Individual Genome Sequence Gene List (By Disease)

Disease	Gene Symbol	Inheritance	Transcript	Callability
17-Beta-Hydroxysteroid Dehydrogenase III Deficiency	HSD17B3	RECESSIVE	NM_000197.1	0.9994
3-Hydroxy-3-Methylglutaryl-CoA Synthase 2 Deficiency	HMGCS2	RECESSIVE	NM_005518.3	0.9996
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	HMGCL	RECESSIVE	NM_000191.2	0.9991
3-Hydroxyacyl-Coenzyme A Dehydrogenase Deficiency	HADH	RECESSIVE	NM_005327.4	0.9996
3-M Syndrome	CUL7	RECESSIVE	NM_014780.4	0.9990
	OBSL1	RECESSIVE	NM_015311.2	0.9967
3-MCC Deficiency	MCCC1	RECESSIVE	NM_020166	0.9997
	MCCC2	RECESSIVE	NM_022132	0.9996
3-Methylglutaconic Aciduria, Type 1	AUH	RECESSIVE	NM_001698.2	0.9996
3-Methylglutaconic Aciduria, Type 2	TAZ	X_LINKED	NM_000116	0.9984
3-Methylglutaconic Aciduria, Type 3	OPA3	RECESSIVE	NM_025136	0.9973
3-Methylglutaconic Aciduria, Type 5	DNAJC19	RECESSIVE	NM_145261.3	0.9997
46,XY DSD/46,XY CGD	DHH	RECESSIVE	NM_021044.2	0.9994
6-Pyruvoyltetrahydropterin Synthase Deficiency	PTS	RECESSIVE	NM_000317.2	0.9997
ABCA4-Related Disorders	ABCA4	RECESSIVE (Rec/Dom)	NM_000350	0.9981
Abetalipoproteinemia	MTTP	RECESSIVE	NM_000253.2	0.9994
ACAD9 Deficiency	ACAD9	RECESSIVE	NM_014049.4	0.9995
Aceruloplasminemia	CP	RECESSIVE	NM_000096.3	0.9992
Achalasia-Addisonianism-Alacrima Syndrome	AAAS	RECESSIVE	NM_015665.5	0.9982
Achondrogenesis	SLC26A2	RECESSIVE	NM_000112.3	0.9994
	TRIP11	RECESSIVE	NM_004239.3	0.9998
Achromatopsia	CNGA3	RECESSIVE	NM_001298.2	0.9989

Achromatopsia	CNGB3	RECESSIVE	NM_019098.4	0.9991
	GNAT2	RECESSIVE	NM_005272.3	0.9997
	PDE6C	RECESSIVE	NM_006204.3	0.9997
Acid Sphingomyelinase Deficiency	SMPD1	RECESSIVE	NM_000543	0.9805
Acid-Labile Subunit Deficiency	IGFALS	RECESSIVE	NM_004970.2	0.9992
Acrocallosal Syndrome	KIF7	RECESSIVE	NM_198525.2	0.9982
Acrodermatitis Enteropathica	SLC39A4	RECESSIVE	NM_130849.2	0.9987
Acrodysostosis	PDE4D	DOMINANT	NM_001104631.1	0.9989
	PRKAR1A	DOMINANT	NM_002734.3	0.9996
Acromesomelic Dysplasia	GDF5	RECESSIVE	NM_000557.2	0.9989
	NPR2	RECESSIVE	NM_003995.3	0.9982
Acromicric Dysplasia	FBN1	DOMINANT	NM_000138.4	0.9997
ACTH Deficiency	TBX19	RECESSIVE	NM_005149.2	0.9994
Acute Hepatic Porphyria	ALAD	RECESSIVE	NM_000031.5	0.9993
Acute Infantile Liver Failure	TRMU	RECESSIVE	NM_018006.4	0.9987
Acute Recurrent Myoglobinuria	LPIN1	RECESSIVE	NM_145693.2	0.9931
Acyl-CoA Dehydrogenase, Short/Branched Chain Deficiency	ACADSB	RECESSIVE	NM_001609.3	0.9981
Adams-Oliver Syndrome	ARHGAP31	DOMINANT	NM_020754.2	0.9993
Adenine Phosphoribosyltransferase Deficiency	APRT	RECESSIVE	NM_000485.2	0.9933
Adenosine Deaminase Deficiency	ADA	RECESSIVE	NM_000022	0.9986
Adenylosuccinase Deficiency	ADSL	RECESSIVE	NM_000026.2	0.9995
Adrenoleukodystrophy, X-linked	ABCD1	X_LINKED	NM_000033.3	0.9962
Adult i Blood Group With or Without Congenital Cataract	GCNT2	RECESSIVE	NM_001491.2	0.9997
Adult Polyglucosan Body Disease	GBE1	RECESSIVE	NM_000158.3	0.9997
Agammaglobulinemia, X-linked	BTK	X_LINKED	NM_000061.2	0.9998
Age-Related Cortical Cataract	EPHA2	DOMINANT	NM_004431.3	0.9991
Aicardi-Goutieres Syndrome	RNASEH2A	RECESSIVE	NM_006397.2	0.9991
	RNASEH2B	RECESSIVE	NM_024570.3	0.9995
	RNASEH2C	RECESSIVE	NM_032193.3	0.9988
	SAMHD1	RECESSIVE	NM_015474.3	0.9996
	TREX1	RECESSIVE	NM_033629.2	0.9983

Alagille Syndrome	JAG1	DOMINANT	NM_000214	0.9993
Aldolase A Deficiency	ALDOA	RECESSIVE	NM_000034.3	0.9988
Alexander Disease	GFAP	DOMINANT	NM_002055.4	0.9990
Alkaptonuria	HGD	RECESSIVE	NM_000187.3	0.9996
Alopecia and T-Cell Immunodeficiency	FOXN1	RECESSIVE	NM_003593.2	0.9977
Alopecia Universalis	HR	RECESSIVE	NM_005144.4	0.9958
Alpha-1 Antitrypsin Deficiency	SERPINA1	RECESSIVE	NM_000295	0.9978
Alpha-B Crystallinopathy	CRYAB	RECESSIVE	NM_001885.1	0.9995
Alpha-Mannosidosis	MAN2B1	RECESSIVE	NM_000528.3	0.9984
Alpha-Methylacyl-CoA Racemase Deficiency	AMACR	RECESSIVE	NM_014324.5	0.9982
Alpha-Sarcoglycanopathy	SGCA	RECESSIVE	NM_000023	0.9989
Alport Syndrome	COL4A3	RECESSIVE (Rec/Dom)	NM_000091.4	0.9996
	COL4A4	RECESSIVE (Rec/Dom)	NM_000092.4	0.9988
ALS2-Related Spectrum Disorders	ALS2	RECESSIVE	NM_020919	0.9996
Alstrom Syndrome	ALMS1	RECESSIVE	NM_015120.4	0.9993
Alternating Hemiplegia of Childhood	ATP1A2	DOMINANT	NM_000702.3	0.9992
	ATP1A3	DOMINANT	NM_152296.4	0.9819
Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins	FOXF1	DOMINANT	NM_001451.2	0.9977
Amelogenesis Imperfecta, Dominant	DLX3	RECESSIVE (Rec/Dom)	NM_005220.2	0.9980
	ENAM	RECESSIVE (Rec/Dom)	NM_031889.2	0.9996
Amelogenesis Imperfecta, Recessive	MMP20	RECESSIVE (Rec/Dom)	NM_004771.3	0.9946
	WDR72	RECESSIVE (Rec/Dom)	NM_182758.2	0.9985
Amish Infantile Epilepsy Syndrome	ST3GAL5	RECESSIVE	NM_003896.3	0.9995
Amish Lethal Microcephaly	SLC25A19	RECESSIVE	NM_021734.4	0.9993
Amyloidosis	GSN	DOMINANT	NM_000177.4	0.9971
Amyotrophic Lateral Sclerosis, Dominant	ANG	DOMINANT	NM_001145.4	0.9995
	FIG4	DOMINANT	NM_014845	0.9995
	FUS	DOMINANT	NM_004960.3	0.9990
	SETX	DOMINANT	NM_015046	0.9995
	SOD1	DOMINANT	NM_000454	0.9999

Amyotrophic Lateral Sclerosis, Dominant	TARDBP	DOMINANT	NM_007375.3	0.9997
	UBQLN2	X_LINKED	NM_013444.3	0.9996
	VAPB	DOMINANT	NM_004738	0.9973
	VCP	DOMINANT	NM_007126.3	0.9994
Amyotrophic Lateral Sclerosis, Recessive	ALS2	RECESSIVE	NM_020919	0.9996
	OPTN	RECESSIVE	NM_021980.4	0.9944
Amyotrophic Lateral Sclerosis/Frontotemporal Dementia	C9orf72	DOMINANT	NM_001256054.1	0.9971
Andersen-Tawil Syndrome	KCNJ2	DOMINANT	NM_000891	0.9927
Angiokeratoma Corporis Diffusum with Arteriovenous Fistulas	KRIT1	DOMINANT	NM_194456.1	0.9984
Aniridia	PAX6	DOMINANT	NM_000280.4	0.9984
Aniridia, Cerebellar Ataxia, And Intellectual Disability	PAX6	RECESSIVE (Rec/Dom)	NM_000280.4	0.9984
ANO5-Related Muscle Diseases	ANO5	RECESSIVE	NM_213599	0.9995
Anophthalmia	PAX6	RECESSIVE	NM_000280.4	0.9984
Anophthalmia/Microphthalmia	RAX	RECESSIVE (Rec/Dom)	NM_013435.2	0.9915
	SIX6	RECESSIVE (Rec/Dom)	NM_007374.2	0.9996
Antenatal Bartter Syndrome	KCNJ1	RECESSIVE	NM_000220.4	0.9997
	SLC12A1	RECESSIVE	NM_000338.2	0.9998
Anterior Segment Mesenchymal Dysgenesis	PITX2	DOMINANT	NM_153427.2	0.9962
Antithrombin-III Deficiency	SERPINC1	DOMINANT	NM_000488	0.9995
APC-Associated Polyposis Disorders	APC	DOMINANT	NM_000038	0.9998
Apert Syndrome	FGFR2	DOMINANT	NM_000141.4	0.9980
Aplasia of Lacrimal and Salivary Glands	FGF10	DOMINANT	NM_004465.1	0.9984
Aplastic Anemia	IFNG	RECESSIVE	NM_000619.2	0.9999
Apolipoprotein A-II Deficiency	APOA2	RECESSIVE	NM_001643.1	0.9987
	TERT	RECESSIVE	NM_198253.2	0.9991
Apolipoprotein C-II Deficiency	APOC2	RECESSIVE	NM_000483.4	0.9986
Arginase Deficiency	ARG1	RECESSIVE	NM_000045	0.9997
Argininosuccinate Lyase Deficiency	ASL	RECESSIVE	NM_000048.3	0.9972
Aromatase Deficiency	CYP19A1	RECESSIVE	NM_031226.2	0.9978
Aromatic L-Amino Acid Decarboxylase Deficiency	DDC	RECESSIVE	NM_000790.3	0.9986
ARSACS	SACS	RECESSIVE	NM_014363.4	0.9990

Arterial Tortuosity Syndrome	SLC2A10	RECESSIVE	NM_030777.3	0.9995
Arthrogryposis Multiplex Congenita	MYH3	DOMINANT	NM_002470.3	0.9993
	TNNI2	DOMINANT	NM_003282.3	0.9942
	TNNT3	DOMINANT	NM_006757.3	0.9986
	TPM2	DOMINANT	NM_003289.3	0.9933
Arthrogryposis, Renal Dysfunction, and Cholestasis Syndrome	VPS33B	RECESSIVE	NM_018668.3	0.9991
Arts Syndrome	PRPS1	X_LINKED	NM_002764	0.9944
Arylsulfatase A Deficiency	ARSA	RECESSIVE	NM_000487	0.9874
Aspartylglucosaminuria	AGA	RECESSIVE	NM_000027	0.9997
Asphyxiating Thoracic Dystrophy	DYNC2H1	RECESSIVE	NM_001080463.1	0.9998
Ataxia Neuropathy Spectrum Disorders	C10orf2	DOMINANT	NM_021830.4	0.9991
	IFT80	RECESSIVE	NM_020800.2	0.9989
	TTC21B	RECESSIVE	NM_024753.4	0.9998
	WDR19	RECESSIVE	NM_025132.3	0.9998
Ataxia with Oculomotor Apraxia	APTX	RECESSIVE	NM_175073.2	0.9966
	SETX	RECESSIVE	NM_015046	0.9995
Ataxia with Vitamin E Deficiency	TTPA	RECESSIVE	NM_000370.3	0.9998
Ataxia-Telangiectasia	ATM	RECESSIVE	NM_000051.3	0.9980
Ataxia-Telangiectasia-Like Disorder	MRE11A	RECESSIVE	NM_005591.3	0.9996
Atelosteogenesis	SLC26A2	RECESSIVE	NM_000112.3	0.9998
Athabaskan Brainstem Dysgenesis Syndrome	HOXA1	RECESSIVE	NM_005522.4	0.9985
Atransferrinemia	TF	RECESSIVE	NM_001063.3	0.9994
Atrial Septal Defect	ACTC1	DOMINANT	NM_005159	0.9980
	MYH6	DOMINANT	NM_002471.3	0.9964
Atrichia with Papular Lesions	HR	RECESSIVE	NM_005144.4	0.9958
Atypical Gaucher Disease	PSAP	DOMINANT	NM_002778.2	0.9993
Atypical Hemolytic-Uremic Syndrome	C3	DOMINANT	NM_000064.2	0.9966
	CD46	RECESSIVE (Rec/Dom)	NM_002389.4	0.9998
	CFB	DOMINANT	NM_001710.5	0.9993
	CFH	DOMINANT (Dom/Rec)	NM_000186.3	0.9997
	CFI	DOMINANT	NM_000204.3	0.9999

Atypical Hemolytic-Uremic Syndrome	THBD	DOMINANT	NM_000361.2	0.9996
Atypical Werner Syndrome	LMNA	DOMINANT	NM_005572.3	0.9982
Auriculocondylar Syndrome	PLCB4	DOMINANT	NM_000933.3	0.9995
Autoimmune Lymphoproliferative Syndrome	CASP10	DOMINANT	NM_032977.3	0.9981
Autoinflammation, Lipodystrophy, and Dermatosis Syndrome	PSMB8	RECESSIVE	NM_148919.3	0.9987
	FAS	DOMINANT	NM_000043.4	0.9997
	FASLG	DOMINANT	NM_000639.1	0.9992
Axenfeld-Rieger Syndrome	PITX2	DOMINANT	NM_153427.2	0.9962
Axonal Neuropathy	GARS	DOMINANT	NM_002047	0.9997
Bardet-Biedl Syndrome	ARL6	RECESSIVE	NM_177976.1	0.9996
	BBS1	RECESSIVE	NM_024649.4	0.9992
	BBS10	RECESSIVE	NM_024685.3	0.9998
	BBS12	RECESSIVE	NM_152618.2	0.9977
	BBS2	RECESSIVE	NM_031885.3	0.9997
	BBS4	RECESSIVE	NM_033028.4	0.9995
	BBS7	RECESSIVE	NM_176824.2	0.9998
	BBS9	RECESSIVE	NM_198428.2	0.9996
	CEP290	RECESSIVE	NM_025114.3	0.9998
	MKKS	RECESSIVE	NM_018848.3	0.9998
	MKS1	RECESSIVE	NM_017777.3	0.9995
	SDCCAG8	RECESSIVE	NM_006642.3	0.9997
	TRIM32	RECESSIVE	NM_012210	0.9951
	TTC8	RECESSIVE	NM_198309.2	0.9997
	WDPCP	RECESSIVE	NM_015910.5	0.9968
Bare Lymphocyte Syndrome, Type II	CIITA	RECESSIVE	NM_000246.3	0.9990
	RFX5	RECESSIVE	NM_000449.3	0.9972
	RFXAP	RECESSIVE	NM_000538.3	0.9996
Bartter Syndrome	BSND	RECESSIVE	NM_057176	0.9990
Basal Laminar Drusen	CFH	DOMINANT	NM_000186.3	0.9997
Beare-Stevenson Syndrome	FGFR2	DOMINANT	NM_000141.4	0.9980
Benign Chronic Pemphigus	ATP2C1	DOMINANT	NM_014382.3	0.9953

Benign Familial Neonatal Infantile Seizures	SCN2A	DOMINANT	NM_021007.2	0.9983
Benign Familial Neonatal Seizures	KCNQ3	DOMINANT	NM_004519.3	0.9987
Benign Hereditary Chorea	NKX2-1	DOMINANT	NM_001079668.2	0.9965
Benign Neonatal Epilepsy	KCNQ3	DOMINANT	NM_004519.3	0.9987
Berardinelli-Seip Congenital Lipodystrophy	AGPAT2	RECESSIVE	NM_006412.3	0.9972
	BSCL2	RECESSIVE	NM_032667.6	0.9989
Bernard-Soulier Syndrome	GP9	RECESSIVE (Rec/Dom)	NM_000174.3	0.9993
Best Vitelliform Macular Dystrophy	BEST1	DOMINANT (Dom/Rec)	NM_004183	0.9992
Beta-Mannosidosis	MANBA	RECESSIVE	NM_005908	0.9963
Beta-Sarcoglycanopathy	SGCB	RECESSIVE	NM_000232	0.9998
Beta-Thalassemia	HBB	RECESSIVE	NM_000518	0.9997
Beta-Ureidopropionase Deficiency	UPB1	RECESSIVE	NM_016327.2	0.9997
BH4-Deficient Hyperphenylalaninemia	PCBD1	RECESSIVE	NM_000281.2	0.9995
	QDPR	RECESSIVE	NM_000320.2	0.9951
Bietti Crystalline Dystrophy	CYP4V2	RECESSIVE	NM_207352.3	0.9988
Biotinidase Deficiency	BTD	RECESSIVE	NM_000060	0.9995
Birk-Barel Intellectual Disability Dysmorphism Syndrome	KCNK9	DOMINANT	NM_016601.2	0.9991
Birt-Hogg-Dube Syndrome	FLCN	DOMINANT	NM_144997.5	0.9970
Blau Syndrome	NOD2	DOMINANT	NM_022162.1	0.9995
Bloom Syndrome	BLM	RECESSIVE	NM_000057.2	0.9997
BMP4-Related Syndromic Microphthalmia	BMP4	DOMINANT	NM_001202.3	0.9831
Bohring-Opitz Syndrome	ASXL1	DOMINANT (Dom/Rec)	NM_015338.5	0.9973
Bosley-Salih-Alorainy Syndrome	HOXA1	RECESSIVE	NM_005522.4	0.9985
Brachydactyly	BMPR1B	DOMINANT (Dom/Rec)	NM_001203.2	0.9970
	GDF5	DOMINANT (Dom/Rec)	NM_000557.2	0.9989
	IHH	DOMINANT	NM_002181.3	0.9982
	ROR2	DOMINANT	NM_004560.3	0.9990
	TRPV4	DOMINANT	NM_021625	0.9968
Brain Small Vessel Disease with Hemorrhage	COL4A1	DOMINANT	NM_001845.4	0.9993
Branchiootorenal Spectrum Disorders	EYA1	DOMINANT	NM_000503.4	0.9976
	SIX1	DOMINANT	NM_005982	0.9984

Breast and Ovarian Cancer Susceptibility	RAD51C	DOMINANT	NM_058216	0.9994
	RAD51D	DOMINANT	NM_002878.3	0.9994
Breast Cancer	BARD1	DOMINANT	NM_000465.2	0.9994
	BRIP1	DOMINANT	NM_032043	0.9975
	CHEK2	DOMINANT	NM_007194	0.9992
Brittle Cornea Syndrome	PRDM5	RECESSIVE	NM_018699.2	0.9995
	ZNF469	RECESSIVE	NM_001127464.1	0.9959
Brody Myopathy	ATP2A1	RECESSIVE (Rec/Dom)	NM_173201.3	0.9953
Brooke-Spiegler Syndrome	CYLD	DOMINANT	NM_015247.2	0.9995
Bruck Syndrome	PLOD2	RECESSIVE	NM_000935.2	0.9997
Brugada Syndrome	CACNA1C	DOMINANT	NM_000719	0.9986
	CACNB2	DOMINANT	NM_201590	0.9968
	GPD1L	DOMINANT	NM_015141	0.9984
	HCN4	DOMINANT	NM_005477.2	0.9963
	KCNE3	DOMINANT	NM_005472	0.9938
	SCN1B	DOMINANT	NM_001037	0.9951
	SCN3B	DOMINANT	NM_018400	0.9995
	SCN5A	DOMINANT	NM_198056	0.9983
Budd-Chiari Syndrome	F5	RECESSIVE (Rec/Dom)	NM_000130	0.9987
	JAK2	RECESSIVE (Rec/Dom)	NM_004972.3	0.9996
Buschke-Ollendorff Syndrome	LEMD3	DOMINANT	NM_014319.4	0.9996
Butyrylcholinesterase Deficiency	BCHE	RECESSIVE	NM_000055.2	0.9999
C Syndrome	CD96	DOMINANT (Dom/Rec)	NM_198196.2	0.9948
C3 Deficiency	C3	RECESSIVE	NM_000064.2	0.9966
Caffey Disease	COL1A1	DOMINANT	NM_000088.3	0.9902
Calpainopathy	CAPN3	RECESSIVE	NM_000070	0.9984
Campomelic Dysplasia	SOX9	DOMINANT	NM_000346.3	0.9964
Canavan Disease	ASPA	RECESSIVE	NM_000049	0.9998
Capillary Malformation-Arteriovenous Malformation Syndrome	RASA1	DOMINANT	NM_002890.2	0.9995
Carbamoylphosphate Synthetase I Deficiency	CPS1	RECESSIVE	NM_001875	0.9993
Cardiofaciocutaneous Syndrome	BRAF	DOMINANT	NM_004333.4	0.9934

Cardiofaciocutaneous Syndrome	KRAS	DOMINANT	NM_004985.3	0.9996
	MAP2K1	DOMINANT	NM_002755.3	0.9985
Cardiomyopathy, ARVC	DSC2	DOMINANT	NM_024422	0.9995
	DSG2	DOMINANT	NM_001943	0.9979
	DSP	DOMINANT	NM_004415	0.9996
	JUP	DOMINANT	NM_002230	0.9952
	PKP2	DOMINANT	NM_004572	0.9996
	RYR2	DOMINANT	NM_001035	0.9982
	TGFB3	DOMINANT	NM_003239.2	0.9994
	TMEM43	DOMINANT	NM_024334	0.9996
Carney Complex	PRKAR1A	DOMINANT	NM_002734.3	0.9996
Carnitine Palmitoyltransferase II Deficiency	CPT2	RECESSIVE	NM_000098	0.9995
Carnitine-Acylcarnitine Translocase Deficiency	SLC25A20	RECESSIVE	NM_000387	0.9995
Carpenter Syndrome	RAB23	RECESSIVE	NM_183227.1	0.9998
Caspase 8 Deficiency	CASP8	RECESSIVE	NM_001228.4	0.9963
Cataract-Microcornea Syndrome	GJA8	DOMINANT	NM_005267.4	0.9994
Cataracts	BFSP2	DOMINANT	NM_003571.2	0.9997
	CRYGD	DOMINANT	NM_006891.3	0.9992
	HSF4	DOMINANT	NM_001538.3	0.9947
	MIP	DOMINANT	NM_012064.3	0.9991
	PITX2	DOMINANT	NM_153427.2	0.9962
Catecholaminergic Polymorphic Ventricular Tachycardia	CASQ2	RECESSIVE	NM_001232.3	0.9987
	RYR2	DOMINANT	NM_001035	0.9982
	TRDN	RECESSIVE	NM_006073.3	0.9994
Caudal Dysgenesis Syndrome	VANGL1	DOMINANT	NM_138959.2	0.9996
Caveolinopathies	CAV3	DOMINANT (Dom/Rec)	NM_033337	0.9991
Cenani-Lenz Syndactyly Syndrome	LRP4	RECESSIVE	NM_002334.3	0.9980
Central Core Disease (Dominant/Recessive)	RYR1	DOMINANT (Dom/Rec)	NM_000540.2	0.9963
Centronuclear Myopathy, Dominant	DNM2	DOMINANT	NM_001005360.2	0.9984
	MYF6	RECESSIVE (Rec/Dom)	NM_002469.2	0.9994
Centronuclear Myopathy, Recessive	BIN1	RECESSIVE (Rec/Dom)	NM_139343.2	0.9981

