Individual Genome Sequence Predisposition and Carrier Screening Test Gene List (By Disease)

Disease	Associated Gene	Mode of Inheritance	Transcript ID
17-Beta-Hydroxysteroid Dehydrogenase III Deficiency	HSD17B3	Autosomal Recessive	NM_000197.1
3-Hydroxy-3-Methylglutaryl-CoA Synthase 2 Deficiency	HMGCS2	Autosomal Recessive	NM_005518.3
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	HMGCL	Autosomal Recessive	NM_000191.2
3-Hydroxyacyl-Coenzyme A Dehydrogenase Deficiency	HADH	Autosomal Recessive	NM_005327.4
3-M Syndrome	CUL7	Autosomal Recessive	NM_014780.4
3-MCC Deficiency	MCCC1	Autosomal Recessive	NM_020166.3
3-MCC Deficiency	MCCC2	Autosomal Recessive	NM_022132.4
3-Methylglutaconic Aciduria, Type 1	AUH	Autosomal Recessive	NM_001698.2
3-Methylglutaconic Aciduria, Type 2	TAZ	X-Linked	NM_000116.3
3-Methylglutaconic Aciduria, Type 3	OPA3	Autosomal Recessive	NM_025136.3
3-Methylglutaconic Aciduria, Type 5	DNAJC19	Autosomal Recessive	NM_145261.3
46,XY DSD/46,XY CGD	DHH	Autosomal Recessive	NM_021044.2
6-Pyruvoyltetrahydropterin Synthase Deficiency	PTS	Autosomal Dominant	NM_000317.2
Abetalipoproteinemia	MTTP	Autosomal Recessive	NM_000253.2
ACAD9 Deficiency	ACAD9	Autosomal Recessive	NM_014049.4
Aceruloplasminemia	CP	Autosomal Recessive	NM_000096.3
Achalasia-Addisonianism-Alacrima Syndrome	AAAS	Autosomal Recessive	NM_015665.5
Achondrogenesis	SLC26A2	Autosomal Dominant	NM_000112.3
Achondrogenesis	TRIP11	Autosomal Dominant	NM_004239.3
Achromatopsia	CNGA3	Autosomal Recessive	NM_001298.2
Achromatopsia	CNGB3	Autosomal Recessive	NM_019098.4

Achromatopsia	GNAT2	Autosomal Recessive	NM_005272.3
Achromatopsia	PDE6C	Autosomal Recessive	NM_006204.3
Acid Sphingomyelinase Deficiency	SMPD1	Autosomal Recessive	NM_000543.4
Acid-Labile Subunit Deficiency	IGFALS	Autosomal Recessive	NM_004970.2
Acrodermatitis Enteropathica	SLC39A4	Autosomal Recessive	NM_130849.2
Acrodysostosis	PDE4D	Autosomal Dominant	NM_001104631.1
Acrodysostosis	PRKAR1A	Autosomal Dominant	NM_002734.3
Acromesomelic Dysplasia	GDF5	Autosomal Recessive	NM_000557.2
Acromesomelic Dysplasia	NPR2	Autosomal Recessive	NM_003995.3
Acromicric Dysplasia	FBN1	Autosomal Dominant	NM_000138.4
ACTH Deficiency	TBX19	Autosomal Recessive	NM_005149.2
Acute Hepatic Porphyria	ALAD	Autosomal Dominant	NM_000031.5
Acute Infantile Liver Failure	TRMU	Autosomal Recessive	NM_018006.4
Acute Recurrent Myoglobinuria	LPIN1	Autosomal Dominant	NM_145693.2
Acyl-CoA Dehydrogenase, Short/Branched Chain Deficiency	ACADSB	Autosomal Recessive	NM_001609.3
Adams-Oliver Syndrome	ARHGAP31	Autosomal Dominant	NM_020754.2
Adenine Phosphoribosyltransferase Deficiency	APRT	Autosomal Recessive	NM_000485.2
Adenosine Deaminase Deficiency	ADA	Autosomal Recessive	NM_000022.2
Adenylosuccinase Deficiency	ADSL	Autosomal Recessive	NM_000026.2
Adult Polyglucosan Body Disease	GBE1	Autosomal Recessive	NM_000158.3
Age-Related Cortical Cataract	EPHA2	Autosomal Dominant	NM_004431.3
Aicardi-Goutieres Syndrome	RNASEH2A	Autosomal Recessive	NM_006397.2
Aicardi-Goutieres Syndrome	RNASEH2B	Autosomal Recessive	NM_024570.3
Aicardi-Goutieres Syndrome	RNASEH2C	Autosomal Recessive	NM_032193.3
Aicardi-Goutieres Syndrome	SAMHD1	Autosomal Recessive	NM_015474.3
Alagille Syndrome	JAG1	Autosomal Dominant	NM_000214.2
Aldolase A Deficiency	ALDOA	Autosomal Recessive	NM_000034.3
Alexander Disease	GFAP	Autosomal Dominant	NM_002055.4

Alkaptonuria	HGD	Autosomal Recessive	NM_000187.3
Alopecia and T-Cell Immunodeficiency	FOXN1	Autosomal Recessive	NM_003593.2
Alopecia Universalis	HR	Autosomal Recessive	NM_005144.4
Alpha-1 Antitrypsin Deficiency	SERPINA1	Autosomal Recessive	NM_000295.4
Alpha-B Crystallinopathy	CRYAB	Autosomal Recessive	NM_001885.1
Alpha-Mannosidosis	MAN2B1	Autosomal Recessive	NM_000528.3
Alpha-Methylacyl-CoA Racemase Deficiency	AMACR	Autosomal Recessive	NM_014324.5
Alpha-Sarcoglycanopathy	SGCA	Autosomal Recessive	NM_000023.2
Alport Syndrome	COL4A3	Mixed	NM_000091.4
Alport Syndrome	COL4A4	Mixed	NM_000092.4
ALS2-Related Spectrum Disorders	ALS2	Autosomal Recessive	NM_020919.3
Alstrom Syndrome	ALMS1	Autosomal Recessive	NM_015120.4
Alternating Hemiplegia of Childhood	ATP1A2	Autosomal Dominant	NM_000702.3
Alternating Hemiplegia of Childhood	ATP1A3	Autosomal Dominant	NM_152296.4
Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins	FOXF1	Autosomal Dominant	NM_001451.2
Alzheimer Disease	APP	Autosomal Dominant	NM_000484.3
Alzheimer Disease	PSEN1	Autosomal Dominant	NM_000021.3
Alzheimer Disease	PSEN2	Autosomal Dominant	NM_000447.2
Amelogenesis Imperfecta, Dominant	DLX3	Autosomal Dominant	NM_005220.2
Amelogenesis Imperfecta, Dominant	ENAM	Autosomal Dominant	NM_031889.2
Amelogenesis Imperfecta, Recessive	MMP20	Autosomal Recessive	NM_004771.3
Amelogenesis Imperfecta, Recessive	WDR72	Autosomal Recessive	NM_182758.2
Amish Infantile Epilepsy Syndrome	ST3GAL5	Autosomal Recessive	NM_003896.3
Amish Lethal Microcephaly	SLC25A19	Autosomal Recessive	NM_021734.4
Amyloidosis	GSN	Autosomal Dominant	NM_000177.4
Amyotrophic Lateral Sclerosis, Dominant	ANG	Autosomal Dominant	NM_001145.4
Amyotrophic Lateral Sclerosis, Dominant	FIG4	Autosomal Dominant	NM_014845.5
Amyotrophic Lateral Sclerosis, Dominant	SETX	Autosomal Dominant	NM_015046.5

Amyotrophic Lateral Sclerosis, Dominant	SOD1	Autosomal Dominant	NM_000454.4
Amyotrophic Lateral Sclerosis, Dominant	TARDBP	Autosomal Dominant	NM_007375.3
Amyotrophic Lateral Sclerosis, Dominant	VAPB	Autosomal Dominant	NM_004738.4
Amyotrophic Lateral Sclerosis, Dominant	VCP	Autosomal Dominant	NM_007126.3
Amyotrophic Lateral Sclerosis, Recessive	ALS2	Autosomal Recessive	NM_020919.3
Amyotrophic Lateral Sclerosis, Recessive	FUS	Autosomal Recessive	NM_004960.3
Amyotrophic Lateral Sclerosis, Recessive	OPTN	Autosomal Recessive	NM_021980.4
Amyotrophic Lateral Sclerosis/Frontotemporal Dementia	C9orf72	Autosomal Dominant	NM_001256054.1
Andersen Syndrome	KCNJ2	Autosomal Dominant	NM_000891.2
Andersen-Tawil Syndrome	KCNJ2	Autosomal Dominant	NM_000891.2
Angiokeratoma Corporis Diffusum with Arteriovenous Fistulas	KRIT1	Autosomal Dominant	NM_194456.1
Aniridia	PAX6	Autosomal Dominant	NM_000280.4
Aniridia, Cerebellar Ataxia, And Mental Retardation	PAX6	Autosomal Recessive	NM_000280.4
Anophthalmia	PAX6	Autosomal Dominant	NM_000280.4
Anophthalmia/Microphthalmia	SIX6	Autosomal Dominant	NM_007374.2
Antenatal Bartter Syndrome	KCNJ1	Autosomal Recessive	NM_000220.4
Antenatal Bartter Syndrome	SLC12A1	Autosomal Recessive	NM_000338.2
Anterior Segment Mesenchymal Dysgenesis	PITX2	Autosomal Dominant	NM_153427.2
Antithrombin-III Deficiency	SERPINC1	Autosomal Dominant	NM_000488.3
APC-Associated Polyposis Disorders	APC	Autosomal Dominant	NM_000038.5
Apert Syndrome	FGFR2	Autosomal Dominant	NM_000141.4
Aplasia of Lacrimal and Salivary Glands	FGF10	Autosomal Dominant	NM_004465.1
Aplastic Anemia	IFNG	Autosomal Recessive	NM_000619.2
Apolipoprotein C-II Deficiency	APOC2	Autosomal Recessive	NM_000483.4
Arginase Deficiency	ARG1	Autosomal Recessive	NM_000045.2
Argininosuccinate Lyase Deficiency	ASL	Autosomal Recessive	NM_000048.3
Aromatase Deficiency	CYP19A1	Autosomal Recessive	NM_031226.2
Aromatic L-Amino Acid Decarboxylase Deficiency	DDC	Autosomal Recessive	NM_000790.3

ARSACS	SACS	Autosomal Recessive	NM_014363.4
Arterial Tortuosity Syndrome	SLC2A10	Autosomal Recessive	NM_030777.3
Arthrogryposis Multiplex Congenita	MYH3	Autosomal Dominant	NM_002470.3
Arthrogryposis Multiplex Congenita	TNNI2	Autosomal Dominant	NM_003282.3
Arthrogryposis Multiplex Congenita	TNNT3	Autosomal Dominant	NM_006757.3
Arthrogryposis Multiplex Congenita	TPM2	Autosomal Dominant	NM_003289.3
Arthrogryposis, Renal Dysfunction, and Cholestasis Syndrome	VPS33B	Autosomal Recessive	NM_018668.3
Arts Syndrome	PRPS1	X-Linked	NM_002764.3
Arylsulfatase A Deficiency	ARSA	Autosomal Recessive	NM_000487.5
Aspartylglycosaminuria	AGA	Autosomal Recessive	NM_000027.3
Asphyxiating Thoracic Dystrophy	DYNC2H1	Autosomal Recessive	NM_001080463.1
Asphyxiating Thoracic Dystrophy	IFT80	Autosomal Recessive	NM_020800.2
Asphyxiating Thoracic Dystrophy	TTC21B	Autosomal Recessive	NM_024753.4
Asphyxiating Thoracic Dystrophy	WDR19	Autosomal Recessive	NM_025132.3
Ataxia Neuropathy Spectrum Disorders	C10orf2	Autosomal Dominant	NM_021830.4
Ataxia with Oculomotor Apraxia	APTX	Autosomal Recessive	NM_175073.2
Ataxia with Oculomotor Apraxia	SETX	Autosomal Recessive	NM_015046.5
Ataxia with Vitamin E Deficiency	TTPA	Autosomal Recessive	NM_000370.3
Ataxia-Telangiectasia	ATM	Autosomal Recessive	NM_000051.3
Ataxia-Telangiectasia-Like Disorder	MRE11A	Autosomal Recessive	NM_005591.3
Atelosteogenesis	SLC26A2	Autosomal Recessive	NM_000112.3
Athabaskan Brainstem Dysgenesis Syndrome	HOXA1	Autosomal Recessive	NM_005522.4
Atransferrinemia	TF	Autosomal Recessive	NM_001063.3
Atrial Septal Defect	ACTC1	Autosomal Dominant	NM_005159.4
Atrial Septal Defect	MYH6	Autosomal Dominant	NM_002471.3
Atrichia with Papular Lesions	HR	Autosomal Recessive	NM_005144.4
Atypical Gaucher Disease	PSAP	Autosomal Dominant	NM_002778.2
Atypical Hemolytic-Uremic Syndrome	C3	Autosomal Dominant	NM_000064.2

Atypical Hemolytic-Uremic Syndrome	CD46	Autosomal Dominant	NM_002389.4
Atypical Hemolytic-Uremic Syndrome	CFB	Autosomal Dominant	NM_001710.5
Atypical Hemolytic-Uremic Syndrome	CFH	Autosomal Dominant	NM_000186.3
Atypical Hemolytic-Uremic Syndrome	CFI	Autosomal Dominant	NM_000204.3
Atypical Hemolytic-Uremic Syndrome	THBD	Autosomal Dominant	NM_000361.2
Atypical Werner Syndrome	LMNA	Autosomal Dominant	NM_005572.3
Auriculocondylar Syndrome	PLCB4	Autosomal Dominant	NM_000933.3
Autism Spectrum Disorders	SHANK2	Autosomal Dominant	NM_012309.3
Autism Spectrum Disorders	SNRPN	Autosomal Dominant	NM_022807.2
Autoimmune Lymphoproliferative Syndrome	CASP10	Autosomal Dominant	NM_032977.3
Autoimmune Lymphoproliferative Syndrome	FAS	Autosomal Dominant	NM_000043.4
Autoimmune Lymphoproliferative Syndrome	FASLG	Autosomal Dominant	NM_000639.1
Autoinflammation, Lipodystrophy, and Dermatosis Syndrome	PSMB8	Autosomal Recessive	NM_148919.3
Axenfeld-Rieger Syndrome	PITX2	Autosomal Dominant	NM_153427.2
Axonal Neuropathy	GARS	Autosomal Dominant	NM_002047.2
Bardet-Biedl Syndrome	ARL6	Autosomal Recessive	NM_177976.1
Bardet-Biedl Syndrome	BBS1	Autosomal Recessive	NM_024649.4
Bardet-Biedl Syndrome	BBS10	Autosomal Recessive	NM_024685.3
Bardet-Biedl Syndrome	BBS12	Autosomal Recessive	NM_152618.2
Bardet-Biedl Syndrome	BBS2	Autosomal Recessive	NM_031885.3
Bardet-Biedl Syndrome	BBS4	Autosomal Recessive	NM_033028.4
Bardet-Biedl Syndrome	BBS7	Autosomal Recessive	NM_176824.2
Bardet-Biedl Syndrome	BBS9	Autosomal Recessive	NM_198428.2
Bardet-Biedl Syndrome	CEP290	Autosomal Recessive	NM_025114.3
Bardet-Biedl Syndrome	MKKS	Autosomal Recessive	NM_018848.3
Bardet-Biedl Syndrome	MKS1	Autosomal Recessive	NM_017777.3
Bardet-Biedl Syndrome	SDCCAG8	Autosomal Recessive	NM_006642.3
Bardet-Biedl Syndrome	TRIM32	Autosomal Recessive	NM_012210.3

Bardet-Biedl Syndrome	TTC8	Autosomal Recessive	NM_198309.2
Bardet-Biedl Syndrome	WDPCP	Autosomal Recessive	NM_015910.5
Bare Lymphocyte Syndrome, Type II	CIITA	Autosomal Recessive	NM_000246.3
Bare Lymphocyte Syndrome, Type II	RFX5	Autosomal Recessive	NM_000449.3
Bare Lymphocyte Syndrome, Type II	RFXAP	Autosomal Recessive	NM_000538.3
Bartter Syndrome	BSND	Autosomal Recessive	NM_057176.2
Basal Laminar Drusen	CFH	Autosomal Recessive	NM_000186.3
Beare-Stevenson Syndrome	FGFR2	Autosomal Dominant	NM_000141.4
Benign Chronic Pemphigus	ATP2C1	Autosomal Dominant	NM_014382.3
Benign Familial Neonatal Infantile Seizures	SCN2A	Autosomal Dominant	NM_021007.2
Benign Familial Neonatal Seizures	KCNQ3	Autosomal Dominant	NM_004519.3
Benign Hereditary Chorea	NKX2-1	Autosomal Dominant	NM_001079668.2
Benign Neonatal Epilepsy	KCNQ3	Autosomal Dominant	NM_004519.3
Berardinelli-Seip Congenital Lipodystrophy	AGPAT2	Autosomal Recessive	NM_006412.3
Berardinelli-Seip Congenital Lipodystrophy	BSCL2	Autosomal Recessive	NM_032667.6
Bernard-Soulier Syndrome	GP9	Autosomal Dominant	NM_000174.3
Best Vitelliform Macular Dystrophy	BEST1	Autosomal Dominant	NM_004183.3
Beta-Mannosidosis	MANBA	Autosomal Recessive	NM_005908.3
Beta-Sarcoglycanopathy	SGCB	Autosomal Recessive	NM_000232.4
Beta-Thalassemia	HBB	Autosomal Recessive	NM_000518.4
Beta-Ureidopropionase Deficiency	UPB1	Autosomal Recessive	NM_016327.2
BH4-Deficient Hyperphenylalaninemia	PCBD1	Autosomal Recessive	NM_000281.2
BH4-Deficient Hyperphenylalaninemia	QDPR	Autosomal Recessive	NM_000320.2
Bietti Crystalline Dystrophy	CYP4V2	Autosomal Recessive	NM_207352.3
Biotinidase Deficiency	BTD	Autosomal Recessive	NM_000060.2
Birk-Barel Mental Retardation Dysmorphism Syndrome	KCNK9	Autosomal Dominant	NM_016601.2
Birt-Hogg-Dube Syndrome	FLCN	Autosomal Dominant	NM_144997.5
Blau Syndrome	NOD2	Autosomal Dominant	NM_022162.1

