Inherited Disease Panel: List of Inherited Diseases and associated genes tested

DISEASE	GENE SYMBOL
Agammaglobulinemia, X-Linked, Type 1	*BTK*
Alagille Syndrome	*JAG1*
Alopecia Universalis Congenita (ALUNC)	*HR*
Alpers Syndrome	*POLG*
Alpha-1-Antitrypsin Deficiency	*SERPINA1*
Alpha-Thalassemia - Southeast Asia	*HBA2*
Alport Syndrome	*COL4A5*
Amyotrophic Lateral Sclerosis (Lou Gehrig's Disease)	*SOD1*
Androgen Insensitivity Syndrome	*AKR1B1*
Angioedema, Hereditary, Types I and II	*SERPING1*
Aniridia	*PAX6*
APC-Associated Polyposis Conditions	*APC*
Argininosuccinate Lyase Deficiency	*ASL*
Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy	*DSC2,DSG2,DSP,JUP,PKP2,RYR2,TMEM43*
Arylsulfatase A Deficiency	*ARSA*
Ataxia with Oculomotor Apraxia Type 2	*APTX*
Ataxia with Vitamin E Deficiency	*TTPA*
Ataxia-Telangiectasia	*ATM*
Atrial Septal Defect	*GATA4*
Autoimmune Polyendocrine Syndrome	*AIRE*
Beta-Hydroxyisobutyryl CoA Deacylase Deficiency (HIBCH Deficiency)	*HIBCH*
Biotinidase Deficiency	*BTD*
Blepharophimosis-Ptosis-Epicanthus Inversus	*FOXL2*
Brachydactyly	*GDF5*
Brachydactyly, Type B1	*ROR2*
Branchiootorenal Spectrum Disorders	*EYA1,SIX1,SIX5*
Brugada Syndrome	*CACNA1C,CACNB2,GPD1L,HCN4,KCNE3, SCN1B,SCN3B,SCN5A*
Campomelic Dysplasia	*SOX9*
Canavan	*ASPA*
Cardiomyopathy (Dilated)	*STARD3, TAZ*
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)	*CASQ2*
Cerebrotendinous Xanthomatosis	*CYP27A1*
Ceroid Lipofuscinoses (Batten Disease)	*PPT1*
Charcot-Marie-Tooth Disease Type 2B	*DNM2*
Charcot-Marie-Tooth Neuropathy Type 1A	*PMP22*
Charcot-Marie-Tooth Neuropathy Type 1B	*MPZ*
Charcot-Marie-Tooth Neuropathy Type 2A	*MFN2*
Charge Syndrome	*CHD7*
Cherubism	*SH3BP2*
Choroideremia	*CHM*
Citrin Deficiency	*SLC25A13*
Coffin-Lowry Syndrome	*RPS6KA3*
Congenital Cataracts, Facial Dysmorphism, and Neuropathy	*CTDP1*
Congenital Disorder of Glycosylation Type 1a	*PMM2*
Congenital Myasthenic Syndromes	*CHRNA1, CHRNB1,CHRND,CHRNE,DOK7,RAPSN, CHAT*
Cornelia de Lange Syndrome	*NIPBL*
Familial Hypertrophic Cardiomyopathy	*ACTC1,CALR3, CAV3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK, MYOZ2, PRKAG2, RPS7, SLC25A4, TNNT2, TPM1, VCL*
Cystinosis	*CTNS*
Darier Disease	*ATP2A2*
Diamond-Blackfan Anemia	*RPL11, RPL35A, RPS10, RPS19, RPS24, RPS26*
Dilated Cardiomyopathy	*DES, LAMP2, LDB3, SGCD, TNNC1, TNNI3*
Dilated Cardiomyopathy 1AA	*ACTN2*
Dilated Cardiomyopathy 1HH	*BAG3*
Dilated Cardiomyopathy 1O	*ABCC9*
