CENTOGENE
GUIDING PRECISION MEDICINE

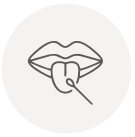
Gene List

Pediatric Panels

CentoCard


EDTA Blood


Ready to use DNA


Buccal Swab


Saliva

Methods

NGS including CNV analysis

TAT

25 business days
15 business days (Oncology)

CentoPediatric Neuro
Comprehensive
2,398 genes

- **Ataxia and cerebellar anomalies** | 193 genes
- **Dystonia, chorea and related movement disorders** | 205 genes
- **Epilepsy** | 1,059 genes
- **Leukodystrophy/ leukoencephalopathy** | 163 genes
- **NDD and ASD** | 2,016 genes
- **Neuromuscular** | 347 genes
- **Spastic paraplegia** | 136 genes
- **Structural CNS abnormalities** | 18 genes

CentoPediatric Dysmorph
Comprehensive
858 genes

- **Arthrogryposis** | 186 genes
- **Ciliopathies** | 131 genes
- **Connective tissue disorders** | 87 genes
- **Lipodystrophies** | 34 genes
- **Overgrowth** | 41 genes
- **RASopathy** | 29 genes
- **VACTERL and VACTERL-like** | 30 genes

CentoPediatric Hem/Imm
Comprehensive
578 genes

- **Blood coagulation** | 107 genes
- **Bone marrow failure** | 156 genes
- **Congenital neutropenia** | 41 genes
- **Cytopenias and congenital anemias** | 145 genes
- **Fanconi anemia** | 27 genes
- **Hemolytic anemia** | 48 genes
- **Immunology** | 472 genes
- **Primary immunodeficiency and ciliary dyskinesia** | 366 genes

CentoPediatric Bone
Comprehensive
608 genes

- **Abnormal mineralization** | 89 genes
- **Craniosynostosis** | 102 genes
- **Non-syndromic short stature** | 16 genes
- **Osteogenesis imperfecta** | 54 genes
- **Short-rib/Asphyxiating thoracic dysplasia** | 31 genes
- **Skeletal dysplasias** | 558 genes

CentoPediatric Cardio
Comprehensive
412 genes

- **Abnormalities of cardiac rhythm** | 43 genes
- **Cardiomyopathies** | 104 genes
- **Structural heart disease** | 121 genes
- **Vascular disorders** | 207 genes

CentoPediatric Metabol
Comprehensive
797 genes

- **Diabetes** | 41 genes
- **Hyperinsulinemia, hypoglycemia** | 69 genes
- **IEM** | 766 genes

CentoPediatric GI
Comprehensive
190 genes

- **Cholestasis** | 79 genes
- **Congenital diarrhea, infantile enterocolitis** | 69 genes
- **Gastrointestinal atresias, anorectal malformations** | 33 genes

CentoPediatric Derma
Comprehensive
295 genes

- **Ectodermal dysplasia** | 77 genes
- **Epidermolysis bullosa** | 45 genes
- **Ichthyosis** | 78 genes
- **Oculocutaneous albinism/ hypopigmentation** | 36 genes
- **Pigmentary skin disorders** | 140 genes
- **Xeroderma pigmentosum** | 15 genes

CentoPediatric Eye
Comprehensive
633 genes

- **Cataracts** | 209 genes
- **Leber congenital amaurosis** | 31 genes
- **Microphthalmia, anophthalmia and anterior dysgenesis** | 119 genes
- **Retinal dystrophy** | 377 genes

CentoPediatric Ear
Comprehensive
447 genes

CentoPediatric DSD
Comprehensive
149 genes

- **CAH** | 11 genes

CentoCancerHereditary Pediatric
136 genes

CentoPediatric Mito
Comprehensive
447 genes

CentoPediatric Nephro
Comprehensive
502 genes

- **aHUS** | 10 genes
- **Kidney malformations** | 23 genes
- **Polycystic kidney** | 39 genes

***Genome Backbone** available

Eye

CentoPediatric Eye Comprehensive | 633 genes

CentoPediatric Eye Comprehensive targets genes associated with a broad spectrum of disorders within pediatric ophthalmology, including conditions such as retinal dystrophies, cataracts, Leber congenital amaurosis, microphthalmia, anophthalmia and anterior dysgenesis. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCA4, ABCB6, ABCC6, ABHD12, ACBD5, ACO2, ACVR1, ADAM9, ADAMTS10, ADAMTS17, ADAMTS18, ADAMTSL4, ADGRV1, ADIPOR1, AFG3L2, AGBL5, AGK, AGPS, AHI1, AHR, AIPL1, ALDH18A1, ALDH1A3, ALDH3A2, ALMS1, ALPK1, ALX1, AMACR, ANAPC1, AP3B1, APTX, ARHGAP35, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARPC4, ARR3, ASPH, ATAD3A, ATF6, ATOH7, AUH, B3GALNT2, B3GLCT, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BFSP1, BFSP2, BLOC1S3, BLOC1S6, BMP4, BMP7, C2CD3, CA4, CABP4, CACNA1F, CACNA2D4, CANT1, CAPN15, CAPN5, CAV1, CC2D2A, CDH2, CDH23, CDH3, CDHR1, CDK9, CDON, CENPF, CEP104, CEP120, CEP164, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CFAP410, CFAP418, CHD7, CHM, CHMP4B, CHRDL1, CIB2, CISD2, CLCC1, CLCN7, CLN3, CLN5, CLN6, CLN8, CLPB, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COG4, COL11A1, COL18A1, COL2A1, COL4A1, COL8A2, COL9A1, COL9A2, COL9A3, COQ2, COQ5, COX7B, CPAMD8, CPLANE1, CRB1, CRPPA, CRX, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CRYGS, CSPP1, CTC1, CTDP1, CTNNB1, CTSD, CWC27, CYP1B1, CYP27A1, CYP4V2, CYP51A1, DAG1, DCT, DGUOK, DHCR7, DHDDS, DHX38, DKC1, DNA2, DNAJC19, DNM1L, DNMBP, DPAGT1, DRAM2, DTNBP1, DYNC2I2, DYRK1A, EDN3, EDNRB, EIF2B2, ELOVL1, ELOVL4, ELP4, EMC1, ENPP1, EPG5, EPHA2, ERCC1, ERCC2, ERCC3, ERCC5, ERCC6, ERCC8, ESPN, ETFA, ETFB, ETFDH, EXOSC2, EYA1, EYS, FAM111A, FAM161A, FAR1, FAT1, FBXL4, FDXR, FKRP, FKTN, FLVCR1, FOSL2, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FRMD7, FTL, FYCO1, FZD4, FZD5, GALE, GALK1, GALM, GALT, GBA1, GCNT2, GDF6, GEMIN4, GFER, GJA1, GJA3, GJA8, GLI2, GLS, GNAT1, GNAT2, GNB3, GNPAT, GPR143, GPR179, GRIP1, GRK1, GRM6, GRN, GTF2H5, GUCA1A, GUCA1B, GUCY2D, HARS1, HCCS, HESX1, HEXA, HGSNAT, HK1, HMX1, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, HSPG2, HTRA2, HYCC1, IARS2, IDH3A, IFT140, IFT172, IFT27, IFT43, IFT54, IFT74, IKBKG, IMPDH1, IMPG1, IMPG2, INPP5E, INPP5K, INTS1, INVS, IQCB1, JAG1, JAM3, KATNIP, KCNJ13, KCNV2, KCTD1, KIAA0586, KIAA0753, KIAA1549, KIF11, KIF3B, KIF7, KIZ, KLHL7, KMT2D, LAMA1, LAMP2, LCA5, LCAT, LEMD2, LETM1, LIM2, LMX1B, LONP1, LRAT, LRIT3, LRMDA, LRP2, LRP5, LRRC32, LSS, LTBP2, LYST, LZTFL1, MAB21L2, MAF, MAK, MAN2B1, MAPRE2, MCOLN1, MECR, MED12, MED25, MED27, MERTK, MFN2, MFRP, MIP, MITF, MKKS, MKS1, MMACHC, MPDZ, MSMO1, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTRFR, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MTTP, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MVK, MYH9, MYO7A, MYOC, MYRF, NAA10, NACC1, NAGLU, NBAS, NDP, NEK2, NEU1, NF2, NHS, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NTF4, NUP188, NYX, OAT, OCA2, OCLN, OCRL, OFD1, OPA1, OPA3, OPN1LW, OPN1MW, OPN1SW, OSTM1, OTX2, P3H2, PANK2, PANK4, PAX2, PAX3, PAX6, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDSS1, PDSS2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGRMC1, PHF6, PHGDH, PHYH, PIGY, PIK3C2A, PISD, PITPNM3, PITX2, PITX3, PLK4, PNKP, PNPLA6, PNPT1, POC1B, POLG, POLG2, POMGNT1, POMGNT2, POMT1, POMT2, PPT1, PQBP1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PRR12, PRSS56, PSMC3, PXDN, RAB18, RAB28, RAB3GAP1, RAB3GAP2, RARB, RAX, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, RECQL4, REEP6, RERE, RGS9, RGS9BP, RHO, RIC1, RIMS1, RIMS2, RIPK4, RLBP1, RNU4ATAC, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RRM2B, RS1, RTN4IP1, SAG, SAMD7, SBF2, SC5D, SCAPER, SCLT1, SDCCAG8, SEC31A, SEMA4A, SERAC1, SETX, SGSH, SHH, SIL1, SIPA1L3, SIX3, SIX6, SLC16A12, SLC24A1, SLC24A5, SLC25A46, SLC2A1, SLC33A1, SLC37A3, SLC38A8, SLC45A2, SLC52A2, SLC66A1, SLC6A6, SLC9A6, SMCHD1, SMG8, SMG9, SMO, SMOC1, SNAI2, SNRNP200, SNX10, SOX10, SOX2, SPATA7, SRD5A3, SREBF1, SSBP1, STN1, STRA6, STX3, SUMF1, TBC1D20, TCIRG1, TCTN1, TCTN2, TCTN3, TDRD7, TEAD1, TEK, TENM3, TFAP2A, TIMM50, TIMM8A, TINF2, TK2, TKFC, TLCD3B, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TMEM70, TNFRSF11A, TNFSF11, TOGARAM1, TOPORS, TPP1, TRAPPC11, TRIM32, TRNT1, TRPM1, TRPM3, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, TWNK, TYMP, TYR, TYRP1, UNC119, UNC45B, USH1C, USH1G, USH2A, USP45, VCAN, VIM, VPS13B, VPS35L, VPS4A, VSX1, VSX2, VWA8, WDPCP, WDR19, WDR37, WFS1, WHRN, WRN, XYLT2, YAP1, ZEB2, ZFYVE26, ZIC2, ZNF408, ZNF423, ZNF513, ZNF526

CentoPediatric Eye Comprehensive Genome | 635 genes

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> This version of the panel is based on genome-sequencing, it includes the non-coding genes *MIR184* and *MIR204*; and *GBA1* conversion analysis with its pseudogene.

Genes Included

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CentoPediatric Eye – Cataracts | 209 genes

CentoPediatric Eye – Cataracts targets genes associated with cataracts. This panel is intended for pediatric patients with clinical features suggestive of cataracts and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

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ERCC5, ERCC6, ERCC8, ETFA, ETFB, ETFDH, EYA1, FAM111A, FAR1, FBXL4, FKTN, FOSL2, FOXE3, FTL, FYCO1, FZD4, GALE, GALK1, GALM, GALT, GCNT2, GEMIN4, GFER, GJA1, GJA3, GJA8, GLS, GNPAT, GTF2H5, HMX1, HSF4, HSPG2, HTRA2, HYCC1, IARS2, INPP5K, INTS1, JAM3, KCTD1, LCAT, LEMD2, LETM1, LIM2, LONP1, LSS, MAF, MAN2B1, MED25, MED27, MIP, MSMO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYH9, NACC1, NDP, NEU1, NF2, NHS, NUP188, OCLN, OCRL, OPA3, P3H2, PANK4, PAX6, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGRMC1, PHGDH, PIGY, PIK3C2A, PISD, PITX3, PNPT1, POLG, POMGNT1, POMT1, POMT2, PSMC3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RDH11, RECQL4, RIC1, SC5D, SEC31A, SIL1, SIPA1L3, SLC16A12, SLC2A1, SLC33A1, SMG8, SRD5A3, SREBF1, TBC1D20, TDRD7, TFAP2A, TKFC, TMEM70, TRAPPC11, TRPM3, UNC45B, VIM, VPS4A, VSX2, WFS1, WRN, XYLT2, ZNF526

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This version of the panel is based on genome sequencing, it includes the non-coding gene *MIR184*.

Genes Included

ABCB6, ABHD12, ADAMTS10, ADAMTS18, ADAMTSL4, AGK, AGPS, ALDH18A1, ANAPC1, ARPC4, ATAD3A, B3GLCT, BCOR, BEST1, BFSP1, BFSP2, CAV1, CDH2, CDK9, CHMP4B, CLN3, COG4, COL11A1, COL18A1, COL2A1, COL4A1, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, CYP51A1, DHCR7, DNMBP, DPAGT1, DYRK1A, EIF2B2, ELP4, EPG5, EPHA2, ERCC1, ERCC2, ERCC3, ERCC5, ERCC6, ERCC8, ETFA, ETFB, ETFDH, EYA1, FAM111A, FAR1, FBXL4, FKTN, FOSL2, FOXE3, FTL, FYCO1, FZD4, GALE, GALK1, GALM, GALT, GCNT2, GEMIN4, GFER, GJA1, GJA3, GJA8, GLS, GNPAT, GTF2H5, HMX1, HSF4, HSPG2, HTRA2, HYCC1, IARS2, INPP5K, INTS1, JAM3, KCTD1, LCAT, LEMD2, LETM1, LIM2, LONP1, LSS, MAF, MAN2B1, MED25, MED27, MIP, MIR184, MSMO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYH9, NACC1, NDP, NEU1, NF2, NHS, NUP188, OCLN, OCRL, OPA3, P3H2, PANK4, PAX6, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGRMC1, PHGDH, PIGY, PIK3C2A, PISD, PITX3, PNPT1, POLG, POMGNT1, POMT1, POMT2, PSMC3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RDH11, RECQL4, RIC1, SC5D, SEC31A, SIL1, SIPA1L3, SLC16A12, SLC2A1, SLC33A1, SMG8, SRD5A3, SREBF1, TBC1D20, TDRD7, TFAP2A, TKFC, TMEM70, TRAPPC11, TRPM3, UNC45B, VIM, VPS4A, VSX2, WFS1, WRN, XYLT2, ZNF526

CentoPediatric Eye – Leber congenital amaurosis | 31 genes

CentoPediatric Eye – Leber congenital amaurosis targets genes associated with Leber congenital amaurosis. This panel is intended for pediatric patients with clinical features suggestive of Leber congenital amaurosis and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AIPL1, ALMS1, BBS4, CABP4, CEP290, CNGA3, CRB1, CRX, CWC27, GUCY2D, IDH3A, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, MT-ND1, MT-ND4, MT-ND6, MYO7A, NMNAT1, PRPH2, RD3, RDH12, RDH5, RPE65, RPGRIP1, SPATA7, TUBB4B, TULP1

CentoPediatric Eye – Microphthalmia, anophthalmia and anterior dysgenesis | 119 genes

CentoPediatric Eye – Microphthalmia, anophthalmia and anterior dysgenesis targets genes associated with microphthalmia, anophthalmia and anterior dysgenesis. This panel is intended for pediatric patients with clinical features suggestive of microphthalmia, anophthalmia and anterior dysgenesis and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic

counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCB6, ADAMTS10, ADAMTS17, ADAMTS18, ADAMTSL4, ALDH1A3, ALX1, ARHGAP35, ASPH, ATOH7, B3GALNT2, B3GLCT, BCOR, BEST1, BMP4, BMP7, CAPN15, CDK9, CDON, CENPF, CHD7, CHRDL1, COL18A1, COL4A1, COL8A2, COX7B, CPAMD8, CRPPA, CRYAA, CRYBA4, CYP1B1, DAG1, ELP4, ERCC1, ERCC2, ERCC5, ERCC6, EYA1, FAT1, FKRP, FKTN, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FZD5, GDF6, GJA1, GLI2, GRIP1, HCCS, HESX1, HMX1, KIF11, KMT2D, LRP5, MAB21L2, MAPRE2, MFRP, MITF, MYRF, NAA10, NDP, NHS, NUP188, OCRL, OTX2, PAX2, PAX6, PDE6D, PITX2, PITX3, PLK4, POMGNT1, POMGNT2, POMT1, POMT2, PQBP1, PRR12, PRSS56, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, RECQL4, RERE, RIPK4, RPGRIP1L, SHH, SIX3, SIX6, SLC38A8, SMCHD1, SMG9, SMO, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, TMEM216, TMEM237, TOGARAM1, TUBB, TUBGCP4, VPS13B, VPS35L, VSX1, VSX2, WDR37, YAP1, ZEB2, ZIC2

CentoPediatric Eye – Retinal dystrophy | 377 genes

CentoPediatric Eye – Retinal dystrophy targets genes associated with retinal dystrophies, both syndromic and non-syndromic. This panel is intended for pediatric patients with clinical features suggestive of retinal dystrophies and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRV1, ADIPOR1, AFG3L2, AGBL5, AHI1, AHR, AIPL1, ALDH3A2, ALMS1, ALPK1, AMACR, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARR3, ATF6, ATOH7, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CFAP410, CFAP418, CHM, CIB2, CISD2, CLCC1, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, COQ2, COQ5, CPLANE1, CRB1, CRPPA, CRX, CSPP1, CTC1, CTNNB1, CTSD, CWC27, CYP4V2, DCT, DHDDS, DHX38, DRAM2, DYNC2I2, ELOVL1, ELOVL4, EMC1, ERCC6, ERCC8, ESPN, EXOSC2, EYS, FAM161A, FDXR, FLVCR1, FRMD7, FZD4, GNAT1, GNAT2, GNB3, GPR143, GPR179, GRK1, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HARS1, HCCS, HGSNAT, HK1, HMX1, IDH3A, IFT140, IFT172, IFT27, IFT43, IFT54, IFT74, IKBKG, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, JAG1, KATNIP, KCNJ13, KCNV2, KIAA0586, KIAA0753, KIAA1549, KIF11, KIF3B, KIF7, KIZ, KLHL7, LAMA1, LAMP2, LCA5, LRAT, LRIT3, LRP2, LRP5, LRRC32, LZTFL1, MAK, MCOLN1, MED12, MERTK, MFRP, MKKS, MKS1, MMACHC, MPDZ, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTRFR, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MTTP, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MVK, MYO7A, NAGLU, NBAS, NDP, NEK2, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1LW, OPN1MW, OPN1SW, OTX2, P3H2, PANK2, PAX2, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDSS1, PDSS2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PISD, PITPNM3, PLK4, PNPLA6, POC1B, POMGNT1, POMGNT2, POMT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGS9, RGS9BP, RHO, RIMS1, RIMS2, RLBP1, RNU4ATAC, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD7, SCAPER, SCLT1, SDCCAG8, SEMA4A, SGSH, SIX6, SLC24A1, SLC25A46, SLC37A3, SLC38A8, SLC45A2, SLC66A1, SLC6A6, SNRNP200, SPATA7, SRD5A3, SSBP1, STN1, STX3, SUMF1, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TINF2, TLCD3B, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOPORS, TPP1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, UNC119, USH1C, USH1G, USH2A, USP45, VCAN, VPS13B, VWA8, WDPCP, WDR19, WFS1, WHRN, ZFYVE26, ZNF408, ZNF423, ZNF513

CentoPediatric Eye – Retinal dystrophy Genome | 378 genes

CentoPediatric Eye – Retinal dystrophy Genome targets genes associated with retinal dystrophies, both syndromic and non-syndromic. This panel is intended for pediatric patients with clinical features suggestive of retinal dystrophies and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This panel version is based on genome sequencing, it includes the non-coding gene *MIR204*.

Genes Included

ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRV1, ADIPOR1, AFG3L2, AGBL5, AHI1, AHR, AIPL1, ALDH3A2, ALMS1, ALPK1, AMACR, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARR3, ATF6, ATOH7, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CFAP410, CFAP418, CHM, CIB2, CISD2, CLCC1, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, COQ2, COQ5, CPLANE1, CRB1, CRPPA, CRX, CSPP1, CTC1, CTNNB1, CTSD, CWC27, CYP4V2, DCT, DHDDS, DHX38, DRAM2, DYNC2I2, ELOVL1, ELOVL4, EMC1, ERCC6, ERCC8, ESPN, EXOSC2, EYS, FAM161A, FDXR, FLVCR1, FRMD7, FZD4, GNAT1, GNAT2, GNB3, GPR143, GPR179, GRK1, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HARS1, HCCS, HGSNAT, HK1, HMX1, IDH3A, IFT140, IFT172, IFT27, IFT43, IFT54, IFT74, IKBKG, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, JAG1, KATNIP, KCNJ13, KCNV2, KIAA0586, KIAA0753, KIAA1549, KIF11, KIF3B, KIF7, KIZ, KLHL7, LAMA1, LAMP2, LCA5, LRAT, LRIT3, LRP2, LRP5, LRRC32, LZTFL1, MAK, MCOLN1, MED12, MERTK, MFRP, MIR204, MKKS, MKS1, MMACHC, MPDZ, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTRFR, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MTTP, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MVK, MYO7A, NAGLU, NBAS, NDP, NEK2, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1LW, OPN1MW, OPN1SW, OTX2, P3H2, PANK2, PAX2, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDSS1, PDSS2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PISD, PITPNM3, PLK4, PNPLA6, POC1B, POMGNT1, POMGNT2, POMT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGS9, RGS9BP, RHO, RIMS1, RIMS2, RLBP1, RNU4ATAC, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD7, SCAPER, SCLT1, SDCCAG8, SEMA4A, SGSH, SIX6, SLC24A1, SLC25A46, SLC37A3, SLC38A8, SLC45A2, SLC66A1, SLC6A6, SNRNP200, SPATA7, SRD5A3, SSBP1, STN1, STX3, SUMF1, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TINF2, TLCD3B, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOPORS, TPP1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, UNC119, USH1C, USH1G, USH2A, USP45, VCAN, VPS13B, VWA8, WDPCP, WDR19, WFS1, WHRN, ZFYVE26, ZNF408, ZNF423, ZNF513

Dysmorphology

CentoPediatric Dysmorph Comprehensive | 858 genes

CentoPediatric Dysmorph Comprehensive targets genes associated with a broad spectrum of disorders within pediatric dysmorphology, including conditions such as overgrowth syndromes, RASopathies, connective tissue disorders, VACTERL and VACTERL-like disorders, lipodystrophies, ciliopathies, and arthrogryposis. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCC6, ABL1, ACTA1, ACTA2, ACTB, ACTG1, ADAMTS10, ADAMTS15, ADAMTS17, ADAMTS2, ADAMTS9, ADAMTSL2, ADAMTSL4, ADCY6, ADGRG6, AEBP1, AGPAT2, AGRN, AHDC1, AHI1, AKT1, AKT2, AKT3, ALDH18A1, ALG3, ALMS1, ALPL, ALX3, ALX4, AMER1, AMH, AMHR2, ANKH, ANKLE2, ANKRD11, ANKS6, ANTXR2, ARFGEF2, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARL6, ARMC9, ARSB, ARSL, ARX, ASCC1, ASPA, ASPM, ASXL1, ASXL2, ASXL3, ATAD1, ATP1A2, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, ATR, AXIN1, AXIN2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALT7, B4GAT1, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCL11A, BCL11B, BCOR, BGN, BICD2, BIN1, BLTP1, BMP1, BMP2, BMP4, BMPR1B, BRAF, BRWD3, BSCL2, BUD13, C1R, C1S, C2CD3, CACNA1E, CANT1, CASK, CAV1, CAVIN1, CBL, CBS, CBY1, CC2D2A, CCN6, CCND2, CCNQ, CDC42, CDC45, CDH1, CDK13, CDK5RAP2, CDKN1C, CDON, CDT1, CDX2, CENPF, CEP104, CEP120, CEP135, CEP152, CEP164, CEP19, CEP290, CEP41, CEP55, CEP63, CEP83, CFAP410, CFAP418, CFL2, CHAT, CHD4, CHD5, CHD7, CHD8, CHMP1A, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CHUK, CILK1, CIT, CNOT1, CNTN1, CNTNAP1, COASY, COL10A1, COL11A1, COL11A2, COL12A1, COL13A1, COL1A1, COL1A2, COL25A1, COL2A1, COL3A1, COL4A5, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COLQ, COMP, COX7B, CPAP, CPE, CPLANE1, CRB2, CREB3L1, CREBBP, CRELD1, CRLF1, CRPPA, CRTAP, CSGALNACT1, CSPP1, CTNND1, CTSK, CUL4B, CUL7, CYB5A, CYP19A1, CYP1B1, CYP26B1, DACT1, DAG1, DCDC2, DCHS1, DDR2, DDX3X, DDX59, DEAF1, DHCR24, DHCR7, DHODH, DIS3L2, DLG4, DLG5, DLL3, DLL4, DMP1, DNM2, DNMT3A, DOCK6, DOK7, DPAGT1, DPF2, DPH1, DPM2, DSE, DVL3, DYM, DYNC1H1, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, DYRK1A, EBP, ECEL1, EDN3, EDNRB, EED, EFEMP2, EFNB1, EFTUD2, EGR2, EIF2AK3, EIF2S3, ELN, ENPP1, EOGT, EPG5, EPHB4, ERBB3, ERCC1, ERCC2, ERCC5, ERCC6, ERCC8, ERF, ERGIC1, ESCO2, EVC, EVC2, EXOC3L2, EXOSC3, EXOSC9, EYA1, EZH2, FAM20C, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FAT4, FBLN5, 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LMX1B, LOX, LRP2, LRP4, LTBP1, LTBP3, LZTFL1, LZTR1, MAB21L2, MAFB, MAGEL2, MAN2B1, MAP2K1, MAP2K2, MAPK1, MAPKBP1, MASP1, MAT2A, MATN3, MBTPS2, MCPH1, MED12, MED13L, MED17, MEGF8, MEIS2, MEOX1, MESP2, MET, MFAP5, MFN2, MFSD2A, MID1, MITF, MKKS, MKS1, MLC1, MMP13, MMP9, MPDZ, MPZ, MRAS, MSMO1, MSX1, MSX2, MTM1, MTOR, MTX2, MUSK, MYBPC1, MYCN, MYH11, MYH2, MYH3, MYH8, MYL11, MYLK, MYMK, MYO18B, MYOD1, NAA10, NALCN, NBAS, NCAPD3, NDE1, NDP, NEB, NECTIN1, NEK1, NEK8, NEK9, NF1, NF2, NFIA, NFIB, NFIX, NHEJ1, NIPBL, NKX3-2, NODAL, NOG, NOTCH1, NOTCH2, NPHP1, NPHP3, NPHP4, NPR2, NR2F2, NRAS, NRG1, NSD1, NSDHL, NSUN2, NUP88, OBSL1, OCRL, OFD1, OGT, OPHN1, ORAI1, ORC1, ORC4, ORC6, OTULIN, OTX2, P3H1, P4HB, PAPSS2, PAX3, PAX6, PAX7, PCNT, PCYT1A, PDE4D, PDE6D, PDGFRB, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGM1, PHEX, PHF6, PHF8, PHYH, PIBF1, PIEZO2, PIGA, PIGV, PIK3C2A, PIK3CA, PIK3R1, PIK3R2, PIP5K1C, PITX2, PJA1, PKD1, PKD2, PKHD1, 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VPS53, VRK1, VSX2, WASHC5, WBP4, WDPCP, WDR19, WDR35, WDR62, WNT1, WNT5A, WNT7A, XPNPEP3, XYLT1, YWHAE, ZBTB20, ZBTB24, ZBTB42, ZBTB7A, ZC4H2, ZDHHC9, ZEB2, ZFPM2, ZIC1, ZIC2, ZIC3, ZMPSTE24, ZNF335, ZNF423, ZNF462, ZNF469, ZNF699, ZSWIM6

CentoPediatric Dysmorph Comprehensive Genome | 860 genes

CentoPediatric Dysmorph Comprehensive Genome targets genes associated with a broad spectrum of disorders within pediatric dysmorphology, including conditions such as overgrowth syndromes, RASopathies, connective tissue disorders, VACTERL and VACTERL-like disorders, lipodystrophies, ciliopathies, and arthrogryposis. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test. This version of the panel is based on genome sequencing, it includes the non-coding genes *CHASERR* and *RNU4-2*; *SMN1* CNV analysis; and *GBA1* conversion analysis with its pseudogene.

Genes Included

ABCC6, ABL1, ACTA1, ACTA2, ACTB, ACTG1, ADAMTS10, ADAMTS15, ADAMTS17, ADAMTS2, ADAMTS9, ADAMTSL2, ADAMTSL4, ADCY6, ADGRG6, AEBP1, AGPAT2, AGRN, AHDC1, AHI1, AKT1, AKT2, AKT3, ALDH18A1, ALG3, ALMS1, ALPL, ALX3, ALX4, AMER1, AMH, AMHR2, ANKH, ANKLE2, ANKRD11, ANKS6, ANTXR2, ARFGEF2, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARL6, ARMC9, ARSB, ARSL, ARX, ASCC1, ASPA, ASPM, ASXL1, ASXL2, ASXL3, ATAD1, ATP1A2, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, ATR, AXIN1, AXIN2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALT7, B4GAT1, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCL11A, BCL11B, BCOR, BGN, BICD2, BIN1, BLTP1, BMP1, BMP2, BMP4, BMPR1B, BRAF, BRWD3, BSCL2, BUD13, C1R, C1S, C2CD3, CACNA1E, CANT1, CASK, CAV1, CAVIN1, CBL, CBS, CBY1, CC2D2A, CCN6, CCND2, CCNQ, CDC42, CDC45, CDH1, CDK13, CDK5RAP2, CDKN1C, CDON, CDT1, CDX2, CENPF, CEP104, CEP120, CEP135, CEP152, CEP164, CEP19, CEP290, CEP41, CEP55, CEP63, CEP83, CFAP410, CFAP418, CFL2, CHASERR, CHAT, CHD4, CHD5, 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CentoPediatric Dysmorph – Arthrogryposis | 186 genes

CentoPediatric Dysmorph – Arthrogryposis targets genes associated with arthrogryposis. This panel is intended for pediatric patients with clinical features suggestive of arthrogryposis and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ACTA1, ADAMTS10, ADAMTS15, ADCY6, ADGRG6, AGRN, ALG3, ANTXR2, ASCC1, ASXL1, ATAD1, ATP1A2, B3GALNT2, B4GAT1, BICD2, BIN1, BLTP1, CACNA1E, CASK, CFL2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHUK, CNTN1, CNTNAP1, COASY, COL12A1, COL25A1, COL6A1, COL6A2, COL6A3, COLQ, CRLF1, CRPPA, DAG1, DHCR24, DNM2, DOK7, DPAGT1, DPM2, DYNC1H1, EBP, ECEL1, EGR2, ERBB3, ERCC5, ERCC6, ERCC8, ERGIC1, EXOSC3, FAM20C, FBN2, FGFR2, FGFR3, FHL1, FILIP1, FKBP10, FKRP, FKTN, FLNA, FLNB, FLNC, FLVCR2, GBA1, GBE1, GFPT1, GLDN, GLE1, GMPPB, HSPG2, IRF6, KAT6B, KCNK3, KIDINS220, KIF21A, KLHL40, KLHL41, KLHL7, LAMA2, LARGE1, LGI3, LGI4, LMNA, LMOD3, LMX1B, MAGEL2, MED12, MET, MPZ, MTM1, MUSK, MYBPC1, MYH2, MYH3, MYH8, MYL11, MYMK, MYOD1, NALCN, NEB, NEK9, NUP88, ORAI1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PIEZO2, PIP5K1C, PLOD1, PLOD2, PMM2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PPP3CA, PRG4, RAPSN, RARS2, RIPK4, RXYLT1, RYR1, SCARF2, SCN1A, SCN4A, SCO2, SCYL2, SELENON, SKI, SLC18A3, SLC29A3, SLC35A3, SLC5A7, SLC6A9, SMAD3, SMAD4, SMN1, SMPD4, STAC3, STIM1, SVIL, SYNE1, TGFB2, TGFB3, TGFBR1, TGFBR2, TK2, TNNI2, TNNT1, TNNT3, TOR1A, TOR1AIP1, TPM2, TPM3, TRIP4, TRPV4, TSEN2, TSEN54, TTN, UBA1, VAMP1, VIPAS39, VPS33B, VRK1, ZBTB42, ZC4H2, ZMPSTE24

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> This version of the panel is based on genome sequencing, it includes *SMN1* CNV analysis; and *GBA1* conversion analysis with its pseudogene.

Genes Included

ACTA1, ADAMTS10, ADAMTS15, ADCY6, ADGRG6, AGRN, ALG3, ANTXR2, ASCC1, ASXL1, ATAD1, ATP1A2, B3GALNT2, B4GAT1, BICD2, BIN1, BLTP1, CACNA1E, CASK, CFL2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHUK, CNTN1, CNTNAP1, COASY, COL12A1, COL25A1, COL6A1, COL6A2, COL6A3, COLQ, CRLF1, CRPPA, DAG1, DHCR24, DNM2, DOK7, DPAGT1, DPM2, DYNC1H1, EBP, ECEL1, EGR2, ERBB3, ERCC5, ERCC6, ERCC8, ERGIC1, EXOSC3, FAM20C, FBN2, FGFR2, FGFR3, FHL1, FILIP1, FKBP10, FKRP, FKTN, FLNA, FLNB, FLNC, FLVCR2, GBA1, GBE1, GFPT1, GLDN, GLE1, GMPPB, HSPG2, IRF6, KAT6B, KCNK3, KIDINS220, KIF21A, KLHL40, KLHL41, KLHL7, LAMA2, LARGE1, LGI3, LGI4, LMNA, LMOD3, LMX1B, MAGEL2, MED12, MET, MPZ, MTM1, MUSK, MYBPC1, MYH2, MYH3, MYH8, MYL11, MYMK, MYOD1, NALCN, NEB, NEK9, NUP88, ORAI1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PIEZO2, PIP5K1C, PLOD1, PLOD2, PMM2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PPP3CA, PRG4, RAPSN, RARS2, RIPK4, RXYLT1, RYR1, SCARF2, SCN1A, SCN4A, SCO2, SCYL2, SELENON, SKI, SLC18A3, SLC29A3, SLC35A3, SLC5A7, SLC6A9, SMAD3, SMAD4, SMN1, SMPD4, STAC3, STIM1, SVIL, SYNE1, TGFB2, TGFB3, TGFBR1, TGFBR2, TK2, TNNI2, TNNT1, TNNT3, TOR1A, TOR1AIP1, TPM2, TPM3, TRIP4, TRPV4, TSEN2, TSEN54, TTN, UBA1, VAMP1, VIPAS39, VPS33B, VRK1, ZBTB42, ZC4H2, ZMPSTE24

CentoPediatric Dysmorph – Ciliopathies | 131 genes

CentoPediatric Dysmorph – Ciliopathies targets genes associated with ciliopathies. This panel is intended for pediatric patients with clinical features suggestive of ciliopathies and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ADAMTS9, AHI1, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C2CD3, CBY1, CC2D2A, CCNQ, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP55, CEP83, CFAP410, CFAP418, CILK1, CPE, CPLANE1, CRB2, CSPP1, DCDC2, DDX59, DHCR7, DLG5, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EVC, EVC2, EXOC3L2, FAN1, GLI2, GLI3, GLIS2, GRK2, HNF1B, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT54, IFT57, IFT74, IFT80, IFT81, INPP5E, INTU, INVS, IQCB1, IQCE, KATNIP, KIAA0586, KIAA0753, KIF14, KIF3B, KIF7, LBR, LZTFL1, MAPKBP1, MKKS, MKS1, NEK1, NEK8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PIBF1, PIK3C2A, PKD1, PKD2, PKHD1, PMM2, PNPLA6, POC1B, PRKACA, PRKACB, PSKH1, RPGRIP1L, SBDS, SCAPER, SCLT1, SDCCAG8, SUFU, TBC1D32, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOGARAM1, TRIM32, TTC21B, TTC8, USP9X, WDPCP, WDR19, WDR35, XPNPEP3, ZIC3, ZNF423

CentoPediatric Dysmorph – Connective tissue disorders | 87 genes

CentoPediatric Dysmorph – Connective tissue disorders targets genes associated with connective tissue disorders. This panel is intended for pediatric patients with clinical features suggestive of connective tissue disorders and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCC6, ABL1, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL2, ADAMTSL4, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, B3GALT6, B3GAT3, B4GALT7, BGN, C1R, C1S, CBS, CHST14, CHST3, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A5, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, CREB3L1, DLG4, DSE, EFEMP2, ELN, ENPP1, FBLN5, FBN1, FBN2, FKBP14, FLNA, FLNB, FOXE3, GORAB, ITGB4, LAMA3, LAMB3, LAMC2, LOX, LRP2, LTBP3, MAT2A, MBTPS2, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, PYCR1, RIN2, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD6, SP7, SPARC, TENT5A, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, UPF3B, VCAN, WNT1, ZDHHC9, ZNF469

CentoPediatric Dysmorph – Lipodystrophies | 34 genes

CentoPediatric Dysmorph – Lipodystrophies targets genes associated with lipodystrophies. This panel is intended for pediatric patients with clinical features suggestive of lipodystrophies and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABL1, AGPAT2, ATP6V0A2, BSCL2, BUD13, CAV1, CAVIN1, FBN1, GRM7, IGF1R, IKBKG, INSR, KCNJ6, LIPE, LMNA, LMNB2, MFN2, MTX2, OTULIN, PCYT1A, PDGFRB, PIK3R1, PLAAT3, PLIN1, PMM2, POLD1, POLR3A, PPARG, PRIM1, PSMB8, SPI1, SPRTN, TOMM7, ZMPSTE24

CentoPediatric Dysmorph – Overgrowth | 41 genes

CentoPediatric Dysmorph - Overgrowth targets genes associated with overgrowth syndromes. This panel is intended for pediatric patients with clinical features suggestive of overgrowth and supports the identification of a molecular diagnosis to inform clinical management,

prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This panel includes MS-MLPA analysis for Beckwith-Wiedemann syndrome (11p15).

