, IFIH1, IFT172, IGF1, IGF1R, IGHMBP2, IGLL1, IKBKB, IL12RB1, IL2RA, IL2RG, IL7R, INS, INSR, INVS, IRF8, ITGA2B, ITGA6, ITGA7, ITGB3, ITGB4, IVD, JAG1, JAGN1, JAK3, JAM3, KAT6A, KAT6B, KBTBD13, KCNE1, KCNH1, KCNH2, KCNJ10, KCNJ11, KCNQ1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIF1B, KLF1, KLHL40, KLHL41, KLHL7, KRAS, KRT5, LAMA2, LAMA3, LAMB3, LAMC2, LAMP2, LAMTOR2, LARS2, LAS1L, LCT, LHX3, LHX4, LIAS, LIG4, LIPA, LIPN, LIPT1, LMBRD1, LMNA, LPIN1, LRBA, LRPPRC, LRRC8A, MAGEL2, MAGT1, MALT1, MAN2B1, MANBA, MAP2K1, MAP2K2, MAT1A, MCCC1, MCCC2, MCEE, MCM4, MCPH1, MECP2, MED12, MEF2C, MEGF10, MFN2, MFSD8, MITF, MKKS, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MPC1, MPI, MPL, MPV17, MPZ, MRPL3, MRPL44, MSMO1, MTHFR, MTM1, MTMR14, MTO1, MTR, MTRR, MUSK, MVK, MYCN, MYH9, NAA10, NAGA, NAGS, NALCN, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NDUFV2, NEB, NEU1, NEUROG3, NEXN, NFKB2, NFU1, NGF, NGLY1, NHEJ1, NIPAL4, NIPBL, NKX2-1, NKX2-5, NLRC4, NLRP3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NR0B1, NR3C2, NRAS, NSD1, NSDHL, NUBPL, OAT, OCLN, OCRL, OPA3, OPHN1, ORC1, ORC4, OTC, OXCT1, PAFAH1B1, PAH, PAX2, PAX3, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH19, PCNT, PDCD10, PDE10A, PDHA1, PDHB, PDHX, PDP1, PDSS2, PDX1, PEPD, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGM1, PHGDH, PHKG2, PHOX2B, PIGA, PIGN, PIGT, PIGV, PIK3CD, PKD2, PKHD1, PKLR, PLCB4, PLEC, PLOD1, PLP1, PMM2, PMP22, PNKP, PNP, PNPLA1, PNPO, PNPT1, POGZ, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU1F1, PPT1, PRDM16, PRKAG2, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PSAP, PSAT1, PSPH, PTPN11, PTPRC, PTRH2, PTS, PURA, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RAC2, RAF1, RAG1, RAG2, RANBP2, RAPSN, RARS2, RB1, RBBP8, RBM8A, RET, RFT1, RFX5, RFX6, RIT1, RMND1, RNASEH2C, RNASET2, RORC, RPS19, RRM2B, RXYLT1, RYR1, SALL1, SATB2, SBDS, SCN1A, SCN2A, SCN4A, SCN5A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SELENON, SERAC1, SERPINC1, SERPING1, SFTPB, SFTPC, SHOC2, SIL1, SIX3, SIX5, SKI, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC26A2, SLC26A3, SLC2A1, SLC30A2, SLC33A1, SLC3A1, SLC4A1, SLC52A1, SLC52A3, SLC5A1, SLC5A5, SLC6A1, SLC6A3, SLC6A5, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNAI2, SNX10, SOS1, SOX10, SOX2, SOX9, SPAST, SPEG, SPINK5, SPINT2, SPR, SPRED1, SPTA1, SPTAN1, SPTB, SRD5A3, ST3GAL3, ST3GAL5, STAR, STAT1, STAT3, STIL, STIM1, STS, STT3B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SYNE1, TACO1, TAT, TAZ, TBC1D24, TBCE, TBX19, TBX5, TCAP, TCN2, TFR2, TG, TGM1, TH, THRA, TJP2, TMCO1, TMEM165, STING1, TMEM70, TNFRSF13B, TNFRSF13C, TNFSF4, TNNT1, TP63, TPM2, TPM3, TPO, TPP1, TRIP11, TRMU, TRPV4, TSC1, TSC2, TSFM, TSHB, TSHR, TSPYL1, TTC7A, TTN, TUBA8, TUBB2A, TWNK, UBA1, UGT1A1, UMPS, UNG, UPB1, UQCRC2, UROD, UROS, WAS, WDPCP, WDR62, WDR73, WFS1, WNK1, WT1, ZAP70, ZEB2, ZFP57, ZNF423	5445	5

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Disease	Gene	Test code	OMIM Gene	Available test methods
17-beta hydroxysteroid dehydrogenase X deficiency	HSD17B10	134	300256	2, 4
Achalasia addisonianism alacrimia syndrome	AAAS	121	605378	2, 3, 4, 8, 9, 17
Acromegaly, predisposition to, due to germline GPR101 mutation	GPR101	2740	300393	2, 4

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Disease	Gene	Test code	OMIM Gene	Available test methods
Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	CYP11A1	2095	118485	2, 4, 17
Adrenocorticotropic hormone deficiency	TBX19	2513	604614	2, 4, 17
Alport syndrome, autosomal recessive	COL4A3	983	120070	2, 3, 4, 8, 9, 17
Alport syndrome, autosomal recessive	COL4A4	984	120131	2, 3, 4, 8, 9, 17
Alport syndrome, X-Linked	COL4A5	985	303630	2, 4, 8, 9, 17
Androgen insensitivity	AR	20	313700	2, 4, 8, 9, 10, 17
Androgen insensitivity, partial, with or without breast cancer	AR	20	313700	2, 4, 8, 9, 10, 17
Androgen-binding protein deficiency	SHBG	2787	182205	2, 4
Arthrogryposis, renal dysfunction, and cholestasis type 2	VIPAS39	2467	613401	2, 3, 4, 8, 9, 17
Bardet-Biedl syndrome type 1	BBS1	31	209901	2, 4, 17
Bardet-Biedl syndrome type 2	BBS2	35	606151	2, 3, 4, 8, 9, 17
Bardet-Biedl syndrome type 3	ARL6	22	608845	2, 4
Bardet-Biedl syndrome type 4	BBS4	36	600374	2, 4, 17
Bardet-Biedl syndrome type 5	BBS5	1120	603650	2, 3, 4, 8, 9, 17
Bardet-Biedl syndrome type 6	MKKS	166	604896	2, 3, 4, 8, 9
Bardet-Biedl syndrome type 7	BBS7	1121	607590	2, 4, 17
Bardet-Biedl syndrome type 8	TTC8	308	608132	2, 4, 17
Bardet-Biedl syndrome type 9	BBS9	1122	607968	2, 4, 17
Bardet-Biedl syndrome type 10	BBS10	32	610148	2, 4
Bardet-Biedl syndrome type 11	TRIM32	307	602290	2, 4, 17
Bardet-Biedl syndrome type 12	BBS12	33	610683	2, 4, 17
Bardet-Biedl syndrome type 13	MKS1	34	609883	2, 3, 4, 8, 9, 17
Bardet-Biedl syndrome type 14	CEP290	642	610142	2, 3, 4, 8, 9
Bardet-Biedl syndrome type 15	WDPCP	1326	613580	2, 4, 17
Bardet-Biedl syndrome, LZTFL1 related	LZTFL1	500	606568	2, 4, 17
Bardet-Biedl syndrome, modifier of, CCDC28B related	CCDC28B	58	610162	2, 4
Bartter syndrome	SLC12A2	1695	600840	2, 4, 17
Bartter syndrome	SLC12A3	1130	600968	2, 4, 8, 9, 17
Bartter syndrome	SLC12A5	1697	606726	2, 4, 17
Bartter syndrome	SLC12A7	1698	604879	2, 4
Bartter syndrome type 1	SLC12A1	1691	600839	2, 3, 4, 8, 9, 17
Bartter syndrome type 2	KCNJ1	1692	600359	2, 4, 8, 9
Bartter syndrome type 3	CLCNKB	1514	602023	2, 4, 8, 9, 17
Bartter syndrome type 4a	BSND	1693	606412	2, 4
Bartter syndrome type 4b	CLCNKA	1694	602024	2, 4
Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia	IARS2	3785	612801	17
Cholestasis, infantile, NR1H4 related	NR1H4	2853	603826	2, 4
Crigler-Najjar syndrome, type 1	UGT1A1	1448	191740	2, 3, 4, 8, 9, 17
Crigler-Najjar syndrome, type 2	UGT1A1	1448	191740	2, 3, 4, 8, 9, 17
Cryptorchidism	RXFP2	2390	606655	2, 4, 17
Cystinosis, nephropathic	CTNS	371	606272	2, 3, 4, 8, 9, 17
Cystinuria	PREPL	1743	609557	2, 4, 8, 9, 17
Cystinuria	SLC3A1	1124	104614	2, 4, 8, 9, 17
Cystinuria	SLC7A9	1125	604144	2, 4, 8, 9, 17
Dent disease	CLCN5	1840	300008	2, 3, 4, 8, 9, 17
Diabetes insipidus, nephrogenic, autosomal	AQP2	19	107777	2, 3, 4, 8, 9
Dubin-Johnson syndrome	ABCC2	1126	601107	2, 3, 4, 8, 9, 17
Endocrine-cerebroosteodysplasia	ICK	3787	612325	17
Epstein syndrome	MYH9	179	160775	2, 4, 8, 9, 17
Estrogen resistance	ESR1	2532	133430	2, 4, 17
Factor XI deficiency	F11	1815	264900	2, 4, 8, 9, 17
Factor XII deficiency	F12	1816	610619	2, 3, 4, 8, 9, 17
Factor XIIIA deficiency	F13A1	2272	134570	2, 3, 4, 8, 9, 17
Fanconi renotubular syndrome type 2	SLC34A1	1776	182309	2, 4, 17
Fanconi-Bickel syndrome	SLC2A2	390	138160	2, 3, 4, 8, 9, 17
Focal segmental glomerulosclerosis and dilated cardiomyopath, MT-TY related	MT-TY	2561	590100	4
Focal segmental glomerulosclerosis type 1	ACTN4	8	604638	2, 4, 17
Focal segmental glomerulosclerosis type 2	TRPC6	1128	603652	2, 4, 17
Focal segmental glomerulosclerosis type 3	CD2AP	1512	604241	2, 4, 17
Focal segmental glomerulosclerosis type 4, susceptibility to	APOL1	2725	603743	2, 4
Focal segmental glomerulosclerosis type 5	INF2	1129	610982	2, 3, 4, 8, 9, 17
Focal segmental glomerulosclerosis type 6	MYO1E	2724	601479	2, 4, 17
Focal segmental glomerulosclerosis type 7	PAX2	1252	167409	2, 3, 4, 8, 9, 17
Focal segmental glomerulosclerosis type 8	ANLN	2726	616027	2, 4, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Focal segmental glomerulosclerosis type 9	CRB2	2727	609720	2, 4
Focal segmental glomerulosclerosis, LAMA5 related	LAMA5	2418	601033	2, 4, 17
FSH releasing protein deficiency	INHBA	2788	147290	2, 4
Gilbert syndrome	UGT1A1	1448	191740	2, 3, 4, 8, 9, 17
Gitelman syndrome	SLC12A3	1130	600968	2, 4, 8, 9, 17
Glomerulocystic kidney disease with hyperuricemia and isosthenuria	UMOD	1139	191845	2, 3, 4, 8, 9, 17
Glucocorticoid deficiency type 4, with or without mineralocorticoid deficiency	NNT	3148	607878	17
Glucocorticoid resistance, generalized	NR3C1	2627	138040	2, 3, 4, 8, 9, 17
Glycine encephalopathy	AMT	1455	238310	2, 4, 8, 9, 17
Glycine encephalopathy	GLDC	1457	238300	2, 4, 8, 9, 17
Glycine N-methyltransferase deficiency	GNMT	2488	606628	2, 4
Goitre, multinodular	KEAP1	2160	606016	2, 4
Growth hormone deficiency	GH1	1517	139250	2, 4, 8, 9
Growth hormone deficiency	GHRHR	1518	139191	2, 4, 8, 9, 17
Growth hormone insensitivity with immunodeficiency	STAT5B	2512	604260	2, 4, 8, 9, 17
Hemolytic uremic syndrome	CFB	1347	138470	2, 4, 17
Hemolytic uremic syndrome	CFH	1213	134370	2, 4, 8, 9, 17
Hemolytic uremic syndrome	CFHR1	1700	134371	2, 4, 8, 9, 17
Hemolytic uremic syndrome	CFHR2	1818	600889	2, 4, 8, 9, 17
Hemolytic uremic syndrome	CFHR3	1819	605336	2, 4, 8, 9, 17
Hemolytic uremic syndrome	CFHR4	1982	605337	2, 4
Hemolytic uremic syndrome	CFHR5	1318	608593	2, 4, 8, 9, 17
Hemolytic uremic syndrome	CFI	1214	217030	2, 4, 8, 9, 17
Hemolytic uremic syndrome	THBD	1287	188040	2, 4
Hemolytic uremic syndrome, atypical type 2, susceptibility to	CD46	1215	120920	2, 4, 8, 9, 17
Hepatic failure, early onset, and neurologic disorder	SCO1	1424	603644	2, 4
Hepatic venoocclusive disease with immunodeficiency	SP110	2398	604457	2, 4, 17
Hyperaldosteronism type 3	KCNJ5	1099	600734	2, 4
Hyperbilirubinemia, familial transient neonatal	UGT1A1	1448	191740	2, 3, 4, 8, 9, 17
Hyperbilirubinemia, Rotor type	SLCO1B1	839	604843	2, 3, 4, 8, 9, 17
Hyperbilirubinemia, Rotor type	SLCO1B3	599	605495	2, 3, 4, 8, 9, 17
Hyperparathyroidism type 1, familial	CDC73	2371	607393	2, 4, 8, 9, 17
Hyperparathyroidism type 2, familial	CDC73	2371	607393	2, 4, 8, 9, 17
Hyperparathyroidism, neonatal severe	CASR	1620	601199	2, 4, 8, 9, 17
Hypertriglyceridemia, transient infantile	GPD1	2821	138420	2, 3, 4, 8, 9
Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis	SARS2	3802	612804	17
Hyperuricemic nephropathy, familial juvenile type 1	UMOD	1139	191845	2, 3, 4, 8, 9, 17
Hypocalcemia, autosomal dominant, with Bartter syndrome	CASR	1620	601199	2, 4, 8, 9, 17
Hypocalciuric hypercalcemia, type 1	CASR	1620	601199	2, 4, 8, 9, 17
Hypogonadotropic hypogonadism	KISS1R	1131	604161	2, 4, 8, 9
Hypogonadotropic hypogonadism	LHB	1774	152780	2, 4
Hypogonadotropic hypogonadism	NSMF	1132	608137	2, 3, 4, 8, 9, 17
Hypogonadotropic hypogonadism type 3 with or without anosmia	PROKR2	229	607123	2, 4, 8, 9
Hypogonadotropic hypogonadism type 7 with or without anosmia	GNRHR	100	138850	2, 4, 8, 9
Hypogonadotropic hypogonadism type 10 with or without anosmia	TAC3	2217	162330	2, 4
Hypogonadotropic hypogonadism type 11 with or without anosmia	TACR3	102	162332	2, 3, 4, 8, 9
Hypogonadotropic hypogonadism type 12 with or without anosmia	GNRH1	101	152760	2, 4, 8, 9
Hypogonadotropic hypogonadism type 15 with or without anosmia	HS6ST1	2632	604846	2, 4, 8, 9
Hypoparathyroidism	PTH	2893	168450	2, 4
Hypoparathyroidism, familial isolated	GCM2	2961	603716	2, 3, 4, 8, 9
Hypoparathyroidism, sensorineural deafness, and renal dysplasia	GATA3	2836	131320	2, 4, 8, 9
Hypoparathyroidism-retardation-dysmorphism syndrome	TBCE	1701	604934	2, 3, 4, 8, 9, 17
Hypophosphatemic rickets	CLCN5	1840	300008	2, 3, 4, 8, 9, 17
Hypothyroidism congenital nongoitrous type 1	TSHR	1470	603372	2, 4, 8, 9, 17
Hypothyroidism congenital nongoitrous type 2, familial	PAX8	1462	167415	2, 4, 8, 9, 17
Hypothyroidism congenital nongoitrous type 4	TSHB	1469	188540	2, 4
Hypothyroidism congenital nongoitrous type 6	THRA	1373	190120	2, 3, 4, 8, 9, 17
Hypothyroidism, isolated, TRHR related	TRHR	2870	188545	2, 4
Hypotonia-cystinuria syndrome	PREPL	1743	609557	2, 4, 8, 9, 17
Hypouricemia, renal type 1	SLC22A12	2295	607096	2, 4
Hypouricemia, renal type 2	SLC2A9	2296	606142	2, 4, 17
Immunodeficiency with natural killer cell deficiency	MCM4	527	602638	2, 4, 17
Immunodeficiency, common variable type 7	CR2	903	120650	2, 4, 17
Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia	MAGT1	160	300715	2, 4, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Infantile liver failure syndrome type 2	NBAS	2605	608025	2, 4, 17
Interstitial nephritis karyomegalic	FAN1	449	613534	2, 4, 17
Intrahepatic cholestasis of pregnancy, NR1H4 related	NR1H4	2853	603826	2, 4
Johanson Blizzard syndrome	UBR1	1471	605981	2, 3, 4, 8, 9, 17
Kallmann syndrome type 1	ANOS1	142	300836	2, 4, 8, 9, 17
Kallmann syndrome type 2	FGFR1	1136	136350	2, 4, 8, 9
Kallmann syndrome type 4	PROK2	228	607002	2, 4, 8, 9
Kallmann syndrome, SEMA3A related	SEMA3A	1123	603961	2, 3, 4, 8, 9, 17
Laron syndrome	GHR	1956	600946	2, 4, 8, 9, 17
Liddle syndrome	SCNN1B	259	600760	2, 3, 4, 8, 9, 17
Liddle syndrome	SCNN1G	260	600761	2, 3, 4, 8, 9, 17
Lipoid congenital adrenal hyperplasia	STAR	1466	600617	2, 3, 4, 8, 9
Lipoprotein glomerulopathy	APOE	18	107741	2, 3, 4, 8, 9
Liver failure transient infantile	TRMU	46	610230	2, 4, 17
Lowe oculocerebrorenal syndrome	OCRL	203	300535	2, 3, 4, 8, 9, 17
LYSINURIC PROTEIN INTOLERANCE	SLC7A7	1929	603593	2, 3, 4, 8, 9, 17
McKusick-Kaufman syndrome	MKKS	166	604896	2, 3, 4, 8, 9
Meckel syndrome type 1	MKS1	34	609883	2, 3, 4, 8, 9, 17
Meckel syndrome type 3	TMEM67	301	609884	2, 4, 17
Meckel syndrome type 4	CEP290	642	610142	2, 3, 4, 8, 9
Meckel syndrome type 8	TCTN2	1407	613846	2, 4, 17
Meckel syndrome type 9	B9D1	1355	614144	2, 4, 17
Meckel syndrome type 10	B9D2	1356	611951	2, 4
Medullary cystic kidney disease type 2	UMOD	1139	191845	2, 3, 4, 8, 9, 17
Menkes disease	ATP7A	25	300011	2, 4, 8, 9, 17
Mirage syndrome	SAMD9	3621	610456	17
Multiple endocrine neoplasia type 1	MEN1	81	613733	2, 4, 8, 9, 17
Nephrogenic syndrome of inapproriate antidiuresis	AVPR2	28	300538	2, 3, 4, 8, 9
Nephrolithiasis type 1	CLCN5	1840	300008	2, 3, 4, 8, 9, 17
Nephrolithiasis/osteoporosis, hypophosphatemic, type 1	SLC34A1	1776	182309	2, 4, 17
Nephrolithiasis/osteoporosis, hypophosphatemic, type 2	SLC9A3R1	2966	604990	2, 4
Nephronophthisis type 1	NPHP1	190	607100	2, 4, 8, 9, 17
Nephronophthisis type 2	INVS	1140	243305	2, 4, 17
Nephronophthisis type 3	NPHP3	1141	608002	2, 4, 17
Nephronophthisis type 4	NPHP4	1142	607215	2, 3, 4, 8, 9, 17
Nephronophthisis type 7	GLIS2	1143	608539	2, 4, 17
Nephronophthisis type 9	NEK8	1391	609799	2, 4, 17
Nephronophthisis type 12	TTC21B	2001	612014	2, 3, 4, 8, 9, 17
Nephronophthisis type 13	WDR19	2424	608151	2, 4, 17
Nephronophthisis type 14	ZNF423	406	604557	2, 4, 17
Nephronophthisis type 15	CEP164	539	614848	2, 3, 4, 8, 9, 17
Nephronophthisis type 16	ANKS6	2070	615370	2, 4, 17
Nephronophthisis type 19	DCDC2	2647	605755	2, 4, 17
Nephronophthisis-like nephropathy type 1	XPNPEP3	1144	613553	2, 4, 17
Nephrosis, Finnish type	NPHS1	191	602716	2, 3, 4, 8, 9, 17
Nephrotic syndrome	NPHS2	1145	604766	2, 3, 4, 8, 9, 17
Nephrotic syndrome type 2	NPHS1	191	602716	2, 3, 4, 8, 9, 17
Nephrotic syndrome type 3	PLCE1	2112	608414	2, 17
Nephrotic syndrome type 5	LAMB2	1702	150325	2, 4, 17
Nephrotic syndrome type 7	DGKE	1533	601440	2, 4, 17
Nephrotic syndrome type 8	ARHGDIA	1703	601925	2, 4
Nephrotic syndrome type 9	COQ8B	2474	615567	2, 4, 17
Obesity	MC4R	1950	155541	2, 4, 8, 9
Obesity due to leptin deficiency	LEP	1951	164160	2, 4, 8, 9
Obesity with adrenal insufficiency and red hair	POMC	222	176830	2, 3, 4, 8, 9
Obesity with impaired prohormone processing	PCSK1	2094	162150	2, 4, 17
Obesity, early-onset, susceptibility to	POMC	222	176830	2, 3, 4, 8, 9
Obesity, severe	PPARG	2566	601487	2, 4, 8, 9, 17
Obesity, susceptibility to, SLC6A14 related	SLC6A14	2700	300444	2, 3, 4, 8, 9, 17
Ovalocytosis	SLC4A1	1704	109270	2, 4, 17
Pancreatitis	CTRC	475	601405	2, 4
Pancreatitis	PRSS1	472	276000	2, 4, 8, 9
Pancreatitis	SPINK1	473	167790	2, 3, 4, 8, 9
Pancreatitis, chronic, early onset	CPA1	2417	114850	2, 4, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Pancreatitis, chronic, protection against	PRSS2	2260	601564	2, 4
Panhypopituitarism, X-linked	SOX3	280	313430	2, 4, 8, 9
Parathyroid adenoma with cystic changes, familial	CDC73	2371	607393	2, 4, 8, 9, 17
Pendred syndrome	SLC26A4	988	605646	2, 4, 8, 9, 17
Pigmented nodular adrenocortical disease type 1, primary	PRKAR1A	1192	188830	2, 3, 4, 8, 9, 17
Pituitary hormone deficiency, combined type 3	LHX3	1146	600577	2, 4, 8, 9, 17
Pituitary hormone deficiency, combined type 4	LHX4	1147	602146	2, 4, 8, 9
Polycystic kidney and hepatic disease	PKHD1	1150	606702	2, 8, 17
Polycystic kidney disease type 1, autosomal dominant	PKD1	1148	601313	2, 3, 4, 8, 9
Polycystic kidney disease type 1, autosomal recessive	PKHD1	1150	606702	2, 8, 17
Polycystic kidney disease type 2, autosomal dominant	PKD2	1149	173910	2, 4, 8, 9, 17
Polycystic liver disease	PRKCSH	2528	177060	2, 4, 17
Polycystic ovary syndrome type 1	SULT2A1	2096	125263	2, 4
Polycystic ovary syndrome type 1	SULT2B1	2097	604125	2, 4
Precocious puberty, male	LHCGR	80	152790	2, 3, 4, 8, 9, 17
Premature ovarian failure type 1	FMR1	588	309550	2, 10
Primary aldosteronism, seizures, and neurologic abnormalities	CACNA1D	2796	114206	17
Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis	CLCN5	1840	300008	2, 3, 4, 8, 9, 17
Pseudohypoaldosteronism, type 1, autosomal dominant	NR3C2	82	600983	2, 3, 4, 8, 9, 17
Pseudohypoaldosteronism, type 1, autosomal recessive	SCNN1A	214	600228	2, 3, 4, 8, 9, 17
Pseudohypoaldosteronism, type 1, autosomal recessive	SCNN1B	259	600760	2, 3, 4, 8, 9, 17
Pseudohypoaldosteronism, type 1, autosomal recessive	SCNN1G	260	600761	2, 3, 4, 8, 9, 17
Pseudohypoaldosteronism, type 2B	WNK4	1705	601844	2, 4, 17
Pseudohypoparathyroidism type 1A	GNAS	1345	139320	2, 4, 8, 9, 17
Pseudohypoparathyroidism type 1B	GNAS	1345	139320	2, 4, 8, 9, 17
Pseudohypoparathyroidism type 1C	GNAS	1345	139320	2, 4, 8, 9, 17
Pseudopseudohypoparathyroidism	GNAS	1345	139320	2, 4, 8, 9, 17
Renal cystic dysplasia, cystic, susceptibility to	BICC1	695	614295	2, 4, 17
Renal dysfunction due to SLC26A1 deficiency	SLC26A1	2176	610130	2, 4
Renal glucosuria	SLC5A2	2731	182381	2, 3, 4, 8, 9, 17
Renal tubular acidosis with deafness	ATP6V1B1	509	192132	2, 3, 4, 8, 9, 17
Renal tubular acidosis, distal, autosomal recessive	ATP6V0A4	1843	605239	2, 3, 4, 8, 9, 17
Renal tubular acidosis, proximal, with ocular abnormalities	SLC4A4	1621	603345	2, 3, 4, 8, 9, 17
Renal tubular acidosis, SLC4A5 related	SLC4A5	1622	606757	2, 3, 4, 8, 9, 17
Renal tubular dysgenesis	ACE	1841	106180	2, 3, 4, 8, 9, 17
Renal tubular dysgenesis	AGT	1830	106150	2, 4
Renal tubular dysgenesis	AGTR1	2471	106165	2, 4
Renal tubular dysgenesis	REN	1152	179820	2, 3, 4, 8, 9
Senior-Loken syndrome type 6	CEP290	642	610142	2, 3, 4, 8, 9
Senior-Loken syndrome type 7	SDCCAG8	1327	613524	2, 4, 17
Senior-Loken syndrome type 8	WDR19	2424	608151	2, 4, 17
SERKAL syndrome	WNT4	122	603490	2, 4, 8, 9
Thromocytopenia-Absent-Radius-Syndrome	RBM8A	238	605313	2, 3, 4, 8, 9
Thyroid dyshormonogenesis type 1	SLC5A5	1465	601843	2, 4, 17
Thyroid dyshormonogenesis type 2A	TPO	1468	606765	2, 4, 8, 9, 17
Thyroid dyshormonogenesis type 3	TG	1467	188450	2, 17
Thyroid dyshormonogenesis type 4	IYD	2347	612025	2, 4
Thyroid dyshormonogenesis type 5	DUOXA2	2346	612772	2, 4
Thyroid dyshormonogenesis type 6	DUOX1	2137	606758	2, 4, 17
Thyroid dyshormonogenesis type 6	DUOX2	2138	606759	2, 4, 17
Thyroid hormone metabolism abnormal	SECISBP2	1863	607693	2, 4, 17
Thyroid hormone resistance	THRB	1403	190160	2, 3, 4, 8, 9
Trifunctional protein deficiency	HADHB	1413	143450	2, 3, 4, 8, 9, 17
Ventriculomegaly with cystic kidney disease	CRB2	2727	609720	2, 4
Vesicoureteral reflux type 2	ROBO2	1153	602431	2, 3, 4, 8, 9
Vesicoureteral reflux type 3	SOX17	1995	610928	2, 4, 8, 9
Wilms tumor type 1, familial	WT1	1558	607102	2, 4, 8, 9, 17
Wilson-Turner syndrome	LAS1L	2752	300964	17
Wolcott-Rallison syndrome	EIF2AK3	1072	604032	2, 4, 17