Bloom Syndrome	BLM	Autosomal Recessive	NM_000057.2
BMP4-Related Syndromic Microphthalmia	BMP4	Autosomal Dominant	NM_001202.3
Bohring-Opitz Syndrome	ASX-Linked1	Autosomal Dominant	NM_015338.5
Bone Mineral Density Variation	LRP4	Autosomal Recessive	NM_002334.3
Bosley-Salih-Alorainy Syndrome	HOXA1	Autosomal Recessive	NM_005522.4
Brachydactyly	BMPR1B	Autosomal Dominant	NM_001203.2
Brachydactyly	GDF5	Autosomal Dominant	NM_000557.2
Brachydactyly	IHH	Autosomal Dominant	NM_002181.3
Brachydactyly	ROR2	Autosomal Dominant	NM_004560.3
Brachyolmia	TRPV4	Autosomal Recessive	NM_021625.4
Brain Small Vessel Disease with Hemorrhage	COL4A1	Autosomal Dominant	NM_001845.4
Branchiootorenal Spectrum Disorders	EYA1	Autosomal Dominant	NM_000503.4
Branchiootorenal Spectrum Disorders	SIX1	Autosomal Dominant	NM_005982.3
Breast and Ovarian Cancer Susceptibility	RAD51C	Autosomal Dominant	NM_058216.1
Breast and Ovarian Cancer Susceptibility	RAD51D	Autosomal Dominant	NM_002878.3
Breast Cancer	BARD1	Autosomal Recessive	NM_000465.2
Breast Cancer	BRIP1	Autosomal Recessive	NM_032043.2
Breast Cancer	CHEK2	Autosomal Recessive	NM_007194.3
Brittle Cornea Syndrome	PRDM5	Autosomal Recessive	NM_018699.2
Brittle Cornea Syndrome	ZNF469	Autosomal Recessive	NM_001127464.1
Brody Myopathy	ATP2A1	Autosomal Dominant	NM_173201.3
Brooke-Spiegler Syndrome	CYLD	Autosomal Dominant	NM_015247.2
Bruck Syndrome	PLOD2	Autosomal Recessive	NM_000935.2
Brugada Syndrome	CACNA1C	Autosomal Dominant	NM_000719.6
Brugada Syndrome	CACNB2	Autosomal Dominant	NM_201590.2
Brugada Syndrome	GPD1L	Autosomal Dominant	NM_015141.3
Brugada Syndrome	KCNE3	Autosomal Dominant	NM_005472.4
Brugada Syndrome	SCN1B	Autosomal Dominant	NM_001037.4

Brugada Syndrome	SCN3B	Autosomal Dominant	NM_018400.3
Brugada Syndrome	SCN5A	Autosomal Dominant	NM_198056.2
Budd-Chiari Syndrome	F5	Autosomal Dominant	NM_000130.4
Budd-Chiari Syndrome	JAK2	Autosomal Dominant	NM_004972.3
Buschke-Ollendorff Syndrome	LEMD3	Autosomal Dominant	NM_014319.4
Butyrylcholinesterase Deficiency	BCHE	Autosomal Recessive	NM_000055.2
C Syndrome	CD96	Autosomal Dominant	NM_198196.2
C3 Deficiency	C3	Autosomal Recessive	NM_000064.2
Caffey Disease	COL1A1	Autosomal Dominant	NM_000088.3
Calpainopathy	CAPN3	Autosomal Recessive	NM_000070.2
Campomelic Dysplasia	SOX9	Autosomal Dominant	NM_000346.3
Canavan Disease	ASPA	Autosomal Recessive	NM_000049.2
Capillary Malformation-Arteriovenous Malformation Syndrome	RASA1	Autosomal Dominant	NM_002890.2
Carbamoylphosphate Synthetase I Deficiency	CPS1	Autosomal Recessive	NM_001875.4
Cardiofaciocutaneous Syndrome	BRAF	Autosomal Dominant	NM_004333.4
Cardiofaciocutaneous Syndrome	KRAS	Autosomal Dominant	NM_004985.3
Cardiofaciocutaneous Syndrome	MAP2K1	Autosomal Dominant	NM_002755.3
Cardiomyopathy, ARVC	DSC2	Autosomal Dominant	NM_024422.3
Cardiomyopathy, ARVC	DSG2	Autosomal Dominant	NM_001943.3
Cardiomyopathy, ARVC	DSP	Autosomal Dominant	NM_004415.2
Cardiomyopathy, ARVC	JUP	Autosomal Dominant	NM_002230.2
Cardiomyopathy, ARVC	PKP2	Autosomal Dominant	NM_004572.3
Cardiomyopathy, ARVC	RYR2	Autosomal Dominant	NM_001035.2
Cardiomyopathy, ARVC	TGFB3	Autosomal Dominant	NM_003239.2
Cardiomyopathy, ARVC	TMEM43	Autosomal Dominant	NM_024334.2
Cardiomyopathy, LVNC	ACTC1	Autosomal Dominant	NM_005159.4
Cardiomyopathy, LVNC	MYH7	Autosomal Dominant	NM_000257.2
Cardiomyopathy, LVNC	TNNT2	Autosomal Dominant	NM_001001430.1

Carney Complex	PRKAR1A	Autosomal Dominant	NM_002734.3
Carnitine Palmitoyltransferase II Deficiency	CPT2	Autosomal Recessive	NM_000098.2
Carnitine-Acylcarnitine Translocase Deficiency	SLC25A20	Autosomal Recessive	NM_000387.5
Carpenter Syndrome	RAB23	Autosomal Recessive	NM_183227.1
Cartilage-Hair Hypoplasia-Anauxetic Dysplasia Spectrum Disorders	RMRP	Autosomal Recessive	NR_003051.3
Caspase 8 Deficiency	CASP8	Autosomal Recessive	NM_001228.4
Cataract-Microcornea Syndrome	GJA8	Autosomal Dominant	NM_005267.4
Cataracts	BFSP2	Autosomal Dominant	NM_003571.2
Cataracts	CRYGD	Autosomal Dominant	NM_006891.3
Cataracts	PITX2	Autosomal Dominant	NM_153427.2
Catecholaminergic Polymorphic Ventricular Tachycardia	CASQ2	Autosomal Dominant	NM_001232.3
Catecholaminergic Polymorphic Ventricular Tachycardia	RYR2	Autosomal Dominant	NM_001035.2
Catecholaminergic Polymorphic Ventricular Tachycardia	TRDN	Autosomal Dominant	NM_006073.3
Caudal Dysgenesis Syndrome	VANGL1	Autosomal Dominant	NM_138959.2
Caveolinopathies	CAV3	Autosomal Dominant	NM_033337.2
Central Core Disease	RYR1	Autosomal Dominant	NM_000540.2
Centronuclear Myopathy, Dominant	DNM2	Autosomal Dominant	NM_001005360.2
Centronuclear Myopathy, Dominant	MYF6	Autosomal Dominant	NM_002469.2
Centronuclear Myopathy, Recessive	BIN1	Autosomal Recessive	NM_139343.2
Cerebellar Ataxia	SYNE1	Autosomal Recessive	NM_033071.3
Cerebellar Ataxia, Cayman type	ATCAY	Autosomal Recessive	NM_033064.4
Cerebellar Hypoplasia	VLDLR	Autosomal Recessive	NM_003383.3
Cerebral Dysgenesis, Neuropathy, Ichthyosis, and Palmoplantar Keratoderma Syndrome	SNAP29	Autosomal Recessive	NM_004782.3
Cerebrooculofacioskeletal Syndrome	ERCC6	Autosomal Recessive	NM_000124.2
Cerebrotendinous Xanthomatosis	CYP27A1	Autosomal Recessive	NM_000784.3
Chanarin-Dorfman Syndrome	ABHD5	Autosomal Recessive	NM_016006.4
Char Syndrome	TFAP2B	Autosomal Dominant	NM_003221.3

Charcot-Marie-Tooth with Vocal Cord Paresis	GDAP1	Autosomal Recessive	NM_018972.2
Charcot-Marie-Tooth, Intermediate	DNM2	Autosomal Dominant	NM_001005360.2
Charcot-Marie-Tooth, Intermediate	GDAP1	Autosomal Dominant	NM_018972.2
Charcot-Marie-Tooth, Intermediate	KARS	Autosomal Dominant	NM_001130089.1
Charcot-Marie-Tooth, Intermediate	MPZ	Autosomal Dominant	NM_000530.6
Charcot-Marie-Tooth, Intermediate	YARS	Autosomal Dominant	NM_003680.3
Charcot-Marie-Tooth, Type 1	EGR2	Autosomal Dominant	NM_000399.3
Charcot-Marie-Tooth, Type 1	LITAF	Autosomal Dominant	NM_004862.3
Charcot-Marie-Tooth, Type 1	MPZ	Autosomal Dominant	NM_000530.6
Charcot-Marie-Tooth, Type 1	NEFL	Autosomal Dominant	NM_006158.3
Charcot-Marie-Tooth, Type 1	PMP22	Autosomal Dominant	NM_000304.2
Charcot-Marie-Tooth, Type 2	AARS	Autosomal Dominant	NM_001605.2
Charcot-Marie-Tooth, Type 2	DYNC1H1	Autosomal Dominant	NM_001376.4
Charcot-Marie-Tooth, Type 2	GARS	Autosomal Dominant	NM_002047.2
Charcot-Marie-Tooth, Type 2	HSPB1	Autosomal Dominant	NM_001540.3
Charcot-Marie-Tooth, Type 2	HSPB8	Autosomal Dominant	NM_014365.2
Charcot-Marie-Tooth, Type 2	KIF1B	Autosomal Dominant	NM_015074.3
Charcot-Marie-Tooth, Type 2	LMNA	Autosomal Dominant	NM_005572.3
Charcot-Marie-Tooth, Type 2	LRSAM1	Autosomal Dominant	NM_138361.5
Charcot-Marie-Tooth, Type 2	MED25	Autosomal Dominant	NM_030973.3
Charcot-Marie-Tooth, Type 2	MFN2	Autosomal Dominant	NM_014874.3
Charcot-Marie-Tooth, Type 2	RAB7A	Autosomal Dominant	NM_004637.5
Charcot-Marie-Tooth, Type 2	TRPV4	Autosomal Dominant	NM_021625.4
Charcot-Marie-Tooth, Type 4	FGD4	Autosomal Recessive	NM_139241.2
Charcot-Marie-Tooth, Type 4	FIG4	Autosomal Recessive	NM_014845.5
Charcot-Marie-Tooth, Type 4	MTMR2	Autosomal Recessive	NM_016156.5
Charcot-Marie-Tooth, Type 4	NDRG1	Autosomal Recessive	NM_006096.3
Charcot-Marie-Tooth, Type 4	PRX	Autosomal Recessive	NM_181882.2

Charcot-Marie-Tooth, Type 4	SBF2	Autosomal Recessive	NM_030962.3
Charcot-Marie-Tooth, Type 4	SH3TC2	Autosomal Recessive	NM_024577.3
Charcot-Marie-Tooth, X-linked	PRPS1	X-Linked	NM_002764.3
CHARGE Syndrome	CHD7	Autosomal Dominant	NM_017780.3
Chediak-Higashi Syndrome	LYST	Autosomal Recessive	NM_000081.2
Cherubism	SH3BP2	Autosomal Dominant	NM_003023.4
Chilblain Lupus	SAMHD1	Autosomal Dominant	NM_015474.3
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter	EIF2B1	Autosomal Recessive	NM_001414.3
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter	EIF2B2	Autosomal Recessive	NM_014239.3
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter	EIF2B3	Autosomal Recessive	NM_020365.4
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter	EIF2B4	Autosomal Recessive	NM_015636.3
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter	EIF2B5	Autosomal Recessive	NM_003907.2
Childhood Restrictive Cardiomyopathy	ACTA1	Autosomal Dominant	NM_001100.3
Childhood Restrictive Cardiomyopathy	ACTC1	Autosomal Dominant	NM_005159.4
Chitotriosidase Deficiency	CHIT1	Autosomal Recessive	NM_003465.2
Cholesterol Ester Storage Disease	LIPA	Autosomal Recessive	NM_000235.2
Chondrocalcinosis	ANKH	Autosomal Dominant	NM_054027.4
Chondrodysplasia	GDF5	Autosomal Recessive	NM_000557.2
Chondrodysplasia	IMPAD1	Autosomal Recessive	NM_017813.4
Chondrodysplasia	PTH1R	Autosomal Recessive	NM_000316.2
Chorea-acanthocytosis	VPS13A	Autosomal Recessive	NM_033305.2
Choreoathetosis, Hypothyroidism, and Neonatal Respiratory Distress	NKX2-1	Autosomal Dominant	NM_001079668.2
Choroidal Dystrophy	PRPH2	Autosomal Dominant	NM_000322.4

Chronic Granulomatous Disease	NCF2	Autosomal Recessive	NM_000433.3
Chronic Granulomatous Disease	NCF4	Autosomal Recessive	NM_013416.3
Chronic Infantile Neurological Cutaneous and Articular Syndrome	NLRP3	Autosomal Dominant	NM_004895.4
Citrin Deficiency	SLC25A13	Autosomal Recessive	NM_014251.2
Citrullinemia	ASS1	Autosomal Recessive	NM_000050.4
Citrullinemia	SLC25A13	Autosomal Recessive	NM_014251.2
Cleft Lip +/- Cleft Palate, Autosomal Dominant	BMP4	Autosomal Dominant	NM_001202.3
Cleft Lip +/- Cleft Palate, Autosomal Dominant	IRF6	Autosomal Dominant	NM_006147.3
Cleft Lip +/- Cleft Palate, Autosomal Dominant	SUMO1	Autosomal Dominant	NM_001005781.1
Cleft Lip +/- Cleft Palate, Autosomal Dominant	TP63	Autosomal Dominant	NM_003722.4
Cleft Lip +/- Cleft Palate, Autosomal Recessive	PVRL1	Autosomal Recessive	NM_002855.4
Cleft Palate, X-Linked	TBX22	X-Linked	NM_001109878.1
Cleidocranial Dysplasia	RUNX2	Autosomal Dominant	NM_001024630.3
Cockayne Syndrome	ERCC6	Autosomal Recessive	NM_000124.2
Cockayne Syndrome	ERCC8	Autosomal Recessive	NM_000082.3
Coenzyme Q10 deficiency, Oculomotor Apraxia Type	APTX	Autosomal Recessive	NM_175073.2
Coenzyme Q10 deficiency, Spinocerebellar Ataxia Type	ADCK3	Autosomal Recessive	NM_020247.4
Coffin-Siris Syndrome	SMARCA4	Autosomal Dominant	NM_001128849.1
Coffin-Siris Syndrome	SMARCB1	Autosomal Dominant	NM_003073.3
Cohen Syndrome	VPS13B	Autosomal Recessive	NM_017890.4
Coloboma, Congenital Heart Disease, Ichthyosiform Dermatosis, Mental Retardation, and Ear Anomalies Syndrome	PIGL	Autosomal Recessive	NM_004278.3
Colorectal Cancer	CHEK2	Autosomal Dominant	NM_007194.3
Combined Deficiency of Factor V and Factor VIII	LMAN1	Autosomal Recessive	NM_005570.3
Combined Deficiency of Factor V and Factor VIII	MCFD2	Autosomal Recessive	NM_139279.5
Combined Oxidative Phosphorylation Deficiency	AARS2	Autosomal Recessive	NM_020745.3
Combined Oxidative Phosphorylation Deficiency	EARS2	Autosomal Recessive	NM_001083614.1
Combined Oxidative Phosphorylation Deficiency	GFM1	Autosomal Recessive	NM_024996.5

Combined Oxidative Phosphorylation Deficiency	MRPS16	Autosomal Recessive	NM_016065.3
Combined Oxidative Phosphorylation Deficiency	MRPS22	Autosomal Recessive	NM_020191.2
Combined Oxidative Phosphorylation Deficiency	TSFM	Autosomal Recessive	NM_001172696.1
Combined Oxidative Phosphorylation Deficiency	TUFM	Autosomal Recessive	NM_003321.4
Combined Pituitary Hormone Deficiency, Dominant	LHX4	Autosomal Dominant	NM_033343.3
Combined Pituitary Hormone Deficiency, Dominant	OTX2	Autosomal Dominant	NM_172337.2
Combined Pituitary Hormone Deficiency, Dominant/Recessive	HESX1	Mixed	NM_003865.2
Combined Pituitary Hormone Deficiency, Recessive	LHX3	Autosomal Recessive	NM_014564.3
Combined Pituitary Hormone Deficiency, Recessive	POU1F1	Autosomal Recessive	NM_000306.2
Combined Pituitary Hormone Deficiency, Recessive	PROP1	Autosomal Recessive	NM_006261.4
Combined Saposin Deficiency	PSAP	Autosomal Recessive	NM_002778.2
Common Variable Immune Deficiency, Dominant	TNFRSF13B	Autosomal Dominant	NM_012452.2
Common Variable Immune Deficiency, Recessive	CD19	Autosomal Recessive	NM_001770.5
Common Variable Immune Deficiency, Recessive	ICOS	Autosomal Recessive	NM_012092.3
Common Variable Immune Deficiency, Recessive	TNFRSF13C	Autosomal Recessive	NM_052945.3
Complement Component C2 Deficiency	C2	Autosomal Recessive	NM_000063.4
Cone Dystrophy	GUCA1A	Autosomal Dominant	NM_000409.3
Cone-Rod Dystrophy, Dominant	CRX	Autosomal Dominant	NM_000554.4
Cone-Rod Dystrophy, Dominant	PITPNM3	Autosomal Dominant	NM_031220.3
Cone-Rod Dystrophy, Dominant	PROM1	Autosomal Dominant	NM_006017.2
Cone-Rod Dystrophy, Dominant	PRPH2	Autosomal Dominant	NM_000322.4
Cone-Rod Dystrophy, Dominant	RAX2	Autosomal Dominant	NM_032753.3
Cone-Rod Dystrophy, Dominant	RIMS1	Autosomal Dominant	NM_014989.5
Cone-Rod Dystrophy, Dominant	UNC119	Autosomal Dominant	NM_005148.3
Cone-Rod Dystrophy, Recessive	ABCA4	Autosomal Recessive	NM_000350.2
Cone-Rod Dystrophy, Recessive	ADAM9	Autosomal Recessive	NM_003816.2
Cone-Rod Dystrophy, Recessive	C8orf37	Autosomal Recessive	NM_177965.3
Cone-Rod Dystrophy, Recessive	CDHR1	Autosomal Recessive	NM_033100.2

Cone-Rod Dystrophy, Recessive	PDE6C	Autosomal Recessive	NM_006204.3
Cone-Rod Dystrophy, Recessive	RPGRIP1	Autosomal Recessive	NM_020366.3
Cone-Rod Dystrophy, Recessive	SEMA4A	Autosomal Recessive	NM_022367.3
Congenital Adrenal Hyperplasia	CYP11B1	Autosomal Recessive	NM_000497.3
Congenital Adrenal Hyperplasia	CYP17A1	Autosomal Recessive	NM_000102.3
Congenital Adrenal Hyperplasia	HSD3B2	Autosomal Recessive	NM_000198.3
Congenital Adrenal Hyperplasia	STAR	Autosomal Recessive	NM_000349.2
Congenital Adrenal Insufficiency	CYP11A1	Autosomal Dominant	NM_000781.2
Congenital Afibrinogenemia	FGA	Autosomal Recessive	NM_021871.2
Congenital Afibrinogenemia	FGB	Autosomal Recessive	NM_005141.4
Congenital Afibrinogenemia	FGG	Autosomal Recessive	NM_000509.4
Congenital Amegakaryocytic Thrombocytopenia	MPL	Autosomal Recessive	NM_005373.2
Congenital Aural Atresia	TSHZ1	Autosomal Dominant	NM_005786.5
Congenital Bile Acid Synthesis Defect	AKR1D1	Autosomal Recessive	NM_005989.3
Congenital Bile Acid Synthesis Defect	CYP7B1	Autosomal Recessive	NM_004820.3
Congenital Cataract	AGK	Autosomal Recessive	NM_018238.3
Congenital Cataract	CRYAA	Autosomal Recessive	NM_000394.2
Congenital Cataract	FYCO1	Autosomal Recessive	NM_024513.3
Congenital Cataract	TDRD7	Autosomal Recessive	NM_014290.2
Congenital Central Hypoventilation Syndrome	PHOX2B	Autosomal Dominant	NM_003924.3
Congenital Contractural Arachnodactyly	FBN2	Autosomal Dominant	NM_001999.3
Congenital Disorders of Glycosylation	ALG11	Autosomal Recessive	NM_001004127.2
Congenital Disorders of Glycosylation	ALG12	Autosomal Recessive	NM_024105.3
Congenital Disorders of Glycosylation	ALG2	Autosomal Recessive	NM_033087.3
Congenital Disorders of Glycosylation	ALG3	Autosomal Recessive	NM_005787.5
Congenital Disorders of Glycosylation	ALG6	Autosomal Recessive	NM_013339.3
Congenital Disorders of Glycosylation	ALG8	Autosomal Recessive	NM_024079.4
Congenital Disorders of Glycosylation	ALG9	Autosomal Recessive	NM_024740.2