Genes Included

AKT1, AKT2, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DIS3L2, DNMT3A, EED, EZH2, GFAP, GPC3, H1-4, HEPACAM, KDM1A, KPTN, MED12, MLC1, MTOR, NFIB, NFIX, NSD1, PDGFRB, PIGA, PIK3CA, PIK3R2, PTCH1, PTEN, RAB39B, RNF125, SETD2, SPRED1, SUZ12, TMEM94, UPF3B, ZBTB20, ZBTB7A

CentoPediatric Dysmorph – RASopathy | 29 genes

CentoPediatric Dysmorph – RASopathy targets genes associated with RASopathies. This panel is intended for pediatric patients with clinical features suggestive of RASopathies and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

BRAF, CBL, CDC42, EPHB4, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NF2, NRAS, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2, STAMBP, SYNGAP1

CentoPediatric Dysmorph – VACTERL and VACTERL-like | 30 genes

CentoPediatric Dysmorph – VACTERL and VACTERL-like targets genes associated with VACTERL and VACTERL-like disorders. This panel is intended for pediatric patients with clinical features suggestive of VACTERL and VACTERL-like disorders and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AXIN1, B9D1, CDX2, CHD4, CHD7, DACT1, DYNC2H1, EFTUD2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FOXF1, FREM2, GLI3, HOXA13, IFT172, MID1, MYCN, PUF60, SALL1, SALL4, SOX2, TBX3, TBX5, TRAP1, WBP4, WDR35

Hemm/Imm

CentoPediatric Hem/Imm Comprehensive | 578 genes

CentoPediatric Hem/Imm Comprehensive targets genes associated with a broad spectrum of disorders within pediatric hematology and immunology, including conditions such as primary immunodeficiency and ciliary dyskinesia, immunological disorders, blood coagulation disorders, bone marrow failure disorders, Fanconi anemia, congenital neutropenia, hemolytic anemia, and cytopenias and congenital anemias disorders. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCB7, ABCG5, ABCG8, ACD, ACP5, ACTB, ACTN1, ACVRL1, ADA, ADA2, ADAM17, ADAMTS13, ADAR, AGR2, AICDA, AIRE, AK1, AK2, ALAS2, ALDOA, ANK1, ANKRD26, ANO6, AP3B1, AP3D1, ARHGEF1, ARPC1B, ARPC5, ATM, ATP6AP1, ATR, ATRX, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S5, BLOC1S6, BPGM, BRCA1, BRCA2, BRIP1, BTK, C1QA, C1QB, C1QC, C2orf69, C3, C4A, C5, C6, C7, C8A, C8B, C9, CARD11, CARD9, CARMIL2, CASP8, CAVIN1, CBL, CCBE1, CCDC39, CCDC40, CCNO, CD19, CD247, CD27, CD36, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD55, CD59, CD70, CD79A, CD79B, CDAN1, CDC42, CDIN1, CEBPA, CEBPE, CFAP298, CFAP300, CFAP74, CFB, CFP, CFTR, CHST14, CIB1, CIITA, CLPB, COG6, COL1A1, COL3A1, COL4A1, COL5A1, COL5A2, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CXCR2, CXCR4, CYB5R3, CYBA, CYBB, CYBC1, CYCS, DAW1, DBR1, DCLRE1B, DCLRE1C, DDX41, DEF6, DGAT1, DHFR, DIAPH1, DKC1, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAJC21, DNAL1, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, DRC1, DRC2, DRC4, DSG1, DTNBP1, DUT, EFL1, ELANE, ELF4, ENG, EPB41, EPB42, EPG5, ERCC4, ERCC6L2, ETV6, EXTL3, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FCGR3A, FCHO1, FCN3, FERMT3, FGA, FGB, FGG, FLI1, FNIP1, FOXJ1, FOXN1, FOXP3, FYB1, G6PC3, G6PD, GAS2L2, GATA1, GATA2, GBA1, GCLC, GFI1, GFI1B, GGCX, GIMAP5, GINS1, GLRX5, GNE, GP1BA, GP1BB, GP6, GP9, GPI, GSR, GSS, HAX1, HBA1, HBA2, HBB, HBD, HELLS, HK1, HMOX1, HPS1, HPS3, HPS4, HPS5, HPS6, HRG, HTRA2, HYDIN, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGHM, IGKC, IGLL1, IKBKB, IKBKG, IKZF1, IKZF2, IKZF3, IKZF5, IL10, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21R, IL2RA, IL2RB, IL2RG, IL6R, IL6ST, IL7, IL7R, IRAK4, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITGA2, ITGA2B, ITGB2, ITGB3, ITK, ITPR3, IVNS1ABP, JAGN1, JAK1, JAK2, JAK3, KCNN4, KDSR, KIF23, KLF1, KLKB1, KNG1, LAMTOR2, LAT, LCK, LCP2, LIG1, LIG4, LMAN1, LPIN2, LRBA, LRRC56, LYST, MAGT1, MALT1, MAN2B1, MAP3K14, MCFD2, MCIDAS, MCM10, MCM4, MCTS1, MECOM, MPEG1, MPIG6B, MPL, MPO, MRE11, MRTFA, MS4A1, MSH6, MSN, MTHFD1, MYD88, MYH9, MYSM1, NAF1, NBEAL2, NBN, NCF1, NCF2, NCF4, NCKAP1L, NEK10, NF1, NFKB1, NFKB2, NFKBIA, NHEJ1, NME5, NOP10, NSMCE3, NT5C3A, OAS1, ODAD1, ODAD2, ODAD3, ODAD4, ORAI1, OTULIN, P2RY12, PALB2, PARN, PAX1, PAX5, PC, PDCD1, PDHX, PEPD, PFKM, PGK1, PGM3, PI4KA, PIEZO1, PIK3CD, PIK3CG, PIK3R1, PKLR, PLA2G4A, PLAT, PLAU, PLCG2, PLG, PMS2, PNP, POLD1, POLD3, POLE, PRF1, PRIM1, PRKCD, PRKDC, PROC, PROS1, PSMB10, PSTPIP1, PTCRA, PTGS1, PTPN11, PTPRC, PUS1, RAB27A, RAC2, RAD51, RAD51C, RAG1, RAG2, RASGRP1, RASGRP2, RBCK1, RBM8A, RECQL4, REL, RELB, RFWD3, RFX5, RFXANK, RFXAP, RHAG, RHOH, RIPK1, RIT1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF113A, RNF168, RNF31, RNU4ATAC, RORC, RPA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RPSA, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, RUNX1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SCNN1A, SCNN1B, SCNN1G, SEC23B, SEC61A1, SERPINC1, SERPIND1, SERPINE1, SERPINF2, SERPING1, SGPL1, SH2D1A, SH3KBP1, SKIC2, SKIC3, SLC19A2, SLC25A38, SLC2A1, SLC35A1, SLC37A4, SLC39A7, SLC45A2, SLC46A1, SLC4A1, SLFN14, SLX4, SMARCAL1, SMARCD2, SNORA31, SOCS1, SOS1, SP110, SPAG1, SPEF2, SPI1, SPINK5, SPPL2A, SPTA1, SPTB, SRC, SRP54, SRP72, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK36, STK4, STN1, STX11, STXBP2, SYK, TAFAZZIN, TAP1, TAP2, TAPBP, TBX1, TBX2, TBX21, TBXA2R, TBXAS1, TCF3, TCIRG1, TCN2, TERC, TERT, TET2, TFRC, TGFB1, THBD, THPO, TICAM1, TINF2, TLR3, TLR7, TLR8, TNF, TNFAIP3, TNFRSF13B, TNFRSF13C, TNFRSF9, TNXB, TOP2B, TOP3A, TP53, TPI1, TPM4, TPP2, TRAC, TRAF3, TREX1, TTC12, TTC7A, TUBB1, TYK2, UBE2T, UNC119, UNC13D, UNC93B1, UNG, UROS, USB1, VKORC1, VPS13B, VPS45, VWF, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, ZAP70, ZBTB24, ZCCHC8, ZMYND10, ZNF341, ZNF699, ZNFX1

CentoPediatric Hem/Imm Comprehensive Genome | 579 genes

CentoPediatric Hem/Imm Comprehensive Genome targets genes associated with a broad spectrum of disorders within pediatric hematology and immunology, including conditions such as primary immunodeficiency and ciliary dyskinesia, immunological disorders, blood coagulation disorders, bone marrow failure disorders, Fanconi anemia, congenital neutropenia, hemolytic anemia, and cytopenias and congenital anemias disorders. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This version of the panel is genome sequencing based, it includes the non-coding gene *RNU7-1*; and *GBA1* conversion analysis with its pseudogene.

Genes Included

ABCB7, ABCG5, ABCG8, ACD, ACP5, ACTB, ACTN1, ACVRL1, ADA, ADA2, ADAM17, ADAMTS13, ADAR, AGR2, AICDA, AIRE, AK1, AK2, ALAS2, ALDOA, ANK1, ANKRD26, ANO6, AP3B1, AP3D1, ARHGEF1, ARPC1B, ARPC5, ATM, ATP6AP1, ATR, ATRX, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S5, BLOC1S6, BPGM, BRCA1, BRCA2, BRIP1, BTK, C1QA, C1QB, C1QC, C2orf69, C3, C4A, C5, C6, C7, C8A, C8B, C9, CARD11, CARD9, CARMIL2, CASP8, CAVIN1, CBL, CCBE1, CCDC39, CCDC40, CCNO, CD19, CD247, CD27, CD36, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD55, CD59, CD70, CD79A, CD79B, CDAN1, CDC42, CDIN1, CEBPA, CEBPE, CFAP298, CFAP300, CFAP74, CFB, CFP, CFTR, CHST14, CIB1, CIITA, CLPB, COG6, COL1A1, COL3A1, COL4A1, COL5A1, COL5A2, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CXCR2, CXCR4, CYB5R3, CYBA, CYBB, CYBC1, CYCS, DAW1, DBR1, DCLRE1B, DCLRE1C, DDX41, DEF6, DGAT1, DHFR, DIAPH1, DKC1, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAJC21, DNAL1, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, DRC1, DRC2, DRC4, DSG1, DTNBP1, DUT, EFL1, ELANE, ELF4, ENG, EPB41, EPB42, EPG5, ERCC4, ERCC6L2, ETV6, EXTL3, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FCGR3A, FCHO1, FCN3, FERMT3, FGA, FGB, FGG, FLI1, FNIP1, FOXJ1, FOXN1, FOXP3, FYB1, G6PC3, G6PD, GAS2L2, GATA1, GATA2, GBA1, GCLC, GFI1, GFI1B, GGCX, GIMAP5, GINS1, GLRX5, GNE, GP1BA, GP1BB, GP6, GP9, GPI, GSR, GSS, HAX1, HBA1, HBA2, HBB, HBD, HELLS, HK1, HMOX1, HPS1, HPS3, HPS4, HPS5, HPS6, HRG, HTRA2, HYDIN, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGHM, IGKC, IGLL1, IKBKB, IKBKG, IKZF1, IKZF2, IKZF3, IKZF5, IL10, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21R, IL2RA, IL2RB, IL2RG, IL6R, IL6ST, IL7, IL7R, IRAK4, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITGA2, ITGA2B, ITGB2, ITGB3, ITK, ITPR3, IVNS1ABP, JAGN1, JAK1, JAK2, JAK3, KCNN4, KDSR, KIF23, KLF1, KLKB1, KNG1, LAMTOR2, LAT, LCK, LCP2, LIG1, LIG4, LMAN1, LPIN2, LRBA, LRRC56, LYST, MAGT1, MALT1, MAN2B1, MAP3K14, MCFD2, MCIDAS, MCM10, MCM4, MCTS1, MECOM, MPEG1, MPIG6B, MPL, MPO, MRE11, MRTFA, MS4A1, MSH6, MSN, MTHFD1, MYD88, MYH9, MYSM1, NAF1, NBEAL2, NBN, NCF1, NCF2, NCF4, NCKAP1L, NEK10, NF1, NFKB1, NFKB2, NFKBIA, NHEJ1, NME5, NOP10, NSMCE3, NT5C3A, OAS1, ODAD1, ODAD2, ODAD3, ODAD4, ORAI1, OTULIN, P2RY12, PALB2, PARN, PAX1, PAX5, PC, PDCD1, PDHX, PEPD, PFKM, PGK1, PGM3, PI4KA, PIEZO1, PIK3CD, PIK3CG, PIK3R1, PKLR, PLA2G4A, PLAT, PLAU, PLCG2, PLG, PMS2, PNP, POLD1, POLD3, POLE, PRF1, PRIM1, PRKCD, PRKDC, PROC, PROS1, PSMB10, PSTPIP1, PTCRA, PTGS1, PTPN11, PTPRC, PUS1, RAB27A, RAC2, RAD51, RAD51C, RAG1, RAG2, RASGRP1, RASGRP2, RBCK1, RBM8A, RECQL4, REL, RELB, RFWD3, RFX5, RFXANK, RFXAP, RHAG, RHOH, RIPK1, RIT1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF113A, RNF168, RNF31, RNU4ATAC, RNU7-1, RORC, RPA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RPSA, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, RUNX1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SCNN1A, SCNN1B, SCNN1G, SEC23B, SEC61A1, SERPINC1, SERPIND1, SERPINE1, SERPINF2, SERPING1, SGPL1, SH2D1A, SH3KBP1, SKIC2, SKIC3, SLC19A2, SLC25A38, SLC2A1, SLC35A1, SLC37A4, SLC39A7, SLC45A2, SLC46A1, SLC4A1, SLFN14, SLX4, SMARCAL1, SMARCD2, SNORA31, SOCS1, SOS1, SP110, SPAG1, SPEF2, SPI1, SPINK5, SPPL2A, SPTA1, SPTB, SRC, SRP54, SRP72, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK36, STK4, STN1, STX11, STXBP2, SYK, TAFAZZIN, TAP1, TAP2, TAPBP, TBX1, TBX2, TBX21, TBXA2R, TBXAS1, TCF3, TCIRG1, TCN2, TERC, TERT, TET2, TFRC, TGFB1, THBD, THPO, TICAM1, TINF2, TLR3, TLR7, TLR8, TNF, TNFAIP3, TNFRSF13B, TNFRSF13C, TNFRSF9, TNXB, TOP2B, TOP3A, TP53, TPI1, TPM4, TPP2, TRAC, TRAF3, TREX1, TTC12, TTC7A, TUBB1, TYK2, UBE2T, UNC119, UNC13D, UNC93B1, UNG, UROS, USB1, VKORC1, VPS13B, VPS45, VWF, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, ZAP70, ZBTB24, ZCCHC8, ZMYND10, ZNF341, ZNF699, ZNFX1

CentoPediatric Hem/Imm – Blood coagulation | 107 genes

CentoPediatric Hem/Imm – Blood coagulation targets genes associated with blood coagulation disorders. This panel is intended for pediatric patients with clinical features suggestive of blood coagulation disorders and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Inversion analysis of intron 1 and 22 of *F8* is included.

Genes Included

ABCG5, ABCG8, ACTB, ACTN1, ACVRL1, ADAMTS13, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S5, BLOC1S6, CD36, CDC42, CHST14, COL1A1, COL3A1, COL5A1, COL5A2, CYCS, DIAPH1, DTNBP1, EFL1, ENG, ETV6, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FGA, FGB, FGG, FLI1, FYB1, GATA1, GBA1, GFI1B, GGCX, GNE, GP1BA, GP1BB, GP6, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, HRG, IKZF5, ITGA2, ITGA2B, ITGB3, KDSR, KLKB1, KNG1, LMAN1, LYST, MCFD2, MECOM, MPIG6B, MPL, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAT, PLAU, PLG, PROC, PROS1, PTGS1, PTPN11, RASGRP2, RBM8A, RNU4ATAC, RUNX1, SERPINC1, SERPIND1, SERPINE1, SERPINF2, SLC35A1, SLC45A2, SLFN14, SRC, SRP54, STIM1, STXBP2, TBXA2R, TBXAS1, THBD, THPO, TNXB, TPM4, TUBB1, VKORC1, VWF, WAS, WIPF1

CentoPediatric Hem/Imm – Bone marrow failure | 156 genes

CentoPediatric Hem/Imm – Bone marrow failure targets genes associated with bone marrow failure disorders. This panel is intended for pediatric patients with clinical features suggestive of bone marrow failure and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCB7, ACD, ACTB, AK2, ALAS2, ANKRD26, AP3B1, ATR, BLM, BLOC1S3, BLOC1S6, BRCA1, BRCA2, BRIP1, CBL, CDAN1, CDIN1, CEBPA, CEBPE, CLPB, CSF3R, CTC1, CXCR4, DDX41, DHFR, DKC1, DNAJC21, DNASE2, DTNBP1, DUT, EFL1, ELANE, ERCC4, ERCC6L2, ETV6, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, G6PC3, GATA1, GATA2, GFI1, GINS1, GLRX5, GP1BA, HAX1, HPS1, HPS3, HPS4, HPS5, HPS6, IFNGR2, IKZF1, ITGA2B, ITK, JAGN1, JAK2, KCNN4, KLF1, LAMTOR2, LIG4, LYST, MAGT1, MECOM, MPIG6B, MPL, MRTFA, MYH9, MYSM1, NAF1, NBN, NF1, NOP10, PALB2, PARN, PAX5, PGM3, PIEZO1, PRF1, PUS1, RAB27A, RAC2, RAD51, RAD51C, RBM8A, RECQL4, RFWD3, RIT1, RMRP, RPA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SEC23B, SH2D1A, SLC19A2, SLC25A38, SLC37A4, SLX4, SMARCD2, SOS1, SRP54, SRP72, STIM1, STN1, STX11, STXBP2, TBXAS1, TCIRG1, TCN2, TERC, TERT, TET2, THPO, TINF2, TP53, TUBB1, UBE2T, UNC13D, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, ZCCHC8

CentoPediatric Hem/Imm – Congenital neutropenia | 41 genes

CentoPediatric Hem/Imm – Congenital neutropenia targets genes associated with congenital neutropenia. This panel is intended for pediatric patients with clinical features suggestive of congenital neutropenia and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AK2, AP3B1, AP3D1, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, HTRA2, IFNGR2, JAGN1, LAMTOR2, LYST, MRTFA, MSN, PGM3, RAC2, SBDS, SLC37A4, SMARCD2, SRP54, SRP72, TAFAZZIN, TCN2, TFRC, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1

CentoPediatric Hem/Imm – Cytopenias and congenital anemias | 145 genes

CentoPediatric Hem/Imm – Cytopenias and congenital anemias targets genes associated with cytopenias and congenital anemias. This panel is intended for pediatric patients with clinical features suggestive of cytopenias and congenital anemias, including neutropenia, bone marrow failure, hemolytic anemia and blood coagulation disorders, and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

MLPA analysis of *HBA1* and *HBA2* is included.

Genes Included

ABCB7, ABCG5, ABCG8, ACD, ACTN1, ADA2, ADAMTS13, AK1, AK2, ALAS2, ALDOA, ANK1, ANKRD26, AP3B1, ATRX, BRCA1, BRCA2, BRIP1, CBL, CD59, CDAN1, CDIN1, CEBPA, CLPB, CSF3R, CTC1, CXCR2, CXCR4, CYCS, DDX41, DHFR, DKC1, DNAJC21, DUT, EFL1, ELANE, EPB41, EPB42, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FLI1, FYB1, G6PC3, G6PD, GATA1, GATA2, GCLC, GFI1, GLRX5, GP1BA, GP1BB, GP9, GPI, GSS, HAX1, HBA1, HBA2, HBB, HBD, HK1, IKZF1, JAGN1, KIF23, KLF1, LAT, LPIN2, MECOM, MPL, MYH9, MYSM1, NBEAL2, NBN, NOP10, NT5C3A, PALB2, PARN, PFKM, PIEZO1, PKLR, PUS1, RAC2, RAD51C, RBM8A, RHAG, RMRP, RPA1, RPL11, RPL15, RPL18, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SEC23B, SEC61A1, SLC19A2, SLC25A38, SLC2A1, SLC4A1, SLFN14, SLX4, SPTA1, SPTB, SRC, SRP54, SRP72, STIM1, STN1, TCIRG1, TCN2, TERC, TERT, THPO, TINF2, TPI1, TUBB1, UBE2T, UROS, USB1, VPS45, WAS, WIPF1, WRAP53

CentoPediatric Hem/Imm – Fanconi anemia | 27 genes

CentoPediatric Hem/Imm – Fanconi anemia targets genes associated with Fanconi anemia. This panel is intended for pediatric patients with clinical features suggestive of Fanconi anemia and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ATR, BLM, BRCA1, BRCA2, BRIP1, CXCR4, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, NBN, PALB2, RAD51, RAD51C, RFWD3, SLX4, TOP3A, UBE2T, XRCC2

CentoPediatric Hem/Imm – Hemolytic anemia | 48 genes

CentoPediatric Hem/Imm – Hemolytic anemia targets genes associated with hemolytic anemia. This panel is intended for pediatric patients with clinical features suggestive of hemolytic anemia and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

MLPA analysis of *HBA1* and *HBA2* is included.

Genes Included

ABCG5, ABCG8, ADA, AK1, ALAS2, ALDOA, ANK1, ATRX, BPGM, CD59, CDAN1, CDIN1, COL4A1, CYB5R3, EPB41, EPB42, G6PD, GATA1, GCLC, GPI, GSR, GSS, HBA1, HBA2, HBB, HBD, HK1, HMOX1, KCNN4, KIF23, KLF1, LPIN2, NT5C3A, PC, PDHX, PFKM, PGK1, PIEZO1, PKLR, RHAG, SEC23B, SLC2A1, SLC4A1, SPTA1, SPTB, THBD, TPI1, UROS

CentoPediatric Hem/Imm – Immunology | 472 genes

CentoPediatric Hem/Imm – Immunology targets genes associated with immune conditions. This panel is intended for pediatric patients with clinical features suggestive of an immune disorder, including primary immunodeficiencies, congenital neutropenia and bone marrow failure syndromes, and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCB7, ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, AGR2, AICDA, AIRE, AK2, ALAS2, ANKRD26, AP3B1, AP3D1, ARHGEF1, ARPC1B, ARPC5, ATM, ATP6AP1, ATR, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S6, BRCA1, BRCA2, BRIP1, BTK, C1QA, C1QB, C1QC, C2orf69, C3, C4A, C5, C6, C7, C8A, C8B, C9, CARD11, CARD9, CARMIL2, CASP8, CAVIN1, CBL, CCBE1, CCDC39, CCDC40, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD55, CD70, CD79A, CD79B, CDAN1, CDC42, CDIN1, CEBPA, CEBPE, CFAP298, CFAP300, CFAP74, CFB, CFP, CFTR, CIB1, CIITA, CLPB, COG6, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DAW1, DBR1, DCLRE1B, DCLRE1C, DDX41, DEF6, DGAT1, DHFR, DKC1, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAJC21, DNAL1, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, DRC1, DRC2, DRC4, DSG1, DTNBP1, DUT, EFL1, ELANE, ELF4, EPG5, ERCC4, ERCC6L2, ETV6, EXTL3, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FCGR3A, FCHO1, FCN3, FERMT3, FNIP1, FOXJ1, FOXN1, FOXP3, G6PC3, GAS2L2, GATA1, GATA2, GFI1, GIMAP5, GINS1, GLRX5, GP1BA, HAX1, HELLS, HMOX1, HPS1, HPS3, HPS4, HPS5, HPS6, HTRA2, HYDIN, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGHM, IGKC, IGLL1, IKBKB, IKBKG, IKZF1, IKZF2, IKZF3, IL10, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21R, IL2RA, IL2RB, IL2RG, IL6R, IL6ST, IL7, IL7R, IRAK4, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITGA2B, ITGB2, ITK, ITPR3, IVNS1ABP, JAGN1, JAK1, JAK2, JAK3, KCNN4, KLF1, LAMTOR2, LAT, LCK, LCP2, LIG1, LIG4, LPIN2, LRBA, LRRC56, LYST, MAGT1, MALT1, MAN2B1, MAP3K14, MCIDAS, MCM10, MCM4, MCTS1, MECOM, MPEG1, MPIG6B, MPL, MPO, MRE11, MRTFA, MS4A1, MSH6, MSN, MTHFD1, MYD88, MYH9, MYSM1, NAF1, NBN, NCF1, NCF2, NCF4, NCKAP1L, NEK10, NF1, NFKB1, NFKB2, NFKBIA, NHEJ1, NME5, NOP10, NSMCE3, OAS1, ODAD1, ODAD2, ODAD3, ODAD4, ORAI1, OTULIN, PALB2, PARN, PAX1, PAX5, PDCD1, PEPD, PGM3, PI4KA, PIEZO1, PIK3CD, PIK3CG, PIK3R1, PLCG2, PMS2, PNP, POLD1, POLD3, POLE, PRF1, PRIM1,

PRKCD, PRKDC, PSMB10, PSTPIP1, PTCRA, PTPRC, PUS1, RAB27A, RAC2, RAD51, RAD51C, RAG1, RAG2, RASGRP1, RBCK1, RBM8A, RECQL4, REL, RELB, RFWD3, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RIT1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF113A, RNF168, RNF31, RNU4ATAC, RORC, RPA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RPSA, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, RUNX1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SCNN1A, SCNN1B, SCNN1G, SEC23B, SEC61A1, SERPING1, SGPL1, SH2D1A, SH3KBP1, SKIC2, SKIC3, SLC19A2, SLC25A38, SLC37A4, SLC39A7, SLC46A1, SLX4, SMARCAL1, SMARCD2, SNORA31, SOCS1, SOS1, SP110, SPAG1, SPEF2, SPI1, SPINK5, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK36, STK4, STN1, STX11, STXBP2, SYK, TAFAZZIN, TAP1, TAP2, TAPBP, TBX1, TBX2, TBX21, TBXAS1, TCF3, TCIRG1, TCN2, TERC, TERT, TET2, TFRC, TGFB1, THPO, TICAM1, TINF2, TLR3, TLR7, TLR8, TNF, TNFAIP3, TNFRSF13B, TNFRSF13C, TNFRSF9, TOP2B, TOP3A, TP53, TPP2, TRAC, TRAF3, TREX1, TTC12, TTC7A, TUBB1, TYK2, UBE2T, UNC119, UNC13D, UNC93B1, UNG, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, ZAP70, ZBTB24, ZCCHC8, ZMYND10, ZNF341, ZNF699, ZNFX1

CentoPediatric Hem/Imm – Immunology Genome | 473 genes

CentoPediatric Hem/Imm – Immunology Genome targets genes associated with immune conditions. This panel is intended for pediatric patients with clinical features suggestive of an immune disorder, including primary immunodeficiencies, congenital neutropenia and bone marrow failure syndromes, and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This version of the panel is based on genome sequencing, and it includes the non-coding gene *RNU7-1*.

Genes Included

ABCB7, ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, AGR2, AICDA, AIRE, AK2, ALAS2, ANKRD26, AP3B1, AP3D1, ARHGEF1, ARPC1B, ARPC5, ATM, ATP6AP1, ATR, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S6, BRCA1, BRCA2, BRIP1, BTK, C1QA, C1QB, C1QC, C2orf69, C3, C4A, C5, C6, C7, C8A, C8B, C9, CARD11, CARD9, CARMIL2, CASP8, CAVIN1, CBL, CCBE1, CCDC39, CCDC40, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD55, CD70, CD79A, CD79B, CDAN1, CDC42, CDIN1, CEBPA, CEBPE, CFAP298, CFAP300, CFAP74, CFB, CFP, CFTR, CIB1, CIITA, CLPB, COG6, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DAW1, DBR1, DCLRE1B, DCLRE1C, DDX41, DEF6, DGAT1, DHFR, DKC1, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAJC21, DNAL1, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, DRC1, DRC2, DRC4, DSG1, DTNBP1, DUT, EFL1, ELANE, ELF4, EPG5, ERCC4, ERCC6L2, ETV6, EXTL3, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FCGR3A, FCHO1, FCN3, FERMT3, FNIP1, FOXJ1, FOXN1, FOXP3, G6PC3, GAS2L2, GATA1, GATA2, GFI1, GIMAP5, GINS1, GLRX5, GP1BA, HAX1, HELLS, HMOX1, HPS1, HPS3, HPS4, HPS5, HPS6, HTRA2, HYDIN, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGHM, IGKC, IGLL1, IKBKB, IKBKG, IKZF1, IKZF2, IKZF3, IL10, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21R, IL2RA, IL2RB, IL2RG, IL6R, IL6ST, IL7, IL7R, IRAK4, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITGA2B, ITGB2, ITK, ITPR3, IVNS1ABP, JAGN1, JAK1, JAK2, JAK3, KCNN4, KLF1, LAMTOR2, LAT, LCK, LCP2, LIG1, LIG4, LPIN2, LRBA, LRRC56, LYST, MAGT1, MALT1, MAN2B1, MAP3K14, MCIDAS, MCM10, MCM4, MCTS1, MECOM, MPEG1, MPIG6B, MPL, MPO, MRE11, MRTFA, MS4A1, MSH6, MSN, MTHFD1, MYD88, MYH9, MYSM1, NAF1, NBN, NCF1, NCF2, NCF4, NCKAP1L, NEK10, NF1, NFKB1, NFKB2, NFKBIA, NHEJ1, NME5, NOP10, NSMCE3, OAS1, ODAD1, ODAD2, ODAD3, ODAD4, ORAI1, OTULIN, PALB2, PARN, PAX1, PAX5, PDCD1, PEPD, PGM3, PI4KA, PIEZO1, PIK3CD, PIK3CG, PIK3R1, PLCG2, PMS2, PNP, POLD1, POLD3, POLE, PRF1, PRIM1, PRKCD, PRKDC, PSMB10, PSTPIP1, PTCRA, PTPRC, PUS1, RAB27A, RAC2, RAD51, RAD51C, RAG1, RAG2, RASGRP1, RBCK1, RBM8A, RECQL4, REL, RELB, RFWD3, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RIT1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF113A, RNF168, RNF31, RNU4ATAC, RNU7-1, RORC, RPA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RPSA, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, RUNX1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SCNN1A, SCNN1B, SCNN1G, SEC23B, SEC61A1, SERPING1, SGPL1, SH2D1A, SH3KBP1, SKIC2, SKIC3, SLC19A2, SLC25A38, SLC37A4, SLC39A7, SLC46A1, SLX4, SMARCAL1, SMARCD2, SNORA31, SOCS1, SOS1, SP110, SPAG1, SPEF2, SPI1, SPINK5, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK36, STK4, STN1, STX11, STXBP2, SYK, TAFAZZIN, TAP1, TAP2, TAPBP, TBX1, TBX2, TBX21, TBXAS1, TCF3, TCIRG1, TCN2, TERC, TERT, TET2, TFRC, TGFB1, THPO, TICAM1, TINF2, TLR3, TLR7, TLR8, TNF, TNFAIP3, TNFRSF13B, TNFRSF13C, TNFRSF9, TOP2B, TOP3A, TP53, TPP2, TRAC, TRAF3, TREX1, TTC12, TTC7A, TUBB1, TYK2, UBE2T, UNC119, UNC13D, UNC93B1, UNG, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, ZAP70, ZBTB24, ZCCHC8, ZMYND10, ZNF341, ZNF699, ZNFX1

CentoPediatric Hem/Imm – Primary immunodeficiency and ciliary dyskinesia | 366 genes

CentoPediatric Hem/Imm – Primary immunodeficiency and ciliary dyskinesia targets genes associated with primary immunodeficiency and ciliary dyskinesia. This panel is intended for pediatric patients with clinical features suggestive of primary immunodeficiency and/or ciliary dyskinesia and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AGR2, AICDA, AIRE, AK2, AP3B1, AP3D1, ARHGEF1, ARPC1B, ARPC5, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S6, BTK, C1QA, C1QB, C1QC, C2orf69, C3, C4A, C5, C6, C7, C8A, C8B, C9, CARD11, CARD9, CARMIL2, CASP8, CAVIN1, CCBE1, CCDC39, CCDC40, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD55, CD70, CD79A, CD79B, CDC42, CEBPE, CFAP298, CFAP300, CFAP74, CFB, CFP, CFTR, CIB1, CIITA, COG6, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CTC1, CTLA4, CTPS1, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DAW1, DBR1, DCLRE1B, DCLRE1C, DEF6, DGAT1, DKC1, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAJC21, DNAL1, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, DRC1, DRC2, DRC4, DSG1, EFL1, ELANE, ELF4, EPG5, ERCC6L2, EXTL3, FADD, FCGR3A, FCHO1, FCN3, FERMT3, FNIP1, FOXJ1, FOXN1, FOXP3, GAS2L2, GATA2, GIMAP5, GINS1, HELLS, HMOX1, HPS1, HPS4, HPS6, HYDIN, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGHM, IGKC, IGLL1, IKBKB, IKBKG, IKZF1, IKZF2, IKZF3, IL10, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21R, IL2RA, IL2RB, IL2RG, IL6R, IL6ST, IL7, IL7R, IRAK4, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITGB2, ITK, ITPR3, IVNS1ABP, JAK1, JAK3, LAMTOR2, LAT, LCK, LCP2, LIG1, LPIN2, LRBA, LRRC56, LYST, MAGT1, MALT1, MAN2B1, MAP3K14, MCIDAS, MCM10, MCM4, MCTS1, MPEG1, MPO, MRE11, MRTFA, MS4A1, MSH6, MSN, MTHFD1, MYD88, MYSM1, NBN, NCF1, NCF2, NCF4, NCKAP1L, NEK10, NFKB1, NFKB2, NFKBIA, NHEJ1, NME5, NOP10, NSMCE3, OAS1, ODAD1, ODAD2, ODAD3, ODAD4, ORAI1, OTULIN, PARN, PAX1, PDCD1, PEPD, PGM3, PI4KA, PIK3CD, PIK3CG, PIK3R1, PLCG2, PMS2, PNP, POLD1, POLD3, POLE, PRIM1, PRKCD, PRKDC, PSMB10, PSTPIP1, PTCRA, PTPRC, RAB27A, RAC2, RAG1, RAG2, RASGRP1, RBCK1, RECQL4, REL, RELB, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF113A, RNF168, RNF31, RNU4ATAC, RORC, RPSA, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, SAMD9, SAMHD1, SASH3, SBDS, SCNN1A, SCNN1B, SCNN1G, SEC61A1, SERPING1, SGPL1, SH2D1A, SH3KBP1, SKIC2, SKIC3, SLC39A7, SLC46A1, SMARCAL1, SMARCD2, SNORA31, SOCS1, SP110, SPAG1, SPEF2, SPI1, SPINK5, SPPL2A, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK36, STK4, STX11, STXBP2, SYK, TAP1, TAP2, TAPBP, TBX1, TBX2, TBX21, TCF3, TCN2, TERC, TERT, TET2, TFRC, TGFB1, TICAM1, TINF2, TLR3, TLR7, TLR8, TNF, TNFAIP3, TNFRSF13B, TNFRSF13C, TNFRSF9, TOP2B, TPP2, TRAC, TRAF3, TREX1, TTC12, TTC7A, TYK2, UNC119, UNC93B1, UNG, WDR1, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24, ZMYND10, ZNF341, ZNF699, ZNFX1

CentoPediatric Hem/Imm – Primary immunodeficiency and ciliary dyskinesia Genome | 367 genes

CentoPediatric Hem/Imm - Primary immunodeficiency and ciliary dyskinesia Genome targets genes associated with primary immunodeficiency and ciliary dyskinesia. This panel is intended for pediatric patients with clinical features suggestive of primary immunodeficiency and/or ciliary dyskinesia and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This version of the panel is based on genome-sequencing, and it includes the non-coding gene RNU7-1.