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› Reproductive Genetics - Panels

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoArrayCyto® 750K	Genome-wide CMA with 750,000 markers		50068	5
CentoArrayCyto® HD	Genome-wide CMA with 2.6 million markers		50001	5
CentoDx®	Targets exonic regions of >6500 genes associated with known clinical phenotypes		50158	5
CentoLCV	Genome wide analysis of structural variants and large copy number changes through sWGS		50118	5
CentoScreen® DUO	Analysis of 330 genes for carrier screening		50143	6
CentoScreen® Paired PACK	Complete panel evaluation of 330 genes for first partner + risk gene analysis for second partner based on the result of first partner		50144	6
CentoScreen® SOLO	Analysis of 330 genes for carrier screening		50145	6
Infertility panel	ANOS1, AR, ARL13B, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BMP15, C8ORF37, CATSPER1, CC2D2A, CCDC28B, CEP164, CEP290, CFTR, CHD7, DUSP6, ENPP1, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FMR1, FSHB, FSHR, GNRH1, GNRHR, HESX1, HEXA, HFE, HS6ST1, IFT172, IFT27, IL17RD, INPP5E, KIF7, KISS1, KISS1R, LEP, LEPR, LHB, LHCGR, LHX3, LHX4, LZTFL1, MKKS, MKS1, MYO7A, NPHP1, NPHP3, NR0B1, NR0B2, NR5A1, NSMF, OFD1, PCSK1, PHF6, PNPLA6, POLR3B, POMC, POU1F1, PPARG, PROK2, PROKR2, PROM1, PROP1, PRPH2, RDH5, RHO, RLBP1, RPGRIP1L, SDCCAG8, SEMA3A, SOX10, SOX2, SOX3, SPRY4, SRY, TAC3, TACR3, TMEM67, TRIM32, TTC21B, TTC8, TUBB8, WDPCP, WDR11, ZP1		5452	6

› Reproductive Genetics

Disease	Gene	Test code	OMIM Gene	Available test methods
46,XX sex reversal type 1	SRY	1880	480000	2, 4, 8, 9
46,XY sex reversal type 8, modifier of	AKR1C4	2659	600451	2, 4
Aromatase deficiency	CYP19A1	1511	107910	2, 3, 4, 8, 9, 17
Azoospermia induced by Y chromosome microdeletions	AZF region	1881		8
Congenital bilateral absence of vas deferens	CFTR	370	602421	2, 4, 8
Cryptorchidism	RXFP2	2390	606655	2, 4, 17
Deafness and male infertility	STRC	958	606440	2, 3, 4, 8, 9
Deafness and male infertility, CATSPER2 related	CATSPER2	1233	607249	2, 4
Disorders of sex development with cleft palate	FOXF2	2049	603250	2, 4, 8, 9
Follicle-stimulating hormone deficiency, isolated	FSHB	1772	136530	2, 4, 8, 9
Guttmacher syndrome	HOXA13	2913	142959	2, 4
Hand-foot-uterus syndrome	HOXA13	2913	142959	2, 4
Hydatidiform mole	NLRP7	1744	609661	2, 3, 4, 8, 9
Hydatidiform mole, recurrent, type 2	KHDC3L	1745	611687	2, 3, 4, 8, 9
Hypogonadotropic hypogonadism	KISS1	1154	603286	2, 4, 8, 9
Hypogonadotropic hypogonadism	NSMF	1132	608137	2, 3, 4, 8, 9, 17
Hypogonadotropic hypogonadism type 6 with or without anosmia	FGF8	1235	600483	2, 3, 4, 8, 9
Hypogonadtropic hypogonadism type 14	WDR11	1447	606417	2, 3, 4, 8, 9, 17
Hypospadias type 1, X-linked	AR	20	313700	2, 4, 8, 9, 10, 17
Hypospadias type 2, X-linked	MAMLD1	1199	300120	2, 3, 4, 8, 9, 17
Leydig cell hypoplasia type 1	LHCGR	80	152790	2, 3, 4, 8, 9, 17
Oligo-astheno-teratozoospermia	NANOS1	1535	608226	2, 4
Oocyte maturation defect	ZP1	2389	195000	2, 4, 17
Oogenesis dysfunction	SOHLH1	474	610224	2, 4
Ovarian dysgenesis type 1	FSHR	1773	136435	2, 3, 4, 8, 9, 17
Ovarian dysgenesis type 2	BMP15	2388	300247	2, 4
Persistent Mullerian duct syndrome type 1	AMH	1833	600957	2, 3, 4, 8, 9
Persistent Mullerian duct syndrome type 2	AMHR2	1834	600956	2, 3, 4, 8, 9, 17
Preeclampsia/eclampsia type 5	CORIN	2613	605236	2, 4, 17
Pregnancy loss, recurrent, C4BPA related	C4BPA	2614	120830	2, 4, 17
Pseudohermaphroditism with gynecomastia	HSD17B3	2088	605573	2, 4
SPGF4	SYCP3	1157	604759	2, 4

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Disease	Gene	Test code	OMIM Gene	Available test methods
SPGF5	AURKC	1158	603495	2, 4
SPGF6	SPATA16	1159	609856	2, 3, 4, 8, 9
SPGF7	CATSPER1	1160	606389	2, 4, 17
SPGF8	NR5A1	1161	184757	2, 4, 8, 9, 17
SPGF9	DPY19L2	1162	613893	2, 4, 17
Testicular anomalies with or without congenital heart disease	GATA4	1209	600576	2, 4, 8, 9, 17

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Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
Blood coagulation panel	ACTN1, ACVRL1, ADAMTS13, ADAMTS2, ANKRD26, AP3B1, BLOC1S3, BLOC1S6, CCM2, CD36, CD40LG, CHST14, COL1A2, COL3A1, COL4A1, CTC1, CYCS, DIAPH1, DTNBP1, EFEMP2, ENG, F10, F11, F13A1, F2, F5, F7, F8, F9, FANCA, FGA, FGB, FGG, FLNA, GATA1, GFI1B, GNE, GP1BA, GP1BB, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, IL2RG, ITGA2B, ITGB3, JAM3, KRIT1, LYST, LYZ, MPL, MYH9, P2RY12, PDCD10, PROC, PROS1, PTPN11, RBM8A, RUNX1, SBDS, SERPINC1, SLC35A1, SLC7A7, SMAD4, STIM1, THBD, VIPAS39, WAS		5435	6
Bone marrow failure / Anemia panel	ABCB7, ACTN1, ADAMTS13, AK2, ALAS2, AMN, ANK1, ANKRD26, AP3B1, ATM, ATRX, BLM, BLOC1S3, BRCA1, BRCA2, BRIP1, C15orf41, CASP10, CBL, CBLIF, CD36, CD40LG, CDAN1, CENPJ, CEP152, CLCN7, CLPB, CSF3R, CTC1, CUBN, CXCR4, CYB5R3, CYCS, DHFR, DIAPH1, DKC1, DTNBP1, ELANE, EPB42, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, G6PC3, G6PD, GATA1, GFI1, GFI1B, GLRX5, GNE, GP1BA, GP1BB, GP9, GPI, GSS, HAX1, HBA1, HBA2, HBB, HFE, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HSPA9, IL2RG, ITGA2B, ITGB3, ITK, JAGN1, KLF1, KRAS, LIG4, LPIN2, LYST, MLH1, MPL, MRE11, MSH2, MSH6, MTR, MTRR, MYH9, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PARN, PC, PDHA1, PDHX, PKLR, PMS2, PRF1, PTPN11, PUS1, RAC2, RAD51C, RBBP8, RBM8A, RIT1, RPL11, RPL15, RPL26, RPL27, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SBDS, SEC23B, SH2D1A, SLC19A2, SLC19A3, SLC25A19, SLC25A38, SLC4A1, SLX4, SPTA1, SPTB, SRP72, STIM1, STX11, STXBP2, TCN2, TERT, TINF2, TP53, TPK1, TRNT1, UBE2T, UNC13D, VPS13B, VPS45, WAS, WRAP53, XIAP, XRCC2, YARS2		5440	6
CentoDx®	Targets exonic regions of >6500 genes associated with known clinical phenotypes		50158	5
CentoLCV	Genome wide analysis of structural variants and large copy number changes through sWGS		50118	5