Congenital Disorders of Glycosylation	B4GALT1	Autosomal Recessive	NM_001497.3
Congenital Disorders of Glycosylation	COG1	Autosomal Recessive	NM_018714.2
Congenital Disorders of Glycosylation	COG4	Autosomal Recessive	NM_015386.2
Congenital Disorders of Glycosylation	COG5	Autosomal Recessive	NM_006348.3
Congenital Disorders of Glycosylation	COG6	Autosomal Recessive	NM_020751.2
Congenital Disorders of Glycosylation	COG7	Autosomal Recessive	NM_153603.3
Congenital Disorders of Glycosylation	COG8	Autosomal Recessive	NM_032382.4
Congenital Disorders of Glycosylation	DDOST	Autosomal Recessive	NM_005216.4
Congenital Disorders of Glycosylation	DOLK	Autosomal Recessive	NM_014908.3
Congenital Disorders of Glycosylation	DPAGT1	Autosomal Recessive	NM_001382.3
Congenital Disorders of Glycosylation	DPM1	Autosomal Recessive	NM_003859.1
Congenital Disorders of Glycosylation	DPM3	Autosomal Recessive	NM_153741.1
Congenital Disorders of Glycosylation	MGAT2	Autosomal Recessive	NM_002408.3
Congenital Disorders of Glycosylation	MPDU1	Autosomal Recessive	NM_004870.3
Congenital Disorders of Glycosylation	MPI	Autosomal Recessive	NM_002435.1
Congenital Disorders of Glycosylation	PGM1	Autosomal Recessive	NM_002633.2
Congenital Disorders of Glycosylation	PMM2	Autosomal Recessive	NM_000303.2
Congenital Disorders of Glycosylation	RFT1	Autosomal Recessive	NM_052859.3
Congenital Disorders of Glycosylation	SLC35C1	Autosomal Recessive	NM_018389.4
Congenital Disorders of Glycosylation	SRD5A3	Autosomal Recessive	NM_024592.4
Congenital Disorders of Glycosylation	TUSC3	Autosomal Recessive	NM_006765.3
Congenital Dyserythropoietic Anemia	CDAN1	Autosomal Dominant	NM_138477.2
Congenital Dyserythropoietic Anemia	KLF1	Autosomal Dominant	NM_006563.3
Congenital Dyserythropoietic Anemia	SEC23B	Autosomal Dominant	NM_006363.4
Congenital Erythropoietic Porphyria	UROS	Autosomal Recessive	NM_000375.2
Congenital Fiber-Type Disproportion	ACTA1	Autosomal Dominant	NM_001100.3
Congenital Fiber-Type Disproportion	TPM3	Autosomal Dominant	NM_152263.2
Congenital Fibrosis of the Extraocular Muscles	KIF21A	Autosomal Dominant	NM_017641.3

Congenital Finnish Nephrosis	NPHS1	Autosomal Recessive	NM_004646.3
Congenital Glutamine Deficiency	GLUL	Autosomal Recessive	NM_002065.5
Congenital Hypomyelinating Neuropathy	MPZ	Autosomal Dominant	NM_000530.6
Congenital Hypomyelination	MPZ	Autosomal Dominant	NM_000530.6
Congenital Hypothyroidism	DUOX2	Autosomal Recessive	NM_014080.4
Congenital Hypothyroidism	IYD	Autosomal Recessive	NM_203395.2
Congenital Hypothyroidism	PAX8	Autosomal Recessive	NM_003466.3
Congenital Hypothyroidism	TPO	Autosomal Recessive	NM_000547.5
Congenital Hypothyroidism	TSHB	Autosomal Recessive	NM_000549.3
Congenital Hypothyroidism	TSHR	Autosomal Recessive	NM_000369.2
Congenital Ichthyosis	ABCA12	Autosomal Recessive	NM_173076.2
Congenital Ichthyosis	ALOX12B	Autosomal Recessive	NM_001139.2
Congenital Ichthyosis	ALOXE3	Autosomal Recessive	NM_021628.2
Congenital Ichthyosis	CYP4F22	Autosomal Recessive	NM_173483.3
Congenital Ichthyosis	NIPAL4	Autosomal Recessive	NM_001099287.1
Congenital Ichthyosis	PNPLA1	Autosomal Recessive	NM_001145717.1
Congenital Ichthyosis	TGM1	Autosomal Recessive	NM_000359.2
Congenital Indifference to Pain	SCN9A	Autosomal Recessive	NM_002977.3
Congenital Insensitivity to Pain with Anhidrosis	NTRK1	Autosomal Recessive	NM_001012331.1
Congenital Lactase Deficiency	LCT	Autosomal Recessive	NM_002299.2
Congenital Muscular Dystrophy, alpha-dystroglycan related	FKTN	Autosomal Recessive	NM_001079802.1
Congenital Muscular Dystrophy, alpha-dystroglycan related	ISPD	Autosomal Recessive	NM_001101426.3
Congenital Muscular Dystrophy, alpha-dystroglycan related	LARGE	Autosomal Recessive	NM_004737.4
Congenital Muscular Dystrophy, alpha-dystroglycan related	POMGNT1	Autosomal Recessive	NM_017739.3
Congenital Muscular Dystrophy, alpha-dystroglycan related	POMT1	Autosomal Recessive	NM_007171.3
Congenital Muscular Dystrophy, alpha-dystroglycan related	POMT2	Autosomal Recessive	NM_013382.5
Congenital Muscular Dystrophy, CKHB-related	CHKB	Autosomal Recessive	NM_005198.4
Congenital Muscular Dystrophy, collagen-related	COL6A1	Mixed	NM_001848.2

Congenital Muscular Dystrophy, collagen-related	COL6A2	Mixed	NM_001849.3
Congenital Muscular Dystrophy, collagen-related	COL6A3	Mixed	NM_004369.3
Congenital Muscular Dystrophy, ITGA7-related	ITGA7	Autosomal Recessive	NM_002206.2
Congenital Muscular Dystrophy, LAMA2-related	LAMA2	Autosomal Recessive	NM_000426.3
Congenital Muscular Dystrophy, LMNA-related	LMNA	Autosomal Dominant	NM_005572.3
Congenital Myasthenic Syndrome, Dominant/Recessive	CHRNA1	Mixed	NM_000079.3
Congenital Myasthenic Syndrome, Dominant/Recessive	CHRNB1	Mixed	NM_000747.2
Congenital Myasthenic Syndrome, Dominant/Recessive	CHRND	Mixed	NM_000751.2
Congenital Myasthenic Syndrome, Dominant/Recessive	CHRNE	Mixed	NM_000080.3
Congenital Myasthenic Syndrome, Recessive	COLQ	Autosomal Recessive	NM_005677.3
Congenital Myasthenic Syndrome, Recessive	GFPT1	Autosomal Recessive	NM_002056.3
Congenital Myasthenic Syndrome, Recessive	MUSK	Autosomal Recessive	NM_005592.3
Congenital Myasthenic Syndrome, Recessive	RAPSN	Autosomal Recessive	NM_005055.4
Congenital Myasthenic Syndrome, Recessive	SCN4A	Autosomal Recessive	NM_000334.4
Congenital Neuromuscular Disease with Uniform Type 1 Fiber	RYR1	Autosomal Dominant	NM_000540.2
Congenital Nuclear Cataract	CRYBB1	Autosomal Recessive	NM_001887.3
Congenital Nuclear Cataract	CRYBB3	Autosomal Recessive	NM_004076.3
Congenital Stationary Night Blindness, Dominant	GNAT1	Autosomal Dominant	NM_144499.2
Congenital Stationary Night Blindness, Dominant	PDE6B	Autosomal Dominant	NM_000283.3
Congenital Stationary Night Blindness, Dominant	RHO	Autosomal Dominant	NM_000539.3
Congenital Stationary Night Blindness, Recessive	CABP4	Autosomal Recessive	NM_145200.3
Congenital Stationary Night Blindness, Recessive	GPR179	Autosomal Recessive	NM_001004334.2
Congenital Stationary Night Blindness, Recessive	SLC24A1	Autosomal Recessive	NM_004727.2
Congenital Stationary Night Blindness, Recessive	TRPM1	Autosomal Recessive	NM_002420.5
Congenital Stromal Corneal Dystrophy	DCN	Autosomal Dominant	NM_001920.3
Congenital Sucrase-Isomaltase Deficiency	SI	Autosomal Recessive	NM_001041.3
Congenital Vertical Talus	HOXD10	Autosomal Dominant	NM_002148.3
Corneal Dystrophy, Dominant	TGFBI	Autosomal Dominant	NM_000358.2

Corneal Dystrophy, Dominant/Recessive	TACSTD2	Autosomal Recessive	NM_002353.2
Corneal Dystrophy, Recessive	CYP4V2	Autosomal Recessive	NM_207352.3
Corneal Dystrophy, Recessive	SLC4A11	Autosomal Recessive	NM_032034.3
Corneal Fleck Dystrophy	PIKFYVE	Autosomal Dominant	NM_015040.3
Cornelia de Lange Syndrome	NIPBL	Autosomal Dominant	NM_133433.3
Cornelia de Lange Syndrome	SMC1A	Autosomal Dominant	NM_006306.2
Cornelia de Lange Syndrome	SMC3	Autosomal Dominant	NM_005445.3
Cortical Dysplasia-Focal Epilepsy Syndrome	CNTNAP2	Autosomal Recessive	NM_014141.5
Cortical Pulverulent Cataract	LIM2	Autosomal Recessive	NM_030657.3
Corticosterone Methyloxidase Type I Deficiency	CYP11B2	Autosomal Recessive	NM_000498.3
Corticosterone Methyloxidase Type II Deficiency	CYP11B2	Autosomal Recessive	NM_000498.3
Cranioectodermal Dysplasia	IFT122	Autosomal Recessive	NM_052985.2
Cranioectodermal Dysplasia	IFT43	Autosomal Recessive	NM_052873.2
Cranioectodermal Dysplasia	WDR19	Autosomal Recessive	NM_025132.3
Cranioectodermal Dysplasia	WDR35	Autosomal Recessive	NM_001006657.1
Craniofacial-Deafness-Hand Syndrome	PAX3	Autosomal Dominant	NM_181457.3
Craniometaphyseal Dysplasia	ANKH	Autosomal Dominant	NM_054027.4
Craniosynostosis	FGFR1	Autosomal Dominant	NM_023110.2
Craniosynostosis	FGFR2	Autosomal Dominant	NM_000141.4
Craniosynostosis	MSX2	Autosomal Dominant	NM_002449.4
Creatine Deficiency Syndromes	GAMT	Autosomal Recessive	NM_000156.4
Creatine Deficiency Syndromes	GATM	Autosomal Recessive	NM_001482.2
Crigler-Najjar Syndrome	UGT1A1	Autosomal Recessive	NM_000463.2
Crohn Disease	NOD2	Autosomal Dominant	NM_022162.1
Crouzon Syndrome	FGFR2	Autosomal Dominant	NM_000141.4
Cutaneous Malignant Melanoma, Dominant	CDK4	Autosomal Dominant	NM_000075.2
Cutaneous Malignant Melanoma, Recessive	MC1R	Autosomal Recessive	NM_002386.3
Cutis Laxa with Severe Pulmonary, Gastrointestinal, and Urinary	LTBP4	Autosomal Recessive	NM_003573.2

Abnormalities			
Cutis Laxa, Dominant	ELN	Autosomal Dominant	NM_000501.2
Cutis Laxa, Dominant/Recessive	FBLN5	Mixed	NM_006329.3
Cutis Laxa, Recessive	ALDH18A1	Autosomal Recessive	NM_002860.3
Cutis Laxa, Recessive	ATP6V0A2	Autosomal Recessive	NM_012463.3
Cutis Laxa, Recessive	PYCR1	Autosomal Recessive	NM_006907.2
Cystathioninuria	CTH	Autosomal Recessive	NM_001902.5
Cystic Fibrosis	CFTR	Autosomal Recessive	NM_000492.3
Cystic Fibrosis-Like Syndrome	SCNN1A	Autosomal Dominant	NM_001038.5
Cystinosis	CTNS	Autosomal Recessive	NM_004937.2
Cystinuria	SLC3A1	Autosomal Recessive	NM_000341.3
Cystinuria	SLC7A9	Autosomal Recessive	NM_014270.4
Cytochrome P450 Oxidoreductase Deficiency	POR	Autosomal Recessive	NM_000941.2
D-2-Hydroxyglutaric Aciduria	D2HGDH	Autosomal Dominant	NM_152783.3
Danon Disease	LAMP2	X-Linked	NM_002294.2
Darier-White Disease	ATP2A2	Autosomal Dominant	NM_001681.3
Delta-Sarcoglycanopathy	SGCD	Autosomal Recessive	NM_000337.5
Dementia, Deafness, and Sensory Neuropathy	DNMT1	Autosomal Dominant	NM_001130823.1
Dense Deposit Disease / Membranoproliferative Glomerulonephritis Type II	CFH	Autosomal Recessive	NM_000186.3
Dense Deposit Disease / Membranoproliferative Glomerulonephritis Type II	CFHR5	Autosomal Recessive	NM_030787.3
Dent Disease	CLCN5	X-Linked	NM_000084.2
Desbuquois Dysplasia	CANT1	Autosomal Recessive	NM_138793.3
Desminopathy	DES	Autosomal Dominant	NM_001927.3
Desmosterolosis	DHCR24	Autosomal Recessive	NM_014762.3
Diabetes Mellitus, Insulin-Resistant, with Acanthosis Nigricans	INSR	Autosomal Dominant	NM_000208.2
Diabetes Mellitus, Neonatal, with Congenital Hypothyroidism	GLIS3	Autosomal Recessive	NM_001042413.1
Diabetes Mellitus, Noninsulin-Dependent, with Acanthosis Nigricans and Hypertension	PPARG	Autosomal Dominant	NM_015869.4

Diabetes, Transient Neonatal	ZFP57	Autosomal Recessive	NM_001109809.2
Diamond-Blackfan Anemia	RPL11	Autosomal Dominant	NM_000975.3
Diamond-Blackfan Anemia	RPL35A	Autosomal Dominant	NM_000996.2
Diamond-Blackfan Anemia	RPL5	Autosomal Dominant	NM_000969.3
Diamond-Blackfan Anemia	RPS10	Autosomal Dominant	NM_001014.4
Diamond-Blackfan Anemia	RPS19	Autosomal Dominant	NM_001022.3
Diamond-Blackfan Anemia	RPS24	Autosomal Dominant	NM_033022.3
Diamond-Blackfan Anemia	RPS26	Autosomal Dominant	NM_001029.3
Diamond-Blackfan Anemia	RPS7	Autosomal Dominant	NM_001011.3
Diaphanospondylodysostosis	BMPER	Autosomal Recessive	NM_133468.4
Diaphyseal Medullary Stenosis with Malignant Fibrous Histiocytoma	MTAP	Autosomal Dominant	NM_002451.3
Diarrhea with Microvillus Atrophy	MYO5B	Autosomal Recessive	NM_001080467.2
Diastrophic Dysplasia	SLC26A2	Autosomal Recessive	NM_000112.3
Dicarboxylicaminoaciduria	SLC1A1	Autosomal Recessive	NM_004170.5
Diffuse Mesangial Sclerosis Syndromes (DMS)	WT1	Autosomal Dominant	NM_024426.4
Dihydropyrimidinase Deficiency	DPYS	Autosomal Recessive	NM_001385.2
Dihydropyrimidine Dehydrogenase Deficiency	DPYD	Autosomal Recessive	NM_000110.3
Dilated Cardiomyopathy with Quadriceps Myopathy	LMNA	Autosomal Dominant	NM_005572.3
Dilated Cardiomyopathy, Dominant	ABCC9	Autosomal Dominant	NM_005691.2
Dilated Cardiomyopathy, Dominant	ACTC1	Autosomal Dominant	NM_005159.4
Dilated Cardiomyopathy, Dominant	ACTN2	Autosomal Dominant	NM_001103.2
Dilated Cardiomyopathy, Dominant	ANKRD1	Autosomal Dominant	NM_014391.2
Dilated Cardiomyopathy, Dominant	BAG3	Autosomal Dominant	NM_004281.3
Dilated Cardiomyopathy, Dominant	CSRP3	Autosomal Dominant	NM_003476.4
Dilated Cardiomyopathy, Dominant	CTF1	Autosomal Dominant	NM_001330.3
Dilated Cardiomyopathy, Dominant	DES	Autosomal Dominant	NM_001927.3
Dilated Cardiomyopathy, Dominant	DSG2	Autosomal Dominant	NM_001943.3
Dilated Cardiomyopathy, Dominant	EYA4	Autosomal Dominant	NM_004100.4