Genes Included

ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AGR2, AICDA, AIRE, AK2, AP3B1, AP3D1, ARHGEF1, ARPC1B, ARPC5, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S6, BTK, C1QA, C1QB, C1QC, C2orf69, C3, C4A, C5, C6, C7, C8A, C8B, C9, CARD11, CARD9, CARMIL2, CASP8, CAVIN1, CCBE1, CCDC39, CCDC40, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD55, CD70, CD79A, CD79B, CDC42, CEBPE, CFAP298, CFAP300, CFAP74, CFB, CFP, CFTR, CIB1, CIITA, COG6, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CTC1, CTLA4, CTPS1, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DAW1, DBR1, DCLRE1B, DCLRE1C, DEF6, DGAT1, DKC1, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAJC21, DNAL1, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, DRC1, DRC2, DRC4, DSG1, EFL1, ELANE, ELF4, EPG5, ERCC6L2, EXTL3, FADD, FCGR3A, FCHO1, FCN3, FERMT3, FNIP1, FOXJ1, FOXN1, FOXP3, GAS2L2, GATA2, GIMAP5, GINS1, HELLS, HMOX1, HPS1, HPS4, HPS6, HYDIN, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGHM, IGKC, IGLL1, IKBKB, IKBKG, IKZF1, IKZF2, IKZF3, IL10, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21R, IL2RA, IL2RB, IL2RG, IL6R, IL6ST, IL7, IL7R, IRAK4, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITGB2, ITK, ITPR3, IVNS1ABP, JAK1, JAK3, LAMTOR2, LAT, LCK, LCP2, LIG1, LPIN2, LRBA, LRRC56, LYST, MAGT1, MALT1, MAN2B1, MAP3K14, MCIDAS, MCM10, MCM4, MCTS1, MPEG1, MPO, MRE11, MRTFA, MS4A1, MSH6, MSN, MTHFD1, MYD88, MYSM1, NBN, NCF1, NCF2, NCF4, NCKAP1L, NEK10, NFKB1, NFKB2, NFKBIA, NHEJ1, NME5, NOP10, NSMCE3, OAS1, ODAD1, ODAD2, ODAD3,

ODAD4, ORAI1, OTULIN, PARN, PAX1, PDCD1, PEPD, PGM3, PI4KA, PIK3CD, PIK3CG, PIK3R1, PLCG2, PMS2, PNP, POLD1, POLD3, POLE, PRIM1, PRKCD, PRKDC, PSMB10, PSTPIP1, PTCRA, PTPRC, RAB27A, RAC2, RAG1, RAG2, RASGRP1, RBCK1, RECQL4, REL, RELB, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF113A, RNF168, RNF31, RNU4ATAC, RNU7-1, RORC, RPSA, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, SAMD9, SAMHD1, SASH3, SBDS, SCNN1A, SCNN1B, SCNN1G, SEC61A1, SERPING1, SGPL1, SH2D1A, SH3KBP1, SKIC2, SKIC3, SLC39A7, SLC46A1, SMARCAL1, SMARCD2, SNORA31, SOCS1, SP110, SPAG1, SPEF2, SPI1, SPINK5, SPPL2A, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK36, STK4, STX11, STXBP2, SYK, TAP1, TAP2, TAPBP, TBX1, TBX2, TBX21, TCF3, TCN2, TERC, TERT, TET2, TFRC, TGFB1, TICAM1, TINF2, TLR3, TLR7, TLR8, TNF, TNFAIP3, TNFRSF13B, TNFRSF13C, TNFRSF9, TOP2B, TPP2, TRAC, TRAF3, TREX1, TTC12, TTC7A, TYK2, UNC119, UNC93B1, UNG, WDR1, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24, ZMYND10, ZNF341, ZNF699, ZNFX1

Mito

CentoPediatric Mito Comprehensive | 447 genes

CentoPediatric Mito Comprehensive targets genes associated with a broad spectrum of disorders within pediatric mitochondrial disorders, including conditions associated with both nuclear and mitochondrial DNA. Targeted diseases include Leigh syndrome, Leber hereditary optic neuropathy, MERRF syndrome, mitochondrial DNA depletion syndrome and combined oxidative phosphorylation deficiency, among others. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AARS2, AASS, ABAT, ABCB6, ABCB7, ABCD1, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ADAR, AFG3L2, AGK, AGXT, AIFM1, AK2, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, AMACR, AMT, APTX, ATAD3A, ATIC, ATP5F1A, ATP5F1B, ATP5F1D, ATP5F1E, ATP5MC3, ATP5PO, AUH, BCKDHA, BCKDHB, BCKDK, BCS1L, BOLA3, BTD, C19orf12, C1QBP, C2orf69, CA5A, CARS2, CHCHD10, CISD2, CLPB, CLPP, COA6, COA7, COA8, COASY, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX15, COX16, COX20, COX4I2, COX5A, COX6A1, COX6A2, COX6B1, COX7B, COXFA4, CPOX, CPS1, CPT1A, CPT2, CRBN, CYB5A, CYB5R3, CYC1, CYCS, CYP11A1, CYP24A1, CYP27A1, CYP27B1, D2HGDH, DARS2, DBT, DGUOK, DHCR24, DHODH, DHTKD1, DLAT, DLD, DNA2, DNAJC19, DNM1L, DNM2, EARS2, ECHS1, ELAC2, ETFA, ETFB, ETFDH, ETHE1, FAH, FARS2, FASTKD2, FBXL4, FDX2, FDXR, FECH, FH, FKBP10, FLAD1, FOXRED1, FXN, GARS1, GCDH, GCSH, GDAP1, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLUD1, GLYCTK, GPI, GPT2, GRHPR, GSR, GTPBP3, HADH, HADHA, HADHB, HAMP, HARS2, HAX1, HCCS, HIBCH, HINT1, HK1, HLCS, HMBS, HMGCL, HMGCS2, HOGA1, HPDL, HSD17B10, HSD17B4, HSD3B2, HSPA9, HSPD1, HTRA2, IARS2, IBA57, IDH2, IDH3A, IDH3B, IFIH1, ISCA1, ISCA2, ISCU, IVD, KARS1, L2HGDH, LAMP2, LARS2, LETM1, LIAS, LIG3, LIPT1, LIPT2, LMBRD1, LONP1, LRPPRC, LYRM4, LYRM7, MAOA, MARS2, MCCC1, MCCC2, MCEE, MDH2, MECR, MFF, MFN2, MGME1, MICOS13, MICU1, MIPEP, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MPC1, MPV17, MRM2, MRPL3, MRPL39, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS34, MSRB3, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTFMT, MTHFD1, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTO1, MTPAP, MTRFR, MT-RNR1, MT-RNR2, MTRR, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, NADK2, NAGS, NARS2, NAXD, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA6, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB10, NDUFB11, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFU1, NGLY1, NNT, NR2F1, NSUN3, NUBPL, OAT, OGDH, OPA1, OPA3, OTC, OXA1L, OXCT1, PAM16, PANK2, PARS2, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PDX1, PET100, PEX11B, PHYH, PITRM1, PKLR, PLA2G6, PMPCA, PMPCB, PNKD, PNPLA8, PNPO, PNPT1, POLG, POLG2, POLRMT, POP1, PPA2, PPOX, PRODH, PRORP, PTCD3, PTPMT1, PTRH2, PTS, PUS1, PYCR1, PYCR2, QARS1, QDPR, QRSL1, RARS1, RARS2, RMND1, RRM2B, RTN4IP1, SACS, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SECISBP2, SERAC1, SFXN4, SLC13A3, SLC16A1, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A21, SLC25A22, SLC25A24, SLC25A26, SLC25A3, SLC25A32, SLC25A36, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC39A8, SLC52A2, SLC52A3, SPG7, SSBP1, STAT2, SUCLA2, SUCLG1, SUOX, SURF1, TACO1, TAFAZZIN, TAMM41, TANGO2, TARS2, TEFM, TFAM, TIMM50, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM65, TMEM70, TOMM70, TPK1, TRIT1, TRMT10C, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TYMP, UNG, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRQ, VARS2, WARS2, WDR45, WFS1, XPNPEP3, YARS2

Metabolic Disorders

CentoPediatric Metabol Comprehensive | 797 genes

CentoPediatric Metabol Comprehensive targets genes associated with a broad spectrum of disorders within pediatric metabolism disorders, including conditions such as IEM, inheritable diabetes, hyperinsulinemia and hypoglycemia. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AARS2, AASS, ABAT, ABCA1, ABCB11, ABCB4, ABCB7, ABCC2, ABCC8, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD5, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACY1, ADA, ADAR, ADK, ADSL, AFG3L2, AGA, AGK, AGL, AGPAT2, AGPS, AGXT, AHCY, AIFM1, AKR1D1, AKT2, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALPL, AMACR, AMN, AMT, APOA1, APOA2, APOB, APOC2, APRT, AQP2, ARG1, ARSA, ARSB, ARSK, ARSL, ASAH1, ASL, ASPA, ASS1, ATAD3A, ATIC, ATP5F1A, ATP5F1B, ATP5F1D, ATP5F1E, ATP5MC3, ATP5PO, ATP6AP1, ATP6V0A2, ATP7A, ATP7B, ATP8B1, AUH, AVP, AVPR2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, BAAT, BCAP31, BCAT2, BCKDHA, BCKDHB, BCKDK, BCS1L, BLK, BOLA3, BRAT1, BSCL2, BTD, C19orf12, C1QBP, CA5A, CACNA1C, CACNA1D, CARS2, CAT, CAV1, CAVIN1, CBLIF, CBS, CD320, CEL, CERS1, CHKB, CHST14, CHST3, CHST6, CHSY1, CISD2, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, CNNM2, CNOT1, COA3, COA6, COA7, COA8, COASY, COG1, COG3, COG4, COG5, COG6, COG7, COG8, COLGALT1, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX15, COX20, COX4I2, COX5A, COX6A1, COX6B1, COX7B, COXFA4, CPOX, CPS1, CPT1A, CPT2, CREBBP, CRPPA, CTH, CTNS, CTSA, CTSD, CTSF, CTSK, CUBN, CYC1, CYP11B1, CYP17A1, CYP19A1, CYP27A1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBH, DBT, DDC, DDOST, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHODH, DHRSX, DHTKD1, DLAT, DLD, DNA2, DNAJC12, DNAJC19, DNM1L, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, EARS2, EBP, ECHS1, EDEM3, EHHADH, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, ELAC2, ENO3, ENPP1, EP300, EPG5, EPM2A, EPRS1, ERAL1, ETFA, ETFB, ETFDH, ETHE1, FA2H, FAH, FARS2, FARSB, FASTKD2, FBP1, FBXL4, FCSK, FDX2, FDXR, FECH, FGF23, FH, FICD, FKRP, FKTN, FLAD1, FMO3, FOLR1, FOXA2, FOXP3, FOXRED1, FTCD, FUCA1, FUT8, G6PC1, G6PD, GAA, GALC, GALE, GALK1, GALM, GALNS, GALNT2, GALNT3, GALT, GAMT, GATA4, GATA6, GATM, GBA1, GBA2, GBE1, GCDH, GCH1, GCK, GCLC, GCSH, GFER, GFM1, GFM2, GFPT1, GK, GLA, GLB1, GLDC, GLIS3, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPB, GNE, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC3, GPD1, GPHN, GRHPR, GRN, GSS, GSTZ1, GTPBP3, GUSB, GYG1, GYS1, GYS2, HAAO, HADH, HADHA, HADHB, HAMP, HCFC1, HEXA, HEXB, HGSNAT, HIBCH, HJV, HK1, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HOGA1, HPD, HPRT1, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HTRA2, HYAL1, IBA57, IDH2, IDS, IDUA, IER3IP1, IL2RA, INS, INSR, ISCA1, ISCA2, ISCU, IVD, KARS1, KCNC1, KCNJ11, KCTD7, KDM6A, KHK, KMT2D, KYNU, L2HGDH, LARGE1, LARS1, LARS2, LCAT, LCT, LDHA, LDLR, LDLRAP1, LETM1, LIAS, LIPA, LIPE, LIPT1, LIPT2, LMBRD1, LMF1, LMNA, LONP1, LPIN1, LPL, LRBA, LRPPRC, LYRM4, LYRM7, MAFA, MAGEL2, MAGT1, MAN1B1, MAN2B1, MAN2B2, MANBA, MAOA, MARS2, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MDH2, MECR, MFF, MFSD8, MGAT2, MGME1, MICOS13, MIPEP, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MNX1, MOCS1, MOCS2, MOGS, MPC1, MPDU1, MPI, MPV17, MRM2, MRPL3, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS34, MRPS7, MSMO1, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTFMT, MTHFR, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTO1, MTR, MTRFR, MT-RNR1, MT-RNR2, MTRR, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MTTP, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MVK, NADK2, NAGA, NAGLU, NAGS, NARS2, NAXD, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA6, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB10, NDUFB11, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEU1, NEUROD1, NEUROG3, NFS1, NFU1, NGLY1, NHLRC1, NKX2-2, NNT, NPC1, NPC2, NSD1, NSDHL, NSUN3, NT5C3A, NUBPL, NUS1, OAT, OCLN, OCRL, ONECUT1, OPA1, OPA3, OPLAH, OTC, OXCT1, PAH, PANK2, PC, PCBD1, PCCA, PCCB, PCK1, PCSK9, PDHA1, PDHB, PDHX, PDIA6, PDP1, PDSS1, PDSS2, PDX1, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGAP2, PGAP3, PGK1, PGM1, PGM3, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PIGN, PIGO, PIGS, PIGV, PKLR, PLA2G6, PLIN1, PLPBP, PMM2, PMPCB, PNP, PNPLA2, PNPLA8, PNPO, PNPT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PPA2, PPARG, PPOX, PPT1, PRKAG2, PRKAG3, PRODH, PRPS1, PSAP, PSAT1, PSPH, PTCD3, PTF1A, PTS, PUS1, PYCR1, PYCR2, PYGL, PYGM, QARS1, QDPR, QRSL1, RARS1, RARS2, RBCK1, RFT1, RFX6, RMND1, RPIA, RRM2B, RTN4IP1, RXYLT1, SAR1B, SARS2, SC5D, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHB, SDHD, SERAC1, SFXN4, SGSH, SHMT2, SI, SLC12A3, SLC13A3, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC2A1, SLC2A2, SLC30A10, SLC31A1, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC40A1, SLC46A1, SLC4A1, SLC5A1, SLC5A6, SLC6A19, SLC6A3, SLC6A8, SLC6A9, SLC7A7, SLC7A9, SMPD1, SPR, SRD5A3, SSBP1, SSR4, ST3GAL3, ST3GAL5, STAT3, STS, STT3A, SUCLA2, SUCLG1, SUGCT, SUMF1, SUOX, SURF1, TACO1, TAFAZZIN, TALDO1, TANGO2, TARS2, TAT, TCN2, TFAM, TFR2, TH, TIMM50, TIMMDC1, TK2, TKFC, TMEM126B, TMEM165, TMEM65, TMEM70, TOP3A, TPK1, TRIM37, TRIT1, TRMT10A, TRMT10C, TRMT5, TRMU, TRPM6, TSFM, TTC19, TTPA, TUFM, TWNK, TYMP, UCP2, UGT1A1, UMPS, UPB1, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRQ, UROD, UROS, VARS2, VIPAS39, VKORC1, VMA12, VMA22, VPS11, VPS16, VPS33A, VPS33B, WARS2, WFS1, XDH, YARS2, YIPF5, ZFP57, ZNF808

CentoPediatric Metabol – Diabetes | 41 genes

CentoPediatric Metabol – Diabetes targets genes associated with inheritable diabetes. This panel is intended for pediatric patients with clinical features suggestive of inheritable diabetes and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This panel includes MS-MLPA analyses for Prader-Willi (15q11) and transient neonatal diabetes mellitus (6q24) syndromes.

Genes Included

ABCC8, BLK, BSCL2, CEL, CISD2, CNOT1, EIF2AK3, EIF2B1, EIF2S3, FICD, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, IL2RA, INS, INSR, KCNJ11, LRBA, MNX1, NEUROD1, NEUROG3, NKX2-2, ONECUT1, PDIA6, PDX1, PPARG, PTF1A, RFX6, SLC19A2, SLC2A2, STAT3, WFS1, YIPF5, ZFP57, ZNF808

CentoPediatric Metabol – Hyperinsulinemia, hypoglycemia | 69 genes

CentoPediatric Metabol – Hyperinsulinemia, hypoglycemia targets genes associated with hyperinsulinemia and hypoglycemia. This panel is intended for pediatric patients with clinical features suggestive of hyperinsulinemia and hypoglycemia and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCC8, ACAT1, ACSF3, AGL, AKT2, ALDOA, ALDOB, BTD, CACNA1C, CACNA1D, CREBBP, ENO3, EP300, FBP1, FOXA2, G6PC1, GAA, GBE1, GCK, GLUD1, GPC3, GYG1, GYS1, GYS2, HADH, HK1, HLCS, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, IVD, KCNJ11, KDM6A, KMT2D, LDHA, MAFA, MAGEL2, MCEE, MMUT, MPV17, NSD1, OXCT1, PC, PCCA, PCCB, PCK1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PMM2, PRKAG2, PRKAG3, PTF1A, PYGL, PYGM, RBCK1, SLC16A1, SLC2A2, SLC37A4, TANGO2, TRMT10A, UCP2

CentoPediatric Metabol – IEM | 766 genes

CentoPediatric Metabol – IEM targets genes associated with inborn errors of metabolism. This panel is intended for pediatric patients with clinical features suggestive of inborn errors of metabolism, including inherited disorders of intermediary metabolism, urea cycle disorders, lysosomal storage diseases and glycogen storage disease, among others, and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AARS2, AASS, ABAT, ABCA1, ABCB11, ABCB4, ABCB7, ABCC2, ABCC8, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD5, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACY1, ADA, ADAR, ADK, ADSL, AFG3L2, AGA, AGK, AGL, AGPAT2, AGPS, AGXT, AHCY, AIFM1, AKR1D1, AKT2, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALPL, AMACR, AMN, AMT, APOA1, APOA2, APOB, APOC2, APRT, AQP2, ARG1, ARSA, ARSB, ARSK, ARSL, ASAH1, ASL, ASPA, ASS1, ATAD3A, ATIC, ATP5F1A, ATP5F1B, ATP5F1D, ATP5F1E, ATP5MC3, ATP5PO, ATP6AP1, ATP6V0A2, ATP7A, ATP7B, ATP8B1, AUH, AVP, AVPR2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, BAAT, BCAP31, BCAT2, BCKDHA, BCKDHB, BCKDK, BCS1L, BOLA3, BRAT1, BSCL2, BTD, C19orf12, C1QBP, CA5A, CARS2, CAT, CAV1, CAVIN1, CBLIF, CBS, CD320, CEL, CERS1, CHKB, CHST14, CHST3, CHST6, CHSY1, CISD2, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, CNNM2, COA3, COA6, COA7, COA8, COASY, COG1, COG3, COG4, COG5, COG6, COG7, COG8, COLGALT1, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX15, COX20, COX4I2, COX5A, COX6A1, COX6B1, COX7B, COXFA4, CPOX, CPS1, CPT1A, CPT2, CRPPA, CTH, CTNS, CTSA, CTSD, CTSF, CTSK, CUBN, CYC1, CYP11B1, CYP17A1, CYP19A1, CYP27A1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBH, DBT, DDC, DDOST, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHODH, DHRSX, DHTKD1, DLAT, DLD, DNA2, DNAJC12, DNAJC19, DNM1L, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, EARS2, EBP, ECHS1, EDEM3, EHHADH, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, ELAC2, ENPP1, EPG5, EPM2A, EPRS1, ERAL1, ETFA, ETFB, ETFDH, ETHE1, FA2H, FAH, FARS2, FARSB, FASTKD2, FBP1, FBXL4, FCSK, FDX2, FDXR, FECH, FGF23, FH, FKRP, FKTN, FLAD1, FMO3, FOLR1, FOXA2, FOXRED1, FTCD, FUCA1,

FUT8, G6PC1, G6PD, GAA, GALC, GALE, GALK1, GALM, GALNS, GALNT2, GALNT3, GALT, GAMT, GATM, GBA1, GBA2, GBE1, GCDH, GCH1, GCK, GCLC, GCSH, GFER, GFM1, GFM2, GFPT1, GK, GLA, GLB1, GLDC, GLIS3, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPB, GNE, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPD1, GPHN, GRHPR, GRN, GSS, GSTZ1, GTPBP3, GUSB, GYG1, GYS1, GYS2, HAAO, HADH, HADHA, HADHB, HAMP, HCFC1, HEXA, HEXB, HGSNAT, HIBCH, HJV, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HOGA1, HPD, HPRT1, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HTRA2, HYAL1, IBA57, IDH2, IDS, IDUA, INS, INSR, ISCA1, ISCA2, ISCU, IVD, KARS1, KCNC1, KCNJ11, KCTD7, KHK, KYNU, L2HGDH, LARGE1, LARS1, LARS2, LCAT, LCT, LDHA, LDLR, LDLRAP1, LETM1, LIAS, LIPA, LIPE, LIPT1, LIPT2, LMBRD1, LMF1, LMNA, LONP1, LPIN1, LPL, LRPPRC, LYRM4, LYRM7, MAGT1, MAN1B1, MAN2B1, MAN2B2, MANBA, MAOA, MARS2, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MDH2, MECR, MFF, MFSD8, MGAT2, MGME1, MICOS13, MIPEP, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPC1, MPDU1, MPI, MPV17, MRM2, MRPL3, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS34, MRPS7, MSMO1, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTFMT, MTHFR, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTO1, MTR, MTRFR, MT-RNR1, MT-RNR2, MTRR, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MTTP, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MVK, NADK2, NAGA, NAGLU, NAGS, NARS2, NAXD, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA6, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB10, NDUFB11, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEU1, NEUROD1, NEUROG3, NFS1, NFU1, NGLY1, NHLRC1, NKX2-2, NNT, NPC1, NPC2, NSDHL, NSUN3, NT5C3A, NUBPL, NUS1, OAT, OCLN, OCRL, OPA1, OPA3, OPLAH, OTC, OXCT1, PAH, PANK2, PC, PCBD1, 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, PGAP2, PGAP3, PGK1, PGM1, PGM3, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PIGN, PIGO, PIGS, PIGV, PKLR, PLA2G6, PLIN1, PLPBP, PMM2, PMPCB, PNP, PNPLA2, PNPLA8, PNPO, PNPT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PPA2, PPOX, PPT1, PRKAG2, PRODH, PRPS1, PSAP, PSAT1, PSPH, PTCD3, PTF1A, PTS, PUS1, PYCR1, PYCR2, PYGL, QARS1, QDPR, QRSL1, RARS1, RARS2, RBCK1, RFT1, RFX6, RMND1, RPIA, RRM2B, RTN4IP1, RXYLT1, SAR1B, SARS2, SC5D, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHB, SDHD, SERAC1, SFXN4, SGSH, SHMT2, SI, SLC12A3, SLC13A3, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC2A1, SLC2A2, SLC30A10, SLC31A1, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC40A1, SLC46A1, SLC4A1, SLC5A1, SLC5A6, SLC6A19, SLC6A3, SLC6A8, SLC6A9, SLC7A7, SLC7A9, SMPD1, SPR, SRD5A3, SSBP1, SSR4, ST3GAL3, ST3GAL5, STS, STT3A, SUCLA2, SUCLG1, SUGCT, SUMF1, SUOX, SURF1, TACO1, TAFAZZIN, TALDO1, TANGO2, TARS2, TAT, TCN2, TFAM, TFR2, TH, TIMM50, TIMMDC1, TK2, TKFC, TMEM126B, TMEM165, TMEM65, TMEM70, TOP3A, TPK1, TRIM37, TRIT1, TRMT10A, TRMT10C, TRMT5, TRMU, TRPM6, TSFM, TTC19, TTPA, TUFM, TWNK, TYMP, UGT1A1, UMPS, UPB1, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRQ, UROD, UROS, VARS2, VIPAS39, VKORC1, VMA12, VMA22, VPS11, VPS16, VPS33A, VPS33B, WARS2, WFS1, XDH, YARS2, ZFP57

Cardiology

CentoPediatric Cardio Comprehensive | 412 genes

CentoPediatric Cardio Comprehensive targets genes associated with a broad spectrum of disorders within pediatric cardiovascular disorders, including conditions such as vascular disorders, cardiomyopathies, abnormalities of cardiac rhythm, and structural heart diseases. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCA1, ABCC6, ABCC9, ABCG5, ABCG8, ABL1, ACAD9, ACADVL, ACP5, ACTA1, ACTA2, ACTC1, ACTN2, ACVRL1, ADA2, ADAMTS10, ADAMTS13, ADAMTS19, ADAMTS2, ADGRG1, AEBP1, AGL, AGXT, ALDH1A2, ALPK3, ANKS6, ANTXR1, APOA1, APOB, ARHGAP31, ARL2BP, ASS1, ATAD3A, ATP7A, ATR, B3GALT6, B3GAT3, B4GALT1, BAG3, BCOR, BRAF, C1R, CACNA1A, CACNA1C, CACNA1D, CALM1, CALM2, CALM3, CASQ2, CAV3, CBL, CBS, CCDC39, CCDC40, CCM2, CD59, CDH2, CFAP298, CFAP300, CFAP53, CFC1, CHD4, CHD7, CIROZ, CISD2, CITED2, CNOT3, COG6, COL1A1, COL1A2, COL3A1, COL4A1, COL4A2, COL5A1, COL5A2, COLGALT1, COQ2, COQ8A, COX15, CPS1, CPT2, CREBBP, CRELD1, CRYAB, CSRP3, CTC1, CTNND1, CYP11B1, DAW1, DES, DLL4, DMD, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJC19, DNAL1, DOCK8, DOLK, DPAGT1, DPM1, DPM3, DRC4, DSC2, DSG2, DSP, EFEMP2, EHMT1, ELAC2, ELN, EMD, ENG, ENPP1, EOGT, EP300, EPAS1, EPG5, EPHB4, EPOR, ERCC6, ERCC8, ESCO2, EVC, EVC2, F10, F13A1, F13B, F2, F5, F7, F8, F9, FASTKD2, FBLN5, FBN1, FBN2, FERMT3, FGA, FGB, FGG, FHL1, FHOD3, FKRP, FKTN, FLNA, FLNC, FLT4, FLVCR2, FOXC1, FOXF1, FOXJ1, FOXP1, FOXRED1, FXN, GAA, GATA4, GATA6, GCDH, GDF1, GDF2, GFI1B, GGCX, GJA5, GLA, GLB1, GLMN, GNAQ, GNB5, GP1BA, GPC3, GUCY1A1, GUSB, GYS1, HADHA, HAMP, HAND1, HAND2, HBB, HCCS, HFE, HJV, HRAS, HSD11B2, HTRA1, IDH2, IL1RN, IL6, ISL1, IVD, JAG1, JAK2, JAM3, JPH2, JUP, KANSL1, KCNA5, KCNE1, KCNH2, KCNJ2, KCNQ1, KDM6A, KDR, KLHL24, KMT2D, KRAS, KRIT1, LAMP2, LARS2, LDB3, LDLR, LMAN1, LMBRD1, LMNA, LOX, LRPPRC, LRRC56, LTBP3, LZTR1, MAP2K1, MAP2K2, MCCC1, MED12, MED13L, MEGF8, MEIS2, MFAP5, MGAT2, MGP, MMACHC, MMP21, MMUT, MPI, MRAS, MRPL3, MTHFR, MT-ND5, MT-ND6, MT-RNR1, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TW, MT-TY, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK, NBEAL2, NEK10, NEXN, NF1, NIPBL, NKX2-5, NODAL, NOTCH1, NOTCH2, NOTCH3, NR2F2, NRAP, NRAS, OCLN, ODAD1, ODAD2, ODAD3, ODAD4, OTC, PARS2, PCCA, PCCB, PCNT, PCSK9, PDCD10, PDE3A, PDE4D, PGM1, PIK3C2A, PIK3CA, PKD1L1, PKP2, PLIN1, PLN, PLOD1, PMM2, PNP, POPDC1, PPP1R13L, PRDM16, PRKAG2, PRKAR1A, PRKG1, PROS1, PTEN, PTPN11, RAF1, RARB, RASA1, RASGRP2, RBM10, RBM20, RIT1, RNF213, ROBO4, RPL3L, RPSA, RRAS2, RYR2, SALL1, SAMHD1, SCN1B, SCN5A, SCNN1B, SCNN1G, SCO2, SDHA, SELENON, SERPINC1, SERPINE1, SETD5, SGCB, SGCG, SHOC2, SKI, SLC19A2, SLC22A5, SLC25A3, SLC25A4, SLC2A10, SLC40A1, SLC4A3, SMAD3, SMAD4, SMAD6, SMAD9, SMARCAL1, SMC3, SOS1, SOS2, SOX2, SPAG1, SPARC, SPEG, SPRED1, SPRED2, STAMBP, STAT1, STAT2, STIM1, STING1, STRA6, TAB2, TAFAZZIN, TBX1, TBX20, TBX3, TBX5, TCAP, TECRL, TEK, TFAP2B, TFR2, TGFB2, TGFB3, TGFBR1, TGFBR2, THBD, THSD1, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TNXB, TPM1, TPP2, TRAF7, TRDN, TREX1, TRIM63, TSC1, TSC2, TSFM, TSPYL1, TTN, TTR, TULP3, USP18, VCL, VHL, VWF, WFS1, WNK4, WRN, YY1AP1, ZIC3

CentoPediatric Cardio Comprehensive Genome | 413 genes

CentoPediatric Cardio Comprehensive Genome targets genes associated with a broad spectrum of disorders within pediatric cardiovascular disorders, including conditions such as vascular disorders, cardiomyopathies, abnormalities of cardiac rhythm, and structural heart diseases. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This version of the panel is genome-sequencing based, it includes the non-coding gene *SNORD118*; and repeat expansion screening of *FXN*.