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Panel name	Genes	Test code	Available test methods
CentoICU®	AARS1, AARS2, AASS, ABAT, ABCA12, ABCA3, ABCB11, ABCC8, ABCD1, ABCD3, ABCD4, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACTA1, ACY1, ADA, ADAMTS13, ADAMTSL2, ADAR, ADK, ADNP, ADSL, AGA, AGK, AGL, AGPAT2, AGPS, AGRN, AGXT, AHCY, AICDA, AIFM1, AIMP1, AKAP9, AKR1D1, AKT2, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX12B, ALOXE3, ALPL, ALS2, AMACR, AMN, AMPD1, AMT, ANK1, ANKRD26, ANKS6, ANTXR1, ANTXR2, AP2S1, AP4B1, AP4E1, AP4M1, AP4S1, APOB, APTX, ARG1, ARL6, ARSA, ARSB, ARX, ASAH1, ASL, ASNS, ASPA, ASPM, ASS1, ATIC, ATP1A3, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATPAF2, ATR, ATRX, AUH, B3GLCT, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCKDK, BCS1L, BDNF, BICD2, BIN1, BLNK, BOLA3, BRAF, BRAT1, BRCA2, BSCL2, BSND, BTD, BTK, C12orf65, CA12, CACNA1C, CACNB2, CALM1, CAMTA1, CASK, CASR, CAST, CAV1, CAV3, CAVIN1, CBS, CCDC103, CCDC114, CCDC78, CD19, CD247, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD59, CD79A, CD79B, CD81, CD96, CDAN1, CDK5RAP2, CDKL5, CDKN1C, CENPJ, CEP152, CEP290, CERS3, CFAP298, CFH, CFHR3, CFL2, CFTR, CHAT, CHD7, CHKB, CHM, CHRNA1, CHRNB1, CHRND, CHRNE, CIDEC, CLCN1, CLCNKA, CLCNKB, CLDN16, CLN3, CLN5, CLN6, CLN8, CLPB, CNTN1, COA5, COG1, COG6, COG7, COL11A1, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A2, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COMP, COQ2, COQ8A, COQ9, CORO1A, COX10, COX15, COX20, COX6B1, CPS1, CPT1A, CPT2, CR2, CRPPA, CRTAP, CTNS, CTPS1, CTSA, CTSD, CUL4B, CXCR4, CYP11B1, CYP11B2, CYP17A1, CYP4F22, CYP7B1, D2HGDH, DBT, DCLRE1C, DDC, DDOST, DDR2, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DIAPH1, DLAT, DLD, DMD, DNA2, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC19, DNM2, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPM2, DPYD, DRC1, DSP, DST, DUOX2, DUOXA2, DYSF, EDN3, EEF1A2, EGR2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELANE, ENPP1, EPB42, EPCAM, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYA1, EYA4, F10, F11, F13A1, F2, F5, F7, F8, F9, FADD, FAH, FANCA, FANCB, FANCC, FANCD2, FANCL, FARS2, FASTKD2, FBN1, FBP1, FBXL4, FGA, FGB, FGFR2, FGFR3, FGG, FH, FIG4, FKBP14, FKRP, FKTN, FOXC1, FOXG1, FOXP3, FOXRED1, FRAS1, FUCA1, G6PC, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GARS1, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GFAP, GFM1, GFPT1, GJA1, GJB2, GJB4, GK, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLUD1, GLYCTK, GMPPB, GNAS, GNE, GNMT, GNPAT, GNPTAB, GP1BA, GP1BB, GP9, GPC3, GPHN, GPSM2, GSS, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBA1, HBA2, HBB, HCFC1, HESX1, HEXA, HEXB, HGD, HGF, HIBCH, HLCS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPGD, HRAS, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, ICOS, IDUA, IER3IP1, IFIH1, IFT172, IGF1, IGF1R, IGHMBP2, IGLL1, IKBKB, IL12RB1, IL2RA, IL2RG, IL7R, INS, INSR, INVS, IRF8, ITGA2B, ITGA6, ITGA7, ITGB3, ITGB4, IVD, JAG1, JAGN1, JAK3, JAM3, KAT6A, KAT6B, KBTBD13, KCNE1, KCNH1, KCNH2, KCNJ10, KCNJ11, KCNQ1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIF1B, KLF1, KLHL40, KLHL41, KLHL7, KRAS, KRT5, LAMA2, LAMA3, LAMB3, LAMC2, LAMP2, LAMTOR2, LARS2, LAS1L, LCT, LHX3, LHX4, LIAS, LIG4, LIPA, LIPN, LIPT1, LMBRD1, LMNA, LPIN1, LRBA, LRPPRC, LRRC8A, MAGEL2, MAGT1, MALT1, MAN2B1, MANBA, MAP2K1, MAP2K2, MAT1A, MCCC1, MCCC2, MCEE, MCM4, MCPH1, MECP2, MED12, MEF2C, MEGF10, MFN2, MFSD8, MITF, MKKS, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MPC1, MPI, MPL, MPV17, MPZ, MRPL3, MRPL44, MSMO1, MTHFR, MTM1, MTMR14, MTO1, MTR, MTRR, MUSK, MVK, MYCN, MYH9, NAA10, NAGA, NAGS, NALCN, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NDUFV2, NEB, NEU1, NEUROG3, NEXN, NFKB2, NFU1, NGF, NGLY1, NHEJ1, NIPAL4, NIPBL, NKX2-1, NKX2-5, NLRC4, NLRP3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NR0B1, NR3C2, NRAS, NSD1, NSDHL, NUBPL, OAT, OCLN, OCRL, OPA3, OPHN1, ORC1, ORC4, OTC, OXCT1, PAFAH1B1, PAH, PAX2, PAX3, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH19, PCNT, PDCD10, PDE10A, PDHA1, PDHB, PDHX, PDP1, PDSS2, PDX1, PEPD, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGM1, PHGDH, PHKG2, PHOX2B, PIGA, PIGN, PIGT, PIGV, PIK3CD, PKD2, PKHD1, PKLR, PLCB4, PLEC, PLOD1, PLP1, PMM2, PMP22, PNKP, PNP, PNPLA1, PNPO, PNPT1, POGZ, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU1F1, PPT1, PRDM16, PRKAG2, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PSAP, PSAT1, PSPH, PTPN11, PTPRC, PTRH2, PTS, PURA, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RAC2, RAF1, RAG1, RAG2, RANBP2, RAPSN, RARS2, RB1, RBBP8, RBM8A, RET, RFT1, RFX5, RFX6, RIT1, RMND1, RNASEH2C, RNASET2, RORC, RPS19, RRM2B, RXYLT1, RYR1, SALL1, SATB2, SBDS, SCN1A, SCN2A, SCN4A, SCN5A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SELENON, SERAC1, SERPINC1, SERPING1, SFTPB, SFTPC, SHOC2, SIL1, SIX3, SIX5, SKI, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC26A2, SLC26A3, SLC2A1, SLC30A2, SLC33A1, SLC3A1, SLC4A1, SLC52A1, SLC52A3, SLC5A1, SLC5A5, SLC6A1, SLC6A3, SLC6A5, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNAI2, SNX10, SOS1, SOX10, SOX2, SOX9, SPAST, SPEG, SPINK5, SPINT2, SPR, SPRED1, SPTA1, SPTAN1, SPTB, SRD5A3, ST3GAL3, ST3GAL5, STAR, STAT1, STAT3, STIL, STIM1, STS, STT3B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SYNE1, TACO1, TAT, TAZ, TBC1D24, TBCE, TBX19, TBX5, TCAP, TCN2, TFR2, TG, TGM1, TH, THRA, TJP2, TMCO1, TMEM165, STING1, TMEM70, TNFRSF13B, TNFRSF13C, TNFSF4, TNNT1, TP63, TPM2, TPM3, TPO, TPP1, TRIP11, TRMU, TRPV4, TSC1, TSC2, TSFM, TSHB, TSHR, TSPYL1, TTC7A, TTN, TUBA8, TUBB2A, TWNK, UBA1, UGT1A1, UMPS, UNG, UPB1, UQCRC2, UROD, UROS, WAS, WDPCP, WDR62, WDR73, WFS1, WNK1, WT1, ZAP70, ZEB2, ZFP57, ZNF423	5445	5

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Disease	Gene	Test code	OMIM Gene	Available test methods
Accelerated tumor formation, susceptibility to	MDM2	2144	164785	2, 4, 8, 9, 17
Adenosine triphosphate, elevated, of erythrocytes	PKLR	481	609712	2, 4, 8, 9
Afibrinogenemia, congenital	FGA	2091	134820	2, 4
Afibrinogenemia, congenital	FGB	2092	134830	2, 3, 4, 8, 9, 17
Afibrinogenemia, congenital	FGG	2093	134850	2, 4, 17
Agammaglobulinemia and isolated hormone deficiency	BTK	2024	300300	2, 4, 8, 9, 17
Agammaglobulinemia type 1, X-linked	BTK	2024	300300	2, 4, 8, 9, 17
Agammaglobulinemia type 2, autosomal recessive	IGLL1	2553	146770	2, 4
Agammaglobulinemia type 3, autosomal recessive	CD79A	2550	112205	2, 4
Agammaglobulinemia type 4, autosomal recessive	BLNK	2606	604515	2, 4, 17
Agammaglobulinemia type 5, autosomal recessive	LRRC8A	2525	608360	2, 4, 17
Agammaglobulinemia type 6, autosomal recessive	CD79B	2551	147245	2, 4
Agammaglobulinemia type 7, autosomal recessive	PIK3R1	2523	171833	2, 4, 17
Alpha-thalassemia/mental retardation syndrome	ATRX	27	300032	2, 4, 8, 9, 17
Anemia, neonatal hemolytic, fatal and near-fatal	SPTB	1855	182870	2, 4, 17
Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive	GLRX5	2315	609588	2, 4
Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive	SLC25A38	2904	610819	2, 4
Anemia, sideroblastic, type 4	HSPA9	3784	600548	17
Anemia, sideroblastic, with ataxia	ABCB7	2729	300135	2, 4, 17
Anemia, sideroblastic, X-linked	ALAS2	2783	301300	2, 4, 17
Anemia, X-linked	GATA1	66	305371	2, 3, 4, 8, 9
Anhaptoglobinemia	HP	2789	140100	2, 17
Bare lymphocyte syndrome, type 2, complementation group C	RFX5	3263	601863	17
Bleeding disorder, platelet-type 8	P2RY12	2860	600515	2, 4
Bleeding disorder, platelet-type 15	ACTN1	2603	102575	2, 4, 17
Bleeding disorder, platelet-type 17	GFI1B	2837	604383	2, 4, 17
Bone marrow failure syndrome type 1	SRP72	756	602122	2, 3, 4, 8, 9, 17
Bone marrow failure syndrome type 2	ERCC6L2	2858	615667	2, 3, 4, 8, 9, 17
Cyanosis, transient neonatal	HBG2	2757	142250	2, 4, 8, 9
Dehydrated hereditary stomatocytosis	PIEZO1	635	611184	2, 3, 4, 8, 9
Delta-beta thalassemia	HBB	112	141900	2, 4, 8, 9
Diamond Blackfan anemia type 15 with mandibulofacial dysostosis	RPS28	2699	603685	2, 3, 4, 8, 9
Diamond-Blackfan anemia type 1	RPS19	1498	603474	2, 4, 8, 9, 17
Diamond-blackfan anemia type 3	RPS24	1565	602412	2, 4, 17
Diamond-Blackfan anemia type 4	RPS17	1564	180472	2, 3, 4, 8, 9
Diamond-Blackfan anemia type 5	RPL35A	1561	180468	2, 4, 8, 9
Diamond-Blackfan anemia type 6	RPL5	1243	603634	2, 4, 8, 9, 17
Diamond-Blackfan anemia type 7	RPL11	1560	604175	2, 4, 8, 9
Diamond-Blackfan anemia type 8	RPS7	1563	603658	2, 4
Diamond-Blackfan anemia type 9	RPS10	1562	603632	2, 4
Diamond-Blackfan anemia type 10	RPS26	1566	603701	2, 4, 8, 9
Diamond-Blackfan anemia type 11	RPL26	2696	603704	2, 3, 4, 8, 9
Diamond-Blackfan anemia type 12	RPL15	2793	604174	2, 3, 4, 8, 9
Diamond-Blackfan anemia type 13	RPS29	2697	603633	2, 3, 4, 8, 9
Diamond-Blackfan anemia type 14 with mandibulofacial dysostosis	TSR2	2698	300945	2, 3, 4, 8, 9
Dyserythropoietic anemia	COX4I2	1030	607976	2, 4
Dyserythropoietic anemia, congenital, type 1B	C15orf41	2036	615626	2, 3, 4, 8, 9, 17
Dyserythropoietic anemia, congenital, type 3	KIF23	2182	605064	2, 3, 4, 8, 9, 17
Dyserythropoietic anemia, congenital, type 4	KLF1	2177	600599	2, 3, 4, 8, 9
Dysprothrombinemia	F2	374	176930	2, 4, 17
Erythrocytosis, familial type 1	EPOR	2800	133171	2, 3, 4, 8, 9
Erythrocytosis, familial type 3	EGLN1	1798	606425	2, 3, 4, 8, 9
Erythrocytosis, familial type 4	EPAS1	1706	603349	2, 3, 4, 8, 9, 17
Factor X deficiency	F10	2170	613872	2, 4, 8, 9, 17
Favism, susceptibility to	G6PD	400	305900	2, 3, 4, 8, 9, 17
Granulomatous disease, chronic, autosomal recessive, cytochrome b-negative	CYBA	2172	608508	2, 4
Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 2	NCF2	2173	608515	2, 4, 17
Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 3	NCF4	2174	601488	2, 4, 17
Granulomatous disease, chronic, X-linked	CYBB	998	300481	2, 3, 4, 8, 9, 17
Hemolytic anemia due to G6PD deficiency	G6PD	400	305900	2, 3, 4, 8, 9, 17
Hemolytic anemia due to triosephosphate isomerase deficiency	TPI1	2902	190450	2, 4, 17
Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	CD59	2663	107271	2, 4, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Hemolytic anemia, Kell-system related	KEL	2812	613883	2, 4, 17
Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency	GPI	3628	172400	17
Hemophilia B	F9	2309	300746	2, 4, 8, 9, 17
Hereditary persistence of fetal hemoglobin	HBG2	2757	142250	2, 4, 8, 9
Hydrops, lactic acidosis, and sideroblastic anemia	LARS2	3633	604544	17
Intrinsic factor deficiency	CBLIF	105	609342	2, 4, 17
Iron-refractory iron deficiency anemia	TMPRSS6	2638	609862	2, 3, 4, 8, 9, 17
Leukemia, acute myeloid form, susceptible due to TERT germline mutation	TERT	292	187270	2, 4, 8, 9, 17
Leukocyte adhesion deficiency type 3	FERMT3	2917	607901	2, 4
Lutheran inhibitor blood group	KLF1	2177	600599	2, 3, 4, 8, 9
Lymphoproliferative syndrome type 1	ITK	1708	186973	2, 4, 8, 9, 17
Lymphoproliferative syndrome, autoimmune, type 5	CTLA4	2704	123890	2, 3, 4, 8, 9
Lymphoproliferative syndrome, X-linked type 1	SH2D1A	1184	300490	2, 4, 8, 9
Lymphoproliferative syndrome, X-linked type 2	XIAP	1185	300079	2, 4, 8, 9, 17
Megaloblastic anemia type 1, Finnish type	CUBN	103	602997	2, 17
Megaloblastic anemia type 1	AMN	104	605799	2, 3, 4, 8, 9, 17
Methemoglobinemia type 1	CYB5R3	2450	613213	2, 3, 4, 8, 9, 17
Myeloproliferative disorder, chronic, with eosinophilia	PDGFRB	1939	173410	2, 3, 4, 8, 9
Neutropenia, severe congenital type 1	ELANE	2066	130130	2, 3, 4, 8, 9
Neutropenia, severe congenital type 3	HAX1	2100	605998	2, 3, 4, 8, 9
Neutropenia, severe congenital type 5, autosomal recessive	VPS45	2810	610035	2, 4, 17
Neutrophilia, hereditary	CSF3R	2017	138971	2, 4, 17
Platelet aggregation disorder	PEAR1	1785	610278	2, 4, 17
Platelet dense granule secretion defect, excessive bleeding	FLI1	2262	193067	2, 4
Platelet disorder with associated myeloid malignancy	RUNX1	71	151385	2, 4, 8, 9, 17
Platelet glycoprotein IV deficiency	CD36	2897	173510	2, 3, 4, 8, 9, 17
Protoporphyria, erythropoietic, X-linked	ALAS2	2783	301300	2, 4, 17
Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency	GGCX	3177	137167	2, 4, 17
Schimke immunoosseous dysplasia	SMARCAL1	1709	606622	2, 4, 17
SCID autosomal recessive T negative B positive type	JAK3	1203	600173	2, 4, 17
Sea-blue histiocyte disease	APOE	18	107741	2, 3, 4, 8, 9
Shwachman-Diamond syndrome	SBDS	252	607444	2, 3, 4, 8, 9
Sickle cell anemia	HBB	112	141900	2, 4, 8, 9
Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay	TRNT1	2905	612907	2, 4, 17
Spherocytosis type 1	ANK1	2009	612641	2, 4, 17
Spherocytosis type 2	SPTB	1855	182870	2, 4, 17
Spherocytosis type 3	SPTA1	1854	182860	2, 17
Spherocytosis type 5	EPB42	1853	177070	2, 4, 17
Stormorken syndrome	STIM1	2391	605921	2, 3, 4, 8, 9, 17
Thalassemia, alpha	HBA1	1519	141800	2, 4, 8, 9
Thalassemia, alpha	HBA2	1520	141850	2, 4, 8, 9
Thalassemia, delta	HBD	2128	142000	2, 4, 8, 9
Thiamine-responsive megaloblastic anemia syndrome	SLC19A2	2171	603941	2, 4
Thrombocytopenia congenital amegakaryocytic	MPL	1204	159530	2, 4, 17
Thrombocytopenia type 2	ANKRD26	2333	610855	2, 4, 17
Thrombocytopenia type 2	MASTL	70	608221	2, 4, 17
Thrombocytopenia type 4	CYCS	2767	123970	2, 4
Thrombocytopenia type 5	ETV6	2929	600618	2, 4, 8, 9
Thrombocytopenia with beta thalassemia X-linked	GATA1	66	305371	2, 3, 4, 8, 9
Thrombocytopenia, neonatal alloimmune	ITGA2B	2077	607759	2, 3, 4, 8, 9, 17
Thrombocytopenia, neonatal alloimmune	ITGB3	2078	173470	2, 3, 4, 8, 9, 17
Thrombocytopenia, X-linked	GATA1	66	305371	2, 3, 4, 8, 9
Thrombocytopenia, X-linked, intermittent	WAS	72	300392	2, 3, 4, 8, 9, 17
Thrombocytosis, familial, JAK2 related	JAK2	625	147796	2, 4, 8, 9, 17
Thrombophilia due to thrombin defect	F2	374	176930	2, 4, 17
Thrombophilia, X-linked, due to factor IX defect	F9	2309	300746	2, 4, 8, 9, 17
Thrombotic thrombocytopenic purpura	ADAMTS13	65	604134	2, 3, 4, 8, 9, 17
Thromboxane synthase deficiency	TBXAS1	2428	274180	2, 4, 17
Vitamin K-dependent clotting factors combined deficiency type 1	GGCX	3177	137167	2, 4, 17
von Willebrand disease	VWF	1328	613160	2, 4, 8, 9, 17
von Willebrand disease platelet type	GP1BA	67	606672	2, 4