Dilated Cardiomyopathy, Dominant	LDB3	Autosomal Dominant	NM_001080116.1
Dilated Cardiomyopathy, Dominant	LMNA	Autosomal Dominant	NM_005572.3
Dilated Cardiomyopathy, Dominant	MYBPC3	Autosomal Dominant	NM_000256.3
Dilated Cardiomyopathy, Dominant	MYH6	Autosomal Dominant	NM_002471.3
Dilated Cardiomyopathy, Dominant	MYH7	Autosomal Dominant	NM_000257.2
Dilated Cardiomyopathy, Dominant	NEXN	Autosomal Dominant	NM_144573.3
Dilated Cardiomyopathy, Dominant	PLN	Autosomal Dominant	NM_002667.3
Dilated Cardiomyopathy, Dominant	PSEN1	Autosomal Dominant	NM_000021.3
Dilated Cardiomyopathy, Dominant	PSEN2	Autosomal Dominant	NM_000447.2
Dilated Cardiomyopathy, Dominant	RBM20	Autosomal Dominant	NM_001134363.1
Dilated Cardiomyopathy, Dominant	SCN5A	Autosomal Dominant	NM_198056.2
Dilated Cardiomyopathy, Dominant	SGCD	Autosomal Dominant	NM_000337.5
Dilated Cardiomyopathy, Dominant	TCAP	Autosomal Dominant	NM_003673.3
Dilated Cardiomyopathy, Dominant	TMPO	Autosomal Dominant	NM_003276.2
Dilated Cardiomyopathy, Dominant	TNNC1	Autosomal Dominant	NM_003280.2
Dilated Cardiomyopathy, Dominant	TNNT2	Autosomal Dominant	NM_001001430.1
Dilated Cardiomyopathy, Dominant	TPM1	Autosomal Dominant	NM_001018005.1
Dilated Cardiomyopathy, Dominant	TTN	Autosomal Dominant	NM_133378.4
Dilated Cardiomyopathy, Dominant	VCL	Autosomal Dominant	NM_014000.2
Dilated Cardiomyopathy, Recessive	FKTN	Autosomal Recessive	NM_001079802.1
Dilated Cardiomyopathy, Recessive	TNNI3	Autosomal Recessive	NM_000363.4
Dilated Cardiomyopathy, X-Linked	DMD	X-Linked	NM_004006.2
Dilated Cardiomyopathy, X-Linked	TAZ	X-Linked	NM_000116.3
Disorders of Intracellular Cobalamin Metabolism	LMBRD1	Autosomal Recessive	NM_018368.3
Disorders of Intracellular Cobalamin Metabolism	MMACHC	Autosomal Recessive	NM_015506.2
Disorders of Intracellular Cobalamin Metabolism	MMADHC	Autosomal Recessive	NM_015702.2
Disorders of Intracellular Cobalamin Metabolism	MTR	Autosomal Recessive	NM_000254.2
Disorders of Intracellular Cobalamin Metabolism	MTRR	Autosomal Recessive	NM_002454.2

Distal Arthrogryposis	MYBPC1	Autosomal Dominant	NM_002465.3
Distal Arthrogryposis	MYH3	Autosomal Dominant	NM_002470.3
Distal Arthrogryposis Multiplex Congenita	TNNI2	Autosomal Dominant	NM_003282.3
Distal Arthrogryposis Multiplex Congenita	TNNT3	Autosomal Dominant	NM_006757.3
Distal Congenital Nonprogressive Spinal Muscular Atrophy	TRPV4	Autosomal Dominant	NM_021625.4
Distal Hereditary Motor Neuronopathy	DCTN1	Autosomal Dominant	NM_004082.4
Distal Hereditary Motor Neuronopathy	HSPB1	Autosomal Dominant	NM_001540.3
Distal Hereditary Motor Neuronopathy	HSPB3	Autosomal Dominant	NM_006308.2
Distal Hereditary Motor Neuronopathy	HSPB8	Autosomal Dominant	NM_014365.2
Distal Myopathy	MATR3	Autosomal Dominant	NM_199189.2
Distal Renal Tubular Acidosis with Progressive Nerve Deafness	ATP6V1B1	Autosomal Recessive	NM_001692.3
Distal Renal Tubular Acidosis, Dominant	SLC4A1	Autosomal Dominant	NM_000342.3
Distal Renal Tubular Acidosis, Recessive	ATP6V0A4	Autosomal Recessive	NM_020632.2
Distal Spinal Muscular Atrophy	GARS	Autosomal Dominant	NM_002047.2
Distal Spinal Muscular Atrophy	PLEKHG5	Autosomal Dominant	NM_020631.4
Donnai-Barrow Syndrome	LRP2	Autosomal Recessive	NM_004525.2
Donohue Syndrome	INSR	Autosomal Recessive	NM_000208.2
Dopamine Beta-Hydroxylase Deficiency	DBH	Autosomal Recessive	NM_000787.3
Dopa-Responsive Dystonia	GCH1	Autosomal Dominant	NM_000161.2
Dopa-Responsive Dystonia	SPR	Autosomal Dominant	NM_003124.4
Doyne Honeycomb Retinal Dystrophy	EFEMP1	Autosomal Dominant	NM_001039348.2
Dravet Syndrome	GABRG2	Autosomal Dominant	NM_000816.3
Dravet Syndrome	SCN9A	Autosomal Dominant	NM_002977.3
DRPLA	ATN1	Autosomal Dominant	NM_001007026.1
Duane Syndrome	CHN1	Autosomal Dominant	NM_001822.5
Dubin-Johnson Syndrome	ABCC2	Autosomal Recessive	NM_000392.3
Dyggve-Melchior-Clausen Syndrome	DYM	Autosomal Recessive	NM_017653.3
Dysalbuminemic Hyperthyroxinemia	ALB	Autosomal Dominant	NM_000477.5

Dyschromatosis Symmetrica Hereditaria	ADAR	Autosomal Dominant	NM_001111.4
Dyskeratosis Congenita, Dominant	TINF2	Autosomal Dominant	NM_001099274.1
Dyskeratosis Congenita, Recessive	CTC1	Autosomal Recessive	NM_025099.5
Dyskeratosis Congenita, Recessive	NHP2	Autosomal Recessive	NM_017838.3
Dyskeratosis Congenita, Recessive	NOP10	Autosomal Recessive	NM_018648.3
Dyskeratosis Congenita, Recessive	WRAP53	Autosomal Recessive	NM_018081.2
Dyssegmental Dysplasia	HSPG2	Autosomal Recessive	NM_005529.5
Dystonia	PRKRA	Mixed	NM_003690.4
Dystonia	SLC2A1	Mixed	NM_006516.2
Dystonia	THAP1	Mixed	NM_018105.2
Dystonia/Parkinsonism, Hypermanganesemia, Polycythemia, and Chronic Liver Disease	SLC30A10	Autosomal Recessive	NM_018713.2
Dystrophic Epidermolysis Bullosa	COL7A1	Autosomal Dominant	NM_000094.3
Early Infantile Epileptic Encephalopathy, Autosomal Dominant	SCN2A	Autosomal Dominant	NM_021007.2
Early Infantile Epileptic Encephalopathy, Autosomal Dominant	SCN8A	Autosomal Dominant	NM_014191.3
Early Infantile Epileptic Encephalopathy, Autosomal Dominant	SPTAN1	Autosomal Dominant	NM_001130438.2
Early Infantile Epileptic Encephalopathy, Autosomal Dominant	STXBP1	Autosomal Dominant	NM_003165.3
Early Infantile Epileptic Encephalopathy, Autosomal Recessive	PLCB1	Autosomal Recessive	NM_015192.2
Early Infantile Epileptic Encephalopathy, Autosomal Recessive	SLC25A22	Autosomal Recessive	NM_024698.5
Early-Onset Primary Dystonia	TOR1A	Autosomal Dominant	NM_000113.2
Ectodermal Dysplasia, Anhidrotic, with T-cell Immunodeficiency	NFKBIA	Autosomal Dominant	NM_020529.2
Ectodermal Dysplasia/Skin Fragility Syndrome	DSP	Autosomal Recessive	NM_004415.2
Ectodermal Dysplasia/Skin Fragility Syndrome	PKP1	Autosomal Recessive	NM_001005337.2
Ectopia Lentis	ADAMTSL4	Autosomal Recessive	NM_019032.4
Ectopia Lentis	FBN1	Autosomal Recessive	NM_000138.4
Ectrodactyly	TP63	Autosomal Dominant	NM_003722.4
EEM Syndrome	CDH3	Autosomal Recessive	NM_001793.4
Ehlers-Danlos Syndrome, Arthrochalasia Type	ADAMTS2	Autosomal Dominant	NM_014244.4

Ehlers-Danlos Syndrome, Arthrochalasia Type	COL1A2	Autosomal Dominant	NM_000089.3
Ehlers-Danlos Syndrome, Classic	COL1A1	Autosomal Dominant	NM_000088.3
Ehlers-Danlos Syndrome, Classic	COL5A1	Autosomal Dominant	NM_000093.3
Ehlers-Danlos Syndrome, Classic	COL5A2	Autosomal Dominant	NM_000393.3
Ehlers-Danlos Syndrome, Kyphoscoliotic Form	PLOD1	Autosomal Recessive	NM_000302.3
Ehlers-Danlos Syndrome, Vascular Type	COL3A1	Autosomal Dominant	NM_000090.3
Elliptocytosis	SPTA1	Autosomal Dominant	NM_003126.2
Elliptocytosis	SPTB	Autosomal Dominant	NM_000347.5
Ellis-van Creveld Syndrome	EVC	Autosomal Recessive	NM_153717.2
Ellis-van Creveld Syndrome	EVC2	Autosomal Recessive	NM_147127.4
Emery-Dreifuss Muscular Dystrophy	LMNA	Autosomal Dominant	NM_005572.3
Emery-Dreifuss Muscular Dystrophy	SYNE1	Autosomal Dominant	NM_033071.3
Emery-Dreifuss Muscular Dystrophy	SYNE2	Autosomal Dominant	NM_182914.2
Endocardial Fibroelastosis	TAZ	Autosomal Dominant	NM_000116.3
Enhanced S-Cone Syndrome	NR2E3	Autosomal Recessive	NM_014249.2
Enlarged Parietal Foramina	ALX4	Autosomal Dominant	NM_021926.3
Enlarged Parietal Foramina	MSX2	Autosomal Dominant	NM_002449.4
Epidermolysis Bullosa Simplex	KRT5	Autosomal Dominant	NM_000424.3
Epidermolysis Bullosa with Pyloric Atresia	ITGA6	Autosomal Recessive	NM_000210.2
Epidermolysis Bullosa with Pyloric Atresia	ITGB4	Autosomal Recessive	NM_001005731.1
Epidermolysis Bullosa, Lethal Acantholytic	DSP	Autosomal Recessive	NM_004415.2
Epidermolytic Hyperkeratosis	KRT1	Autosomal Dominant	NM_006121.3
Epidermolytic Palmoplantar Keratoderma	KRT9	Autosomal Dominant	NM_000226.3
Epilepsy with Neurodevelopmental Defects	GRIN2A	Autosomal Dominant	NM_000833.3
Epileptic Encephalopathy	MAPK10	Autosomal Dominant	NM_138982.2
Epimerase Deficiency Galactosemia	GALE	Autosomal Recessive	NM_000403.3
Episodic Ataxia	CACNB4	Autosomal Dominant	NM_000726.3
Episodic Ataxia	KCNA1	Autosomal Dominant	NM_000217.2

Episodic Ataxia	SLC1A3	Autosomal Dominant	NM_004172.4
Epstein Syndrome	MYH9	Autosomal Dominant	NM_002473.4
Erosive Vitreoretinopathy	VCAN	Autosomal Dominant	NM_004385.4
Erythrocyte AMP Deaminase Deficiency	AMPD3	Autosomal Recessive	NM_001025389.1
Erythrokeratodermia Variabilis	GJB3	Mixed	NM_024009.2
Erythropoietic Protoporphyria	FECH	Autosomal Dominant	NM_000140.3
Escobar Syndrome	CHRNG	Autosomal Recessive	NM_005199.4
Essential Fructosuria	KHK	Autosomal Recessive	NM_000221.2
Essential Thrombocythemia	MPL	Autosomal Dominant	NM_005373.2
Essential Thrombocythemia	THPO	Autosomal Dominant	NM_000460.2
Ethylmalonic Encephalopathy	ETHE1	Autosomal Recessive	NM_014297.3
Exocrine Pancreatic Insufficiency, Dyserythropoietic Anemia, and Calvarial Hyperostosis	COX4I2	Autosomal Recessive	NM_032609.2
Fabry Disease	GLA	X-Linked	NM_000169.2
Factor V Cambridge Thrombophilia	F5	Autosomal Dominant	NM_000130.4
Factor V Deficiency	F5	Autosomal Recessive	NM_000130.4
Factor V Leiden Thrombophilia	F5	Autosomal Dominant	NM_000130.4
Factor V R2 Mutation Thrombophilia	F5	Autosomal Dominant	NM_000130.4
Factor VII Deficiency	F7	Autosomal Recessive	NM_000131.3
Factor VII Marburg I Variant Thrombophilia	HABP2	Autosomal Dominant	NM_004132.3
Factor X Deficiency	F10	Autosomal Recessive	NM_000504.3
Factor XI Deficiency	F11	Autosomal Dominant	NM_000128.3
Factor XII Deficiency	F12	Autosomal Dominant	NM_000505.3
Factor XIII Subunit A Deficiency	F13A1	Autosomal Recessive	NM_000129.3
Factor XIII Subunit B Deficiency	F13B	Autosomal Recessive	NM_001994.2
Familial Atrial Fibrillation	ABCC9	Autosomal Dominant	NM_005691.2
Familial Atrial Fibrillation	GJA5	Autosomal Dominant	NM_005266.5
Familial Atrial Fibrillation	KCNA5	Autosomal Dominant	NM_002234.3

Familial Atrial Fibrillation	KCNE2	Autosomal Dominant	NM_172201.1
Familial Atrial Fibrillation	KCNJ2	Autosomal Dominant	NM_000891.2
Familial Atrial Fibrillation	KCNQ1	Autosomal Dominant	NM_000218.2
Familial Atypical Mycobacteriosis, Autosomal Dominant	STAT1	Autosomal Dominant	NM_007315.3
Familial Atypical Mycobacteriosis, Autosomal Recessive	IFNGR1	Autosomal Recessive	NM_000416.2
Familial Atypical Mycobacteriosis, Autosomal Recessive	IL12B	Autosomal Recessive	NM_002187.2
Familial Atypical Mycobacteriosis, Autosomal Recessive	IL12RB1	Autosomal Recessive	NM_005535.1
Familial Atypical Mycobacteriosis, Autosomal Recessive	TYK2	Autosomal Recessive	NM_003331.4
Familial Bone Marrow Failure	SRP72	Autosomal Dominant	NM_006947.3
Familial Candidiasis, Dominant	IL17F	Autosomal Dominant	NM_052872.3
Familial Candidiasis, Recessive	IL17RA	Autosomal Recessive	NM_014339.6
Familial Cerebral Cavernous Malformation	KRIT1	Autosomal Dominant	NM_194456.1
Familial Cerebral Cavernous Malformation	PDCD10	Autosomal Dominant	NM_145860.1
Familial Chloride Diarrhea	SLC26A3	Autosomal Recessive	NM_000111.2
Familial Cold Autoinflammatory Syndrome	NLRP12	Autosomal Dominant	NM_144687.2
Familial Cold Autoinflammatory Syndrome	NLRP3	Autosomal Dominant	NM_004895.4
Familial Cylindromatosis	CYLD	Autosomal Dominant	NM_015247.2
Familial Dysautonomia	IKBKAP	Autosomal Recessive	NM_003640.3
Familial Encephalopathy with Neuroserpin Inclusion Bodies	SERPINI1	Autosomal Dominant	NM_005025.4
Familial Erythrocytosis	EGLN1	Autosomal Dominant	NM_022051.2
Familial Erythrocytosis	EPAS1	Autosomal Dominant	NM_001430.4
Familial Erythrocytosis	EPOR	Autosomal Dominant	NM_000121.3
Familial Exudative Vitreoretinopathy	FZD4	Autosomal Dominant	NM_012193.3
Familial Exudative Vitreoretinopathy	TSPAN12	Autosomal Dominant	NM_012338.3
Familial Febrile Seizures	SCN9A	Autosomal Dominant	NM_002977.3
Familial Hemiplegic Migraine	ATP1A2	Autosomal Dominant	NM_000702.3
Familial Hemiplegic Migraine	SCN1A	Autosomal Dominant	NM_001165963.1
Familial Hemophagocytic Lymphohistiocytosis	PRF1	Autosomal Recessive	NM_001083116.1

Familial Hemophagocytic Lymphohistiocytosis	STX11	Autosomal Recessive	NM_003764.3
Familial Hemophagocytic Lymphohistiocytosis	STXBP2	Autosomal Recessive	NM_006949.3
Familial Hemophagocytic Lymphohistiocytosis	UNC13D	Autosomal Recessive	NM_199242.2
Familial High Density Lipoprotein Deficiency	ABCA1	Autosomal Dominant	NM_005502.3
Familial High Density Lipoprotein Deficiency	APOA1	Autosomal Dominant	NM_000039.1
Familial Horizontal Gaze Palsy with Progressive Scoliosis	ROBO3	Autosomal Recessive	NM_022370.3
Familial Hyperaldosteronism	KCNJ5	Autosomal Dominant	NM_000890.3
Familial Hypercholesterolemia	APOB	Autosomal Dominant	NM_000384.2
Familial Hypercholesterolemia	LDLR	Autosomal Dominant	NM_000527.4
Familial Hypercholesterolemia	LDLRAP1	Autosomal Dominant	NM_015627.2
Familial Hypercholesterolemia	PCSK9	Autosomal Dominant	NM_174936.3
Familial Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome	PRKAG2	Autosomal Dominant	NM_016203.3
Familial Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome	TNNI3	Autosomal Dominant	NM_000363.4
Familial Hypocalciuric Hypercalcemia	CASR	Autosomal Dominant	NM_000388.3
Familial Idiopathic Basal Ganglia Calcification	SLC20A2	Autosomal Dominant	NM_006749.4
Familial Infantile Myoclonic Epilepsy	TBC1D24	Autosomal Recessive	NM_020705.2
Familial Intrahepatic Cholestasis	ABCB11	Autosomal Recessive	NM_003742.2
Familial Intrahepatic Cholestasis	ABCB4	Autosomal Recessive	NM_000443.3
Familial Intrahepatic Cholestasis	ATP8B1	Autosomal Recessive	NM_005603.4
Familial Isolated Hypoparathyroidism	CASR	Autosomal Dominant	NM_000388.3
Familial Isolated Hypoparathyroidism	GCM2	Autosomal Dominant	NM_004752.3
Familial Isolated Hypoparathyroidism	PTH	Autosomal Dominant	NM_000315.2
Familial Isolated Noncompaction of Left Ventricular Myocardium, Dominant	DTNA	Autosomal Dominant	NM_032978.6
Familial Isolated Noncompaction of Left Ventricular Myocardium, X-Linked	TAZ	X-Linked	NM_000116.3
Familial Isolated Pituitary Adenomas	AIP	Autosomal Dominant	NM_003977.2
Familial Juvenile Hyperuricemic Nephropathy	REN	Autosomal Dominant	NM_000537.3