Genes Included

ABCA1, ABCC6, ABCC9, ABCG5, ABCG8, ABL1, ACAD9, ACADVL, ACP5, ACTA1, ACTA2, ACTC1, ACTN2, ACVRL1, ADA2, ADAMTS10, ADAMTS13, ADAMTS19, ADAMTS2, ADGRG1, AEBP1, AGL, AGXT, ALDH1A2, ALPK3, ANKS6, ANTXR1, APOA1, APOB, ARHGAP31, ARL2BP, ASS1, ATAD3A, ATP7A, ATR, B3GALT6, B3GAT3, B4GALT1, BAG3, BCOR, BRAF, C1R, CACNA1A, CACNA1C, CACNA1D, CALM1, CALM2, CALM3, CASQ2, CAV3, CBL, CBS, CCDC39, CCDC40, CCM2, CD59, CDH2, CFAP298, CFAP300, CFAP53, CFC1, CHD4, CHD7, CIROZ, CISD2, CITED2, CNOT3, COG6, COL1A1, COL1A2, COL3A1, COL4A1, COL4A2, COL5A1, COL5A2, COLGALT1, COQ2, COQ8A, COX15, CPS1, CPT2, CREBBP, CRELD1, CRYAB, CSRP3, CTC1, CTNND1, CYP11B1, DAW1, DES, DLL4, DMD, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5,

DNAH9, DNAI1, DNAI2, DNAJC19, DNAL1, DOCK8, DOLK, DPAGT1, DPM1, DPM3, DRC4, DSC2, DSG2, DSP, EFEMP2, EHMT1, ELAC2, ELN, EMD, ENG, ENPP1, EOGT, EP300, EPAS1, EPG5, EPHB4, EPOR, ERCC6, ERCC8, ESCO2, EVC, EVC2, F10, F13A1, F13B, F2, F5, F7, F8, F9, FASTKD2, FBLN5, FBN1, FBN2, FERMT3, FGA, FGB, FGG, FHL1, FHOD3, FKRP, FKTN, FLNA, FLNC, FLT4, FLVCR2, FOXC1, FOXF1, FOXJ1, FOXP1, FOXRED1, FXN, GAA, GATA4, GATA6, GCDH, GDF1, GDF2, GFI1B, GGCX, GJA5, GLA, GLB1, GLMN, GNAQ, GNB5, GP1BA, GPC3, GUCY1A1, GUSB, GYS1, HADHA, HAMP, HAND1, HAND2, HBB, HCCS, HFE, HJV, HRAS, HSD11B2, HTRA1, IDH2, IL1RN, IL6, ISL1, IVD, JAG1, JAK2, JAM3, JPH2, JUP, KANSL1, KCNA5, KCNE1, KCNH2, KCNJ2, KCNQ1, KDM6A, KDR, KLHL24, KMT2D, KRAS, KRIT1, LAMP2, LARS2, LDB3, LDLR, LMAN1, LMBRD1, LMNA, LOX, LRPPRC, LRRC56, LTBP3, LZTR1, MAP2K1, MAP2K2, MCCC1, MED12, MED13L, MEGF8, MEIS2, MFAP5, MGAT2, MGP, MMACHC, MMP21, MMUT, MPI, MRAS, MRPL3, MTHFR, MT-ND5, MT-ND6, MT-RNR1, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TW, MT-TY, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK, NBEAL2, NEK10, NEXN, NF1, NIPBL, NKX2-5, NODAL, NOTCH1, NOTCH2, NOTCH3, NR2F2, NRAP, NRAS, OCLN, ODAD1, ODAD2, ODAD3, ODAD4, OTC, PARS2, PCCA, PCCB, PCNT, PCSK9, PDCD10, PDE3A, PDE4D, PGM1, PIK3C2A, PIK3CA, PKD1L1, PKP2, PLIN1, PLN, PLOD1, PMM2, PNP, POPDC1, PPP1R13L, PRDM16, PRKAG2, PRKAR1A, PRKG1, PROS1, PTEN, PTPN11, RAF1, RARB, RASA1, RASGRP2, RBM10, RBM20, RIT1, RNF213, ROBO4, RPL3L, RPSA, RRAS2, RYR2, SALL1, SAMHD1, SCN1B, SCN5A, SCNN1B, SCNN1G, SCO2, SDHA, SELENON, SERPINC1, SERPINE1, SETD5, SGCB, SGCG, SHOC2, SKI, SLC19A2, SLC22A5, SLC25A3, SLC25A4, SLC2A10, SLC40A1, SLC4A3, SMAD3, SMAD4, SMAD6, SMAD9, SMARCAL1, SMC3, SNORD118, SOS1, SOS2, SOX2, SPAG1, SPARC, SPEG, SPRED1, SPRED2, STAMBP, STAT1, STAT2, STIM1, STING1, STRA6, TAB2, TAFAZZIN, TBX1, TBX20, TBX3, TBX5, TCAP, TECRL, TEK, TFAP2B, TFR2, TGFB2, TGFB3, TGFBR1, TGFBR2, THBD, THSD1, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TNXB, TPM1, TPP2, TRAF7, TRDN, TREX1, TRIM63, TSC1, TSC2, TSFM, TSPYL1, TTN, TTR, TULP3, USP18, VCL, VHL, VWF, WFS1, WNK4, WRN, YY1AP1, ZIC3

CentoPediatric Cardio – Abnormalities of cardiac rhythm | 43 genes

CentoPediatric Cardio - Abnormalities of cardiac rhythm targets genes associated with abnormalities of cardiac rhythm. This panel is intended for pediatric patients with clinical features suggestive of abnormalities of cardiac rhythm and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ACTC1, CACNA1C, CACNA1D, CALM1, CALM2, CALM3, CASQ2, CAV3, DES, DSC2, DSG2, DSP, EMD, GAA, GLA, GNB5, GPC3, HADHA, JUP, KCNA5, KCNE1, KCNH2, KCNJ2, KCNQ1, LAMP2, LMNA, MT-ND5, MYH7, MYL4, NKX2-5, PKP2, POPDC1, PRKAG2, RYR2, SCN5A, SLC4A3, TBX3, TBX5, TECRL, TNNI3K, TRDN, TSPYL1, TTR

CentoPediatric Cardio – Cardiomyopathies | 104 genes

CentoPediatric Cardio – Cardiomyopathies targets genes associated with inheritable cardiomyopathies. This panel is intended for pediatric patients with clinical features suggestive of inheritable cardiomyopathies and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ACADVL, ACTA1, ACTC1, ACTN2, AGL, ALPK3, ATAD3A, BAG3, BRAF, CACNA1C, COX15, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, ELAC2, EPG5, FHL1, FHOD3, FKRP, FKTN, FLNC, FOXRED1, FXN, GAA, GLA, GLB1, GUSB, HADHA, HAMP, HFE, HJV, HRAS, IDH2, JPH2, JUP, KLHL24, LAMP2, LDB3, LMNA, LZTR1, MAP2K1, MAP2K2, MRPL3, MT-ND6, MT-RNR1, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TW, MT-TY, MYBPC3, MYH7, MYL2, MYL3, NEXN, NRAP, NRAS, PKP2, PLN, PPP1R13L, PRDM16, PRKAG2, PTPN11, RAF1, RBM20, RPL3L, SCN5A, SCO2, SDHA, SELENON, SGCB, SGCG, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLC40A1, SOS1, SPEG, TAFAZZIN, TCAP, TFR2, TMEM43, TMEM70, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TSFM, TTN, TTR, TULP3, VCL

CentoPediatric Cardio – Cardiomyopathies Genome | 104 genes

CentoPediatric Cardio - Cardiomyopathies Genome targets genes associated with inheritable cardiomyopathies. This panel is intended for pediatric patients with clinical features suggestive of inheritable cardiomyopathies and supports the identification of a molecular diagnosis

to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This version of the panel is genome sequencing based, it includes repeat expansion screening of *FXN*.

Genes Included

ACADVL, ACTA1, ACTC1, ACTN2, AGL, ALPK3, ATAD3A, BAG3, BRAF, CACNA1C, COX15, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, ELAC2, EPG5, FHL1, FHOD3, FKRP, FKTN, FLNC, FOXRED1, FXN, GAA, GLA, GLB1, GUSB, HADHA, HAMP, HFE, HJV, HRAS, IDH2, JPH2, JUP, KLHL24, LAMP2, LDB3, LMNA, LZTR1, MAP2K1, MAP2K2, MRPL3, MT-ND6, MT-RNR1, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TW, MT-TY, MYBPC3, MYH7, MYL2, MYL3, NEXN, NRAP, NRAS, PKP2, PLN, PPP1R13L, PRDM16, PRKAG2, PTPN11, RAF1, RBM20, RPL3L, SCN5A, SCO2, SDHA, SELENON, SGCB, SGCG, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLC40A1, SOS1, SPEG, TAFAZZIN, TCAP, TFR2, TMEM43, TMEM70, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TSFM, TTN, TTR, TULP3, VCL

CentoPediatric Cardio – Structural heart disease | 121 genes

CentoPediatric Cardio - Structural heart disease targets genes associated with structural heart diseases. This panel is intended for pediatric patients with clinical features suggestive of structural heart diseases and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABL1, ADAMTS10, ADAMTS19, ALDH1A2, ANKS6, ARHGAP31, ARL2BP, B3GAT3, BCOR, BRAF, CBL, CCDC39, CCDC40, CDH2, CFAP298, CFAP300, CFAP53, CFC1, CHD7, CIROZ, CITED2, COL1A2, COL3A1, CREBBP, CRELD1, CTNND1, DAW1, DLL4, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAL1, DRC4, EHMT1, ELN, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLT4, FOXC1, FOXJ1, FOXP1, GATA4, GATA6, GDF1, GPC3, HAND1, HAND2, HCCS, HRAS, ISL1, JAG1, KANSL1, KDM6A, KMT2D, KRAS, LRRC56, MAP2K1, MAP2K2, MED12, MED13L, MEGF8, MEIS2, MGP, MMP21, MRAS, MYH6, NEK10, NIPBL, NKX2-5, NODAL, NOTCH1, NOTCH2, NR2F2, NRAS, ODAD1, ODAD2, ODAD3, ODAD4, PKD1L1, PTPN11, RAF1, RARB, RBM10, RIT1, RPSA, SALL1, SHOC2, SKI, SLC2A10, SMAD3, SMAD6, SMC3, SOS1, SOS2, SOX2, SPAG1, SPRED1, SPRED2, STRA6, TAB2, TBX1, TBX20, TBX3, TBX5, TFAP2B, TRAF7, ZIC3

CentoPediatric Cardio – Vascular disorders | 207 genes

CentoPediatric Cardio - Vascular disorders targets genes associated with vascular disorders. This panel is intended for pediatric patients with clinical features suggestive of vascular disorders and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCA1, ABCC6, ABCC9, ABCG5, ABCG8, ACAD9, ACP5, ACTA2, ACVRL1, ADA2, ADAMTS13, ADAMTS2, ADGRG1, AEBP1, AGXT, ANTXR1, APOA1, APOB, ASS1, ATP7A, ATR, B3GALT6, B4GALT1, BRAF, C1R, CACNA1A, CBL, CBS, CCM2, CD59, CHD4, CISD2, CNOT3, COG6, COL1A1, COL3A1, COL4A1, COL4A2, COL5A1, COL5A2, COLGALT1, COQ2, COQ8A, CPS1, CTC1, CYP11B1, DOCK8, DPAGT1, DPM1, DPM3, EFEMP2, ELN, ENG, ENPP1, EPAS1, EPHB4, EPOR, ERCC6, ERCC8, ESCO2, F10, F13A1, F13B, F2, F5, F7, F8, F9, FASTKD2, FBLN5, FBN1, FERMT3, FGA, FGB, FGG, FLNA, FLT4, FLVCR2, FOXF1, GAA, GCDH, GDF2, GFI1B, GGCX, GJA5, GLA, GLMN, GNAQ, GP1BA, GUCY1A1, GYS1, HBB, HRAS, HSD11B2, HTRA1, IL1RN, IL6, IVD, JAG1, JAK2, JAM3, KANSL1, KCNA5, KCNJ2, KCNQ1, KDR, KRAS, KRIT1, LARS2, LDLR, LMAN1, LMBRD1, LMNA, LOX, LRPPRC, LTBP3, LZTR1, MAP2K1, MCCC1, MFAP5, MGAT2, MMACHC, MMUT, MPI, MTHFR, MYBPC3, MYH11, MYH7, MYLK, NBEAL2, NF1, NOTCH1, NOTCH2, NOTCH3, NRAS, OCLN, OTC, PARS2, PCCA, PCCB, PCNT, PCSK9, PDCD10, PDE3A, PDE4D, PGM1, PIK3C2A, PIK3CA, PLIN1, PLOD1, PMM2, PNP, PRKAR1A, PRKG1, PROS1, PTEN, PTPN11, RAF1, RASA1, RASGRP2, RBM20, RNF213, ROBO4, RRAS2, SAMHD1, SCN1B, SCN5A, SCNN1B, SCNN1G, SERPINC1, SERPINE1, SETD5, SHOC2, SLC19A2, SLC2A10, SMAD3, SMAD4, SMAD9, SMARCAL1, SOS1, SOS2, SPARC, SPRED2, STAMBP, STAT1, STAT2, STIM1, STING1, TEK, TGFB2, TGFB3, TGFBR1, TGFBR2, THBD, THSD1, TNXB, TPP2, TREX1, TSC1, TSC2, USP18, VHL, VWF, WFS1, WNK4, WRN, YY1AP1

CentoPediatric Cardio – Vascular disorders Genome | 208 genes

CentoPediatric Cardio - Vascular disorders Genome targets genes associated with vascular disorders. This panel is intended for pediatric patients with clinical features suggestive of vascular disorders and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This version of the panel is genome sequencing based, it includes the non-coding gene *SNORD118*.

Genes Included

ABCA1, ABCC6, ABCC9, ABCG5, ABCG8, ACAD9, ACP5, ACTA2, ACVRL1, ADA2, ADAMTS13, ADAMTS2, ADGRG1, AEBP1, AGXT, ANTXR1, APOA1, APOB, ASS1, ATP7A, ATR, B3GALT6, B4GALT1, BRAF, C1R, CACNA1A, CBL, CBS, CCM2, CD59, CHD4, CISD2, CNOT3, COG6, COL1A1, COL3A1, COL4A1, COL4A2, COL5A1, COL5A2, COLGALT1, COQ2, COQ8A, CPS1, CTC1, CYP11B1, DOCK8, DPAGT1, DPM1, DPM3, EFEMP2, ELN, ENG, ENPP1, EPAS1, EPHB4, EPOR, ERCC6, ERCC8, ESCO2, F10, F13A1, F13B, F2, F5, F7, F8, F9, FASTKD2, FBLN5, FBN1, FERMT3, FGA, FGB, FGG, FLNA, FLT4, FLVCR2, FOXF1, GAA, GCDH, GDF2, GFI1B, GGCX, GJA5, GLA, GLMN, GNAQ, GP1BA, GUCY1A1, GYS1, HBB, HRAS, HSD11B2, HTRA1, IL1RN, IL6, IVD, JAG1, JAK2, JAM3, KANSL1, KCNA5, KCNJ2, KCNQ1, KDR, KRAS, KRIT1, LARS2, LDLR, LMAN1, LMBRD1, LMNA, LOX, LRPPRC, LTBP3, LZTR1, MAP2K1, MCCC1, MFAP5, MGAT2, MMACHC, MMUT, MPI, MTHFR, MYBPC3, MYH11, MYH7, MYLK, NBEAL2, NF1, NOTCH1, NOTCH2, NOTCH3, NRAS, OCLN, OTC, PARS2, PCCA, PCCB, PCNT, PCSK9, PDCD10, PDE3A, PDE4D, PGM1, PIK3C2A, PIK3CA, PLIN1, PLOD1, PMM2, PNP, PRKAR1A, PRKG1, PROS1, PTEN, PTPN11, RAF1, RASA1, RASGRP2, RBM20, RNF213, ROBO4, RRAS2, SAMHD1, SCN1B, SCN5A, SCNN1B, SCNN1G, SERPINC1, SERPINE1, SETD5, SHOC2, SLC19A2, SLC2A10, SMAD3, SMAD4, SMAD9, SMARCAL1, SOS1, SOS2, SPARC, SPRED2, STAMBP, STAT1, STAT2, STIM1, STING1, TEK, TGFB2, TGFB3, TGFBR1, TGFBR2, THBD, THSD1, TNXB, TPP2, TREX1, TSC1, TSC2, USP18, VHL, VWF, WFS1, WNK4, WRN, YY1AP1

Neurology

CentoPediatric Neuro Comprehensive | 2,398 genes

CentoPediatric Neuro Comprehensive targets genes associated with a broad spectrum of disorders within pediatric neurology, including conditions such as neurodevelopmental delay and autism disorders, ataxia and cerebellar anomalies, spastic paraplegia, epilepsies, neuromuscular disorders, leukodystrophy/leukoencephalopathies, dystonia, chorea and related movement disorders, and structural CNS abnormalities. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AAAS, AARS1, AARS2, AASS, ABAT, ABCA2, ABCB7, ABCC8, ABCC9, ABCD1, ABCD4, ABHD12, ABHD16A, ABHD5, ACACA, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD5, ACBD6, ACER3, ACO2, ACOX1, ACOX2, ACP5, ACSF3, ACSL4, ACTA1, ACTB, ACTG1, ACTG2, ACTL6A, ACTL6B, ACTN2, ACVR1, ACY1, ADA, ADAM22, ADAMTS10, ADAMTSL2, ADAR, ADARB1, ADAT3, ADCY5, ADD1, ADD3, ADGRA2, ADGRG1, ADGRG6, ADGRL1, ADGRV1, ADH5, ADK, ADNP, ADPRS, ADSL, ADSS1, AFF2, AFF3, AFF4, AFG2A, AFG2B, AFG3L2, AGA, AGAP1, AGK, AGL, AGMO, AGO1, AGO2, AGPAT3, AGRN, AGTPBP1, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, AJAP1, AKT1, AKT3, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALMS1, ALPL, ALS2, ALX3, ALX4, AMER1, AMFR, AMMECR1, AMPD2, AMT, ANAPC1, ANAPC7, ANK2, ANK3, ANKLE2, ANKRD11, ANKRD17, ANKS1B, ANO10, ANO3, ANO4, ANO5, ANTXR1, AP1B1, AP1G1, AP1S1, AP1S2, AP2M1, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APTX, AR, ARCN1, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGAP35, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL6, ARL6IP1, ARMC9, ARPC4, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASCC1, ASCC3, ASCL1, ASH1L, ASL, ASNS, ASPA, ASPM, ASS1, ASTN1, ASXL1, ASXL2, ASXL3, ATAD1, ATAD3A, ATCAY, ATG4D, ATG7, ATIC, ATL1, ATM, ATN1, ATP11A, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A1, ATP2A2, ATP2B1, ATP2B3, ATP5F1A, ATP5F1E, ATP5MC3, ATP5PO, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP6V1B2, ATP7A, ATP7B, ATP8A2, ATP9A, ATR, ATRX, ATXN10, ATXN2L, ATXN7L3, AUH, AUTS2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BAG3, BAP1, BAZ2B, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCL11A, BCL11B, BCOR, BCORL1, BCS1L, BET1, BICD2, BICRA, BIN1, BLM, BLOC1S1, BLTP1, BMP4, BMPER, BOLA3, BORCS5, BORCS8, BPTF, BRAF, BRAT1, BRCA1, BRCA2, BRD4, BRF1, BRPF1, BRSK2, BRWD3, BSCL2, BSND, BTD, BUB1, BUB1B, C12orf57, C19orf12, C2CD3, C2orf69, CA2, 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COG3, COG4, COG5, COG6, COG7, COG8, COL12A1, COL13A1, COL18A1, COL25A1, COL2A1, COL3A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A3, COLEC10, COLEC11, COLGALT1, COLQ, COPB2, COQ2, COQ4, COQ6, COQ8A, COQ9, CORO1A, COX10, COX15, COX20, COX4I2, COX6A1, COX6A2, COX6B1, COX7B, CP, CPAP, CPE, CPLANE1, CPLX1, CPS1, CPSF3, CPT1A, CPT2, CRADD, CRB2, CRBN, CREBBP, CRELD1, CRLF1, CRPPA, CRYAB, CSDE1, CSF1R, CSNK2A1, CSNK2B, CSPP1, CSTB, CTBP1, CTC1, CTCF, CTDP1, CTNNA2, CTNNB1, CTNND2, CTNS, CTR9, CTSA, CTSD, CTSF, CTU2, CUL3, CUL4B, CUX1, CUX2, CWC27, CWF19L1, CYB5R3, CYC1, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DAGLA, DALRD3, DARS1, DARS2, DBT, DCAF17, DCC, DCHS1, DCPS, DCX, DDB1, DDC, DDHD1, DDHD2, DDOST, DDX11, DDX17, DDX23, DDX3X, DDX59, DDX6, DEAF1, DEGS1, DENND5A, DENND5B, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHRSX, DHTKD1, DHX16, DHX30, DHX37, DHX9, DIAPH1, DIP2C, DIS3L2, DKC1, DLAT, DLD, DLG3, DLG4, DLL1, DMBX1, DMD, DMPK, DMXL2, DNA2, DNAJB2, DNAJB4, DNAJB6, DNAJC12, DNAJC17, DNAJC19, DNAJC3, DNAJC5, DNAJC6, DNM1, DNM1L, DNM2, DNMT1, DNMT3A, DNMT3B, DOCK3, DOCK4, DOCK6, DOCK7, DOHH, DOK7, DOLK, DONSON, DPAGT1, DPF2, DPH1, DPH2, DPH5, DPM1, DPM2, DPM3, DPYD, DPYS, DPYSL2, DPYSL5, DRG1, DROSHA, DST, DSTYK, DTNA, DTYMK, DUOXA2, DVL3, DYM, DYNC1H1, DYNC1I2, DYRK1A, DYSF, EARS2, EBF3, EBP, ECEL1, ECHS1, ECM1, EDEM3, EED, EEF1A2, EEF1B2, EEF2, EEFSEC, EFNB1, EFTUD2, EGR2, EHMT1, EIF2A, EIF2AK2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4A2, EIF4A3, EIF5A, ELAC2, ELOVL1, ELOVL4, ELP1, ELP2, EMC1, EMC10, EMD, EMG1, EMILIN1, EML1, ENTPD1, EP300, EPG5, EPHA7, EPM2A, EPRS1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERGIC3, ERI1, ERLIN1, ERLIN2, ESAM, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOC2, EXOC7, EXOC8, EXOSC2, EXOSC3, EXOSC5, EXOSC8, EXOSC9, EXT2, EXTL3, EZH1, EZH2, F2, F5, FA2H, FAH, FAM111A, FAM111B, FAM177A1, FAM20C, FAM50A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FAR1, FARS2, FARSA, FARSB, FASTKD2, FAT4, FBN1, FBRSL1, FBXL3, FBXL4, FBXO11, FBXO28, FBXO38, FBXO7, FBXW11, FBXW7, FCSK, FDFT1, FDX2, FDXR, FEM1B, FEM1C, FERRY3, FGD1, FGD4, FGF12, FGF13, FGF14, FGFR1, FGFR2, FGFR3, FH, FHL1, FIBP, FICD, FIG4, FILIP1, FITM2, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNC, FLVCR1, FLVCR2, FMN2, FMR1, FOLR1, FOSL2, FOXG1, FOXP1, FOXP2, FOXP4, FOXR1, FOXRED1, FRA10AC1, FRAS1, FREM2, FRMD5, FRMPD4, FRRS1L, FRYL, FTCD, FTH1, FTL, FTO, FTSJ1, FUCA1, FUT8, FXN, FXR1, FXYD2, FZR1, GAA, GABBR1, GABBR2, GABRA1, GABRA2, GABRA3, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALE, GALNS, GALNT2, GALT, GAMT, GAN, GARS1, GATA6, GATAD2B, GATM, GBA1, GBA2, GBE1, GCDH, GCH1, GCSH, GDAP1, GDI1, GEMIN4,

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RPGRIP1L, RPIA, RPL10, RPS19, RPS23, RPS28, RPS6KA3, RRM2B, RSPRY1, RSRC1, RTEL1, RTN2, RTN4IP1, RTTN, RUBCN, RUSC2, RXYLT1, RYR1, RYR2, SACS, SALL1, SAMD12, SAMD9, SAMHD1, SARS1, SARS2, SASS6, SATB1, SATB2, SBDS, SBF1, SBF2, SC5D, SCAF4, SCAMP5, SCAPER, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN8A, SCN9A, SCO1, SCO2, SCYL1, SDCCAG8, SDHA, SDHAF1, SDHB, SDHD, SEC23A, SEC24D, SEC31A, SELENOI, SELENON, SEMA6B, SEPHS1, SEPSECS, SERAC1, SERPINI1, SET,

SETBP1, SETD1A, SETD1B, SETD2, SETD5, SETX, SFXN4, SGCA, SGCB, SGCD, SGCE, SGCG, SGPL1, SGSH, SGSM3, SH3TC2, SHANK1, SHANK2, SHANK3, SHH, SHMT2, SHOC2, SHQ1, SHROOM4, SIAH1, SIGMAR1, SIK1, SIL1, SIN3A, SIN3B, SIX3, SKI, SKIC3, SLC10A7, SLC12A1, SLC12A2, SLC12A3, SLC12A5, SLC12A6, SLC13A3, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC18A2, SLC18A3, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A4, SLC25A42, SLC25A46, SLC2A1, SLC2A10, SLC2A2, SLC30A10, SLC30A9, SLC31A1, SLC32A1, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35B2, SLC35C1, SLC38A3, SLC39A14, SLC39A8, SLC44A1, SLC45A1, SLC46A1, SLC4A10, SLC4A4, SLC52A2, SLC52A3, SLC5A5, SLC5A6, SLC5A7, SLC6A1, SLC6A17, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SLC7A7, SLC9A1, SLC9A6, SLC9A7, SLX4, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCA5, SMARCB1, SMARCC2, SMARCD1, SMARCD2, SMARCE1, SMC1A, SMC3, SMCHD1, SMG8, SMG9, SMN1, SMOC1, SMPD1, SMPD4, SMS, SNAP25, SNAP29, SNF8, SNIP1, SNRPB, SNUPN, SNX14, SNX27, SON, SOS1, SOS2, SOX10, SOX11, SOX2, SOX3, SOX4, SOX5, SOX6, SPART, SPAST, SPECC1L, SPEG, SPEN, SPG11, SPG7, SPOP, SPOUT1, SPR, SPRED1, SPRED2, SPTAN1, SPTBN1, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SPTSSA, SQSTM1, SRCAP, SRD5A3, SRP54, SRPK3, SRRM2, SRSF1, SSR4, ST3GAL3, ST3GAL5, STAC3, STAG1, STAG2, STAMBP, STARD7, STAT1, STEEP1, STIL, STIM1, STN1, STRA6, STRADA, STT3A, STUB1, STX1A, STX1B, STX3, STXBP1, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SUPT16H, SUPV3L1, SURF1, SUZ12, SVBP, SVIL, SYN1, SYNCRIP, SYNE1, SYNGAP1, SYNJ1, SYP, SYT1, SYT2, SZT2, TAC3, TACO1, TAF1, TAF13, TAF1C, TAF2, TAF4, TAF6, TAF8, TAFAZZIN, TAMM41, TANC2, TANGO2, TAOK1, TAOK2, TARS2, TASP1, TAT, TBC1D20, TBC1D23, TBC1D24, TBC1D2B, TBC1D7, TBCD, TBCE, TBCK, TBL1XR1, TBR1, TBX1, TBX2, TCAP, TCEAL1, TCF12, TCF20, TCF4, TCF7L2, TCN2, TCTN1, TCTN2, TCTN3, TDP2, TECPR2, TEFM, TELO2, TENM3, TERT, TET3, TFAP2A, TFE3, TFG, TG, TGFB1, TGFBR1, TGFBR2, TGIF1, TH, THAP1, THG1L, THOC2, THOC6, THRA, THUMPD1, TIAM1, TIMM50, TIMM8A, TINF2, TK2, TKFC, TKT, TLK2, TMCO1, TMEM106B, TMEM107, TMEM126A, TMEM138, TMEM147, TMEM151A, TMEM163, TMEM165, TMEM216, TMEM218, TMEM222, TMEM231, TMEM237, TMEM240, TMEM63A, TMEM63B, TMEM63C, TMEM67, TMEM70, TMEM94, TMTC3, TMX2, TNIK, TNNC2, TNNI1, TNNI2, TNNT1, TNNT3, TNPO2, TNPO3, TNR, TNRC6B, TOE1, TOGARAM1, TOMM70, TONSL, TOP2B, TOR1A, TOR1AIP1, TP53RK, TP73, TPK1, TPM2, TPM3, TPO, TPP1, TPP2, TPRKB, TRA2B, TRAF7, TRAIP, TRAK1, TRAPPC10, TRAPPC11, TRAPPC12, TRAPPC4, TRAPPC6B, TRAPPC9, TRDN, TREX1, TRIM2, TRIM32, TRIM8, TRIO, TRIP12, TRIP13, TRIP4, TRIT1, TRMT1, TRMT10A, TRMT5, TRNT1, TRPC5, TRPM3, TRPM6, TRPV4, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TSPOAP1, TTC19, TTC5, TTC8, TTI1, TTI2, TTN, TTPA, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP4, TUBGCP6, TUFM, TUSC3, TWIST1, TWIST2, TWNK, TXN2, TYMP, U2AF2, UBA1, UBA2, UBA5, UBAP1, UBAP2L, UBE2A, UBE3A, UBE3B, UBE3C, UBE4A, UBR1, UBR7, UBTF, UCHL1, UFC1, UFM1, UFSP2, UGDH, UGP2, UMPS, UNC13A, UNC45A, UNC45B, UNC80, UPB1, UPF1, UPF3B, UQCC2, UQCRC2, UQCRQ, USP18, USP27X, USP7, USP9X, VAC14, VAMP1, VAMP2, VARS1, VARS2, VCP, VIPAS39, VLDLR, VMA12, VMA21, VMA22, VPS11, VPS13B, VPS13D, VPS16, VPS33A, VPS33B, VPS35, VPS35L, VPS37A, VPS41, VPS4A, VPS50, VPS51, VPS53, VRK1, VWA1, WAC, WARS1, WARS2, WASF1, WASHC4, WASHC5, WBP11, WBP4, WDFY3, WDPCP, WDR1, WDR11, WDR26, WDR37, WDR4, WDR45, WDR45B, WDR47, WDR5, WDR62, WDR73, WDR81, WDR83OS, WFS1, WIPI2, WLS, WNK1, WNK3, WNT1, WNT5A, WWOX, XK, XPA, XPNPEP3, XRCC4, XYLT1, YAP1, YARS1, YARS2, YIF1B, YIPF5, YWHAG, YY1, YY1AP1, ZBTB11, ZBTB18, ZBTB20, ZBTB24, ZBTB47, ZBTB7A, ZC3H14, ZC4H2, ZDHHC9, ZEB2, ZFHX3, ZFHX4, ZFX, ZFYVE26, ZIC1, ZIC2, ZMIZ1, ZMYM2, ZMYM3, ZMYND11, ZMYND8, ZNF142, ZNF148, ZNF292, ZNF335, ZNF341, ZNF407, ZNF423, ZNF462, ZNF526, ZNF668, ZNF699, ZNF711, ZNFX1, ZNHIT3, ZNRF3, ZPR1, ZSCAN10, ZSWIM6

CentoPediatric Neuro Comprehensive Genome | 2,404 genes

CentoPediatric Neuro Comprehensive Genome targets genes associated with a broad spectrum of disorders within pediatric neurology, including conditions such as neurodevelopmental delay and autism disorders, ataxia and cerebellar anomalies, spastic paraplegia, epilepsies, neuromuscular disorders, leukodystrophy/leukoencephalopathies, dystonia, chorea and related movement disorders, and structural CNS abnormalities. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

> This version of the panel is based on genome sequencing, it includes non-coding genes *CHASERR, RNU2-2, RNU4-2, RNU7-1, MIR17HG, SNORD118*; repeat expansion screening of the genes *ATXN10, FMR1, FXN, PPP2R2B, CSTB* and *DMPK*; *SMN1* CNV analysis; and *GBA1* conversion analysis with its pseudogene.

Genes Included

AAAS, AARS1, AARS2, AASS, ABAT, ABCA2, ABCB7, ABCC8, ABCC9, ABCD1, ABCD4, ABHD12, ABHD16A, ABHD5, ACACA, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD5, ACBD6, ACER3, ACO2, ACOX1, ACOX2, ACP5, ACSF3, ACSL4, ACTA1, ACTB, ACTG1, ACTG2, ACTL6A, ACTL6B, ACTN2, ACVR1, ACY1, ADA, ADAM22, ADAMTS10, ADAMTSL2, ADAR, ADARB1, ADAT3, ADCY5, ADD1, ADD3, ADGRA2, ADGRG1, ADGRG6, ADGRL1, ADGRV1, ADH5, ADK, ADNP, ADPRS, ADSL, ADSS1, AFF2, AFF3, AFF4, AFG2A, AFG2B, AFG3L2, AGA, AGAP1, AGK, AGL, AGMO, AGO1, AGO2, AGPAT3, AGRN, AGTPBP1, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, AJAP1, AKT1, AKT3, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALMS1, ALPL, ALS2, ALX3, ALX4, AMER1, AMFR, AMMECR1, AMPD2, AMT, ANAPC1, ANAPC7, ANK2, ANK3, ANKLE2, ANKRD11, ANKRD17, ANKS1B, ANO10, ANO3, ANO4, ANO5, ANTXR1, AP1B1,

AP1G1, AP1S1, AP1S2, AP2M1, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APTX, AR, ARCN1, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGAP35, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL6, ARL6IP1, ARMC9, ARPC4, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASCC1, ASCC3, ASCL1, ASH1L, ASL, ASNS, ASPA, ASPM, ASS1, ASTN1, ASXL1, ASXL2, ASXL3, ATAD1, ATAD3A, ATCAY, ATG4D, ATG7, ATIC, ATL1, ATM, ATN1, ATP11A, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A1, ATP2A2, ATP2B1, ATP2B3, ATP5F1A, ATP5F1E, ATP5MC3, ATP5PO, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP6V1B2, ATP7A, ATP7B, ATP8A2, ATP9A, ATR, ATRX, ATXN10, ATXN2L, ATXN7L3, AUH, AUTS2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BAG3, BAP1, BAZ2B, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCL11A, BCL11B, BCOR, BCORL1, BCS1L, BET1, BICD2, BICRA, BIN1, BLM, BLOC1S1, BLTP1, BMP4, BMPER, BOLA3, BORCS5, BORCS8, BPTF, BRAF, BRAT1, BRCA1, BRCA2, BRD4, BRF1, BRPF1, BRSK2, BRWD3, BSCL2, BSND, BTD, BUB1, BUB1B, C12orf57, C19orf12, C2CD3, C2orf69, CA2, CA5A, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA1S, CACNA2D2, CAD, CAMK2A, CAMK2B, CAMK2D, CAMSAP1, CAMTA1, CANT1, CAPN1, CAPN15, CAPN3, CAPRIN1, CAPZA2, CARS1, CARS2, CASK, CASP2, CASR, CAV1, CAV3, CAVIN1, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC186, CCDC32, CCDC47, CCDC82, CCDC88A, CCDC88C, CCM2, CCND2, CDC42, CDC42BPB, CDH11, CDH2, CDK13, CDK19, CDK5RAP2, CDK8, CDK9, CDKL5, CDKN1C, CDON, CELF2, CELF4, CENPF, CEP104, CEP120, CEP135, CEP152, CEP19, CEP290, CEP295, CEP41, CEP55, CEP57, CEP63, CEP83, CEP85L, CERS1, CERT1, CFAP418, CFL2, CHAMP1, CHAT, CHCHD10, CHD1, CHD2, CHD3, CHD4, CHD5, CHD7, CHD8, CHKA, CHKB, CHMP1A, CHMP2B, CHRNA1, CHRNA2, CHRNA4, CHRNB1, CHRNB2, CHRND, CHRNE, CHRNG, CHST14, CHSY1, CIC, CIT, CKAP2L, CLCN1, CLCN2, CLCN3, CLCN4, CLCN6, CLCNKA, CLCNKB, CLDN11, CLDN16, CLDN19, CLIC2, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, 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DIAPH1, DIP2C, DIS3L2, DKC1, DLAT, DLD, DLG3, DLG4, DLL1, DMBX1, DMD, DMPK, DMXL2, DNA2, DNAJB2, DNAJB4, DNAJB6, DNAJC12, DNAJC17, DNAJC19, DNAJC3, DNAJC5, DNAJC6, DNM1, DNM1L, DNM2, DNMT1, DNMT3A, DNMT3B, DOCK3, DOCK4, DOCK6, DOCK7, DOHH, DOK7, DOLK, DONSON, DPAGT1, DPF2, DPH1, DPH2, DPH5, DPM1, DPM2, DPM3, DPYD, DPYS, DPYSL2, DPYSL5, DRG1, DROSHA, DST, DSTYK, DTNA, DTYMK, DUOXA2, DVL3, DYM, DYNC1H1, DYNC1I2, DYRK1A, DYSF, EARS2, EBF3, EBP, ECEL1, ECHS1, ECM1, EDEM3, EED, EEF1A2, EEF1B2, EEF2, EEFSEC, EFNB1, EFTUD2, EGR2, EHMT1, EIF2A, EIF2AK2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4A2, EIF4A3, EIF5A, ELAC2, ELOVL1, ELOVL4, ELP1, ELP2, EMC1, EMC10, EMD, EMG1, EMILIN1, EML1, ENTPD1, EP300, EPG5, EPHA7, EPM2A, EPRS1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERGIC3, ERI1, ERLIN1, ERLIN2, ESAM, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOC2, EXOC7, EXOC8, EXOSC2, EXOSC3, EXOSC5, EXOSC8, EXOSC9, EXT2, EXTL3, EZH1, EZH2, F2, F5, FA2H, FAH, 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CentoPediatric Neuro – Ataxia and cerebellar anomalies | 193 genes

CentoPediatric Neuro – Ataxia and cerebellar anomalies targets genes associated with ataxia and cerebellar anomalies. This panel is intended for pediatric patients with clinical features suggestive of ataxia and cerebellar anomalies and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AAAS, ABCA2, ABCB7, ABHD12, ACBD6, ACO2, ADGRG1, ADPRS, AGTPBP1, ALDH5A1, AP1S2, APTX, ARSA, ASL, ATAD3A, ATCAY, ATG7, ATM, ATP1A3, ATP2B3, ATP6V0A1, ATP8A2, ATXN10, CA8, CACNA1A, CACNA1G, CAMTA1, CAPRIN1, CASK, CHMP1A, CLCN2, CLN5, CLN6, COA7, COASY, COG5, COQ4, COQ8A, COX20, CTBP1, CWF19L1, DAGLA, DARS2, DKC1, DLG4, DNAJC19, DNAJC3, DOCK3, DPYSL5, EBF3, EEFSEC, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPM2A, EXOSC3, EXOSC5, FA2H, FBXL4, FDXR, FEM1C, FGF14, FLVCR1, FOLR1, FRMD5, FTH1, FXN, GBA2, GEMIN5, GFAP, GJC2, GPAA1, GRID2, GRM1, GRN, HEXA, HEXB, HMBS, INTS11, IRF2BPL, ITPR1, KCNA1, KCNA2, KCNC3, KCNJ10, KCNN2, KIF1A, KIF1C, LAMA1, LETM1, LIG3, LNPK, MAG, MAPK8IP3, MFSD8, MINPP1, MORC2, MRE11, MSTO1, MTFMT, MTTP, MVK, NAXE, NFASC, NKX2-1, NKX6-2, NPC1, NPC2, NUS1, OGDHL, OPA1, OPA3, OPHN1, PEX16, PEX6, PI4KA, PITRM1, PLA2G6, PMPCA, PMPCB, PNKP, PNPLA6, PNPT1, POLG, POLR3A, POLR3B, POU4F1, PPP2R2B, PRDM13, PRDX3, PRKCG, PRRT2, PTF1A, RARS2, RELN, RNF220, ROBO3, RORA, SACS, SCN2A, SCN8A, SCYL1, SEPSECS, SETX, SIL1, SLC17A5, SLC1A3, SLC25A46, SLC2A1, SLC44A1, SLC52A2, SLC9A1, SLC9A6, SMPD4, SNAP25, SNX14, SPR, SPTAN1, SPTBN2, SQSTM1, STUB1, SVBP, SYNE1, TANGO2, TBC1D23, TDP2, TECPR2, THG1L, TMEM106B, TMEM240, TOE1, TPP1, TSEN15, TSEN2, TSEN54, TTC19, TTPA, TUBA1A, TUBB4A, TWNK, UBTF, UCHL1, VAMP1, VLDLR, VPS13D, VPS41, VPS53, VRK1, WDR73, WDR81, WWOX

CentoPediatric Neuro – Ataxia and cerebellar anomalies Genome | 193 genes

CentoPediatric Neuro - Ataxia and cerebellar anomalies Genome targets genes associated with ataxia and cerebellar anomalies. This panel is intended for pediatric patients with clinical features suggestive of ataxia and cerebellar anomalies and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This version of the panel is based on genome sequencing; it includes repeat expansion screening of *ATXN10, FXN, and PPP2R2B*.

