DISEASE	GENE SYMBOL	DISEASE	GENE SYMBOL
Agammaglobulinemia, X-Linked, Type 1	***BTK***	Hunter Syndrome (MPSII)	***IDS, IDUA***
Alagille Syndrome	***JAG1***	Hydroxymethylbilane Synthase (HMBS) Deficiency	***HMBS***
Alopecia Universalis Congenita (ALUNC)	***HR***	Hypochondroplasia	***FGFR3***
Alpers Syndrome	***POLG***	Hypophosphatasia	***ALPL***
Alpha-1-Antitrypsin Deficiency	***SERPINA1***	Inclusion Body Myopathy 2	***GNE***
Alpha-Thalassemia - Southeast Asia	***HBA2***	Inherited Deafness	***COLL11A2, KCNQ4***
Alport Syndrome	***COL4A5***	Inherited Deafness, Top Genes	***GJB2, GJB3, GJB6***
Amyotrophic Lateral Sclerosis (Lou Gehrig's Disease)	***SOD1***	Juvenile Polyposis Syndrome	***BMPR1A, SMAD4***
Androgen Insensitivity Syndrome	***AKR1B1***	Leber Congenital Amaurosis	***AIPL1, CEP290. CRB1, GUCY2D, IMPDH1, RDH12, RPE65, RPGRIP1***
Angioedema, Hereditary, Types I and II	***SERPING1***	Li-Fraumeni Syndrome	***CHEK2, TP53***
Aniridia	***PAX6***	Limb-Girdle Muscular Dystrophy Type 2A - Calpainopathy	***CAPN3***
APC-Associated Polyposis Conditions	***APC***	Limb-Girdle Muscular Dystrophy, Type 1B	***LMNA***
Argininosuccinate Lyase Deficiency	***ASL***	Lissencephaly 1	***PAFAH1B1***
Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy	***DSC2,DSG2,DSP,JUP,PKP2,RYR2,TMEM43***	Long QT Syndrome, Autosomal Dominant	***AKAP9, KCNE1, KCNE2, KCNH2, KCNQ1, SCN4B,SNTA1, ANK2***
Arylsulfatase A Deficiency	***ARSA***	Lowe Syndrome	***OCRL***
Ataxia with Oculomotor Apraxia Type 2	***APTX***	Malignant Hyperthermia Susceptibility	***RYR1***
Ataxia with Vitamin E Deficiency	***TTPA***	Maple Syrup Urine Disease	***BCKDHA, BCKDHB, DBT,DLD***
Ataxia-Telangiectasia	***ATM***	Marfan Syndrome	***FBN1***
Atrial Septal Defect	***GATA4***	MECP2-Rett Syndrome	***MECP2***
Autoimmune Polyendocrine Syndrome	***AIRE***	Menkes/ATP7A-Related Copper Transport Disease	***ATP7A***
Beta-Hydroxyisobutyryl CoA Deacylase Deficiency (HIBCH Deficiency)	***HIBCH***	Methylmalonic Acidemia	***MMAA, MMAB,MMACHC,MUT***
Biotinidase Deficiency	***BTD***	Mucolipidosis II	***GNPTAB***
Blepharophimosis-Ptosis-Epicanthus Inversus	***FOXL2***	Multiple Endocrine Neoplasia Type 1	***MEN1***
Brachydactyly	***GDF5***	Multiple Endocrine Neoplasia Type 2	***RET***
Brachydactyly, Type B1	***ROR2***	Myotonia Congenita	***CLCN1***
Branchiootorenal Spectrum Disorders	***EYA1,SIX1,SIX5***	Nemaline Myopathy	***TNNT1***
Brugada Syndrome	***CACNA1C,CACNB2,GPD1L,HCN4,KCNE3,SCN1B,SCN3B,SCN5A***	Neonatal Adrenoleucodystrophy	***PEX5***
Campomelic Dysplasia	***SOX9***	Neurofibromatosis Type 1	***NF1***
Canavan	***ASPA***	Neurofibromatosis Type 2	***NF2***
Cardiomyopathy (Dilated)	***STARD3, TAZ***	Niemann-Pick Disease Type C1	***NPC1***
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)	***CASQ2***	Niemann-Pick Disease Type C2	***NPC2***
Cerebrotendinous Xanthomatosis	***CYP27A1***	Noonan Syndrome	***KRAS,NRAS,PTPN11, RAF1, SOS1***
Ceroid Lipofuscinoses (Batten Disease)	***PPT1***	Ocular Albinism, X-Linked	***GPR143***
Charcot-Marie-Tooth Disease Type 2B	***DNM2***	Oculocutaneous Albinism Type 1	***TYR***
Charcot-Marie-Tooth Neuropathy Type 1A	***PMP22***	Oculocutaneous Albinism Type 2	***MCR1, OCA2***
Charcot-Marie-Tooth Neuropathy Type 1B	***MPZ***	Oculopharyngeal Muscular Dystrophy	***PABPN1***
Charcot-Marie-Tooth Neuropathy Type 2A	***MFN2***	Ornithine Transcarbamylase Deficiency	***OTC***
Charge Syndrome	***CHD7***	Osteogenesis Imperfecta	***COL1A1, COL1A2***
Cherubism	***SH3BP2***	Parkinson Disease	***FBX07,LRRK2,PINK1,SNCA***
Choroideremia	***CHM***	Parkinson-Dementia Syndrome	***MAPT***
Citrin Deficiency	***SLC25A13***	Pendred Syndrome/Syndromic Deafness	***SLC26A4***
Coffin-Lowry Syndrome	***RPS6KA3***	Peroxisome Biogenesis, Zellweger	***PEX10, PEX13, PEX14, PEX19, PEX26, PEX3***
Congenital Cataracts, Facial Dysmorphism, and Neuropathy	***CTDP1***	Phenylketonuria (PKU)	***PAH***