Familial Lipoprotein Lipase Deficiency	LPL	Autosomal Recessive	NM_000237.2
Familial Mediterranean Fever	MEFV	Autosomal Recessive	NM_000243.2
Familial Paroxysmal Nonkinesigenic Dyskinesia	PNKD	Autosomal Dominant	NM_015488.4
Familial Partial Lipodystrophy	LMNA	Autosomal Dominant	NM_005572.3
Familial Partial Lipodystrophy	PPARG	Autosomal Dominant	NM_015869.4
Familial Periodic Fever	TNFRSF1A	Autosomal Dominant	NM_001065.3
Familial Platelet Disorder with Propensity to Acute Myelogenous Leukemia	RUNX1	Autosomal Dominant	NM_001754.4
Familial Pulmonary Fibrosis	SFTPC	Autosomal Dominant	NM_003018.3
Familial Restrictive Cardiomyopathy	TNNI3	Autosomal Dominant	NM_000363.4
Familial Restrictive Cardiomyopathy	TNNT2	Autosomal Dominant	NM_001001430.1
Familial Spinal Neurofibromatosis	NF1	Autosomal Dominant	NM_000267.3
Familial Temporal Lobe Epilepsy	CPA6	Autosomal Dominant	NM_020361.4
Familial Thrombotic Thrombocytopenia Purpura	ADAMTS13	Autosomal Recessive	NM_139025.3
Familial Transthyretin Amyloidosis	TTR	Autosomal Dominant	NM_000371.3
Familial Visceral Amyloidosis	APOA1	Autosomal Dominant	NM_000039.1
Familial Visceral Amyloidosis	FGA	Autosomal Dominant	NM_021871.2
Familial Visceral Amyloidosis	LYZ	Autosomal Dominant	NM_000239.2
Fanconi Anemia	BRCA2	Autosomal Recessive	NM_000059.3
Fanconi Anemia	BRIP1	Autosomal Recessive	NM_032043.2
Fanconi Anemia	FANCA	Autosomal Recessive	NM_000135.2
Fanconi Anemia	FANCC	Autosomal Recessive	NM_000136.2
Fanconi Anemia	FANCD2	Autosomal Recessive	NM_033084.3
Fanconi Anemia	FANCE	Autosomal Recessive	NM_021922.2
Fanconi Anemia	FANCF	Autosomal Recessive	NM_022725.3
Fanconi Anemia	FANCG	Autosomal Recessive	NM_004629.1
Fanconi Anemia	FANCI	Autosomal Recessive	NM_001113378.1
Fanconi Anemia	FANCL	Autosomal Recessive	NM_001114636.1
Fanconi Anemia	FANCM	Autosomal Recessive	NM_020937.2

Fanconi Anemia	PALB2	Autosomal Recessive	NM_024675.3
Fanconi Anemia	RAD51C	Autosomal Recessive	NM_058216.1
Fanconi Anemia	SLX4	Autosomal Recessive	NM_032444.2
Fanconi Anemia, X-Linked	FANCB	X-Linked	NM_001018113.1
Fanconi-Bickel Syndrome	SLC2A2	Autosomal Recessive	NM_000340.1
Farber Lipogranulomatosis	ASAH1	Autosomal Recessive	NM_177924.3
Fatal Infantile Cardioencephalomyopathy	SCO2	Autosomal Recessive	NM_005138.2
Fatal Infantile Cardioencephalomyopathy	SCO2	Autosomal Recessive	NM_005138.2
Fatal Infantile Lactic Acidosis	SUCLG1	Autosomal Recessive	NM_003849.3
Fatty Acid Hydroxylase-Associated Neurodegeneration	FA2H	Autosomal Recessive	NM_024306.4
Fechtner Syndrome	MYH9	Autosomal Dominant	NM_002473.4
Feingold Syndrome	MIR17HG	Autosomal Dominant	NR_027350.1
Fetal Akinesia Deformation Sequence	RAPSN	Autosomal Recessive	NM_005055.4
Fibrochondrogenesis	COL11A1	Autosomal Recessive	NM_001854.3
Fibrochondrogenesis	COL11A2	Autosomal Recessive	NM_080680.2
Fibrodysplasia Ossificans Progressiva	ACVR1	Autosomal Dominant	NM_001105.4
Fibular Hypoplasia and Complex Brachydactyly	GDF5	Autosomal Recessive	NM_000557.2
FLNB-Related Spectrum Disorders	FLNB	Mixed	NM_001457.3
Floating-Harbor Syndrome	SRCAP	Autosomal Dominant	NM_006662.2
Focal Cortical Dysplasia of Taylor	TSC1	Autosomal Dominant	NM_000368.4
Focal Segmental Glomerulosclerosis	CD2AP	Autosomal Recessive	NM_012120.2
Focal Segmental Glomerulosclerosis	TRPC6	Autosomal Recessive	NM_004621.5
Formiminotransferase Deficiency	FTCD	Autosomal Recessive	NM_006657.2
Foveal Hypoplasia and Presenile Cataract Syndrome	PAX6	Autosomal Dominant	NM_000280.4
Frank-ter Haar Syndrome	SH3PXD2B	Autosomal Recessive	NM_001017995.2
Fraser Syndrome	FRAS1	Autosomal Recessive	NM_025074.6
Fraser Syndrome	FREM2	Autosomal Recessive	NM_207361.4
Fraser Syndrome	GRIP1	Autosomal Recessive	NM_021150.3

Free Sialic Acid Storage Disorders	SLC17A5	Autosomal Recessive	NM_012434.4
Freeman-Sheldon Syndrome	MYH3	Autosomal Recessive	NM_002470.3
Friedreich Ataxia	FXN	Autosomal Recessive	NM_000144.4
Frontotemporal Dementia	CHMP2B	Autosomal Dominant	NM_014043.3
Frontotemporal Dementia	GRN	Autosomal Dominant	NM_002087.2
Frontotemporal Dementia	TARDBP	Autosomal Dominant	NM_007375.3
Fructose 1,6 Bisphosphatase Deficiency	FBP1	Autosomal Dominant	NM_000507.3
Fucosidosis	FUCA1	Autosomal Recessive	NM_000147.4
Fumarate Hydratase Deficiency	FH	Autosomal Recessive	NM_000143.3
Fundus Albipunctatus	PRPH2	Mixed	NM_000322.4
Fundus Albipunctatus	RDH5	Mixed	NM_002905.3
Fundus Albipunctatus	RLBP1	Mixed	NM_000326.4
Furlong Syndrome	TGFBR1	Autosomal Dominant	NM_004612.2
GABA-Transaminase Deficiency	ABAT	Autosomal Recessive	NM_020686.5
Galactokinase Deficiency	GALK1	Autosomal Recessive	NM_000154.1
Galactosemia	GALT	Autosomal Recessive	NM_000155.2
Galactosialidosis	CTSA	Autosomal Recessive	NM_000308.2
Gamma-Sarcoglycanopathy	SGCG	Autosomal Recessive	NM_000231.2
Gastrointestinal Stromal Tumor	KIT	Autosomal Dominant	NM_000222.2
Gastrointestinal Stromal Tumor	PDGFRA	Autosomal Dominant	NM_006206.4
Geleophysic Dysplasia	ADAMTSL2	Autosomal Dominant	NM_014694.3
Geleophysic Dysplasia	FBN1	Autosomal Dominant	NM_000138.4
Generalized Arterial Calcification of Infancy	ENPP1	Autosomal Recessive	NM_006208.2
Generalized Epilepsy and Paroxysmal Dyskinesia	KCNMA1	Autosomal Dominant	NM_002247.3
Generalized Epilepsy with Febrile Seizures Plus	GABRG2	Autosomal Dominant	NM_000816.3
Generalized Epilepsy with Febrile Seizures Plus	SCN1B	Autosomal Dominant	NM_001037.4
Generalized Epilepsy with Febrile Seizures Plus	SCN9A	Autosomal Dominant	NM_002977.3
Generalized Pustular Psoriasis	IL36RN	Autosomal Recessive	NM_012275.2

Genetic Prion Diseases	PRNP	Autosomal Dominant	NM_000311.3
Geroderma Osteodysplasticum	GORAB	Autosomal Recessive	NM_152281.2
Giant Axonal Neuropathy	GAN	Autosomal Recessive	NM_022041.3
Gilbert Syndrome	UGT1A1	Autosomal Recessive	NM_000463.2
Gingival Fibromatosis	SOS1	Autosomal Dominant	NM_005633.3
Gitelman Syndrome	SLC12A3	Autosomal Recessive	NM_000339.2
Glaucoma	MYOC	Autosomal Dominant	NM_000261.1
Global Cerebral Hypomyelination	SLC25A12	Autosomal Recessive	NM_003705.4
Glomuvenous Malformation	GLMN	Autosomal Dominant	NM_053274.2
Glucocorticoid Deficiency	MC2R	Autosomal Recessive	NM_000529.2
Glucocorticoid Deficiency	MRAP	Autosomal Recessive	NM_178817.3
Glucocorticoid Resistance	NR3C1	Autosomal Dominant	NM_001018077.1
Glucocorticoid-Remediable Aldosteronism	CYP11B1	Autosomal Dominant	NM_000497.3
Glucocorticoid-Remediable Aldosteronism	CYP11B2	Autosomal Dominant	NM_000498.3
Glucose Transporter Type 1 Deficiency Syndrome	SLC2A1	Autosomal Dominant	NM_006516.2
Glucose-6-Phosphate Dehydrogenase Deficiency	G6PD	X-Linked	NM_001042351.1
Glucose-Galactose Malabsorption	SLC5A1	Autosomal Recessive	NM_000343.3
Glutaricacidemia	GCDH	Autosomal Recessive	NM_000159.2
Glutathione Synthetase Deficiency	GSS	Autosomal Recessive	NM_000178.2
Glycine Encephalopathy	AMT	Autosomal Recessive	NM_000481.3
Glycine Encephalopathy	GLDC	Autosomal Recessive	NM_000170.2
Glycogen Storage Disease of Heart, Lethal Congenital	PRKAG2	Autosomal Dominant	NM_016203.3
Glycogen Storage Disease Type 0, Liver	GYS2	Autosomal Recessive	NM_021957.3
Glycogen Storage Disease Type 0, Muscle	GYS1	Autosomal Recessive	NM_002103.4
Glycogen Storage Disease Type I	G6PC	Autosomal Recessive	NM_000151.3
Glycogen Storage Disease Type I	SLC37A4	Autosomal Recessive	NM_001164277.1
Glycogen Storage Disease Type III	AGL	Autosomal Recessive	NM_000642.2
Glycogen Storage Disease Type IV	GBE1	Autosomal Recessive	NM_000158.3

Glycogen Storage Disease Type V	PYGM	Autosomal Recessive	NM_005609.2
Glycogen Storage Disease Type VI	PYGL	Autosomal Recessive	NM_002863.4
Glycogen Storage Disease Type VII	PFKM	Autosomal Recessive	NM_000289.5
Glycogen Storage Disease Type X	PGAM2	Autosomal Recessive	NM_000290.3
Glycogen Storage Disease Type XIII	ENO3	Autosomal Recessive	NM_053013.3
Glycogen Storage Disease Type XIV	PGM1	Autosomal Recessive	NM_002633.2
Glycogen Storage Disease XI	LDHA	Autosomal Recessive	NM_005566.3
Glycogen Storage Disease, Type II	GAA	Autosomal Recessive	NM_000152.3
Glycoprotein 1a Deficiency	ITGA2	Autosomal Dominant	NM_002203.3
GM1 Gangliosidosis	GLB1	Autosomal Recessive	NM_000404.2
GM2 Activator Deficiency	GM2A	Autosomal Recessive	NM_000405.4
Goldberg-Shprintzen Megacolon Syndrome	KIAA1279	Autosomal Recessive	NM_015634.3
Gracile Syndrome	BCS1L	Autosomal Recessive	NM_004328.4
Gray Platelet Syndrome	NBEAL2	Autosomal Recessive	NM_015175.2
Greenberg Dysplasia	LBR	Autosomal Recessive	NM_002296.3
Greig Cephalopolysyndactyly Syndrome	GLI3	Autosomal Dominant	NM_000168.5
Griscelli Syndrome	RAB27A	Autosomal Dominant	NM_004580.4
Growth Retardation, Developmental Delay, Coarse Facies, and Early Death	FTO	Autosomal Recessive	NM_001080432.2
GTP Cyclohydrolase 1 Deficiency (GTPCH)	GCH1	Autosomal Recessive	NM_000161.2
Guanidinoacetate Methyltransferase Deficiency	GAMT	Autosomal Recessive	NM_000156.4
Haim-Munk Syndrome	CTSC	Autosomal Recessive	NM_001814.4
Hawkinsinuria	HPD	Autosomal Dominant	NM_002150.2
Hemoglobin E	HBB	Autosomal Recessive	NM_000518.4
Hemolytic Anemia	SLC4A1	Autosomal Recessive	NM_000342.3
Hennekam Lymphangiectasia-Lymphedema Syndrome	CCBE1	Autosomal Recessive	NM_133459.3
Hepatic Failure, Early-Onset, and Neurologic Disorder due to Cytochrome C Oxidase Deficiency	SCO1	Autosomal Recessive	NM_004589.2
Hepatic Lipase Deficiency	LIPC	Autosomal Recessive	NM_000236.2

Hepatic Veno-occlusive Disease with Immunodeficiency	SP110	Autosomal Recessive	NM_004509.3
Hepatocerebral Mitochondrial DNA Depletion Syndrome	MPV17	Autosomal Recessive	NM_002437.4
Hereditary Angioedema	F12	Autosomal Dominant	NM_000505.3
Hereditary Angioedema	SERPING1	Autosomal Dominant	NM_000062.2
Hereditary Angiopathy with Nephropathy, Aneurysms, and Muscle Cramps	COL4A1	Autosomal Dominant	NM_001845.4
Hereditary Breast and Ovarian Cancer	BRCA1	Autosomal Dominant	NM_007294.3
Hereditary Breast and Ovarian Cancer	BRCA2	Autosomal Dominant	NM_000059.3
Hereditary Coproporphyria	CPOX	Autosomal Dominant	NM_000097.5
Hereditary Diffuse Gastric Cancer	CDH1	Autosomal Dominant	NM_004360.3
Hereditary Diffuse Leukoencephalopathy with Spheroids	CSF1R	Autosomal Dominant	NM_005211.3
Hereditary Essential Tremor	DRD3	Autosomal Dominant	NM_000796.3
Hereditary Folate Malabsorption	SLC46A1	Autosomal Recessive	NM_080669.4
Hereditary Fructose Intolerance	ALDOB	Autosomal Recessive	NM_000035.3
Hereditary Hemochromatosis	HFE	Autosomal Recessive	NM_000410.3
Hereditary Hemochromatosis	SLC40A1	Autosomal Recessive	NM_014585.5
Hereditary Hemochromatosis	TFR2	Autosomal Recessive	NM_003227.3
Hereditary Hemorrhagic Telangiectasia	ACVRL1	Autosomal Dominant	NM_000020.2
Hereditary Hemorrhagic Telangiectasia	ENG	Autosomal Dominant	NM_000118.2
Hereditary Hemorrhagic Telangiectasia	SMAD4	Autosomal Dominant	NM_005359.5
Hereditary Keratitis	PAX6	Autosomal Dominant	NM_000280.4
Hereditary Leiomyomatosis and Renal Cell Cancer	FH	Autosomal Dominant	NM_000143.3
Hereditary Motor and Sensory Neuropathy	MFN2	Autosomal Recessive	NM_014874.3
Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum	SLC12A6	Autosomal Recessive	NM_133647.1
Hereditary Multiple Osteochondromatosis	EXT1	Autosomal Dominant	NM_000127.2
Hereditary Multiple Osteochondromatosis	EXT2	Autosomal Dominant	NM_207122.1
Hereditary Myopathy with Early Respiratory Failure	TTN	Autosomal Dominant	NM_133378.4
Hereditary Neuropathy with Liability to Pressure Palsies	PMP22	Autosomal Dominant	NM_000304.2

Hereditary Pancreatitis	CFTR	Autosomal Dominant	NM_000492.3
Hereditary Pancreatitis	CTRC	Autosomal Dominant	NM_007272.2
Hereditary Pancreatitis	SPINK1	Autosomal Dominant	NM_003122.3
Hereditary Paraganglioma-Pheochromocytoma Syndrome	MAX	Autosomal Dominant	NM_002382.4
Hereditary Paraganglioma-Pheochromocytoma Syndrome	SDHAF2	Autosomal Dominant	NM_017841.2
Hereditary Paraganglioma-Pheochromocytoma Syndrome	SDHB	Autosomal Dominant	NM_003000.2
Hereditary Persistence of Fetal Hemoglobin	HBB	Autosomal Recessive	NM_000518.4
Hereditary Persistence of Fetal Hemoglobin	HBD	Autosomal Recessive	NM_000519.3
Hereditary Pyropoikilocytosis	SPTA1	Autosomal Recessive	NM_003126.2
Hereditary Sensory and Autonomic Neuropathy, Type IC	SPTLC2	Autosomal Dominant	NM_004863.3
Hereditary Sensory and Autonomic Neuropathy, Type II	FAM134B	Autosomal Recessive	NM_001034850.2
Hereditary Sensory and Autonomic Neuropathy, Type II	KIF1A	Autosomal Recessive	NM_004321.6
Hereditary Sensory and Autonomic Neuropathy, Type II	SPTLC1	Autosomal Recessive	NM_006415.2
Hereditary Sensory and Autonomic Neuropathy, Type II	WNK1	Autosomal Recessive	NM_018979.3
Hereditary Sensory and Autonomic Neuropathy, Type IV	NTRK1	Autosomal Recessive	NM_001012331.1
Hereditary Sensory and Autonomic Neuropathy, Type V	NGF	Autosomal Recessive	NM_002506.2
Hereditary Sensory and Autonomic Neuropathy, Type VI	DST	Autosomal Recessive	NM_001723.5
Hereditary Sideroblastic Anemia	SLC25A38	Autosomal Recessive	NM_017875.2
Hereditary Sideroblastic Anemia with Myopathy and Lactic Acidosis	YARS2	Autosomal Recessive	NM_001040436.2
Hereditary Sideroblastic Anemia with Spinocerebellar Ataxia	ABCB7	X-Linked	NM_004299.4
Heritable Pulmonary Arterial Hypertension	BMPR2	Autosomal Dominant	NM_001204.6
Heritable Pulmonary Arterial Hypertension	SMAD9	Autosomal Dominant	NM_001127217.2
Hermanskly-Pudlak Syndrome	AP3B1	Autosomal Recessive	NM_003664.4
Hermanskly-Pudlak Syndrome	DTNBP1	Autosomal Recessive	NM_032122.4
Hermanskly-Pudlak Syndrome	HPS1	Autosomal Recessive	NM_000195.3
Hermanskly-Pudlak Syndrome	HPS3	Autosomal Recessive	NM_032383.3
Hermanskly-Pudlak Syndrome	HPS4	Autosomal Recessive	NM_022081.5
Hermanskly-Pudlak Syndrome	HPS5	Autosomal Recessive	NM_181507.1

Hermanskly-Pudlak Syndrome	HPS6	Autosomal Recessive	NM_024747.5
Heterotaxy Syndrome	ACVR2B	Autosomal Dominant	NM_001106.3
Heterotaxy Syndrome	NODAL	Autosomal Dominant	NM_018055.4
Hexosaminidase A Deficiency	HEXA	Autosomal Recessive	NM_000520.4
Hidrotic Ectodermal Dysplasia	GJB6	Autosomal Dominant	NM_006783.4
Hirschsprung Disease, Dominant	EDN3	Autosomal Dominant	NM_207034.1
Hirschsprung Disease, Dominant	RET	Autosomal Dominant	NM_020975.4
Hirschsprung Disease, Recessive	EDNRB	Autosomal Recessive	NM_000115.3
Histidinemia	HAL	Autosomal Recessive	NM_002108.3
Holocarboxylase Synthetase Deficiency	HLCS	Autosomal Recessive	NM_000411.6
Holoprosencephaly	CDON	Autosomal Dominant	NM_016952.4
Holoprosencephaly	FOXH1	Autosomal Dominant	NM_003923.2
Holoprosencephaly	NODAL	Autosomal Dominant	NM_018055.4
Holoprosencephaly	PTCH1	Autosomal Dominant	NM_000264.3
Holoprosencephaly	TGIF1	Autosomal Dominant	NM_173208.1
Holt-Oram Syndrome	TBX5	Autosomal Dominant	NM_000192.3
Homocystinuria	CBS	Autosomal Recessive	NM_000071.2
Humerospinal Dysostosis	CHST3	Autosomal Recessive	NM_004273.4
Hutchinson-Gilford Progeria Syndrome	LMNA	Autosomal Dominant	NM_005572.3
Hydrolethalus Syndrome	HYLS1	Autosomal Recessive	NM_145014.2
Hydroxymethylbilane Synthase Deficiency	HMBS	Autosomal Dominant	NM_000190.3
Hyper IgD Syndrome	MVK	Autosomal Recessive	NM_000431.2
Hyper IgE Syndrome	DOCK8	Autosomal Recessive	NM_203447.3
Hyper IgE Syndrome	STAT3	Autosomal Recessive	NM_139276.2
Hyperalphalipoproteinemia	CETP	Autosomal Dominant	NM_000078.2
Hypercholanemia	BAAT	Autosomal Recessive	NM_001701.3
Hyperekplexia	GLRA1	Mixed	NM_000171.3
Hyperekplexia	GLRB	Mixed	NM_000824.4