Genes Included

AAAS, ABCA2, ABCB7, ABHD12, ACBD6, ACO2, ADGRG1, ADPRS, AGTPBP1, ALDH5A1, AP1S2, APTX, ARSA, ASL, ATAD3A, ATCAY, ATG7, ATM, ATP1A3, ATP2B3, ATP6V0A1, ATP8A2, ATXN10, CA8, CACNA1A, CACNA1G, CAMTA1, CAPRIN1, CASK, CHMP1A, CLCN2, CLN5, CLN6, COA7, COASY, COG5, COQ4, COQ8A, COX20, CTBP1, CWF19L1, DAGLA, DARS2, DKC1, DLG4, DNAJC19, DNAJC3, DOCK3, DPYSL5, EBF3, EEFSEC, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPM2A, EXOSC3, EXOSC5, FA2H, FBXL4, FDXR, FEM1C, FGF14, FLVCR1, FOLR1, FRMD5, FTH1, FXN, GBA2, GEMIN5, GFAP, GJC2, GPAA1, GRID2, GRM1, GRN, HEXA, HEXB, HMBS, INTS11, IRF2BPL, ITPR1, KCNA1, KCNA2, KCNC3, KCNJ10, KCNN2, KIF1A, KIF1C, LAMA1, LETM1, LIG3, LNPK, MAG, MAPK8IP3, MFSD8, MINPP1, MORC2, MRE11, MSTO1, MTFMT, MTTP, MVK, NAXE, NFASC, NKX2-1, NKX6-2, NPC1, NPC2, NUS1, OGDHL, OPA1, OPA3, OPHN1, PEX16, PEX6, PI4KA, PITRM1, PLA2G6, PMPCA, PMPCB, PNKP, PNPLA6, PNPT1, POLG, POLR3A, POLR3B, POU4F1, PPP2R2B, PRDM13, PRDX3, PRKCG, PRRT2, PTF1A, RARS2, RELN, RNF220, ROBO3, RORA, SACS, SCN2A, SCN8A, SCYL1, SEPSECS, SETX, SIL1, SLC17A5, SLC1A3, SLC25A46, SLC2A1, SLC44A1, SLC52A2, SLC9A1, SLC9A6, SMPD4, SNAP25, SNX14, SPR,

SPTAN1, SPTBN2, SQSTM1, STUB1, SVBP, SYNE1, TANGO2, TBC1D23, TDP2, TECPR2, THG1L, TMEM106B, TMEM240, TOE1, TPP1, TSEN15, TSEN2, TSEN54, TTC19, TTPA, TUBA1A, TUBB4A, TWNK, UBTF, UCHL1, VAMP1, VLDLR, VPS13D, VPS41, VPS53, VRK1, WDR73, WDR81, WWOX

CentoPediatric Neuro – Dystonia, chorea and related movement disorders | 205 genes

CentoPediatric Neuro - Dystonia, chorea and related movement disorders targets genes associated with dystonia, chorea and related movement disorders. This panel is intended for pediatric patients with clinical features suggestive of dystonia, chorea and related movement disorders and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Repeat expansion analysis of *ARX*.

Genes Included

ABAT, ABCB7, ACBD6, ACER3, ACOX1, ACSF3, ACTB, ADAR, ADCY5, AFG2B, AFG3L2, ALDH18A1, ANO10, ANO3, AP1S2, APTX, ARSA, ARX, ASL, ATCAY, ATM, ATP13A2, ATP1A2, ATP1A3, ATP5MC3, ATP7B, AUH, BCAP31, BCS1L, C19ORF12, CA8, CACNA1A, CACNA1G, CLN3, CLN5, CLN8, CLPB, COASY, COX10, COX15, COX20, CTSD, CWF19L1, DCAF17, DCC, DDC, DHDDS, DLAT, DLD, DNAJC12, DNAJC6, ECHS1, EIF2AK2, ELOVL4, FA2H, FBXO7, FGF14, FITM2, FOLR1, FOXG1, FOXRED1, FTL, FUCA1, GBA1, GCDH, GCH1, GJC2, GLB1, GLRA1, GLRB, GM2A, GNAL, GNAO1, GNB1, GRID2, GRIN1, GRM1, GTPBP2, HCFC1, HEXA, HIBCH, HNRNPH1, HPCA, HPRT1, HSD17B10, HSPD1, HTRA2, IFIH1, IMPDH2, IRF2BPL, ITPR1, KCNA1, KCNC3, KCND3, KCNMA1, KCNQ2, KCTD17, KIF1C, KMT2B, L2HGDH, LRPPRC, MAL, MARS2, MECR, MED27, MRE11, MTFMT, NDUFA1, NDUFA12, NDUFA2, NDUFAF5, NDUFAF6, NDUFS1, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NGLY1, NKX2-1, NKX6-2, NPC1, NPC2, NUP54, NUS1, OPA3, PANK2, PCCA, PCCB, PCDH12, PDE10A, PDE2A, PDGFB, PDHA1, PDHX, PET100, PINK1, PLA2G6, PLP1, PNKD, PNKP, PNPT1, POLR3A, PRKCG, PRKRA, PRRT2, PTS, QDPR, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SACS, SCN1A, SCN8A, SERAC1, SETX, SGCE, SHQ1, SIL1, SLC16A2, SLC18A2, SLC19A3, SLC1A3, SLC2A1, SLC30A10, SLC30A9, SLC39A14, SLC6A3, SLC6A5, SLC6A8, SNX14, SPR, SQSTM1, STUB1, SUCLA2, SUOX, SURF1, SYT1, TAF1, TARS2, TBC1D24, TH, THAP1, TIMM8A, TMEM151A, TMEM240, TOR1A, TPK1, TPP1, TREX1, TSPOAP1, TUBB4A, UBTF, VAC14, VAMP1, VAMP2, VPS13D, VPS16, VPS41, VPS4A, WDR45, WWOX, YIF1B, YY1, ZSWIM6

CentoPediatric Neuro – Dystonia, chorea and related movement disorders Genome | 207 genes

CentoPediatric Neuro - Dystonia, chorea and related movement disorders Genome targets genes associated with dystonia, chorea and related movement disorders. This panel is intended for pediatric patients with clinical features suggestive of dystonia, chorea and related movement disorders and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This genome-based panel includes the non-coding genes *RNU7-1* and *SNORD118*; and *GBA1* conversion analysis with its pseudogene.
Repeat expansion analysis of *ARX*.

Genes Included

ABAT, ABCB7, ACBD6, ACER3, ACOX1, ACSF3, ACTB, ADAR, ADCY5, AFG2B, AFG3L2, ALDH18A1, ANO10, ANO3, AP1S2, APTX, ARSA, ARX, ASL, ATCAY, ATM, ATP13A2, ATP1A2, ATP1A3, ATP5MC3, ATP7B, AUH, BCAP31, BCS1L, C19ORF12, CA8, CACNA1A, CACNA1G, CLN3, CLN5, CLN8, CLPB, COASY, COX10, COX15, COX20, CTSD, CWF19L1, DCAF17, DCC, DDC, DHDDS, DLAT, DLD, DNAJC12, DNAJC6, ECHS1, EIF2AK2, ELOVL4, FA2H, FBXO7, FGF14, FITM2, FOLR1, FOXG1, FOXRED1, FTL, FUCA1, GBA1, GCDH, GCH1, GJC2, GLB1, GLRA1, GLRB, GM2A, GNAL, GNAO1, GNB1, GRID2, GRIN1, GRM1, GTPBP2, HCFC1, HEXA, HIBCH, HNRNPH1, HPCA, HPRT1, HSD17B10, HSPD1, HTRA2, IFIH1, IMPDH2, IRF2BPL, ITPR1, KCNA1, KCNC3, KCND3, KCNMA1, KCNQ2, KCTD17, KIF1C, KMT2B, L2HGDH, LRPPRC, MAL, MARS2, MECR, MED27, MRE11, MTFMT, NDUFA1, NDUFA12, NDUFA2, NDUFAF5, NDUFAF6, NDUFS1, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NGLY1, NKX2-1, NKX6-2, NPC1, NPC2, NUP54, NUS1, OPA3, PANK2, PCCA, PCCB, PCDH12, PDE10A, PDE2A, PDGFB, PDHA1, PDHX, PET100, PINK1, PLA2G6, PLP1, PNKD, PNKP, PNPT1, POLR3A, PRKCG, PRKRA, PRRT2, PTS, QDPR, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNU7-1, SACS, SCN1A, SCN8A, SERAC1, SETX, SGCE, SHQ1, SIL1, SLC16A2, SLC18A2, SLC19A3, SLC1A3, SLC2A1, SLC30A10, SLC30A9, SLC39A14, SLC6A3, SLC6A5, SLC6A8, SNORD118, SNX14, SPR, SQSTM1, STUB1, SUCLA2, SUOX, SURF1, SYT1, TAF1, TARS2, TBC1D24, TH, THAP1, TIMM8A, TMEM151A, TMEM240, TOR1A, TPK1, TPP1, TREX1, TSPOAP1, TUBB4A, UBTF, VAC14, VAMP1, VAMP2, VPS13D, VPS16, VPS41, VPS4A, WDR45, WWOX, YIF1B, YY1, ZSWIM6

CentoPediatric Neuro – Epilepsy | 1,059 genes

CentoPediatric Neuro – Epilepsy targets genes associated with epilepsies. This panel is intended for pediatric patients with clinical features suggestive of epilepsies and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AARS1, AARS2, ABAT, ABCA2, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADVL, ACOX1, ACTL6B, ACY1, ADA, ADAM22, ADAMTS10, ADAMTSL2, ADAR, ADARB1, ADAT3, ADD1, ADGRG1, ADGRL1, ADGRV1, ADPRS, ADSL, AFF3, AFG2A, AFG2B, AFG3L2, AGA, AGK, AGMO, AGO1, AIFM1, AIMP1, AIMP2, AJAP1, AKT1, AKT3, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALPL, AMPD2, AMT, ANK2, ANK3, ANKRD11, ANO4, AP1G1, AP2M1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APTX, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGEF9, ARID1B, ARSA, ARSB, ARV1, ARX, ASAH1, ASH1L, ASL, ASNS, ASPA, ASS1, ASTN1, ASXL1, ASXL3, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B1, ATP5F1A, ATP5PO, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP7A, ATP7B, ATRX, AUH, B3GALNT2, B3GLCT, B4GALT1, BAP1, BCAP31, BCKDHA, BCKDHB, BCORL1, BCS1L, BET1, BLOC1S1, BLTP1, BOLA3, BORCS8, BRAF, BRAT1, BSCL2, BTD, C12orf57, C19orf12, C2orf69, CA5A, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D2, CAD, CAMK2D, CAMSAP1, CAPRIN1, CARS2, CASK, CASR, CBL, CBS, CC2D2A, CCDC186, CCDC88A, CCDC88C, CCND2, CDC42BPB, CDK19, CDKL5, CELF2, CEP85L, CERS1, CERT1, CHD2, CHD4, CHD5, CHKA, CHMP2B, CHRNA2, CHRNA4, CHRNB2, CIC, CLCN2, CLCN3, CLCN4, CLCN6, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, CLTC, CNKSR2, CNNM2, CNPY3, CNTN2, CNTNAP1, CNTNAP2, COA7, COA8, COASY, COG1, COG3, COG4, COG5, COG6, COG7, COG8, COL18A1, COL2A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ4, COQ6, COQ8A, COQ9, COX10, COX15, COX20, COX6B1, CP, CPLX1, CPS1, CPSF3, CPT1A, CPT2, CREBBP, CRELD1, CRPPA, CSF1R, CSNK2A1, CSNK2B, CSTB, CTC1, CTNNA2, CTNS, CTSA, CTSD, CTSF, CTU2, CUL3, CUL4B, CUX1, CUX2, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DALRD3, DARS1, DARS2, DBT, DCAF17, DCX, DDC, DDX3X, DEAF1, DEGS1, DENND5A, DENND5B, DEPDC5, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHRSX, DHX16, DHX30, DIAPH1, DKC1, DLAT, DLD, DLL1, DMBX1, DMXL2, DNAJC5, DNAJC6, DNM1, DNM1L, DOCK6, DOCK7, DOLK, DPAGT1, DPH5, DPM1, DPM2, DPM3, DPYD, DPYS, DROSHA, DTYMK, DYM, DYNC1H1, DYRK1A, EARS2, ECHS1, EEF1A2, EFTUD2, EHMT1, EIF2A, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4A2, EMC10, EML1, ENTPD1, EPG5, EPM2A, EPRS1, ERCC6, ERCC8, ESAM, ETFA, ETFB, ETFDH, ETHE1, EXOC7, EXOSC3, EXT2, F2, F5, FA2H, FAH, FAM50A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FBXO11, FBXO28, FCSK, FDFT1, FDX2, FGF12, FGF13, FGFR3, FH, FIG4, FKRP, FKTN, FLNA, FLVCR2, FOLR1, FOXG1, FOXRED1, FRMD5, FRRS1L, FTL, FUCA1, FUT8, FXYD2, FZR1, GAA, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALNS, GALNT2, GALT, GAMT, GAN, GATAD2B, GATM, GBA1, GCDH, GCH1, GCSH, GFAP, GFER, GFM1, GFM2, GFPT1, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLI3, GLRA1, GLRA2, GLRB, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GNAO1, GNAQ, GNB1, GNB2, GNB5, GNE, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GOT2, GPAA1, GPC3, GPHN, GRIA2, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GRN, GSS, GTPBP2, GTPBP3, GUSB, H3-3A, H3-3B, HACE1, HADHA, HADHB, HAX1, HCCS, HCFC1, HCN1, HCN2, HEATR5B, HECTD4, HECW2, HEPACAM, HERC2, HEXA, HEXB, HGSNAT, HIBCH, HID1, HIKESHI, HLCS, HMGCL, HMGCS2, HNRNPH2, HNRNPR, HNRNPU, HOXA1, HPDL, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HTRA1, HTRA2, HYCC1, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IKBKG, INO80, IQSEC2, IRF2BPL, ISCA2, ITPA, IVD, JAG1, JAKMIP1, JAM3, KARS1, KAT5, KAT8, KATNB1, KCNA1, KCNA2, KCNA3, KCNB1, KCNB2, KCNC1, KCNC2, KCND2, KCNH1, KCNH5, KCNJ10, KCNJ11, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD3, KCTD7, KDM6B, KIF1A, KIF2A, KIF5A, KIF5C, KIFBP, KLHL20, KMT2E, KMT5B, KPTN, KRAS, L2HGDH, LAMA2, LAMB1, LAMP2, LARGE1, LARS1, LAT, LDB3, LETM1, LGI1, LIAS, LIPT1, LIPT2, LMAN2L, LMBRD2, LMNB1, LMNB2, LNPK, LRPPRC, LSS, LYRM7, LYST, MACF1, MADD, MAF, MAGI2, MAGT1, MAN1B1, MAN2B1, MANBA, MAP2K1, MAP2K2, MARS2, MAST1, MAST3, MAST4, MBD5, MBOAT7, MCCC1, MCCC2, MCM3AP, MCOLN1, MDH2, MECP2, MECR, MED11, MED12, MED17, MED27, MEF2C, MFF, MFN2, MFSD8, MGAT2, MGME1, MINPP1, MLC1, MLPH, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MRPS22, MT-CO3, MTFMT, MTHFR, MTHFS, MTOR, MTR, MTRFR, MT-TL1, MYO5A, NACC1, NAGA, NAGLU, NAGS, NAPB, NARS1, NARS2, NAXD, NAXE, NBAS, NBEA, NCDN, NDE1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NECAP1, NEDD4L, NEU1, NEUROD2, NEXMIF, NF1, NFU1, NGLY1, NHLRC1, NID1, NOTCH1, NOTCH3, NPC1, NPC2, NPRL2, NPRL3, NR4A2, NRAS, NRROS, NRXN1, NSD1, NSD2, NSDHL, NSF, NSRP1, NTRK2, NUBPL, NUP214, NUS1, OAT, OCLN, OCRL, OGDHL, OPA1, OPA3, OPHN1, OSGEP, OTC, OTUD6B, OTUD7A, OTX2, OXR1, P4HTM, PABPC1, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PANK2, PARP6, PARS2, PC, PCCA, PCCB, PCDH12, PCDH19, PCDHGC4, PCLO, PCYT2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PGM1, PGM2L1, PHACTR1, PHGDH, PHYH, PIDD1, PIGA, PIGB, PIGG, PIGK, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGV, PIK3CA, PIK3R2, PIP5K1C, PLA2G6, PLAA, PLCB1, PLEKHG2, PLK1, PLP1, PLPBP, PLXNA1, PMM2, PMPCB, PNKP, PNPO, PNPT1, POLG, POLG2, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPFIBP1, PPIL1, PPP1R3F, PPP2CA, PPP2R1A, PPP3CA, PPT1, PRICKLE2, PRMT7, PRODH, PRPF8, PRPS1, PRRT2, PSAP, PSAT1, PSMB8, PSPH, PTCD3, PTCH1, PTEN, PTF1A, PTPN23, PTS, PUM1, PURA, PYCR2, QARS1, QDPR, RAB11A, RAB11B, RAB18, RAB27A, RAB3GAP1, RAB3GAP2, RAB5C, RAC3, RAI1, RALA, RALGAPA1, RALGAPB, RANBP2, RARS1, RARS2, RELN, RFT1, RHOBTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF2, RNU4ATAC, ROGDI, RORA, RORB, RPIA, RRM2B, RTN4IP1, RTTN, RUBCN, RUSC2, RYR2, SAMD12, SAMHD1, SARS1, SATB1, SATB2, SCAF4, SCAMP5, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEC24D, SEC31A, SELENOI, SEMA6B, SEPSECS, SERAC1, SERPINI1, SETBP1, SETD1A, SETD1B, SETD5, SGCE, SGSH, SHH, SHQ1, SIK1, SIX3, SLC12A3, SLC12A5, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A4, SLC2A1, SLC31A1, SLC32A1, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC38A3,

SLC39A8, SLC45A1, SLC5A6, SLC6A1, SLC6A19, SLC6A5, SLC6A8, SLC7A7, SLC9A6, SMARCA2, SMARCC2, SMC1A, SMPD1, SMS, SNAP25, SNF8, SNIP1, SNX27, SON, SOX10, SPART, SPG11, SPG7, SPR, SPTAN1, SPTBN1, SPTBN4, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STAG1, STAMBP, STARD7, STAT1, STIL, STRADA, STT3A, STX1B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYN1, SYNCRIP, SYNE1, SYNGAP1, SYNJ1, SZT2, TACO1, TAF8, TANC2, TANGO2, TBC1D20, TBC1D24, TBC1D2B, TBCD, TBCE, TBCK, TBL1XR1, TCF4, TDP2, TEFM, TELO2, TET3, TFE3, TGFB1, TGIF1, TIAM1, TIMM50, TIMM8A, TINF2, TK2, TMEM106B, TMEM126A, TMEM165, TMEM222, TMEM63B, TMEM70, TMX2, TNPO2, TPK1, TPP1, TRA2B, TRAF7, TRAK1, TRAPPC12, TRAPPC4, TRAPPC6B, TRAPPC9, TREX1, TRIM8, TRIP13, TRIT1, TRPM3, TRPM6, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TSFM, TTC19, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUFM, TUSC3, TWNK, TXN2, TYMP, U2AF2, UBA5, UBAP2L, UBE2A, UBE3A, UBR7, UFC1, UFM1, UFSP2, UGDH, UGP2, UMPS, UNC80, UPB1, UQCRQ, USP18, USP7, VAMP2, VARS1, VARS2, VLDLR, VMA12, VMA22, VPS11, VPS50, WARS2, WASF1, WDR37, WDR45, WDR45B, WDR62, WDR73, WFS1, WNK3, WWOX, XK, YIF1B, YIPF5, YWHAG, ZBTB18, ZBTB47, ZDHHC9, ZEB2, ZFYVE26, ZIC2, ZMIZ1, ZMYM2, ZNF142, ZNF335, ZNFX1

CentoPediatric Neuro – Epilepsy Genome | 1,062 genes

CentoPediatric Neuro - Epilepsy Genome targets genes associated with epilepsies. This panel is intended for pediatric patients with clinical features suggestive of epilepsies and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

> This version of the panel is based on genome-sequencing; it includes the non-coding genes *RNU2-2*, *RNU4-2* and *SNORD118*; repeat expansion screening of *CSTB*; and *GBA1* conversion analysis with its pseudogene.

Genes Included

AARS1, AARS2, ABAT, ABCA2, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADVL, ACOX1, ACTL6B, ACY1, ADA, ADAM22, ADAMTS10, ADAMTSL2, ADAR, ADARB1, ADAT3, ADD1, ADGRG1, ADGRL1, ADGRV1, ADPRS, ADSL, AFF3, AFG2A, AFG2B, AFG3L2, AGA, AGK, AGMO, AGO1, AIFM1, AIMP1, AIMP2, AJAP1, AKT1, AKT3, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALPL, AMPD2, AMT, ANK2, ANK3, ANKRD11, ANO4, AP1G1, AP2M1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APTX, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGEF9, ARID1B, ARSA, ARSB, ARV1, ARX, ASAH1, ASH1L, ASL, ASNS, ASPA, ASS1, ASTN1, ASXL1, ASXL3, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B1, ATP5F1A, ATP5PO, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP7A, ATP7B, ATRX, AUH, B3GALNT2, B3GLCT, B4GALT1, BAP1, BCAP31, BCKDHA, BCKDHB, BCORL1, BCS1L, BET1, BLOC1S1, BLTP1, BOLA3, BORCS8, BRAF, BRAT1, BSCL2, BTD, C12orf57, C19orf12, C2orf69, CA5A, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D2, CAD, CAMK2D, CAMSAP1, CAPRIN1, CARS2, CASK, CASR, CBL, CBS, CC2D2A, CCDC186, CCDC88A, CCDC88C, CCND2, CDC42BPB, CDK19, CDKL5, CELF2, CEP85L, CERS1, CERT1, CHD2, CHD4, CHD5, CHKA, CHMP2B, CHRNA2, CHRNA4, CHRNB2, CIC, CLCN2, CLCN3, CLCN4, CLCN6, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, CLTC, CNKSR2, CNNM2, CNPY3, CNTN2, CNTNAP1, CNTNAP2, COA7, COA8, COASY, COG1, COG3, COG4, COG5, COG6, COG7, COG8, COL18A1, COL2A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ4, COQ6, COQ8A, COQ9, COX10, COX15, COX20, COX6B1, CP, CPLX1, CPS1, CPSF3, CPT1A, CPT2, CREBBP, CRELD1, CRPPA, CSF1R, CSNK2A1, CSNK2B, CSTB, CTC1, CTNNA2, CTNS, CTSA, CTSD, CTSF, CTU2, CUL3, CUL4B, CUX1, CUX2, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DALRD3, DARS1, DARS2, DBT, DCAF17, DCX, DDC, DDX3X, DEAF1, DEGS1, DENND5A, DENND5B, DEPDC5, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHRSX, DHX16, DHX30, DIAPH1, DKC1, DLAT, DLD, DLL1, DMBX1, DMXL2, DNAJC5, DNAJC6, DNM1, DNM1L, DOCK6, DOCK7, DOLK, DPAGT1, DPH5, DPM1, DPM2, DPM3, DPYD, DPYS, DROSHA, DTYMK, DYM, DYNC1H1, DYRK1A, EARS2, ECHS1, EEF1A2, EFTUD2, EHMT1, EIF2A, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4A2, EMC10, EML1, ENTPD1, EPG5, EPM2A, EPRS1, ERCC6, ERCC8, ESAM, ETFA, ETFB, ETFDH, ETHE1, EXOC7, EXOSC3, EXT2, F2, F5, FA2H, FAH, FAM50A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FBXO11, FBXO28, FCSK, FDFT1, FDX2, FGF12, FGF13, FGFR3, FH, FIG4, FKRP, FKTN, FLNA, FLVCR2, FOLR1, FOXG1, FOXRED1, FRMD5, FRRS1L, FTL, FUCA1, FUT8, FXYD2, FZR1, GAA, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALNS, GALNT2, GALT, GAMT, GAN, GATAD2B, GATM, GBA1, GCDH, GCH1, GCSH, GFAP, GFER, GFM1, GFM2, GFPT1, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLI3, GLRA1, GLRA2, GLRB, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GNAO1, GNAQ, GNB1, GNB2, GNB5, GNE, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GOT2, GPAA1, GPC3, GPHN, GRIA2, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GRN, GSS, GTPBP2, GTPBP3, GUSB, H3-3A, H3-3B, HACE1, HADHA, HADHB, HAX1, HCCS, HCFC1, HCN1, HCN2, HEATR5B, HECTD4, HECW2, HEPACAM, HERC2, HEXA, HEXB, HGSNAT, HIBCH, HID1, HIKESHI, HLCS, HMGCL, HMGCS2, HNRNPH2, HNRNPR, HNRNPU, HOXA1, HPDL, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HTRA1, HTRA2, HYCC1, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IKBKG, INO80, IQSEC2, IRF2BPL, ISCA2, ITPA, IVD, JAG1, JAKMIP1, JAM3, KARS1, KAT5, KAT8, KATNB1, KCNA1, KCNA2, KCNA3, KCNB1, KCNB2, KCNC1, KCNC2, KCND2, KCNH1, KCNH5, KCNJ10, KCNJ11, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD3, KCTD7, KDM6B, KIF1A, KIF2A, KIF5A, KIF5C, KIFBP, KLHL20, KMT2E, KMT5B, KPTN, KRAS, L2HGDH, LAMA2, LAMB1, LAMP2, LARGE1, LARS1, LAT, LDB3, LETM1, LGI1, LIAS, LIPT1, LIPT2, LMAN2L, LMBRD2, LMNB1, LMNB2, LNPK, LRPPRC, LSS, LYRM7, LYST, MACF1, MADD, MAF, MAGI2, MAGT1, MAN1B1, MAN2B1, MANBA, MAP2K1, MAP2K2, MARS2, MAST1, MAST3, MAST4, MBD5, MBOAT7, MCCC1, MCCC2, MCM3AP, MCOLN1, MDH2, MECP2, MECR, MED11, MED12, MED17, MED27, MEF2C, MFF, MFN2, MFSD8, MGAT2, MGME1, MINPP1, MLC1, MLPH, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MRPS22, MT-CO3, MTFMT, MTHFR, MTHFS, MTOR, MTR, MTRFR, MT-TL1, MYO5A, NACC1, NAGA, NAGLU, NAGS, NAPB, NARS1, NARS2, NAXD, NAXE, NBAS, NBEA, NCDN, NDE1, NDUFA1, NDUFA10, NDUFA11,

NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NECAP1, NEDD4L, NEU1, NEUROD2, NEXMIF, NF1, NFU1, NGLY1, NHLRC1, NID1, NOTCH1, NOTCH3, NPC1, NPC2, NPRL2, NPRL3, NR4A2, NRAS, NRROS, NRXN1, NSD1, NSD2, NSDHL, NSF, NSRP1, NTRK2, NUBPL, NUP214, NUS1, OAT, OCLN, OCRL, OGDHL, OPA1, OPA3, OPHN1, OSGEP, OTC, OTUD6B, OTUD7A, OTX2, OXR1, P4HTM, PABPC1, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PANK2, PARP6, PARS2, PC, PCCA, PCCB, PCDH12, PCDH19, PCDHGC4, PCLO, PCYT2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PGM1, PGM2L1, PHACTR1, PHGDH, PHYH, PIDD1, PIGA, PIGB, PIGG, PIGK, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGV, PIK3CA, PIK3R2, PIP5K1C, PLA2G6, PLAA, PLCB1, PLEKHG2, PLK1, PLP1, PLPBP, PLXNA1, PMM2, PMPCB, PNKP, PNPO, PNPT1, POLG, POLG2, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPFIBP1, PPIL1, PPP1R3F, PPP2CA, PPP2R1A, PPP3CA, PPT1, PRICKLE2, PRMT7, PRODH, PRPF8, PRPS1, PRRT2, PSAP, PSAT1, PSMB8, PSPH, PTCD3, PTCH1, PTEN, PTF1A, PTPN23, PTS, PUM1, PURA, PYCR2, QARS1, QDPR, RAB11A, RAB11B, RAB18, RAB27A, RAB3GAP1, RAB3GAP2, RAB5C, RAC3, RAI1, RALA, RALGAPA1, RALGAPB, RANBP2, RARS1, RARS2, RELN, RFT1, RHOBTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF2, RNU2-2, RNU4-2, RNU4ATAC, ROGDI, RORA, RORB, RPIA, RRM2B, RTN4IP1, RTTN, RUBCN, RUSC2, RYR2, SAMD12, SAMHD1, SARS1, SATB1, SATB2, SCAF4, SCAMP5, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEC24D, SEC31A, SELENOI, SEMA6B, SEPSECS, SERAC1, SERPINI1, SETBP1, SETD1A, SETD1B, SETD5, SGCE, SGSH, SHH, SHQ1, SIK1, SIX3, SLC12A3, SLC12A5, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A4, SLC2A1, SLC31A1, SLC32A1, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC38A3, SLC39A8, SLC45A1, SLC5A6, SLC6A1, SLC6A19, SLC6A5, SLC6A8, SLC7A7, SLC9A6, SMARCA2, SMARCC2, SMC1A, SMPD1, SMS, SNAP25, SNF8, SNIP1, SNORD118, SNX27, SON, SOX10, SPART, SPG11, SPG7, SPR, SPTAN1, SPTBN1, SPTBN4, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STAG1, STAMBP, STARD7, STAT1, STIL, STRADA, STT3A, STX1B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYN1, SYNCRIP, SYNE1, SYNGAP1, SYNJ1, SZT2, TACO1, TAF8, TANC2, TANGO2, TBC1D20, TBC1D24, TBC1D2B, TBCD, TBCE, TBCK, TBL1XR1, TCF4, TDP2, TEFM, TELO2, TET3, TFE3, TGFB1, TGIF1, TIAM1, TIMM50, TIMM8A, TINF2, TK2, TMEM106B, TMEM126A, TMEM165, TMEM222, TMEM63B, TMEM70, TMX2, TNPO2, TPK1, TPP1, TRA2B, TRAF7, TRAK1, TRAPPC12, TRAPPC4, TRAPPC6B, TRAPPC9, TREX1, TRIM8, TRIP13, TRIT1, TRPM3, TRPM6, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TSFM, TTC19, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUFM, TUSC3, TWNK, TXN2, TYMP, U2AF2, UBA5, UBAP2L, UBE2A, UBE3A, UBR7, UFC1, UFM1, UFSP2, UGDH, UGP2, UMPS, UNC80, UPB1, UQCRQ, USP18, USP7, VAMP2, VARS1, VARS2, VLDLR, VMA12, VMA22, VPS11, VPS50, WARS2, WASF1, WDR37, WDR45, WDR45B, WDR62, WDR73, WFS1, WNK3, WWOX, XK, YIF1B, YIPF5, YWHAG, ZBTB18, ZBTB47, ZDHHC9, ZEB2, ZFYVE26, ZIC2, ZMIZ1, ZMYM2, ZNF142, ZNF335, ZNFX1

CentoPediatric Neuro – Leukodystrophy/leukoencephalopathy | 163 genes

CentoPediatric Neuro – Leukodystrophy/leukoencephalopathy targets genes associated with leukodystrophy/leukoencephalopathies. This panel is intended for pediatric patients with clinical features suggestive of leukodystrophy/leukoencephalopathies and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AARS2, ABCD1, ACBD5, ACER3, ACOX1, ADAR, ADGRG1, AIFM1, AIMP1, AIMP2, ALDH3A2, AP4B1, AP4E1, AP4M1, AP4S1, ARSA, ASPA, BCAP31, BOLA3, BPTF, CLCN2, CLDN11, CNTNAP1, COA7, COA8, COL4A1, COL4A2, COLGALT1, COQ2, COQ9, COX10, COX15, COX6B1, CSF1R, CTC1, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DDHD2, DEGS1, DGUOK, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, ERCC6, ERCC8, FA2H, FARS2, FBXL4, FDFT1, FDX2, FOLR1, FOXRED1, FUCA1, GALC, GBE1, GFAP, GFM1, GJC2, HEPACAM, HIBCH, HIKESHI, HSD17B4, HSPD1, HTRA1, HYCC1, IBA57, ISCA2, JAM3, KARS1, L2HGDH, LMNB1, LRPPRC, LYRM7, MARS2, MLC1, MOCS1, MRPL44, MRPS16, MRPS22, MTFMT, MTHFS, NAXD, NAXE, NDUFA1, NDUFA10, NDUFA12, NDUFA2, NDUFA9, NDUFAF2, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB8, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NFU1, NKX6-2, NOTCH3, NT5C2, NUBPL, OCLN, PC, PDHA1, PEX1, PEX10, PEX12, PEX13, PEX5, PLAA, PLEKHG2, PLP1, POLR1A, POLR1C, POLR3A, POLR3B, PSAP, PYCR2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, RNF220, SAMHD1, SCO1, SDHAF1, SERAC1, SLC16A2, SLC17A5, SLC19A3, SLC1A4, SLC6A9, SOX10, SPART, STN1, SUMF1, SURF1, TMEM106B, TMEM63A, TREX1, TTC19, TUBB4A, TUFM, TWNK, UFM1, VPS11, YY1, ZFYVE26

CentoPediatric Neuro – Leukodystrophy/leukoencephalopathy Genome | 164 genes

CentoPediatric Neuro - Leukodystrophy/leukoencephalopathy Genome targets genes associated with leukodystrophy/leukoencephalopathies. This panel is intended for pediatric patients with clinical features suggestive of leukodystrophy/leukoencephalopathies and supports the identification of a molecular diagnosis to inform clinical management, prognosis,

and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This version of the panel is genome-sequencing based and it includes the non-coding gene *SNORD118*.