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Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoArrayCyto® 750K	Genome-wide CMA with 750,000 markers		50068	5
CentoArrayCyto® HD	Genome-wide CMA with 2.6 million markers		50001	5
CentoDx®	Targets exonic regions of >6500 genes associated with known clinical phenotypes		50158	5
CentoDysmorph	ABCB6, ABCC6, ACTA1, ACTA2, ACTB, ACTG1, ACVR2B, ADAMTS2, ADAMTSL2, ADGRG1, AFF4, AGPS, AGRN, AHI1, AKT3, ALDH18A1, ALDH1A3, ALG2, ALPL, AMPD2, ANKH, ANKRD11, ANO5, AP4M1, ARFGEF2, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, ARMC4, ARSL, ARX, ASPM, ASXL1, ASXL3, ATP6V0A2, ATP7A, ATR, ATRX, B3GALNT2, B3GALT6, B3GAT3, B4GALT7, B4GAT1, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BGN, BIN1, BMP1, BMP4, BMPR1B, BRAF, C8orf37, CANT1, CASK, CASR, CBL, CBS, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC28B, CCDC39, CCDC40, CCDC65, CCM2, CCNO, CDK5RAP2, CDKN1C, CDON, CENPF, CENPJ, CEP135, CEP152, CEP164, CEP290, CEP41, CEP63, CFAP298, CFAP53, CFC1, CFL2, CHAT, CHD7, CHMP1A, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHSY1, CILK1, CLCN5, COASY, COL10A1, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, COLQ, COMP, COX7B, CPLANE1, CREB3L1, CREBBP, CRELD1, CRIPT, CRPPA, CRTAP, CSPP1, CUL7, CYP1B1, DAG1, DCX, DDR2, DHCR24, DHCR7, DMP1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DNM2, DOK7, DPAGT1, DRC1, DSE, DYM, DYNC1H1, DYNC2H1, DYRK1A, EBP, ECE1, ECEL1, EDN3, EDNRB, EFEMP2, EFNB1, EFTUD2, EGR2, EIF2AK3, ELN, ENPP1, ERCC1, ERCC2, ERCC5, ERCC6, ERF, ESCO2, EVC, EVC2, EXOSC3, FAS, FBLN5, FBN1, FBN2, FGF23, FGF8, FGFR1, FGFR2, FGFR3, FHL1, FKBP10, FKBP14, FKRP, FKTN, FLCN, FLNA, FLNB, FOXC1, FOXE3, FOXH1, FOXL2, FRAS1, FREM1, FREM2, GBA, GBE1, GDF1, GDF3, GDF5, GDF6, GDNF, GFPT1, GJA1, GLE1, GLI2, GLI3, GMPPB, GNAS, GNPAT, GORAB, GRIP1, HCCS, HDAC8, HESX1, HEXA, HMGB3, HMX1, HRAS, HSPG2, HUWE1, HYDIN, HYLS1, IER3IP1, IFITM5, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IL11RA, INPP5E, INPPL1, IRF6, ITGB4, KAT6B, KATNB1, KBTBD13, KDM5C, KDM6A, KIF11, KIF2A, KIF5C, KIF7, KIT, KLHL40, KLHL41, KMT2A, KMT2D, KNL1, KRAS, KRIT1, L1CAM, LAMA2, LAMA3, LAMB1, LAMB3, LAMC2, LARGE1, LBR, LEFTY2, LEP, LEPR, LIFR, LMNA, LMOD3, LMX1B, LOX, LRP2, LRP5, LRRC6, LZTFL1, LZTR1, MAB21L2, MAGEL2, MAP2K1, MAP2K2, MASP1, MAT2A, MATN3, MBTPS2, MCPH1, MED12, MED17, MEGF8, MEIS2, MEOX1, MFAP5, MFRP, MFSD2A, MITF, MKKS, MKS1, MMP13, MMP21, MMP9, MSMO1, MSX1, MSX2, MTM1, MUSK, MYBPC1, MYCN, MYH11, MYH2, MYH3, MYH8, MYLK, MYO18B, MYO7A, NAA10, NALCN, NDE1, NDP, NEB, NECTIN1, NEK1, NEK8, NF1, NF2, NHEJ1, NIPBL, NKX2-5, NKX3-2, NME8, NODAL, NOG, NOTCH1, NPHP1, NPHP3, NR0B2, NRAS, NRG1, NSDHL, OCRL, OFD1, ORC1, OTX2, P3H1, PAFAH1B1, PAX2, PAX3, PAX6, PCNT, PDCD10, PDE6D, PEX7, PHC1, PHEX, PHF6, PIEZO2, PIGV, PITX2, PKD1L1, PKD2, PKHD1, PLK4, PLOD1, PLOD2, PMM2, PNKP, PNPLA6, POMC, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PPARG, PPIB, PQBP1, PRDM5, PRKAR1A, PRKG1, PROM1, PRPH2, PRSS56, PTCH1, PTH1R, PTPN11, PYCR1, PYCR2, QARS1, RAB18, RAB23, RAB3GAP1, RAB3GAP2, RAD21, RAF1, RAPSN, RARB, RARS2, RASA1, RASA2, RAX, RBBP8, RBP4, RDH5, RELN, RET, RHO, RIN2, RIT1, RLBP1, ROR2, RPGRIP1L, RPL10, RPS6KA3, RSPH1, RSPH4A, RSPH9, RUNX2, RXYLT1, RYR1, SALL1, SALL2, SALL4, SASS6, SATB2, SBDS, SCN4A, SDCCAG8, SELENON, SEPSECS, SERPINF1, SERPINH1, SHH, SHOC2, SHROOM4, SIX3, SIX6, SKI, SLC25A19, SLC26A2, SLC2A10, SLC34A3, SLC35D1, SLC39A13, SLC5A7, SLC9A6, SMAD2, SMAD3, SMAD4, SMAD6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMCHD1, SMOC1, SNAI2, SOS1, SOS2, SOX10, SOX2, SOX9, SP7, SPAG1, SPRED1, STAMBP, STIL, STRA6, SUMO1, TAF6, TBC1D20, TBC1D24, TBX5, TCF12, TCF4, TCTN1, TCTN2, TCTN3, TENM3, TFAP2A, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TK2, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNFRSF11B, TNNI2, TNNT1, TNNT3, TNXB, TOR1A, TP63, TPM2, TPM3, TRIM32, TRIP11, TRMT10A, TRPS1, TRPV4, TSC1, TSC2, TSEN2, TSEN54, TTC21B, TTC8, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP4, TUBGCP6, TWIST1, TYR, UBA1, VAMP1, VIPAS39, VLDLR, VPS13B, VPS53, VRK1, VSX2, WDPCP, WDR19, WDR34, WDR35, WDR60, WDR62, WNT1, WNT5A, WNT7A, YWHAE, ZEB2, ZIC2, ZIC3, ZMYND10, ZNF335, ZNF423, ZNF469		5429	6

1: Hotspot Testing
2: Carrier Testing (point mutation)
3: Carrier Testing (del/dup)
4: Single Gene Sequencing (Sanger)
5: NGS Panel
6: NGS Panel + CNV
7: NGS Panel Genomic
8: Deletion/Duplication with qPCR/MLPA
9: Sequencing + Deletion/Duplication
10: Repeat Expansions
11: Somatic Mutation Analysis
12: Biochemical Enzyme Analysis
13: Biomarker Analysis
14: Biochemical Enzyme Panel
15: Biochemical Genetics Panel Plus
16: Single Gene Sequencing (NGS)
17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoLCV	Genome wide analysis of structural variants and large copy number changes through sWGS		50118	5
Ciliopathies panel	ACVR2B, AGPS, AHI1, ALPL, ANKH, ANO5, ARHGAP31, ARL13B, ARL6, ARMC4, ARSL, ATR, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BMP1, BMPR1B, C8ORF37, CANT1, CASR, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC28B, CCDC39, CCDC40, CCDC65, CCNO, CDKN1C, CENPF, CENPJ, CEP152, CEP164, CEP290, CEP41, CEP63, CFAP298, CFAP53, CFC1, CHSY1, CILK1, CLCN5, COL10A1, COL9A3, COMP, CPLANE1, CRELD1, CRTAP, CSPP1, DDR2, DHCR7, DMP1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DYM, DYNC2H1, EBP, EIF2AK3, ENPP1, ESCO2, EVC, EVC2, FGF23, FKBP10, FLNB, GDF1, GDF5, GLI2, GLI3, GNAS, GNPAT, HEXA, HSPG2, HYDIN, HYLS1, IFITM5, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, INPP5E, INPPL1, KIF7, LBR, LEFTY2, LEP, LEPR, LIFR, LRP5, LRRC6, LZTFL1, MATN3, MKKS, MKS1, MMP13, MMP21, MMP9, MYO7A, NEK1, NEK8, NKX2-5, NKX3-2, NME8, NODAL, NOG, NPHP1, NPHP3, NR0B2, NSDHL, OFD1, P3H1, PDE6D, PEX7, PHEX, PHF6, PKD1L1, PKD2, PKHD1, PLOD2, PMM2, PNPLA6, POMC, PPARG, PPIB, PRKAR1A, PROM1, PRPH2, PTH1R, RBBP8, RDH5, RHO, RLBP1, ROR2, RPGRIP1L, RSPH1, RSPH4A, RSPH9, RUNX2, SALL1, SALL4, SBDS, SDCCAG8, SERPINF1, SERPINH1, SLC26A2, SLC34A3, SLC35D1, SOX9, SPAG1, TBX5, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNFRSF11B, TRIM32, TRIP11, TRPS1, TRPV4, TTC21B, TTC8, WDPCP, WDR19, WDR34, WDR35, WDR60, WNT5A, WNT7A, ZIC3, ZMYND10, ZNF423		5431	6
Noonan - RASophathies panel	BRAF, CBL, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NF2, NRAS, PTPN11, RAF1, RASA2, RIT1, SHOC2, SMARCB1, SOS1, SOS2, SPRED1, TSC1, TSC2		5461	6

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6: NGS Panel + CNV
7: NGS Panel Genomic
8: Deletion/Duplication with qPCR/MLPA
9: Sequencing + Deletion/Duplication
10: Repeat Expansions
11: Somatic Mutation Analysis
12: Biochemical Enzyme Analysis
13: Biomarker Analysis
14: Biochemical Enzyme Panel
15: Biochemical Genetics Panel Plus
16: Single Gene Sequencing (NGS)
17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Test code	Available test methods
CentoICU®	AARS1, AARS2, AASS, ABAT, ABCA12, ABCA3, ABCB11, ABCC8, ABCD1, ABCD3, ABCD4, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACTA1, ACY1, ADA, ADAMTS13, ADAMTSL2, ADAR, ADK, ADNP, ADSL, AGA, AGK, AGL, AGPAT2, AGPS, AGRN, AGXT, AHCY, AICDA, AIFM1, AIMP1, AKAP9, AKR1D1, AKT2, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX12B, ALOXE3, ALPL, ALS2, AMACR, AMN, AMPD1, AMT, ANK1, ANKRD26, ANKS6, ANTXR1, ANTXR2, AP2S1, AP4B1, AP4E1, AP4M1, AP4S1, APOB, APTX, ARG1, ARL6, ARSA, ARSB, ARX, ASAH1, ASL, ASNS, ASPA, ASPM, ASS1, ATIC, ATP1A3, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATPAF2, ATR, ATRX, AUH, B3GLCT, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCKDK, BCS1L, BDNF, BICD2, BIN1, BLNK, BOLA3, BRAF, BRAT1, BRCA2, BSCL2, BSND, BTD, BTK, C12orf65, CA12, CACNA1C, CACNB2, CALM1, CAMTA1, CASK, CASR, CAST, CAV1, CAV3, CAVIN1, CBS, CCDC103, CCDC114, CCDC78, CD19, CD247, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD59, CD79A, CD79B, CD81, CD96, CDAN1, CDK5RAP2, CDKL5, CDKN1C, CENPJ, CEP152, CEP290, CERS3, CFAP298, CFH, CFHR3, CFL2, CFTR, CHAT, CHD7, CHKB, CHM, CHRNA1, CHRNB1, CHRND, CHRNE, CIDEC, CLCN1, CLCNKA, CLCNKB, CLDN16, CLN3, CLN5, CLN6, CLN8, CLPB, CNTN1, COA5, COG1, COG6, COG7, COL11A1, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A2, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COMP, COQ2, COQ8A, COQ9, CORO1A, COX10, COX15, COX20, COX6B1, CPS1, CPT1A, CPT2, CR2, CRPPA, CRTAP, CTNS, CTPS1, CTSA, CTSD, CUL4B, CXCR4, CYP11B1, CYP11B2, CYP17A1, CYP4F22, CYP7B1, D2HGDH, DBT, DCLRE1C, DDC, DDOST, DDR2, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DIAPH1, DLAT, DLD, DMD, DNA2, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC19, DNM2, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPM2, DPYD, DRC1, DSP, DST, DUOX2, DUOXA2, DYSF, EDN3, EEF1A2, EGR2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELANE, ENPP1, EPB42, EPCAM, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYA1, EYA4, F10, F11, F13A1, F2, F5, F7, F8, F9, FADD, FAH, FANCA, FANCB, FANCC, FANCD2, FANCL, FARS2, FASTKD2, FBN1, FBP1, FBXL4, FGA, FGB, FGFR2, FGFR3, FGG, FH, FIG4, FKBP14, FKRP, FKTN, FOXC1, FOXG1, FOXP3, FOXRED1, FRAS1, FUCA1, G6PC, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GARS1, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GFAP, GFM1, GFPT1, GJA1, GJB2, GJB4, GK, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLUD1, GLYCTK, GMPPB, GNAS, GNE, GNMT, GNPAT, GNPTAB, GP1BA, GP1BB, GP9, GPC3, GPHN, GPSM2, GSS, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBA1, HBA2, HBB, HCFC1, HESX1, HEXA, HEXB, HGD, HGF, HIBCH, HLCS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPGD, HRAS, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, ICOS, IDUA, IER3IP1, IFIH1, IFT172, IGF1, IGF1R, IGHMBP2, IGLL1, IKBKB, IL12RB1, IL2RA, IL2RG, IL7R, INS, INSR, INVS, IRF8, ITGA2B, ITGA6, ITGA7, ITGB3, ITGB4, IVD, JAG1, JAGN1, JAK3, JAM3, KAT6A, KAT6B, KBTBD13, KCNE1, KCNH1, KCNH2, KCNJ10, KCNJ11, KCNQ1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIF1B, KLF1, KLHL40, KLHL41, KLHL7, KRAS, KRT5, LAMA2, LAMA3, LAMB3, LAMC2, LAMP2, LAMTOR2, LARS2, LAS1L, LCT, LHX3, LHX4, LIAS, LIG4, LIPA, LIPN, LIPT1, LMBRD1, LMNA, LPIN1, LRBA, LRPPRC, LRRC8A, MAGEL2, MAGT1, MALT1, MAN2B1, MANBA, MAP2K1, MAP2K2, MAT1A, MCCC1, MCCC2, MCEE, MCM4, MCPH1, MECP2, MED12, MEF2C, MEGF10, MFN2, MFSD8, MITF, MKKS, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MPC1, MPI, MPL, MPV17, MPZ, MRPL3, MRPL44, MSMO1, MTHFR, MTM1, MTMR14, MTO1, MTR, MTRR, MUSK, MVK, MYCN, MYH9, NAA10, NAGA, NAGS, NALCN, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NDUFV2, NEB, NEU1, NEUROG3, NEXN, NFKB2, NFU1, NGF, NGLY1, NHEJ1, NIPAL4, NIPBL, NKX2-1, NKX2-5, NLRC4, NLRP3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NR0B1, NR3C2, NRAS, NSD1, NSDHL, NUBPL, OAT, OCLN, OCRL, OPA3, OPHN1, ORC1, ORC4, OTC, OXCT1, PAFAH1B1, PAH, PAX2, PAX3, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH19, PCNT, PDCD10, PDE10A, PDHA1, PDHB, PDHX, PDP1, PDSS2, PDX1, PEPD, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGM1, PHGDH, PHKG2, PHOX2B, PIGA, PIGN, PIGT, PIGV, PIK3CD, PKD2, PKHD1, PKLR, PLCB4, PLEC, PLOD1, PLP1, PMM2, PMP22, PNKP, PNP, PNPLA1, PNPO, PNPT1, POGZ, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU1F1, PPT1, PRDM16, PRKAG2, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PSAP, PSAT1, PSPH, PTPN11, PTPRC, PTRH2, PTS, PURA, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RAC2, RAF1, RAG1, RAG2, RANBP2, RAPSN, RARS2, RB1, RBBP8, RBM8A, RET, RFT1, RFX5, RFX6, RIT1, RMND1, RNASEH2C, RNASET2, RORC, RPS19, RRM2B, RXYLT1, RYR1, SALL1, SATB2, SBDS, SCN1A, SCN2A, SCN4A, SCN5A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SELENON, SERAC1, SERPINC1, SERPING1, SFTPB, SFTPC, SHOC2, SIL1, SIX3, SIX5, SKI, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC26A2, SLC26A3, SLC2A1, SLC30A2, SLC33A1, SLC3A1, SLC4A1, SLC52A1, SLC52A3, SLC5A1, SLC5A5, SLC6A1, SLC6A3, SLC6A5, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNAI2, SNX10, SOS1, SOX10, SOX2, SOX9, SPAST, SPEG, SPINK5, SPINT2, SPR, SPRED1, SPTA1, SPTAN1, SPTB, SRD5A3, ST3GAL3, ST3GAL5, STAR, STAT1, STAT3, STIL, STIM1, STS, STT3B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SYNE1, TACO1, TAT, TAZ, TBC1D24, TBCE, TBX19, TBX5, TCAP, TCN2, TFR2, TG, TGM1, TH, THRA, TJP2, TMCO1, TMEM165, STING1, TMEM70, TNFRSF13B, TNFRSF13C, TNFSF4, TNNT1, TP63, TPM2, TPM3, TPO, TPP1, TRIP11, TRMU, TRPV4, TSC1, TSC2, TSFM, TSHB, TSHR, TSPYL1, TTC7A, TTN, TUBA8, TUBB2A, TWNK, UBA1, UGT1A1, UMPS, UNG, UPB1, UQCRC2, UROD, UROS, WAS, WDPCP, WDR62, WDR73, WFS1, WNK1, WT1, ZAP70, ZEB2, ZFP57, ZNF423	5445	5