Cerebellar Ataxia	SYNE1	RECESSIVE	NM_033071.3	0.9997
Cerebellar Ataxia, Cayman type	ATCAY	RECESSIVE	NM_033064.4	0.9974
Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL)	NOTCH3	DOMINANT	NM_000435.2	0.9978
Cerebral Dysgenesis, Neuropathy, Ichthyosis, and Palmoplantar Keratoderma Syndrome	SNAP29	RECESSIVE	NM_004782.3	0.9904
Cerebrooculofacioskeletal Syndrome	ERCC6	RECESSIVE	NM_000124.2	0.9998
Cerebrotendinous Xanthomatosis	CYP27A1	RECESSIVE	NM_000784	0.9990
CFTR-Related Disorders	CFTR	RECESSIVE	NM_000492	0.9981
Chanarin-Dorfman Syndrome	ABHD5	RECESSIVE	NM_016006.4	0.9991
Char Syndrome	TFAP2B	DOMINANT	NM_003221.3	0.9988
Charcot-Marie-Tooth with Vocal Cord Paresis	GDAP1	RECESSIVE	NM_018972	0.9995
Charcot-Marie-Tooth, Intermediate	DNM2	DOMINANT	NM_001005360.2	0.9984
	GDAP1	RECESSIVE	NM_018972	0.9995
	KARS	RECESSIVE	NM_001130089.1	0.9996
	MPZ	DOMINANT	NM_000530	0.9865
	YARS	DOMINANT	NM_003680.3	0.9989
Charcot-Marie-Tooth, Type 1	EGR2	DOMINANT	NM_000399	0.9992
	LITAF	DOMINANT	NM_004862	0.9985
	MPZ	DOMINANT	NM_000530	0.9865
	NEFL	DOMINANT	NM_006158	0.9993
	PMP22	DOMINANT	NM_000304	0.9994
Charcot-Marie-Tooth, Type 2	AARS	DOMINANT	NM_001605	0.9994
	DYNC1H1	DOMINANT	NM_001376.4	0.9994
	GARS	DOMINANT	NM_002047	0.9997
	HSPB1	DOMINANT	NM_001540	0.9955
	HSPB8	DOMINANT	NM_014365	0.9986
	KIF1B	DOMINANT	NM_015074	0.9987
	LMNA	DOMINANT	NM_005572.3	0.9982
	LRSAM1	DOMINANT (Dom/Rec)	NM_138361.5	0.9975
	MED25	RECESSIVE	NM_030973	0.9983

Charcot-Marie-Tooth, Type 2	MFN2	DOMINANT (Dom/Rec)	NM_014874	0.9976
	RAB7A	DOMINANT	NM_004637	0.9986
	TRPV4	DOMINANT	NM_021625	0.9968
Charcot-Marie-Tooth, Type 4	FGD4	RECESSIVE	NM_139241	0.9997
	FIG4	RECESSIVE	NM_014845	0.9995
	MTMR2	RECESSIVE	NM_016156	0.9997
	NDRG1	RECESSIVE	NM_006096	0.9984
	PRX	RECESSIVE	NM_181882.2	0.9941
	SBF2	RECESSIVE	NM_030962	0.9997
	SH3TC2	RECESSIVE	NM_024577	0.9985
Charcot-Marie-Tooth, X-linked	GJB1	X_LINKED	NM_000166.5	0.9961
	PRPS1	X_LINKED	NM_002764	0.9944
CHARGE Syndrome	CHD7	DOMINANT	NM_017780.3	0.9997
Chediak-Higashi Syndrome	LYST	RECESSIVE	NM_000081.2	0.9997
Cherubism	SH3BP2	DOMINANT (Dom/Rec)	NM_003023.4	0.9973
Chilblain Lupus	SAMHD1	DOMINANT	NM_015474.3	0.9996
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter	EIF2B1	RECESSIVE	NM_001414.3	0.9996
	EIF2B2	RECESSIVE	NM_014239.3	0.9981
	EIF2B3	RECESSIVE	NM_020365.4	0.9996
	EIF2B4	RECESSIVE	NM_015636.3	0.9992
	EIF2B5	RECESSIVE	NM_003907.2	0.9995
Childhood Restrictive Cardiomyopathy	ACTA1	DOMINANT	NM_001100.3	0.9974
	ACTC1	DOMINANT	NM_005159	0.9980
Chitotriosidase Deficiency	CHIT1	RECESSIVE	NM_003465.2	0.9992
Cholesterol Ester Storage Disease	LIPA	RECESSIVE	NM_000235	0.9997
Chondrocalcinosis	ANKH	DOMINANT	NM_054027.4	0.9994
Chondrodysplasia	GDF5	RECESSIVE	NM_000557.2	0.9989
	IMPAD1	RECESSIVE	NM_017813.4	0.9997
	PTH1R	RECESSIVE	NM_000316.2	0.9959
Chorea-acanthocytosis	VPS13A	RECESSIVE	NM_033305.2	0.9997

Choreoathetosis, Hypothyroidism, and Neonatal Respiratory Distress	NKX2-1	DOMINANT	NM_001079668.2	0.9965
Choroidal Dystrophy	PRPH2	DOMINANT (Dom/Rec)	NM_000322	0.9989
Chronic Granulomatous Disease	NCF2	RECESSIVE	NM_000433.3	0.9997
	NCF4	RECESSIVE	NM_013416.3	0.9992
Chronic Infantile Neurological Cutaneous and Articular Syndrome	NLRP3	DOMINANT	NM_004895	0.9994
CHST3-Related Skeletal Dysplasia	CHST3	RECESSIVE	NM_004273.4	0.9992
Citrin Deficiency	SLC25A13	RECESSIVE	NM_014251	0.9998
Citrullinemia	ASS1	RECESSIVE	NM_054012	0.9975
Cleft Lip +/- Cleft Palate, Autosomal Dominant	BMP4	DOMINANT	NM_001202.3	0.9959
	IRF6	DOMINANT	NM_006147.3	0.9995
	SUMO1	DOMINANT	NM_001005781.1	0.9998
	TP63	DOMINANT	NM_003722.4	0.9997
Cleft Lip +/- Cleft Palate, Autosomal Recessive	NECTIN1	RECESSIVE	NM_002855.4	0.9902
Cleft Palate, X-Linked	TBX22	X_LINKED	NM_001109878.1	0.9998
Cleidocranial Dysplasia	RUNX2	DOMINANT	NM_001024630.3	0.9989
Cockayne Syndrome	ERCC6	RECESSIVE	NM_000124.2	0.9998
	ERCC8	RECESSIVE	NM_000082.3	0.9996
Coenzyme Q10 deficiency, Oculomotor Apraxia Type	APTX	RECESSIVE	NM_175073.2	0.9966
Coenzyme Q10 deficiency, Spinocerebellar Ataxia Type	ADCK3	RECESSIVE	NM_020247.4	0.9983
Coffin-Siris Syndrome	SMARCA4	DOMINANT	NM_001128849.1	0.9984
	SMARCB1	DOMINANT	NM_003073.3	0.9914
Cohen Syndrome	VPS13B	RECESSIVE	NM_017890.4	0.9986
Collagen Type VI-Related Disorders	COL6A1	DOMINANT (Dom/Rec)	NM_001848	0.9918
	COL6A2	DOMINANT (Dom/Rec)	NM_001849	0.9975
	COL6A3	DOMINANT (Dom/Rec)	NM_004369	0.9988
Coloboma, Congenital Heart Disease, Ichthyosiform Dermatosis, Intellectual Disability, and Ear Anomalies Syndrome	PIGL	RECESSIVE	NM_004278.3	0.9996
Colorectal Cancer	CHEK2	DOMINANT	NM_007194	0.9992

Combined Deficiency of Factor V and Factor VIII	LMAN1	RECESSIVE	NM_005570.3	0.9995
	MCFD2	RECESSIVE	NM_139279.5	0.9906
Combined Oxidative Phosphorylation Deficiency	AARS2	RECESSIVE	NM_020745.3	0.9992
	AIFM1	X_LINKED	NM_004208.3	0.9948
	C12orf65	RECESSIVE	NM_152269.4	0.9995
	EARS2	RECESSIVE	NM_001083614.1	0.9995
	GFM1	RECESSIVE	NM_024996.5	0.9999
	MRPS16	RECESSIVE	NM_016065.3	0.9971
	MRPS22	RECESSIVE	NM_020191.2	0.9995
	TSFM	RECESSIVE	NM_001172696.1	0.9997
	TUFM	RECESSIVE	NM_003321.4	0.9994
Combined Pituitary Hormone Deficiency, Dominant	LHX4	DOMINANT	NM_033343.3	0.9979
	OTX2	DOMINANT	NM_172337.2	0.9995
Combined Pituitary Hormone Deficiency, Dominant/Recessive	HESX1	RECESSIVE (Rec/Dom)	NM_003865.2	0.9997
Combined Pituitary Hormone Deficiency, Recessive	LHX3	RECESSIVE	NM_014564.3	0.9985
	POU1F1	RECESSIVE (Rec/Dom)	NM_000306.2	0.9997
	PROP1	RECESSIVE	NM_006261.4	0.9835
Combined Saposin Deficiency	PSAP	RECESSIVE	NM_002778.2	0.9993
Common Variable Immune Deficiency, Dominant	TNFRSF13B	DOMINANT (Dom/Rec)	NM_012452.2	0.9955
Common Variable Immune Deficiency, Recessive	CD19	DOMINANT (Dom/Rec)	NM_001770.5	0.9988
	ICOS	RECESSIVE	NM_012092.3	0.9997
	TNFRSF13C	RECESSIVE	NM_052945.3	0.9983
Complement Component C2 Deficiency	C2	RECESSIVE	NM_000063.4	0.9989
Cone Dystrophy	GUCA1A	DOMINANT	NM_000409.3	0.9994
Cone-Rod Dystrophy, Dominant	CRX	DOMINANT	NM_000554	0.9939
	PITPNM3	DOMINANT	NM_031220.3	0.9876
	PROM1	DOMINANT	NM_006017.2	0.9995
	PRPH2	DOMINANT	NM_000322	0.9989
	RAX2	DOMINANT	NM_032753.3	0.9970
	RIMS1	DOMINANT	NM_014989.5	0.9997
	UNC119	DOMINANT	NM_005148.3	0.9970

Cone-Rod Dystrophy, Recessive	ADAM9	RECESSIVE	NM_003816.2	0.9995
	C8orf37	RECESSIVE	NM_177965.3	0.9993
	CDHR1	RECESSIVE	NM_033100.2	0.9990
	PDE6C	RECESSIVE	NM_006204.3	0.9997
	RPGRIP1	RECESSIVE	NM_020366.3	0.9995
	SEMA4A	RECESSIVE	NM_022367	0.9965
Congenital Adrenal Hyperplasia	CYP11B1	RECESSIVE	NM_000497.3	0.9992
	CYP17A1	RECESSIVE	NM_000102.3	0.9991
	HSD3B2	RECESSIVE	NM_000198.3	0.9994
	STAR	RECESSIVE	NM_000349.2	0.9920
Congenital Adrenal Insufficiency	CYP11A1	RECESSIVE (Rec/Dom)	NM_000781.2	0.9993
Congenital Afibrinogenemia	FGA	RECESSIVE	NM_021871.2	0.9998
	FGB	RECESSIVE	NM_005141.4	0.9990
	FGG	RECESSIVE	NM_000509.4	0.9999
Congenital Amegakaryocytic Thrombocytopenia	MPL	RECESSIVE	NM_005373.2	0.9972
Congenital Aural Atresia	TSHZ1	DOMINANT	NM_005786.5	0.9915
Congenital Bile Acid Synthesis Defect	AKR1D1	RECESSIVE	NM_005989.3	0.9997
	CYP7B1	RECESSIVE	NM_004820.3	0.9996
Congenital Cataract	AGK	RECESSIVE	NM_018238.3	0.9997
	CRYAA	RECESSIVE	NM_000394.2	0.9984
	FYCO1	RECESSIVE	NM_024513.3	0.9995
	TDRD7	RECESSIVE	NM_014290.2	0.9997
Congenital Central Hypoventilation Syndrome	PHOX2B	DOMINANT	NM_003924	0.9983
Congenital Contractural Arachnodactyly	FBN2	DOMINANT	NM_001999.3	0.9998
Congenital Disorders of Glycosylation	ALG11	RECESSIVE	NM_001004127.2	0.9998
	ALG12	RECESSIVE	NM_024105.3	0.9984
	ALG2	RECESSIVE	NM_033087.3	0.9998
	ALG3	RECESSIVE	NM_005787.5	0.9971
	ALG6	RECESSIVE	NM_013339.3	0.9998
	ALG8	RECESSIVE	NM_024079.4	0.9962
	ALG9	RECESSIVE	NM_024740.2	0.9957

Congenital Disorders of Glycosylation	B4GALT1	RECESSIVE	NM_001497.3	0.9966
	COG1	RECESSIVE	NM_018714.2	0.9916
	COG4	RECESSIVE	NM_015386.2	0.9988
	COG5	RECESSIVE	NM_006348.3	0.9996
	COG6	RECESSIVE	NM_020751.2	0.9998
	COG7	RECESSIVE	NM_153603.3	0.9963
	COG8	RECESSIVE	NM_032382.4	0.9994
	DDOST	RECESSIVE	NM_005216.4	0.9986
	DOLK	RECESSIVE	NM_014908.3	0.9985
	DPAGT1	RECESSIVE	NM_001382.3	0.9969
	DPM1	RECESSIVE	NM_003859.1	0.9942
	DPM2	RECESSIVE	NM_003863.3	0.9988
	DPM3	RECESSIVE	NM_153741.1	0.9996
	MGAT2	RECESSIVE	NM_002408.3	0.9995
	MOGS	RECESSIVE	NM_006302.2	0.9990
	MPDU1	RECESSIVE	NM_004870.3	0.9986
	MPI	RECESSIVE	NM_002435	0.9996
	PGM1	RECESSIVE	NM_002633.2	0.9992
	PMM2	RECESSIVE	NM_000303.2	0.9916
	RFT1	RECESSIVE	NM_052859.3	0.9993
	SLC35A1	RECESSIVE	NM_006416.4	0.9997
	SLC35C1	RECESSIVE	NM_018389.4	0.9935
	SRD5A3	RECESSIVE	NM_024592.4	0.9996
	TMEM165	RECESSIVE	NM_018475.3	0.9997
	TUSC3	RECESSIVE	NM_006765.3	0.9998
Congenital Dyserythropoietic Anemia	CDAN1	RECESSIVE	NM_138477.2	0.9987
	KLF1	DOMINANT	NM_006563.3	0.9992
	SEC23B	RECESSIVE	NM_006363.4	0.9996
Congenital Erythropoietic Porphyria	UROS	RECESSIVE	NM_000375.2	0.9996
Congenital Fiber-Type Disproportion	ACTA1	DOMINANT	NM_001100.3	0.9974
	TPM3	RECESSIVE	NM_152263.2	0.9980

Congenital Fibrosis of the Extraocular Muscles	KIF21A	DOMINANT	NM_017641.3	0.9997
Congenital Finnish Nephrosis	NPHS1	RECESSIVE	NM_004646.3	0.9990
Congenital Glutamine Deficiency	GLUL	RECESSIVE	NM_002065.5	0.9993
Congenital heart defects 1, Nonsyndromic, 1	ZIC3	X-LINKED	NM_003413.3	0.9934
Congenital Hypomyelinating Neuropathy	MPZ	RECESSIVE	NM_000530	0.9865
Congenital Hypomyelination	MPZ	RECESSIVE	NM_000530	0.9865
Congenital Hypothyroidism	DUOX2	RECESSIVE	NM_014080.4	0.9985
	IYD	RECESSIVE	NM_203395	0.9994
	PAX8	RECESSIVE	NM_003466	0.9991
	TPO	RECESSIVE	NM_000547	0.9994
	TSHB	RECESSIVE	NM_000549.3	0.9999
	TSHR	RECESSIVE	NM_000369	0.9994
Congenital Ichthyosis	ABCA12	RECESSIVE	NM_173076.2	0.9996
	ALOX12B	RECESSIVE	NM_001139.2	0.9982
	ALOXE3	RECESSIVE	NM_021628.2	0.9982
	CYP4F22	RECESSIVE	NM_173483.3	0.9902
	NIPAL4	RECESSIVE	NM_001099287.1	0.9993
	PNPLA1	RECESSIVE	NM_001145717.1	0.9987
	TGM1	RECESSIVE	NM_000359.2	0.9906
Congenital Indifference to Pain	SCN9A	RECESSIVE	NM_002977.3	0.9997
Congenital Insensitivity to Pain with Anhidrosis	NTRK1	RECESSIVE	NM_001012331.1	0.9982
Congenital Lactase Deficiency	LCT	RECESSIVE	NM_002299.2	0.9994
Congenital Muscular Dystrophy, alpha-dystroglycan related	ISPD	RECESSIVE	NM_001101426.3	0.9946
	LARGE	RECESSIVE	NM_004737.4	0.9988
	POMGNT1	RECESSIVE	NM_017739	0.9993
Congenital Muscular Dystrophy, CHKB-related	CHKB	RECESSIVE	NM_005198.4	0.9969
Congenital Muscular Dystrophy, ITGA7-related	ITGA7	RECESSIVE	NM_002206.2	0.9952
Congenital Muscular Dystrophy, LAMA2-related	LAMA2	RECESSIVE	NM_000426.3	0.9991
Congenital Muscular Dystrophy, LMNA-related	LMNA	DOMINANT	NM_005572.3	0.9982
Congenital Myasthenic Syndrome, Dominant/Recessive	CHRNA1	RECESSIVE	NM_000079.3	0.9996
	CHRNB1	RECESSIVE	NM_000747.2	0.9989

Congenital Myasthenic Syndrome, Dominant/Recessive	CHRND	RECESSIVE	NM_000751.2	0.9986
	CHRNE	RECESSIVE	NM_000080.3	0.9815
Congenital Myasthenic Syndrome, Recessive	COLQ	RECESSIVE	NM_005677.3	0.9968
	GFPT1	RECESSIVE	NM_002056.3	0.9997
	MUSK	RECESSIVE	NM_005592.3	0.9997
	RAPSN	RECESSIVE	NM_005055.4	0.9984
	SCN4A	RECESSIVE	NM_000334.4	0.9976
Congenital Neuromuscular Disease with Uniform Type 1 Fiber	RYR1	RECESSIVE	NM_000540.2	0.9963
Congenital Nuclear Cataract	CRYBB1	RECESSIVE	NM_001887.3	0.9994
	CRYBB3	RECESSIVE	NM_004076.3	0.9995
Congenital Stationary Night Blindness, Dominant	GNAT1	DOMINANT	NM_144499.2	0.9989
	PDE6B	DOMINANT	NM_000283	0.9942
	RHO	DOMINANT	NM_000539	0.9982
Congenital Stationary Night Blindness, Recessive	CABP4	RECESSIVE	NM_145200.3	0.9977
	GPR179	RECESSIVE	NM_001004334.2	0.9989
	LRIT3	RECESSIVE	NM_198506.3	0.9997
	SLC24A1	RECESSIVE	NM_004727.2	0.9995
	TRPM1	RECESSIVE	NM_002420.5	0.9997
Congenital Stationary Night Blindness, X-linked	NYX	X-LINKED	NM_022567.2	0.9988
Congenital Stromal Corneal Dystrophy	DCN	DOMINANT	NM_001920.3	0.9997
Congenital Sucrase-Isomaltase Deficiency	SI	RECESSIVE	NM_001041.3	0.9980
Congenital Vertical Talus	HOXD10	DOMINANT	NM_002148.3	0.9967
Corneal Dystrophy, Dominant	TGFBI	DOMINANT	NM_000358.2	0.9993
Corneal Dystrophy, Dominant/Recessive	TACSTD2	DOMINANT (Dom/Rec)	NM_002353.2	0.9969
Corneal Dystrophy, Recessive	CYP4V2	RECESSIVE	NM_207352.3	0.9988
	SLC4A11	RECESSIVE	NM_032034.3	0.9978
Corneal Fleck Dystrophy	PIKFYVE	DOMINANT	NM_015040.3	0.9998
Cornelia de Lange Syndrome	NIPBL	DOMINANT	NM_133433.3	0.9967
	SMC1A	X_LINKED	NM_006306.2	0.9982
	SMC3	DOMINANT	NM_005445.3	0.9998
Cortical Dysplasia-Focal Epilepsy Syndrome	CNTNAP2	RECESSIVE	NM_014141.5	0.9986

Cortical Pulverulent Cataract	LIM2	RECESSIVE	NM_030657.3	0.9994
Corticosterone Methyloxidase Type I Deficiency	CYP11B2	RECESSIVE	NM_000498.3	0.9988
Corticosterone Methyloxidase Type II Deficiency	CYP11B2	RECESSIVE	NM_000498.3	0.9988
Cranioectodermal Dysplasia	IFT122	RECESSIVE	NM_052985.2	0.9994
	IFT43	RECESSIVE	NM_052873.2	0.9994
	WDR19	RECESSIVE	NM_025132.3	0.9998
	WDR35	RECESSIVE	NM_001006657.1	0.9998
Craniofacial-Deafness-Hand Syndrome	PAX3	DOMINANT	NM_181457.3	0.9996
Craniometaphyseal Dysplasia	ANKH	DOMINANT	NM_054027.4	0.9994
Craniosynostosis	FGFR1	DOMINANT	NM_023110.2	0.9991
	FGFR2	DOMINANT	NM_000141.4	0.9980
	MSX2	DOMINANT	NM_002449.4	0.9993
Crigler-Najjar Syndrome	UGT1A1	RECESSIVE	NM_000463	0.9995
Crohn Disease	NOD2	DOMINANT	NM_022162.1	0.9995
Crouzon Syndrome	FGFR2	DOMINANT	NM_000141.4	0.9980
Cutaneous Malignant Melanoma, Dominant	CDK4	DOMINANT	NM_000075	0.9994
Cutis Laxa with Severe Pulmonary, Gastrointestinal, and Urinary Abnormalities	LTBP4	RECESSIVE	NM_003573.2	0.9986
Cutis Laxa, Dominant	ELN	DOMINANT	NM_000501.2	0.9931
Cutis Laxa, Dominant/Recessive	FBLN5	DOMINANT (Dom/Rec)	NM_006329.3	0.9994
Cutis Laxa, Recessive	ALDH18A1	RECESSIVE	NM_002860.3	0.9996
	ATP6V0A2	RECESSIVE	NM_012463.3	0.9995
	EFEMP2	RECESSIVE	NM_016938.4	0.9955
	PYCR1	RECESSIVE	NM_006907.2	0.9992
Cystathioninuria	CTH	RECESSIVE	NM_001902.5	0.9998
Cystic Fibrosis-Like Syndrome	SCNN1A	DOMINANT	NM_001038.5	0.9970
Cystinosis	CTNS	RECESSIVE	NM_004937	0.9955
Cystinuria	SLC3A1	RECESSIVE	NM_000341	0.9998
	SLC7A9	RECESSIVE	NM_014270	0.9990
Cytochrome P450 Oxidoreductase Deficiency	POR	RECESSIVE	NM_000941.2	0.9988
D-2-Hydroxyglutaric Aciduria	D2HGDH	RECESSIVE	NM_152783.3	0.9941

Danon Disease	LAMP2	X_LINKED	NM_002294	0.9997
Darier-White Disease	ATP2A2	DOMINANT	NM_001681.3	0.9963
Delta-Sarcoglycanopathy	SGCD	RECESSIVE	NM_000337	0.9990
Dementia, Deafness, and Sensory Neuropathy	DNMT1	DOMINANT	NM_001130823.1	0.9990
Dense Deposit Disease / Membranoproliferative Glomerulonephritis Type II	CFH	RECESSIVE	NM_000186.3	0.9997
	CFHR5	RECESSIVE	NM_030787.3	0.9989
Dent Disease	CLCN5	X_LINKED	NM_000084.2	0.9997
Desbuquois Dysplasia	CANT1	RECESSIVE	NM_138793.3	0.9988
Desminopathy	DES	RECESSIVE (Rec/Dom)	NM_001927	0.9986
Desmosterolosis	DHCR24	RECESSIVE	NM_014762.3	0.9952
Diabetes Mellitus, Insulin-Resistant, with Acanthosis Nigricans	INSR	RECESSIVE (Rec/Dom)	NM_000208.2	0.9917
Diabetes Mellitus, Neonatal, with Congenital Hypothyroidism	GLIS3	RECESSIVE	NM_001042413.1	0.9980
Diabetes Mellitus, Noninsulin-Dependent, with Acanthosis Nigricans and Hypertension	PPARG	DOMINANT	NM_015869.4	0.9950
Diamond-Blackfan Anemia	RPL11	DOMINANT	NM_000975.3	0.9997
	RPL35A	DOMINANT	NM_000996.2	0.9997
	RPL5	DOMINANT	NM_000969.3	0.9997
	RPS10	DOMINANT	NM_001014.4	0.9993
	RPS19	DOMINANT	NM_001022.3	0.9989
	RPS24	DOMINANT	NM_033022.3	0.9983
	RPS26	DOMINANT	NM_001029.3	0.9995
	RPS7	DOMINANT	NM_001011.3	0.9991
Diaphanospondylodysostosis	BMPER	RECESSIVE	NM_133468.4	0.9968
Diaphyseal Medullary Stenosis with Malignant Fibrous Histiocytoma	MTAP	DOMINANT	NM_002451.3	0.9992
Diarrhea with Microvillus Atrophy	MYO5B	RECESSIVE	NM_001080467.2	0.9962
Diastrophic Dysplasia	SLC26A2	RECESSIVE	NM_000112.3	0.9998
Dicarboxylicaminoaciduria	SLC1A1	RECESSIVE	NM_004170.5	0.9997
Diffuse Mesangial Sclerosis Syndromes (DMS)	WT1	DOMINANT	NM_024426.4	0.9923
Dihydropyrimidinase Deficiency	DPYS	RECESSIVE	NM_001385.2	0.9979
Dihydropyrimidine Dehydrogenase Deficiency	DPYD	RECESSIVE	NM_000110.3	0.9997