Genes Included

AARS2, ABCD1, ACBD5, ACER3, ACOX1, ADAR, ADGRG1, AIFM1, AIMP1, AIMP2, ALDH3A2, AP4B1, AP4E1, AP4M1, AP4S1, ARSA, ASPA, BCAP31, BOLA3, BPTF, CLCN2, CLDN11, CNTNAP1, COA7, COA8, COL4A1, COL4A2, COLGALT1, COQ2, COQ9, COX10, COX15, COX6B1, CSF1R, CTC1, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DDHD2, DEGS1, DGUOK, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, ERCC6, ERCC8, FA2H, FARS2, FBXL4, FDFT1, FDX2, FOLR1, FOXRED1, FUCA1, GALC, GBE1, GFAP, GFM1, GJC2, HEPACAM, HIBCH, HIKESHI, HSD17B4, HSPD1, HTRA1, HYCC1, IBA57, ISCA2, JAM3, KARS1, L2HGDH, LMNB1, LRPPRC, LYRM7, MARS2, MLC1, MOCS1, MRPL44, MRPS16, MRPS22, MTFMT, MTHFS, NAXD, NAXE, NDUFA1, NDUFA10, NDUFA12, NDUFA2, NDUFA9, NDUFAF2, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB8, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NFU1, NKX6-2, NOTCH3, NT5C2, NUBPL, OCLN, PC, PDHA1, PEX1, PEX10, PEX12, PEX13, PEX5, PLAA, PLEKHG2, PLP1, POLR1A, POLR1C, POLR3A, POLR3B, PSAP, PYCR2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, RNF220, SAMHD1, SCO1, SDHAF1, SERAC1, SLC16A2, SLC17A5, SLC19A3, SLC1A4, SLC6A9, SNORD118, SOX10, SPART, STN1, SUMF1, SURF1, TMEM106B, TMEM63A, TREX1, TTC19, TUBB4A, TUFM, TWNK, UFM1, VPS11, YY1, ZFYVE26

CentoPediatric Neuro – NDD and ASD | 2,016 genes

CentoPediatric Neuro – NDD and ASD targets genes associated with neurodevelopmental delay and autism spectrum disorders. This panel is intended for pediatric patients with clinical features suggestive of NDD and ASD and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AAAS, AARS1, AASS, ABAT, ABCA2, ABCC9, ABCD1, ABCD4, ABHD16A, ABHD5, ACACA, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD6, ACER3, ACO2, ACOX1, ACOX2, ACP5, ACSL4, ACTB, ACTG1, ACTL6A, ACTL6B, ACVR1, ACY1, ADAM22, ADAMTS10, ADAR, ADARB1, ADAT3, ADCY5, ADD3, ADGRA2, ADGRG1, ADGRL1, ADH5, ADK, ADNP, ADPRS, ADSL, AFF2, AFF3, AFF4, AFG2A, AFG2B, AFG3L2, AGA, AGAP1, AGMO, AGO1, AGO2, AGPAT3, AGTPBP1, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, AKT3, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALMS1, ALX3, ALX4, AMER1, AMMECR1, AMPD2, AMT, ANAPC1, ANAPC7, ANK2, ANK3, ANKLE2, ANKRD11, ANKRD17, ANKS1B, ANO10, ANO4, ANTXR1, AP1B1, AP1G1, AP1S1, AP1S2, AP2M1, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, APC2, ARCN1, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGAP35, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL6, ARMC9, ARPC4, ARSA, ARSL, ARV1, ARX, ASAH1, ASCC3, ASCL1, ASH1L, ASL, ASNS, ASPA, ASPM, ASS1, ASTN1, ASXL1, ASXL2, ASXL3, ATAD1, ATAD3A, ATCAY, ATG4D, ATG7, ATIC, ATL1, ATM, ATN1, ATP11A, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A2, ATP2B1, ATP2B3, ATP5F1E, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP6V1B2, ATP7A, ATP8A2, ATP9A, ATR, ATRX, ATXN2L, ATXN7L3, AUH, AUTS2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BAP1, BAZ2B, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCL11A, BCL11B, BCOR, BCORL1, BCS1L, BICD2, BICRA, BIN1, BLM, BLOC1S1, BLTP1, BMP4, BMPER, BOLA3, BORCS5, BORCS8, BPTF, BRAF, BRAT1, BRCA1, BRCA2, BRD4, BRF1, BRPF1, BRSK2, BRWD3, BSCL2, BSND, BTD, BUB1, BUB1B, C12orf57, C19orf12, C2CD3, C2orf69, CA2, CA5A, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1I, CACNA2D2, CAD, CAMK2A, CAMK2B, CAMK2D, CAMSAP1, CAMTA1, CANT1, CAPN15, CAPRIN1, CAPZA2, CARS1, CARS2, CASK, CASP2, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC186, CCDC32, CCDC47, CCDC82, CCDC88A, CCDC88C, CCND2, CDC42, CDC42BPB, CDH11, CDH2, CDK13, CDK19, CDK5RAP2, CDK8, CDK9, CDKL5, CDKN1C, CDON, CELF2, CELF4, CENPF, CEP104, CEP120, CEP135, CEP152, CEP19, CEP290, CEP295, CEP41, CEP55, CEP57, CEP63, CEP83, CEP85L, CERS1, CERT1, CFAP418, CHAMP1, CHD1, CHD2, CHD3, CHD4, CHD5, CHD7, CHD8, CHKA, CHKB, CHMP1A, CHST14, CHSY1, CIC, CIT, CKAP2L, CLCN2, CLCN3, CLCN4, CLCN6, CLCNKA, CLCNKB, CLDN11, CLIC2, CLN3, CLN5, CLN6, CLN8, CLPB, CLTC, CNKSR2, CNNM2, CNOT1, CNOT2, CNOT3, CNPY3, CNTNAP1, CNTNAP2, COA3, COA7, COA8, COASY, COG1, COG3, COG4, COG5, COG6, COG7, COG8, COL18A1, COL3A1, COL4A1, COL4A2, COLEC10, COLEC11, COLGALT1, COPB2, COQ2, COQ4, COQ8A, COQ9, CORO1A, COX10, COX15, COX20, COX4I2, COX6A2, COX6B1, COX7B, CPAP, CPE, CPLANE1, CPLX1, CPS1, CPSF3, CPT2, CRADD, CRB2, CRBN, CREBBP, CRELD1, CRLF1, CRPPA, CSDE1, CSF1R, CSNK2A1, CSNK2B, CSPP1, CSTB, CTBP1, CTC1, CTCF, CTDP1, CTNNA2, CTNNB1, CTNND2, CTR9, CTSA, CTSD, CTU2, CUL3, CUL4B, CUX1, CUX2, CWC27, CWF19L1, CYB5R3, CYC1, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DAGLA, DALRD3, DARS1, DARS2, DBT, DCAF17, DCC, DCHS1, DCPS, DCX, DDB1, DDC, DDHD2, DDOST, DDX11, DDX17, DDX23, DDX3X, DDX59, DDX6, DEAF1, DEGS1, DENND5A, DENND5B, DEPDC5, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHRSX, DHTKD1, DHX30, DHX37, DHX9, DIAPH1, DIP2C, DIS3L2, DKC1, DLAT, DLD, DLG3, DLG4, DLL1, DMBX1, DMD, DMXL2, DNA2, DNAJB4, DNAJC12, DNAJC17, DNAJC19, DNAJC3, DNM1, DNM1L, DNMT3A, DNMT3B, DOCK3, DOCK4, DOCK6, DOCK7, DOHH, DOLK, DONSON, DPAGT1, DPF2, DPH1, DPH2, DPH5, DPM1, DPM2, DPM3, DPYD, DPYS, DPYSL2, DPYSL5, DRG1, DST, DSTYK, DTYMK, DUOXA2, DVL3, DYM, DYNC1H1, DYNC1I2, DYRK1A, EARS2, EBF3, EBP, ECHS1, ECM1, EDEM3, EED, EEF1A2, EEF1B2, EEF2, EEFSEC, EFNB1, EFTUD2, EHMT1, EIF2AK2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4A2, EIF4A3, EIF5A, ELAC2,

ELOVL4, ELP2, EMC1, EMC10, EMG1, EMILIN1, EML1, ENTPD1, EP300, EPG5, EPHA7, EPRS1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERGIC3, ERI1, ERLIN2, ESAM, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOC2, EXOC7, EXOC8, EXOSC2, EXOSC3, EXOSC5, EXOSC8, EXOSC9, EXT2, EXTL3, EZH1, EZH2, FAM111A, FAM177A1, FAM20C, FAM50A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FAR1, FARS2, FARSA, FARSB, FASTKD2, FAT4, FBN1, FBRSL1, FBXL3, FBXL4, FBXO11, FBXO28, FBXW11, FBXW7, FCSK, FDFT1, FEM1B, FEM1C, FERRY3, FGD1, FGF12, FGF13, FGF14, FGFR1, FGFR2, FGFR3, FH, FIBP, FICD, FIG4, FILIP1, FITM2, FKRP, FKTN, FLNA, FLVCR1, FLVCR2, FMN2, FMR1, FOLR1, FOSL2, FOXG1, FOXP1, FOXP2, FOXP4, FOXR1, FOXRED1, FRA10AC1, FRAS1, FREM2, FRMD5, FRMPD4, FRRS1L, FRYL, FTCD, FTO, FTSJ1, FUCA1, FUT8, FZR1, GABBR1, GABBR2, GABRA1, GABRA2, GABRA3, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALE, GALNT2, GALT, GAMT, GAN, GATA6, GATAD2B, GATM, GBA1, GBA2, GCDH, GCH1, GCSH, GDI1, GEMIN4, GEMIN5, GFAP, GFER, GFM1, GFM2, GIGYF1, GJA1, GJB1, GJC2, GK, GLB1, GLDC, GLE1, GLI2, GLI3, GLIS3, GLRA2, GLS, GLUL, GLYCTK, GM2A, GMNN, GMPPA, GMPPB, GNAI1, GNAO1, GNAS, GNB1, GNB2, GNB5, GNE, GNPAT, GNPTAB, GNPTG, GNS, GON4L, GORAB, GOT2, GPAA1, GPATCH11, GPC3, GPC4, GPHN, GPI, GPSM2, GPT2, GRIA1, GRIA2, GRIA3, GRIA4, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRIP1, GRM1, GRM7, GSS, GSX2, GTF2E2, GTF2H5, GTF3C3, GTF3C5, GTPBP2, GTPBP3, GUCY2D, GUSB, H1-4, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HAAO, HACE1, HADHA, HADHB, HAX1, HCCS, HCFC1, HCN1, HCN2, HDAC3, HDAC4, HDAC8, HEATR3, HEATR5B, HECTD4, HECW2, HELLS, HEPACAM, HERC1, HERC2, HESX1, HEXA, HEXB, HGSNAT, HHAT, HIBCH, HID1, HIKESHI, HIRA, HIVEP2, HK1, HLCS, HMGB1, HMGCL, HNMT, HNRNPD, HNRNPH1, HNRNPH2, HNRNPK, HNRNPR, HNRNPU, HOXA1, HPD, HPDL, HPRT1, HRAS, HS2ST1, HSD17B10, HSD17B4, HSPA9, HSPD1, HSPG2, HTRA2, HUWE1, HYCC1, IARS1, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IFT140, IFT172, IFT27, IFT43, IFT52, IFT56, IFT74, 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RXYLT1, SACS, SALL1, SAMD9, SAMHD1, SARS1, SARS2, SASS6, SATB1, SATB2, SBDS, SBF1, SC5D, SCAF4, SCAMP5, SCAPER, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCO1, SCO2, SCYL1, SDCCAG8, SDHA, SDHAF1, SDHB, SDHD, SEC23A, SEC31A, SELENOI, SEMA6B, SEPHS1, SEPSECS, SERAC1, SET, SETBP1, SETD1A, SETD1B, SETD2, SETD5, SFXN4, SGPL1, SGSH, SGSM3, SHANK1, SHANK2, SHANK3, SHH, SHMT2, SHOC2, SHQ1, SHROOM4, SIAH1, SIK1, SIL1, SIN3A, SIN3B, SIX3, SKI, SKIC3, SLC10A7,

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CentoPediatric Neuro – NDD and ASD Genome | 2,021 genes

CentoPediatric Neuro - NDD and ASD Genome targets genes associated with neurodevelopmental delay and autism spectrum disorders. This panel is intended for pediatric patients with clinical features suggestive of NDD and ASD and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

> This version of the panel is genome sequencing based; it includes the non-coding genes: *CHASERR, RNU4-2, RNU7-1, MIR17HG* and *SNORD118*; repeat expansion screening analysis of *FMR1*; and *GBA1* conversion analysis with its pseudogene.

Genes Included

AAAS, AARS1, AASS, ABAT, ABCA2, ABCC9, ABCD1, ABCD4, ABHD16A, ABHD5, ACACA, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD6, ACER3, ACO2, ACOX1, ACOX2, ACP5, ACSL4, ACTB, ACTG1, ACTL6A, ACTL6B, ACVR1, ACY1, ADAM22, ADAMTS10, ADAR, ADARB1, ADAT3, ADCY5, ADD3, ADGRA2, ADGRG1, ADGRL1, ADH5, ADK, ADNP, ADPRS, ADSL, AFF2, AFF3, AFF4, AFG2A, AFG2B, AFG3L2, AGA, AGAP1, AGMO, AGO1, AGO2, AGPAT3, AGTPBP1, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, AKT3, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALMS1, ALX3, ALX4, AMER1, AMMECR1, AMPD2, AMT, ANAPC1, ANAPC7, ANK2, ANK3, ANKLE2, ANKRD11, ANKRD17, ANKS1B, ANO10, ANO4, ANTXR1, AP1B1, AP1G1, AP1S1, AP1S2, AP2M1, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, APC2, ARCN1, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGAP35, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL6, ARMC9, ARPC4, ARSA, ARSL, ARV1, ARX, ASAH1, ASCC3, ASCL1, ASH1L, ASL, ASNS, ASPA, ASPM, ASS1, ASTN1, ASXL1, ASXL2, ASXL3, ATAD1, ATAD3A, ATCAY, ATG4D, ATG7, ATIC, ATL1, ATM, ATN1, ATP11A, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A2, ATP2B1, ATP2B3, ATP5F1E, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP6V1B2, ATP7A, ATP8A2, ATP9A, ATR, ATRX, ATXN2L, ATXN7L3, AUH, AUTS2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BAP1, BAZ2B, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCL11A, BCL11B, BCOR, BCORL1, BCS1L, BICD2, BICRA, BIN1, BLM, BLOC1S1, BLTP1, BMP4, BMPER, BOLA3, BORCS5, BORCS8, BPTF, BRAF, BRAT1, BRCA1, BRCA2, BRD4, BRF1, BRPF1, BRSK2, BRWD3, BSCL2, BSND, BTD, BUB1, BUB1B, C12orf57, C19orf12, C2CD3, C2orf69, CA2, CA5A, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1I, CACNA2D2, CAD, CAMK2A, CAMK2B, CAMK2D, CAMSAP1, CAMTA1, CANT1, CAPN15, CAPRIN1, CAPZA2, CARS1, CARS2, CASK, CASP2, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC186, CCDC32, CCDC47, CCDC82, CCDC88A, CCDC88C, CCND2, CDC42, CDC42BPB, CDH11, CDH2, CDK13, CDK19, CDK5RAP2, CDK8, CDK9, CDKL5, CDKN1C, CDON, CELF2, CELF4, CENPF, CEP104, CEP120, CEP135, CEP152, CEP19, CEP290, CEP295, CEP41, CEP55, CEP57, CEP63, CEP83, CEP85L, CERS1, CERT1, CFAP418, CHAMP1, CHASERR, CHD1, CHD2, CHD3, CHD4, CHD5, CHD7, CHD8, CHKA, CHKB, CHMP1A, CHST14, CHSY1, CIC, CIT, CKAP2L, CLCN2, CLCN3, CLCN4, CLCN6, CLCNKA, CLCNKB, CLDN11, CLIC2, CLN3, CLN5,

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FIG4, FILIP1, FITM2, FKRP, FKTN, FLNA, FLVCR1, FLVCR2, FMN2, FMR1, FOLR1, FOSL2, FOXG1, FOXP1, FOXP2, FOXP4, FOXR1, FOXRED1, FRA10AC1, FRAS1, FREM2, FRMD5, FRMPD4, FRRS1L, FRYL, FTCD, FTO, FTSJ1, FUCA1, FUT8, FZR1, GABBR1, GABBR2, GABRA1, GABRA2, GABRA3, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALE, GALNT2, GALT, GAMT, GAN, GATA6, GATAD2B, GATM, GBA1, GBA2, GCDH, GCH1, GCSH, GDI1, GEMIN4, GEMIN5, GFAP, GFER, GFM1, GFM2, GIGYF1, GJA1, GJB1, GJC2, GK, GLB1, GLDC, GLE1, GLI2, GLI3, GLIS3, GLRA2, GLS, GLUL, GLYCTK, GM2A, GMNN, GMPPA, GMPPB, GNAI1, GNAO1, GNAS, GNB1, GNB2, GNB5, GNE, GNPAT, GNPTAB, GNPTG, GNS, GON4L, GORAB, GOT2, GPAA1, GPATCH11, GPC3, GPC4, GPHN, GPI, GPSM2, GPT2, GRIA1, GRIA2, GRIA3, GRIA4, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRIP1, GRM1, GRM7, GSS, GSX2, GTF2E2, GTF2H5, GTF3C3, GTF3C5, GTPBP2, GTPBP3, GUCY2D, GUSB, H1-4, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HAAO, HACE1, HADHA, HADHB, HAX1, HCCS, HCFC1, HCN1, HCN2, HDAC3, HDAC4, HDAC8, 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MMACHC, MMADHC, MMGT1, MMUT, MN1, MOCS1, MOCS2, MOGS, MORC2, MPC1, MPDU1, MPDZ, MPLKIP, MPV17, MRAS, MRPL3, MRPS2, MRPS22, MRPS34, MSL2, MSL3, MSMO1, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTFMT, MTHFD1, MTHFR, MTHFS, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTO1, MTOR, MTR, MTRFR, MT-RNR1, MT-RNR2, MTRR, MTSS2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MVK, MYCBP2, MYCN, MYH10, MYH3, MYMK, MYO5A, MYO9A, MYRF, MYT1L, NAA10, NAA15, NAA20, NACC1, NADK2, NAE1, NAGA, NAGLU, NAGS, NALCN, NANS, NAPB, NARS1, NARS2, NBEA, NBN, NCAPD2, NCAPD3, NCAPG2, NCDN, NCKAP1, NCOR1, NDE1, NDP, NDST1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFAF1, NDUFAF2, NDUFAF5, NDUFB8, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEDD4L, NEMF, NEU1, NEUROD2, NEUROG1, NEXMIF, NF1, NFASC, NFIA, NFIB, NFIX, NFU1, NGF, NGLY1, NHS, NIPBL, NKAP, NKX2-1, NKX2-5, NLGN3, NLGN4X, NMNAT1, NONO, NOTCH2, NOVA2, NPC1, NPC2, NPHP1, NPHP3, NPRL2, NR2F1, NR2F2, NR4A2, NRAS, NRCAM, NRROS, NRXN1, NRXN2, NSD1, NSD2, NSDHL, NSF, NSMCE3, NSRP1, NSUN2, NT5C2, NTNG2, NTRK1, NTRK2, NUBPL, NUDT2, NUP107, NUP133, NUP188, NUP214, NUP62, NUP85, NUS1, OCLN, OCRL, ODC1, OFD1, OGDHL, OGT, OPA1, OPA3, OPHN1, ORC1, OSGEP, OTC, OTUD5, OTUD6B, OTUD7A, OTX2, OXR1, P4HTM, PABPC1, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PAK3, PALS1, PAM16, PAN2, PANK2, PARN, PARP6, PARS2, PAX1, PAX3, PAX6, PAX8, PBX1, PC, PCBP2, PCCA, PCCB, PCDH12, PCDH19, PCDHGC4, PCGF2, PCNT, PCYT2, PDCD6IP, PDE10A, PDE2A, PDE4D, PDE6D, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDZD8, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PGM2L1, PGM3, PHACTR1, PHF21A, PHF6, PHF8, PHGDH, PHIP, PHKA2, PHKG2, PI4K2A, PI4KA, PIBF1, PIDD1, PIEZO2, PIGA, PIGB, PIGG, PIGK, PIGL, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGV, PIGY, PIK3C2A, PIK3R2, PIP5K1C, PISD, PITRM1, PITX3, PJA1, PLA2G6, PLAA, PLAAT3, PLCB1, PLCH1, PLEKHG2, PLK1, PLK4, PLP1, PLPBP, PLXNA1, PLXNA2, PLXNB2, PMM2, PMPCA, PMPCB, PNKP, PNP, PNPLA6, PNPLA8, PNPO, PNPT1, POGZ, POLA1, POLG, POLG2, POLR1C, POLR1D, POLR2A, POLR3A, POLR3B, POLRMT, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POP1, PORCN, POT1, POU1F1, POU3F2, POU3F3, POU4F1, PPFIA3, PPFIBP1, PPIL1, PPM1D, PPOX, PPP1CB, PPP1R12A,

PPP1R15B, PPP1R21, PPP1R3F, PPP2CA, PPP2R1A, PPP2R3C, PPP2R5C, PPP2R5D, PPP3CA, PPT1, PQBP1, PRDM13, PRDX1, PRICKLE2, PRIM1, PRKACB, PRKAR1A, PRKAR1B, PRKD1, PRKDC, PRMT7, PRODH, PRORP, PRPS1, PRR12, PRSS12, PRUNE1, PSAP, PSMB8, PSMC3, PSMC5, PSMD12, PSPH, PTCH1, PTCHD1, PTDSS1, PTEN, PTF1A, PTPA, PTPN11, PTPN23, PTPN4, PTRH2, PTRHD1, PTS, PUF60, PUM1, PURA, PUS1, PUS3, PUS7, PYCR1, PYCR2, QARS1, QDPR, QRICH1, RAB11A, RAB11B, RAB14, RAB18, RAB23, RAB39B, RAB3GAP1, RAB3GAP2, RAC1, RAC3, RAD21, RAD50, RAD51, RAF1, RAI1, RALA, RALGAPA1, RAP1B, RAP1GDS1, RARB, RARS1, RARS2, RAX, RBBP5, RBBP8, RBFOX1, RBL2, RBM10, RBM28, RBPJ, RBSN, RECQL4, RELN, RERE, RFT1, RFX3, RFX4, RFX7, RHOBTB2, RIC1, RIN2, RIT1, RLIM, RMND1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF125, RNF13, RNF168, RNF2, RNF220, RNPC3, RNU4-2, RNU4ATAC, RNU7-1, ROBO1, ROGDI, ROR2, RORA, RPGRIP1L, RPIA, RPL10, RPS19, RPS23, RPS28, RPS6KA3, RRM2B, RSPRY1, RSRC1, RTEL1, RTN4IP1, RTTN, RUBCN, RUSC2, RXYLT1, SACS, SALL1, SAMD9, SAMHD1, SARS1, SARS2, SASS6, SATB1, SATB2, SBDS, SBF1, SC5D, SCAF4, SCAMP5, SCAPER, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCO1, SCO2, SCYL1, SDCCAG8, SDHA, SDHAF1, SDHB, SDHD, SEC23A, SEC31A, SELENOI, SEMA6B, SEPHS1, SEPSECS, SERAC1, SET, SETBP1, SETD1A, SETD1B, SETD2, SETD5, SFXN4, SGPL1, SGSH, SGSM3, SHANK1, SHANK2, SHANK3, SHH, SHMT2, SHOC2, SHQ1, SHROOM4, SIAH1, SIK1, SIL1, SIN3A, SIN3B, SIX3, SKI, SKIC3, SLC10A7, SLC12A1, SLC12A2, SLC12A5, SLC12A6, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A4, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC25A26, SLC25A42, SLC25A46, SLC2A1, SLC2A10, SLC2A2, SLC30A9, SLC31A1, SLC32A1, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35B2, SLC35C1, SLC38A3, SLC39A14, SLC39A8, SLC45A1, SLC46A1, SLC4A10, SLC4A4, SLC5A5, SLC5A6, SLC5A7, SLC6A1, SLC6A17, SLC6A19, SLC6A3, SLC6A8, SLC6A9, SLC7A7, SLC9A1, SLC9A6, SLC9A7, SLX4, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCA5, SMARCB1, SMARCC2, SMARCD1, SMARCD2, SMARCE1, SMC1A, SMC3, SMG8, SMG9, SMOC1, SMPD1, SMPD4, SMS, SNAP25, SNAP29, SNF8, SNIP1, SNORD118, SNRPB, SNX14, SNX27, SON, SOS1, SOS2, SOX10, SOX11, SOX2, SOX3, SOX4, SOX5, SOX6, SPART, SPECC1L, SPEN, SPG11, SPOP, SPOUT1, SPR, SPRED1, SPRED2, SPTAN1, SPTBN1, SPTBN2, SPTBN4, SRCAP, SRD5A3, SRP54, SRPK3, SRRM2, SRSF1, SSR4, ST3GAL3, ST3GAL5, STAC3, STAG1, STAG2, STAMBP, STEEP1, STIL, STRA6, STRADA, STT3A, STX1A, STX1B, STX3, STXBP1, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SUPT16H, SUPV3L1, SURF1, SUZ12, SVBP, SYN1, SYNCRIP, SYNE1, SYNGAP1, SYNJ1, SYP, SYT1, SZT2, TAC3, TACO1, TAF1, TAF13, TAF1C, TAF2, TAF4, TAF6, TAF8, TANC2, TANGO2, TAOK1, TAOK2, TARS2, TASP1, TAT, TBC1D20, TBC1D23, TBC1D24, TBC1D2B, TBC1D7, TBCD, TBCE, TBCK, TBL1XR1, TBR1, TBX1, TBX2, TCEAL1, TCF12, TCF20, TCF4, TCF7L2, TCN2, TCTN1, TCTN2, TCTN3, TDP2, TECPR2, TEFM, TELO2, TENM3, TERT, TET3, TFAP2A, TFE3, TG, TGFB1, TGFBR1, TGFBR2, TGIF1, TH, THG1L, THOC2, THOC6, THRA, THUMPD1, TIAM1, TIMM50, TIMM8A, TINF2, TKFC, TKT, TLK2, TMCO1, TMEM106B, TMEM107, TMEM138, TMEM147, TMEM163, TMEM165, TMEM216, TMEM218, TMEM222, TMEM231, TMEM237, TMEM240, TMEM63B, TMEM63C, TMEM67, TMEM70, TMEM94, TMTC3, TMX2, TNIK, TNPO2, TNR, TNRC6B, TOE1, TOGARAM1, TOMM70, TONSL, TOP2B, TOR1A, TP53RK, TP73, TPO, TPP1, TPP2, TPRKB, TRA2B, TRAF7, TRAIP, TRAK1, TRAPPC10, TRAPPC11, TRAPPC12, TRAPPC4, TRAPPC6B, TRAPPC9, TREX1, TRIM8, TRIO, TRIP12, TRIP4, TRIT1, TRMT1, TRMT10A, TRMT5, TRNT1, TRPC5, TRPM3, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TSPOAP1, TTC19, TTC5, TTC8, TTI1, TTI2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP4, TUBGCP6, TUSC3, TWIST1, TWIST2, TWNK, U2AF2, UBA2, UBA5, UBAP2L, UBE2A, UBE3A, UBE3B, UBE3C, UBE4A, UBR1, UBR7, UBTF, UFC1, UFM1, UFSP2, UGDH, UGP2, UMPS, UNC45A, UNC80, UPB1, UPF1, UPF3B, UQCC2, UQCRC2, UQCRQ, USP27X, USP7, USP9X, VAMP2, VARS1, VARS2, VIPAS39, VLDLR, VMA12, VMA22, VPS11, VPS13B, VPS13D, VPS16, VPS33A, VPS33B, VPS35, VPS35L, VPS37A, VPS41, VPS4A, VPS50, VPS51, VPS53, VRK1, WAC, WARS1, WARS2, WASF1, WASHC4, WASHC5, WBP11, WBP4, WDFY3, WDPCP, WDR1, WDR11, WDR26, WDR37, WDR4, WDR45, WDR45B, WDR47, WDR5, WDR62, WDR73, WDR81, WDR83OS, WFS1, WIPI2, WLS, WNK3, WNT1, WNT5A, WWOX, XPA, XPNPEP3, XRCC4, XYLT1, YAP1, YARS1, YIF1B, YIPF5, YWHAG, YY1, YY1AP1, ZBTB11, ZBTB18, ZBTB20, ZBTB24, ZBTB47, ZBTB7A, ZC3H14, ZC4H2, ZDHHC9, ZEB2, ZFHX3, ZFHX4, ZFX, ZFYVE26, ZIC1, ZIC2, ZMIZ1, ZMYM2, ZMYM3, ZMYND11, ZMYND8, ZNF142, ZNF148, ZNF292, ZNF335, ZNF341, ZNF407, ZNF423, ZNF462, ZNF526, ZNF668, ZNF699, ZNF711, ZNFX1, ZNHIT3, ZNRF3, ZPR1, ZSCAN10, ZSWIM6

CentoPediatric Neuro – Neuromuscular | 347 genes

CentoPediatric Neuro – Neuromuscular targets genes associated with neuromuscular disorders. This panel is intended for pediatric patients with clinical features suggestive of neuromuscular disorders and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AARS1, ABHD5, ACAD9, ACADM, ACADVL, ACTA1, ACTG2, ACTN2, ADGRG6, ADSS1, AGL, AGRN, AHCY, AIFM1, ALDOA, ALG14, ALG2, ALS2, ANO5, AR, ASAH1, ASCC1, ASCC3, ATAD1, ATL1, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BET1, BICD2, BIN1, BSCL2, CACNA1A, CACNA1S, CAPN3, CAV1, CAV3, CAVIN1, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCN1, CNTN1, CNTNAP1, COL12A1, COL13A1, COL25A1, COL4A1, COL6A1, COL6A2, COL6A3, COL9A3, COLQ, COQ2, COQ4, COQ8A, COX6A1, COX6A2, CRLF1, CRPPA, CRYAB, CTDP1, DAG1, DES, DHCR24, DHTKD1, DHX16, DMD, DMPK, DNA2, DNAJB2, DNAJB4, DNAJB6, DNM2, DNMT1, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DTNA, DYNC1H1, DYSF, ECEL1, EGR2, ELP1, EMD, EPG5, ERCC5, ERCC6, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FAM111B, FBXO38, FDX2, FGD4, FHL1, FIG4, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GAN, GARS1, GBA1, GBE1, GDAP1, GFER, GFPT1, GGPS1, GJB1, GLDN, GLE1, GLRA1, GLRB, GMPPB, GNB4, GNE, GOLGA2, GOSR2, GYG1, GYS1, HACD1, HADHA, HADHB, HINT1, HK1, HMGCR, HNRNPA2B1, HNRNPDL, HSPB1, HSPB8, HSPG2, HTRA2, IGHMBP2, INF2, INPP5K, ISCU, ITGA7, JAG2, KAT6B, KBTBD13, KCNA1, KCNJ2, KIF1A, KIF5A, KLHL40, KLHL41, KLHL7, KY, LAMA2, LAMA5, LAMB2, LAMP2, LARGE1, LDHA, LETM1, LGI4, LITAF, LMNA, LMOD3, LPIN1, LRP4, LRSAM1, MAGEL2, MAP3K20, MARS1, MEGF10, MFN2,

MICU1, MLIP, MOCS1, MPV17, MPZ, MSTO1, MT-CO1, MT-CO2, MTM1, MTMR14, MTMR2, MT-TL1, MUSK, MYBPC1, MYBPC3, MYH14, MYH2, MYH3, MYH7, MYH8, MYL2, MYMK, MYO18B, MYO9A, MYOD1, MYPN, NALCN, NDRG1, NEB, NEFL, NTRK1, OBSCN, OPA1, OPA3, ORAI1, PAX7, PDK3, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PIP5K1C, PLEC, PLEKHG5, PLOD2, PMM2, PMP22, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC1, POPDC3, PREPL, PRKAG2, PRPS1, PRX, PUS1, PYGM, PYROXD1, RAB7A, RAPSN, RBCK1, REEP1, RETREG1, RFC4, RRM2B, RXYLT1, RYR1, SBF1, SBF2, SCN4A, SELENON, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC16A1, SLC18A3, SLC22A5, SLC25A1, SLC25A20, SLC25A4, SLC25A42, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SLC6A5, SMCHD1, SMN1, SMPD4, SNAP25, SNUPN, SPEG, SPG11, SPTBN4, SPTLC1, SPTLC2, SRPK3, STAC3, STIM1, SUCLA2, SVIL, SYNE1, SYT2, TAFAZZIN, TAMM41, TANGO2, TBCK, TCAP, TFG, TK2, TNNC2, TNNI1, TNNI2, TNNT1, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRDN, TRIM2, TRIM32, TRIP4, TRPV4, TSEN2, TSEN54, TSFM, TTN, TWNK, TYMP, UBA1, UNC13A, UNC45B, VAMP1, VCP, VIPAS39, VMA21, VPS33B, VRK1, VWA1, WNK1, YARS1, YARS2, ZC4H2

CentoPediatric Neuro – Neuromuscular Genome | 347 genes

CentoPediatric Neuro - Neuromuscular Genome targets genes associated with neuromuscular disorders. This panel is intended for pediatric patients with clinical features suggestive of neuromuscular disorders and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

> This version of the panel is based on genome sequencing; it includes repeat expansion screening of *DMPK*; *SMN1* CNV analysis; and *GBA1* conversion analysis with its pseudogene.