Dilated Cardiomyopathy 1P	*PLN*
Double Cortex Syndrome	*DCX*
Duane Syndrome - Autosomal Dominant	*SALL4*
Duchenne/Becker Muscular Dystrophy	*DMD*
Dysferlinopathy	*DYSF*
Dyskeratosis Congenita	*DKC1*
Early-Onset Familial Alzheimer Disease	*APP, PSEN1, PSEN2*
Ehlers-Danlos Syndrome	*COL3A1, COL5A1, COL5A2*
Ehlers-Danlos Syndrome, Hypermobility Type	*TNXB, PLOD1, EMD*
Epidermolysis Bullosa Simplex	*COL7A1, ITGB4, KRT14, KRT5, LAMB3, PLEC*
Exostoses, Multiple, Type 1	*EXT1*
Fabry Disease	*GLA*
Facioscapulohumeral Muscular Dystrophy	*FRG1*
Familial Dysautonomia (HSAN III)	*IKBKAP*
Familial Transthyretin Amyloidosis	*TTR*
Fanconi Anemia	*FANCA, FANCC, FANCF, FANCG*
FGFR-Related Craniosynostosis Syndromes	*FGFR1*
Friedreich Ataxia	*FXN*
FRMD7-Related Infantile Nystagmus	*FRMD7*
Fryns Syndrome	*MED12*
Galactosemia	*GALT*
Gaucher Disease	*GBA*
Glycine Encephalopathy	*AMT, GCSH, GLDC*
Glycogen Storage Disease Type VI	*GBE1*
Hemophilia A	*F8*
Hemophilia B	*F9*
Hereditary Hemorrhagic Telangiectasia	*ENG*
Hexosaminidase A Deficiency	*HEXA*
HFE-Associated Hereditary Hemochromatosis	*HFE*
Holoprosencephaly-7 & Basal Cell Nevus Syndrome	*PTCH1*
Holt-Oram Syndrome	*TBX5*
Hunter Syndrome (MPSII)	*IDS, IDUA*

DISEASE	GENE SYMBOL
Hydroxymethylbilane Synthase (HMBS) Deficiency	*HMBS*
Hypochondroplasia	*FGFR3*
Hypophosphatasia	*ALPL*
Inclusion Body Myopathy 2	*GNE*
Inherited Deafness	*COLL11A2, KCNQ4*
Inherited Deafness, Top Genes	*GJB2, GJB3, GJB6*
Juvenile Polyposis Syndrome	*BMPR1A, SMAD4*
Leber Congenital Amaurosis	*AIPL1, CEP290. CRB1, GUCY2D, IMPDH1, RDH12, RPE65, RPGRIP1*
Li-Fraumeni Syndrome	*CHEK2, TP53*
Limb-Girdle Muscular Dystrophy Type 2A - Calpainopathy	*CAPN3*
Limb-Girdle Muscular Dystrophy, Type 1B	*LMNA*
Lissencephaly 1	*PAFAH1B1*
Long QT Syndrome, Autosomal Dominant	*AKAP9, KCNE1, KCNE2, KCNH2, KCNQ1, SCN4B,SNTA1, ANK2*
Lowe Syndrome	*OCRL*
Malignant Hyperthermia Susceptibility	*RYR1*
Maple Syrup Urine Disease	*BCKDHA, BCKDHB, DBT,DLD*
Marfan Syndrome	*FBN1*
MECP2-Rett Syndrome	*MECP2*
Menkes/ATP7A-Related Copper Transport Disease	*ATP7A*
Methylmalonic Acidemia	*MMAA, MMAB,MMACHC,MUT*
Mucolipidosis II	*GNPTAB*
Multiple Endocrine Neoplasia Type 1	*MEN1*
Multiple Endocrine Neoplasia Type 2	*RET*
Myotonia Congenita	*CLCN1*
Nemaline Myopathy	*TNNT1*
Neonatal Adrenoleucodystrophy	*PEX5*
Neurofibromatosis Type 1	*NF1*
Neurofibromatosis Type 2	*NF2*
Niemann-Pick Disease Type C1	*NPC1*