Hyperekplexia	SLC6A5	Mixed	NM_004211.3
Hypereosinophilic Syndrome	PDGFRA	Autosomal Dominant	NM_006206.4
Hyperferritinemia Cataract Syndrome	FTL	Autosomal Dominant	NM_000146.3
Hyperglycinuria	SLC6A20	Autosomal Dominant	NM_020208.3
Hypergonadotropic Hypogonadism	LHCGR	Autosomal Recessive	NM_000233.3
Hyperinsulinism, Dominant	GCK	Autosomal Dominant	NM_000162.3
Hyperinsulinism, Dominant/Recessive	ABCC8	Mixed	NM_000352.3
Hyperinsulinism, Dominant/Recessive	GLUD1	Mixed	NM_005271.3
Hyperinsulinism, Dominant/Recessive	HADH	Mixed	NM_005327.4
Hyperinsulinism, Dominant/Recessive	HNF4A	Mixed	NM_000457.4
Hyperinsulinism, Dominant/Recessive	KCNJ11	Mixed	NM_000525.3
Hyperinsulinism, Dominant/Recessive	SLC16A1	Mixed	NM_003051.3
Hyperkalemic Periodic Paralysis	SCN4A	Autosomal Dominant	NM_000334.4
Hypermethioninemia	ADK	Autosomal Recessive	NM_001123.3
Hypermethioninemia	AHCY	Autosomal Recessive	NM_000687.2
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	SLC25A15	Autosomal Recessive	NM_014252.3
Hyperparathyroidism	MEN1	Autosomal Dominant	NM_130799.2
Hyperparathyroidism-Jaw Tumor Syndrome	CDC73	Autosomal Dominant	NM_024529.4
Hyperphosphatasia with Mental Retardation Syndrome	PIGV	Autosomal Recessive	NM_017837.3
Hyperphosphatemic Familial Tumoral Calcinosis	FGF23	Autosomal Recessive	NM_020638.2
Hyperphosphatemic Familial Tumoral Calcinosis	GALNT3	Autosomal Recessive	NM_004482.3
Hyperphosphatemic Familial Tumoral Calcinosis	KL	Autosomal Recessive	NM_004795.3
Hyperpigmentation, Cutaneous, with Hypertrichosis, Hepatosplenomegaly, Heart Anomalies, Hearing Loss, and Hypogonadism	SLC29A3	Autosomal Recessive	NM_018344.5
Hyperprolinemia	ALDH4A1	Autosomal Recessive	NM_003748.3
Hypertrichotic Osteochondrodysplasia	ABCC9	Autosomal Dominant	NM_005691.2
Hypertrophic Cardiomyopathy	ACTC1	Autosomal Dominant	NM_005159.4
Hypertrophic Cardiomyopathy	ACTN2	Autosomal Dominant	NM_001103.2

Hypertrophic Cardiomyopathy	CSRP3	Autosomal Dominant	NM_003476.4
Hypertrophic Cardiomyopathy	LAMP2	Autosomal Dominant	NM_002294.2
Hypertrophic Cardiomyopathy	MYBPC3	Autosomal Dominant	NM_000256.3
Hypertrophic Cardiomyopathy	MYH6	Autosomal Dominant	NM_002471.3
Hypertrophic Cardiomyopathy	MYH7	Autosomal Dominant	NM_000257.2
Hypertrophic Cardiomyopathy	MYL2	Autosomal Dominant	NM_000432.3
Hypertrophic Cardiomyopathy	MYL3	Autosomal Dominant	NM_000258.2
Hypertrophic Cardiomyopathy	MYLK2	Autosomal Dominant	NM_033118.3
Hypertrophic Cardiomyopathy	MYOZ2	Autosomal Dominant	NM_016599.3
Hypertrophic Cardiomyopathy	NEXN	Autosomal Dominant	NM_144573.3
Hypertrophic Cardiomyopathy	PLN	Autosomal Dominant	NM_002667.3
Hypertrophic Cardiomyopathy	TCAP	Autosomal Dominant	NM_003673.3
Hypertrophic Cardiomyopathy	TNNC1	Autosomal Dominant	NM_003280.2
Hypertrophic Cardiomyopathy	TNNI3	Autosomal Dominant	NM_000363.4
Hypertrophic Cardiomyopathy	TNNT2	Autosomal Dominant	NM_001001430.1
Hypertrophic Cardiomyopathy	TPM1	Autosomal Dominant	NM_001018005.1
Hypertrophic Cardiomyopathy	TTN	Autosomal Dominant	NM_133378.4
Hyperuricemia, Pulmonary Hypertension, Renal Failure, and Alkalosis	SARS2	Autosomal Recessive	NM_017827.3
Hypocalcemia	CASR	Autosomal Dominant	NM_000388.3
Hypochromic Microcytic Anemia with Iron Overload	SLC11A2	Autosomal Recessive	NM_000617.2
Hypohidrotic Ectodermal Dysplasia, Dominant	EDAR	Autosomal Dominant	NM_022336.3
Hypohidrotic Ectodermal Dysplasia, Recessive	EDARADD	Autosomal Recessive	NM_145861.2
Hypokalemic Periodic Paralysis	CACNA1S	Autosomal Dominant	NM_000069.2
Hypokalemic Periodic Paralysis	KCNE3	Autosomal Dominant	NM_005472.4
Hypokalemic Periodic Paralysis	SCN4A	Autosomal Dominant	NM_000334.4
Hypomagnesemia with Secondary Hypocalcemia	TRPM6	Autosomal Recessive	NM_017662.4
Hypomyelination and Congenital Cataract	FAM126A	Autosomal Recessive	NM_032581.3
Hypoparathyroidism, Sensorineural Deafness, and Renal Disease	GATA3	Autosomal Dominant	NM_001002295.1

Hypoparathyroidism-Retardation-Dysmorphism Syndrome	TBCE	Autosomal Recessive	NM_003193.3
Hypophosphatasia	ALPL	Autosomal Dominant	NM_000478.4
Hypophosphatemic Nephrolithiasis/Osteoporosis	SLC34A1	Autosomal Recessive	NM_003052.4
Hypophosphatemic Rickets, Dominant	FGF23	Autosomal Dominant	NM_020638.2
Hypophosphatemic Rickets, Recessive	DMP1	Autosomal Recessive	NM_004407.3
Hypophosphatemic Rickets, Recessive	ENPP1	Autosomal Recessive	NM_006208.2
Hypoplastic Left Heart Syndrome	GJA1	Autosomal Recessive	NM_000165.3
Ichthyosis Bullosa of Siemens	KRT2	Autosomal Dominant	NM_000423.2
Ichthyosis, Hystrix-like, with Deafness	GJB2	Autosomal Dominant	NM_004004.5
Immunodeficiency due to Defect in CD3-Gamma	CD3G	Autosomal Recessive	NM_000073.2
Immunodeficiency with Hyper-IgM	AICDA	Autosomal Recessive	NM_020661.2
Immunodeficiency with Hyper-IgM	CD40	Autosomal Recessive	NM_001250.4
Immunodeficiency with Hyper-IgM	UNG	Autosomal Recessive	NM_080911.2
Immunodeficiency-Centromeric Instability-Facial Anomalies Syndrome	DNMT3B	Autosomal Recessive	NM_006892.3
Inclusion Body Myopathy, Dominant	MYH2	Autosomal Dominant	NM_017534.5
Inclusion Body Myopathy, Dominant	VCP	Autosomal Dominant	NM_007126.3
Inclusion Body Myopathy, Recessive	GNE	Autosomal Recessive	NM_005476.5
Infantile Hypercalcemia	CYP24A1	Autosomal Dominant	NM_000782.4
Infantile Nystagmus	FRMD7	X-Linked	NM_194277.2
Inflammatory Bowel Disease	IL10RA	Autosomal Recessive	NM_001558.3
Inflammatory Bowel Disease	IL10RB	Autosomal Recessive	NM_000628.4
Inherited Erythromelalgia	SCN9A	Autosomal Dominant	NM_002977.3
Inherited Systemic Hyalinosis	ANTXR2	Autosomal Recessive	NM_058172.5
Insulin-Like Growth Factor I Deficiency	IGF1	Autosomal Recessive	NM_000618.3
Insulin-Like Growth Factor I Deficiency	IGF1R	Autosomal Recessive	NM_000875.3
Intellectual Disability Syndrome	KANSL1	Autosomal Dominant	NM_001193466.1
Interleukin 1 Receptor Antagonist Deficiency	IL1RN	Autosomal Recessive	NM_173841.2
Interleukin 2 Receptor Alpha Chain Deficiency	IL2RA	Autosomal Recessive	NM_000417.2

Intrinsic Factor Deficiency	GIF	Autosomal Recessive	NM_005142.2
IRAK4 Deficiency	IRAK4	Autosomal Recessive	NM_016123.3
Iris Hypoplasia	PITX2	Autosomal Dominant	NM_153427.2
Iron Overload	FTH1	Autosomal Dominant	NM_002032.2
Iron-Refractory Iron Deficiency Anemia	TMPRSS6	Autosomal Recessive	NM_153609.2
Isobutyryl-CoA Dehydrogenase Deficiency	ACAD8	Autosomal Recessive	NM_014384.2
Isolated Congenital Digital Clubbing	HPGD	Autosomal Recessive	NM_000860.5
Isolated Coronal Synostosis	FGFR2	Autosomal Dominant	NM_000141.4
Isolated Follicle-Stimulating Hormone Deficiency	FSHB	Autosomal Recessive	NM_000510.2
Isolated GnRH Deficiency	GNRH1	Autosomal Recessive	NM_000825.3
Isolated GnRH Deficiency	GNRHR	Autosomal Recessive	NM_000406.2
Isolated GnRH Deficiency	TACR3	Autosomal Recessive	NM_001059.2
Isolated Growth Hormone Deficiency	GH1	Autosomal Recessive	NM_000515.3
Isolated Growth Hormone Deficiency	GHRHR	Autosomal Recessive	NM_000823.3
Isolated Hyperparathyroidism	CDC73	Autosomal Dominant	NM_024529.4
Isolated Microphthalmia	MFRP	Autosomal Recessive	NM_031433.2
Isolated Microphthalmia	VSX2	Autosomal Recessive	NM_182894.2
Isolated Nonsyndromic Congenital Heart Disease	JAG1	Autosomal Dominant	NM_000214.2
Isolated Persistent Hypermethioninemia	MAT1A	Autosomal Dominant	NM_000429.2
Isovaleric Acidemia	IVD	Autosomal Recessive	NM_002225.3
Jackson-Weiss Syndrome	FGFR2	Autosomal Dominant	NM_000141.4
Jalili Syndrome	CNNM4	Autosomal Recessive	NM_020184.3
Jawad Syndrome	RBBP8	Autosomal Recessive	NM_002894.2
Jervell and Lange-Nielsen Syndrome	KCNE1	Autosomal Recessive	NM_000219.3
Jervell and Lange-Nielsen Syndrome	KCNQ1	Autosomal Recessive	NM_000218.2
Joubert Syndrome	AHI1	Autosomal Recessive	NM_017651.4
Joubert Syndrome	ARL13B	Autosomal Recessive	NM_182896.2
Joubert Syndrome	C5orf42	Autosomal Recessive	NM_023073.3

Joubert Syndrome	CC2D2A	Autosomal Recessive	NM_001080522.2
Joubert Syndrome	CEP290	Autosomal Recessive	NM_025114.3
Joubert Syndrome	CEP41	Autosomal Recessive	NM_018718.2
Joubert Syndrome	NPHP1	Autosomal Recessive	NM_000272.3
Joubert Syndrome	RPGRIP1L	Autosomal Recessive	NM_015272.2
Joubert Syndrome	TCTN1	Autosomal Recessive	NM_001082538.2
Joubert Syndrome	TCTN2	Autosomal Recessive	NM_024809.4
Joubert Syndrome	TMEM138	Autosomal Recessive	NM_016464.4
Joubert Syndrome	TMEM216	Autosomal Recessive	NM_001173990.2
Joubert Syndrome	TMEM237	Autosomal Recessive	NM_001044385.2
Joubert Syndrome	TMEM67	Autosomal Recessive	NM_153704.5
Joubert Syndrome	TTC21B	Autosomal Recessive	NM_024753.4
Junctional Epidermolysis Bullosa	COL17A1	Autosomal Recessive	NM_000494.3
Junctional Epidermolysis Bullosa	LAMA3	Autosomal Recessive	NM_000227.3
Junctional Epidermolysis Bullosa	LAMB3	Autosomal Recessive	NM_000228.2
Junctional Epidermolysis Bullosa	LAMC2	Autosomal Recessive	NM_005562.2
Juvenile Hereditary Hemochromatosis	HAMP	Autosomal Recessive	NM_021175.2
Juvenile Hereditary Hemochromatosis	HFE2	Autosomal Recessive	NM_213653.3
Juvenile Myoclonic Epilepsy	CACNB4	Autosomal Dominant	NM_000726.3
Juvenile Myoclonic Epilepsy	EFHC1	Autosomal Dominant	NM_018100.3
Juvenile Myoclonic Epilepsy	GABRA1	Autosomal Dominant	NM_000806.5
Juvenile Paget Disease	TNFRSF11B	Autosomal Recessive	NM_002546.3
Juvenile Polyposis	ENG	Autosomal Dominant	NM_000118.2
Juvenile Polyposis	SMAD4	Autosomal Dominant	NM_005359.5
Kabuki Syndrome	MLL2	Autosomal Dominant	NM_003482.3
Kallmann Syndrome	CHD7	Autosomal Dominant	NM_017780.3
Kallmann Syndrome	FGFR1	Autosomal Dominant	NM_023110.2
Kallmann Syndrome	PROKR2	Autosomal Dominant	NM_144773.2

Kanzaki disease	NAGA	Autosomal Recessive	NM_000262.2
KAT6B-Related Spectrum Disorders	KAT6B	Autosomal Dominant	NM_012330.3
Keratitis-Ichthyosis-Deafness Syndrome	GJB2	Mixed	NM_004004.5
Keratoconus	VSX1	Autosomal Dominant	NM_014588.5
Ketothiolase Deficiency	ACAT1	Autosomal Recessive	NM_000019.3
Keutel Syndrome	MGP	Autosomal Recessive	NM_000900.3
Kindler Syndrome	FERMT1	Autosomal Recessive	NM_017671.4
Kleefstra Syndrome	EHMT1	Autosomal Dominant	NM_024757.4
Klippel-Feil Syndrome	GDF6	Autosomal Dominant	NM_001001557.2
Knobloch Syndrome	COL18A1	Autosomal Recessive	NM_130445.2
Krabbe Disease	GALC	Autosomal Recessive	NM_000153.3
Krabbe Disease	PSAP	Autosomal Recessive	NM_002778.2
Kufor-Rakeb Syndrome	ATP13A2	Autosomal Recessive	NM_022089.2
Lacrimo-Auriculo-Dento-Digital Syndrome	FGF10	Autosomal Dominant	NM_004465.1
Lacrimo-Auriculo-Dento-Digital Syndrome	FGFR2	Autosomal Dominant	NM_000141.4
Lactate Dehydrogenase B Deficiency	LDHB	Autosomal Recessive	NM_002300.6
Lactose Intolerance	LCT	Autosomal Recessive	NM_002299.2
Laing Distal Myopathy	MYH7	Autosomal Dominant	NM_000257.2
Langer-Giedion Syndrome	EXT1	Autosomal Dominant	NM_000127.2
Langer-Giedion Syndrome	TRPS1	Autosomal Dominant	NM_014112.2
L-Arginine:Glycine Amidinotransferase Deficiency	GATM	Autosomal Recessive	NM_001482.2
Larsen Syndrome	CHST3	Autosomal Recessive	NM_004273.4
Laryngoonychocutaneous Syndrome	LAMA3	Autosomal Recessive	NM_000227.3
Lathosterolosis	SC5DL	Autosomal Recessive	NM_006918.4
LCHAD Deficiency	HADHA	Autosomal Recessive	NM_000182.4
Leber Congenital Amaurosis	AIPL1	Autosomal Recessive	NM_014336.3
Leber Congenital Amaurosis	CEP290	Autosomal Recessive	NM_025114.3
Leber Congenital Amaurosis	CRB1	Autosomal Recessive	NM_201253.2

Leber Congenital Amaurosis	CRX	Autosomal Recessive	NM_000554.4
Leber Congenital Amaurosis	IMPDH1	Autosomal Recessive	NM_000883.3
Leber Congenital Amaurosis	KCNJ13	Autosomal Recessive	NM_002242.4
Leber Congenital Amaurosis	LCA5	Autosomal Recessive	NM_181714.3
Leber Congenital Amaurosis	LRAT	Autosomal Recessive	NM_004744.3
Leber Congenital Amaurosis	RD3	Autosomal Recessive	NM_183059.2
Leber Congenital Amaurosis	RPE65	Autosomal Recessive	NM_000329.2
Leber Congenital Amaurosis	RPGRIP1	Autosomal Recessive	NM_020366.3
Leber Congenital Amaurosis	SPATA7	Autosomal Recessive	NM_018418.4
Leber Congenital Amaurosis	TULP1	Autosomal Recessive	NM_003322.3
Lecithin Cholesterol Acyltransferase Deficiency	LCAT	Autosomal Recessive	NM_000229.1
Legius Syndrome	SPRED1	Autosomal Dominant	NM_152594.2
Leigh Syndrome	BCS1L	Autosomal Recessive	NM_004328.4
Leigh Syndrome	COX10	Autosomal Recessive	NM_001303.3
Leigh Syndrome	DLD	Autosomal Recessive	NM_000108.3
Leigh Syndrome	LRPPRC	Autosomal Recessive	NM_133259.3
Leigh Syndrome	NDUFA10	Autosomal Recessive	NM_004544.3
Leigh Syndrome	NDUFAF2	Autosomal Recessive	NM_174889.4
Leigh Syndrome	NDUFS1	Autosomal Recessive	NM_005006.6
Leigh Syndrome	NDUFS3	Autosomal Recessive	NM_004551.2
Leigh Syndrome	NDUFS4	Autosomal Recessive	NM_002495.2
Leigh Syndrome	NDUFS7	Autosomal Recessive	NM_024407.4
Leigh Syndrome	NDUFS8	Autosomal Recessive	NM_002496.3
Leigh Syndrome	NDUFV1	Autosomal Recessive	NM_007103.3
Leigh Syndrome	SCO1	Autosomal Recessive	NM_004589.2
LEOPARD Syndrome	BRAF	Autosomal Dominant	NM_004333.4
LEOPARD Syndrome	PTPN11	Autosomal Dominant	NM_002834.3
LEOPARD Syndrome	RAF1	Autosomal Dominant	NM_002880.3