Genes Included

AARS1, ABHD5, ACAD9, ACADM, ACADVL, ACTA1, ACTG2, ACTN2, ADGRG6, ADSS1, AGL, AGRN, AHCY, AIFM1, ALDOA, ALG14, ALG2, ALS2, ANO5, AR, ASAH1, ASCC1, ASCC3, ATAD1, ATL1, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BET1, BICD2, BIN1, BSCL2, CACNA1A, CACNA1S, CAPN3, CAV1, CAV3, CAVIN1, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCN1, CNTN1, CNTNAP1, COL12A1, COL13A1, COL25A1, COL4A1, COL6A1, COL6A2, COL6A3, COL9A3, COLQ, COQ2, COQ4, COQ8A, COX6A1, COX6A2, CRLF1, CRPPA, CRYAB, CTDP1, DAG1, DES, DHCR24, DHTKD1, DHX16, DMD, DMPK, DNA2, DNAJB2, DNAJB4, DNAJB6, DNM2, DNMT1, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DTNA, DYNC1H1, DYSF, ECEL1, EGR2, ELP1, EMD, EPG5, ERCC5, ERCC6, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FAM111B, FBXO38, FDX2, FGD4, FHL1, FIG4, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GAN, GARS1, GBA1, GBE1, GDAP1, GFER, GFPT1, GGPS1, GJB1, GLDN, GLE1, GLRA1, GLRB, GMPPB, GNB4, GNE, GOLGA2, GOSR2, GYG1, GYS1, HACD1, HADHA, HADHB, HINT1, HK1, HMGCR, HNRNPA2B1, HNRNPDL, HSPB1, HSPB8, HSPG2, HTRA2, IGHMBP2, INF2, INPP5K, ISCU, ITGA7, JAG2, KAT6B, KBTBD13, KCNA1, KCNJ2, KIF1A, KIF5A, KLHL40, KLHL41, KLHL7, KY, LAMA2, LAMA5, LAMB2, LAMP2, LARGE1, LDHA, LETM1, LGI4, LITAF, LMNA, LMOD3, LPIN1, LRP4, LRSAM1, MAGEL2, MAP3K20, MARS1, MEGF10, MFN2, MICU1, MLIP, MOCS1, MPV17, MPZ, MSTO1, MT-CO1, MT-CO2, MTM1, MTMR14, MTMR2, MT-TL1, MUSK, MYBPC1, MYBPC3, MYH14, MYH2, MYH3, MYH7, MYH8, MYL2, MYMK, MYO18B, MYO9A, MYOD1, MYPN, NALCN, NDRG1, NEB, NEFL, NTRK1, OBSCN, OPA1, OPA3, ORAI1, PAX7, PDK3, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PIP5K1C, PLEC, PLEKHG5, PLOD2, PMM2, PMP22, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC1, POPDC3, PREPL, PRKAG2, PRPS1, PRX, PUS1, PYGM, PYROXD1, RAB7A, RAPSN, RBCK1, REEP1, RETREG1, RFC4, RRM2B, RXYLT1, RYR1, SBF1, SBF2, SCN4A, SELENON, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC16A1, SLC18A3, SLC22A5, SLC25A1, SLC25A20, SLC25A4, SLC25A42, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SLC6A5, SMCHD1, SMN1, SMPD4, SNAP25, SNUPN, SPEG, SPG11, SPTBN4, SPTLC1, SPTLC2, SRPK3, STAC3, STIM1, SUCLA2, SVIL, SYNE1, SYT2, TAFAZZIN, TAMM41, TANGO2, TBCK, TCAP, TFG, TK2, TNNC2, TNNI1, TNNI2, TNNT1, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRDN, TRIM2, TRIM32, TRIP4, TRPV4, TSEN2, TSEN54, TSFM, TTN, TWNK, TYMP, UBA1, UNC13A, UNC45B, VAMP1, VCP, VIPAS39, VMA21, VPS33B, VRK1, VWA1, WNK1, YARS1, YARS2, ZC4H2

CentoPediatric Neuro – Spastic paraplegia | 136 genes

CentoPediatric Neuro – Spastic paraplegia targets genes associated with spastic paraplegia. This panel is intended for pediatric patients with clinical features suggestive of spastic paraplegia and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCD1, ABHD16A, ACBD6, ACER3, ADAR, AFG2B, AFG3L2, AIMP1, ALDH18A1, ALDH3A2, ALS2, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, ARG1, ARL6IP1, ATAD3A, ATL1, ATP13A2, ATXN10, B4GALNT1, BCAS3, BLOC1S1, BORCS8, BSCL2, C19ORF12, CAPN1, CCDC82, CLDN11, CTNNB1, CYP2U1, CYP7B1, DARS1, DDHD1, DDHD2, DDX3X, DSTYK, ELOVL1, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAR1, FARS2, FICD, FXN, GAD1, GALC, GBA2, GCH1, GJA1, GJC2, GLRX5, GPT2, HACE1, HECTD4, HIKESHI, HMBS, HPDL, HSPD1, IBA57, IFIH1, KCNA2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KLC2, KPNA3, L1CAM, LETM1, LYST, MAG, MAPK8IP3, MARS1, MARS2, MTPAP, MTRFR, NDUFA12, NIPA1, NKX6-2, NRCAM, NSRP1, NT5C2, OPA3, PCDH12, PCYT2, PGAP1, PI4KA, PLP1, PNPLA6, POLR3A, PPFIBP1, PPP2R2B, RAB3GAP2, REEP1, REEP2, RETREG1, RHOB, RINT1, RNASEH2B, RNF170, RTN2, SACS, SERAC1, SLC16A2, SLC1A4, SLC25A15, SLC25A46, SLC2A1, SLC33A1, SPART, SPAST, SPG11, SPG7, SPTAN1, SPTSSA, STN1, TAF8, TECPR2, TFG, TMEM63C, TUBB4A, UBAP1, UCHL1, VAMP1, VPS37A, WASHC5, WDR45B, ZEB2, ZFYVE26

CentoPediatric Neuro – Spastic paraplegia Genome | 137 genes

CentoPediatric Neuro – Spastic paraplegia Genome targets genes associated with spastic paraplegia. This panel is intended for pediatric patients with clinical features suggestive of spastic paraplegia and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

> This version of the panel is based on genome sequencing, it includes the non-coding gene *RNU7-1*, and repeat expansion screening of *ATXN10, FXN, PPP2R2B*.

Genes Included

ABCD1, ABHD16A, ACBD6, ACER3, ADAR, AFG2B, AFG3L2, AIMP1, ALDH18A1, ALDH3A2, ALS2, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, ARG1, ARL6IP1, ATAD3A, ATL1, ATP13A2, ATXN10, B4GALNT1, BCAS3, BLOC1S1, BORCS8, BSCL2, C19ORF12, CAPN1, CCDC82, CLDN11, CTNNB1, CYP2U1, CYP7B1, DARS1, DDHD1, DDHD2, DDX3X, DSTYK, ELOVL1, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAR1, FARS2, FICD, FXN, GAD1, GALC, GBA2, GCH1, GJA1, GJC2, GLRX5, GPT2, HACE1, HECTD4, HIKESHI, HMBS, HPDL, HSPD1, IBA57, IFIH1, KCNA2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KLC2, KPNA3, L1CAM, LETM1, LYST, MAG, MAPK8IP3, MARS1, MARS2, MTPAP, MTRFR, NDUFA12, NIPA1, NKX6-2, NRCAM, NSRP1, NT5C2, OPA3, PCDH12, PCYT2, PGAP1, PI4KA, PLP1, PNPLA6, POLR3A, PPFIBP1, PPP2R2B, RAB3GAP2, REEP1, REEP2, RETREG1, RHOB, RINT1, RNASEH2B, RNF170, RNU7-1, RTN2, SACS, SERAC1, SLC16A2, SLC1A4, SLC25A15, SLC25A46, SLC2A1, SLC33A1, SPART, SPAST, SPG11, SPG7, SPTAN1, SPTSSA, STN1, TAF8, TECPR2, TFG, TMEM63C, TUBB4A, UBAP1, UCHL1, VAMP1, VPS37A, WASHC5, WDR45B, ZEB2, ZFYVE26

CentoPediatric Neuro – Structural CNS abnormalities | 18 genes

CentoPediatric Neuro - Structural CNS abnormalities targets genes associated with structural CNS abnormalities. This panel is intended for pediatric patients with clinical features suggestive of structural CNS abnormalities, including lissencephaly and polymicrogyria, among others, and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ANKLE2, ARFGEF2, ARX, ASPM, CCM2, COL18A1, DCX, FLNA, KIF26A, KIFBP, MAN2C1, PAFAH1B1, PAX6, PDCD10, RAB3GAP1, RELN, TUBA1A, VLDLR

Gastrointestinal

CentoPediatric GI Comprehensive | 190 genes

CentoPediatric GI Comprehensive targets genes associated with a broad spectrum of disorders within pediatric gastrointestinal disorders, including conditions such as cholestasis, gastrointestinal atresias, anorectal malformations, congenital diarrhea and infantile enterocolitis. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCB11, ABCB4, ABCC2, ABCG5, ABCG8, ACOX2, ADA, ADAM17, ADAMTS3, ADK, AGR2, AICDA, AKR1D1, ALDOB, ALPI, AMACR, ANGPTL3, ANO1, AP1S1, APOB, ARX, ATP7B, ATP8B1, BAAT, BACH2, BCS1L, BTK, CASK, CC2D2A, CCBE1, CCNQ, CD3G, CD40LG, CD55, CDK9, CDX2, CENPF, CFTR, CHD7, CLDN1, CLMP, COG7, CTLA4, CYBA, CYBB, CYP27A1, CYP7B1, DCDC2, DCLRE1C, DGAT1, DGUOK, DHCR7, DIS3L2, DOCK8, EFTUD2, EGFR, EPCAM, FAH, FANCB, FANCC, FAT4, FERMT1, FLNA, FOXF1, FOXP3, G6PC3, GALE, GALK1, GALM, GALT, GBA1, GBE1, GLI3, GUCY2C, HADHA, HNF1B, HPS1, HPS4, HPS6, HSD3B7, IARS1, ICOS, IKBKG, IL10, IL10RA, IL10RB, IL2RA, IL2RG, INVS, ITGB2, JAG1, KCNMA1, KMT2D, LCT, LIG4, LIPA, LRBA, MED12, MEFV, MID1, MKS1, MNX1, MPI, MPV17, MTTP, MVK, MYCN, MYH14, MYLK, MYO5B, NCF1, NCF2, NEUROG3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR1H4, OTULIN, PCSK1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PI4KA, PIK3CD, PKHD1, PLVAP, POLG, PTF1A, RAG1, RAG2, RECQL4, RFX6, RTEL1, SALL1, SAMD9, SAR1B, SCYL1, SERPINA1, SH2D1A, SI, SKIC2, SKIC3, SLC10A1, SLC10A2, SLC25A13, SLC26A3, SLC37A4, SLC39A4, SLC5A1, SLC9A3, SMPD1, SON, SOX2, SPINK5, SPINT2, STAT1, STAT3, STX3, STXBP2, TALDO1, TERT, TGFBR1, TGFBR2, TJP2, TMEM216, TMPRSS15, TRMU, TTC7A, UGT1A1, VIPAS39, VPS33B, WAS, WNT2B, XIAP, ZIC3, ZNF699

CentoPediatric GI – Cholestasis | 79 genes

CentoPediatric GI – Cholestasis targets genes associated with cholestasis. This panel is intended for pediatric patients with clinical features suggestive of cholestasis and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCB11, ABCB4, ABCC2, ABCG5, ABCG8, ACOX2, ADK, AKR1D1, ALDOB, AMACR, ATP7B, ATP8B1, BAAT, BCS1L, CC2D2A, CFTR, CLDN1, COG7, CYP27A1, CYP7B1, DCDC2, DGUOK, DHCR7, FAH, GALE, GALK1, GALM, GALT, GBA1, GBE1, HADHA, HNF1B, HSD3B7, IARS1, INVS, JAG1, LIPA, MKS1, MPI, MPV17, MVK, MYO5B, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR1H4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PKHD1, POLG, SCYL1, SERPINA1, SKIC3, SLC10A1, SLC10A2, SLC25A13, SMPD1, TALDO1, TJP2, TMEM216, TRMU, UGT1A1, VIPAS39, VPS33B

CentoPediatric GI – Congenital diarrhea, infantile enterocolitis | 69 genes

CentoPediatric GI – Congenital diarrhea, infantile enterocolitis targets genes associated with congenital diarrhea and infantile enterocolitis. This panel is intended for pediatric patients with clinical features suggestive of congenital diarrhea and infantile enterocolitis and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ADA, ADAM17, ADAMTS3, AGR2, AICDA, ALPI, ANGPTL3, ANO1, AP1S1, APOB, ARX, BACH2, BTK, CCBE1, CD3G, CD40LG, CD55, CLMP, CTLA4, CYBA, CYBB, CYP27A1, DCLRE1C, DGAT1, DOCK8, EGFR, EPCAM, FAT4, FERMT1, FLNA, FOXP3, G6PC3, GUCY2C, HPS1, HPS4, HPS6, ICOS, IKBKG, IL10, IL10RA, IL10RB, IL2RA, IL2RG, ITGB2, KMT2D, LCT, LIG4, LIPA, LRBA, MEFV, MTTP, MVK, MYO5B, NCF1, NCF2, NEUROG3, OTULIN, PCSK1,

PIK3CD, PLVAP, RAG1, RAG2, RFX6, RTEL1, SAMD9, SAR1B, SH2D1A, SI, SKIC2, SKIC3, SLC10A2, SLC26A3, SLC37A4, SLC39A4, SLC5A1, SLC9A3, SPINT2, STAT1, STAT3, STX3, STXBP2, TERT, TGFBR1, TGFBR2, TMPRSS15, TTC7A, WAS, WNT2B, XIAP

CentoPediatric GI – Gastrointestinal atresias, anorectal malformations | 33 genes

CentoPediatric GI – Gastrointestinal atresias, anorectal malformations targets genes associated with gastrointestinal atresias and anorectal malformations. This panel is intended for pediatric patients with clinical features suggestive of gastrointestinal atresias and anorectal malformations and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

CASK, CCNQ, CDK9, CDX2, CENPF, CHD7, CLMP, DHCR7, DIS3L2, EFTUD2, FANCB, FANCC, FOXF1, GLI3, KCNMA1, MED12, MID1, MNX1, MYCN, MYH14, MYLK, PI4KA, PTF1A, RECQL4, RFX6, SALL1, SON, SOX2, SPINK5, SPINT2, TTC7A, ZIC3, ZNF699

Dermatology

CentoPediatric Derma Comprehensive | 295 genes

CentoPediatric Derma Comprehensive targets genes associated with a broad spectrum of disorders within pediatric dermatology, including conditions such as ichthyosis, ectodermal dysplasia, epidermolysis bullosa, xeroderma pigmentosum, oculocutaneous albinism/hypopigmentation disorders and other pigmentary skin disorders. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

A2ML1, AAGAB, ABCA12, ABCB6, ABCD4, ABHD5, ADAM10, ADAR, AIRE, ALDH3A2, ALOX12B, ALOXE3, ANAPC1, AP1S1, AP3B1, AP3D1, APCDD1, AQP5, ARSL, ASPRV1, ATP2A2, AXIN2, BAP1, BCS1L, BLOC1S3, BLOC1S5, BLOC1S6, BNC2, BRAF, BRCA1, BRCA2, BRIP1, C2, C3orf52, C5, CACNA1F, CARD14, CBL, CD151, CDH3, CDK4, CDKN2A, CDSN, CIB1, CLCN7, CLDN1, COL17A1, COL7A1, CORO1A, COX7B, CSTA, CYP4F22, DCT, DDB2, DDX3X, DES, DKC1, DSC2, DSC3, DSG1, DSG2, DSG3, DSG4, DSP, DST, DTNBP1, EBP, EDA, EDAR, EDARADD, EDN3, EDNRB, EGFR, ELOVL1, ELOVL4, ENPP1, EPG5, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, EVC, EVC2, EXPH5, FAM111B, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FERMT1, FLG, FLG2, FLNA, GJA1, GJB2, GJB3, GJB4, GJB6, GNA11, GNAI3, GNAQ, GNAS, GPNMB, GPR143, GRHL2, GTF2E2, GTF2H5, HCCS, HOXC13, HPS1, HPS3, HPS4, HPS5, HPS6, HR, HRAS, IFT122, IFT43, IKBKG, IRF4, ITGA3, ITGA6, ITGB4, JUP, KANK2, KDSR, KIT, KITLG, KLHL24, KRAS, KREMEN1, KRT1, KRT10, KRT14, KRT16, KRT17, KRT2, KRT25, KRT5, KRT6A, KRT6B, KRT6C, KRT71, KRT74, KRT81, KRT83, KRT85, KRT9, LAMA3, LAMB3, LAMC2, LEF1, LIPH, LORICRIN, LPAR6, LRMDA, LRP6, LSS, LTV1, LYST, LZTR1, MAP2K1, MAP2K2, MBTPS2, MC1R, MITF, MLH1, MLPH, MPLKIP, MSH2, MSH6, MSMO1, MSX1, MTOR, MT-TS1, MYO5A, NAXD, NDUFB11, NECTIN1, NECTIN4, NF1, NF2, NFKB2, NFKBIA, NIPAL4, NOP10, NRAS, OCA2, OFD1, OSMR, PALB2, PAX3, PAX9, PERP, PEX7, PHF6, PHYH, PIGL, PIK3CA, PKP1, PLEC, PMS2, PNPLA1, POFUT1, POGLUT1, POLH, POMP, PORCN, PPARG, PPP1CB, PRKAR1A, PRKD1, PSENEN, PTEN, PTPN11, RAB27A, RAD51C, RAF1, RASA2, RECQL4, RHBDF2, RIPK4, RIT1, RMRP, RNF113A, RPL21, RSPO1, RSPO4, SAMD9, SASH1, SDR9C7, SERPINB7, SERPINB8, SHOC2, SKIC2, SLC24A5, SLC27A4, SLC29A3, SLC39A4, SLC39A7, SLC45A2, SLURP1, SLX4, SMARCAD1, SMARCAL1, SNAI2, SNAP29, SNRPE, SOS1, SOS2, SOX10, SOX18, SPINK5, SPRED1, SREBF1, ST14, STK11, STS, SULT2B1, SUMF1, TAT, TERC, TERT, TFE3, TGM1, TGM5, TINF2, TMC6, TMC8, TP63, TRPV3, TSC1, TSC2, TSPEAR, TYR, TYRP1, UBE2T, USB1, USP9X, WDR19, WDR35, WNT10A, WRAP53, XPA, XPC, XRCC2

CentoPediatric Derma – Ectodermal dysplasia | 77 genes

CentoPediatric Derma – Ectodermal dysplasia targets genes associated with ectodermal dysplasia. This panel is intended for pediatric patients with clinical features suggestive of ectodermal dysplasia and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCA12, AIRE, ALOX12B, ALOXE3, ANAPC1, APCDD1, AXIN2, BCS1L, C2, C3orf52, C5, CDH3, CDSN, CLDN1, CYP4F22, DSC3, DSG4, DSP, EDA, EDAR, EDARADD, ERCC2, EVC, EVC2, GJB2, GJB6, GRHL2, HOXC13, HR, IFT122, IFT43, IKBKG, ITGA6, ITGB4, JUP, KREMEN1, KRT14, KRT25, KRT71, KRT74, KRT81, KRT83, KRT85, LEF1, LIPH, LPAR6, LRP6, LSS, MBTPS2, MPLKIP, MSX1, NECTIN1, NECTIN4, NFKB2, NFKBIA, NIPAL4, PAX9, PKP1, PNPLA1, PORCN, PPARG, PRKD1, RIPK4, RMRP, RPL21, RSPO4, SDR9C7, SKIC2, SNRPE, SREBF1, ST14, TGM1, TP63, TSPEAR, WDR19, WDR35, WNT10A

CentoPediatric Derma – Epidermolysis bullosa | 45 genes

CentoPediatric Derma – Epidermolysis bullosa targets genes associated with epidermolysis bullosa. This panel is intended for pediatric patients with clinical features suggestive of epidermolysis bullosa and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ATP2A2, CARD14, CD151, CDSN, CIB1, COL17A1, COL7A1, CORO1A, CSTA, DSC3, DSG1, DSG3, DSP, DST, EDA, EGFR, EXPH5, FERMT1, FLG2, IKBKG, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT1, KRT10, KRT14, KRT2, KRT5, LAMA3, LAMB3, LAMC2, NAXD, PKP1, PLEC, SERPINB8, SLC39A4, SLC39A7, SPINK5, TGM5, TMC6, TMC8, TP63, TRPV3

CentoPediatric Derma – Ichthyosis | 78 genes

CentoPediatric Derma – Ichthyosis targets genes associated with ichthyosis. This panel is intended for pediatric patients with clinical features suggestive of ichthyosis and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AAGAB, ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AQP5, ASPRV1, CARD14, CDSN, CLDN1, CSTA, CYP4F22, DES, DSC2, DSG1, DSG2, DSP, EBP, ELOVL1, ELOVL4, ENPP1, ERCC2, FLG, FLG2, GJA1, GJB2, GJB3, GJB4, GJB6, GTF2E2, JUP, KANK2, KDSR, KRT1, KRT10, KRT14, KRT16, KRT17, KRT2, KRT6A, KRT6B, KRT6C, KRT9, LIPH, LORICRIN, MBTPS2, MPLKIP, MSMO1, MT-TS1, NIPAL4, OSMR, PERP, PEX7, PHYH, PIGL, PNPLA1, POMP, RHBDF2, RSPO1, SASH1, SDR9C7, SERPINB7, SLC27A4, SLURP1, SMARCAD1, SNAP29, SPINK5, SREBF1, ST14, STS, SULT2B1, SUMF1, TAT, TGM1, TGM5, TRPV3

CentoPediatric Derma – Oculocutaneous albinism/hypopigmentation | 36 genes

CentoPediatric Derma – Oculocutaneous albinism/hypopigmentation targets genes associated with oculocutaneous albinism/hypopigmentation. This panel is intended for pediatric patients with clinical features suggestive of oculocutaneous albinism/hypopigmentation and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, CACNA1F, CLCN7, DCT, DTNBP1, EDN3, EDNRB, EPG5, GNAI3, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, KITLG, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SNAI2, SOX10, TYR, TYRP1

CentoPediatric Derma – Pigmentary skin disorders | 140 genes

CentoPediatric Derma – Pigmentary skin disorders targets genes associated with pigmentary skin disorders. This panel is intended for pediatric patients with clinical features suggestive of pigmentary skin disorders, including both hypo- and hyperpigmentation, and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

A2ML1, ABCB6, ABCD4, ADAM10, ADAR, ANAPC1, AP3B1, ARSL, BAP1, BNC2, BRAF, BRCA1, BRCA2, BRIP1, CBL, CDK4, CDKN2A, CIB1, COX7B, DDB2, DDX3X, DKC1, EDN3, EDNRB, ENPP1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111B, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FLNA, GJA1, GJB3, GJB4, GNA11, GNAQ, GNAS, GPNMB, GTF2E2, GTF2H5, HCCS, HPS1, HRAS, IKBKG, IRF4, KIT, KITLG, KRAS, KRT10, KRT14, KRT5, LTV1, LYST, LZTR1, MAP2K1, MAP2K2, MC1R, MITF, MLH1, MLPH, MPLKIP, MSH2, MSH6, MTOR, MYO5A, NDUFB11, NF1, NF2, NOP10, NRAS, OCA2, OFD1, OSMR, PALB2, PAX3, PHF6, PIK3CA, PMS2, POFUT1, POGLUT1, POLH, PORCN, PPP1CB, PRKAR1A, PSENEN, PTEN, PTPN11, RAB27A, RAD51C, RAF1, RASA2, RECQL4, RIT1, RNF113A, SAMD9, SASH1, SHOC2, SKIC2, SLC24A5, SLC29A3, SLC45A2, SLX4, SMARCAL1, SNAI2, SOS1, SOS2, SOX10, SOX18, SPRED1, STK11, TERC, TERT, TFE3, TINF2, TMC6, TMC8, TSC1, TSC2, TYR, TYRP1, UBE2T, USB1, USP9X, WRAP53, XPA, XPC, XRCC2

CentoPediatric Derma – Xeroderma pigmentosum | 15 genes

CentoPediatric Derma - Xeroderma pigmentosum targets genes associated with xeroderma pigmentosum. This panel is intended for pediatric patients with clinical features suggestive of xeroderma pigmentosum and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, GTF2E2, GTF2H5, MPLKIP, POLH, RNF113A, XPA, XPC

Nephrology

CentoPediatric Nephro Comprehensive | 502 genes

CentoPediatric Nephro Comprehensive targets genes associated with a broad spectrum of disorders within pediatric nephrology, including conditions such as polycystic kidney disorders, kidney malformations and the atypical hemolytic uremic syndrome. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCB11, ABCB4, ABCC2, ACE, ACP5, ACTG2, ACTN4, ADAMTS13, AGPS, AGT, AGTR1, AGXT, AHI1, AIPL1, AKR1D1, ALDOB, ALG8, ALG9, ALPL, AMER1, ANKH, ANKS6, ANLN, ANO5, ANOS1, AP2S1, ARHGAP31, ARHGDIA, ARL13B, ARL3, ARL6, ARMC5, ARMC9, ARSL, ATP6V0A4, ATP6V1B1, ATP8B1, ATR, AVPR2, B9D1, B9D2, BAAT, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BICC1, BMP1, BMP4, BMPR1B, BNC2, BSND, C2CD3, C3, CA2, CABP4, CANT1, CASR, CC2D2A, CCDC39, CCDC40, CCN6, CCNO, CCNQ, CD2AP, CD46, CD59, CDKN1C, CENPF, CEP120, CEP152, CEP164, CEP290, CEP41, CEP55, CEP83, CFAP298, CFAP418, CFAP53, CFB, CFH, CFHR5, CFI, CFTR, CHD7, CHRNA3, CHST3, CHSY1, CILK1, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, COL10A1, COL4A1, COL4A3, COL4A4, COL4A5, COL9A3, COMP, COQ2, COQ6, COQ8B, COQ9, CPAP, CPLANE1, CRB1, CRB2, CRELD1, CRTAP, CRX, CSPP1, CTNS, CTU2, CUBN, CUL3, CWC27, CYP7B1, DCDC2, DDR2, DDX59, DGKE, DGUOK, DHCR7, DICER1, DLL3, DMP1, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAJB11, DNAL1, DRC1, DRC2, DSTYK, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DZIP1L, EBP, EIF2AK3, EMP2, ENPP1, ESCO2, EVC, EVC2, EXT1, EXT2, EYA1, FAH, FAM20C, FAN1, FANCB, FAS, FASLG, FAT4, FEZF1, FGF17, FGF23, FGF8, FGFR1, FGFR2, FKBP10, FLNB, FN1, FOXC2, FOXP1, FRAS1, FREM1, FREM2, FSHB, FXYD2, GANAB, GATA3, GDF1, GDF5, GFM1, GHR, GLA, GLI2, GLI3, GLIS2, GLIS3, GNA11, GNAS, GNPAT, GNRH1, GNRHR, GPC3, GPC6, GREB1L, GRIP1, GUCY2D, HAAO, HAMP, HESX1, HEXA, HFE, HNF1B, HNF4A, HOXA13, HOXD13, HPSE2, HSD11B2, HSD3B7, HSPG2, HYDIN, HYLS1, IFITM5, IFT122, IFT140, IFT172, IFT27, IFT43, IFT54, IFT80, IFT81, IHH, IMPDH1, INF2, INPP5E, INPPL1, INVS, IQCB1, ITGA3, ITGA8, JAG1, KANK2, KCNJ1, KCNJ10, KCNJ13, KCNJ5, KDM6A, KIAA0586, KIF14, KIF22, KIF7, KISS1, KISS1R, KLHL3, KMT2D, KYNU, LAGE3, LAMB2, LBR, LCA5, LCAT, LCT, LEP, LEPR, LHB, LHX3, LHX4, LIFR, LMF1, LMX1B, LRAT, LRIG2, LRP4, LRP5, LZTFL1, MAFB, MAGI2, MAPKBP1, MATN3, MCEE, MERTK, MESP2, MGP, MKKS, MKS1, MMAA, MMAB, MMADHC, MMP13, MMP21, MMP9, MMUT, MPV17, MUC1, MYH9, MYO1E, MYO5B, MYO7A, MYOCD, NADSYN1, NBAS, NEK1, NEK8, NEUROG3, NIPBL, NKX2-5, NKX3-2, NMNAT1, NODAL, NOG, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPR2, NR0B1, NR1H4, NR3C2, NSDHL, NUP107, NUP93, OBSL1, OCRL, ODAD1, ODAD2, ODAD3, OFD1, OSGEP, OTX2, P3H1, PAPSS2, PAX2, PBX1, PCSK1, PDE4D, PDE6D, PDSS2, PEX1, PEX10, PEX12, PEX2, PEX26, PEX5, PEX6, PEX7, PHEX, PHF6, PIBF1, PKD1, PKD1L1, PKD2, PKHD1, PLCE1, PLOD2, PMM2, PNPLA6, POLG, POLR3B, POMC, POU1F1, PPARG, PPIB, PRKAR1A, PRKCSH, PROK2, PROKR2, PROM1, PROP1, PRPH2, PTH1R, PTHLH, PTPRO, PUF60, RBBP8, RD3, RDH12, RDH5, REN, RET, RHO, RLBP1, RMND1, RNF216, ROBO1, ROBO2, ROR2, RPE65, RPGRIP1, RPGRIP1L, RRM2B, RSPH1, RSPH4A, RSPH9, RUNX2, SALL1, SALL4, SBDS, SCARB2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SEMA3A, SERPINA1, SERPINF1, SERPINH1, SGPL1, SH3PXD2B, SIX1, SIX2, SKIC3, SLC12A1, SLC12A3, SLC25A13, SLC25A15, SLC26A2, SLC26A3, SLC2A2, SLC34A1, SLC34A3, SLC35D1, SLC4A1, SLC4A4, SLIT2, SMARCAL1, SMPD1, SOX10, SOX11, SOX17, SOX2, SOX3, SOX9, SPAG1, SPATA7, SPINT2, SPRY4, STRA6, SUFU, TAC3, TACR3, TBC1D1, TBX15, TBX18, TBX3, TBX5, TCTN1, TCTN2, TCTN3, TFR2, THBD, TJP2, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNFRSF11B, TP53RK, TPRKB, TRAP1, TRIM32, TRIP11, TRMU, TRPC6, TRPS1, TRPV4, TSC1, TSC2, TTC21B, TTC8, TULP1, TXNDC15, UGT1A1, UMOD, VHL, VIPAS39, VPS33B, WDR11, WDR19, WDR35, WDR4, WDR73, WNK1, WNK4, WNT4, WNT5A, WNT7A, WT1, XPNPEP3, XYLT1, ZIC3, ZMYND10, ZNF423

CentoPediatric Nephro – aHUS | 10 genes

CentoPediatric Nephro – aHUS targets genes associated with the atypical hemolytic uremic syndrome (aHUS). This panel is intended for pediatric patients with clinical features suggestive of atypical hemolytic uremic syndrome (aHUS) and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This panel includes MLPA analysis for aHUS (*CFH, CFHR1, CFHR2, CFHR3, CFHR5*).

Genes Included

ADAMTS13, C3, CD46, CD59, CFB, CFH, CFHR5, CFI, DGKE, THBD

CentoPediatric Nephro – Kidney malformations | 23 genes

CentoPediatric Nephro – Kidney malformations targets genes associated with kidney malformations. This panel is intended for pediatric patients with clinical features suggestive of kidney malformations and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ACE, ACTG2, AGT, BMP4, CCNQ, DSTYK, EYA1, FANCB, FOXC2, FREM1, FREM2, GATA3, GREB1L, HNF1B, KYNU, PAX2, PBX1, PUF60, REN, ROBO2, SALL1, SIX1, WT1

CentoPediatric Nephro – Polycystic kidney | 39 genes

CentoPediatric Nephro - Polycystic kidney targets genes associated with polycystic kidney disorders. This panel is intended for pediatric patients with clinical features suggestive of polycystic kidney disease and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This panel includes Sanger sequencing and MLPA analysis of *PKD1*.

Genes Included

ALG8, ALG9, ANKS6, BICC1, CEP164, CEP290, CEP83, CRB2, DCDC2, DZIP1L, GLIS2, HNF1B, IFT140, IFT172, INVS, IQCB1, JAG1, MAPKBP1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PMM2, RPGRIP1L, SDCCAG8, SEC61A1, TMEM67, TSC1, TSC2, TTC21B, VHL, WDR19, ZNF423

Osteology

CentoPediatric Bone Comprehensive | 608 genes

CentoPediatric Bone Comprehensive targets genes associated with a broad spectrum of disorders within pediatric osteology, including conditions such as skeletal dysplasias, non-syndromic short stature disorders, abnormal mineralization disorders, osteogenesis imperfecta, short-rib/asphyxiating thoracic dysplasia, and craniosynostosis. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCC6, ABCC9, ACAN, ACP5, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL2, ADAMTSL4, AFF3, AFF4, AGA, AGPS, AHDC1, AIFM1, AKT2, ALG12, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANAPC1, ANKH, ANO5, ANTXR2, AP2S1, ARCN1, ARHGAP31, ARID1A, ARID1B, ARSB, ARSK, ARSL, ASAH1, ASXL1, ASXL2, ASXL3, ATP6V0A2, ATP7A, ATR, AXIN1, AXIN2, B3GALT6, B3GAT3, B3GLCT, B4GALT7, BANF1, BCL11B, BCOR, BCS1L, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BNIP1, BPNT2, BRAF, C2CD3, CA2, CANT1, CASR, CBFB, CC2D2A, CCDC8, CCN6, CCNQ, CDC45, CDC73, CDH3, CDK13, CDKN1C, CDT1, CEP120, CEP152, CEP290, CFAP410, CHD5, CHD7, CHST11, CHST14, CHST3, CHSY1, CILK1, CKAP2L, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COMP, COPB2, CREB3L1, CREBBP, CRLF1, CRTAP, CSF1R, CSGALNACT1, CSPP1, CTSA, CTSC, CTSK, CUL7, CWC27, CYP26B1, CYP27B1, CYP2R1, DDR2, DDRGK1, DHCR24, DHCR7, DHODH, DLL3, DLL4, DLX3, DLX5, DMP1, DNA2, DNAJC21, DNMT3A, DOCK6, DONSON, DPH1, DPM1, DSE, DVL1, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EDN1, EDNRA, EED, EFL1, EFNB1, EFTUD2, EIF2AK3, EIF4A3, ELMO2, EN1, ENPP1, EOGT, EP300, ERF, ERI1, ESCO2, EVC, EVC2, EXOC6B, EXT1, EXT2, EXTL3, EZH2, FAH, FAM111A, FAM20C, FANCB, FAR1, FBN1, FBN2, FBXO11, FERMT3, FGD1, FGF10, FGF16, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FKBP14, FLNA, FLNB, FMN1, FN1, FREM1, FUCA1, FZD2, GALNS, GALNT3, GBA1, GCM2, GDF5, GDF6, GH1, GHR, GHRHR, GHSR, GJA1, GLB1, GLI1, GLI2, GLI3, GMNN, GNAI3, GNAS, GNPAT, GNPNAT1, GNPTAB, GNPTG, GNS, GORAB, GPAA1, GPC3, GPC4, GPC6, GPX4, GRK2, GSC, GUSB, GZF1, HAAO, HDAC4, HDAC8, HEATR3, HES7, HESX1, HGSNAT, HHAT, HNRNPK, HOXA13, HOXD13, HPGD, HRAS, HS2ST1, HSPA9, HSPG2, HUWE1, HYAL1, HYLS1, IARS2, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT54, IFT80, IFT81, IGF1, IGF1R, IGF2, IGFALS, IHH, IKBKG, IL11RA, IL1RN, IL6ST, INPPL1, INTU, IRX5, JAG1, KAT6A, KAT6B, KCNJ2, KDELR2, KIAA0586, KIAA0753, KIAA0825, KIF22, KIF5B, KIF7, KMT2D, KRAS, KYNU, LAMA5, LARP7, LBR, LEMD3, LFNG, LHX3, LHX4, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LRRK1, LTBP1, LTBP2, LTBP3, LYSET, MAB21L2, MAFB, MAN2B1, MANBA, MAP2K1, MAP3K20, MAP3K7, MAPKAPK5, MASP1, MATN3, MBTPS1, MBTPS2, MCM7, MECOM, MEGF8, MEOX1, MESD, MESP2, MGP, MID1, MKS1, MMP13, MMP2, MMP9, MNX1, MSX2, MTAP, MTX2, MYCN, MYH3, MYO18B, NADSYN1, NAGLU, NANS, NBAS, NEK1, NEU1, NF1, NFIA, NFIX, NIPBL, NKX3-2, NLRP3, NMNAT1, NOG, NOTCH1, NOTCH2, NPR2, NPR3, NRAS, NRCAM, NSD1, NSDHL, NSMCE2, NT5E, NXN, OBSL1, OCRL, OFD1, ORC1, ORC4, ORC6, OSGEP, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PDGFRB, PEX14, PEX19, PEX5, PEX7, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PISD, PITX1, PJA1, PKDCC, PLCB4, PLEKHM1, PLOD2, PLS3, POC1A, POLE, POLR1A, POLR1B, POLR1C, POLR1D, POLR3A, POLR3B, POLR3GL, POP1, POR, PORCN, POU1F1, PPIB, PPP1CB, PPP3CA, PRKACA, PRKACB, PRKAR1A, PRKG2, PRMT7, PROP1, PRRX1, PSAT1, PSPH, PTCH1, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RAD21, RASGRP2, RBBP8, RBM8A, RBPJ, RECQL4, RIGI, RINT1, RIPPLY2, RMP64, RMRP, RNU4ATAC, ROR2, RPGRIP1L, RPL13, RSPO2, RSPRY1, RUNX2, SALL1, SALL4, SBDS, SC5D, SCARF2, SCUBE3, SEC23A, SEC24D, SERPINF1, SERPINH1, SETBP1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHH, SHOC2, SHOX, SIK3, SIX1, SIX2, SKI, SLC10A7, SLC17A5, SLC25A24, SLC26A2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SLC35B2, SLC35D1, SLC39A13, SLC4A2, SLCO2A1, SMAD2, SMAD3, SMAD4, SMAD6, SMARCA4, SMARCAL1, SMARCB1, SMARCE1, SMC1A, SMC3, SMCHD1, SMO, SMOC1, SMS, SNRPB, SNX10, SOST, SOX3, SOX6, SOX9, SP7, SPARC, SPECC1L, SPRY1, SRCAP, SRP54, STAMBP, STAT3, STAT5B, STT3A, SUCO, SULF1, SUMF1, SUZ12, TAB2, TAPT1, TBC1D24, TBCE, TBX15, TBX2, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCF12, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TFAP2B, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TLK2, TMCO1, TMEM165, TMEM216, TMEM231, TMEM38B, TMEM53, TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TOP2B, TP63, TRAF7, TRAIP, TRAPPC2, TREM2, TRIM37, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TUBGCP6, TWIST1, TXNL4A, TYROBP, UBA2, UFSP2, UNC45A, VDR, VPS33A, VPS35L, WBP11, WDPCP, WDR19, WDR35, WNT1, WNT10B, WNT11, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, ZBTB20, ZEB2, ZIC1, ZIC3, ZMPSTE24, ZNF462, ZNF687, ZSWIM6

CentoPediatric Bone Comprehensive Genome | 609 genes

CentoPediatric Bone Comprehensive Genome targets genes associated with a broad spectrum of disorders within pediatric osteology, including conditions such as skeletal dysplasias, non-syndromic short stature disorders, abnormal mineralization disorders, osteogenesis imperfecta, short-rib/asphyxiating thoracic dysplasia, and craniosynostosis. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

> This version of the panel is based on genome sequencing, it includes the non-coding gene *MIR140*; and *GBA1* conversion analysis with its pseudogene.