Dilated Cardiomyopathy, Dominant	ABCC9	DOMINANT	NM_005691	0.9880
	ACTC1	DOMINANT	NM_005159	0.9980
	ACTN2	DOMINANT	NM_001103	0.9991
	ANKRD1	DOMINANT	NM_014391.2	0.9930
	BAG3	DOMINANT	NM_004281.3	0.9992
	CSRP3	DOMINANT	NM_003476.4	0.9912
	CTF1	DOMINANT	NM_001330	0.9892
	DES	DOMINANT	NM_001927	0.9986
	DSG2	DOMINANT	NM_001943	0.9979
	EYA4	DOMINANT	NM_172105	0.9992
	LDB3	DOMINANT	NM_001080116	0.9928
	LMNA	DOMINANT	NM_005572.3	0.9982
	MYBPC3	DOMINANT	NM_000256	0.9947
	MYH6	DOMINANT	NM_002471.3	0.9964
	MYH7	DOMINANT	NM_000257	0.9987
	NEXN	DOMINANT	NM_144573	0.9997
	PLN	DOMINANT	NM_002667	0.9998
	PSEN1	DOMINANT	NM_000021	0.9997
	PSEN2	DOMINANT	NM_000447	0.9991
	RBM20	DOMINANT	NM_001134363	0.9970
	SCN5A	DOMINANT	NM_198056	0.9983
	SGCD	DOMINANT	NM_000337	0.9990
	TCAP	DOMINANT	NM_003673	0.9985
	TMPO	DOMINANT	NM_003276.2	0.9997
	TNNC1	DOMINANT	NM_003280	0.9989
	TNNT2	DOMINANT	NM_001001430	0.9991
	TPM1	DOMINANT	NM_001018005	0.9987
	TTN	DOMINANT	NM_133378	0.9885
	VCL	DOMINANT	NM_014000	0.9988
Dilated Cardiomyopathy, Recessive	FKTN	RECESSIVE	NM_001079802	0.9993
	TNNI3	RECESSIVE	NM_000363	0.9987

Dilated Cardiomyopathy, X-Linked	DMD	X_LINKED	NM_004006	0.9997
	TAZ	X_LINKED	NM_000116	0.9984
Disorders of Intracellular Cobalamin Metabolism	LMBRD1	RECESSIVE	NM_018368.3	0.9996
	MMACHC	RECESSIVE	NM_015506.2	0.9954
	MMADHC	RECESSIVE	NM_015702	0.9998
	MTR	RECESSIVE	NM_000254.2	0.9989
	MTRR	RECESSIVE	NM_002454.2	0.9995
Distal Arthrogryposis	MYBPC1	DOMINANT	NM_002465.3	0.9996
Distal Arthrogryposis Multiplex Congenita	TNNI2	DOMINANT	NM_003282.3	0.9942
	TNNT3	DOMINANT	NM_006757.3	0.9986
Distal Congenital Nonprogressive Spinal Muscular Atrophy	TRPV4	DOMINANT	NM_021625	0.9968
Distal Hereditary Motor Neuronopathy	DCTN1	DOMINANT	NM_004082.4	0.9990
	HSPB1	DOMINANT	NM_001540	0.9955
	HSPB3	DOMINANT	NM_006308.2	0.9997
	HSPB8	DOMINANT	NM_014365	0.9986
Distal Myopathy	MATR3	DOMINANT	NM_199189.2	0.9994
Distal Renal Tubular Acidosis with Progressive Nerve Deafness	ATP6V1B1	RECESSIVE	NM_001692.3	0.9939
Distal Renal Tubular Acidosis, Dominant	SLC4A1	DOMINANT	NM_000342.3	0.9913
Distal Renal Tubular Acidosis, Recessive	ATP6V0A4	RECESSIVE	NM_020632.2	0.9994
Distal Spinal Muscular Atrophy	GARS	DOMINANT	NM_002047	0.9997
	PLEKHG5	RECESSIVE	NM_020631.4	0.9965
Donnai-Barrow Syndrome	LRP2	RECESSIVE	NM_004525.2	0.9983
Donohue Syndrome	INSR	RECESSIVE	NM_000208.2	0.9917
Dopamine Beta-Hydroxylase Deficiency	DBH	RECESSIVE	NM_000787.3	0.9988
Dopa-Responsive Dystonia	GCH1	DOMINANT	NM_000161.2	0.9995
	SPR	RECESSIVE (Rec/Dom)	NM_003124.4	0.9995
Doyne Honeycomb Retinal Dystrophy	EFEMP1	DOMINANT	NM_001039348.2	0.9997
Dravet Syndrome	GABRG2	DOMINANT	NM_000816.3	0.9997
	SCN9A	DOMINANT	NM_002977.3	0.9997
Duane Syndrome	CHN1	DOMINANT	NM_001822.5	0.9996
Dubin-Johnson Syndrome	ABCC2	RECESSIVE	NM_000392.3	0.9996

Dyggve-Melchior-Clausen Syndrome	DYM	RECESSIVE	NM_017653.3	0.9989
Dysalbuminemic Hyperthyroxinemia	ALB	DOMINANT	NM_000477.5	0.9998
Dyschromatosis Symmetrica Hereditaria	ADAR	DOMINANT	NM_001111.4	0.9981
Dysferlinopathy	DYSF	RECESSIVE	NM_003494	0.9990
Dyskeratosis Congenita, Dominant	TINF2	DOMINANT	NM_001099274.1	0.9987
Dyskeratosis Congenita, Recessive	CTC1	RECESSIVE	NM_025099.5	0.9986
	NHP2	RECESSIVE	NM_017838.3	0.9983
	NOP10	RECESSIVE	NM_018648.3	0.9887
	TERT	RECESSIVE	NM_198253.2	0.9991
	WRAP53	RECESSIVE	NM_018081.2	0.9977
Dyssegmental Dysplasia	HSPG2	RECESSIVE	NM_005529.5	0.9966
Dystonia	PRKRA	RECESSIVE	NM_003690.4	0.9988
	SLC2A1	DOMINANT	NM_006516.2	0.9994
	THAP1	DOMINANT	NM_018105.2	0.9997
	TUBB4A	DOMINANT	NM_006087.2	0.9919
Dystonia/Parkinsonism, Hypermanganesemia, Polycythemia, and Chronic Liver Disease	SLC30A10	RECESSIVE	NM_018713.2	0.9995
Dystrophic Epidermolysis Bullosa	COL7A1	RECESSIVE (Rec/Dom)	NM_000094.3	0.9979
Early Infantile Epileptic Encephalopathy, Autosomal Dominant	SCN2A	DOMINANT	NM_021007.2	0.9983
	SCN8A	DOMINANT	NM_014191.3	0.9991
	SPTAN1	DOMINANT	NM_001130438.2	0.9990
	STXBP1	DOMINANT	NM_003165.3	0.9992
Early Infantile Epileptic Encephalopathy, Autosomal Recessive	PLCB1	RECESSIVE	NM_015192.2	0.9996
	SLC25A22	RECESSIVE	NM_024698.5	0.9967
Early Onset Familial Alzheimer Disease	APP	DOMINANT	NM_000484	0.9968
	PSEN1	DOMINANT	NM_000021	0.9997
	PSEN2	DOMINANT	NM_000447	0.9991
Early-Onset Primary Dystonia	TOR1A	DOMINANT	NM_000113.2	0.9982
Ectodermal Dysplasia, Anhidrotic, with T-cell Immunodeficiency	NFKBIA	DOMINANT	NM_020529.2	0.9989
Ectodermal Dysplasia/Skin Fragility Syndrome	DSP	RECESSIVE	NM_004415	0.9996
	PKP1	RECESSIVE	NM_001005337.2	0.9965

Ectopia Lentis	ADAMTSL4	RECESSIVE	NM_019032.4	0.9980
	FBN1	DOMINANT	NM_000138.4	0.9997
Ectrodactyly	TP63	DOMINANT	NM_003722.4	0.9997
EEM Syndrome	CDH3	RECESSIVE	NM_001793.4	0.9973
Ehlers-Danlos Syndrome, Arthrochalasia Type	COL1A2	DOMINANT	NM_000089	0.9995
Ehlers-Danlos Syndrome, Dermatosparaxis Type	ADAMTS2	RECESSIVE	NM_014244	0.9990
Ehlers-Danlos Syndrome, Kyphoscoliotic Form	PLOD1	RECESSIVE	NM_000302	0.9959
Ehlers-Danlos Syndrome, Type VIIA	COL1A1	DOMINANT	NM_000088.3	0.9902
	COL5A1	DOMINANT	NM_000093	0.9984
	COL5A2	DOMINANT	NM_000393	0.9997
Ehlers-Danlos Syndrome, Vascular Type	COL3A1	DOMINANT	NM_000090	0.9997
Elliptocytosis	SPTA1	DOMINANT	NM_003126	0.9996
	SPTB	DOMINANT	NM_000347	0.9968
Ellis-van Creveld Syndrome	EVC	RECESSIVE	NM_153717.2	0.9767
	EVC2	RECESSIVE	NM_147127.4	0.9992
Emery-Dreifuss Muscular Dystrophy	LMNA	DOMINANT	NM_005572.3	0.9982
	SYNE1	DOMINANT	NM_033071.3	0.9997
	SYNE2	DOMINANT	NM_182914.2	0.9985
Endocardial Fibroelastosis	TAZ	X_LINKED	NM_000116	0.9984
Enhanced S-Cone Syndrome	NR2E3	RECESSIVE	NM_014249	0.9986
Enlarged Parietal Foramina	ALX4	DOMINANT	NM_021926.3	0.9993
	MSX2	DOMINANT	NM_002449.4	0.9993
Epidermolysis Bullosa Simplex	KRT5	DOMINANT	NM_000424.3	0.9929
Epidermolysis Bullosa with Pyloric Atresia	ITGA6	RECESSIVE	NM_000210.2	0.9993
	ITGB4	RECESSIVE	NM_001005731.1	0.9923
Epidermolysis Bullosa, Lethal Acantholytic	DSP	RECESSIVE	NM_004415	0.9996
Epidermolytic Hyperkeratosis	KRT1	DOMINANT	NM_006121.3	0.9954
Epidermolytic Palmoplantar Keratoderma	KRT9	DOMINANT	NM_000226.3	0.9939
Epilepsy with Neurodevelopmental Defects	GRIN2A	DOMINANT	NM_000833.3	0.9981
Epileptic Encephalopathy	MAPK10	DOMINANT	NM_138982.2	0.9987
	PNKP	RECESSIVE	NM_007254.3	0.9892

Epimerase Deficiency Galactosemia	GALE	RECESSIVE	NM_000403.3	0.9987
Episodic Ataxia	CACNB4	DOMINANT	NM_000726.3	0.9992
	KCNA1	DOMINANT	NM_000217.2	0.9963
	SLC1A3	DOMINANT	NM_004172.4	0.9997
Erosive Vitreoretinopathy	VCAN	DOMINANT	NM_004385.4	0.9991
Erythrocyte AMP Deaminase Deficiency	AMPD3	RECESSIVE	NM_001025389.1	0.9983
Erythrokeratodermia Variabilis	GJB3	DOMINANT (Dom/Rec)	NM_024009	0.9981
Erythropoietic Protoporphyria	FECH	DOMINANT (Dom/Rec)	NM_000140.3	0.9991
Escobar Syndrome	CHRNG	RECESSIVE	NM_005199.4	0.9967
Essential Fructosuria	KHK	RECESSIVE	NM_000221.2	0.9987
Essential Thrombocythemia	MPL	DOMINANT	NM_005373.2	0.9972
	THPO	DOMINANT	NM_000460.2	0.9988
Ethylmalonic Encephalopathy	ETHE1	RECESSIVE	NM_014297.3	0.9038
Exocrine Pancreatic Insufficiency, Dyserythropoietic Anemia, and Calvarial Hyperostosis	COX4I2	RECESSIVE	NM_032609.2	0.9987
Fabry Disease	GLA	X_LINKED	NM_000169	0.9995
Factor V Cambridge Thrombophilia	F5	DOMINANT	NM_000130	0.9987
Factor V Deficiency	F5	RECESSIVE	NM_000130	0.9987
Factor V Leiden Thrombophilia	F5	DOMINANT	NM_000130	0.9987
Factor V R2 Mutation Thrombophilia	F5	DOMINANT	NM_000130	0.9987
Factor VII Deficiency	F7	RECESSIVE	NM_000131	0.9983
Factor VII Marburg I Variant Thrombophilia	HABP2	DOMINANT	NM_004132.3	0.9988
Factor X Deficiency	F10	RECESSIVE	NM_000504.3	0.9985
Factor XI Deficiency (Dominant/Recessive)	F11	RECESSIVE (Rec/Dom)	NM_000128	0.9994
Factor XII Deficiency	F12	RECESSIVE	NM_000505.3	0.9986
Factor XIII Subunit A Deficiency	F13A1	RECESSIVE	NM_000129.3	0.9996
Factor XIII Subunit B Deficiency	F13B	RECESSIVE	NM_001994.2	0.9997
Familial Atrial Fibrillation	ABCC9	DOMINANT	NM_005691	0.9880
	GJA5	DOMINANT	NM_005266.5	0.9996
	KCNA5	DOMINANT	NM_002234.3	0.9988
	KCNE2	DOMINANT	NM_172201	0.9998

Familial Atrial Fibrillation	KCNJ2	DOMINANT	NM_000891	0.9927
	KCNQ1	DOMINANT	NM_000218	0.9984
Familial Atypical Mycobacteriosis, Autosomal Dominant	STAT1	DOMINANT	NM_007315.3	0.9996
Familial Atypical Mycobacteriosis, Autosomal Recessive	IFNGR1	RECESSIVE	NM_000416.2	0.9996
	IL12B	RECESSIVE	NM_002187.2	0.9996
	IL12RB1	RECESSIVE	NM_005535.1	0.9984
	TYK2	RECESSIVE	NM_003331.4	0.9985
Familial Bone Marrow Failure	SRP72	DOMINANT	NM_006947.3	0.9980
Familial Candidiasis, Dominant	IL17F	DOMINANT	NM_052872.3	0.9998
Familial Candidiasis, Recessive	CARD9	RECESSIVE	NM_052813.4	0.9988
	IL17RA	RECESSIVE	NM_014339.6	0.9959
Familial Cerebral Cavernous Malformation	KRIT1	DOMINANT	NM_194456.1	0.9984
	PDCD10	DOMINANT	NM_145860.1	0.9991
Familial Chloride Diarrhea	SLC26A3	RECESSIVE	NM_000111.2	0.9997
Familial Cold Autoinflammatory Syndrome	NLRP12	DOMINANT	NM_144687	0.9964
	NLRP3	DOMINANT	NM_004895	0.9994
Familial Cylindromatosis	CYLD	DOMINANT	NM_015247.2	0.9995
Familial Dysautonomia	IKBKAP	RECESSIVE	NM_003640	0.9991
Familial Encephalopathy with Neuroserpin Inclusion Bodies	SERPINI1	DOMINANT	NM_005025.4	0.9998
Familial Erythrocytosis	EGLN1	DOMINANT	NM_022051.2	0.9994
	EPAS1	DOMINANT	NM_001430.4	0.9939
	EPOR	DOMINANT	NM_000121.3	0.9989
Familial Exudative Vitreoretinopathy	FZD4	DOMINANT	NM_012193.3	0.9993
	TSPAN12	DOMINANT	NM_012338.3	0.9994
Familial Febrile Seizures	SCN9A	DOMINANT	NM_002977.3	0.9990
Familial Hemiplegic Migraine	ATP1A2	DOMINANT	NM_000702.3	0.9997
	SCN1A	DOMINANT	NM_001165963.1	0.9992
Familial Hemophagocytic Lymphohistiocytosis	PRF1	RECESSIVE	NM_001083116.1	0.9998
	STX11	RECESSIVE	NM_003764.3	0.9994
	STXBP2	RECESSIVE	NM_006949.3	0.9976
	UNC13D	RECESSIVE	NM_199242.2	0.9991

Familial High Density Lipoprotein Deficiency	ABCA1	DOMINANT	NM_005502.3	0.9994
	APOA1	DOMINANT	NM_000039.1	0.9991
Familial Horizontal Gaze Palsy with Progressive Scoliosis	ROBO3	RECESSIVE	NM_022370.3	0.9971
Familial Hyperaldosteronism	KCNJ5	DOMINANT	NM_000890.3	0.9937
Familial Hypercholesterolemia	APOB	DOMINANT	NM_000384	0.9997
	LDLR	DOMINANT	NM_000527	0.9951
	LDLRAP1	RECESSIVE	NM_015627.2	0.9992
	PCSK9	DOMINANT	NM_174936	0.9984
Familial Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome	PRKAG2	DOMINANT	NM_016203	0.9936
	TNNI3	DOMINANT	NM_000363	0.9987
Familial Hypobetalipoproteinemia	APOB	RECESSIVE (Rec/Dom)	NM_000384	0.9997
	PCSK9	DOMINANT (Dom/Rec)	NM_174936	0.9984
Familial Hypocalciuric Hypercalcemia	CASR	DOMINANT	NM_000388.3	0.9994
Familial Idiopathic Basal Ganglia Calcification	SLC20A2	DOMINANT	NM_006749.4	0.9975
Familial Infantile Myoclonic Epilepsy	TBC1D24	RECESSIVE	NM_020705.2	0.9969
Familial Intrahepatic Cholestasis	ABCB11	RECESSIVE	NM_003742.2	0.9996
	ABCB4	RECESSIVE	NM_000443.3	0.9994
	ATP8B1	RECESSIVE	NM_005603.4	0.9993
Familial Isolated Hypoparathyroidism	CASR	DOMINANT	NM_000388.3	0.9994
	GCM2	RECESSIVE	NM_004752.3	0.9998
	PTH	DOMINANT (Dom/Rec)	NM_000315.2	0.9999
Familial Isolated Pituitary Adenomas	AIP	DOMINANT	NM_003977.2	0.9990
Familial Juvenile Hyperuricemic Nephropathy	REN	DOMINANT	NM_000537.3	0.9992
Familial Lipoprotein Lipase Deficiency	LPL	RECESSIVE	NM_000237.2	0.9995
Familial Mediterranean Fever	MEFV	RECESSIVE	NM_000243	0.9891
Familial Paroxysmal Nonkinesigenic Dyskinesia	PNKD	DOMINANT	NM_015488.4	0.9990
Familial Partial Lipodystrophy	LMNA	DOMINANT	NM_005572.3	0.9982
	PPARG	DOMINANT	NM_015869.4	0.9950
	PTRF	RECESSIVE	NM_012232.5	0.9967
Familial Periodic Fever	TNFRSF1A	DOMINANT	NM_001065.3	0.9992

Familial Platelet Disorder with Propensity to Acute Myelogenous Leukemia	RUNX1	DOMINANT	NM_001754.4	0.9954
Familial Pulmonary Fibrosis	SFTPC	DOMINANT	NM_003018.3	0.9992
	TERT	DOMINANT	NM_198253.2	0.9991
Familial Restrictive Cardiomyopathy	TNNI3	DOMINANT	NM_000363	0.9987
	TNNT2	DOMINANT	NM_001001430	0.9991
Familial Spinal Neurofibromatosis	NF1	DOMINANT	NM_000267	0.9956
Familial Temporal Lobe Epilepsy	CPA6	DOMINANT	NM_020361.4	0.9998
Familial Thrombotic Thrombocytopenia Purpura	ADAMTS13	RECESSIVE	NM_139025.3	0.9992
Familial Transthyretin Amyloidosis	TTR	DOMINANT	NM_000371	0.9998
Familial Visceral Amyloidosis	APOA1	DOMINANT	NM_000039.1	0.9991
	FGA	DOMINANT	NM_021871.2	0.9998
	LYZ	DOMINANT	NM_000239.2	0.9923
Fanconi Anemia	BRCA2	RECESSIVE	NM_000059	0.9998
	BRIP1	RECESSIVE	NM_032043	0.9975
	FANCA	RECESSIVE	NM_000135	0.9994
	FANCC	RECESSIVE	NM_000136	0.9988
	FANCD2	RECESSIVE	NM_033084	0.9987
	FANCE	RECESSIVE	NM_021922	0.9991
	FANCF	RECESSIVE	NM_022725	0.9995
	FANCG	RECESSIVE	NM_004629	0.9950
	FANCI	RECESSIVE	NM_001113378	0.9996
	FANCL	RECESSIVE	NM_001114636.1	0.9999
	FANCM	RECESSIVE	NM_020937	0.9998
	PALB2	RECESSIVE	NM_024675	0.9996
	RAD51C	RECESSIVE	NM_058216	0.9994
	SLX4	RECESSIVE	NM_032444	0.9980
Fanconi Anemia, X-Linked	FANCB	X_LINKED	NM_001018113	0.9998
Fanconi-Bickel Syndrome	SLC2A2	RECESSIVE	NM_000340.1	0.9998
Farber Lipogranulomatosis	ASAH1	RECESSIVE	NM_177924	0.9976
Fatal Infantile Cardioencephalomyopathy	SCO2	RECESSIVE	NM_005138.2	0.9991

Fatal Infantile Lactic Acidosis	SUCLG1	RECESSIVE	NM_003849.3	0.9966
Fatty Acid Hydroxylase-Associated Neurodegeneration	FA2H	RECESSIVE	NM_024306.4	0.9988
Fetal Akinesia Deformation Sequence	RAPSN	RECESSIVE	NM_005055.4	0.9984
Fibrochondrogenesis	COL11A1	RECESSIVE (Rec/Dom)	NM_001854.3	0.9996
	COL11A2	RECESSIVE (Rec/Dom)	NM_080680	0.9952
Fibrodysplasia Ossificans Progressiva	ACVR1	DOMINANT	NM_001105.4	0.9948
Fibular Hypoplasia and Complex Brachydactyly	GDF5	RECESSIVE	NM_000557.2	0.9989
FLNB-Related Spectrum Disorders	FLNB	DOMINANT (Dom/Rec)	NM_001457.3	0.9992
Floating-Harbor Syndrome	SRCAP	DOMINANT	NM_006662.2	0.9986
Focal Cortical Dysplasia of Taylor	TSC1	DOMINANT	NM_000368.4	0.9977
Focal Segmental Glomerulosclerosis	CD2AP	DOMINANT	NM_012120.2	0.9977
	INF2	DOMINANT	NM_022489.3	0.9811
	TRPC6	DOMINANT	NM_004621.5	0.9996
Formiminotransferase Deficiency	FTCD	RECESSIVE	NM_006657.2	0.9919
Foveal Hypoplasia and Presenile Cataract Syndrome	PAX6	DOMINANT	NM_000280.4	0.9984
Frank-ter Haar Syndrome	SH3PXD2B	RECESSIVE	NM_001017995.2	0.9967
Fraser Syndrome	FRAS1	RECESSIVE	NM_025074.6	0.9994
	FREM2	RECESSIVE	NM_207361.4	0.9995
	GRIP1	RECESSIVE	NM_021150.3	0.9992
Free Sialic Acid Storage Disorders	SLC17A5	RECESSIVE	NM_012434	0.9990
Freeman-Sheldon Syndrome	MYH3	DOMINANT (Dom/Rec)	NM_002470.3	0.9993
Frontotemporal Dementia	CHMP2B	DOMINANT	NM_014043	0.9997
	GRN	DOMINANT	NM_002087	0.9984
	TARDBP	DOMINANT	NM_007375.3	0.9997
Fructose 1,6 Bisphosphatase Deficiency	FBP1	RECESSIVE	NM_000507.3	0.9978
Fucosidosis	FUCA1	RECESSIVE	NM_000147	0.9990
Fukuyama Congenital Muscular Dystrophy	FKTN	RECESSIVE	NM_001079802	0.9993
Fumarate Hydratase Deficiency	FH	RECESSIVE	NM_000143.3	0.9993
Fundus Albipunctatus	PRPH2	DOMINANT (Dom/Rec)	NM_000322	0.9989
	RDH5	DOMINANT (Dom/Rec)	NM_002905.3	0.9996
	RLBP1	DOMINANT (Dom/Rec)	NM_000326.4	0.9995