Congenital Disorder of Glycosylation Type 1a	***PMM2***	Polycystic Kidney Disease, Autosomal Dominant	***PKD1, PKD2, PKHD1***
Congenital Myasthenic Syndromes	***CHRNA1, CHRNB1,CHRND,CHRNE,DOK7,RAPSN,CHAT***	Pompe Disease -GSD II	***GAA***
Cornelia de Lange Syndrome	***NIPBL***	Primary Ciliary Dyskinesia	***CCDC39,CCDC40, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, RSPH4A, RSPH9, TXNDC3***
Familial Hypertrophic Cardiomyopathy (extra charge for multiple gene analysis)	***ACTC1,CALR3, CAV3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK, MYOZ2, PRKAG2, RPS7, SLC25A4, TNNT2, TPM1, VCL***	Retinitis Pigmentosa (extra charge for multiple gene analysis)	***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***
Cystinosis	***CTNS***	Retinoblastoma0	***RB1***
Darier Disease	***ATP2A2***	Saethre-Chotzen Syndrome	***TWIST1***
Diamond-Blackfan Anemia	***RPL11, RPL35A, RPS10, RPS19, RPS24, RPS26***	SCN9A-Related Inherited Erythromelalgia	***SCN9A***
Dilated Cardiomyopathy	***DES, LAMP2, LDB3, SGCD, TNNC1, TNNI3***	Severe Combined Immunodeficiency	***ADA, RAG1, RAG2***
Dilated Cardiomyopathy 1AA	***ACTN2***	Short QT Syndrome	***KCNJ2***
Dilated Cardiomyopathy 1HH	***BAG3***	Sickle Cell Disease Beta-Thalassemia	***HBB***
Dilated Cardiomyopathy 1O	***ABCC9***	Smith-Lemli-Opitz Syndrome	***DHCR7***
Dilated Cardiomyopathy 1P	***PLN***	Smith-Magenis Syndrome	***RAI1***
Double Cortex Syndrome	***DCX***	Sotos Syndrome	***NSD1***
Duane Syndrome - Autosomal Dominant	***SALL4***	Spastic Paraplegia 7	***SPG7***
Duchenne/Becker Muscular Dystrophy	***DMD***	Spastic Paraplegia 8	***KIAA0196***
Dysferlinopathy	***DYSF***	Spastic Paraplegia Type 1 - L1 Syndrome	***L1CAM***
Dyskeratosis Congenita	***DKC1***	Spastic Paraplegia-3A	***ATL1***
Early-Onset Familial Alzheimer Disease	***APP, PSEN1, PSEN2***	Spinocerebellar Ataxia 1	***ATXN1, ATXN2, ATXN7***
Ehlers-Danlos Syndrome	***COL3A1, COL5A1, COL5A2***	Stickler Syndrome	***COL9A1***
Ehlers-Danlos Syndrome, Hypermobility Type	***TNXB, PLOD1, EMD***	Stickler Syndrome, AD	***COL11A1, COL2A1***
Epidermolysis Bullosa Simplex	***COL7A1, ITGB4, KRT14, KRT5, LAMB3, PLEC***	Supravalvular Aortic Stenosis	***ELN***
Exostoses, Multiple, Type 1	***EXT1***	Tetralogy of Fallot	***NKX2-5***
Fabry Disease	***GLA***	Thoracic Aortic Aneurysms and Aortic Dissections	***ACTA2, COL4A1, MYH11, SMAD3, TGFBR1, TGFBR2***
Facioscapulohumeral Muscular Dystrophy	***FRG1***	Treacher Collins Syndrome	***TCOF1***
Familial Dysautonomia (HSAN III)	***IKBKAP***	Trimethylaminuria	***FMO3***
Familial Transthyretin Amyloidosis	***TTR***	Tuberous Sclerosis Complex	***TSC1, TSC2***
Fanconi Anemia	***FANCA, FANCC, FANCF, FANCG***	Turcot Syndrome	***MLH1, MSH2***
FGFR-Related Craniosynostosis Syndromes	***FGFR1***	Usher Syndrome Type 1	***CDH23, MYO7A, PCDH15, USH1C***
Friedreich Ataxia	***FXN***	Usher Syndrome Type 2	***USH2A***
FRMD7-Related Infantile Nystagmus	***FRMD7***	Very Long Chain Acyl-Coenzyme A Dehydrogenase Deficiency	***ACADVL***
Fryns Syndrome	***MED12***	von Hippel-Lindau Syndrome	***VHL***
Galactosemia	***GALT***	Waardenburg Syndrome, Type 1	***PAX3***
Gaucher Disease	***GBA***	Werner Syndrome	***WRN***
Glycine Encephalopathy	***AMT, GCSH, GLDC***	Wilms Tumor, Classic	***WT1, GPC3***
Glycogen Storage Disease Type VI	***GBE1***	Wilson Disease	***ATP7B***
Hemophilia A	***F8***	Wiskott-Aldrich Syndrome	***WAS***
Hemophilia B	***F9***	X-Linked Adrenoleukodystrophy	***ABCD1***
Hereditary Hemorrhagic Telangiectasia	***ENG***	X-Linked Dystonia-Parkinsonism	***TAF1***
Hexosaminidase A Deficiency	***HEXA***	X-Linked Juvenile Retinoschisis	***RS1***
HFE-Associated Hereditary Hemochromatosis	***HFE***	X-Linked Myotubular Myopathy	***MTM1***
Holoprosencephaly-7 & Basal Cell Nevus Syndrome	***PTCH1***	X-Linked SCIDS	***IL2RG***
Holt-Oram Syndrome	***TBX5***	Zellweger Syndrome	***PEX1***