Genes Included

ABCC6, ABCC9, ACAN, ACP5, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL2, ADAMTSL4, AFF3, AFF4, AGA, AGPS, AHDC1, AIFM1, AKT2, ALG12, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANAPC1, ANKH, ANO5, ANTXR2, AP2S1, ARCN1, ARHGAP31, ARID1A, ARID1B, ARSB, ARSK, ARSL, ASAH1, ASXL1, ASXL2, ASXL3, ATP6V0A2, ATP7A, ATR, AXIN1, AXIN2, B3GALT6, B3GAT3, B3GLCT, B4GALT7, BANF1, BCL11B, BCOR, BCS1L, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BNIP1, BPNT2, BRAF, C2CD3, CA2, CANT1, CASR, CBFB, CC2D2A, CCDC8, CCN6, CCNQ, CDC45, CDC73, CDH3, CDK13, CDKN1C, CDT1, CEP120, CEP152, CEP290, CFAP410, CHD5, CHD7, CHST11, CHST14, CHST3, CHSY1, CILK1, CKAP2L, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COMP, COPB2, CREB3L1, CREBBP, CRLF1, CRTAP, CSF1R, CSGALNACT1, CSPP1, CTSA, CTSC, CTSK, CUL7, CWC27, CYP26B1, CYP27B1, CYP2R1, DDR2, DDRGK1, DHCR24, DHCR7, DHODH, DLL3, DLL4, DLX3, DLX5, DMP1, DNA2, DNAJC21, DNMT3A, DOCK6, DONSON, DPH1, DPM1, DSE, DVL1, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EDN1, EDNRA, EED, EFL1, EFNB1, EFTUD2, EIF2AK3, EIF4A3, ELMO2, EN1, ENPP1, EOGT, EP300, ERF, ERI1, ESCO2, EVC, EVC2, EXOC6B, EXT1, EXT2, EXTL3, EZH2, FAH, FAM111A, FAM20C, FANCB, FAR1, FBN1, FBN2, FBXO11, FERMT3, FGD1, FGF10, FGF16, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FKBP14, FLNA, FLNB, FMN1, FN1, FREM1, FUCA1, FZD2, GALNS, GALNT3, GBA1, GCM2, GDF5, GDF6, GH1, GHR, GHRHR, GHSR, GJA1, GLB1, GLI1, GLI2, GLI3, GMNN, GNAI3, GNAS, GNPAT, GNPNAT1, GNPTAB, GNPTG, GNS, GORAB, GPAA1, GPC3, GPC4, GPC6, GPX4, GRK2, GSC, GUSB, GZF1, HAAO, HDAC4, HDAC8, HEATR3, HES7, HESX1, HGSNAT, HHAT, HNRNPK, HOXA13, HOXD13, HPGD, HRAS, HS2ST1, HSPA9, HSPG2, HUWE1, HYAL1, HYLS1, IARS2, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT54, IFT80, IFT81, IGF1, IGF1R, IGF2, IGFALS, IHH, IKBKG, IL11RA, IL1RN, IL6ST, INPPL1, INTU, IRX5, JAG1, KAT6A, KAT6B, KCNJ2, KDELR2, KIAA0586, KIAA0753, KIAA0825, KIF22, KIF5B, KIF7, KMT2D, KRAS, KYNU, LAMA5, LARP7, LBR, LEMD3, LFNG, LHX3, LHX4, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LRRK1, LTBP1, LTBP2, LTBP3, LYSET, MAB21L2, MAFB, MAN2B1, MANBA, MAP2K1, MAP3K20, MAP3K7, MAPKAPK5, MASP1, MATN3, MBTPS1, MBTPS2, MCM7, MECOM, MEGF8, MEOX1, MESD, MESP2, MGP, MID1, MIR140, MKS1, MMP13, MMP2, MMP9, MNX1, MSX2, MTAP, MTX2, MYCN, MYH3, MYO18B, NADSYN1, NAGLU, NANS, NBAS, NEK1, NEU1, NF1, NFIA, NFIX, NIPBL, NKX3-2, NLRP3, NMNAT1, NOG, NOTCH1, NOTCH2, NPR2, NPR3, NRAS, NRCAM, NSD1, NSDHL, NSMCE2, NT5E, NXN, OBSL1, OCRL, OFD1, ORC1, ORC4, ORC6, OSGEP, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PDGFRB, PEX14, PEX19, PEX5, PEX7, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PISD, PITX1, PJA1, PKDCC, PLCB4, PLEKHM1, PLOD2, PLS3, POC1A, POLE, POLR1A, POLR1B, POLR1C, POLR1D, POLR3A, POLR3B, POLR3GL, POP1, POR, PORCN, POU1F1, PPIB, PPP1CB, PPP3CA, PRKACA, PRKACB, PRKAR1A, PRKG2, PRMT7, PROP1, PRRX1, PSAT1, PSPH, PTCH1, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RAD21, RASGRP2, RBBP8, RBM8A, RBPJ, RECQL4, RIGI, RINT1, RIPPLY2, RMP64, RMRP, RNU4ATAC, ROR2, RPGRIP1L, RPL13, RSPO2, RSPRY1, RUNX2, SALL1, SALL4, SBDS, SC5D, SCARF2, SCUBE3, SEC23A, SEC24D, SERPINF1, SERPINH1, SETBP1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHH, SHOC2, SHOX, SIK3, SIX1, SIX2, SKI, SLC10A7, SLC17A5, SLC25A24, SLC26A2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SLC35B2, SLC35D1, SLC39A13, SLC4A2, SLCO2A1, SMAD2, SMAD3, SMAD4, SMAD6, SMARCA4, SMARCAL1, SMARCB1, SMARCE1, SMC1A, SMC3, SMCHD1, SMO, SMOC1, SMS, SNRPB, SNX10, SOST, SOX3, SOX6, SOX9, SP7, SPARC, SPECC1L, SPRY1, SRCAP, SRP54, STAMBP, STAT3, STAT5B, STT3A, SUCO, SULF1, SUMF1, SUZ12, TAB2, TAPT1, TBC1D24, TBCE, TBX15, TBX2, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCF12, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TFAP2B, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TLK2, TMCO1, TMEM165, TMEM216, TMEM231, TMEM38B, TMEM53, TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TOP2B, TP63, TRAF7, TRAIP, TRAPPC2, TREM2, TRIM37, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TUBGCP6, TWIST1, TXNL4A, TYROBP, UBA2, UFSP2, UNC45A, VDR, VPS33A, VPS35L, WBP11, WDPCP, WDR19, WDR35, WNT1, WNT10B, WNT11, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, ZBTB20, ZEB2, ZIC1, ZIC3, ZMPSTE24, ZNF462, ZNF687, ZSWIM6

CentoPediatric Bone – Abnormal mineralization | 89 genes

CentoPediatric Bone – Abnormal mineralization targets genes associated with abnormal mineralization. This panel is intended for pediatric patients with clinical features suggestive of abnormal mineralization, including hypo-mineralization and hyper-mineralization, and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCC6, ALPL, AMER1, ANKH, ANO5, AP2S1, ARCN1, B3GALT6, B3GAT3, B4GALT7, BMP1, CA2, CASR, CLCN5, CLCN7, COL1A1, COL1A2, COPB2, CREB3L1, CRTAP, CTSK, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, FKBP10, GALNT3, GJA1, GNAS, GORAB, GPAA1, HPGD, IFITM5, KDELR2, KIF5B, LEMD3, LRP4, LRP5, MBTPS2, MESD, MTAP, NBAS, NOTCH2, OCRL, OSTM1, P3H1, P4HB, PHEX, PLEKHM1, PLOD2, PLS3, PPIB, PTDSS1, PTH1R, SEC24D, SERPINF1, SERPINH1, SGMS2, SH3PXD2B, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SLC4A2, SLCO2A1, SNX10, SOST, SP7, SPARC, SUCO, TAPT1, TBXAS1, TCIRG1, TENT5A, TGFB1, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TRPV6, UNC45A, VDR, WNT1, WNT11, XYLT2, ZBTB20

CentoPediatric Bone – Craniosynostosis | 102 genes

CentoPediatric Bone – Craniosynostosis targets genes associated with craniosynostosis, both syndromic and non-syndromic. This panel is intended for pediatric patients with clinical features suggestive of craniosynostosis and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ADAMTSL4, AHDC1, ALPL, ALX4, ASXL1, ASXL3, AXIN2, B3GAT3, BCL11B, BRAF, CDC45, CDK13, CDT1, CHD5, CHD7, COLEC11, CTSK, CYP26B1, DPH1, EFNB1, ERF, ESCO2, FAM20C, FBN1, FBXO11, FGF10, FGF9, FGFR1, FGFR2, FGFR3, FLNA, FREM1, GLI3, GNAS, GNPTAB, GPC3, HNRNPK, HUWE1, IFT122, IFT140, IFT43, IHH, IL11RA, IL6ST, IRX5, JAG1, KAT6A, KAT6B, KMT2D, KRAS, LTBP1, MAN2B1, MASP1, MEGF8, MSX2, NFIA, ORC1, ORC4, ORC6, P4HB, PHEX, PJA1, POR, PPP1CB, PPP3CA, PRRX1, PTCH1, PTPN11, RAB23, RECQL4, RSPRY1, RUNX2, SCARF2, SEC24D, SHOC2, SIX1, SIX2, SKI, SLC25A24, SMAD2, SMAD3, SMAD6, SOX6, SPECC1L, SPRY1, STAT3, TCF12, TCOF1, TFAP2B, TGFB2, TGFB3, TGFBR1, TGFBR2, TLK2, TMCO1, TRAF7, TWIST1, WDR19, WDR35, ZEB2, ZIC1, ZNF462

CentoPediatric Bone – Non-syndromic short stature | 16 genes

CentoPediatric Bone – Non-syndromic short stature targets genes associated with non-syndromic short stature. This panel is intended for pediatric patients with clinical features suggestive of non-syndromic short stature and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

GH1, GHR, GHRHR, GHSR, HESX1, IGF1, IGF1R, IGF2, IGFALS, LHX3, LHX4, POU1F1, PROP1, SHOX, SOX3, STAT5B

CentoPediatric Bone – Osteogenesis imperfecta | 54 genes

CentoPediatric Bone – Osteogenesis imperfecta targets genes associated with osteogenesis imperfecta. This panel is intended for pediatric patients with clinical features suggestive of osteogenesis imperfecta and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ALPL, ARCN1, B3GALT6, B3GAT3, B4GALT7, BMP1, CASR, CLCN5, COL1A1, COL1A2, COPB2, CREB3L1, CRTAP, CYP27B1, CYP2R1, DMP1, ENPP1, FGF23, FKBP10, GORAB, IFITM5, KDELR2, KIF5B, LRP5, MBTPS2, MESD, NBAS, NOTCH2, P3H1, P4HB, PHEX, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SGMS2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SP7, SPARC, SUCO, TAPT1, TENT5A, TMEM38B, TRPV6, UNC45A, VDR, WNT1, WNT11, XYLT2

CentoPediatric Bone – Short-rib/Asphyxiating thoracic dysplasia | 31 genes

CentoPediatric Bone – Short-rib/Asphyxiating thoracic dysplasia targets genes associated with short-rib/asphyxiating thoracic dysplasia. This panel is intended for pediatric patients with clinical features suggestive of short-rib/asphyxiating thoracic dysplasia and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

C2CD3, CEP120, CFAP410, CILK1, CSPP1, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EVC, EVC2, FGFR3, GLI2, HYLS1, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, KIAA0586, KIF7, NEK1, OFD1, TCTN3, TRPV6, TTC21B, WDR19, WDR35

CentoPediatric Bone – Skeletal dysplasias | 558 genes

CentoPediatric Bone – Skeletal dysplasias targets genes associated with skeletal dysplasias. This panel is intended for pediatric patients with clinical features suggestive of skeletal dysplasias and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABCC9, ACAN, ACP5, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL2, AFF3, AFF4, AGA, AGPS, AIFM1, AKT2, ALG12, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANAPC1, ANKH, ANO5, ANTXR2, ARCN1, ARHGAP31, ARID1A, ARID1B, ARSB, ARSK, ARSL, ASAH1, ASXL1, ASXL2, ATP6V0A2, ATP7A, ATR, AXIN1, B3GALT6, B3GAT3, B3GLCT, B4GALT7, BANF1, BCL11B, BCOR, BCS1L, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BNIP1, BPNT2, C2CD3, CA2, CANT1, CASR, CBFB, CC2D2A, CCDC8, CCN6, CCNQ, CDC45, CDC73, CDH3, CDKN1C, CDT1, CEP120, CEP152, CEP290, CFAP410, CHST11, CHST14, CHST3, CHSY1, CILK1, CKAP2L, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COMP, COPB2, CREB3L1, CREBBP, CRLF1, CRTAP, CSF1R, CSGALNACT1, CSPP1, CTSA, CTSC, CTSK, CUL7, CWC27, CYP26B1, CYP27B1, CYP2R1, DDR2, DDRGK1, DHCR24, DHCR7, DHODH, DLL3, DLL4, DLX3, DLX5, DMP1, DNA2, DNAJC21, DNMT3A, DOCK6, DONSON, DPM1, DSE, DVL1, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EDN1, EDNRA, EED, EFL1, EFNB1, EFTUD2, EIF2AK3, EIF4A3, ELMO2, EN1, ENPP1, EOGT, EP300, ERF, ERI1, ESCO2, EVC, EVC2, EXOC6B, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20C, FANCB, FAR1, FBN1, FBN2, FERMT3, FGD1, FGF10, FGF16, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FKBP14, FLNA, FLNB, FMN1, FN1, FUCA1, FZD2, GALNS, GALNT3, GBA1, GCM2, GDF5, GDF6, GHR, GJA1, GLB1, GLI1, GLI2, GLI3, GMNN, GNAI3, GNAS, GNPAT, GNPNAT1, GNPTAB, GNPTG, GNS, GORAB, GPC3, GPC4, GPC6, GPX4, GRK2, GSC, GUSB, GZF1, HAAO, HDAC4, HDAC8, HEATR3, HES7, HGSNAT, HHAT, HNRNPK, HOXA13, HOXD13, HPGD, HRAS, HS2ST1, HSPA9, HSPG2, HYAL1, IARS2, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT54, IFT80, IFT81, IHH, IKBKG, IL11RA, IL1RN, IL6ST, INPPL1, INTU, KAT6B, KCNJ2, KDELR2, KIAA0586, KIAA0753, KIAA0825, KIF22, KIF5B, KIF7, KYNU, LAMA5, LARP7, LBR, LEMD3, LFNG, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LRRK1, LTBP1, LTBP2, LTBP3, LYSET, MAB21L2, MAFB, MAN2B1, MANBA, MAP2K1, MAP3K20, MAP3K7, MAPKAPK5, MASP1, MATN3, MBTPS1, MBTPS2, MCM7, MECOM, MEGF8, MEOX1, MESD, MESP2, MGP, MID1, MKS1, MMP13, MMP2, MMP9, MNX1, MSX2, MTAP, MTX2, MYCN, MYH3, MYO18B, NADSYN1, NAGLU, NANS, NBAS, NEK1, NEU1, NF1, NFIX, NIPBL, NKX3-2, NLRP3, NMNAT1, NOG, NOTCH1, NOTCH2, NPR2, NPR3, NRAS, NRCAM, NSD1, NSDHL, NSMCE2, NT5E, NXN, OBSL1, OCRL, OFD1, ORC1, ORC4, ORC6, OSGEP, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PDGFRB, PEX14, PEX19, PEX5, PEX7, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PISD, PITX1, PKDCC, PLCB4, PLEKHM1, PLOD2, PLS3, POC1A, POLE, POLR1A, POLR1B, POLR1C, POLR1D, POLR3A, POLR3B, POLR3GL, POP1, POR, PORCN, PPIB, PPP3CA, PRKACA, PRKACB, PRKAR1A, PRKG2, PRMT7, PRRX1, PSAT1, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RAD21, RASGRP2, RBBP8, RBM8A, RBPJ, RECQL4, RIGI, RINT1, RIPPLY2, RMP64, RMRP, RNU4ATAC, ROR2, RPGRIP1L, RPL13, RSPO2, RSPRY1, RUNX2, SALL1, SALL4, SBDS, SC5D, SCARF2, SCUBE3, SEC23A, SEC24D, SERPINF1, SERPINH1, SETBP1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHH, SHOX, SIK3, SIX1, SIX2, SKI, SLC10A7, SLC17A5, SLC25A24, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35B2, SLC35D1, SLC39A13, SLC4A2, SLCO2A1, SMAD2, SMAD3, SMAD4, SMAD6, SMARCA4, SMARCAL1, SMARCB1, SMARCE1, SMC1A, SMC3, SMCHD1, SMO, SMOC1, SMS, SNRPB, SNX10, SOST, SOX9, SP7, SPARC, SPECC1L, SRCAP, SRP54, STAMBP, STT3A, SULF1, SUMF1, SUZ12, TAB2, TAPT1, TBC1D24, TBCE, TBX15, TBX2, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCF12, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TFAP2B, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TMCO1, TMEM165, TMEM216, TMEM231, TMEM38B, TMEM53, TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TOP2B, TP63, TRAF7, TRAIP, TRAPPC2, TREM2, TRIM37, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TUBGCP6, TWIST1, TXNL4A, TYROBP, UBA2, UFSP2, UNC45A, VDR, VPS33A, VPS35L, WBP11, WDPCP, WDR19, WDR35, WNT1, WNT10B, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, ZIC1, ZIC3, ZMPSTE24, ZNF462, ZNF687, ZSWIM6

CentoPediatric Bone – Skeletal dysplasias Genome | 559 genes

CentoPediatric Bone - Skeletal dysplasias Genome targets genes associated with skeletal dysplasias. This panel is intended for pediatric patients with clinical features suggestive of skeletal dysplasias and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

> This version of the panel is based on genome sequencing, it includes the non-coding gene *MIR140*; and *GBA1* conversion analysis with its pseudogene.

Genes Included

ABCC9, ACAN, ACP5, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL2, AFF3, AFF4, AGA, AGPS, AIFM1, AKT2, ALG12, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANAPC1, ANKH, ANO5, ANTXR2, ARCN1, ARHGAP31, ARID1A, ARID1B, ARSB, ARSK, ARSL, ASAH1, ASXL1, ASXL2, ATP6V0A2, ATP7A, ATR, AXIN1, B3GALT6, B3GAT3, B3GLCT, B4GALT7, BANF1, BCL11B, BCOR, BCS1L, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BNIP1, BPNT2, C2CD3, CA2, CANT1, CASR, CBFB, CC2D2A, CCDC8, CCN6, CCNQ, CDC45, CDC73, CDH3, CDKN1C, CDT1, CEP120, CEP152, CEP290, CFAP410, CHST11, CHST14, CHST3, CHSY1, CILK1, CKAP2L, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COMP, COPB2, CREB3L1, CREBBP, CRLF1, CRTAP, CSF1R, CSGALNACT1, CSPP1, CTSA, CTSC, CTSK, CUL7, CWC27, CYP26B1, CYP27B1, CYP2R1, DDR2, DDRGK1, DHCR24, DHCR7, DHODH, DLL3, DLL4, DLX3, DLX5, DMP1, DNA2, DNAJC21, DNMT3A, DOCK6, DONSON, DPM1, DSE, DVL1, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EDN1, EDNRA, EED, EFL1, EFNB1, EFTUD2, EIF2AK3, EIF4A3, ELMO2, EN1, ENPP1, EOGT, EP300, ERF, ERI1, ESCO2, EVC, EVC2, EXOC6B, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20C, FANCB, FAR1, FBN1, FBN2, FERMT3, FGD1, FGF10, FGF16, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FKBP14, FLNA, FLNB, FMN1, FN1, FUCA1, FZD2, GALNS, GALNT3, GBA1, GCM2, GDF5, GDF6, GHR, GJA1, GLB1, GLI1, GLI2, GLI3, GMNN, GNAI3, GNAS, GNPAT, GNPNAT1, GNPTAB, GNPTG, GNS, GORAB, GPC3, GPC4, GPC6, GPX4, GRK2, GSC, GUSB, GZF1, HAAO, HDAC4, HDAC8, HEATR3, HES7, HGSNAT, HHAT, HNRNPK, HOXA13, HOXD13, HPGD, HRAS, HS2ST1, HSPA9, HSPG2, HYAL1, IARS2, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT54, IFT80, IFT81, IHH, IKBKG, IL11RA, IL1RN, IL6ST, INPPL1, INTU, KAT6B, KCNJ2, KDELR2, KIAA0586, KIAA0753, KIAA0825, KIF22, KIF5B, KIF7, KYNU, LAMA5, LARP7, LBR, LEMD3, LFNG, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LRRK1, LTBP1, LTBP2, LTBP3, LYSET, MAB21L2, MAFB, MAN2B1, MANBA, MAP2K1, MAP3K20, MAP3K7, MAPKAPK5, MASP1, MATN3, MBTPS1, MBTPS2, MCM7, MECOM, MEGF8, MEOX1, MESD, MESP2, MGP, MID1, MIR140, MKS1, MMP13, MMP2, MMP9, MNX1, MSX2, MTAP, MTX2, MYCN, MYH3, MYO18B, NADSYN1, NAGLU, NANS, NBAS, NEK1, NEU1, NF1, NFIX, NIPBL, NKX3-2, NLRP3, NMNAT1, NOG, NOTCH1, NOTCH2, NPR2, NPR3, NRAS, NRCAM, NSD1, NSDHL, NSMCE2, NT5E, NXN, OBSL1, OCRL, OFD1, ORC1, ORC4, ORC6, OSGEP, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PDGFRB, PEX14, PEX19, PEX5, PEX7, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PISD, PITX1, PKDCC, PLCB4, PLEKHM1, PLOD2, PLS3, POC1A, POLE, POLR1A, POLR1B, POLR1C, POLR1D, POLR3A, POLR3B, POLR3GL, POP1, POR, PORCN, PPIB, PPP3CA, PRKACA, PRKACB, PRKAR1A, PRKG2, PRMT7, PRRX1, PSAT1, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RAD21, RASGRP2, RBBP8, RBM8A, RBPJ, RECQL4, RIGI, RINT1, RIPPLY2, RMP64, RMRP, RNU4ATAC, ROR2, RPGRIP1L, RPL13, RSPO2, RSPRY1, RUNX2, SALL1, SALL4, SBDS, SC5D, SCARF2, SCUBE3, SEC23A, SEC24D, SERPINF1, SERPINH1, SETBP1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHH, SHOX, SIK3, SIX1, SIX2, SKI, SLC10A7, SLC17A5, SLC25A24, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35B2, SLC35D1, SLC39A13, SLC4A2, SLCO2A1, SMAD2, SMAD3, SMAD4, SMAD6, SMARCA4, SMARCAL1, SMARCB1, SMARCE1, SMC1A, SMC3, SMCHD1, SMO, SMOC1, SMS, SNRPB, SNX10, SOST, SOX9, SP7, SPARC, SPECC1L, SRCAP, SRP54, STAMBP, STT3A, SULF1, SUMF1, SUZ12, TAB2, TAPT1, TBC1D24, TBCE, TBX15, TBX2, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCF12, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TFAP2B, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TMCO1, TMEM165, TMEM216, TMEM231, TMEM38B, TMEM53, TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TOP2B, TP63, TRAF7, TRAIP, TRAPPC2, TREM2, TRIM37, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TUBGCP6, TWIST1, TXNL4A, TYROBP, UBA2, UFSP2, UNC45A, VDR, VPS33A, VPS35L, WBP11, WDPCP, WDR19, WDR35, WNT1, WNT10B, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, ZIC1, ZIC3, ZMPSTE24, ZNF462, ZNF687, ZSWIM6

Differences of sex development

CentoPediatric DSD Comprehensive | 149 genes

CentoPediatric DSD Comprehensive targets genes associated with a broad spectrum of disorders within pediatric differences of sex development, including conditions such as congenital adrenal hyperplasia, gonadal dysgenesis, androgen insensitivity syndrome and 5-alpha-reductase 2 deficiency. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

AMH, AMHR2, ANOS1, AR, ARL6, ARMC5, ARX, ATRX, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BMP15, BMP4, BMP7, CCNQ, CDK9, CDKN1C, CEP41, CHD4, CHD7, CILK1, CREBBP, CUL7, CYB5A, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP21A2, DHCR24, DHCR7, DHH, DHX37, DYNC2H1, DYNC2I1, EFNB1, EPG5, ERCC3, ESCO2, ESR2, EVC, EVC2, FAT4, FBXL4, FEZF1, FGF17, FGF8, FGFR1, FGFR2, FIG4, FOXL2, FRAS1, FREM2, FSHB, FSHR, GATA4, GLI3, GNRH1, GNRHR, GPC3, GRIP1, HCCS, HHAT, HNF1B, HOXA13, HSD17B3, HSD17B4, HSD3B2, INSL3, IRF6, KISS1, KISS1R, LEP, LEPR, LHB, LHCGR, LMNA, MAMLD1, MAP3K1, MCM9, MED12, MID1, MKKS, MKRN3, MKS1, MYRF, NEK1, NR0B1, NR5A1, OPHN1, PBX1, PCNT, PCSK1, PDE4D, PDE8B, PEX1, POLR3B, POR, PPP2R3C, PRKAR1A, PROK2, PROKR2, PROP1, PSMC3IP, PTDSS1, PTPN11, RIPK4, RNF216, ROR2, RSPO1, SALL1, SAMD9, SEMA3A, SETBP1, SGPL1, SOS1, SOX10, SOX2, SOX3, SOX9, SRD5A2, SRY, STAR, TAC3, TACR3, TBX15, TMEM70, TOE1, TP63, TRAIP, TRIM32, TSPYL1, TTC8, TWIST2, UBR1, WDR11, WNT4, WNT5A, WNT7A, WT1, ZEB2, ZFPM2

CentoPediatric DSD – CAH | 11 genes

CentoPediatric DSD - CAH targets genes associated with congenital adrenal hyperplasia. This panel is intended for pediatric patients with clinical features suggestive of congenital adrenal hyperplasia and supports the identification of a molecular diagnosis to inform clinical management, prognosis, and genetic counseling. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

This panel includes Sanger sequencing and MLPA analysis of *CYP21A2*.

Genes Included

ARMC5, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP21A2, HSD3B2, PDE8B, POR, PRKAR1A, STAR

Ear

CentoPediatric Ear Comprehensive | 311 genes

CentoPediatric Ear Comprehensive targets genes associated with a broad spectrum of disorders within pediatric hearing disorders, including both isolated (non-syndromic) hearing loss and syndromic forms linked to conditions such as Usher syndrome, Pendred syndrome, and Waardenburg syndrome. This panel is intended for pediatric patients with complex, overlapping, or nonspecific clinical presentations and facilitates a comprehensive molecular evaluation to support diagnosis, guide management, and inform family risk assessment. The panel is designed to include genes associated with early/childhood-onset disorders; conditions with exclusively adult-onset presentation are not within the scope of this test.

Genes Included

ABHD12, ABHD5, ACOX1, ACTB, ACTG1, ADGRV1, AFG2A, AFG2B, AIFM1, ALMS1, AMMECR1, ANKH, AP1B1, AP1S1, ATOH1, ATP11A, ATP1A3, ATP2B2, ATP6V0A4, ATP6V1B1, ATP6V1B2, BCAP31, BCS1L, BDP1, BSND, BTD, CABP2, CACNA1D, CD151, CD164, CDC14A, CDC42, CDH23, CEACAM16, CEP250, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLDN9, CLIC5, CLPP, CLRN1, CLRN2, COCH, COG4, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, DCAF17, DCDC2, DHRSX, DIAPH1, DLX5, DMXL2, EDN1, EDN3, EDNRA, EDNRB, EFTUD2, EIF3F, EPS8, EPS8L2, ERAL1, ESPN, ESRRB, EYA1, EYA4, FDXR, FGF3, FITM2, FOXC1, FOXI1, GATA3, GDF6, GGPS1, GIPC3, GJA1, GJB2, GJB3, GJB6, GNAI3, GPR156, GPSM2, GREB1L, GRHL2, GRXCR1, GRXCR2, GSDME, HAAO, HARS1, HARS2, HGF, HOMER2, HOXA2, HOXB1, HSD17B4, ILDR1, KARS1, KCNE1, KCNJ10, KCNJ16, KCNQ1, KCNQ4, KDM3B, KITLG, KMT2D, LARS2, LETM1, LHFPL5, LHX3, LMX1A, LOXHD1, LRP2, LRTOMT, MAFB, MAN2B1, MANBA, MAP1B, MARVELD2, MASP1, MET, MGP, MINAR2, MIR96, MITF, MN1, MORC2, MPZL2, MRPS2, MSRB3, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NF2, NLRP12, NLRP3, NOG, NR2F1, OGDHL, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX1, PAX2, PAX3, PBX1, PCDH15, PCGF2, PDSS1, PDZD7, PEX1, PEX11B, PEX12, PEX13, PEX14, PEX2, PEX26, PEX5, PEX6, PEX7, PHYH, PISD, PJVK, PKHD1L1, PLCB4, PLS1, PLXNB2, PMP22, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PPIP5K2, PRPS1, PSMC3, PTPRQ, RAI1, RDX, REST, RFC4, RIPOR2, RMND1, RNF220, ROR1, RPS6KA3, S1PR2, SALL1, SALL4, SEMA3E, SERAC1, SERPINB6, SGPL1, SIX1, SLC12A2, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLC4A11, SLC52A2, SLC52A3, SLC9A1, SLITRK6, SMAD4, SMPX, SNAI2, SOX10, SOX2, SPATC1L, SPNS2, SPTBN4, STAG2, STRC, STX4, STXBP3, SUCLA2, SUCLG1, SYNE4, TBC1D24, TBL1X, TBX1, TCOF1, TECTA, TFAP2A, THBS1, TIMM8A, TMC1, TMEM126A, TMEM132E, TMEM43, TMIE, TMPRSS3, TMTC4, TNC, TPRN, TRIOBP, TRMT10C, TRMU, TRRAP, TSHZ1, TSPEAR, TUBB4B, TWNK, UBR1, USH1C, USH1G, USH2A, USP48, WBP2, WFS1, WHRN, XPA, XYLT2, YARS1

Oncology

CentoCancerHereditary Pediatric | 136 genes

Cancer occurs in people of all ages, with some being nearly exclusively tied to childhood. CentoCancerHereditary Pediatric is our comprehensive solution to detect genes associated with pediatric cancer. The gene list has been carefully curated by internal and external experts to cover the most common forms of pediatric cancer, such as leukemia, malignant brain tumors, lymphomas, bone cancer, neuroblastoma, Wilms tumor, and rhabdomyosarcoma. Germline variants identified by this panel will help to define prognosis, differentiate patient/family risk, and guide treatment decisions. Spotting cancer early increases the chances of survival.

TAT

15 Business Days

Genes Included

AIP, AKT1, ALK, ANKRD26, APC, ATM, BAP1, BLM, BMPR1A, BRAF, BUB1B, CBL, CD40LG, CDC73, CDKN1B, CDKN1C, CEBPA, COL7A1, DDB2, DDX41, DICER1, DIS3L2, DKC1, DNAJC21, EFL1, EGLN1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, GATA2, GPC3, HAVCR2, HRAS, IKZF1, ITK, KRAS, LZTR1, MAD1L1, MAP2K1, MAP2K2, MAX, MBD4, MECOM, MEN1, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NHP2, NKX2-1, NOP10, NPM1, NRAS, NSD1, PALB2, PAX5, PHOX2B, PMS2, POLH, POT1, PRF1, PRKAR1A, PTCH1, PTEN, PTPN11, RAD51C, RAF1, RASA2, RB1, RECQL4, REST, RET, RHBDF2, RIT1, RPL5, RPS7, RRAS, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SHOC2, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SOS1, SOS2, SRP54, STK11, SUFU, TERC, TERT, TINF2, TMEM127, TP53, TRIM28, TRIM37, TRIP13, TSC1, TSC2, VHL, WAS, WRAP53, WRN, WT1, XPA, XPC, XRCC2