Furlong Syndrome	TGFBR1	DOMINANT	NM_004612.2	0.9992
GABA-Transaminase Deficiency	ABAT	RECESSIVE	NM_020686.5	0.9965
Galactokinase Deficiency	GALK1	RECESSIVE	NM_000154	0.9990
Galactosemia	GALT	RECESSIVE	NM_000155	0.9994
Galactosialidosis	CTSA	RECESSIVE	NM_000308	0.9991
Gamma-Sarcoglycanopathy	SGCG	RECESSIVE	NM_000231	0.9963
Gastrointestinal Stromal Tumor	KIT	DOMINANT	NM_000222.2	0.9997
	PDGFRA	DOMINANT	NM_006206.4	0.9995
Geleophysic Dysplasia	ADAMTSL2	RECESSIVE	NM_014694.3	0.9991
	FBN1	DOMINANT	NM_000138.4	0.9997
Generalized Arterial Calcification of Infancy	ENPP1	RECESSIVE (Rec/Dom)	NM_006208.2	0.9956
Generalized Epilepsy and Paroxysmal Dyskinesia	KCNMA1	DOMINANT	NM_002247.3	0.9994
Generalized Epilepsy with Febrile Seizures Plus	GABRG2	DOMINANT	NM_000816.3	0.9997
	SCN1B	DOMINANT	NM_001037	0.9951
	SCN9A	DOMINANT	NM_002977.3	0.9997
Generalized Pustular Psoriasis	IL36RN	RECESSIVE	NM_012275.2	0.9983
Genetic Prion Diseases	PRNP	DOMINANT	NM_000311.3	0.9987
Geroderma Osteodysplasticum	GORAB	RECESSIVE	NM_152281.2	0.9998
Giant Axonal Neuropathy	GAN	RECESSIVE	NM_022041.3	0.9993
Gilbert Syndrome	UGT1A1	RECESSIVE	NM_000463	0.9995
Gingival Fibromatosis	SOS1	DOMINANT	NM_005633.3	0.9998
Gitelman Syndrome	SLC12A3	RECESSIVE	NM_000339.2	0.9962
Glaucoma	MYOC	DOMINANT	NM_000261.1	0.9997
Global Cerebral Hypomyelination	SLC25A12	RECESSIVE	NM_003705.4	0.9998
Glomuvenous Malformation	GLMN	DOMINANT	NM_053274.2	0.9993
Glucocorticoid Deficiency	MC2R	RECESSIVE	NM_000529.2	0.9955
	MRAP	RECESSIVE	NM_178817.3	0.9996
Glucocorticoid Resistance	NR3C1	DOMINANT	NM_001018077.1	0.9969
Glucocorticoid-Remediable Aldosteronism	CYP11B1	DOMINANT	NM_000497.3	0.9992
	CYP11B2	DOMINANT	NM_000498.3	0.9988
Glucose Transporter Type 1 Deficiency Syndrome	SLC2A1	DOMINANT	NM_006516.2	0.9994

Glucose-6-Phosphate Dehydrogenase Deficiency	G6PD	X_LINKED	NM_001042351	0.9943
Glucose-Galactose Malabsorption	SLC5A1	RECESSIVE	NM_000343.3	0.9994
Glutaric Acidemia	GCDH	RECESSIVE	NM_000159	0.9978
Glutathione Synthetase Deficiency	GSS	RECESSIVE	NM_000178.2	0.9993
Glycine Encephalopathy	AMT	RECESSIVE	NM_000481.3	0.9969
	GLDC	RECESSIVE	NM_000170.2	0.9995
Glycogen Storage Disease of Heart, Lethal Congenital	PRKAG2	RECESSIVE	NM_016203	0.9936
Glycogen Storage Disease Type 0, Liver	GYS2	RECESSIVE	NM_021957.3	0.9998
Glycogen Storage Disease Type 0, Muscle	GYS1	RECESSIVE	NM_002103.4	0.9990
Glycogen Storage Disease Type I	G6PC	RECESSIVE	NM_000151.3	0.9989
	SLC37A4	RECESSIVE	NM_001164277.1	0.9995
Glycogen Storage Disease Type III	AGL	RECESSIVE	NM_000642.2	0.9994
Glycogen Storage Disease Type IV	GBE1	RECESSIVE	NM_000158.3	0.9997
Glycogen Storage Disease Type V	PYGM	RECESSIVE	NM_005609.2	0.9990
Glycogen Storage Disease Type VI	PYGL	RECESSIVE	NM_002863.4	0.9992
Glycogen Storage Disease Type VII	PFKM	RECESSIVE	NM_000289.5	0.9992
Glycogen Storage Disease Type X	PGAM2	RECESSIVE	NM_000290.3	0.9970
Glycogen Storage Disease Type XIII	ENO3	RECESSIVE	NM_053013.3	0.9971
Glycogen Storage Disease Type XIV	PGM1	RECESSIVE	NM_002633.2	0.9992
Glycogen Storage Disease XI	LDHA	RECESSIVE	NM_005566.3	0.9996
Glycogen Storage Disease, Type II	GAA	RECESSIVE	NM_000152	0.9987
Glycoprotein 1a Deficiency	ITGA2	DOMINANT	NM_002203.3	0.9970
GM1 Gangliosidosis	GLB1	RECESSIVE	NM_000404	0.9993
GM2 Activator Deficiency	GM2A	RECESSIVE	NM_000405	0.9992
GNE-Related Myopathy	GNE	RECESSIVE	NM_005476.5	0.9981
Goldberg-Shprintzen Megacolon Syndrome	KIF1BP	RECESSIVE	NM_015634.3	0.9996
Gracile Syndrome	BCS1L	RECESSIVE	NM_004328.4	0.9989
Gray Platelet Syndrome	NBEAL2	RECESSIVE	NM_015175.2	0.9982
Greenberg Dysplasia	LBR	RECESSIVE	NM_002296.3	0.9995
Greig Cephalopolysyndactyly Syndrome	GLI3	DOMINANT	NM_000168.5	0.9984
Griscelli Syndrome	RAB27A	RECESSIVE	NM_004580.4	0.9941

Growth Retardation, Developmental Delay, Coarse Facies, and Early Death	FTO	RECESSIVE	NM_001080432.2	0.9991
GTP Cyclohydrolase 1 Deficiency (GTPCH)	GCH1	RECESSIVE	NM_000161.2	0.9995
Guanidinoacetate Methyltransferase Deficiency	GAMT	RECESSIVE	NM_000156	0.9972
Haim-Munk Syndrome	CTSC	RECESSIVE	NM_001814.4	0.9995
Hawkinsinuria	HPD	DOMINANT	NM_002150	0.9978
Hemoglobin E	HBB	RECESSIVE	NM_000518	0.9997
Hemolytic Anemia	SLC4A1	DOMINANT	NM_000342.3	0.9913
Hemophilia A, FVIII Deficiency	F8	X_LINKED	NM_000132.3	0.9841
Hemophilia B, Factor IX Deficiency	F9	X_LINKED	NM_000133.3	0.9997
Hennekam Lymphangiectasia-Lymphedema Syndrome	CCBE1	RECESSIVE	NM_133459.3	0.9946
Hepatic Failure, Early-Onset, and Neurologic Disorder due to Cytochrome C Oxidase Deficiency	SCO1	RECESSIVE	NM_004589.2	0.9996
Hepatic Lipase Deficiency	LIPC	RECESSIVE	NM_000236.2	0.9982
Hepatic Veno-occlusive Disease with Immunodeficiency	SP110	RECESSIVE	NM_004509.3	0.9995
Hepatocerebral Mitochondrial DNA Depletion Syndrome	MPV17	RECESSIVE	NM_002437.4	0.9992
Hereditary Angioedema	F12	DOMINANT	NM_000505.3	0.9986
	SERPING1	DOMINANT	NM_000062.2	0.9928
Hereditary Angiopathy with Nephropathy, Aneurysms, and Muscle Cramps	COL4A1	DOMINANT	NM_001845.4	0.9993
Hereditary Breast and Ovarian Cancer	BRCA1	DOMINANT	NM_007294	0.9986
	BRCA2	DOMINANT	NM_000059	0.9998
Hereditary Coproporphyria	CPOX	DOMINANT	NM_000097.5	0.9996
Hereditary Diffuse Gastric Cancer	CDH1	DOMINANT	NM_004360	0.9993
Hereditary Diffuse Leukoencephalopathy with Spheroids	CSF1R	DOMINANT	NM_005211.3	0.9977
Hereditary Essential Tremor	DRD3	DOMINANT	NM_000796.3	0.9996
Hereditary Folate Malabsorption	SLC46A1	RECESSIVE	NM_080669.4	0.9990
Hereditary Fructose Intolerance	ALDOB	RECESSIVE	NM_000035.3	0.9997
Hereditary Hemochromatosis	HFE	RECESSIVE	NM_000410	0.9982
	SLC40A1	DOMINANT	NM_014585	0.9999
	TFR2	RECESSIVE	NM_003227.3	0.9949

Hereditary Hemorrhagic Telangiectasia	ACVRL1	DOMINANT	NM_000020	0.9978
	ENG	DOMINANT	NM_000118	0.9978
	SMAD4	DOMINANT	NM_005359	0.9984
Hereditary Keratitis	PAX6	DOMINANT	NM_000280.4	0.9984
Hereditary Leiomyomatosis and Renal Cell Cancer	FH	DOMINANT	NM_000143.3	0.9993
Hereditary Motor and Sensory Neuropathy	MFN2	DOMINANT	NM_014874	0.9976
Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum	SLC12A6	RECESSIVE	NM_133647.1	0.9982
Hereditary Multiple Osteochondromatosis	EXT1	DOMINANT	NM_000127.2	0.9994
	EXT2	DOMINANT	NM_207122.1	0.9997
Hereditary Myopathy with Early Respiratory Failure	TTN	DOMINANT	NM_133378	0.9885
Hereditary Neuralgic Amyotrophy (HNA)	SEPT9	DOMINANT	NM_006640.4	0.9914
Hereditary Neuropathy with Liability to Pressure Palsies	PMP22	DOMINANT	NM_000304	0.9994
Hereditary Pancreatitis	CTRC	DOMINANT	NM_007272.2	0.9987
	SPINK1	RECESSIVE	NM_003122.3	0.9999
Hereditary Paraganglioma-Pheochromocytoma Syndrome	MAX	DOMINANT	NM_002382.4	0.9986
	SDHA	DOMINANT	NM_004168.2	0.9991
	SDHAF2	DOMINANT	NM_017841.2	0.9996
	SDHB	DOMINANT	NM_003000.2	0.9995
	SDHC	DOMINANT	NM_003001.3	0.9991
	SDHD	DOMINANT	NM_003002.2	0.9982
Hereditary Persistence of Fetal Hemoglobin	HBB	RECESSIVE	NM_000518	0.9997
	HBD	RECESSIVE	NM_000519.3	0.9997
Hereditary Pyropoikilocytosis	SPTA1	RECESSIVE	NM_003126	0.9996
Hereditary Sensory and Autonomic Neuropathy, Type IA	SPTLC1	DOMINANT	NM_006415.2	0.9984
Hereditary Sensory and Autonomic Neuropathy, Type IC	SPTLC2	DOMINANT	NM_004863.3	0.9994
Hereditary Sensory and Autonomic Neuropathy, Type II	FAM134B	RECESSIVE	NM_001034850.2	0.9990
	KIF1A	RECESSIVE	NM_004321.6	0.9979
	WNK1	RECESSIVE	NM_018979.3	0.9983
Hereditary Sensory and Autonomic Neuropathy, Type V	NGF	RECESSIVE	NM_002506.2	0.9990
Hereditary Sensory and Autonomic Neuropathy, Type VI	DST	RECESSIVE	NM_001723.5	0.9997

Hereditary Sideroblastic Anemia	SLC25A38	RECESSIVE	NM_017875.2	0.9996
Hereditary Sideroblastic Anemia with Myopathy and Lactic Acidosis	YARS2	RECESSIVE	NM_001040436.2	0.9985
Heritable Pulmonary Arterial Hypertension	BMPR2	DOMINANT	NM_001204.6	0.9975
	SMAD9	DOMINANT	NM_001127217.2	0.9971
Hermansky-Pudlak Syndrome	AP3B1	RECESSIVE	NM_003664.4	0.9996
	BLOC1S3	RECESSIVE	NM_212550.3	0.9957
	BLOC1S6	RECESSIVE	NM_012388.2	0.9996
	DTNBP1	RECESSIVE	NM_032122.4	0.9995
	HPS1	RECESSIVE	NM_000195.3	0.9976
	HPS3	RECESSIVE	NM_032383.3	0.9996
	HPS4	RECESSIVE	NM_022081.5	0.9974
	HPS5	RECESSIVE	NM_181507.1	0.9994
	HPS6	RECESSIVE	NM_024747.5	0.9994
Heterotaxy Syndrome	ACVR2B	DOMINANT	NM_001106.3	0.9963
	LEFTY2	DOMINANT	NM_003240.3	0.9930
	NODAL	DOMINANT	NM_018055.4	0.9995
Hexosaminidase A Deficiency	HEXA	RECESSIVE	NM_000520	0.9994
Hidrotic Ectodermal Dysplasia	GJB6	DOMINANT	NM_006783	0.9994
Hirschsprung Disease, Dominant	EDN3	DOMINANT	NM_207034.1	0.9983
	GDNF	DOMINANT	NM_000514.3	0.9958
	RET	DOMINANT	NM_020975	0.9990
Hirschsprung Disease, Recessive	EDNRB	RECESSIVE	NM_000115.3	0.9996
Histidinemia	HAL	RECESSIVE	NM_002108.3	0.9996
Holocarboxylase Synthetase Deficiency	HLCS	RECESSIVE	NM_000411.6	0.9963
Holoprosencephaly	CDON	DOMINANT	NM_016952.4	0.9977
	FOXH1	DOMINANT	NM_003923.2	0.9895
	GLI2	DOMINANT	NM_005270.4	0.9984
	NODAL	DOMINANT	NM_018055.4	0.9995
	PTCH1	DOMINANT	NM_000264.3	0.9972
	TGIF1	DOMINANT	NM_173208.1	0.9971

Holt-Oram Syndrome	TBX5	DOMINANT	NM_000192.3	0.9993
Homocystinuria	CBS	RECESSIVE	NM_000071.2	0.9954
Hutchinson-Gilford Progeria Syndrome	LMNA	DOMINANT (Dom/Rec)	NM_005572.3	0.9982
Hydrolethalus Syndrome	HYLS1	RECESSIVE	NM_145014.2	0.9861
Hydroxymethylbilane Synthase Deficiency	HMBS	DOMINANT	NM_000190.3	0.9966
Hyper IgD Syndrome	MVK	RECESSIVE	NM_000431.2	0.9993
Hyper IgE Syndrome	DOCK8	RECESSIVE	NM_203447.3	0.9980
	STAT3	DOMINANT	NM_139276.2	0.9917
Hyperalphalipoproteinemia	CETP	DOMINANT	NM_000078.2	0.9990
Hypercholanemia	BAAT	RECESSIVE	NM_001701.3	0.9997
Hyperekplexia	GLRA1	DOMINANT (Dom/Rec)	NM_000171.3	0.9991
	GLRB	DOMINANT (Dom/Rec)	NM_000824.4	0.9996
	SLC6A5	DOMINANT (Dom/Rec)	NM_004211.3	0.9990
Hypereosinophilic Syndrome	PDGFRA	DOMINANT	NM_006206.4	0.9995
Hyperferritinemia Cataract Syndrome	FTL	DOMINANT	NM_000146.3	0.9863
Hyperglycinuria	SLC6A20	DOMINANT	NM_020208.3	0.9972
Hypergonadotropic Hypogonadism	LHCGR	RECESSIVE	NM_000233.3	0.9995
Hyperinsulinism, Dominant	GCK	DOMINANT	NM_000162	0.9990
	GLUD1	DOMINANT	NM_005271.3	0.9987
	HNF4A	DOMINANT	NM_000457.4	0.9914
	SLC16A1	DOMINANT	NM_003051	0.9990
Hyperinsulinism, Dominant/Recessive	ABCC8	RECESSIVE	NM_000352	0.9992
	HADH	RECESSIVE	NM_005327.4	0.9996
	KCNJ11	RECESSIVE	NM_000525	0.9936
Hyperkalemic Periodic Paralysis	SCN4A	DOMINANT	NM_000334.4	0.9976
Hypermethioninemia	ADK	RECESSIVE	NM_001123.3	0.9997
	AHCY	RECESSIVE	NM_000687.2	0.9991
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	SLC25A15	RECESSIVE	NM_014252.3	0.9996
Hyperparathyroidism	MEN1	DOMINANT	NM_130799.2	0.9953
Hyperparathyroidism-Jaw Tumor Syndrome	CDC73	DOMINANT	NM_024529.4	0.9999

Hyperphosphatasia with Intellectual Disability Syndrome	PIGO	RECESSIVE	NM_032634.3	0.9981
	PIGV	RECESSIVE	NM_017837.3	0.9994
Hyperphosphatemic Familial Tumoral Calcinosis	FGF23	RECESSIVE	NM_020638.2	0.9931
	GALNT3	RECESSIVE	NM_004482.3	0.9998
	KL	RECESSIVE	NM_004795.3	0.9995
Hyperpigmentation, Cutaneous, with Hypertrichosis, Hepatosplenomegaly, Heart Anomalies, Hearing Loss, and Hypogonadism	SLC29A3	RECESSIVE	NM_018344.5	0.9991
Hyperprolinemia	ALDH4A1	RECESSIVE	NM_003748.3	0.9962
Hypertrichotic Osteochondrodysplasia	ABCC9	RECESSIVE	NM_005691	0.9880
Hypertrophic Cardiomyopathy	ACTC1	DOMINANT	NM_005159	0.9980
	ACTN2	DOMINANT	NM_001103	0.9991
	CAV3	DOMINANT	NM_033337	0.9991
	CSRP3	DOMINANT	NM_003476.4	0.9912
	MYBPC3	DOMINANT	NM_000256	0.9947
	MYH6	DOMINANT	NM_002471.3	0.9964
	MYH7	DOMINANT	NM_000257	0.9987
	MYL2	DOMINANT	NM_000432	0.9992
	MYL3	DOMINANT	NM_000258	0.9983
	MYLK2	DOMINANT	NM_033118.3	0.9955
	MYOZ2	DOMINANT	NM_016599	0.9998
	NEXN	DOMINANT	NM_144573	0.9997
	PLN	DOMINANT	NM_002667	0.9998
	TCAP	DOMINANT	NM_003673	0.9985
	TNNC1	DOMINANT	NM_003280	0.9989
	TNNI3	DOMINANT	NM_000363	0.9987
	TNNT2	DOMINANT	NM_001001430	0.9991
	TPM1	DOMINANT	NM_001018005	0.9987
	TTN	DOMINANT	NM_133378	0.9885
Hyperuricemia, Pulmonary Hypertension, Renal Failure, and Alkalosis	SARS2	RECESSIVE	NM_017827.3	0.9984

Hypocalcemia	CASR	DOMINANT	NM_000388.3	0.9994
Hypochromic Microcytic Anemia with Iron Overload	SLC11A2	RECESSIVE	NM_000617.2	0.9991
Hypohidrotic Ectodermal Dysplasia, Dominant	EDAR	RECESSIVE	NM_022336.3	0.9992
Hypohidrotic Ectodermal Dysplasia, Recessive	EDARADD	RECESSIVE	NM_145861.2	0.9974
Hypokalemic Periodic Paralysis	CACNA1S	DOMINANT	NM_000069.2	0.9979
	SCN4A	DOMINANT	NM_000334.4	0.9976
Hypomagnesemia with Secondary Hypocalcemia	TRPM6	RECESSIVE	NM_017662.4	0.9996
Hypomyelination and Congenital Cataract	FAM126A	RECESSIVE	NM_032581.3	0.9996
Hypoparathyroidism, Sensorineural Deafness, and Renal Disease	GATA3	DOMINANT	NM_001002295.1	0.9961
Hypoparathyroidism-Retardation-Dysmorphism Syndrome	TBCE	RECESSIVE	NM_003193.3	0.9989
Hypophosphatasia	ALPL	DOMINANT (Dom/Rec)	NM_000478.4	0.9947
Hypophosphatemic Nephrolithiasis/Osteoporosis	SLC34A1	DOMINANT	NM_003052.4	0.9962
Hypophosphatemic Rickets, Dominant	FGF23	DOMINANT	NM_020638.2	0.9931
Hypophosphatemic Rickets, Recessive	DMP1	RECESSIVE	NM_004407.3	0.9996
	ENPP1	RECESSIVE	NM_006208.2	0.9956
Hypoplastic Left Heart Syndrome	GJA1	RECESSIVE	NM_000165.3	0.9996
Ichthyosis Bullosa of Siemens	KRT2	DOMINANT	NM_000423.2	0.9991
Ichthyosis, Hystrix-like, with Deafness	GJB2	DOMINANT	NM_004004	0.9995
Immunodeficiency due to Defect in CD3-Gamma	CD3G	RECESSIVE	NM_000073.2	0.9941
Immunodeficiency with Hyper-IgM	AICDA	RECESSIVE	NM_020661.2	0.9939
	CD40	RECESSIVE	NM_001250.4	0.9984
	UNG	RECESSIVE	NM_080911.2	0.9988
Immunodeficiency-Centromeric Instability-Facial Anomalies Syndrome	DNMT3B	RECESSIVE	NM_006892.3	0.9985
Inclusion Body Myopathy, Dominant	MYH2	RECESSIVE	NM_017534.5	0.9997
	VCP	DOMINANT	NM_007126.3	0.9994
Infantile Hypercalcemia	CYP24A1	RECESSIVE	NM_000782.4	0.9975
Infantile Nystagmus	FRMD7	X-LINKED	NM_194277.2	0.9999
Infantile Spinal Muscular Atrophy, X-Linked	UBA1	X-LINKED	NM_003334	0.9994
Inflammatory Bowel Disease	IL10RA	RECESSIVE	NM_001558.3	0.9993

Inflammatory Bowel Disease	IL10RB	RECESSIVE	NM_000628.4	0.9941
Inherited Erythromelalgia	SCN9A	DOMINANT	NM_002977.3	0.9997
Inherited Systemic Hyalinosis	ANTXR2	RECESSIVE	NM_058172.5	0.9995
Insulin-Like Growth Factor I Deficiency	IGF1	RECESSIVE	NM_000618.3	0.9997
Insulin-Like Growth Factor I Resistance	IGF1R	RECESSIVE	NM_000875.3	0.9969
Intellectual Disability Syndrome	KANSL1	DOMINANT	NM_001193466.1	0.9890
Intellectual Disability with Language Impairment and Autistic Features	FOXP1	DOMINANT	NM_032682.5	0.9885
Intellectual Disability, Dominant	DYNC1H1	DOMINANT	NM_001376.4	0.9994
	GRIN2B	DOMINANT	NM_000834.3	0.9993
	KIF1A	DOMINANT	NM_004321.6	0.9979
	MBD5	DOMINANT	NM_018328.4	0.9994
Intellectual Disability, Recessive	MAN1B1	RECESSIVE	NM_016219.4	0.9989
	NSUN2	RECESSIVE	NM_017755.5	0.9989
	PRSS12	RECESSIVE	NM_003619.3	0.9992
	TRAPPC9	RECESSIVE	NM_031466.5	0.9967
Intellectual Disability, Stereotypic Movements, Epilepsy, and/or Cerebral Malformations	MEF2C	DOMINANT	NM_002397.4	0.9962
Intellectual Disability, X-linked	ARHGEF6	X_LINKED	NM_004840.2	0.9995
	BRWD3	X_LINKED	NM_153252.4	0.9982
	IL1RAPL1	X_LINKED	NM_014271.3	0.9993
	PAK3	X_LINKED	NM_002578.3	0.9985
	RAB39B	X_LINKED	NM_171998.2	0.9994
	YP	X_LINKED	NM_003179.2	0.9979
	UF3B	X_LINKED	NM_080632.2	0.9972
	ZN41	X_LINKED	NM_153380.2	0.9994
	ZNF74	X_LINKED	NM_001039891.2	0.9992
	ZNF71	X_LINKED	NM_021998.4	0.9991
	ZNF8	X_LINKED	NM_007137.3	0.9995
Interleukin 1 Receptor Antagonist Deficiency	IL1RN	RECESSIVE	NM_173841.2	0.9997
Interleukin 2 Receptor Alpha Chain Deficiency	IL2RA	RECESSIVE	NM_000417.2	0.9996