1: Hotspot Testing
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4: Single Gene Sequencing (Sanger)
5: NGS Panel
6: NGS Panel + CNV
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8: Deletion/Duplication with qPCR/MLPA
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10: Repeat Expansions
11: Somatic Mutation Analysis
12: Biochemical Enzyme Analysis
13: Biomarker Analysis
14: Biochemical Enzyme Panel
15: Biochemical Genetics Panel Plus
16: Single Gene Sequencing (NGS)
17: Single Gene Sequencing + CNV (NGS)

Disease	Gene	Test code	OMIM Gene	Available test methods
Achondrogenesis type 2	COL2A1	943	120140	2, 4, 8, 9, 17
Acrodysostosis type 1, with or without hormone resistance	PRKAR1A	1192	188830	2, 3, 4, 8, 9, 17
Acromelic frontonasal dysostosis	ZSWIM6	2454	615951	2, 3, 4, 8, 9
Acromesomelic dysplasia, Maroteaux type	NPR2	1969	108961	2, 3, 4, 8, 9, 17
Acromicric dysplasia	FBN1	950	134797	2, 8, 17
Adams-Oliver syndrome type 6	DLL4	3049	605185	2, 4
ADULT syndrome, split hand-foot malformation	TP63	304	603273	2, 3, 4, 8, 9, 17
Alacrima, achalasia and mental retardation syndrome	GMPPA	2162	615495	2, 4, 17
Alagille syndrome type 1	JAG1	1206	601920	2, 4, 8, 9, 17
Alagille syndrome type 2	NOTCH2	1207	600275	2, 3, 4, 8, 9, 17
Alazami syndrome	LARP7	2187	612026	2, 3, 4, 8, 9, 17
Alpha-thalassemia/mental retardation syndrome	ATRX	27	300032	2, 4, 8, 9, 17
Ankyloblepharon-ectodermal defects-cleft lip/palate	TP63	304	603273	2, 3, 4, 8, 9, 17
Anterior segment mesenchymal dysgenesis	PITX3	2571	602669	2, 4
Antley-Bixler syndrome	FGFR2	1730	176943	2, 4, 8, 9, 17
Apert syndrome	FGFR2	1730	176943	2, 4, 8, 9, 17
Athabaskan brainstem dysgenesis syndrome	HOXA1	1208	142955	2, 4, 8, 9
Atrial septal defect type 2	GATA4	1209	600576	2, 4, 8, 9, 17
Atrial septal defect with atrioventricular conduction defects	NKX2-5	1210	600584	2, 4, 8, 9
Atrioventricular septal defect, partial with heterotaxy syndrome	CRELD1	2039	607170	2, 4
Auriculocondylar syndrome type 1	GNAI3	2916	139370	2, 3, 4, 8, 9
Auriculocondylar syndrome type 2	PLCB4	1015	600810	2, 3, 4, 8, 9, 17
Axenfeld-Rieger syndrome type 1	PITX2	216	601542	2, 4, 8, 9
Axenfeld-Rieger syndrome type 3	FOXC1	888	601090	2, 4, 8, 9
Bainbridge-Ropers syndrome	ASXL3	2111	615115	2, 4, 17
Band-like calcification with simplified gyration and polymicrogyria	OCLN	1710	602876	2, 3, 4, 8, 9, 17
Baraitser-Winter syndrome type 1	ACTB	779	102630	2, 3, 4, 8, 9
Baraitser-Winter syndrome type 2	ACTG1	989	102560	2, 3, 4, 8, 9
Basal cell nevus syndrome	PTCH1	1216	601309	2, 4, 8, 9, 17
Basal cell nevus syndrome	SUFU	2360	607035	2, 3, 4, 8, 9, 17
Basal ganglia calcification type 1, ideopathic	SLC20A2	1329	158378	2, 3, 4, 8, 9, 17
Basal ganglia calcification type 4	PDGFRB	1939	173410	2, 3, 4, 8, 9
Basal ganglia calcification type 5, idiopathic	PDGFB	1801	190040	2, 3, 4, 8, 9
Basal ganglia calcification type 6, idiopathic	XPR1	2881	605237	2, 3, 4, 8, 9
Beta-ureidopropionase deficiency	UPB1	2466	606673	2, 3, 4, 8, 9, 17
Bifid nose	FREM1	1534	608944	2, 3, 4, 8, 9, 17
Birt-Hogg-Dube syndrome	FLCN	55	607273	2, 4, 8, 9, 17
Bjornstad syndrome	BCS1L	1423	603647	2, 3, 4, 8, 9, 17
Blau syndrome	NOD2	2310	605956	2, 3, 4, 8, 9, 17
Bohring-Opitz syndrome	ASXL1	2280	612990	2, 3, 4, 8, 9, 17
Bone marrow failure syndrome type 2	ERCC6L2	2858	615667	2, 3, 4, 8, 9, 17
Brachydactyly type A1C	GDF5	1920	601146	2, 4, 8, 9
Brachydactyly type A2	BMP2	1491	112261	2, 4
Brachydactyly type A2	BMPR1B	1324	603248	2, 4, 17
Brachydactyly type B1	ROR2	244	602337	2, 4, 8, 9, 17
Brachydactyly type E1	HOXD13	1975	142989	2, 4, 8, 9
Brachydactyly-mental retardation syndrome	HDAC4	1363	605314	2, 4, 17
Brachydactyly-syndactyly syndrome	HOXD13	1975	142989	2, 4, 8, 9
Branchiooculofacial syndrome	TFAP2A	2509	107580	2, 4, 17
C syndrome	CD96	1839	606037	2, 3, 4, 8, 9, 17
Campomelic dysplasia	SOX9	1218	608160	2, 3, 4, 8, 9
Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	PRG4	2524	604283	2, 3, 4, 8, 9
Camurati-Engelmann disease	TGFB1	293	190180	2, 4, 17
Cantu syndrome	ABCC9	1082	601439	2, 17
Carpenter syndrome	RAB23	2423	606144	2, 3, 4, 8, 9
Carpenter syndrome type 2	MEGF8	2422	604267	2, 3, 4, 8, 9, 17
Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia	IARS2	3785	612801	17
Caudal regression syndrome	VANGL1	3816	610132	17
Central hypoventilation syndrome with or without Hirschsprung disease	PHOX2B	740	603851	2, 3, 4, 8, 9, 10
Central hypoventilation syndrome, congenital	ASCL1	712	100790	2, 4, 8, 9
Central hypoventilation syndrome, congenital	BDNF	1494	113505	2, 4, 8, 9

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Central hypoventilation syndrome, congenital	ECE1	1497	600423	2, 4, 17
Central hypoventilation syndrome, congenital	EDN3	1493	131242	2, 4, 8, 9
Central hypoventilation syndrome, congenital	GDNF	1492	600837	2, 4, 8, 9
Central hypoventilation syndrome, congenital	GFRA1	1496	601496	2, 4, 8, 9
Central hypoventilation syndrome, congenital	MECP2	163	300005	2, 4, 8, 9
Central hypoventilation syndrome, congenital	PHOX2A	1033	602753	2, 3, 4, 8, 9
Central hypoventilation syndrome, congenital	RET	240	164761	2, 4, 8, 9, 17
Central hypoventilation syndrome, congenital	ZEB2	1495	605802	2, 4, 8, 9, 17
Cerebral dysgenesis, neuropathy, ichthyosis, palmoplantar keratoderma syndrome	SNAP29	1220	604202	2, 4
Cerebrooculofacioskeletal syndrome type 1	ERCC6	1822	609413	2, 3, 4, 8, 9, 17
Cerebrooculofacioskeletal syndrome type 4	ERCC1	1330	126380	2, 4, 17
Char syndrome	TFAP2B	3243	601601	17
CHARGE syndrome	CHD7	1221	608892	2, 4, 8, 9, 17
CHIME syndrome	PIGL	1331	605947	2, 4
Chondrodysplasia with joint dislocations, GPAPP type	IMPAD1	2943	614010	2, 4
Chondrodysplasia, Blomstrand type	PTH1R	2447	168468	2, 3, 4, 8, 9, 17
Choroideremia	CHM	1333	300390	2, 4, 8, 9, 17
Chrondrodysplasia, acromesomelic, with genital anomalies	BMPR1B	1324	603248	2, 4, 17
Chudley-McCullough syndrome	GPSM2	1713	609245	2, 3, 4, 8, 9, 17
Cleft palate, cardiac defects, and mental retardation	MEIS2	3667	601740	17
Club foot	PITX1	734	602149	2, 3, 4, 8, 9
Cockayne syndrome type A	ERCC8	1862		2, 3, 4, 8, 9, 17
Cockayne syndrome type B	ERCC6	1822	609413	2, 3, 4, 8, 9, 17
CODAS syndrome	LONP1	2601	605490	2, 3, 4, 8, 9, 17
Coffin-Siris syndrome, SMARCE1 related	SMARCE1	1510	603111	2, 3, 4, 8, 9, 17
Cold-induced sweating syndrome	CRLF1	2126	604237	2, 4
Cold-induced sweating syndrome type 2	CLCF1	2127	607672	2, 4
Congenital heart defects and ectodermal dysplasia	PRKD1	3795	605435	17
Congenital heart disease and transposition of the great arteries	FOXH1	2041	603621	2, 4
Congenital short-bowel syndrome	CLMP	1334	611693	2, 4
Corpus callosum, agenesis of, with abnormal genitalia	ARX	24	300382	2, 4, 8, 9, 17
Cortical malformations, occipital	LAMC3	1714	604349	2, 4, 17
Costello syndrome	HRAS	1222	190020	2, 3, 4, 8, 9
Cousin syndrome	TBX15	2484	604127	2, 4
Craniodiaphyseal dysplasia, autosomal dominant	SOST	2427	605740	2, 4
Cranioectodermal dysplasia type 1	IFT122	2421	606045	2, 4, 17
Cranioectodermal dysplasia type 2	WDR35	2425	613602	2, 4, 17
Cranioectodermal dysplasia type 3	IFT43	2420	614068	2, 4, 17
Cranioectodermal dysplasia type 4	WDR19	2424	608151	2, 4, 17
Craniofacial and neuro-developmental abnormalities	DISP1	2281	607502	2, 4, 17
Craniofacial and neuro-developmental abnormalities, JAG2 related	JAG2	2019	602570	2, 3, 4, 8, 9, 17
Craniofacial anomalies and anterior segment dysgenesis syndrome	VSX1	2900	605020	2, 4
Craniofrontonasal syndrome	EFNB1	1846	300035	2, 4, 8, 9
Craniometaphyseal dysplasia	ANKH	2084	605145	2, 4, 17
Craniosynostosis and dental anomalies	IL11RA	2185	600939	2, 4, 17
Craniosynostosis type 1	TWIST1	1541	601622	2, 4, 8, 9
Craniosynostosis type 2	MSX2	171	123101	2, 4, 8, 9
Craniosynostosis type 3	TCF12	2047	600480	2, 4, 17
Craniosynostosis type 4	ERF	2419	611888	2, 4
Craniosynostosis type 6	ZIC1	2198	600470	2, 4, 8, 9
Craniosynostosis, FGFR1 related	FGFR1	1136	136350	2, 4, 8, 9
Craniosynostosis, nonspecific	FGFR2	1730	176943	2, 4, 8, 9, 17
Crouzon syndrome	FGFR2	1730	176943	2, 4, 8, 9, 17
Currarino syndrome	MNX1	1223	142994	2, 3, 4, 8, 9
Czech dysplasia	COL2A1	943	120140	2, 4, 8, 9, 17
Dandy-Walker malformation and occipital cephaloceles, LAMC1 related	LAMC1	2629	150290	2, 4, 17
D-bifunctional protein deficiency	HSD17B4	1095	601860	2, 3, 4, 8, 9, 17
De Sanctis-Cacchione syndrome	ERCC6	1822	609413	2, 3, 4, 8, 9, 17
Desanto-Shinawi syndrome	WAC	3447	615049	17
Desbuquois dysplasia type 1	CANT1	2568	613165	2, 4
Desbuquois dysplasia type 2	XYLT1	2483	608124	2, 4, 17
Desmosterolosis	DHCR24	2621	606418	2, 4, 17
Diamond Blackfan anemia type 15 with mandibulofacial dysostosis	RPS28	2699	603685	2, 3, 4, 8, 9
Diamond-Blackfan anemia type 14 with mandibulofacial dysostosis	TSR2	2698	300945	2, 3, 4, 8, 9

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DiGeorge syndrome	TBX1	1224	602054	2, 4, 8, 9, 17
Donnai-Barrow syndrome	LRP2	1904	600073	2, 17
DOOR syndrome	TBC1D24	783	613577	2, 3, 4, 8, 9, 17
Duane Retraction syndrome	SALL4	251	607343	2, 3, 4, 8, 9, 17
Dysmorphism, HMG20B related	HMG20B	2011	605535	2, 4
Ectodactyly, ectodermal dysplasia, and cleft lip/palate syndrome type 3	TP63	304	603273	2, 3, 4, 8, 9, 17
Ellis-van Creveld syndrome	EVC2	2274	607261	2, 3, 4, 8, 9, 17
Ellis-van Creveld syndrome	EVC	2273	604831	2, 3, 4, 8, 9, 17
Epiphyseal dysplasia, multiple, with myopia and deafness	COL2A1	943	120140	2, 4, 8, 9, 17
Faciogenital dysplasia	FGD1	315	300546	2, 4, 8, 9, 17
Fanconi anemia, complementation group Q	ERCC4	2543	133520	2, 4, 17
Feingold syndrome	MYCN	1226	164840	2, 4, 8, 9
Fetal akinesia deformation sequence	DOK7	1227	610285	2, 3, 4, 8, 9, 17
Fetal akinesia deformation sequence	RAPSN	1228	601592	2, 3, 4, 8, 9
Fibrochondrogenesis type 1	COL11A1	944	120280	2, 8, 17
Fibular aplasia or hypoplasia, femoral bowing and poly-, syn-, and oligodactyly	WNT7A	327	601570	2, 3, 4, 8, 9
FILS syndrome	POLE	2876	174762	2, 17
Fraser syndrome	FRAS1	2531	607830	2, 17
Fraser syndrome	FREM2	1756	608945	2, 4, 17
Fraser syndrome	GRIP1	1212	604597	2, 3, 4, 8, 9, 17
Frontonasal dysplasia type 1	ALX3	2197	606014	2, 4, 8, 9
Frontonasal dysplasia type 2	ALX4	666	605420	2, 4, 8, 9
Fumarase deficiency	FH	1335	136850	2, 4, 8, 9, 17
Galloway-Mowat syndrome	WDR73	2405	616144	2, 4, 17
Geleophysic dysplasia type 2	FBN1	950	134797	2, 8, 17
Genitopatellar syndrome	KAT6B	1336	605880	2, 3, 4, 8, 9, 17
Glass syndrome	SATB2	2292	612313	2, 4
Goldberg-Shprintzen megacolon syndrome	KIF1BP	1229	609367	2, 4, 17
GRACILE syndrome	BCS1L	1423	603647	2, 3, 4, 8, 9, 17
Greenberg skeletal dysplasia	LBR	2446	600024	2, 4, 17
Greig cephalopolysyndactyly syndrome	GLI3	128	165240	2, 3, 4, 8, 9, 17
Growth hormone insensitivity, partial	GHR	1956	600946	2, 4, 8, 9, 17
Growth retardation with deafness and mental retardation due to IGF1 deficiency	IGF1	1677	147440	2, 4, 8, 9, 17
Growth retardation, developmental delay, facial dysmorphism	FTO	3354	610966	17
Guttmacher syndrome	HOXA13	2913	142959	2, 4
Hamamy syndrome	IRX5	1088	606195	2, 4
Hand-foot-uterus syndrome	HOXA13	2913	142959	2, 4
Hartsfield syndrome	FGFR1	1136	136350	2, 4, 8, 9
Heart-hand syndrome, Slovenian type	LMNA	158	150330	2, 4, 8, 9, 17
Helsmoortel-van der Aa syndrome	ADNP	2400	611386	2, 4
Hennekam lymphangiectasia-lymphedema syndrome type 2	FAT4	2580	612411	2, 4, 17
Heterotaxy, visceral type 1	ZIC3	2044	300265	2, 4, 8, 9
Heterotaxy, visceral type 2	CFC1	2038	605194	2, 4, 8, 9, 17
Heterotaxy, visceral type 4	ACVR2B	2037	602730	2, 4, 8, 9, 17
Heterotaxy, visceral type 5	NODAL	2043	601265	2, 4, 8, 9
Heterotaxy, visceral type 6	CFAP53	2888	614759	2, 4, 17
Heterotaxy, visceral type 7	MMP21	2889	608416	2, 4
Heterotaxy, visceral type 8, autosomal	PKD1L1	2886	609721	17
Heterotaxy, visceral, BCL9L related	BCL9L	2641	609004	2, 4
Hirschsprung disease	ECE1	1497	600423	2, 4, 17
Hirschsprung disease	EDN3	1493	131242	2, 4, 8, 9
Hirschsprung disease	EDNRB	194	131244	2, 4, 8, 9, 17
Hirschsprung disease	KIF1BP	1229	609367	2, 4, 17
Hirschsprung disease	NRG1	1500	142445	2, 4, 17
Hirschsprung disease	NRTN	1501	602018	2, 4, 8, 9
Hirschsprung disease	RET	240	164761	2, 4, 8, 9, 17
Hirschsprung disease	ZEB2	1495	605802	2, 4, 8, 9, 17
Hirschsprung disease, type 3, susceptibility to	GDNF	1492	600837	2, 4, 8, 9
Holoprosencephaly type 2	SIX3	1338	603714	2, 4, 8, 9
Holoprosencephaly type 3	SHH	1339	600725	2, 4, 8, 9, 17
Holoprosencephaly type 4	TGIF1	1715	602630	2, 4, 8, 9, 17
Holoprosencephaly type 5	ZIC2	1340	603073	2, 4, 8, 9, 17
Holoprosencephaly type 11	CDON	1717	608707	2, 4, 17
Holoprosencephaly-type 9	GLI2	1716	165230	2, 4, 8, 9, 17