Leptin Deficiency	LEP	Autosomal Recessive	NM_000230.2
Leptin Receptor Deficiency	LEPR	Autosomal Recessive	NM_002303.5
Lethal Arthrogryposis With Anterior Horn Cell Disease	GLE1	Autosomal Dominant	NM_001003722.1
Lethal Congenital Contracture Syndrome	GLE1	Autosomal Recessive	NM_001003722.1
Lethal Encephalopathy	DNM1L	Autosomal Dominant	NM_012062.3
Lethal Restrictive Dermopathy	LMNA	Autosomal Recessive	NM_005572.3
Lethal Restrictive Dermopathy	ZMPSTE24	Autosomal Recessive	NM_005857.4
Leukocyte Adhesion Deficiency	ITGB2	Autosomal Recessive	NM_000211.3
Leukodystrophy, Adult-Onset	LMNB1	Autosomal Dominant	NM_005573.3
Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation	DARS2	Autosomal Recessive	NM_018122.4
Leukoencephalopathy, Cystic, without Megalencephaly	RNASET2	Autosomal Recessive	NM_003730.4
Leydig Cell Hypoplasia/Agenesis	LHCGR	Autosomal Recessive	NM_000233.3
Liddle Syndrome	SCNN1B	Autosomal Dominant	NM_000336.2
Li-Fraumeni Syndrome	TP53	Autosomal Dominant	NM_000546.4
LIG4 Syndrome	LIG4	Autosomal Recessive	NM_002312.3
Limb-Girdle Muscular Dystrophy, Dominant	CAV3	Autosomal Dominant	NM_033337.2
Limb-Girdle Muscular Dystrophy, Dominant	DNAJB6	Autosomal Dominant	NM_058246.3
Limb-Girdle Muscular Dystrophy, Dominant	MYOT	Autosomal Dominant	NM_006790.2
Limb-Girdle Muscular Dystrophy, Recessive	ANO5	Autosomal Recessive	NM_213599.2
Limb-Girdle Muscular Dystrophy, Recessive	CAPN3	Autosomal Recessive	NM_000070.2
Limb-Girdle Muscular Dystrophy, Recessive	DYSF	Autosomal Recessive	NM_003494.3
Limb-Girdle Muscular Dystrophy, Recessive	LMNA	Autosomal Recessive	NM_005572.3
Limb-Girdle Muscular Dystrophy, Recessive	POMGNT1	Autosomal Recessive	NM_017739.3
Limb-Girdle Muscular Dystrophy, Recessive	SGCA	Autosomal Recessive	NM_000023.2
Limb-Girdle Muscular Dystrophy, Recessive	SGCB	Autosomal Recessive	NM_000232.4
Limb-Girdle Muscular Dystrophy, Recessive	SGCD	Autosomal Recessive	NM_000337.5
Limb-Girdle Muscular Dystrophy, Recessive	SGCG	Autosomal Recessive	NM_000231.2

Limb-Girdle Muscular Dystrophy, Recessive	TCAP	Autosomal Recessive	NM_003673.3
Limb-Girdle Muscular Dystrophy, Recessive	TRIM32	Autosomal Recessive	NM_012210.3
Limb-Girdle Muscular Dystrophy, Recessive	TTN	Autosomal Recessive	NM_133378.4
Limb-Girdle Myasthenia with Tubular Aggregates	GFPT1	Autosomal Recessive	NM_002056.3
Lipoatrophy with Diabetes, Hepatic Steatosis, Hypertrophic Cardiomyopathy, and Leukomelanodermic Papules	LMNA	Autosomal Dominant	NM_005572.3
Lissencephaly, Dominant	TUBA1A	Autosomal Dominant	NM_006009.3
Lissencephaly, Recessive	NDE1	Autosomal Recessive	NM_001143979.1
Lissencephaly, Recessive	RELN	Autosomal Recessive	NM_005045.3
Lissencephaly/Subcortical Band Heterotopia	PAFAH1B1	Autosomal Dominant	NM_000430.3
Localized AR Hypotrichosis	DSG4	Autosomal Recessive	NM_177986.3
Loeys-Dietz Syndrome	SMAD3	Autosomal Dominant	NM_005902.3
Loeys-Dietz Syndrome	TGFBR1	Autosomal Dominant	NM_004612.2
Loeys-Dietz Syndrome	TGFBR2	Autosomal Dominant	NM_003242.5
Long QT Syndrome	AKAP9	Autosomal Dominant	NM_005751.4
Long QT Syndrome	ANK2	Autosomal Dominant	NM_001148.4
Long QT Syndrome	CAV3	Autosomal Dominant	NM_033337.2
Long QT Syndrome	KCNE1	Autosomal Dominant	NM_000219.3
Long QT Syndrome	KCNE2	Autosomal Dominant	NM_172201.1
Long QT Syndrome	KCNJ5	Autosomal Dominant	NM_000890.3
Long QT Syndrome	KCNQ1	Autosomal Dominant	NM_000218.2
Long QT Syndrome	SCN4B	Autosomal Dominant	NM_174934.3
Long QT Syndrome	SCN5A	Autosomal Dominant	NM_198056.2
Long QT Syndrome	SNTA1	Autosomal Dominant	NM_003098.2
Long-Chain Acyl-CoA Dehydrogenase Deficiency	ACADL	Autosomal Recessive	NM_001608.3
Lung Cancer	EGFR	Mixed	NM_005228.3
Lymphoproliferative Syndrome	ITK	Autosomal Recessive	NM_005546.3
Lynch Syndrome	EPCAM	Autosomal Dominant	NM_002354.2

Lynch Syndrome	MLH1	Autosomal Dominant	NM_000249.3
Lynch Syndrome	MLH3	Autosomal Dominant	NM_001040108.1
Lynch Syndrome	MSH2	Autosomal Dominant	NM_000251.1
Lynch Syndrome	MSH6	Autosomal Dominant	NM_000179.2
Lynch Syndrome	PMS1	Autosomal Dominant	NM_000534.4
Lysinuric Protein Intolerance	SLC7A7	Autosomal Recessive	NM_001126106.2
Macular Corneal Dystrophy	CHST6	Autosomal Recessive	NM_021615.4
Macular Degeneration	ABCA4	Autosomal Dominant	NM_000350.2
Macular Degeneration	ARMS2	Autosomal Dominant	NM_001099667.1
Macular Degeneration	C2	Autosomal Dominant	NM_000063.4
Macular Degeneration	C3	Autosomal Dominant	NM_000064.2
Macular Degeneration	CFB	Autosomal Dominant	NM_001710.5
Macular Degeneration	CFH	Autosomal Dominant	NM_000186.3
Macular Degeneration	ERCC6	Autosomal Dominant	NM_000124.2
Macular Degeneration	FBLN5	Autosomal Dominant	NM_006329.3
Macular Degeneration	HMCN1	Autosomal Dominant	NM_031935.2
Macular Degeneration	RAX2	Autosomal Dominant	NM_032753.3
Mainzer-Saldino Syndrome	IFT140	Autosomal Recessive	NM_014714.3
Majeed Syndrome	LPIN2	Autosomal Recessive	NM_014646.2
Mal de Meleda	SLURP1	Autosomal Recessive	NM_020427.2
Male Infertility	CATSPER1	Autosomal Recessive	NM_053054.3
Male-Limited Precocious Puberty	LHCGR	Autosomal Dominant	NM_000233.3
Malignant Hyperthermia Susceptibility	CACNA1S	Autosomal Dominant	NM_000069.2
Malignant Hyperthermia Susceptibility	RYR1	Autosomal Dominant	NM_000540.2
Malonyl-CoA Decarboxylase Deficiency	MLYCD	Autosomal Recessive	NM_012213.2
Mandibuloacral Dysplasia	LMNA	Autosomal Recessive	NM_005572.3
Mandibuloacral Dysplasia	ZMPSTE24	Autosomal Recessive	NM_005857.4
Manitoba Oculotrichoanal Syndrome	FREM1	Autosomal Recessive	NM_144966.5

Mannose-Binding Protein Deficiency	MBL2	Autosomal Recessive	NM_000242.2
Maple Syrup Urine Disease	BCKDHA	Autosomal Recessive	NM_000709.3
Maple Syrup Urine Disease	BCKDHB	Autosomal Recessive	NM_183050.2
Maple Syrup Urine Disease	DBT	Autosomal Recessive	NM_001918.2
Maple Syrup Urine Disease	DLD	Autosomal Recessive	NM_000108.3
MAPT-Related Spectrum Disorders	MAPT	Autosomal Dominant	NM_005910.5
Marfan Syndrome	FBN1	Autosomal Dominant	NM_000138.4
Marinesco-Sjogren Syndrome	SIL1	Autosomal Recessive	NM_022464.4
Marshall Syndrome	COL11A1	Mixed	NM_001854.3
Martsolf Syndrome	RAB3GAP2	Autosomal Recessive	NM_012414.3
MASP2 Deficiency	MASP2	Autosomal Recessive	NM_006610.3
MASS Syndrome	FBN1	Autosomal Dominant	NM_000138.4
Mast Cell Disease	KIT	Autosomal Dominant	NM_000222.2
Maturity Onset Diabetes of the Young	BLK	Autosomal Dominant	NM_001715.2
Maturity Onset Diabetes of the Young	GCK	Autosomal Dominant	NM_000162.3
Maturity Onset Diabetes of the Young	HNF1A	Autosomal Dominant	NM_000545.5
Maturity Onset Diabetes of the Young	HNF4A	Autosomal Dominant	NM_000457.4
Maturity Onset Diabetes of the Young	INS	Autosomal Dominant	NM_000207.2
Maturity Onset Diabetes of the Young	KLF11	Autosomal Dominant	NM_003597.4
Maturity Onset Diabetes of the Young	NEUROD1	Autosomal Dominant	NM_002500.4
Maturity Onset Diabetes of the Young	PAX4	Autosomal Dominant	NM_006193.2
May-Hegglin Anomaly	MYH9	Autosomal Dominant	NM_002473.4
MCAD Deficiency	ACADM	Autosomal Recessive	NM_000016.4
McKusick-Kaufman Syndrome	MKKS	Autosomal Recessive	NM_018848.3
Meacham Syndrome	WT1	Autosomal Dominant	NM_024426.4
Meckel Syndrome	B9D1	Autosomal Recessive	NM_015681.3
Meckel Syndrome	B9D2	Autosomal Recessive	NM_030578.3
Meckel Syndrome	CC2D2A	Autosomal Recessive	NM_001080522.2

Meckel Syndrome	CEP290	Autosomal Recessive	NM_025114.3
Meckel Syndrome	MKS1	Autosomal Recessive	NM_017777.3
Meckel Syndrome	NPHP3	Autosomal Recessive	NM_153240.4
Meckel Syndrome	RPGRIP1L	Autosomal Recessive	NM_015272.2
Meckel Syndrome	TCTN2	Autosomal Recessive	NM_024809.4
Meckel Syndrome	TMEM216	Autosomal Recessive	NM_001173990.2
Meckel Syndrome	TMEM67	Autosomal Recessive	NM_153704.5
Medulloblastoma	SUFU	Autosomal Dominant	NM_016169.3
Megalencephalic Leukoencephalopathy with Subcortical Cysts	HEPACAM	Autosomal Recessive	NM_152722.4
Megalencephalic Leukoencephalopathy with Subcortical Cysts	MLC1	Autosomal Recessive	NM_015166.3
Megaloblastic Anemia	CUBN	Autosomal Recessive	NM_001081.3
Meier-Gorlin Syndrome	CDC6	Autosomal Recessive	NM_001254.3
Meier-Gorlin Syndrome	ORC1	Autosomal Recessive	NM_004153.3
Meier-Gorlin Syndrome	ORC6	Autosomal Recessive	NM_014321.3
Mental Retardation with Language Impairment and Autistic Features	FOXP1	Autosomal Dominant	NM_032682.5
Mental Retardation, Dominant	DYNC1H1	Autosomal Dominant	NM_001376.4
Mental Retardation, Dominant	GRIN2B	Autosomal Dominant	NM_000834.3
Mental Retardation, Dominant	KIF1A	Autosomal Dominant	NM_004321.6
Mental Retardation, Dominant	MBD5	Autosomal Dominant	NM_018328.4
Mental Retardation, Recessive	MAN1B1	Autosomal Recessive	NM_016219.4
Mental Retardation, Recessive	NSUN2	Autosomal Recessive	NM_017755.5
Mental Retardation, Recessive	TRAPPC9	Autosomal Recessive	NM_031466.5
Mental Retardation, Stereotypic Movements, Epilepsy, and/or Cerebral Malformations	MEF2C	Autosomal Dominant	NM_002397.4
Mental Retardation, X-linked	ARHGEF6	X-Linked	NM_004840.2
Mental Retardation, X-linked	IL1RAPL1	X-Linked	NM_014271.3
Mental Retardation, X-linked	RAB39B	X-Linked	NM_171998.2
Mental Retardation, X-linked	ZNF81	X-Linked	NM_007137.3

Metachondromatosis	PTPN11	Autosomal Dominant	NM_002834.3
Metachromatic Leukodystrophy	PSAP	Autosomal Recessive	NM_002778.2
Metaphyseal Anadysplasia	MMP13	Autosomal Dominant	NM_002427.3
Metaphyseal Chondrodysplasia	COL10A1	Autosomal Dominant	NM_000493.3
Metaphyseal Chondrodysplasia	PTH1R	Autosomal Dominant	NM_000316.2
Metatropic Dysplasia	TRPV4	Autosomal Dominant	NM_021625.4
Methylmalonate Semialdehyde Dehydrogenase Deficiency	ALDH6A1	Autosomal Recessive	NM_005589.2
Methylmalonic Acidemia	MCEE	Autosomal Recessive	NM_032601.3
Methylmalonic Acidemia	MMAA	Autosomal Recessive	NM_172250.2
Methylmalonic Acidemia	MMAB	Autosomal Recessive	NM_052845.3
Methylmalonic Acidemia	MMADHC	Autosomal Recessive	NM_015702.2
Methylmalonic Acidemia	MUT	Autosomal Recessive	NM_000255.3
Mevalonicaciduria	MVK	Autosomal Recessive	NM_000431.2
MHC Class II Deficiency	RFXANK	Autosomal Recessive	NM_003721.2
Microcephalic Osteodysplastic Primordial Dwarfism	PCNT	Autosomal Recessive	NM_006031.5
Microcephalic Osteodysplastic Primordial Dwarfism	RNU4ATAC	Autosomal Recessive	NR_023343.1
Microcephaly, Cortical Malformations, and Mental Retardation	WDR62	Autosomal Recessive	NM_001083961.1
Microtia, Hearing Impairment, and Cleft Palate	HOXA2	Autosomal Recessive	NM_006735.3
Mitochondrial Complex V (ATP Synthase) Deficiency, Nuclear Type	ATPAF2	Autosomal Recessive	NM_145691.3
Mitochondrial Complex V (ATP Synthase) Deficiency, Nuclear Type	TMEM70	Autosomal Recessive	NM_017866.5
Mitochondrial DNA Depletion Syndrome	C10orf2	Autosomal Recessive	NM_021830.4
Mitochondrial DNA Depletion Syndrome	DGUOK	Autosomal Recessive	NM_080916.2
Mitochondrial DNA Depletion Syndrome	MPV17	Autosomal Recessive	NM_002437.4
Mitochondrial DNA Depletion Syndrome	RRM2B	Autosomal Recessive	NM_015713.4
Mitochondrial DNA Depletion Syndrome	SUCLA2	Autosomal Recessive	NM_003850.2
Mitochondrial DNA Depletion Syndrome	SUCLG1	Autosomal Recessive	NM_003849.3
Mitochondrial DNA Depletion Syndrome	TK2	Autosomal Recessive	NM_004614.4
Mitochondrial Membrane Protein-Associated Neurodegeneration	C19orf12	Autosomal Recessive	NM_001031726.3

Mitochondrial Neurogastrointestinal Encephalopathy Disease	TYMP	Autosomal Recessive	NM_001953.4
Mitochondrial Phosphate Carrier Deficiency	SLC25A3	Autosomal Recessive	NM_005888.3
Mitochondrial Respiratory Chain Complex I Deficiency	FOXRED1	Autosomal Recessive	NM_017547.3
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFA10	Autosomal Recessive	NM_004544.3
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFA11	Autosomal Recessive	NM_175614.4
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFA2	Autosomal Recessive	NM_002488.4
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFAF1	Autosomal Recessive	NM_016013.3
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFAF2	Autosomal Recessive	NM_174889.4
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFAF3	Autosomal Recessive	NM_199069.1
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFAF4	Autosomal Recessive	NM_014165.3
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS1	Autosomal Recessive	NM_005006.6
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS2	Autosomal Recessive	NM_004550.4
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS3	Autosomal Recessive	NM_004551.2
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS4	Autosomal Recessive	NM_002495.2
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS6	Autosomal Recessive	NM_004553.4
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS7	Autosomal Recessive	NM_024407.4
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS8	Autosomal Recessive	NM_002496.3
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFV1	Autosomal Recessive	NM_007103.3
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFV2	Autosomal Recessive	NM_021074.4
Mitochondrial Respiratory Chain Complex I Deficiency	NUBPL	Autosomal Recessive	NM_025152.2
Mitochondrial Respiratory Chain Complex II Deficiency	SDHAF1	Autosomal Recessive	NM_001042631.2
Mitochondrial Respiratory Chain Complex III Deficiency	BCS1L	Autosomal Recessive	NM_004328.4
Mitochondrial Respiratory Chain Complex III Deficiency	TTC19	Autosomal Recessive	NM_017775.3
Mitochondrial Respiratory Chain Complex III Deficiency	UQCRQ	Autosomal Recessive	NM_014402.4
Mitochondrial Respiratory Chain Complex IV Deficiency	COX10	Autosomal Recessive	NM_001303.3
Mitochondrial Respiratory Chain Complex IV Deficiency	COX15	Autosomal Recessive	NM_004376.5
Mitochondrial Respiratory Chain Complex IV Deficiency	COX6B1	Autosomal Recessive	NM_001863.4
Mitochondrial Respiratory Chain Complex IV Deficiency	FASTKD2	Autosomal Recessive	NM_014929.3