Intrinsic Factor Deficiency	GIF	RECESSIVE	NM_005142.2	0.9997
IRAK4 Deficiency	IRAK4	RECESSIVE	NM_016123.3	0.9986
Iris Hypoplasia	PITX2	DOMINANT	NM_153427.2	0.9962
Iron Overload	FTH1	DOMINANT	NM_002032.2	0.9997
Iron-Refractory Iron Deficiency Anemia	TMPRSS6	RECESSIVE	NM_153609.2	0.9957
Isobutyryl-CoA Dehydrogenase Deficiency	ACAD8	RECESSIVE	NM_014384.2	0.9994
Isolated Congenital Digital Clubbing	HPGD	RECESSIVE	NM_000860.5	0.9996
Isolated Coronal Synostosis	FGFR2	DOMINANT	NM_000141.4	0.9980
Isolated Follicle-Stimulating Hormone Deficiency	FSHB	RECESSIVE	NM_000510.2	0.9998
Isolated GnRH Deficiency	GNRH1	RECESSIVE	NM_000825.3	0.9998
	GNRHR	RECESSIVE	NM_000406.2	0.9992
	TACR3	RECESSIVE	NM_001059.2	0.9996
Isolated Growth Hormone Deficiency	BTK	X-LINKED	NM_000061.2	0.9998
	GHRHR	RECESSIVE	NM_000823.3	0.9992
Isolated Growth Hormone Deficiency (Dominant/Recessive)	GH1	RECESSIVE	NM_000515.3	0.9992
Isolated Hyperparathyroidism	CDC73	DOMINANT	NM_024529.4	0.9999
Isolated Microphthalmia	MFRP	RECESSIVE	NM_031433.2	0.9974
	VSX2	RECESSIVE	NM_182894.2	0.9988
Isolated Nonsyndromic Congenital Heart Disease	JAG1	DOMINANT	NM_000214	0.9993
Isolated Persistent Hypermethioninemia	MAT1A	RECESSIVE	NM_000429	0.9993
Isovaleric Acidemia	IVD	RECESSIVE	NM_002225	0.9984
Jackson-Weiss Syndrome	FGFR2	DOMINANT	NM_000141.4	0.9980
Jalili Syndrome	CNNM4	RECESSIVE	NM_020184.3	0.9983
Jawad Syndrome	RBBP8	RECESSIVE	NM_002894.2	0.9995
Jervell and Lange-Nielsen Syndrome	KCNE1	RECESSIVE	NM_000219	0.9993
	KCNQ1	RECESSIVE	NM_000218	0.9984
Joubert Syndrome	AHI1	RECESSIVE	NM_017651.4	0.9997
	CEP41	RECESSIVE	NM_018718.2	0.9957
	INPP5E	RECESSIVE	NM_019892.4	0.9943
	NPHP1	RECESSIVE	NM_000272.3	0.9978
	RPGRIP1L	RECESSIVE	NM_015272.2	0.9995

Joubert Syndrome	TCTN1	RECESSIVE	NM_001082538.2	0.9996
	TCTN2	RECESSIVE	NM_024809.4	0.9945
	TMEM138	RECESSIVE	NM_016464.4	0.9993
	TMEM216	RECESSIVE	NM_001173990.2	0.9995
	TMEM237	RECESSIVE	NM_001044385.2	0.9998
	TMEM67	RECESSIVE	NM_153704.5	0.9989
	TTC21B	RECESSIVE	NM_024753.4	0.9998
Junctional Epidermolysis Bullosa	COL17A1	RECESSIVE	NM_000494.3	0.9990
	LAMA3	RECESSIVE	NM_000227.3	0.9995
	LAMB3	RECESSIVE	NM_000228.2	0.9994
	LAMC2	RECESSIVE	NM_005562.2	0.9992
Juvenile Hereditary Hemochromatosis	HAMP	RECESSIVE	NM_021175	0.9995
	HFE2	RECESSIVE	NM_213653	0.9993
Juvenile Myoclonic Epilepsy (Dominant/Recessive)	CACNB4	RECESSIVE	NM_000726.3	0.9992
	EFHC1	RECESSIVE	NM_018100.3	0.9974
	GABRA1	RECESSIVE	NM_000806.5	0.9946
Juvenile Paget Disease	TNFRSF11B	RECESSIVE	NM_002546.3	0.9989
Juvenile Polyposis	BMPR1A	DOMINANT	NM_004329.2	0.9992
	ENG	DOMINANT	NM_000118	0.9978
	SMAD4	DOMINANT	NM_005359	0.9984
Kabuki Syndrome	KMT2D	DOMINANT	NM_003482.3	0.9887
Kallmann Syndrome	CHD7	DOMINANT	NM_017780.3	0.9986
	FGFR1	DOMINANT	NM_023110.2	0.9991
	PROKR2	DOMINANT	NM_144773.2	0.9992
Kanzaki disease	NAGA	RECESSIVE	NM_000262	0.9995
KAT6B-Related Spectrum Disorders	KAT6B	DOMINANT	NM_012330.3	0.9989
KCNQ2-Related Disorders	KCNQ2	DOMINANT	NM_172107.2	0.9908
Keratitis-Ichthyosis-Deafness Syndrome	GJB2	DOMINANT	NM_004004	0.9995
Ketothiolase Deficiency	ACAT1	RECESSIVE	NM_000019.3	0.9955
Keutel Syndrome	MGP	RECESSIVE	NM_000900.3	0.9997
Kindler Syndrome	FERMT1	RECESSIVE	NM_017671.4	0.9989

Kleefstra Syndrome	EHMT1	DOMINANT	NM_024757.4	0.9993
Klippel-Feil Syndrome	GDF6	DOMINANT	NM_001001557.2	0.9961
Knobloch Syndrome	COL18A1	RECESSIVE	NM_130445.2	0.9978
Kohlschutter-Tonz syndrome	ROGDI	RECESSIVE	NM_024589.1	0.9988
Krabbe Disease	GALC	RECESSIVE	NM_000153	0.9997
	PSAP	RECESSIVE	NM_002778.2	0.9993
Kufor-Rakeb Syndrome	ATP13A2	RECESSIVE	NM_022089.2	0.9979
Lacrimo-Auriculo-Dento-Digital Syndrome	FGF10	DOMINANT	NM_004465.1	0.9984
	FGFR2	DOMINANT	NM_000141.4	0.9980
Lactate Dehydrogenase B Deficiency	LDHB	RECESSIVE	NM_002300.6	0.9995
Lactose Intolerance	LCT	RECESSIVE	NM_002299.2	0.9994
Laing Distal Myopathy	MYH7	DOMINANT	NM_000257	0.9987
L-Arginine:Glycine Amidinotransferase Deficiency	GATM	RECESSIVE	NM_001482	0.9995
Laryngoonychocutaneous Syndrome	LAMA3	RECESSIVE	NM_000227.3	0.9995
Lathosterolosis	SC5D	RECESSIVE	NM_006918.4	0.9976
LCHAD Deficiency	HADHA	RECESSIVE	NM_000182	0.9995
Leber Congenital Amaurosis	AIPL1	RECESSIVE	NM_014336.3	0.9991
	CEP290	RECESSIVE	NM_025114.3	0.9998
	CRB1	RECESSIVE	NM_201253	0.9998
	CRX	DOMINANT	NM_000554	0.9939
	IMPDH1	DOMINANT	NM_000883.3	0.9984
	KCNJ13	RECESSIVE	NM_002242.4	0.9998
	LCA5	RECESSIVE	NM_181714.3	0.9998
	LRAT	RECESSIVE	NM_004744	0.9975
	RD3	RECESSIVE	NM_183059.2	0.9989
	RPE65	RECESSIVE	NM_000329	0.9998
	RPGRIP1	RECESSIVE	NM_020366.3	0.9995
	SPATA7	RECESSIVE	NM_018418.4	0.9983
	TULP1	RECESSIVE	NM_003322	0.9977
Lecithin Cholesterol Acyltransferase Deficiency	LCAT	RECESSIVE	NM_000229.1	0.9986
Left Ventricular Noncompaction Cardiomyopathy	ACTC1	DOMINANT	NM_005159	0.9980

Left Ventricular Noncompaction Cardiomyopathy	DTNA	DOMINANT	NM_032978	0.9997
	LDB3	DOMINANT	NM_001080116	0.9928
	MYBPC3	DOMINANT	NM_000256	0.9947
	MYH7	DOMINANT	NM_000257	0.9987
	TAZ	DOMINANT	NM_000116	0.9984
	TNNT2	DOMINANT	NM_001001430	0.9991
Legius Syndrome	SPRED1	DOMINANT	NM_152594.2	0.9993
Leigh Syndrome	BCS1L	RECESSIVE	NM_004328.4	0.9989
	COX10	RECESSIVE	NM_001303.3	0.9984
	DLD	RECESSIVE	NM_000108.3	0.9998
	LRPPRC	RECESSIVE	NM_133259.3	0.9985
	NDUFA10	RECESSIVE	NM_004544.3	0.9979
	NDUFAF2	RECESSIVE	NM_174889.4	0.9998
	NDUFS1	RECESSIVE	NM_005006.6	0.9982
	NDUFS3	RECESSIVE	NM_004551.2	0.9993
	NDUFS4	RECESSIVE	NM_002495.2	0.9997
	NDUFS7	RECESSIVE	NM_024407.4	0.9849
	NDUFS8	RECESSIVE	NM_002496.3	0.9987
	NDUFV1	RECESSIVE	NM_007103.3	0.9973
	SCO1	RECESSIVE	NM_004589.2	0.9996
	SDHA	RECESSIVE	NM_004168.2	0.9991
	SURF1	RECESSIVE	NM_003172.2	0.9994
LEOPARD Syndrome	BRAF	DOMINANT	NM_004333.4	0.9934
	PTPN11	DOMINANT	NM_002834.3	0.9996
	RAF1	DOMINANT	NM_002880.3	0.9991
Leptin Deficiency	LEP	RECESSIVE	NM_000230.2	0.9987
Leptin Receptor Deficiency	LEPR	RECESSIVE	NM_002303.5	0.9985
Lethal Arthrogryposis With Anterior Horn Cell Disease	GLE1	RECESSIVE	NM_001003722.1	0.9996
Lethal Congenital Contracture Syndrome	GLE1	RECESSIVE	NM_001003722.1	0.9996
Lethal Encephalopathy	DNM1L	DOMINANT	NM_012062.3	0.9993
Lethal Restrictive Dermopathy	LMNA	RECESSIVE	NM_005572.3	0.9982

Lethal Restrictive Dermopathy	ZMPSTE24	RECESSIVE	NM_005857.4	0.9998
Leukocyte Adhesion Deficiency	ITGB2	RECESSIVE	NM_000211.3	0.9974
Leukodystrophy, Adult-Onset	LMNB1	DOMINANT	NM_005573.3	0.9939
Leukodystrophy, hypomyelinating 6	TUBB4A	DOMINANT	NM_006087.2	0.9919
Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation	DARS2	RECESSIVE	NM_018122.4	0.9994
Leukoencephalopathy, Cystic, without Megalencephaly	RNASET2	RECESSIVE	NM_003730.4	0.9978
Leydig Cell Hypoplasia/Agenesis	LHCGR	RECESSIVE	NM_000233.3	0.9995
Liddle Syndrome	SCNN1B	DOMINANT	NM_000336.2	0.9990
	SCNN1G	DOMINANT	NM_001039.3	0.9987
Li-Fraumeni Syndrome	TP53	DOMINANT	NM_000546	0.9974
LIG4 Syndrome	LIG4	RECESSIVE	NM_002312.3	0.9879
Limb-Girdle Muscular Dystrophy, Dominant	DNAJB6	DOMINANT	NM_058246.3	0.9995
	LMNA	DOMINANT (Dom/Rec)	NM_005572.3	0.9982
Limb-Girdle Muscular Dystrophy, Recessive	POMGNT1	RECESSIVE	NM_017739	0.9993
	POMT1	RECESSIVE	NM_007171	0.9993
	POMT2	RECESSIVE	NM_013382.5	0.9980
	TCAP	RECESSIVE	NM_003673	0.9985
	TRIM32	RECESSIVE	NM_012210	0.9951
	TTN	RECESSIVE	NM_133378	0.9885
Lipoatrophy with Diabetes, Hepatic Steatosis, Hypertrophic Cardiomyopathy, and Leukomelanodermic Papules	LMNA	DOMINANT	NM_005572.3	0.9982
Lissencephaly, Dominant	TUBA1A	DOMINANT	NM_006009.3	0.9957
Lissencephaly, Recessive	NDE1	RECESSIVE	NM_001143979.1	0.9974
	RELN	RECESSIVE	NM_005045.3	0.9988
Lissencephaly/Subcortical Band Heterotopia	PAFAH1B1	DOMINANT	NM_000430.3	0.9908
Localized AR Hypotrichosis	DSG4	RECESSIVE	NM_177986.3	0.9997
Loeys-Dietz Syndrome	SMAD3	DOMINANT	NM_005902.3	0.9974
	TGFB2	DOMINANT	NM_001135599.2	0.9995
	TGFBR1	DOMINANT	NM_004612.2	0.9992
	TGFBR2	DOMINANT	NM_003242.5	0.9995

Long QT Syndrome	AKAP9	DOMINANT	NM_005751	0.9997
	ANK2	DOMINANT	NM_001148	0.9997
	CAV3	DOMINANT	NM_033337	0.9991
	KCNE1	DOMINANT	NM_000219	0.9993
	KCNE2	DOMINANT	NM_172201	0.9998
	KCNH2	DOMINANT	NM_000238.3	0.9936
	KCNQ1	DOMINANT	NM_000218	0.9984
	SCN4B	DOMINANT	NM_174934	0.9958
	SCN5A	DOMINANT	NM_198056	0.9983
	SNTA1	DOMINANT	NM_003098	0.9986
Long-Chain Acyl-CoA Dehydrogenase Deficiency	ACADL	RECESSIVE	NM_001608	0.9997
LRRK2-Related Parkinson Disease	LRRK2	DOMINANT	NM_198578	0.9998
Lung Cancer	EGFR	DOMINANT	NM_005228.3	0.9993
Lymphoproliferative Syndrome	ITK	RECESSIVE	NM_005546.3	0.9996
	SH2D1A	X_LINKED	NM_002351.4	0.9995
	XIAP	X_LINKED	NM_001167.3	0.9977
Lynch Syndrome	EPCAM	DOMINANT	NM_002354.2	0.9993
	MLH1	DOMINANT	NM_000249	0.9982
	MLH3	DOMINANT	NM_001040108	0.9996
	MSH2	DOMINANT	NM_000251	0.9942
	MSH6	DOMINANT	NM_000179	0.9986
	PMS1	DOMINANT	NM_000534	0.9997
	PMS2	DOMINANT	NM_000535.5	0.9573
Lysinuric Protein Intolerance	SLC7A7	RECESSIVE	NM_001126106.2	0.9994
Macular Corneal Dystrophy	CHST6	RECESSIVE	NM_021615.4	0.9944
Macular Degeneration	ARMS2	DOMINANT	NM_001099667.1	0.9938
	C2	DOMINANT	NM_000063.4	0.9989
	C3	DOMINANT	NM_000064.2	0.9966
	CFB	DOMINANT	NM_001710.5	0.9993
	CFH	DOMINANT	NM_000186.3	0.9997
	ERCC6	DOMINANT	NM_000124.2	0.9998

Macular Degeneration	FBLN5	DOMINANT	NM_006329.3	0.9994
	HMCN1	DOMINANT	NM_031935.2	0.9998
	HTRA1	DOMINANT	NM_002775.4	0.9994
	RAX2	DOMINANT	NM_032753.3	0.9970
Mainzer-Saldino Syndrome	IFT140	RECESSIVE	NM_014714.3	0.9989
Majeed Syndrome	LPIN2	RECESSIVE	NM_014646.2	0.9993
Mal de Meleda	SLURP1	RECESSIVE	NM_020427.2	0.9995
Male Infertility	CATSPER1	RECESSIVE	NM_053054.3	0.9958
Male-Limited Precocious Puberty	LHCGR	DOMINANT	NM_000233.3	0.9995
Malignant Hyperthermia Susceptibility	CACNA1S	DOMINANT	NM_000069.2	0.9979
	RYR1	DOMINANT	NM_000540.2	0.9963
Malignant Melanoma Susceptibility	MC1R	DOMINANT	NM_002386.3	0.9987
Malonyl-CoA Decarboxylase Deficiency	MLYCD	RECESSIVE	NM_012213.2	0.9990
Mandibuloacral Dysplasia	LMNA	RECESSIVE	NM_005572.3	0.9982
	ZMPSTE24	RECESSIVE	NM_005857.4	0.9998
Manitoba Oculotrichoanal Syndrome	FREM1	RECESSIVE	NM_144966.5	0.9989
Mannose-Binding Lectin Deficiency	MBL2	DOMINANT (Dom/Rec)	NM_000242.2	0.9998
Maple Syrup Urine Disease	BCKDHA	RECESSIVE	NM_000709	0.9978
	BCKDHB	RECESSIVE	NM_183050	0.9987
	DBT	RECESSIVE	NM_001918	0.9950
	DLD	RECESSIVE	NM_000108.3	0.9998
MAPT-Related Spectrum Disorders	MAPT	DOMINANT	NM_005910	0.9959
Marfan Syndrome	FBN1	DOMINANT	NM_000138.4	0.9997
	TGFBR2	DOMINANT	NM_003242.5	0.9995
Marinesco-Sjogren Syndrome	SIL1	RECESSIVE	NM_022464.4	0.9995
Marshall Syndrome	COL11A1	DOMINANT (Dom/Rec)	NM_001854.3	0.9996
Martsolf Syndrome	RAB3GAP2	RECESSIVE	NM_012414.3	0.9991
MASP2 Deficiency	MASP2	RECESSIVE	NM_006610.3	0.9989
MASS Syndrome	FBN1	DOMINANT	NM_000138.4	0.9997
Mast Cell Disease	KIT	DOMINANT	NM_000222.2	0.9997
Maturity Onset Diabetes of the Young	BLK	DOMINANT	NM_001715.2	0.9996

Maturity Onset Diabetes of the Young	GCK	DOMINANT	NM_000162	0.9990
	HNF1A	DOMINANT	NM_000545	0.9965
	HNF4A	DOMINANT	NM_000457.4	0.9914
	INS	DOMINANT	NM_000207.2	0.9982
	KCNJ11	DOMINANT	NM_000525	0.9936
	KLF11	DOMINANT	NM_003597.4	0.9981
	NEUROD1	DOMINANT	NM_002500.4	0.9951
	PAX4	DOMINANT	NM_006193.2	0.9982
MCAD Deficiency	ACADM	RECESSIVE	NM_000016	0.9989
McKusick-Kaufman Syndrome	MKKS	RECESSIVE	NM_018848.3	0.9998
Meacham Syndrome	WT1	DOMINANT	NM_024426.4	0.9959
Meckel Syndrome	B9D1	RECESSIVE	NM_015681.3	0.9995
	B9D2	RECESSIVE	NM_030578.3	0.9994
	CC2D2A	RECESSIVE	NM_001080522.2	0.9997
	CEP290	RECESSIVE	NM_025114.3	0.9998
	MKS1	RECESSIVE	NM_017777.3	0.9995
	NPHP3	RECESSIVE	NM_153240.4	0.9976
	RPGRIP1L	RECESSIVE	NM_015272.2	0.9995
	TCTN2	RECESSIVE	NM_024809.4	0.9945
	TMEM216	RECESSIVE	NM_001173990.2	0.9995
	TMEM67	RECESSIVE	NM_153704.5	0.9989
Medulloblastoma	SUFU	DOMINANT	NM_016169.3	0.9979
Megalencephalic Leukoencephalopathy with Subcortical Cysts	HEPACAM	RECESSIVE (Rec/Dom)	NM_152722.4	0.9919
	MLC1	RECESSIVE (Rec/Dom)	NM_015166.3	0.9938
Megaloblastic Anemia	CUBN	RECESSIVE	NM_001081.3	0.9996
Meier-Gorlin Syndrome	CDC6	RECESSIVE	NM_001254.3	0.9998
	ORC1	RECESSIVE	NM_004153.3	0.9996
	ORC6	RECESSIVE	NM_014321.3	0.9998
Metachondromatosis	PTPN11	DOMINANT	NM_002834.3	0.9996
Metachromatic Leukodystrophy	PSAP	RECESSIVE	NM_002778.2	0.9993
Metaphyseal Anadysplasia	MMP13	DOMINANT (Dom/Rec)	NM_002427.3	0.9992

Metaphyseal Anadysplasia	MMP9	RECESSIVE (Rec/Dom)	NM_004994.2	0.9973
Metaphyseal Chondrodysplasia	COL10A1	DOMINANT	NM_000493.3	0.9993
	PTH1R	DOMINANT	NM_000316.2	0.9959
Metatropic Dysplasia	TRPV4	DOMINANT	NM_021625	0.9968
Methylmalonate Semialdehyde Dehydrogenase Deficiency	ALDH6A1	RECESSIVE	NM_005589.2	0.9938
Methylmalonic Acidemia	MCEE	RECESSIVE	NM_032601	0.9998
	MMAA	RECESSIVE	NM_172250	0.9997
	MMAB	RECESSIVE	NM_052845	0.9940
	MMADHC	RECESSIVE	NM_015702	0.9998
	MUT	RECESSIVE	NM_000255	0.9998
Mevalonicaciduria	MVK	RECESSIVE	NM_000431.2	0.9993
MHC Class II Deficiency	RFXANK	RECESSIVE	NM_003721.2	0.9972
Microcephalic Osteodysplastic Primordial Dwarfism	PCNT	RECESSIVE	NM_006031.5	0.9991
Microtia, Hearing Impairment, and Cleft Palate	HOXA2	RECESSIVE	NM_006735.3	0.9995
Mitochondrial Complex V (ATP Synthase) Deficiency, Nuclear Type	ATPAF2	RECESSIVE	NM_145691.3	0.9979
	TMEM70	RECESSIVE	NM_017866.5	0.9995
Mitochondrial DNA Depletion Syndrome	C10orf2	RECESSIVE	NM_021830.4	0.9991
	DGUOK	RECESSIVE	NM_080916.2	0.9997
	MPV17	RECESSIVE	NM_002437.4	0.9992
	RRM2B	RECESSIVE (Rec/Dom)	NM_015713.4	0.9948
	SUCLA2	RECESSIVE	NM_003850.2	0.9998
	SUCLG1	RECESSIVE	NM_003849.3	0.9966
	TK2	RECESSIVE	NM_004614.4	0.9990
Mitochondrial Membrane Protein-Associated Neurodegeneration	C19orf12	RECESSIVE	NM_001031726.3	0.9966
Mitochondrial Neurogastrointestinal Encephalopathy Disease	TYMP	RECESSIVE	NM_001953.4	0.9990
Mitochondrial Phosphate Carrier Deficiency	SLC25A3	RECESSIVE	NM_005888.3	0.9996
Mitochondrial Respiratory Chain Complex I Deficiency	FOXRED1	RECESSIVE	NM_017547.3	0.9959
	NDUFA10	RECESSIVE	NM_004544.3	0.9979
	NDUFA11	RECESSIVE	NM_175614.4	0.9985
	NDUFA2	RECESSIVE	NM_002488.4	0.9999