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Holt-Oram syndrome	TBX5	1230	601620	2, 4, 8, 9, 17
Hutchinson-Gilford progeria	LMNA	158	150330	2, 4, 8, 9, 17
Hydranencephaly, Fowler type	FLVCR2	2340	610865	2, 4, 17
Hydrolethalus syndrome	HYLS1	139	610693	2, 4
Hypermethioninemia due to adenosine kinase deficiency	ADK	2602	102750	2, 4, 17
Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	AHCY	2485	180960	2, 4, 17
Hyperphosphatasia with mental retardation syndrome type 1	PIGV	2497	610274	2, 4
Hyperphosphatasia with mental retardation syndrome type 2	PIGO	1251	614730	2, 4, 17
Hyperphosphatasia with mental retardation syndrome type 3	PGAP2	2519	615187	2, 4
Hyperphosphatasia with mental retardation syndrome type 4	PGAP3	2453	611801	2, 4
Hyperphosphatasia with mental retardation syndrome type 5	PIGW	2396	610275	2, 4
Hypogonadism, alopecia, Diabetes mellitus, mental retardation and extrapyramidal syndrome	DCAF17	1231	612515	2, 3, 4, 8, 9, 17
Hypogonadotropic hypogonadism type 6 with or without anosmia	FGF8	1235	600483	2, 3, 4, 8, 9
Hypospadias type 1, X-linked	AR	20	313700	2, 4, 8, 9, 10, 17
Hypospadias type 2, X-linked	MAMLD1	1199	300120	2, 3, 4, 8, 9, 17
IMAGE syndrome	CDKN1C	1164	600856	2, 4, 8, 9
Immunodeficiency-centromeric instability-facial anomalies syndrome type 2	ZBTB24	2464	614064	2, 4
Intestinal atresia, multiple	TTC7A	1718	609332	2, 3, 4, 8, 9, 17
IVIC syndrome	SALL4	251	607343	2, 3, 4, 8, 9, 17
Jackson-Weiss syndrome	FGFR1	1136	136350	2, 4, 8, 9
Jackson-Weiss syndrome	FGFR2	1730	176943	2, 4, 8, 9, 17
Jawad syndrome	RBBP8	790	604124	2, 4, 17
Kabuki syndrome type 1	KMT2D	1232	602113	2, 4, 8, 9, 17
Kabuki syndrome type 2	KDM6A	720	300128	2, 3, 4, 8, 9, 17
Kagami-Ogata syndrome	paternal UPD chr. 14	2935		8
Kallmann syndrome type 2	FGFR1	1136	136350	2, 4, 8, 9
Kallmann syndrome type 4	PROK2	228	607002	2, 4, 8, 9
Kallmann syndrome type 5	CHD7	1221	608892	2, 4, 8, 9, 17
KBG syndrome	ANKRD11	1842	611192	2, 3, 4, 8, 9, 17
Keutel syndrome	MGP	1925	154870	2, 4
Kleefstra syndrome	EHMT1	1236	607001	2, 4, 8, 9, 17
Klippel-Feil syndrome type 1, autosomal dominant	GDF6	1997	601147	2, 4, 8, 9
Klippel-Feil syndrome type 2, autosomal dominant	MEOX1	2473	600147	2, 3, 4, 8, 9
Klippel-Feil syndrome type 3, autosomal dominant	GDF3	1948	606522	2, 4, 8, 9
Klippel-Feil syndrome type 4, autosomal dominant, with myopathy and facial dysmorphism	MYO18B	2816	607295	2, 17
Kniest dysplasia	COL2A1	943	120140	2, 4, 8, 9, 17
Koolen syndrome	KANSL1	1079	612452	2, 3, 4, 8, 9, 17
LADD syndrome	FGF10	1453	602115	2, 4, 8, 9
LADD syndrome	FGFR2	1730	176943	2, 4, 8, 9, 17
Langer-Giedion syndrome	EXT1	1733	608177	2, 4, 8, 9, 17
Langer-Giedion syndrome	TRPS1	120	604386	2, 4, 8, 9, 17
Larsen syndrome	FLNB	1884	603381	2, 4, 17
Left-right axis malformations	LEFTY2	2042	601877	2, 4
Legionnaire disease, susceptibility to	TLR5	3810	603031	17
Lenz-Majewski hyperostotic dwarfism	PTDSS1	2426	612792	2, 4, 17
LEOPARD syndrome type 3	BRAF	1078	164757	2, 4, 17
Lethal congenital contracture syndrome type 1	GLE1	2878	603371	2, 4, 17
Lethal congenital contracture syndrome type 4	MYBPC1	992	160794	2, 3, 4, 8, 9, 17
Limb-mammary syndrome	TP63	304	603273	2, 3, 4, 8, 9, 17
Lissencephaly type 1	PAFAH1B1	1238	601545	2, 4, 8, 9, 17
Lissencephaly type 2 (Norman-Roberts type)	RELN	63	600514	2, 17
Lissencephaly type 3	TUBA1A	1239	602529	2, 4
Lissencephaly type 5	LAMB1	1537	150240	2, 3, 4, 8, 9, 17
Lissencephaly, X-linked type 1	DCX	813	300121	2, 4, 8, 9, 17
Lissencephaly, X-linked type 2	ARX	24	300382	2, 4, 8, 9, 17
Lujan-Fryns syndrome	MED12	164	300188	2, 3, 4, 8, 9, 17
Lymphedema-distichiasis syndrome	FOXC2	1817	602402	2, 4, 8, 9
Macrocephaly, alopecia, cutis laxa, and scoliosis	RIN2	3059	610222	2, 4, 17
Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome	POLD1	2875	174761	2, 3, 4, 8, 9, 17
Mandibuloacral dysplasia	LMNA	158	150330	2, 4, 8, 9, 17
Mandibulofacial dysostosis with microcephaly	EFTUD2	1103	603892	2, 3, 4, 8, 9, 17
Marden-Walker syndrome	PIEZO2	2397	613629	2, 3, 4, 8, 9, 17
Marfan lipodystrophy syndrome	FBN1	950	134797	2, 8, 17

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Marfan syndrome	FBN1	950	134797	2, 8, 17
Martsolf syndrome	RAB3GAP2	1619	609275	2, 17
MASS syndrome	FBN1	950	134797	2, 8, 17
McLeod syndrome with or without chronic granulomatous disease	XK	2208	314850	2, 3, 4, 8, 9
Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome	PIK3R2	1885	603157	2, 3, 4, 8, 9, 17
Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome type 2	AKT3	1337	611223	2, 4, 17
Meier-Gorlin syndrome type 1	ORC1	1891	601902	2, 3, 4, 8, 9, 17
Meier-Gorlin syndrome type 2	ORC4	3543	603056	17
Meier-Gorlin syndrome type 4	CDT1	1927	605525	2, 4
Mental retardation and distinctive facial features with or without cardiac defects	MED13L	2912	608771	2, 4, 17
Mental retardation, autosomal dominant type 13	DYNC1H1	1279	600112	2, 17
Mental retardation, truncal obesity, retinal dystrophy, and micropenis	INPP5E	639	613037	2, 4, 17
Mental retardation, X-linked syndromic, Turner type	HUWE1	853	300697	2, 17
Metaphyseal anadysplasia type 1	MMP13	2087	600108	2, 4, 17
Metaphyseal anadysplasia type 2	MMP9	2086	120361	2, 4, 17
Microcephalic osteodysplastic primordial dwarfism type 2	PCNT	1869	605925	2, 3, 4, 8, 9, 17
Microcephaly and chorioretinopathy with or without mental retardation	TUBGCP6	751	610053	2, 3, 4, 8, 9, 17
Microcephaly with cortical malformations, autosomal recessive type 2	WDR62	1242	613583	2, 3, 4, 8, 9, 17
Microcephaly with epilepsy and diabetes syndrome	IER3IP1	806	609382	2, 4
Microcephaly with or without chorioretinopathy, Lymphedema, or Mental retardation, MCLMR	KIF11	1343	148760	2, 4, 17
Microcephaly with symplified gyral pattern and insulin-dependant diabetes	GFM2	1182	606544	2, 4, 17
Microcephaly, Amish type	SLC25A19	487	606521	2, 4
Microcephaly, AP4M1 related	AP4M1	1069	602296	2, 4, 17
Microcephaly, autosomal recessive type 1	MCPH1	659	607117	2, 4, 8, 9, 17
Microcephaly, autosomal recessive type 3	CDK5RAP2	1341	608201	2, 4, 8, 9, 17
Microcephaly, autosomal recessive type 4	KNL1	2016	609173	2, 4, 17
Microcephaly, autosomal recessive type 5	ASPM	660	605481	2, 4, 8, 9, 17
Microcephaly, autosomal recessive type 6	CENPJ	1342	609279	2, 4, 8, 9, 17
Microcephaly, autosomal recessive type 7	STIL	661	181590	2, 4
Microcephaly, autosomal recessive type 8	CEP135	1014	611423	2, 3, 4, 8, 9, 17
Microcephaly, autosomal recessive type 9	CEP152	1077	613529	2, 4, 17
Microcephaly, autosomal recessive type 11	PHC1	2637	602978	2, 4, 17
Microcephaly, autosomal recessive type 12	CDK6	2150	603368	2, 4, 17
Microcephaly, autosomal recessive type 13	CENPE	2635	117143	2, 4, 17
Microcephaly, CEP63 related	CEP63	738	614724	2, 4, 17
Microcephaly, MRE11A related	MRE11	1151	600814	2, 3, 4, 8, 9, 17
Microcephaly, MSMO1 related	MSMO1	938	607545	2, 4
Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	QARS1	2745	603727	2, 4, 17
Microcephaly, short stature, and polymicrogyria with seizures	RTTN	1719	610436	2, 4, 17
Microcephaly, TUBB2B related	TUBB2B	477	612850	2, 4
Miller Dieker lissencephaly syndrome	YWHAE	64	605066	2, 4, 8, 9
Mitochondrial complex III deficiency, nuclear type 7	UQCC2	2873	614461	2, 4
Mitochondrial myopathy and sideroblastic anemia type 1	PUS1	44	608109	2, 4
Mosaic variegated aneuploidy syndrome type 2	CEP57	2915	607951	2, 4
Mulibrey nanism	TRIM37	2939	605073	2, 3, 4, 8, 9, 17
Multiple congenital anomalies-hypotonia-seizures syndrome type 1	PIGN	1916	606097	2, 3, 4, 8, 9, 17
Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects	B3GAT3	1897	606374	2, 4
Myhre syndrome	SMAD4	277	600993	2, 4, 8, 9, 17
Native American myopathy	STAC3	2665	615521	2, 4, 17
Neuroaxonal neurodegeneration, infantile, with facial dysmophism	NALCN	2363	611549	2, 17
Neuroblastoma type 3, susceptibility to, familial	ALK	1747	105590	2, 4, 17
Nicolaides Baraitser syndrome	SMARCA2	946	600014	2, 3, 4, 8, 9, 17
Nijmegen breakage syndrome	NBN	344	602667	2, 4, 17
Noonan syndrom like	SHOC2	1244	602775	2, 3, 4, 8, 9, 17
Noonan syndrome type 1	PTPN11	1245	176876	2, 3, 4, 8, 9, 17
Noonan syndrome type 3	KRAS	152	190070	2, 3, 4, 8, 9
Noonan syndrome type 4	SOS1	1246	182530	2, 4, 17
Noonan syndrome type 5	RAF1	1247	164760	2, 3, 4, 8, 9
Noonan syndrome type 6	NRAS	1248	164790	2, 4
Noonan syndrome type 7	BRAF	1078	164757	2, 4, 17
Noonan syndrome type 8	RIT1	2392	609591	2, 4
Noonan syndrome type 10	LZTR1	2963	600574	2, 4, 8, 9, 17
Noonan syndrome-like disorder with or without juvenile meylomonocytic leukemia	CBL	115	165360	2, 3, 4, 8, 9, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Oculodentodigital dysplasia	GJA1	1250	121014	2, 4, 8, 9
Ogden syndrome	NAA10	2351	300013	2, 4, 17
Olmsted syndrome	TRPV3	739	607066	2, 4, 17
Opitz-Kaveggia syndrome	MED12	164	300188	2, 3, 4, 8, 9, 17
Orofacial cleft type 5	MSX1	170	142983	2, 4, 8, 9
Orofacial cleft type 6	IRF6	2826	607199	2, 4, 8, 9, 17
Orofacial cleft type 7	NECTIN1	2828	600644	2, 4
Orofacial cleft type 10	SUMO1	2827	601912	2, 4
Orofacial cleft type 11	BMP4	891	112262	2, 4, 8, 9
Orofaciodigital syndrome type 4	TCTN3	2200	613847	2, 3, 4, 8, 9, 17
Orofaciodigital syndrome type 5	DDX59	2110	615464	2, 3, 4, 8, 9
Orofaciodigital syndrome type 6	CPLANE1	637	614571	2, 17
Orofaciodigital syndrome type 14	C2CD3	2278	615944	2, 4, 17
Osteoglophonic dysplasia	FGFR1	1136	136350	2, 4, 8, 9
Otofaciocervical syndrome	EYA1	125	601653	2, 4, 8, 9, 17
Otospondylomegaepiphyseal dysplasia	COL2A1	943	120140	2, 4, 8, 9, 17
Otospondylomegaepiphyseal dysplasia	COL11A2	945	120290	2, 4, 17
Pallister-Hall syndrome	GLI3	128	165240	2, 3, 4, 8, 9, 17
Papillorenal syndrome	PAX2	1252	167409	2, 3, 4, 8, 9, 17
Parietal foramina type 1	MSX2	171	123101	2, 4, 8, 9
Pelger-Huet anomaly	LBR	2446	600024	2, 4, 17
Pelvic organ prolapse, LAMC1 related	LAMC1	2629	150290	2, 4, 17
Perlman Syndrome	DIS3L2	1344	614184	2, 3, 4, 8, 9, 17
Pfeiffer syndrome	FGFR1	1136	136350	2, 4, 8, 9, 17
Pfeiffer syndrome	FGFR2	1730	176943	2, 4, 8, 9, 17
Phelan-McDermid syndrome	chr. 22q13.3	2899		8
Pitt-Hopkins syndrome	NRXN1	1254	600565	2, 4, 8, 9, 17
Pitt-Hopkins syndrome	TCF4	1253	602272	2, 4, 8, 9, 17
Pituitary hormone deficiency type 1	POU1F1	1255	173110	2, 4, 8, 9
Pituitary hormone deficiency type 2	PROP1	1256	601538	2, 4, 8, 9
Platyspondylic skeletal dysplasia, Torrance type	COL2A1	943	120140	2, 4, 8, 9, 17
Polyhydramnios, megalencephaly, and symptomatic epilepsy	STRADA	3710	608626	17
Polymicrogyria asymmetric	TUBB2B	477	612850	2, 4
Polymicrogyria bilateral frontoparietal	ADGRG1	1720	604110	2, 3, 4, 8, 9, 17
Polymicrogyria bilateral occipital	NR2E1	631	603849	2, 4
Polymicrogyria with optic nerve hypoplasia	TUBA8	1721	605742	2, 3, 4, 8, 9
Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis	PI4KA	2762	600286	2, 4
Popliteal pterygium syndrome type 1	IRF6	2826	607199	2, 4, 8, 9, 17
Popliteal pterygium syndrome, lethal type	RIPK4	1974	605706	2, 4
Porencephaly, familial	COL4A1	1257	120130	2, 3, 4, 8, 9, 17
Poretti-Boltshauser syndrome	LAMA1	2498	150320	2, 17
Postaxial acrofacial dysostosis	DHODH	1258	126064	2, 3, 4, 8, 9, 17
Prolidase deficiency	PEPD	2206	613230	2, 4, 17
Prune belly syndrome	CHRM3	2857	118494	2, 4
Rapp-Hodgkin syndrome	TP63	304	603273	2, 3, 4, 8, 9, 17
Retinal dystrophy, juvenile cataracts, and short stature syndrome	RDH11	3797	607849	17
Rett syndrome	MECP2	163	300005	2, 4, 8, 9
Rett syndrome, congenital variant	FOXG1	1259	164874	2, 4, 8, 9
Rhizomelic chondrodysplasia punctata type 2	GNPAT	1957	602744	2, 4, 17
Rhizomelic chondrodysplasia punctata type 3	AGPS	1374	603051	2, 4, 17
Rhizomelic chondrodysplasia punctata type 5	PEX5	681	600414	2, 3, 4, 8, 9, 17
Ritscher-Schinzel syndrome type 1	WASHC5	493	610657	2, 3, 4, 8, 9, 17
RNA processing related disorders	HNRNPU	1913	602869	2, 4, 17
Roberts syndrome	ESCO2	1260	609353	2, 3, 4, 8, 9, 17
Robinow syndrome, autosomal dominant type 1	WNT5A	123	164975	2, 3, 4, 8, 9, 17
Robinow syndrome, autosomal dominant type 2	DVL1	2675	601365	2, 4
Robinow syndrome, autosomal recessive	ROR2	244	602337	2, 4, 8, 9, 17
Robinow-Sorauf syndrome	TWIST1	1541	601622	2, 4, 8, 9
Rubinstein-Taybi syndrome	CREBBP	1261	600140	2, 4, 8, 9, 17
Rubinstein-Taybi syndrome	EP300	1262	602700	2, 4, 8, 9, 17
Saethre-Chotzen syndrome	FGFR2	1730	176943	2, 4, 8, 9, 17
Saethre-Chotzen syndrome	TWIST1	1541	601622	2, 4, 8, 9
SC Phocomelia syndrome	ESCO2	1260	609353	2, 3, 4, 8, 9, 17
Scaphocephaly, maxillary retrusion, and mental retardation	FGFR2	1730	176943	2, 4, 8, 9, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Schaaf-Yang syndrome	MAGEL2	2820	605283	2, 4, 8, 9, 17
Schinzel-Giedion midface retraction syndrome	SETBP1	2203	611060	2, 4, 17
Schizencephaly	EMX2	1722	600035	2, 3, 4, 8, 9
Schneckenbecken dysplasia	SLC35D1	2448	610804	2, 4, 17
Sclerosteosis type 1	SOST	2427	605740	2, 4
Seckel syndrome	ATRIP	1852	606605	2, 4, 17
Seckel syndrome type 1	ATR	1851	601215	2, 17
Seckel syndrome type 2	RBBP8	790	604124	2, 4, 17
Seckel syndrome type 4	CENPJ	1342	609279	2, 4, 8, 9, 17
Seckel syndrome type 5	CEP152	1077	613529	2, 4, 17
Seckel syndrome type 6	CEP63	738	614724	2, 4, 17
Seckel syndrome type 7	NIN	2636	608684	2, 4, 17
SED congenita	COL2A1	943	120140	2, 4, 8, 9, 17
Seizures, scoliosis, and macrocephaly syndrome	EXT2	2375	608210	2, 4, 8, 9, 17
Septooptic dysplasia	HESX1	1265	601802	2, 4, 8, 9
SERKAL syndrome	WNT4	122	603490	2, 4, 8, 9
Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	NHEJ1	1211	611290	2, 3, 4, 8, 9
Short stature syndrome	SHOX	1360	312865	2, 4, 8, 9
Short stature, microcephaly, and endocrine dysfunction	XRCC4	2798	194363	2, 3, 4, 8, 9
Short stature, optic nerve atrophy, and Pelger-Huet anomaly	NBAS	2605	608025	2, 4, 17
SHORT syndrome	PIK3R1	2523	171833	2, 4, 17
Short-rib thoracic dysplasia type 2 with or without polydactyly	IFT80	2440	611177	2, 4, 17
Short-rib thoracic dysplasia type 3 with or without polydactyly	DYNC2H1	2438	603297	2, 17
Short-rib thoracic dysplasia type 4 with or without polydactyly	TTC21B	2001	612014	2, 3, 4, 8, 9, 17
Short-rib thoracic dysplasia type 5 with or without polydactyly	WDR19	2424	608151	2, 4, 17
Short-rib thoracic dysplasia type 6 with or without polydactyly	NEK1	1984	604588	2, 4, 17
Short-rib thoracic dysplasia type 7 with or without polydactyly	WDR35	2425	613602	2, 4, 17
Short-rib thoracic dysplasia type 8 with or without polydactyly	WDR60	2444	615462	2, 4, 17
Short-rib thoracic dysplasia type 10 with or without polydactyly	IFT172	2441	607386	2, 4, 17
Short-rib thoracic dysplasia type 11 with or without polydactyly	WDR34	2442	613363	2, 4, 17
Shprintzen-Goldberg syndrome	SKI	88	164780	2, 3, 4, 8, 9, 17
Silver-Russell syndrome	chr. 11p15	92		8
Silver-Russell syndrome	maternal UPD chr. 7	2933		8
Skeletal abnormalities, CBFB related	CBFB	1996	121360	2, 4
Skeletal defects, genital hypoplasia, and mental retardation	ZBTB16	3818	176797	17
SMED Strudwick type	COL2A1	943	120140	2, 4, 8, 9, 17
Smith-Lemli-Opitz syndrome	DHCR7	1809	602858	2, 3, 4, 8, 9, 17
Sotos syndrome type 1	NSD1	201	606681	2, 4, 8, 9, 17
Sotos-like syndrome	NFIX	1277	164005	2, 3, 4, 8, 9, 17
Speech-language disorder type 1	FOXP2	1266	605317	2, 3, 4, 8, 9, 17
Spina bifida folate sensitive	MTRR	982	602568	2, 4, 17
Split-hand/foot malformation type 1 with sensorineural hearing loss	DLX5	2249	600028	2, 3, 4, 8, 9
Split-hand/foot malformation type 6	WNT10B	2250	601906	2, 3, 4, 8, 9
Spondylocostal dysostosis type 5	TBX6	2802	602427	2, 4
Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	B3GALT6	3056	615291	2, 4
Spondylo-megaepiphyseal-metaphyseal dysplasia	NKX3-2	2085	602183	2, 4, 8, 9
Spondyloperipheral dysplasia	COL2A1	943	120140	2, 4, 8, 9, 17
Stiff skin syndrome	FBN1	950	134797	2, 8, 17
Stuve-Wiedemann syndrome	LIFR	1267	151443	2, 3, 4, 8, 9, 17
Syndactyly type 1	HOXD13	1975	142989	2, 4, 8, 9
Syndactyly type 5	HOXD13	1975	142989	2, 4, 8, 9
Syndactyly, mesoaxial synostotic, with phalangeal reduction	BHLHA9	2968	615416	2, 4, 8, 9
TANC2 related brain disorders	TANC2	1943	615047	2, 4, 17
Tarp syndrome	RBM10	3739	300080	17
Temple syndrome	maternal UPD chr. 14	2934		8
Temple-Baraitser syndrome	KCNH1	2792	603305	2, 4, 17
Temtamy preaxial brachydactyly syndrome	CHSY1	3771	608183	17
Temtamy syndrome	C12orf57	2129	615140	2, 4
Tetraamelia, autosomal recessive	WNT3	1268	165330	2, 4
Three M syndrome type 1	CUL7	1269	609577	2, 3, 4, 8, 9, 17
Three M syndrome type 2	OBSL1	1952	610991	2, 3, 4, 8, 9, 17
Three M syndrome type 3	CCDC8	2468	614145	2, 4