Niemann-Pick Disease Type C2	*NPC2*
Noonan Syndrome	*KRAS,NRAS,PTPN11, RAF1, SOS1*
Ocular Albinism, X-Linked	*GPR143*
Oculocutaneous Albinism Type 1	*TYR*
Oculocutaneous Albinism Type 2	*MCR1, OCA2*
Oculopharyngeal Muscular Dystrophy	*PABPN1*
Ornithine Transcarbamylase Deficiency	*OTC*
Osteogenesis Imperfecta	*COL1A1, COL1A2*
Parkinson Disease	*FBX07,LRRK2,PINK1,SNCA*
Parkinson-Dementia Syndrome	*MAPT*
Pendred Syndrome/Syndromic Deafness	*SLC26A4*
Peroxisome Biogenesis, Zellweger	*PEX10, PEX13, PEX14, PEX19, PEX26, PEX3*
Phenylketonuria (PKU)	*PAH*
Polycystic Kidney Disease, Autosomal Dominant	*PKD1, PKD2, PKHD1*
Pompe Disease -GSD II	*GAA*
Primary Ciliary Dyskinesia	*CCDC39,CCDC40, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, RSPH4A, RSPH9, TXNDC3*
Retinitis Pigmentosa	*ABCA4, ARL6, BEST1, CA4, CERKL, CNGB1, CRX, EYS, FSCN2, KLHL7,LRAT, MAPRE2, MERTK, NR2E3, NUNDT19, PRCD, PROM1, PRPF31, PRPF8, PRPH2, RHO, RP9, RPGR,SEMA4A, SNRNP200, TOPORS, TULP1*
Retinoblastoma0	*RB1*
Saethre-Chotzen Syndrome	*TWIST1*
SCN9A-Related Inherited Erythromelalgia	*SCN9A*
Severe Combined Immunodeficiency	*ADA, RAG1, RAG2*
Short QT Syndrome	*KCNJ2*
Sickle Cell Disease Beta-Thalassemia	*HBB*
Smith-Lemli-Opitz Syndrome	*DHCR7*
Smith-Magenis Syndrome	*RAI1*
Sotos Syndrome	*NSD1*
Spastic Paraplegia 7	*SPG7*
Spastic Paraplegia 8	*KIAA0196*
Spastic Paraplegia Type 1 - L1 Syndrome	*L1CAM*
Spastic Paraplegia-3A	*ATL1*
Spinocerebellar Ataxia 1	*ATXN1, ATXN2, ATXN7*
Stickler Syndrome	*COL9A1*
Stickler Syndrome, AD	*COL11A1, COL2A1*
Supravalvular Aortic Stenosis	*ELN*
Tetralogy of Fallot	*NKX2-5*
Thoracic Aortic Aneurysms and Aortic Dissections	*ACTA2, COL4A1, MYH11, SMAD3, TGFBR1, TGFBR2*
Treacher Collins Syndrome	*TCOF1*
Trimethylaminuria	*FMO3*
Tuberous Sclerosis Complex	*TSC1, TSC2*
Turcot Syndrome	*MLH1, MSH2*
Usher Syndrome Type 1	*CDH23, MYO7A, PCDH15, USH1C*
Usher Syndrome Type 2	*USH2A*
Very Long Chain Acyl-Coenzyme A Dehydrogenase Deficiency	*ACADVL*
von Hippel-Lindau Syndrome	*VHL*
Waardenburg Syndrome, Type 1	*PAX3*
Werner Syndrome	*WRN*
Wilms Tumor, Classic	*WT1, GPC3*
Wilson Disease	*ATP7B*
Wiskott-Aldrich Syndrome	*WAS*
X-Linked Adrenoleukodystrophy	*ABCD1*
X-Linked Dystonia-Parkinsonism	*TAF1*
X-Linked Juvenile Retinoschisis	*RS1*
X-Linked Myotubular Myopathy	*MTM1*
X-Linked SCIDS	*IL2RG*
Zellweger Syndrome	*PEX1*

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