Mitochondrial Respiratory Chain Complex I Deficiency	NDUFAF1	RECESSIVE	NM_016013.3	0.9995
	NDUFAF2	RECESSIVE	NM_174889.4	0.9999
	NDUFAF3	RECESSIVE	NM_199069.1	0.9984
	NDUFAF4	RECESSIVE	NM_014165.3	0.9996
	NDUFS1	RECESSIVE	NM_005006.6	0.9982
	NDUFS2	RECESSIVE	NM_004550.4	0.9994
	NDUFS3	RECESSIVE	NM_004551.2	0.9993
	NDUFS4	RECESSIVE	NM_002495.2	0.9997
	NDUFS6	RECESSIVE	NM_004553.4	0.9997
	NDUFS7	RECESSIVE	NM_024407.4	0.9849
	NDUFS8	RECESSIVE	NM_002496.3	0.9987
	NDUFV1	RECESSIVE	NM_007103.3	0.9973
	NDUFV2	RECESSIVE	NM_021074.4	0.9995
	NUBPL	RECESSIVE	NM_025152.2	0.9939
Mitochondrial Respiratory Chain Complex II Deficiency	SDHA	RECESSIVE	NM_004168.2	0.9991
	SDHAF1	RECESSIVE	NM_001042631.2	0.9992
Mitochondrial Respiratory Chain Complex III Deficiency	BCS1L	RECESSIVE	NM_004328.4	0.9989
	TTC19	RECESSIVE	NM_017775.3	0.9957
	UQCRQ	RECESSIVE	NM_014402.4	0.9993
Mitochondrial Respiratory Chain Complex IV Deficiency	COX10	RECESSIVE	NM_001303.3	0.9984
	COX15	RECESSIVE	NM_004376.5	0.9996
	COX6B1	RECESSIVE	NM_001863.4	0.9990
	FASTKD2	RECESSIVE	NM_014929.3	0.9970
	SCO1	RECESSIVE	NM_004589.2	0.9995
	SCO2	RECESSIVE	NM_005138.2	0.9991
	TACO1	RECESSIVE	NM_016360.3	0.9995
Molybdenum Cofactor Deficiency	MOCS1	RECESSIVE	NM_005943.5	0.9988
	MOCS2	RECESSIVE	NM_176806.2	0.9986
Monilethrix	KRT83	DOMINANT	NM_002282.3	0.9990
Monogenic Non-Syndromic Obesity	LEP	RECESSIVE	NM_000230.2	0.9987
	LEPR	RECESSIVE	NM_002303.5	0.9985

Monogenic Non-Syndromic Obesity	PCSK1	RECESSIVE	NM_000439.4	0.9988
	POMC	RECESSIVE (Rec/Dom)	NM_001035256.1	0.9972
Mononeuropathy of the Median Nerve	SH3TC2	RECESSIVE (Rec/Dom)	NM_024577	0.9985
Mowat-Wilson Syndrome	ZEB2	DOMINANT	NM_014795.3	0.9985
Moyamoya Disease	ACTA2	DOMINANT	NM_001613.2	0.9995
Muckle-Wells Syndrome	NLRP3	DOMINANT	NM_004895	0.9994
Mucolipidosis, Type I	NEU1	RECESSIVE	NM_000434.3	0.9990
Mucolipidosis, Type II	GNPTAB	RECESSIVE	NM_024312	0.9997
Mucolipidosis, Type III Alpha/Beta	GNPTAB	RECESSIVE	NM_024312	0.9997
Mucolipidosis, Type III Gamma	GNPTG	RECESSIVE	NM_032520	0.9982
Mucolipidosis, Type IV	MCOLN1	RECESSIVE	NM_020533.2	0.9992
Mucopolysaccharidosis, Type I	IDUA	RECESSIVE	NM_000203	0.9970
Mucopolysaccharidosis, Type III	GNS	RECESSIVE	NM_002076	0.9996
	HGSNAT	RECESSIVE	NM_152419	0.9409
	NAGLU	RECESSIVE	NM_000263	0.9991
	SGSH	RECESSIVE	NM_000199	0.9909
Mucopolysaccharidosis, Type IV	GALNS	RECESSIVE	NM_000512	0.9965
Mucopolysaccharidosis, Type IX	HYAL1	RECESSIVE	NR_047690.1	0.9873
	GLB1	RECESSIVE	NM_000404	0.9993
Mucopolysaccharidosis, Type VI	ARSB	RECESSIVE	NM_000046	0.9956
Mucopolysaccharidosis, Type VII	GUSB	RECESSIVE	NM_000181	0.9978
Mulibrey Nanism	TRIM37	RECESSIVE	NM_015294.3	0.9997
Multicentric Carpotarsal Osteolysis Syndrome	MAFB	DOMINANT	NM_005461.3	0.9993
Multicentric Osteolysis-Nodulosis-Arthropathy (MONA) Spectrum Disorders	MMP2	RECESSIVE	NM_004530.4	0.9993
Multiminicore Disease	RYR1	RECESSIVE	NM_000540.2	0.9963
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFA	RECESSIVE	NM_000126.3	0.9996
	ETFDH	RECESSIVE	NM_004453.2	0.9995
Multiple Cutaneous and Mucosal Venous Malformations	TEK	DOMINANT	NM_000459.3	0.9995
Multiple Cutaneous and Uterine Leiomyomas	FH	DOMINANT	NM_000143.3	0.9993
Multiple Endocrine Neoplasia	CDKN1B	DOMINANT	NM_004064.3	0.9974

Multiple Endocrine Neoplasia	MEN1	DOMINANT	NM_130799.2	0.9953
	RET	DOMINANT	NM_020975	0.9990
Multiple Epiphyseal Dysplasia with Early-Onset Diabetes Mellitus	EIF2AK3	RECESSIVE	NM_004836.5	0.9997
Multiple Epiphyseal Dysplasia, Dominant	COL9A1	DOMINANT	NM_001851.4	0.9995
	COL9A2	DOMINANT	NM_001852.3	0.9951
	COL9A3	DOMINANT	NM_001853.3	0.9962
	COMP	DOMINANT	NM_000095.2	0.9957
	MATN3	DOMINANT	NM_002381.4	0.9994
Multiple Epiphyseal Dysplasia, Recessive	SLC26A2	RECESSIVE	NM_000112.3	0.9998
Multiple Familial Trichoepithelioma	CYLD	DOMINANT	NM_015247.2	0.9995
Multiple Mitochondrial Dysfunctions Syndrome	BOLA3	RECESSIVE	NM_212552.2	0.9992
	NFU1	RECESSIVE	NM_001002755.2	0.9996
Multiple Pterygium Syndrome	CHRNA1	RECESSIVE	NM_000079.3	0.9996
	CHRND	RECESSIVE	NM_000751.2	0.9986
	CHRNG	RECESSIVE	NM_005199.4	0.9967
Multiple Sulfatase Deficiency	SUMF1	RECESSIVE	NM_182760	0.9935
Multiple Synostoses Syndrome	FGF9	DOMINANT	NM_002010.2	0.9996
	GDF5	DOMINANT	NM_000557.2	0.9989
Multisystemic Smooth Muscle Dysfunction Syndrome	ACTA2	DOMINANT	NM_001613.2	0.9995
MYH9-related disorder	MYH9	DOMINANT	NM_002473	0.9987
MYH-Associated Polyposis	MUTYH	RECESSIVE	NM_001048171.1	0.9993
Myhre Syndrome	SMAD4	DOMINANT	NM_005359	0.9984
Myoadenylate Deaminase Deficiency	AMPD1	RECESSIVE	NM_000036.2	0.9995
Myoclonus-Dystonia	SGCE	DOMINANT	NM_003919.2	0.9988
Myofibrillar Myopathy, Dominant	BAG3	DOMINANT	NM_004281.3	0.9992
	CRYAB	DOMINANT (Dom/Rec)	NM_001885.1	0.9995
	DES	DOMINANT (Dom/Rec)	NM_001927	0.9986
	DNAJB6	DOMINANT	NM_058246.3	0.9995
	LDB3	DOMINANT	NM_001080116	0.9928
	MYOT	DOMINANT	NM_006790	0.9996

Myokymia	KCNA1	DOMINANT	NM_000217.2	0.9963
Myopathy, Early-Onset, Areflexia, Respiratory Distress, and Dysphagia	MEGF10	RECESSIVE	NM_032446.2	0.9994
Myopathy, Lactic Acidosis, and Sideroblastic Anemia	PUS1	RECESSIVE	NM_025215.5	0.9968
	YARS2	RECESSIVE	NM_001040436.2	0.9985
Myosin Storage Myopathy	MYH7	RECESSIVE (Rec/Dom)	NM_000257	0.9987
Myostatin-Related Muscle Hypertrophy	MSTN	DOMINANT (Dom/Rec)	NM_005259.2	0.9998
Myotilinopathy	MYOT	DOMINANT	NM_006790	0.9996
Myotonia Congenita	CLCN1	DOMINANT (Dom/Rec)	NM_000083.2	0.9983
N-Acetylglutamate Synthase Deficiency	NAGS	RECESSIVE	NM_153006.2	0.9989
Nager Syndrome	SF3B4	DOMINANT (Dom/Rec)	NM_005850.4	0.9988
Nail-Patella Syndrome	LMX1B	DOMINANT	NM_002316.3	0.9973
Natural Killer Cell and Glucocorticoid Deficiency with DNA Repair Defect	MCM4	RECESSIVE	NM_005914.3	0.9961
Naxos Disease	JUP	RECESSIVE	NM_002230	0.9952
Nemaline Myopathy	ACTA1	RECESSIVE	NM_001100.3	0.9974
	TPM3	RECESSIVE	NM_152263.2	0.9980
Nemaline Myopathy, Dominant	KBTBD13	DOMINANT	NM_001101362.2	0.9992
	TPM2	DOMINANT	NM_003289.3	0.9933
Nemaline Myopathy, Recessive	CFL2	RECESSIVE	NM_021914.7	0.9998
	NEB	RECESSIVE	NM_004543.4	0.8742
	TNNT1	RECESSIVE	NM_003283.4	0.9888
Neonatal Severe Primary Hyperparathyroidism	CASR	RECESSIVE	NM_000388.3	0.9994
Nephrogenic Diabetes Insipidus	AQP2	RECESSIVE (Rec/Dom)	NM_000486.5	0.9990
	AVPR2	X_LINKED	NM_000054.4	0.9993
Nephronophthisis	GLIS2	RECESSIVE	NM_032575.2	0.9955
	INVS	RECESSIVE	NM_014425.3	0.9993
	NEK8	RECESSIVE	NM_178170.2	0.9934
	NPHP1	RECESSIVE	NM_000272.3	0.9978
	NPHP3	RECESSIVE	NM_153240.4	0.9976
	NPHP4	RECESSIVE	NM_015102.3	0.9970

Nephronophthisis	RPGRIP1L	RECESSIVE	NM_015272.2	0.9995
	TMEM67	RECESSIVE	NM_153704.5	0.9989
Nephronophthisis-Like Nephropathy	XPNPEP3	RECESSIVE	NM_022098.3	0.9959
Nephropathic Cystinosis	CTNS	RECESSIVE	NM_004937	0.9955
Nephrotic Syndrome	LAMB2	RECESSIVE	NM_002292.3	0.9993
	PLCE1	RECESSIVE	NM_016341.3	0.9990
Nestor-Guillermo Progeria Syndrome	BANF1	RECESSIVE	NM_001143985.1	0.9976
Netherton Syndrome	SPINK5	RECESSIVE	NM_006846.3	0.9994
Neural Tube Defect	VANGL1	DOMINANT	NM_138959.2	0.9996
Neural Tube Defects, Folate-Sensitive	MTHFR	RECESSIVE	NM_005957.4	0.9988
Neuroblastoma	KIF1B	DOMINANT	NM_015074	0.9987
	PHOX2B	DOMINANT	NM_003924	0.9983
Neuroblastoma Susceptibility	ALK	DOMINANT	NM_004304.4	0.9948
Neurodegeneration due to Cerebral Folate Transport Deficiency	FOLR1	RECESSIVE	NM_016725.2	0.9997
Neuroferritinopathy	FTL	DOMINANT	NM_000146.3	0.9863
Neurofibromatosis, Type 1	NF1	DOMINANT	NM_000267	0.9956
Neurofibromatosis, Type 2	NF2	DOMINANT	NM_000268	0.9965
Neurofibromatosis-Noonan Syndrome	NF1	DOMINANT	NM_000267	0.9956
Neurogenic Scapuloperoneal Syndrome	DES	DOMINANT	NM_001927	0.9986
Neurologic Disorders/Seipinopathy	BSCL2	DOMINANT	NM_032667.6	0.9989
Neuronal Ceroid-Lipofuscinosis, Dominant/Recessive	CLN3	RECESSIVE	NM_001042432	0.9987
	CLN5	RECESSIVE	NM_006493.2	0.9992
	CTSD	RECESSIVE	NM_001909.4	0.9987
Neuronal Ceroid-Lipofuscinosis, Recessive	CLN6	RECESSIVE	NM_017882	0.9965
	DNAJC5	DOMINANT	NM_025219.2	0.9987
	MFSD8	RECESSIVE	NM_152778	0.9996
	PPT1	RECESSIVE	NM_000310	0.9994
	TPP1	RECESSIVE	NM_000391	0.9991
Neutral Lipid Storage Disease with Myopathy	PNPLA2	RECESSIVE	NM_020376.3	0.9919
Nevoid Basal Cell Carcinoma Syndrome	PTCH1	DOMINANT	NM_000264.3	0.9972

Newfoundland Rod-Cone Dystrophy	RLBP1	RECESSIVE	NM_000326.4	0.9995
Nicolaides-Baraitser Syndrome	SMARCA2	DOMINANT	NM_003070.3	0.9988
Niemann-Pick Disease Type C	NPC1	RECESSIVE	NM_000271	0.9972
	NPC2	RECESSIVE	NM_006432	0.9996
Nijmegen Breakage Syndrome	NBN	RECESSIVE	NM_002485.4	0.9998
Nocturnal Frontal Lobe Epilepsy	CHRNA2	DOMINANT	NM_000742.3	0.9970
Nonautoimmune Hyperthyroidism	TSHR	DOMINANT	NM_000369	0.9994
Non-Classic Cystic Fibrosis-Like Syndrome	SCNN1B	DOMINANT	NM_000336.2	0.9990
Nonepidermolytic Palmoplantar Hyperkeratosis	KRT1	DOMINANT	NM_006121.3	0.9954
Nonspecific Cardiac Conduction Defect	SCN1B	DOMINANT	NM_001037	0.9951
Nonsyndromic Hearing Loss, Dominant	COCH	DOMINANT	NM_004086	0.9942
	COL11A2	DOMINANT	NM_080680	0.9952
	EYA4	DOMINANT	NM_172105	0.9992
	GJB2	DOMINANT	NM_004004	0.9995
	GJB3	DOMINANT	NM_024009	0.9981
	GJB6	DOMINANT	NM_006783	0.9994
	MYH14	DOMINANT	NM_024729	0.9916
	MYH9	DOMINANT	NM_002473	0.9987
	MYO1A	DOMINANT	NM_005379	0.9993
	MYO6	DOMINANT	NM_004999	0.9997
	MYO7A	DOMINANT	NM_000260	0.9980
	POU4F3	DOMINANT	NM_002700	0.9991
	SIX1	DOMINANT	NM_005982	0.9984
	SLC17A8	DOMINANT	NM_139319	0.9970
	TECTA	DOMINANT	NM_005422	0.9995
	TJP2	DOMINANT	NM_004817	0.9988
	TMC1	DOMINANT	NM_138691	0.9987
	WFS1	DOMINANT	NM_006005	0.9984
Nonsyndromic Hearing Loss, Mixed	DFNA5	DOMINANT	NM_004403.2	0.9997
	DIAPH1	DOMINANT	NM_005219.4	0.9881
	FOXI1	RECESSIVE	NM_012188.4	0.9994

Nonsyndromic Hearing Loss, Mixed	KCNJ10	RECESSIVE	NM_002241.4	0.9976
Nonsyndromic Hearing Loss, Recessive	CDH23	RECESSIVE	NM_022124	0.9973
	CLDN14	RECESSIVE	NM_144492	0.9963
	DFNB31	RECESSIVE	NM_015404	0.9992
	DFNB59	RECESSIVE	NM_001042702	0.9985
	ESRRB	RECESSIVE	NM_004452	0.9942
	GJB2	RECESSIVE	NM_004004	0.9995
	GJB6	RECESSIVE	NM_006783	0.9994
	GPSM2	RECESSIVE	NM_013296	0.9996
	GRXCR1	RECESSIVE	NM_001080476	0.9997
	HGF	RECESSIVE	NM_000601.4	0.9964
	LHFPL5	RECESSIVE	NM_182548	0.9899
	LOXHD1	RECESSIVE	NM_144612	0.9991
	LRTOMT	RECESSIVE	NM_001145308	0.9963
	MARVELD2	RECESSIVE	NM_001038603	0.9985
	MYO15A	RECESSIVE	NM_016239	0.9964
	MYO3A	RECESSIVE	NM_017433	0.9980
	MYO6	RECESSIVE	NM_004999	0.9997
	MYO7A	RECESSIVE	NM_000260	0.9980
	OTOF	RECESSIVE	NM_194248	0.9969
	PCDH15	RECESSIVE	NM_033056	0.9997
	RDX	RECESSIVE	NM_002906	0.9996
	SLC26A4	RECESSIVE	NM_000441	0.9957
	SLC26A5	RECESSIVE	NM_198999	0.9996
	TECTA	RECESSIVE	NM_005422	0.9995
	TMC1	RECESSIVE	NM_138691	0.9987
	TMIE	RECESSIVE	NM_147196	0.9984
	TMPRSS3	RECESSIVE	NM_024022	0.9995
	USH1C	RECESSIVE	NM_005709	0.9889
Nonsyndromic Hearing Loss, X-Linked	POU3F4	X_LINKED	NM_000307	0.9991
	PRPS1	X_LINKED	NM_002764	0.9944

Nonsyndromic Hearing Loss, X-Linked	SMPX	X_LINKED	NM_014332.2	0.9999
Nonsyndromic Trigonocephaly	FGFR1	DOMINANT	NM_023110.2	0.9991
Noonan Syndrome	BRAF	DOMINANT	NM_004333.4	0.9934
	KRAS	DOMINANT	NM_004985.3	0.9996
	MAP2K1	DOMINANT	NM_002755.3	0.9985
	NRAS	DOMINANT	NM_002524.4	0.9996
	PTPN11	DOMINANT	NM_002834.3	0.9996
	RAF1	DOMINANT	NM_002880.3	0.9991
	SOS1	DOMINANT	NM_005633.3	0.9998
Noonan-Like Syndrome Disorder	CBL	DOMINANT	NM_005188.3	0.9990
Noonan-Like Syndrome with Loose Anagen Hair	SHOC2	DOMINANT	NM_007373.3	0.9878
Norepinephrine Transporter Deficiency	SLC6A2	DOMINANT	NM_001043.3	0.9978
North American Indian Childhood Cirrhosis	UTP4	RECESSIVE	NM_032830.2	0.9995
Obesity	MC4R	DOMINANT	NM_005912.2	0.9998
	PPARG	DOMINANT	NM_015869.4	0.9950
Occult Macular Dystrophy	RP1L1	DOMINANT	NM_178857.5	0.9810
Oculocutaneous Albinism	OCA2	RECESSIVE	NM_000275.2	0.9992
	SLC45A2	RECESSIVE	NM_016180.3	0.9997
	TYR	RECESSIVE	NM_000372.4	0.9996
	TYRP1	RECESSIVE	NM_000550.2	0.9997
Oculodentodigital Dysplasia	GJA1	DOMINANT (Dom/Rec)	NM_000165.3	0.9996
Odontoonychodermal Dysplasia	WNT10A	RECESSIVE	NM_025216.2	0.9985
Oguchi Disease	SAG	RECESSIVE	NM_000541	0.9995
Oligodontia-Colorectal Cancer Syndrome	AXIN2	DOMINANT	NM_004655.3	0.9983
Omenn Syndrome	DCLRE1C	RECESSIVE	NM_001033855.1	0.9930
	RAG1	RECESSIVE	NM_000448	0.9998
	RAG2	RECESSIVE	NM_000536	0.9997
Omodysplasia	GPC6	RECESSIVE	NM_005708.3	0.9996
Optic Atrophy, Dominant	OPA1	DOMINANT	NM_015560.2	0.9998
	OPA3	DOMINANT	NM_025136	0.9973
Optic Atrophy, Recessive	TMEM126A	RECESSIVE	NM_032273.3	0.9999

Ornithine Aminotransferase Deficiency	OAT	RECESSIVE	NM_000274.3	0.9887
Ornithine Transcarbamylase Deficiency	OTC	X_LINKED	NM_000531	0.9999
Orofacial Cleft	BMP4	DOMINANT	NM_001202.3	0.9959
Orotic Aciduria	UMPS	RECESSIVE	NM_000373.3	0.9995
Osteogenesis Imperfecta, Dominant	COL1A1	DOMINANT	NM_000088.3	0.9902
	COL1A2	DOMINANT	NM_000089	0.9995
Osteogenesis Imperfecta, Recessive	BMP1	RECESSIVE	NM_006129.4	0.9936
	CRTAP	RECESSIVE	NM_006371.4	0.9996
	FKBP10	RECESSIVE	NM_021939.3	0.9881
	P3H1	RECESSIVE	NM_022356.3	0.9993
	PPIB	RECESSIVE	NM_000942.4	0.9984
	SERPINF1	RECESSIVE	NM_002615.5	0.9985
	SERPINH1	RECESSIVE	NM_001235.3	0.9973
	SP7	RECESSIVE	NM_001173467.1	0.9958
Osteoglophonic Dysplasia	FGFR1	DOMINANT	NM_023110.2	0.9991
Osteopetrosis	CLCN7	RECESSIVE	NM_001287.5	0.9985
	OSTM1	RECESSIVE	NM_014028.3	0.9936
	TCIRG1	RECESSIVE	NM_006019.3	0.9958
	TNFRSF11A	RECESSIVE	NM_003839.3	0.9938
	TNFSF11	RECESSIVE	NM_003701.3	0.9998
Osteopetrosis with Renal Tubular Acidosis	CA2	RECESSIVE	NM_000067.2	0.9963
Otofaciocervical Syndrome	EYA1	DOMINANT	NM_000503.4	0.9976
Otospondylomegaepiphyseal Dysplasia	COL11A2	RECESSIVE	NM_080680	0.9952
OTX2-Related Syndromic Microphthalmia	OTX2	DOMINANT	NM_172337.2	0.9995
Ovarian Dysgenesis	BMP15	X_LINKED	NM_005448.2	0.9994
	FSHR	RECESSIVE	NM_000145.3	0.9996
Ovarian Hyperstimulation Syndrome	FSHR	DOMINANT	NM_000145.3	0.9996
Paget Disease of Bone	SQSTM1	DOMINANT	NM_003900.4	0.9970
	TNFRSF11A	DOMINANT	NM_003839.3	0.9938
PALB2-Related Cancer Susceptibility	PALB2	DOMINANT	NM_024675	0.9996
Pallister-Hall Syndrome	GLI3	DOMINANT	NM_000168.5	0.9984

Palmoplantar Keratoderma, Mutilating, with Periorificial Keratotic Plaques	TRPV3	DOMINANT	NM_145068.3	0.9983
Pancreatic Cancer	PALLD	DOMINANT	NM_001166108.1	0.9967
Pantothenate Kinase-Associated Neurodegeneration	PANK2	RECESSIVE	NM_153638.2	0.9988
Papillary Renal Carcinoma	MET	DOMINANT	NM_001127500.1	0.9996
Papillon-Lefevre Syndrome	CTSC	RECESSIVE	NM_001814.4	0.9995
Paraganglioma and Gastric Stromal Sarcoma	SDHB	DOMINANT	NM_003000.2	0.9995
Paramyotonia Congenita of Von Eulenburg	SCN4A	DOMINANT	NM_000334.4	0.9976
Parathyroid Carcinoma	CDC73	DOMINANT	NM_024529.4	0.9999
Parkes Weber Syndrome	RASA1	DOMINANT	NM_002890.2	0.9995
Parkinson Disease, Dominant	HTRA2	DOMINANT	NM_013247.4	0.9991
	SNCA	DOMINANT	NM_000345	0.9964
	UCHL1	DOMINANT	NM_004181.4	0.9994
	VPS35	DOMINANT	NM_018206.4	0.9959
Parkinson Disease, Dominant/Recessive	NR4A2	DOMINANT (Dom/Rec)	NM_006186.3	0.9995
	SNCAIP	DOMINANT (Dom/Rec)	NM_005460.2	0.9993
Parkinson Disease, Juvenile	PARK2	RECESSIVE (Rec/Dom)	NM_004562	0.9952
Parkinson Disease, Recessive	FBXO7	RECESSIVE (Rec/Dom)	NM_012179.3	0.9995
	PARK7	RECESSIVE (Rec/Dom)	NM_007262.4	0.9997
	PINK1	RECESSIVE (Rec/Dom)	NM_032409	0.9990
Paroxysmal Extreme Pain Disorder	SCN9A	DOMINANT	NM_002977.3	0.9997
Paroxysmal Familial Ventricular Fibrillation	SCN5A	DOMINANT	NM_198056	0.9983
Partial Epilepsy with Auditory Features	LGI1	DOMINANT	NM_005097.2	0.9997
Partial Isolated Growth Hormone Deficiency (Dominant/Recessive)	GHSR	RECESSIVE	NM_198407.2	0.9998
Patterned Dystrophy of Retinal Pigment Epithelium	PRPH2	DOMINANT (Dom/Rec)	NM_000322	0.9989
PCWH Syndrome	SOX10	DOMINANT (Dom/Rec)	NM_006941.3	0.9944
Peeling Skin Syndrome	TGM5	RECESSIVE	NM_201631.3	0.9994
Pendred Syndrome	FOXI1	RECESSIVE	NM_012188.4	0.9994
	KCNJ10	RECESSIVE	NM_002241.4	0.9976
	SLC26A4	RECESSIVE	NM_000441	0.9957