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Disease	Gene	Test code	OMIM Gene	Available test methods
Toe syndactyly, telecanthus, and anogenital and renal malformations	FAM58A	1270	300708	2, 4
Tooth agenesis, selective type 1	MSX1	170	142983	2, 4, 8, 9
Tooth agenesis, selective type 3	PAX9	1205	167416	2, 3, 4, 8, 9
Townes-Brocks syndrome	SALL1	1271	602218	2, 3, 4, 8, 9
Transposition of great arteries, dextro-looped 3	GDF1	2040	602880	2, 3, 4, 8, 9
Transposition of the great arteries, dextro-looped 1	MED13L	2912	608771	2, 4, 17
Treacher Collins syndrome type 1	TCOF1	2130	606847	2, 4, 8, 9, 17
Treacher Collins syndrome type 2	POLR1D	2178	613715	2, 4, 8, 9
Treacher Collins syndrome type 3	POLR1C	2179	610060	2, 4
Trigonocephaly type 1	FGFR1	1136	136350	2, 4, 8, 9
Ulna and fibula, absence of, with severe limb deficiency	WNT7A	327	601570	2, 3, 4, 8, 9
Ulnar-Mammary syndrome	TBX3	1272	601621	2, 3, 4, 8, 9
Urofacial syndrome	LRIG2	1524	608869	2, 4, 17
Urofacial syndrome type 1	HPSE2	3783	613469	17
Van den Ende-Gupta syndrome	SCARF2	1882	613619	2, 4, 17
van der Woude syndrome type 1	IRF6	2826	607199	2, 4, 8, 9, 17
van der Woude syndrome type 2	GRHL3	1892	608317	2, 4
Van Maldergem syndrome type 2	FAT4	2580	612411	2, 4, 17
Vater association	HOXD13	1975	142989	2, 4, 8, 9
Vici syndrome	EPG5	2090	615068	2, 17
Visceral myopathy	ACTG2	2882	102545	2, 3, 4, 8, 9
Warburg micro syndrome type 1	RAB3GAP1	237	602536	2, 3, 4, 8, 9, 17
Warburg micro syndrome type 2	RAB3GAP2	1619	609275	2, 17
Warsaw breakage syndrome	DDX11	2690	601150	2, 4, 17
Weaver syndrome	EZH2	1872	601573	2, 3, 4, 8, 9, 17
Webb-Dattani syndrome	ARNT2	2781	606036	2, 4, 17
Weill-Marchesani syndrome type 3	LTBP2	887	602091	2, 4, 17
Weill-Marchesani syndrome, dominant type 2	FBN1	950	134797	2, 8, 17
Werner syndrome	WRN	2545	604611	2, 17
Wiedemann-Steiner syndrome	KMT2A	1432	159555	2, 3, 4, 8, 9, 17
Williams-Beuren syndrome	chr. 7q11.23	2760		8
Witkop syndrome	MSX1	170	142983	2, 4, 8, 9
XFE progeroid syndrome	ERCC4	2543	133520	2, 4, 17
ZIC5 related brain disorders	ZIC5	2199		2, 4

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› Oncology - Panels

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
BRCA1, BRCA2 panel	BRCA1, BRCA2	BRCA1, BRCA2	5469	5, 8
BRCA1, BRCA2 panel	BRCA1, BRCA2	BRCA1, BRCA2	5469	5, 8
BRCA1, BRCA2 panel	BRCA1, BRCA2	BRCA1, BRCA2	5469	5, 8
BRCA1, BRCA2 panel Combi (with MLPA)	BRCA1, BRCA2	BRCA2, BRCA1	5390	6
BRCA1, BRCA2 panel Combi (with MLPA)	BRCA1, BRCA2	BRCA2, BRCA1	5390	6
BRCA1, BRCA2 panel Combi (with MLPA)	BRCA1, BRCA2	BRCA2, BRCA1	5390	6
BRCA1, BRCA2 panel Plus	BRCA1, BRCA2	BRCA2, BRCA1	5471	6
BRCA1, BRCA2 panel Plus	BRCA1, BRCA2	BRCA2, BRCA1	5471	6
BRCA1, BRCA2 panel Plus	BRCA1, BRCA2	BRCA2, BRCA1	5471	6
BRCA1, BRCA2 somatic mutation analysis	BRCA1, BRCA2		5468	11
BRCA1, BRCA2 somatic mutation analysis	BRCA1, BRCA2		5468	11
BRCA1, BRCA2 somatic mutation analysis	BRCA1, BRCA2		5468	11
CentoBreast®	ABRAXAS1, ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, FANCC, MEN1, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, PALB2, PMS1, PMS2, PTEN, RAD50, RAD51C, RAD51D, RECQL, SMARCA4, STK11, TP53, XRCC2		5441	6
CentoBreast®	ABRAXAS1, ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, FANCC, MEN1, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, PALB2, PMS1, PMS2, PTEN, RAD50, RAD51C, RAD51D, RECQL, SMARCA4, STK11, TP53, XRCC2		5441	6
CentoBreast®	ABRAXAS1, ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, FANCC, MEN1, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, PALB2, PMS1, PMS2, PTEN, RAD50, RAD51C, RAD51D, RECQL, SMARCA4, STK11, TP53, XRCC2		5441	6
CentoCancer®	ABRAXAS1, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, DICER1, DIS3L2, EPCAM, FANCC, FH, FLCN, GALNT12, HNF1B, HOXB13, KIT, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS1, PMS2, POLD1, POLE, POT1, PRSS1, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RECQL, RET, RNF43, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, STK11, TGFBR2, TP53, TSC1, TSC2, VHL, WT1, XRCC2, XRCC3		5442	6
CentoCancer®	ABRAXAS1, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, DICER1, DIS3L2, EPCAM, FANCC, FH, FLCN, GALNT12, HNF1B, HOXB13, KIT, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS1, PMS2, POLD1, POLE, POT1, PRSS1, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RECQL, RET, RNF43, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, STK11, TGFBR2, TP53, TSC1, TSC2, VHL, WT1, XRCC2, XRCC3		5442	6
CentoCancer®	ABRAXAS1, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, DICER1, DIS3L2, EPCAM, FANCC, FH, FLCN, GALNT12, HNF1B, HOXB13, KIT, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS1, PMS2, POLD1, POLE, POT1, PRSS1, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RECQL, RET, RNF43, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, STK11, TGFBR2, TP53, TSC1, TSC2, VHL, WT1, XRCC2, XRCC3		5442	6

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6: NGS Panel + CNV
7: NGS Panel Genomic
8: Deletion/Duplication with qPCR/MLPA
9: Sequencing + Deletion/Duplication
10: Repeat Expansions
11: Somatic Mutation Analysis
12: Biochemical Enzyme Analysis
13: Biomarker Analysis
14: Biochemical Enzyme Panel
15: Biochemical Genetics Panel Plus
16: Single Gene Sequencing (NGS)
17: Single Gene Sequencing + CNV (NGS)

Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
CentoCancer® comprehensive panel	ABRAXAS1, ACVRL1, AKT1, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CHEK2, CTNNA1, DDX41, DICER1, DIS3L2, EGFR, EPCAM, ETV6, EXT1, EXT2, FANCC, FH, FLCN, GALNT12, GATA2, GPC3, GREM1, HNF1A, HNF1B, HOXB13, HRAS, KIF1B, KIT, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PDGFRA, PHOX2B, PIK3CA, PMS1, PMS2, POLD1, POLE, POT1, PRKAR1A, PRSS1, PTCH1, PTCH2, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL, REST, RET, RNF43, RPS20, RUNX1, SAMD9L, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TERT, TGFBR2, TMEM127, TP53, TRIP13, TSC1, TSC2, VHL, WRN, WT1, XRCC2, XRCC3		5462	6
CentoCancer® comprehensive panel	ABRAXAS1, ACVRL1, AKT1, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CHEK2, CTNNA1, DDX41, DICER1, DIS3L2, EGFR, EPCAM, ETV6, EXT1, EXT2, FANCC, FH, FLCN, GALNT12, GATA2, GPC3, GREM1, HNF1A, HNF1B, HOXB13, HRAS, KIF1B, KIT, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PDGFRA, PHOX2B, PIK3CA, PMS1, PMS2, POLD1, POLE, POT1, PRKAR1A, PRSS1, PTCH1, PTCH2, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL, REST, RET, RNF43, RPS20, RUNX1, SAMD9L, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TERT, TGFBR2, TMEM127, TP53, TRIP13, TSC1, TSC2, VHL, WRN, WT1, XRCC2, XRCC3		5462	6
CentoCancer® comprehensive panel	ABRAXAS1, ACVRL1, AKT1, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CHEK2, CTNNA1, DDX41, DICER1, DIS3L2, EGFR, EPCAM, ETV6, EXT1, EXT2, FANCC, FH, FLCN, GALNT12, GATA2, GPC3, GREM1, HNF1A, HNF1B, HOXB13, HRAS, KIF1B, KIT, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PDGFRA, PHOX2B, PIK3CA, PMS1, PMS2, POLD1, POLE, POT1, PRKAR1A, PRSS1, PTCH1, PTCH2, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL, REST, RET, RNF43, RPS20, RUNX1, SAMD9L, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TERT, TGFBR2, TMEM127, TP53, TRIP13, TSC1, TSC2, VHL, WRN, WT1, XRCC2, XRCC3		5462	6
CentoColon	APC, ATM, AXIN2, BLM, BMPR1A, BRCA1, BRCA2, CDH1, CDKN2A, CHEK2, EPCAM, FLCN, GALNT12, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, POLD1, POLE, PRSS1, PTEN, RNF43, SMAD4, STK11, TGFBR2, TP53, VHL		5443	6
CentoColon	APC, ATM, AXIN2, BLM, BMPR1A, BRCA1, BRCA2, CDH1, CDKN2A, CHEK2, EPCAM, FLCN, GALNT12, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, POLD1, POLE, PRSS1, PTEN, RNF43, SMAD4, STK11, TGFBR2, TP53, VHL		5443	6
CentoColon	APC, ATM, AXIN2, BLM, BMPR1A, BRCA1, BRCA2, CDH1, CDKN2A, CHEK2, EPCAM, FLCN, GALNT12, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, POLD1, POLE, PRSS1, PTEN, RNF43, SMAD4, STK11, TGFBR2, TP53, VHL		5443	6
Myeloid tumor panel	ASXL1, CEBPA, DNMT3A, ETV6, EZH2, IDH1, IDH2, KIT, KRAS, NPM1, NRAS, PTPN11, RAD21, RUNX1, SF3B1, SMC1A, SMC3, STAG2, TET2, TP53, U2AF1, WT1		5453	11
Myeloid tumor panel	ASXL1, CEBPA, DNMT3A, ETV6, EZH2, IDH1, IDH2, KIT, KRAS, NPM1, NRAS, PTPN11, RAD21, RUNX1, SF3B1, SMC1A, SMC3, STAG2, TET2, TP53, U2AF1, WT1		5453	11

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Panel name	Genes	Deletion / duplication testing (genes analyzed)	Test code	Available test methods
Myeloid tumor panel	ASXL1, CEBPA, DNMT3A, ETV6, EZH2, IDH1, IDH2, KIT, KRAS, NPM1, NRAS, PTPN11, RAD21, RUNX1, SF3B1, SMC1A, SMC3, STAG2, TET2, TP53, U2AF1, WT1		5453	11
Solid tumor panel	ABL1, AKT1, AKT2, AKT3, ALK, APC, AR, ARAF, ARID1A, ASXL1, ATM, ATR, ATRX, AXL, BAP1, BRAF, BRCA1, BRCA2, BTK, CBL, CCND1, CDH1, CDK12, CDK4, CDK6, CDKN1B, CDKN2A, CDKN2B, CHEK1, CHEK2, CREBBP, CSF1R, CTNNB1, DDR2, EGFR, ERBB2, ERBB3, ERBB4, ERCC2, ESR1, EZH2, FANCA, FANCD2, FANCI, FBXW7, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, FOXL2, GATA2, GNA11, GNAQ, GNAS, H3-3A, H3C2, HNF1A, HRAS, IDH1, IDH2, JAK1, JAK2, JAK3, KDR, KEAP1, KIT, KMT2A, KMT2C, KMT2D, KNSTRN, KRAS, MAGOH, MAP2K1, MAP2K2, MAP2K4, MAPK1, MAX, MDM4, MED12, MEN1, MET, MLH1, MPL, MRE11, MSH2, MSH6, MTOR, MYC, MYCN, MYD88, NBN, NF1, NF2, NFE2L2, NOTCH1, NOTCH2, NOTCH3, NRAS, NTRK1, NTRK2, NTRK3, PALB2, PDGFRA, PDGFRB, PIK3CA, PIK3CB, PIK3R1, PMS2, POLE, PPP2R1A, PTCH1, PTEN, PTPN11, RAC1, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAF1, RB1, RBM10, RET, RHEB, RHOA, RIT1, RNF43, ROS1, SETD2, SF3B1, SLX4, SMAD4, SMARCA4, SMARCB1, SMO, SPOP, SRC, STAT3, STK11, TERT, TOP1, TP53, TSC1, TSC2, TSHR, U2AF1, VHL, XPO1		5457	11
Solid tumor panel	ABL1, AKT1, AKT2, AKT3, ALK, APC, AR, ARAF, ARID1A, ASXL1, ATM, ATR, ATRX, AXL, BAP1, BRAF, BRCA1, BRCA2, BTK, CBL, CCND1, CDH1, CDK12, CDK4, CDK6, CDKN1B, CDKN2A, CDKN2B, CHEK1, CHEK2, CREBBP, CSF1R, CTNNB1, DDR2, EGFR, ERBB2, ERBB3, ERBB4, ERCC2, ESR1, EZH2, FANCA, FANCD2, FANCI, FBXW7, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, FOXL2, GATA2, GNA11, GNAQ, GNAS, H3-3A, H3C2, HNF1A, HRAS, IDH1, IDH2, JAK1, JAK2, JAK3, KDR, KEAP1, KIT, KMT2A, KMT2C, KMT2D, KNSTRN, KRAS, MAGOH, MAP2K1, MAP2K2, MAP2K4, MAPK1, MAX, MDM4, MED12, MEN1, MET, MLH1, MPL, MRE11, MSH2, MSH6, MTOR, MYC, MYCN, MYD88, NBN, NF1, NF2, NFE2L2, NOTCH1, NOTCH2, NOTCH3, NRAS, NTRK1, NTRK2, NTRK3, PALB2, PDGFRA, PDGFRB, PIK3CA, PIK3CB, PIK3R1, PMS2, POLE, PPP2R1A, PTCH1, PTEN, PTPN11, RAC1, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAF1, RB1, RBM10, RET, RHEB, RHOA, RIT1, RNF43, ROS1, SETD2, SF3B1, SLX4, SMAD4, SMARCA4, SMARCB1, SMO, SPOP, SRC, STAT3, STK11, TERT, TOP1, TP53, TSC1, TSC2, TSHR, U2AF1, VHL, XPO1		5457	11
Solid tumor panel	ABL1, AKT1, AKT2, AKT3, ALK, APC, AR, ARAF, ARID1A, ASXL1, ATM, ATR, ATRX, AXL, BAP1, BRAF, BRCA1, BRCA2, BTK, CBL, CCND1, CDH1, CDK12, CDK4, CDK6, CDKN1B, CDKN2A, CDKN2B, CHEK1, CHEK2, CREBBP, CSF1R, CTNNB1, DDR2, EGFR, ERBB2, ERBB3, ERBB4, ERCC2, ESR1, EZH2, FANCA, FANCD2, FANCI, FBXW7, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, FOXL2, GATA2, GNA11, GNAQ, GNAS, H3-3A, H3C2, HNF1A, HRAS, IDH1, IDH2, JAK1, JAK2, JAK3, KDR, KEAP1, KIT, KMT2A, KMT2C, KMT2D, KNSTRN, KRAS, MAGOH, MAP2K1, MAP2K2, MAP2K4, MAPK1, MAX, MDM4, MED12, MEN1, MET, MLH1, MPL, MRE11, MSH2, MSH6, MTOR, MYC, MYCN, MYD88, NBN, NF1, NF2, NFE2L2, NOTCH1, NOTCH2, NOTCH3, NRAS, NTRK1, NTRK2, NTRK3, PALB2, PDGFRA, PDGFRB, PIK3CA, PIK3CB, PIK3R1, PMS2, POLE, PPP2R1A, PTCH1, PTEN, PTPN11, RAC1, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAF1, RB1, RBM10, RET, RHEB, RHOA, RIT1, RNF43, ROS1, SETD2, SF3B1, SLX4, SMAD4, SMARCA4, SMARCB1, SMO, SPOP, SRC, STAT3, STK11, TERT, TOP1, TP53, TSC1, TSC2, TSHR, U2AF1, VHL, XPO1		5457	11