Periventricular Heterotopia	ARFGEF2	RECESSIVE	NM_006420.2	0.9994
Perlman Syndrome	DIS3L2	RECESSIVE	NM_152383.4	0.9981
Permanent Neonatal Diabetes Mellitus	ABCC8	RECESSIVE	NM_000352	0.9992
	GCK	RECESSIVE (Rec/Dom)	NM_000162	0.9990
Permanent neonatal diabetes mellitus (PNDM)	PTF1A	RECESSIVE	NM_178161.2	0.9994
Peroxisomal Bifunctional Enzyme Deficiency	HSD17B4	RECESSIVE	NM_000414.3	0.9997
Perrault Syndrome	HSD17B4	RECESSIVE	NM_000414.3	0.9997
Perry Syndrome	DCTN1	DOMINANT	NM_004082.4	0.9990
Peters Anomaly	CYP1B1	RECESSIVE	NM_000104.3	0.9997
	PAX6	DOMINANT	NM_000280.4	0.9984
	PITX2	DOMINANT	NM_153427.2	0.9962
Peters Plus Syndrome	B3GLCT	RECESSIVE	NM_194318.3	0.9958
Peutz-Jeghers Syndrome	STK11	DOMINANT	NM_000455	0.9965
Pfeiffer Syndrome	FGFR1	DOMINANT	NM_023110.2	0.9991
	FGFR2	DOMINANT	NM_000141.4	0.9980
Phenylalanine Hydroxylase Deficiency	PAH	RECESSIVE	NM_000277	0.9996
Pheochromocytoma	KIF1B	DOMINANT	NM_015074	0.9987
	RET	DOMINANT	NM_020975	0.9990
	TMEM127	DOMINANT	NM_017849.3	0.9995
Phosphoenolpyruvate Carboxykinase Deficiency	PCK1	RECESSIVE	NM_002591.3	0.9988
Phosphoglycerate Dehydrogenase Deficiency	PHGDH	RECESSIVE	NM_006623.3	0.9987
Phosphoribosylpyrophosphate Synthetase Superactivity	PRPS1	X-LINKED	NM_002764	0.9944
Phosphorylase Kinase Deficiency	PHKA1	X-LINKED	NM_002637.3	0.9998
	PHKB	RECESSIVE	NM_000293.2	0.9971
	PHKG2	RECESSIVE	NM_000294.2	0.9981
Phosphoserine Aminotransferase Deficiency	PSAT1	RECESSIVE	NM_058179.2	0.9997
Phosphoserine Phosphatase Deficiency	PSPH	RECESSIVE	NM_004577.3	0.9974
Piebald Trait	KIT	DOMINANT	NM_000222.2	0.9997
	SNAI2	DOMINANT	NM_003068.4	0.9999
Pierson Syndrome	LAMB2	RECESSIVE	NM_002292.3	0.9993
Pigmented Paravenous Chorioretinal Atrophy	CRB1	DOMINANT	NM_201253	0.9998

Pineal Hyperplasia, Insulin-Resistant Diabetes Mellitus, and Somatic Abnormalities	INSR	DOMINANT	NM_000208.2	0.9917
Pitt-Hopkins Syndrome	TCF4	DOMINANT	NM_001083962.1	0.9980
Pitt-Hopkins-Like Syndrome	CNTNAP2	RECESSIVE	NM_014141.5	0.9986
	NRXN1	RECESSIVE	NM_001135659.1	0.9989
Pituitary Dwarfism II	GHR	RECESSIVE	NM_000163.4	0.9991
PITX2-Related Eye Abnormalities	PITX2	DOMINANT	NM_153427.2	0.9962
PLA2G6-Associated Neurodegeneration	PLA2G6	RECESSIVE	NM_003560.2	0.9985
Plasminogen Activator Inhibitor-1 Deficiency	SERPINE1	RECESSIVE (Rec/Dom)	NM_000602	0.9992
Platelet Glycoprotein IV Deficiency	CD36	RECESSIVE	NM_001001547.2	0.9989
Pleuropulmonary Blastoma	DICER1	DOMINANT	NM_177438.2	0.9997
Pol III-Related Leukodystrophy	POLR3A	RECESSIVE	NM_007055.3	0.9992
	POLR3B	RECESSIVE	NM_018082.5	0.9994
POLG-Related Spectrum Disorders	POLG	RECESSIVE (Rec/Dom)	NM_002693.2	0.9987
Polycystic Kidney Disease, Autosomal Dominant	PKD2	DOMINANT	NM_000297	0.9994
Polycystic Kidney Disease, Autosomal Recessive	PKHD1	RECESSIVE	NM_138694	0.9996
Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy	TREM2	RECESSIVE	NM_018965.3	0.9993
	TYROBP	RECESSIVE	NM_003332.3	0.9986
Polycystic Liver Disease	PRKCSH	DOMINANT	NM_002743.2	0.9987
	SEC63	DOMINANT	NM_007214.4	0.9937
Polydactyly	GLI3	DOMINANT	NM_000168.5	0.9984
Polymicrogyria	ADGRG1	RECESSIVE	NM_005682.5	0.9979
Polyneuropathy, Hearing Loss, Ataxia, Retinitis Pigmentosa and Cataract (PHARC)	ABHD12	RECESSIVE	NM_001042472.2	0.9970
Pontocerebellar Hypoplasia	EXOSC3	RECESSIVE	NM_016042.3	0.9997
	RARS2	RECESSIVE	NM_020320.3	0.9979
	SEPSECS	RECESSIVE	NM_016955.3	0.9996
	TSEN2	RECESSIVE	NM_025265.3	0.9996
	TSEN34	RECESSIVE	NM_024075.3	0.9991
	TSEN54	RECESSIVE	NM_207346.2	0.9976
	VRK1	RECESSIVE	NM_003384.2	0.9998

Popliteal Pterygium Syndrome	IRF6	DOMINANT	NM_006147.3	0.9995
	RIPK4	RECESSIVE	NM_020639.2	0.9991
Porencephaly	COL4A1	DOMINANT	NM_001845.4	0.9993
	COL4A2	DOMINANT	NM_001846.2	0.9981
Porphyria Cutanea Tarda	UROD	DOMINANT	NM_000374.4	0.9987
Postaxial Acrofacial Dysostosis	DHODH	RECESSIVE	NM_001361.4	0.9992
Posterior Column Ataxia with Retinitis Pigmentosa	FLVCR1	RECESSIVE	NM_014053.3	0.9910
Posterior Polar Cataract	CRYAB	DOMINANT	NM_001885.1	0.9995
Posterior Polymorphous Corneal Dystrophy	VSX1	DOMINANT	NM_014588.5	0.9978
Potassium-Aggravated Myotonia	SCN4A	DOMINANT	NM_000334.4	0.9976
Prader-Willi-Like Syndrome	SIM1	RECESSIVE	NM_005068.2	0.9997
Premature Ovarian Failure	FIGLA	DOMINANT	NM_001004311.3	0.9982
	NOBOX	DOMINANT	NM_001080413.3	0.9965
Premature Ovarian Failure 2B	POF1B	X-LINKED	NM_024921.3	0.9999
Primary Ciliary Dyskinesia	CCDC103	RECESSIVE	NM_213607.2	0.9994
	CCDC39	RECESSIVE	NM_181426.1	0.9954
	CCDC40	RECESSIVE	NM_017950.3	0.9863
	DNAAF1	RECESSIVE	NM_178452.4	0.9978
	DNAAF2	RECESSIVE	NM_018139.2	0.9992
	DNAAF3	RECESSIVE	NM_001256714.1	0.9974
	DNAH11	RECESSIVE	NM_001277115.1	0.9992
	DNAH5	RECESSIVE	NM_001369.2	0.9987
	DNAI1	RECESSIVE	NM_012144.2	0.9992
	DNAI2	RECESSIVE	NM_023036.4	0.9977
	DNAL1	RECESSIVE	NM_031427.3	0.9986
	LRRC6	RECESSIVE	NM_012472.3	0.9997
	RSPH4A	RECESSIVE	NM_001010892.2	0.9997
	RSPH9	RECESSIVE	NM_152732.4	0.9993
Primary Coenzyme Q10 Deficiency	COQ2	RECESSIVE	NM_015697.7	0.9998
	COQ9	RECESSIVE	NM_020312.3	0.9995
	PDSS1	RECESSIVE	NM_014317.3	0.9987

Primary Coenzyme Q10 Deficiency	PDSS2	RECESSIVE	NM_020381.3	0.9994
Primary Congenital Glaucoma	CYP1B1	RECESSIVE	NM_000104.3	0.9997
	LTBP2	RECESSIVE	NM_000428.2	0.9992
Primary Hyperoxaluria	AGXT	RECESSIVE	NM_000030.2	0.9987
	GRHPR	RECESSIVE	NM_012203.1	0.9994
	HOGA1	RECESSIVE	NM_138413.3	0.9978
Primary Hypertrophic Osteoarthropathy	HPGD	RECESSIVE	NM_000860.5	0.9996
Primary Hypomagnesemia	CLDN16	RECESSIVE	NM_006580.3	0.9997
Primary Lymphedema with Myelodysplasia	GATA2	DOMINANT	NM_032638.4	0.9991
Primary Microcephaly 2 With or Without Cortical Malformations	WDR62	RECESSIVE	NM_001083961.1	0.9988
Primary Microcephaly, Recessive	ASPM	RECESSIVE	NM_018136.4	0.9999
	CASC5	RECESSIVE	NM_144508.3	0.9997
	CDK5RAP2	RECESSIVE	NM_018249.5	0.9996
	CENPJ	RECESSIVE	NM_018451.4	0.9995
	CEP152	RECESSIVE	NM_014985.3	0.9973
	MCPH1	RECESSIVE	NM_024596.3	0.9968
	STIL	RECESSIVE	NM_003035.2	0.9997
Primary Open Angle Glaucoma	MYOC	RECESSIVE	NM_000261.1	0.9997
	OPTN	DOMINANT	NM_021980.4	0.9944
	WDR36	DOMINANT	NM_139281.2	0.9992
Progeroid Laminopathies	LMNA	DOMINANT	NM_005572.3	0.9982
Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	C10orf2	DOMINANT	NM_021830.4	0.9991
Progressive Familial Heart Block	SCN5A	DOMINANT	NM_198056	0.9983
	TRPM4	DOMINANT	NM_017636.3	0.9988
Progressive Familial Intrahepatic Cholestasis	ABCB4	RECESSIVE	NM_000443.3	0.9994
Progressive Myoclonic Epilepsy	GOSR2	RECESSIVE	NM_004287.3	0.9996
	KCTD7	RECESSIVE	NM_153033.4	0.9991
	PRICKLE2	RECESSIVE	NM_198859.3	0.9982
Progressive Myoclonus Epilepsy with Ataxia	PRICKLE1	RECESSIVE	NM_153026.2	0.9970

Progressive Myoclonus Epilepsy, Lafora type	NHLRC1	RECESSIVE	NM_198586	0.9993
Progressive Pseudorheumatoid Arthropathy of Childhood	WISP3	RECESSIVE	NM_003880.3	0.9995
Prolidase Deficiency	PEPD	RECESSIVE	NM_000285.3	0.9991
Proliferative Vasculopathy And Hydranencephaly-Hydrocephaly Syndrome	FLVCR2	RECESSIVE	NM_017791.2	0.9992
Proopiomelanocortin Deficiency	POMC	RECESSIVE	NM_001035256.1	0.9972
Propionic Acidemia	PCCA	RECESSIVE	NM_000282	0.9991
	PCCB	RECESSIVE	NM_000532	0.9969
Proprotein Convertase-1 Deficiency	PCSK1	RECESSIVE	NM_000439.4	0.9988
Protein C Deficiency	PROC	DOMINANT	NM_000312	0.9991
Protein S Deficiency	PROS1	DOMINANT (Dom/Rec)	NM_000313	0.9998
Prothrombin Deficiency	F2	RECESSIVE	NM_000506	0.9984
Prothrombin-Related Thrombophilia	F2	DOMINANT	NM_000506	0.9984
Proximal Renal Tubular Acidosis with Ocular Abnormalities	SLC4A4	RECESSIVE	NM_003759.3	0.9984
Pseudoachondroplasia	COMP	DOMINANT	NM_000095.2	0.9957
Pseudohypoaldosteronism, Type I, Dominant	CUL3	DOMINANT	NM_003590.4	0.9987
	KLHL3	DOMINANT	NM_017415.2	0.9957
	NR3C2	DOMINANT	NM_000901.4	0.9972
	STX16	DOMINANT	NM_001001433.2	0.9981
Pseudohypoaldosteronism, Type I, Recessive	SCNN1A	RECESSIVE	NM_001038.5	0.9970
	SCNN1B	RECESSIVE	NM_000336.2	0.9990
	SCNN1G	RECESSIVE	NM_001039.3	0.9987
Pseudohypoaldosteronism, Type II	WNK1	DOMINANT	NM_018979.3	0.9983
	WNK4	DOMINANT	NM_032387.4	0.9850
Pseudoinflammatory Fundus Dystrophy	TIMP3	DOMINANT	NM_000362.4	0.9995
Pseudoneonatal Adrenoleukodystrophy	ACOX1	RECESSIVE	NM_004035.6	0.9967
PTEN Hamartoma Tumor Syndrome	PTEN	DOMINANT	NM_000314.4	0.9963
Pulmonary Surfactant Metabolism Dysfunction, Dominant	SFTPC	DOMINANT	NM_003018.3	0.9992
Pulmonary Surfactant Metabolism Dysfunction, Recessive	ABCA3	RECESSIVE	NM_001089.2	0.9982
	SFTPB	RECESSIVE	NM_198843.2	0.9975
Purine Nucleoside Phosphorylase Deficiency	PNP	RECESSIVE	NM_000270.3	0.9996

Pycnodysostosis	CTSK	RECESSIVE	NM_000396.3	0.9997
Pyogenic Sterile Arthritis, Pyoderma Gangrenosum, and Acne	PSTPIP1	DOMINANT	NM_003978.3	0.9985
Pyridoxamine 5-Prime-Phosphate Oxidase Deficiency	PNPO	RECESSIVE	NM_018129.3	0.9945
Pyridoxine-Dependent Epilepsy	ALDH7A1	RECESSIVE	NM_001182.4	0.9965
Pyridoxine-Refractory Sideroblastic Anemia	SLC25A38	RECESSIVE	NM_017875.2	0.9996
Pyruvate Carboxylase Deficiency	PC	RECESSIVE	NM_000920.3	0.9989
Pyruvate Dehydrogenase Complex Deficiency	DLAT	RECESSIVE	NM_001931.4	0.9996
	DLD	RECESSIVE	NM_000108.3	0.9998
	PDHB	RECESSIVE	NM_000925.3	0.9996
	PDHX	RECESSIVE	NM_003477.2	0.9992
Pyruvate Dehydrogenase Deficiency	PDHA1	X-LINKED	NM_000284.3	0.9991
Pyruvate Dehydrogenase Phosphatase Deficiency	PDP1	RECESSIVE	NM_018444.3	0.9992
Pyruvate Kinase Deficiency	PKLR	RECESSIVE	NM_000298.5	0.9989
Quebec Platelet Disorder	PLAU	DOMINANT	NM_002658.3	0.9994
Rapid-Onset Dystonia-Parkinsonism	ATP1A3	DOMINANT	NM_152296.4	0.9819
Recurrent Hydatidiform Mole	NLRP7	RECESSIVE	NM_206828.3	0.9915
Refsum Disease	PEX7	RECESSIVE	NM_000288	0.9996
	PHYH	RECESSIVE	NM_006214	0.9993
Renal Adysplasia	RET	DOMINANT	NM_020975	0.9990
	UPK3A	DOMINANT	NM_006953.3	0.9982
Renal Cysts and Diabetes Syndrome	HNF1B	DOMINANT	NM_000458.2	0.9897
Renal Glucosuria	SLC5A2	RECESSIVE (Rec/Dom)	NM_003041.3	0.9990
Renal Hypomagnesemia, Dominant	CNNM2	DOMINANT	NM_017649.4	0.9992
	FXYD2	DOMINANT	NM_001680.4	0.9982
Renal Hypomagnesemia, Recessive	CLDN19	RECESSIVE	NM_148960.2	0.9929
	EGF	RECESSIVE	NM_001963.4	0.9996
Renal Hypouricemia	SLC22A12	RECESSIVE	NM_144585.2	0.9981
	SLC2A9	DOMINANT (Dom/Rec)	NM_020041.2	0.9968
Renal Tubular Dysgenesis	ACE	RECESSIVE	NM_000789.3	0.9957
	AGT	RECESSIVE	NM_000029.3	0.9994
	AGTR1	RECESSIVE	NM_031850.3	0.9999

Renal Tubular Dysgenesis	REN	RECESSIVE	NM_000537.3	0.9992
Renal-Hepatic-Pancreatic Dysplasia	NPHP3	RECESSIVE	NM_153240.4	0.9976
Renpenning Syndrome	PQBP1	X_LINKED	NM_005710.2	0.9976
Retinal Cone Dystrophy	CACNA2D4	RECESSIVE	NM_172364.4	0.9992
	KCNV2	RECESSIVE	NM_133497.3	0.9993
	PDE6H	RECESSIVE	NM_006205.2	0.9996
Retinal Degeneration	C1QTNF5	DOMINANT	NM_015645.3	0.9957
Retinal Dystrophy	LRAT	RECESSIVE	NM_004744	0.9975
	OTX2	DOMINANT	NM_172337.2	0.9995
Retinal Macular Dystrophy	PROM1	DOMINANT	NM_006017.2	0.9995
Retinal Vasculopathy with Cerebral Leukodystrophy	TREX1	DOMINANT	NM_033629.2	0.9983
Retinitis Pigmentosa, Dominant	AIPL1	DOMINANT	NM_014336.3	0.9991
	CA4	DOMINANT	NM_000717	0.9986
	CLRN1	RECESSIVE	NM_174878.2	0.9996
	CRX	DOMINANT	NM_000554	0.9939
	GUCA1B	DOMINANT	NM_002098	0.9988
	IMPDH1	DOMINANT	NM_000883.3	0.9984
	KLHL7	DOMINANT	NM_001031710	0.9997
	NRL	DOMINANT	NM_006177	0.9984
	PRPF3	DOMINANT	NM_004698	0.9994
	PRPF31	DOMINANT	NM_015629	0.9971
	PRPF6	DOMINANT	NM_012469.3	0.9995
	PRPF8	DOMINANT	NM_006445	0.9994
	PRPH2	DOMINANT	NM_000322	0.9989
	ROM1	DOMINANT	NM_000327	0.9968
	RP1	DOMINANT	NM_006269	0.9998
	RP9	DOMINANT	NM_203288.1	0.9991
	SNRNP200	DOMINANT	NM_014014.4	0.9994
	TOPORS	DOMINANT	NM_005802.4	0.9998
	ZNF513	RECESSIVE	NM_144631.5	0.9932
Retinitis Pigmentosa, Dominant/Recessive	RHO	DOMINANT (Dom/Rec)	NM_000539	0.9982

Retinitis Pigmentosa, Dominant/Recessive	SEMA4A	DOMINANT (Dom/Rec)	NM_022367	0.9965
Retinitis Pigmentosa, Recessive	AIPL1	RECESSIVE	NM_014336.3	0.9991
	ARL6	RECESSIVE	NM_177976.1	0.9996
	BEST1	RECESSIVE	NM_004183	0.9992
	C2orf71	RECESSIVE	NM_001029883.2	0.9957
	C8orf37	RECESSIVE	NM_177965.3	0.9993
	CERKL	RECESSIVE	NM_201548	0.9998
	CNGA1	RECESSIVE	NM_000087	0.9996
	CNGB1	RECESSIVE	NM_001297	0.9952
	CRB1	RECESSIVE	NM_201253	0.9998
	DHDDS	RECESSIVE	NM_024887.3	0.9976
	EYS	RECESSIVE	NM_001142800	0.9998
	FAM161A	RECESSIVE	NM_032180.2	0.9995
	IDH3B	RECESSIVE	NM_006899.3	0.9995
	IMPG2	RECESSIVE	NM_016247.3	0.9943
	LRAT	RECESSIVE	NM_004744	0.9975
	MAK	RECESSIVE	NM_001242957.1	0.9991
	MERTK	RECESSIVE	NM_006343	0.9935
	NR2E3	RECESSIVE	NM_014249	0.9986
	PDE6A	RECESSIVE	NM_000440	0.9966
	PDE6B	RECESSIVE	NM_000283	0.9942
	PDE6G	RECESSIVE	NM_002602.3	0.9997
	PRCD	RECESSIVE	NM_001077620.2	0.9976
	PROM1	RECESSIVE	NM_006017.2	0.9995
	RBP3	RECESSIVE	NM_002900.2	0.9993
	RDH12	RECESSIVE	NM_152443	0.9998
	RGR	RECESSIVE	NM_001012720	0.9992
	RLBP1	RECESSIVE	NM_000326.4	0.9995
	RPE65	RECESSIVE	NM_000329	0.9998
	SAG	RECESSIVE	NM_000541	0.9995
	SPATA7	RECESSIVE	NM_018418.4	0.9983

Retinitis Pigmentosa, Recessive	TTC8	RECESSIVE	NM_198309.2	0.9997
	TULP1	RECESSIVE	NM_003322	0.9977
	USH2A	RECESSIVE	NM_007123	0.9997
Retinitis Pigmentosa, X-linked	RP2	X_LINKED	NM_006915	0.9998
Retinoblastoma	RB1	DOMINANT	NM_000321.2	0.9977
Revesz Syndrome	TINF2	DOMINANT	NM_001099274.1	0.9987
Rhabdoid Tumor Predisposition Syndrome	SMARCB1	DOMINANT	NM_003073.3	0.9914
Rhizomelic Chondrodysplasia Punctata	AGPS	RECESSIVE	NM_003659.3	0.9992
	GNPAT	RECESSIVE	NM_014236.3	0.9998
	PEX7	RECESSIVE	NM_000288	0.9996
Ribose 5-Phosphate Isomerase Deficiency	RPIA	RECESSIVE	NM_144563.2	0.9984
Ring Dermoid of Cornea	PITX2	DOMINANT	NM_153427.2	0.9962
Roberts Syndrome	ESCO2	RECESSIVE	NM_001017420.2	0.9990
Robinow Syndrome	ROR2	RECESSIVE	NM_004560.3	0.9990
	WNT5A	DOMINANT	NM_003392.4	0.9989
Romano-Ward Syndrome	AKAP9	DOMINANT	NM_005751	0.9997
	CAV3	DOMINANT	NM_033337	0.9991
	KCNE1	DOMINANT	NM_000219	0.9993
	KCNE2	DOMINANT	NM_172201	0.9998
	KCNJ5	DOMINANT	NM_000890.3	0.9937
	KCNQ1	DOMINANT	NM_000218	0.9984
	SCN4B	DOMINANT	NM_174934	0.9958
	SCN5A	DOMINANT	NM_198056	0.9983
	SNTA1	DOMINANT	NM_003098	0.9986
Rotor Syndrome	SLCO1B1	RECESSIVE	NM_006446.4	0.9966
	SLCO1B3	RECESSIVE	NM_019844.3	0.9999
Roussy-Levy Syndrome	MPZ	DOMINANT	NM_000530	0.9865
Rubinstein-Taybi Syndrome	EP300	DOMINANT	NM_001429.3	0.9972
Saethre-Chotzen Syndrome	FGFR2	DOMINANT	NM_000141.4	0.9980
Salih Myopathy	TTN	RECESSIVE	NM_133378	0.9885
SALL4-Related Spectrum Disorders	SALL4	DOMINANT	NM_020436.3	0.9994

Salla Disease	SLC17A5	RECESSIVE	NM_012434	0.9990
Sandhoff Disease	HEXB	RECESSIVE	NM_000521	0.9985
SCAD Deficiency	ACADS	RECESSIVE	NM_000017	0.9991
Scapuloperoneal Myopathy	MYH7	DOMINANT	NM_000257	0.9987
Scapuloperoneal Spinal Muscular Atrophy	TRPV4	DOMINANT	NM_021625	0.9968
Schimke Immunoosseous Dysplasia	SMARCAL1	RECESSIVE	NM_014140.3	0.9988
Schindler Disease	NAGA	RECESSIVE	NM_000262	0.9995
Schinzel-Giedion Midface Retraction Syndrome	SETBP1	DOMINANT	NM_015559.2	0.9868
Schnyder Crystalline Corneal Dystrophy	UBIAD1	DOMINANT	NM_013319.2	0.9990
Schopf-Schulz-Passarge Syndrome	WNT10A	RECESSIVE	NM_025216.2	0.9985
Schwannomatosis	SMARCB1	DOMINANT	NM_003073.3	0.9914
Schwartz-Jampel Syndrome	HSPG2	RECESSIVE	NM_005529.5	0.9966
Sclerosing Bone Dysplasias	SOST	RECESSIVE	NM_025237.2	0.9954
SCN1A-Related Seizure Disorders	SCN1A	DOMINANT	NM_001165963.1	0.9998
Seckel Syndrome	ATR	RECESSIVE	NM_001184.3	0.9990
	CENPJ	RECESSIVE	NM_018451.4	0.9995
	CEP152	RECESSIVE	NM_014985.3	0.9973
	RBBP8	RECESSIVE	NM_002894.2	0.9995
Seizures, Sensorineural Deafness, Ataxia, Intellectual Disability, and Electrolyte Imbalance Syndrome	KCNJ10	RECESSIVE	NM_002241.4	0.9976
Selective Tooth Agenesis	PAX9	DOMINANT	NM_006194.3	0.9991
	WNT10A	DOMINANT	NM_025216.2	0.9985
Sengers Syndrome	AGK	RECESSIVE	NM_018238.3	0.9997
Senior-Loken Syndrome	CEP290	RECESSIVE	NM_025114.3	0.9998
	IQCB1	RECESSIVE	NM_001023570.2	0.9998
	NPHP1	RECESSIVE	NM_000272.3	0.9978
	NPHP4	RECESSIVE	NM_015102.3	0.9970
	SDCCAG8	RECESSIVE	NM_006642.3	0.9997
Sensory Neuropathy with Spastic Paraplegia	CCT5	RECESSIVE	NM_012073.3	0.9966
SEPN1-Related Disorders	SEPN1	RECESSIVE (Rec/Dom)	NM_020451.2	0.9961
Septooptic Dysplasia	HESX1	RECESSIVE (Rec/Dom)	NM_003865.2	0.9997

Severe Combined Immune Deficiency	CD3D	RECESSIVE	NM_000732.4	0.9994
	CD3E	RECESSIVE	NM_000733.3	0.9808
	IL2RG	X_LINKED	NM_000206.2	0.9993
	IL7R	RECESSIVE	NM_002185.3	0.9998
	JAK3	RECESSIVE	NM_000215.3	0.9929
	RAG1	RECESSIVE	NM_000448	0.9998
	RAG2	RECESSIVE	NM_000536	0.9997
	ZAP70	RECESSIVE	NM_001079.3	0.9991
Severe Combined Immunodeficiency with Sensitivity to Ionizing Radiation	LIG4	RECESSIVE	NM_002312.3	0.9879
Severe Congenital Neutropenia	G6PC3	RECESSIVE	NM_138387.3	0.9990
	GFI1	DOMINANT	NM_005263.3	0.9992
	HAX1	RECESSIVE	NM_006118.3	0.9994
Short QT Syndrome	KCNJ2	DOMINANT	NM_000891	0.9927
	KCNQ1	DOMINANT	NM_000218	0.9984
Short Rib Polydactyly Syndrome	DYNC2H1	RECESSIVE	NM_001080463.1	0.9998
	NEK1	RECESSIVE	NM_012224.2	0.9997
	WDR35	RECESSIVE	NM_001006657.1	0.9998
Sialuria	GNE	DOMINANT	NM_005476.5	0.9981
Sick Sinus Syndrome	HCN4	RECESSIVE	NM_005477.2	0.9963
	SCN5A	RECESSIVE	NM_198056	0.9983
Sickle Cell Disease	HBB	RECESSIVE	NM_000518	0.9997
Sideroblastic Anemia and Ataxia	ABCB7	X_LINKED	NM_004299.4	0.9996
	ALAS2	X_LINKED	NM_000032.4	0.9995
Sitosterolemia	ABCG5	RECESSIVE	NM_022436.2	0.9980
	ABCG8	RECESSIVE	NM_022437.2	0.9993
Sjogren-Larsson Syndrome	ALDH3A2	RECESSIVE	NM_000382.2	0.9995
Skin Fragility-Woolly Hair Syndrome	DSP	RECESSIVE	NM_004415	0.9996
SLC6A4-Related Behavior Disorders	SLC6A4	DOMINANT	NM_001045.5	0.9992
Small Fiber Neuropathy	SCN9A	DOMINANT	NM_002977.3	0.9997
Small Patella Syndrome	TBX4	DOMINANT	NM_018488.2	0.9981

Smith-Lemli-Opitz Syndrome	DHCR7	RECESSIVE	NM_001360	0.9987
Smith-McCort Dysplasia	DYM	RECESSIVE	NM_017653.3	0.9989
	RAB33B	RECESSIVE	NM_031296.1	0.9958
Sotos Syndrome	NSD1	DOMINANT	NM_022455.4	0.9960
Spastic Paraplegia, Dominant	ATL1	DOMINANT	NM_015915.4	0.9997
	HSPD1	DOMINANT	NM_002156.4	0.9992
	KIAA0196	DOMINANT	NM_014846.3	0.9996
	KIF5A	DOMINANT	NM_004984.2	0.9982
	NIPA1	DOMINANT	NM_144599.4	0.9977
	REEP1	DOMINANT	NM_022912.2	0.9952
	RTN2	DOMINANT	NM_206900.1	0.9980
	SLC33A1	DOMINANT	NM_004733.3	0.9998
	SPAST	DOMINANT	NM_014946.3	0.9994
	ZFYVE27	DOMINANT	NM_001002261.3	0.9990
Spastic Paraplegia, Recessive	AP5Z1	RECESSIVE	NM_014855.2	0.9958
	CYP7B1	RECESSIVE	NM_004820.3	0.9996
	KIF1A	RECESSIVE	NM_004321.6	0.9979
	PNPLA6	RECESSIVE	NM_006702.4	0.9973
	SPG11	RECESSIVE	NM_025137.3	0.9997
	SPG21	RECESSIVE	NM_016630.3	0.9992
	SPG7	RECESSIVE	NM_003119.2	0.9985
	ZFYVE26	RECESSIVE	NM_015346.3	0.9979
Spastic Quadriplegic Cerebral Palsy	GAD1	RECESSIVE	NM_000817.2	0.9982
Speech-Language Disorder	FOXP2	DOMINANT	NM_014491.3	0.9994
Spermatogenic Failure	AURKC	RECESSIVE	NM_001015878.1	0.9641
	SPATA16	RECESSIVE	NM_031955.5	0.9996
	SYCP3	DOMINANT	NM_153694.4	0.9985
Spherocytosis, Dominant	ANK1	DOMINANT (Dom/Rec)	NM_000037	0.9979
	SLC4A1	DOMINANT	NM_000342.3	0.9913
	SPTB	DOMINANT (Dom/Rec)	NM_000347	0.9968
Spherocytosis, Recessive	EPB42	RECESSIVE	NM_000119	0.9992

Spherocytosis, Recessive	SPTA1	RECESSIVE	NM_003126	0.9965
Spheroid Body Myopathy	MYOT	DOMINANT	NM_006790	0.9996
Spinal Muscular Atrophy	IGHMBP2	RECESSIVE	NM_002180	0.9921
Spinal Muscular Atrophy, Dominant	VAPB	DOMINANT	NM_004738	0.9973
Spinocerebellar Ataxia, Dominant	AFG3L2	DOMINANT	NM_006796.2	0.9998
	DYNC1H1	DOMINANT	NM_001376.4	0.9994
	FGF14	DOMINANT	NM_004115.3	0.9997
	ITPR1	DOMINANT	NM_002222.5	0.9982
	PDYN	DOMINANT	NM_024411.4	0.9992
	PRKCG	DOMINANT	NM_002739.3	0.9857
	TGM6	DOMINANT	NM_198994.2	0.9993
	TTBK2	DOMINANT	NM_173500.3	0.9991
Spinocerebellar Ataxia, Recessive	ADCK3	RECESSIVE	NM_020247.4	0.9983
	ANO10	RECESSIVE	NM_018075.3	0.9989
	C10orf2	RECESSIVE	NM_021830.4	0.9991
	SPTBN2	RECESSIVE (Rec/Dom)	NM_006946.2	0.9919
	TDP1	RECESSIVE	NM_018319.3	0.9992
Split-Hand/Foot Malformation	FBXW4	DOMINANT	NM_022039.3	0.9977
Spondylocostal Dysostosis	DLL3	RECESSIVE	NM_016941.3	0.9988
	MESP2	RECESSIVE	NM_001039958.1	0.9896
Spondyloepimetaphyseal Dysplasia	DDR2	RECESSIVE	NM_006182.2	0.9982
	MMP13	DOMINANT	NM_002427.3	0.9992
Spondyloepiphyseal Dysplasia	TRAPPC2	X-LINKED	NM_001011658.3	0.9822
Spondylometaphyseal Dysplasia	TRPV4	DOMINANT	NM_021625	0.9968
Spontaneous Pneumothorax	FLCN	DOMINANT	NM_144997.5	0.9970
Stargardt Disease, Dominant	ELOVL4	DOMINANT	NM_022726	0.9997
	PROM1	DOMINANT	NM_006017.2	0.9995
Stargardt Disease, Recessive	CNGB3	RECESSIVE	NM_019098.4	0.9991
Steroid 5-Alpha-Reductase Deficiency	SRD5A2	RECESSIVE	NM_000348.3	0.9996
Steroid-Resistant Nephrotic Syndrome	NPHS2	RECESSIVE	NM_014625.2	0.9992
Stickler Syndrome, Dominant	COL11A1	DOMINANT	NM_001854.3	0.9996

Stickler Syndrome, Dominant	COL11A2	DOMINANT	NM_080680	0.9952
	COL2A1	DOMINANT	NM_001844.4	0.9990
Stickler Syndrome, Recessive	COL9A1	RECESSIVE	NM_001851.4	0.9995
	COL9A2	RECESSIVE	NM_001852.3	0.9951
Stiff Skin Syndrome	FBN1	DOMINANT	NM_000138.4	0.9997
Striatal Degeneration	PDE8B	DOMINANT	NM_003719.3	0.9989
Stuve-Wiedemann Syndrome	LIFR	RECESSIVE	NM_002310.5	0.9996
Succinic Semialdehyde Dehydrogenase Deficiency	ALDH5A1	RECESSIVE	NM_001080.3	0.9994
Succinyl-CoA:3-Oxoacid CoA Transferase Deficiency	OXCT1	RECESSIVE	NM_000436.3	0.9998
Sulfate Transporter-Related Osteochondrodysplasia	SLC26A2	RECESSIVE	NM_000112.3	0.9998
Sulfocysteinuria	SUOX	RECESSIVE	NM_000456.2	0.9995
Supravalvular Aortic Stenosis	ELN	DOMINANT	NM_000501.2	0.9931
Susceptibility to Nonalcoholic Fatty Liver Disease	PNPLA3	DOMINANT	NM_025225.2	0.9989
Syndactyly	DLL3	RECESSIVE	NM_016941.3	0.9988
	GJA1	DOMINANT	NM_000165.3	0.9996
Syndromic Microphthalmia, Dominant	BMP4	DOMINANT	NM_001202.3	0.9959
	OTX2	DOMINANT	NM_172337.2	0.9995
Syndromic Microphthalmia, Recessive	STRA6	RECESSIVE	NM_022369.3	0.9986
Systemic Primary Carnitine Deficiency	SLC22A5	RECESSIVE	NM_003060	0.9990
Tangier Disease	ABCA1	RECESSIVE	NM_005502.3	0.9994
Tetra-Amelia Syndrome	WNT3	RECESSIVE	NM_030753.3	0.9944
Thiamine Metabolism Dysfunction Syndrome	SLC19A3	RECESSIVE	NM_025243.3	0.9990
Thiamine-Responsive Megaloblastic Anemia Syndrome	SLC19A2	RECESSIVE	NM_006996.2	0.9997
Thoracic Aortic Aneurysms and Aortic Dissections	ACTA2	DOMINANT	NM_001613.2	0.9995
	FBN1	DOMINANT	NM_000138.4	0.9997
	MYH11	DOMINANT	NM_002474.2	0.9987
	MYLK	DOMINANT	NM_053025.3	0.9986
	SMAD3	DOMINANT	NM_005902.3	0.9974
	TGFBR1	DOMINANT	NM_004612.2	0.9992
	TGFBR2	DOMINANT	NM_003242.5	0.9995
Thrombasthenia of Glanzmann and Naegeli	ITGA2B	RECESSIVE	NM_000419.3	0.9985

Thrombasthenia of Glanzmann and Naegeli	ITGB3	RECESSIVE	NM_000212.2	0.9996
Thrombocytopenia	ANKRD26	DOMINANT	NM_014915.2	0.9998
	CYCS	DOMINANT	NM_018947.5	0.9990
	MASTL	DOMINANT	NM_032844.3	0.9894
Thyroid Dyshormonogenesis	TG	RECESSIVE	NM_003235.4	0.9986
Thyroid Hormone Resistance	THRB	DOMINANT (Dom/Rec)	NM_000461.4	0.9968
Thyroid Hormonogenesis Defect	SLC5A5	RECESSIVE	NM_000453.2	0.9936
Tietz Syndrome	MITF	DOMINANT	NM_000248.3	0.9997
Timothy Syndrome	CACNA1C	DOMINANT	NM_000719	0.9986
Tourette Syndrome	SLITRK1	DOMINANT	NM_052910.1	0.9887
Townes-Brocks Syndrome	SALL1	DOMINANT	NM_002968.2	0.9974
TP63-Related Spectrum Disorders	TP63	DOMINANT	NM_003722.4	0.9997
Transaldolase Deficiency	TALDO1	RECESSIVE	NM_006755.1	0.9976
Transcobalamin II Deficiency	TCN2	RECESSIVE	NM_000355.3	0.9980
Transient Familial Neonatal Hyperbilirubinemia	UGT1A1	RECESSIVE (Rec/Dom)	NM_000463	0.9995
Transient Neonatal Diabetes, Dominant	ABCC8	DOMINANT	NM_000352	0.9992
	KCNJ11	DOMINANT	NM_000525	0.9936
Transient Neonatal Diabetes, Dominant/Recessive	INS	DOMINANT	NM_000207.2	0.9982
Transient Neonatal Diabetes, Recessive	GCK	RECESSIVE	NM_000162	0.9990
	ZFP57	RECESSIVE	NM_001109809.2	0.9990
Treacher Collins Syndrome, Dominant	POLR1D	DOMINANT (Dom/Rec)	NM_015972.3	0.9993
	TCOF1	DOMINANT	NM_001135243.1	0.9983
Treacher Collins Syndrome, Recessive	POLR1C	RECESSIVE	NM_203290.2	0.9949
Trichohepatoenteric Syndrome	SKIV2L	RECESSIVE	NM_006929.4	0.9993
Trichorhinophalangeal Syndrome	TRPS1	DOMINANT	NM_014112.2	0.9970
Trifunctional Protein Deficiency	HADHA	RECESSIVE	NM_000182	0.9995
	HADHB	RECESSIVE	NM_000183	0.9996
Trimethylaminuria	FMO3	RECESSIVE	NM_006894.5	0.9997
Triosephosphate Isomerase Deficiency	TPI1	RECESSIVE	NM_000365.5	0.9955
Triphalangeal Thumb-Polysyndactyly Syndrome	LMBR1	DOMINANT	NM_022458.3	0.9995
Trismus-Pseudocamptodactyly Syndrome	MYH8	DOMINANT	NM_002472.2	0.9997

Troyer syndrome	SPG20	RECESSIVE	NM_015087.4	0.9991
Tryptophan Hydroxylase Deficiency	TPH2	DOMINANT	NM_173353.3	0.9825
Tuberous Sclerosis	TSC1	DOMINANT	NM_000368.4	0.9977
	TSC2	DOMINANT	NM_000548.3	0.9968
Tumor Predisposition Syndrome	BAP1	DOMINANT	NM_004656.2	0.9984
Tylosis with Esophageal Cancer	RHBDF2	DOMINANT	NM_024599.5	0.9982
Type II Collagenopathies	COL2A1	DOMINANT	NM_001844.4	0.9990
Tyrosine Hydroxylase Deficiency	TH	RECESSIVE	NM_000360.3	0.9952
Tyrosinemia	FAH	RECESSIVE	NM_000137	0.9830
	HPD	RECESSIVE	NM_002150	0.9978
	TAT	RECESSIVE	NM_000353	0.9993
Udd Distal Myopathy	TTN	DOMINANT	NM_133378	0.9885
Ulnar-Mammary Syndrome	TBX3	DOMINANT	NM_005996.3	0.9989
Unverricht-Lundborg Disease	CSTB	RECESSIVE	NM_000100.3	0.9987
Uromodulin-associated kidney disease	UMOD	DOMINANT	NM_003361.2	0.9975
Usher Syndrome	ADGRV1	RECESSIVE	NM_032119.3	0.9997
	CDH23	RECESSIVE	NM_022124	0.9973
	CLRN1	RECESSIVE	NM_174878.2	0.9996
	DFNB31	RECESSIVE	NM_015404	0.9992
	HARS	RECESSIVE	NM_002109.4	0.9993
	MYO7A	RECESSIVE	NM_000260	0.9980
	PCDH15	RECESSIVE	NM_033056	0.9997
	USH1C	RECESSIVE	NM_005709	0.9889
	USH1G	RECESSIVE	NM_173477.2	0.9984
	USH2A	RECESSIVE	NM_007123	0.9997
VACTERL Association with Hydrocephalus	FANCB	X_LINKED	NM_001018113	0.9998
VACTERL Association, X-linked	ZIC3	X_LINKED	NM_003413.3	0.9934
van der Woude Syndrome	IRF6	DOMINANT	NM_006147.3	0.9995
Variegate Porphyria	PPOX	DOMINANT	NM_000309.3	0.9988
Vesicoureteral Reflux	ROBO2	DOMINANT	NM_002942.4	0.9991
Vitamin D-Dependent Rickets	CYP27B1	RECESSIVE	NM_000785.3	0.9992

Vitamin D-Dependent Rickets	VDR	RECESSIVE	NM_001017535.1	0.9975
Vitamin K-Dependent Clotting Factors	GGCX	RECESSIVE	NM_000821.5	0.9992
	VKORC1	RECESSIVE	NM_024006.4	0.9976
Vitelliform Macular Dystrophy	PRPH2	DOMINANT	NM_000322	0.9989
Vitreoretinochoroidopathy	BEST1	DOMINANT	NM_004183	0.9992
Vitreoretinopathy	VCAN	DOMINANT	NM_004385.4	0.9991
VLCAD Deficiency	ACADVL	RECESSIVE	NM_000018	0.9982
VLDLR-Related Cerebellar Hypoplasia	VLDLR	RECESSIVE	NM_003383.3	0.9933
Vohwinkel Syndrome	GJB2	DOMINANT	NM_004004	0.9995
Von Hippel-Lindau Syndrome	VHL	DOMINANT	NM_000551.3	0.9805
von Willebrand Disease (Dominant/Recessive)	VWF	DOMINANT (Dom/Rec)	NM_000552.3	0.9969
VSX2-related Microphthalmia	VSX2	RECESSIVE	NM_182894.2	0.9988
Waardenburg Syndrome	EDN3	DOMINANT (Dom/Rec)	NM_207034.1	0.9983
	EDNRB	DOMINANT (Dom/Rec)	NM_000115.3	0.9996
	MITF	DOMINANT (Dom/Rec)	NM_000248.3	0.9997
	PAX3	DOMINANT (Dom/Rec)	NM_181457.3	0.9996
	SNAI2	DOMINANT (Dom/Rec)	NM_003068.4	0.9999
	SOX10	DOMINANT (Dom/Rec)	NM_006941.3	0.9944
Wagner Syndrome	VCAN	DOMINANT	NM_004385.4	0.9991
WAGR Syndrome	PAX6	DOMINANT	NM_000280.4	0.9984
	WT1	DOMINANT	NM_024426.4	0.9923
Walker-Warburg Syndrome	LARGE	RECESSIVE	NM_004737.4	0.9988
Warburg Micro Syndrome	RAB18	RECESSIVE	NM_021252.4	0.9996
	RAB3GAP1	RECESSIVE	NM_012233.2	0.9984
	RAB3GAP2	RECESSIVE	NM_012414.3	0.9991
Watson Syndrome	NF1	DOMINANT	NM_000267	0.9956
Weaver Syndrome	EZH2	DOMINANT	NM_004456.4	0.9996
	NSD1	DOMINANT	NM_022455.4	0.9960
Weill-Marchesani Syndrome (Dominant/Recessive)	ADAMTS10	RECESSIVE	NM_030957.2	0.9966
	FBN1	DOMINANT	NM_000138.4	0.9997
	LTBP2	RECESSIVE	NM_000428.2	0.9992

Weill-Marchesani-Like Syndrome	ADAMTS17	RECESSIVE	NM_139057.2	0.9961
Weissenbacher-Zweymuller Syndrome	COL11A2	DOMINANT	NM_080680	0.9952
Werner Syndrome	WRN	RECESSIVE	NM_000553.4	0.9963
Weyers Acrofacial Dysostosis	EVC	DOMINANT	NM_153717.2	0.9767
WFS1-Related Spectrum Disorders	WFS1	RECESSIVE (Rec/Dom)	NM_006005	0.9984
White Sponge Nevus of Cannon	KRT13	DOMINANT	NM_002274.3	0.9994
	KRT4	DOMINANT	NM_002272.3	0.9994
Wilms Tumor	WT1	DOMINANT	NM_024426.4	0.9923
Wilson Disease	ATP7B	RECESSIVE	NM_000053	0.9993
Wolff-Parkinson-White Syndrome	PRKAG2	DOMINANT	NM_016203	0.9936
Wolf-Hirschhorn Syndrome	WHSC1	DOMINANT	NM_133330.2	0.9990
Wolman Disease	LIPA	RECESSIVE	NM_000235	0.9997
Woodhouse-Sakati Syndrome	DCAF17	RECESSIVE	NM_025000.3	0.9998
Xanthinuria	XDH	RECESSIVE	NM_000379.3	0.9994
Xeroderma Pigmentosum	DDB2	RECESSIVE	NM_000107.2	0.9987
	ERCC1	RECESSIVE	NM_202001.2	0.9976
	ERCC2	RECESSIVE	NM_000400.3	0.9889
	ERCC3	RECESSIVE	NM_000122.1	0.9996
	ERCC4	RECESSIVE	NM_005236.2	0.9997
	ERCC5	RECESSIVE	NM_000123.3	0.9995
	POLH	RECESSIVE	NM_006502.2	0.9966
	XPA	RECESSIVE	NM_000380.3	0.9999
	XPC	RECESSIVE	NM_004628.4	0.9991
X-linked Heterotaxy, Visceral, 1	ZIC3	X_LINKED	NM_003413.3	0.9934
X-Linked Hypophosphatemia	PHEX	X_LINKED	NM_000444.4	0.9995
Zaspopathy	LDB3	DOMINANT	NM_001080116	0.9928
Zellweger Syndrome	PEX1	RECESSIVE	NM_000466.2	0.9998
	PEX10	RECESSIVE	NM_153818.1	0.9993
	PEX12	RECESSIVE	NM_000286.2	0.9997
	PEX13	RECESSIVE	NM_002618.3	0.9998
	PEX14	RECESSIVE	NM_004565.2	0.9895

Zellweger Syndrome	PEX16	RECESSIVE	NM_004813.2	0.9992
	PEX19	RECESSIVE	NM_002857.3	0.9997
	PEX2	RECESSIVE	NM_000318.2	0.9995
	PEX26	RECESSIVE	NM_017929.5	0.9967
	PEX3	RECESSIVE	NM_003630.2	0.9997
	PEX5	RECESSIVE	NM_001131025.1	0.9825
	PEX6	RECESSIVE	NM_000287.3	0.9888
Zonular Pulverulent Cataract	GJA3	DOMINANT (Dom/Rec)	NM_021954.3	0.9993
	GJA8	DOMINANT (Dom/Rec)	NM_005267.4	0.9994