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Disease	Gene	Test code	OMIM Gene	Available test methods
Acute myeloid leukemia, somatic, DNMT3A related	DNMT3A	30014	602769	2, 11
Basal cell nevus syndrome	PTCH1	1216	601309	2, 4, 8, 9, 17
Basal cell nevus syndrome	SUFU	2360	607035	2, 3, 4, 8, 9, 17
Basal cell nevus syndrome due to germline PTCH2 mutation	PTCH2	2687	603673	2, 4, 17
Beckwith-Wiedemann syndrome	CDKN1C	1164	600856	2, 4, 8, 9
Beckwith-Wiedemann syndrome	chr. 11p15	92		8
Beckwith-Wiedemann syndrome	H19	1750	103280	2, 4, 8, 9
Beckwith-Wiedemann syndrome	KCNQ1OT1	1749	604115	8
BRAF somatic Hotspot: c.1799T>A p.V600E	BRAF	46008	164757	1
BRAF, selective sequencing of exon 15	BRAF	45013	164757	1
Breast cancer, male, susceptibility to	BRCA2	379	600185	2, 4, 8, 9, 16
Breast cancer, RINT1 related	RINT1	2626	610089	2, 4, 17
Breast cancer, susceptibility to	BARD1	986	601593	2, 4, 17
Breast cancer, susceptibility to	PALB2	388	610355	2, 4, 8, 9, 17
Breast cancer, susceptibility to	RECQL	3060	600537	2, 4
Breast cancer, susceptibility to	XRCC3	2332	600675	2, 4, 17
Breast-ovarian cancer	BRCA1	1165	113705	2, 4, 8, 9, 16
Breast-ovarian cancer	RAD51C	1167	602774	2, 4, 8, 9, 17
Breast-ovarian cancer, familial, susceptibility to, type 4	RAD51D	1168	602954	2, 4, 8, 9, 17
Breast-ovarian cancer, familial, type 2	BRCA2	379	600185	2, 4, 8, 9, 16
CALR, selective sequencing of exon 9	CALR	45019	109091	1
Carcinoid tumors, intestinal	SDHD	1190	602690	2, 4, 8, 9
Carney complex type 1	PRKAR1A	1192	188830	2, 3, 4, 8, 9, 17
Cell cycle disorder, CDC20 related	CDC20	2478	603618	2, 4
Colorectal cancer, hereditary	NRAS	1248	164790	2, 4
Colorectal cancer, hereditary nonpolyposis type 1	MSH2	1170	609309	2, 4, 8, 9, 17
Colorectal cancer, hereditary nonpolyposis type 2	MLH1	1171	120436	2, 4, 8, 9, 17
Colorectal cancer, hereditary nonpolyposis type 4	PMS2	1172	600259	2, 4, 8, 9
Colorectal cancer, hereditary nonpolyposis type 5	MSH6	1173	600678	2, 4, 8, 9, 17
Colorectal cancer, hereditary nonpolyposis type 6	TGFBR2	295	190182	2, 3, 4, 8, 9, 17
Colorectal cancer, hereditary nonpolyposis type 7	MLH3	1174	604395	2, 4, 17
Colorectal cancer, hereditary nonpolyposis type 8	EPCAM	1175	185535	2, 4, 8, 9, 17
Colorectal cancer, hereditary, susceptibility to	CCND1	2134	168461	2, 4, 8, 9
Cowden syndrome type 1	PTEN	1176	601728	2, 4, 8, 9, 17
Cowden syndrome type 3	SDHD	1190	602690	2, 4, 8, 9
Cowden syndrome type 5	PIK3CA	917	171834	2, 3, 4, 8, 9, 17
Cowden syndrome type 6	AKT1	1832	164730	2, 3, 4, 8, 9, 17
Cutaneous telangiectasia and cancer syndrome, familial	ATR	1851	601215	2, 17
Cylindromatosis, familial	CYLD	2541	605018	2, 3, 4, 8, 9, 17
Desmoid disease, hereditary	APC	1163	611731	2, 4, 8, 9, 17
EGFR somatic Hotspot: c.2573T>G, p.L858R	EGFR	46003	131550	1
EGFR, selective sequencing of exons 18-21	EGFR	45017	131550	1
Endometrial cancer, familial, MSH6 related	MSH6	1173	600678	2, 4, 8, 9, 17
Familial adenomatous polyposis coli	APC	1163	611731	2, 4, 8, 9, 17
Familial adenomatous polyposis type 2	MUTYH	1169	604933	2, 4, 8, 9, 17
Familial adenomatous polyposis type 3	NTHL1	2874	602656	2, 3, 4, 8, 9
Familial adenomatous polyposis type 4	MSH3	56	600887	2, 3, 4, 8, 9, 17
Gastric cancer, hereditary diffuse	CDH1	1177	192090	2, 4, 8, 9, 17
Gastrointestinal stromal tumor, familial	KIT	1178	164920	2, 4, 8, 9, 17
Glioblastoma type 3, susceptibility to, due to BRCA2 germline mutation	BRCA2	379	600185	2, 4, 8, 9, 16
Granulomatous disease, chronic, autosomal recessive, cytochrome b- positive, type 1	NCF1	1968	608512	2, 3, 4, 8, 9, 17
Hemangioma capillary infantile	ANTXR1	1707	606410	2, 3, 8, 17
Hemangioma, capillary infantile, familial, susceptibility to	KDR	2141	191306	2, 4, 17
Hereditary breast and ovarian cancer syndrome, RAD50 related	RAD50	2321	604040	2, 4, 8, 9, 17
Hereditary Retinoblastoma	RB1	1643	614041	2, 4, 8, 9, 17
Hurthle cell thyroid carcinoma, due to germline NDUFA13 mutation	NDUFA13	40	609435	2, 4
IDH1, selective sequencing of exon 4	IDH1	45015	147700	1
IDH2, selective sequencing of exon 4	IDH2	45016	147650	1
JAK2, selective sequencing of exons 12, 14 and 16	JAK2	45014	147796	1
Juvenile myelomonocytic leukemia, due to CBL germline mutation	CBL	115	165360	2, 3, 4, 8, 9, 17
Juvenile polyposis syndrome	BMPR1A	1179	601299	2, 4, 8, 9, 17
Juvenile polyposis syndrome	SMAD4	277	600993	2, 4, 8, 9, 17

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Disease	Gene	Test code	OMIM Gene	Available test methods
Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	SMAD4	277	600993	2, 4, 8, 9, 17
KIT, selective sequencing of exons 8, 9, 11, 13 and 17	KIT	45018	164920	1
KRAS somatic Hotspot: c.35G>A, p.G12D	KRAS	46005	190070	1
KRAS somatic Hotspot: c.35G>T p.G12V	KRAS	46006	190070	1
KRAS somatic Hotspot: c.38G>A, p.G13D	KRAS	46007	190070	1
KRAS, selective sequencing of exon 2 and 3	KRAS	45026	190070	1
Leukemia, acute lymphoblastic, susceptibility to, due to PAX5 germline mutation	PAX5	2377	167414	2, 4, 8, 9
Leukemia, acute myelogenous	JAK2	30031	147796	11
Leukemia, acute myelogenous	KRAS	30035	190070	11
Leukemia, acute myeloid	KIT	30033	164920	11
Leukemia, acute myeloid	RUNX1	30049	151385	11
Leukemia, acute myeloid, somatic	CEBPA	30010	116897	11
Leukemia, acute promyelocytic, PL2F/RARA type	ZBTB16	3818	176797	17
Leukemia, juvenile myelomonocytic	PTPN11	30047	176876	11
Leukemia, lymphoblastic and myeloid, EZH2 related	EZH2	30018	601573	11
Leukemia, myeloid acute form, due to CEBPA germline mutation	CEBPA	1934	116897	2, 4, 8, 9
Li-Fraumeni syndrome type 1	TP53	1180	191170	2, 4, 8, 9, 17
Li-Fraumeni syndrome type 2	CHEK2	1181	604373	2, 4, 8, 9, 17
Medulloblastoma, desmoplastic, familial	SUFU	2360	607035	2, 3, 4, 8, 9, 17
Medulloblastoma, due to BRCA2 germline mutation	BRCA2	379	600185	2, 4, 8, 9, 16
Melanoma and neural system tumor syndrome, familial	CDKN2A	1186	600160	2, 4, 8, 9, 17
Melanoma, cutaneous malignant	MC1R	1274	155555	2, 4, 8, 9
Melanoma, cutaneous malignant	MITF	777	156845	2, 4, 8, 9, 17
Melanoma, cutaneous malignant, familial	CDKN2A	1186	600160	2, 4, 8, 9, 17
Melanoma, cutaneous malignant, familial type 6, susceptibility to	XRCC3	2332	600675	2, 4, 17
Melanoma, cutaneous malignant, familial type 10, susceptibility to	POT1	2331	606478	2, 4, 17
Melanoma, cutaneous malignant, familial, CDK4 related	CDK4	1187	123829	2, 4, 8, 9
Meningioma, familial, PDGFB related	PDGFB	1801	190040	2, 3, 4, 8, 9
Meningioma, familial, susceptibility to	SMARCE1	1510	603111	2, 3, 4, 8, 9
Meningioma, familial, susceptibility to	SUFU	2360	607035	2, 3, 4, 8, 9, 17
Meningioma, MN1 deficiency related	MN1	2359	156100	2, 4
Mismatch repair cancer syndrome	MLH1	1171	120436	2, 4, 8, 9, 17
Mismatch repair cancer syndrome	MSH2	1170	609309	2, 4, 8, 9, 17
Mismatch repair cancer syndrome	MSH6	1173	600678	2, 4, 8, 9, 17
Mismatch repair cancer syndrome	PMS2	1172	600259	2, 4, 8, 9
MMR genes methylation analysis	MMR genes	3088		8
MPL, selective sequencing of exon 10	MPL	45021	159530	1
Muir-Torre syndrome	MLH1	1171	120436	2, 4, 8, 9, 17
Muir-Torre syndrome	MSH2	1170	609309	2, 4, 8, 9, 17
Multiple endocrine neoplasia type 1, CDKN2B related	CDKN2B	1474	600431	2, 4, 8, 9
Multiple endocrine neoplasia type 2A	RET	240	164761	2, 4, 8, 9
Multiple endocrine neoplasia type 2B	RET	240	164761	2, 4, 8, 9
Multiple endocrine neoplasia type 4	CDKN1B	2608	600778	2, 4, 8, 9
Myelodysplastic syndrome, somatic	TET2	30055	612839	11
Myelofibrosis, somatic	JAK2	30031	147796	11
Myxoma, intracardiac	PRKAR1A	1192	188830	2, 3, 4, 8, 9, 17
Neurofibromatosis type 1 -like syndrome	SPRED1	281	609291	2, 4, 8, 9, 17
Neurofibromatosis type 1	NF1	182	613113	2, 8, 17
Neurofibromatosis type 2	NF2	183	607379	2, 4, 8, 9, 17
Nonpolyposis hereditary colon cancer, PMS1 related	PMS1	57	600258	2, 3, 4, 8, 9, 17
Nonsmall cell lung cancer, familial, susceptibility to	EGFR	1183	131550	2, 4, 8, 9, 17
NPM1, selective sequencing of exon 11	NPM1	45022	164040	1
NRAS somatic Hotspot: c.34G>T, p.G12C	NRAS	46012	164790	1
NRAS somatic Hotspot: c.35G>A, p.G12D	NRAS	46009	164790	1
NRAS somatic Hotspot: c.181C>A p.Q61K	NRAS	46010	164790	1
NRAS somatic Hotspot: c.182A>G, p.Q61R	NRAS	46011	164790	1
NRAS, selective sequencing of exons 2 and 3	NRAS	45025	164790	1
Pancreatic cancer type 2, susceptibility to	BRCA2	379	600185	2, 4, 8, 9, 16
Pancreatic cancer type 3, susceptibility to	PALB2	388	610355	2, 4, 8, 9, 17
Pancreatic cancer, susceptibility to, type 4	BRCA1	1165	113705	2, 4, 8, 9, 16
Pancreatic cancer/melanoma syndrome, familial	CDKN2A	1186	600160	2, 4, 8, 9, 17
Paraganglioma and gastric stromal sarcoma	SDHD	1190	602690	2, 4, 8, 9
Paraganglioma and gastric stromal sarcoma	SDHD	1190	602690	2, 4, 8, 9
Paragangliomas type 1, with or without deafness	SDHD	1190	602690	2, 4, 8, 9

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11: Somatic Mutation Analysis
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Disease	Gene	Test code	OMIM Gene	Available test methods
Paragangliomas type 4	SDHB	261	185470	2, 4, 8, 9, 17
Paragangliomas type 5	SDHA	1188	600857	2, 3, 4, 8, 9, 17
PDGFRA, selective sequencing of exons 12, 14 and 18	PDGFRA	45028	173490	1
PDGFRA, selective sequencing of exons 12, 14 and 18	PDGFRA	45028	173490	1
Peutz-Jeghers syndrome	STK11	1195	602216	2, 4, 8, 9, 17
Pheochromocytoma type 1	SDHD	1190	602690	2, 4, 8, 9
Pheochromocytoma type 2	SDHB	261	185470	2, 4, 8, 9, 17
Pheochromocytoma type 3	SDHC	1189	602413	2, 4, 8, 9
Pheochromocytoma type 5	SDHAF2	1191	613019	2, 4, 8, 9
Pheochromocytoma type 8	TMEM127	1193	613403	2, 4
Pheochromocytoma type 9	MAX	1194	154950	2, 4, 8, 9, 17
Pituitary adenoma, ACTH-secreting, due to AIP germline mutation	AIP	1557	605555	2, 4, 8, 9
Pituitary adenoma, growth hormone-secreting, due to AIP germline mutation	AIP	1557	605555	2, 4, 8, 9
Pituitary adenoma, prolactin-secreting, due to AIP germline mutation	AIP	1557	605555	2, 4, 8, 9
Pleuropulmonary blastoma	DICER1	99	606241	2, 3, 4, 8, 9, 17
Polycythemia vera, somatic	JAK2	30031	147796	11
Polyposis syndrome, hereditary mixed	GREM1	727	603054	2, 4, 8, 9
Polyposis syndrome, hereditary mixed type 2	BMPR1A	1179	601299	2, 4, 8, 9, 17
Prostate cancer	BRCA2	379	600185	2, 4, 8, 9, 16
Prostate cancer	STAG1	1910	604358	2, 17
Prostate cancer	ZNF783	2030		2, 4
Prostate cancer, familial, association with	HOXB13	2736	604607	2, 4
Prostate cancer, hereditary type 1	RNASEL	1198	180435	2, 4, 17
Prostate cancer, hereditary type 2, susceptibility to	ELAC2	1197	605367	2, 4, 17
Renal cell carcinoma, due to HNF1A germline mutation	HNF1A	450	142410	2, 4, 8, 9, 17
Renal cell carcinoma, papillary type 1, familial	MET	1201	164860	2, 4, 8, 9, 17
RET, selective sequencing of exons 5, 8, 10, 11 and 13-16	RET	2773	164761	1
Spiegler-Brooke syndrome	CYLD	2541	605018	2, 3, 4, 8, 9, 17
Thrombocythemia type 3, somatic	JAK2	30031	147796	11
Thyroid cancer type 2, nonmedullary, susceptibility to	SRGAP1	2920	606523	2, 4
Trichoepithelioma, multiple familial, type 1	CYLD	2541	605018	2, 3, 4, 8, 9, 17
Tumor predisposition syndrome	BAP1	2540	603089	2, 4, 8, 9, 17
Tumor predisposition syndrome, ARL11 related	ARL11	2732	609351	2, 4
Tylosis with esophageal cancer	RHBDF2	1049	614404	2, 3, 4, 8, 9, 17
von Hippel-Lindau syndrome	VHL	322	608537	2, 4, 8, 9
Wilms tumor, familial, due to BRCA2 mutation	BRCA2	379	600185	2, 4, 8, 9, 16

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