Mitochondrial Respiratory Chain Complex IV Deficiency	SCO1	Autosomal Recessive	NM_004589.2
Mitochondrial Respiratory Chain Complex IV Deficiency	SCO2	Autosomal Recessive	NM_005138.2
Miyoshi Muscular Dystrophy	ANO5	Autosomal Recessive	NM_213599.2
Molybdenum Cofactor Deficiency	MOCS1	Autosomal Recessive	NM_005943.5
Molybdenum Cofactor Deficiency	MOCS2	Autosomal Recessive	NM_176806.2
Monilethrix	KRT83	Autosomal Dominant	NM_002282.3
Monogenic Non-Syndromic Obesity	LEP	Autosomal Recessive	NM_000230.2
Monogenic Non-Syndromic Obesity	LEPR	Autosomal Recessive	NM_002303.5
Monogenic Non-Syndromic Obesity	PCSK1	Autosomal Recessive	NM_000439.4
Monogenic Non-Syndromic Obesity	POMC	Autosomal Recessive	NM_001035256.1
Mononeuropathy of the Median Nerve	SH3TC2	Autosomal Recessive	NM_024577.3
Mowat-Wilson Syndrome	ZEB2	Autosomal Dominant	NM_014795.3
Moyamoya Disease	ACTA2	Autosomal Dominant	NM_001613.2
MTHFR Deficiency	MTHFR	Autosomal Dominant	NM_005957.4
MTHFR Thermolabile Variant	MTHFR	Autosomal Dominant	NM_005957.4
Muckle-Wells Syndrome	NLRP3	Autosomal Dominant	NM_004895.4
Mucolipidosis, Type I	NEU1	Autosomal Recessive	NM_000434.3
Mucolipidosis, Type II	GNPTAB	Autosomal Recessive	NM_024312.4
Mucolipidosis, Type III	GNPTG	Autosomal Recessive	NM_032520.4
Mucolipidosis, Type IV	MCOLN1	Autosomal Recessive	NM_020533.2
Mucopolysaccharidosis, Type I	IDUA	Autosomal Recessive	NM_000203.3
Mucopolysaccharidosis, Type III	GNS	Autosomal Recessive	NM_002076.3
Mucopolysaccharidosis, Type III	HGSNAT	Autosomal Recessive	NM_152419.2
Mucopolysaccharidosis, Type III	NAGLU	Autosomal Recessive	NM_000263.3
Mucopolysaccharidosis, Type III	SGSH	Autosomal Recessive	NM_000199.3
Mucopolysaccharidosis, Type IV	GALNS	Autosomal Recessive	NM_000512.4
Mucopolysaccharidosis, Type IV	GLB1	Autosomal Recessive	NM_000404.2
Mucopolysaccharidosis, Type IX	HYAL1	Autosomal Recessive	NR_047690.1

Mucopolysaccharidosis, Type VI	ARSB	Autosomal Recessive	NM_000046.3
Mucopolysaccharidosis, Type VII	GUSB	Autosomal Recessive	NM_000181.3
Mulibrey Nanism	TRIM37	Autosomal Recessive	NM_015294.3
Multicentric Carpotarsal Osteolysis Syndrome	MAFB	Autosomal Dominant	NM_005461.3
Multicentric Osteolysis of Torg	MMP2	Autosomal Recessive	NM_004530.4
Multicentric Osteolysis, Nodulosis, and Arthropathy	MMP2	Autosomal Recessive	NM_004530.4
Multiminicore Disease	RYR1	Autosomal Recessive	NM_000540.2
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFA	Autosomal Recessive	NM_000126.3
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFDH	Autosomal Recessive	NM_004453.2
Multiple Cutaneous and Mucosal Venous Malformations	TEK	Autosomal Dominant	NM_000459.3
Multiple Cutaneous and Uterine Leiomyomas	FH	Autosomal Dominant	NM_000143.3
Multiple Endocrine Neoplasia	CDKN1B	Autosomal Dominant	NM_004064.3
Multiple Endocrine Neoplasia	MEN1	Autosomal Dominant	NM_130799.2
Multiple Endocrine Neoplasia	RET	Autosomal Dominant	NM_020975.4
Multiple Epiphyseal Dysplasia with Early-Onset Diabetes Mellitus	EIF2AK3	Autosomal Recessive	NM_004836.5
Multiple Epiphyseal Dysplasia, Dominant	COL9A1	Autosomal Dominant	NM_001851.4
Multiple Epiphyseal Dysplasia, Dominant	COL9A2	Autosomal Dominant	NM_001852.3
Multiple Epiphyseal Dysplasia, Dominant	COL9A3	Autosomal Dominant	NM_001853.3
Multiple Epiphyseal Dysplasia, Dominant	COMP	Autosomal Dominant	NM_000095.2
Multiple Epiphyseal Dysplasia, Dominant	MATN3	Autosomal Dominant	NM_002381.4
Multiple Epiphyseal Dysplasia, Recessive	SLC26A2	Autosomal Recessive	NM_000112.3
Multiple Familial Trichoepithelioma	CYLD	Autosomal Dominant	NM_015247.2
Multiple Mitochondrial Dysfunctions Syndrome	BOLA3	Autosomal Recessive	NM_212552.2
Multiple Mitochondrial Dysfunctions Syndrome	NFU1	Autosomal Recessive	NM_001002755.2
Multiple Pterygium Syndrome	CHRNA1	Autosomal Recessive	NM_000079.3
Multiple Pterygium Syndrome	CHRND	Autosomal Recessive	NM_000751.2
Multiple Pterygium Syndrome	CHRNG	Autosomal Recessive	NM_005199.4
Multiple Sulfatase Deficiency	SUMF1	Autosomal Recessive	NM_182760.3

Multiple Synostoses Syndrome	FGF9	Autosomal Dominant	NM_002010.2
Multiple Synostoses Syndrome	GDF5	Autosomal Dominant	NM_000557.2
Multisystemic Smooth Muscle Dysfunction Syndrome	ACTA2	Autosomal Dominant	NM_001613.2
MYH9-related disorder	MYH9	Autosomal Dominant	NM_002473.4
MYH-Associated Polyposis	MUTYH	Autosomal Recessive	NM_001048171.1
Myhre Syndrome	SMAD4	Autosomal Dominant	NM_005359.5
Myoadenylate Deaminase Deficiency	AMPD1	Autosomal Recessive	NM_000036.2
Myoclonus-Dystonia	SGCE	Autosomal Dominant	NM_003919.2
Myofibrillar Myopathy, Dominant	BAG3	Autosomal Dominant	NM_004281.3
Myofibrillar Myopathy, Dominant	DNAJB6	Autosomal Dominant	NM_058246.3
Myofibrillar Myopathy, Dominant	LDB3	Autosomal Dominant	NM_001080116.1
Myofibrillar Myopathy, Dominant	MYOT	Autosomal Dominant	NM_006790.2
Myofibrillar Myopathy, Mixed	DES	Mixed	NM_001927.3
Myofibrillar Myopathy, Recessive	CRYAB	Autosomal Recessive	NM_001885.1
Myokymia	KCNA1	Autosomal Dominant	NM_000217.2
Myopathy, Early-Onset, Areflexia, Respiratory Distress, and Dysphagia	MEGF10	Autosomal Recessive	NM_032446.2
Myopathy, Lactic Acidosis, and Sideroblastic Anemia	YARS2	Autosomal Recessive	NM_001040436.2
Myosclerosis	COL6A2	Autosomal Recessive	NM_001849.3
Myosin Storage Myopathy	MYH7	Autosomal Dominant	NM_000257.2
Myostatin-Related Muscle Hypertrophy	MSTN	Autosomal Recessive	NM_005259.2
Myotonia Congenita	CLCN1	Autosomal Dominant	NM_000083.2
N-Acetylglutamate Synthase Deficiency	NAGS	Autosomal Recessive	NM_153006.2
Nager Syndrome	SF3B4	Mixed	NM_005850.4
Nail-Patella Syndrome	LMX1B	Autosomal Dominant	NM_002316.3
Natural Killer Cell and Glucocorticoid Deficiency with DNA Repair Defect	MCM4	Autosomal Recessive	NM_005914.3
Naxos Disease	JUP	Autosomal Recessive	NM_002230.2
Nemaline Myopathy	TNNT1	Mixed	NM_003283.4
Nemaline Myopathy	TPM2	Mixed	NM_003289.3

Nemaline Myopathy	TPM3	Mixed	NM_152263.2
Nemaline Myopathy, Dominant	ACTA1	Autosomal Dominant	NM_001100.3
Nemaline Myopathy, Dominant	KBTBD13	Autosomal Dominant	NM_001101362.2
Nemaline Myopathy, Recessive	CFL2	Autosomal Recessive	NM_021914.7
Nemaline Myopathy, Recessive	NEB	Autosomal Recessive	NM_004543.4
Neonatal Severe Primary Hyperparathyroidism	CASR	Autosomal Recessive	NM_000388.3
Nephrogenic Diabetes Insipidus	AQP2	Autosomal Dominant	NM_000486.5
Nephronophthisis	GLIS2	Autosomal Recessive	NM_032575.2
Nephronophthisis	INVS	Autosomal Recessive	NM_014425.3
Nephronophthisis	NEK8	Autosomal Recessive	NM_178170.2
Nephronophthisis	NPHP1	Autosomal Recessive	NM_000272.3
Nephronophthisis	NPHP3	Autosomal Recessive	NM_153240.4
Nephronophthisis	NPHP4	Autosomal Recessive	NM_015102.3
Nephronophthisis	RPGRIP1L	Autosomal Recessive	NM_015272.2
Nephronophthisis	TMEM67	Autosomal Recessive	NM_153704.5
Nephronophthisis-Like Nephropathy	XPNPEP3	Autosomal Recessive	NM_022098.3
Nephropathic Cystinosis	CTNS	Autosomal Recessive	NM_004937.2
Nephrotic Syndrome	LAMB2	Autosomal Recessive	NM_002292.3
Nephrotic Syndrome	PLCE1	Autosomal Recessive	NM_016341.3
Nestor-Guillermo Progeria Syndrome	BANF1	Autosomal Recessive	NM_001143985.1
Netherton Syndrome	SPINK5	Autosomal Recessive	NM_006846.3
Neural Tube Defect	VANGL1	Autosomal Dominant	NM_138959.2
Neural Tube Defects, Folate-Sensitive	MTHFR	Autosomal Dominant	NM_005957.4
Neuroblastoma	KIF1B	Autosomal Dominant	NM_015074.3
Neuroblastoma	PHOX2B	Autosomal Dominant	NM_003924.3
Neuroblastoma Susceptibility	ALK	Autosomal Dominant	NM_004304.4
Neurodegeneration	PLA2G6	Autosomal Recessive	NM_003560.2
Neurodegeneration due to Cerebral Folate Transport Deficiency	FOLR1	Autosomal Recessive	NM_016725.2

Neuroferritinopathy	FTL	Autosomal Dominant	NM_000146.3
Neurofibromatosis, Type 1	NF1	Autosomal Dominant	NM_000267.3
Neurofibromatosis, Type 2	NF2	Autosomal Dominant	NM_000268.3
Neurofibromatosis-Noonan Syndrome	NF1	Autosomal Dominant	NM_000267.3
Neurogenic Scapuloperoneal Syndrome	DES	Autosomal Dominant	NM_001927.3
Neurologic Disorders/Seipinopathy	BSCL2	Autosomal Dominant	NM_032667.6
Neuronal Ceroid-Lipofuscinosis, Dominant/Recessive	CLN3	Mixed	NM_001042432.1
Neuronal Ceroid-Lipofuscinosis, Dominant/Recessive	CLN5	Mixed	NM_006493.2
Neuronal Ceroid-Lipofuscinosis, Dominant/Recessive	CTSD	Mixed	NM_001909.4
Neuronal Ceroid-Lipofuscinosis, Recessive	CLN6	Autosomal Recessive	NM_017882.2
Neuronal Ceroid-Lipofuscinosis, Recessive	DNAJC5	Autosomal Recessive	NM_025219.2
Neuronal Ceroid-Lipofuscinosis, Recessive	MFSD8	Autosomal Recessive	NM_152778.2
Neuronal Ceroid-Lipofuscinosis, Recessive	PPT1	Autosomal Recessive	NM_000310.3
Neuronal Ceroid-Lipofuscinosis, Recessive	TPP1	Autosomal Recessive	NM_000391.3
Neutral Lipid Storage Disease with Myopathy	PNPLA2	Autosomal Recessive	NM_020376.3
Nevoid Basal Cell Carcinoma Syndrome	PTCH1	Autosomal Dominant	NM_000264.3
Newfoundland Rod-Cone Dystrophy	RLBP1	Autosomal Recessive	NM_000326.4
Nicolaides-Baraitser Syndrome	SMARCA2	Autosomal Dominant	NM_003070.3
Nicolaides-Baraitser Syndrome	SMARCA2	Autosomal Dominant	NM_003070.3
Niemann-Pick Disease	NPC1	Autosomal Recessive	NM_000271.4
Niemann-Pick Disease	NPC2	Autosomal Recessive	NM_006432.3
Nijmegen Breakage Syndrome	NBN	Autosomal Recessive	NM_002485.4
Nocturnal Frontal Lobe Epilepsy	CHRNA2	Autosomal Dominant	NM_000742.3
Nonaka Myopathy	GNE	Autosomal Recessive	NM_005476.5
Nonautoimmune Hyperthyroidism	TSHR	Autosomal Dominant	NM_000369.2
Non-Classic Cystic Fibrosis-Like Syndrome	SCNN1B	Autosomal Dominant	NM_000336.2
Nonepidermolytic Palmoplantar Hyperkeratosis	KRT1	Autosomal Dominant	NM_006121.3
Nonspecific Cardiac Conduction Defect	SCN1B	Autosomal Dominant	NM_001037.4

Nonsyndromic Hearing Loss, Dominant	COCH	Autosomal Dominant	NM_004086.2
Nonsyndromic Hearing Loss, Dominant	COL11A2	Autosomal Dominant	NM_080680.2
Nonsyndromic Hearing Loss, Dominant	EYA4	Autosomal Dominant	NM_004100.4
Nonsyndromic Hearing Loss, Dominant	GJB3	Autosomal Dominant	NM_024009.2
Nonsyndromic Hearing Loss, Dominant	MYH14	Autosomal Dominant	NM_024729.3
Nonsyndromic Hearing Loss, Dominant	MYH9	Autosomal Dominant	NM_002473.4
Nonsyndromic Hearing Loss, Dominant	MYO1A	Autosomal Dominant	NM_005379.2
Nonsyndromic Hearing Loss, Dominant	POU4F3	Autosomal Dominant	NM_002700.2
Nonsyndromic Hearing Loss, Dominant	SIX1	Autosomal Dominant	NM_005982.3
Nonsyndromic Hearing Loss, Dominant	SLC17A8	Autosomal Dominant	NM_139319.2
Nonsyndromic Hearing Loss, Dominant	TJP2	Autosomal Dominant	NM_004817.3
Nonsyndromic Hearing Loss, Dominant	WFS1	Autosomal Dominant	NM_006005.3
Nonsyndromic Hearing Loss, Mixed	DFNA5	Mixed	NM_004403.2
Nonsyndromic Hearing Loss, Mixed	DIAPH1	Mixed	NM_005219.4
Nonsyndromic Hearing Loss, Mixed	FOXI1	Mixed	NM_012188.4
Nonsyndromic Hearing Loss, Mixed	HGF	Mixed	NM_000601.4
Nonsyndromic Hearing Loss, Mixed	KCNJ10	Mixed	NM_002241.4
Nonsyndromic Hearing Loss, Mixed	MIR96	Mixed	NR_029512.1
Nonsyndromic Hearing Loss, Recessive	CDH23	Autosomal Recessive	NM_022124.5
Nonsyndromic Hearing Loss, Recessive	CLDN14	Autosomal Recessive	NM_144492.2
Nonsyndromic Hearing Loss, Recessive	DFNB31	Autosomal Recessive	NM_015404.3
Nonsyndromic Hearing Loss, Recessive	DFNB59	Autosomal Recessive	NM_001042702.3
Nonsyndromic Hearing Loss, Recessive	ESRRB	Autosomal Recessive	NM_004452.3
Nonsyndromic Hearing Loss, Recessive	GJB2	Autosomal Recessive	NM_004004.5
Nonsyndromic Hearing Loss, Recessive	GJB6	Autosomal Recessive	NM_006783.4
Nonsyndromic Hearing Loss, Recessive	GPSM2	Autosomal Recessive	NM_013296.4
Nonsyndromic Hearing Loss, Recessive	GRXCR1	Autosomal Recessive	NM_001080476.2
Nonsyndromic Hearing Loss, Recessive	LHFPL5	Autosomal Recessive	NM_182548.3

Nonsyndromic Hearing Loss, Recessive	LOXHD1	Autosomal Recessive	NM_144612.6
Nonsyndromic Hearing Loss, Recessive	LRTOMT	Autosomal Recessive	NM_001145308.2
Nonsyndromic Hearing Loss, Recessive	MARVELD2	Autosomal Recessive	NM_001038603.2
Nonsyndromic Hearing Loss, Recessive	MYO15A	Autosomal Recessive	NM_016239.3
Nonsyndromic Hearing Loss, Recessive	MYO3A	Autosomal Recessive	NM_017433.4
Nonsyndromic Hearing Loss, Recessive	MYO6	Autosomal Recessive	NM_004999.3
Nonsyndromic Hearing Loss, Recessive	MYO7A	Autosomal Recessive	NM_000260.3
Nonsyndromic Hearing Loss, Recessive	OTOF	Autosomal Recessive	NM_194248.2
Nonsyndromic Hearing Loss, Recessive	PCDH15	Autosomal Recessive	NM_033056.3
Nonsyndromic Hearing Loss, Recessive	RDX	Autosomal Recessive	NM_002906.3
Nonsyndromic Hearing Loss, Recessive	SLC26A4	Autosomal Recessive	NM_000441.1
Nonsyndromic Hearing Loss, Recessive	SLC26A5	Autosomal Recessive	NM_198999.2
Nonsyndromic Hearing Loss, Recessive	TECTA	Autosomal Recessive	NM_005422.2
Nonsyndromic Hearing Loss, Recessive	TMC1	Autosomal Recessive	NM_138691.2
Nonsyndromic Hearing Loss, Recessive	TMIE	Autosomal Recessive	NM_147196.2
Nonsyndromic Hearing Loss, Recessive	TMPRSS3	Autosomal Recessive	NM_024022.2
Nonsyndromic Hearing Loss, Recessive	USH1C	Autosomal Recessive	NM_005709.3
Nonsyndromic Hearing Loss, X-Linked	POU3F4	X-Linked	NM_000307.3
Nonsyndromic Hearing Loss, X-Linked	PRPS1	X-Linked	NM_002764.3
Nonsyndromic Hearing Loss, X-Linked	SMPX	X-Linked	NM_014332.2
Nonsyndromic Trigonocephaly	FGFR1	Autosomal Dominant	NM_023110.2
Noonan Syndrome	BRAF	Autosomal Dominant	NM_004333.4
Noonan Syndrome	KRAS	Autosomal Dominant	NM_004985.3
Noonan Syndrome	MAP2K1	Autosomal Dominant	NM_002755.3
Noonan Syndrome	NRAS	Autosomal Dominant	NM_002524.4
Noonan Syndrome	PTPN11	Autosomal Dominant	NM_002834.3
Noonan Syndrome	RAF1	Autosomal Dominant	NM_002880.3
Noonan Syndrome	SOS1	Autosomal Dominant	NM_005633.3

Noonan-Like Syndrome Disorder	CBL	Autosomal Dominant	NM_005188.3
Noonan-Like Syndrome with Loose Anagen Hair	SHOC2	Autosomal Dominant	NM_007373.3
Norepinephrine Transporter Deficiency	SLC6A2	Autosomal Dominant	NM_001043.3
Normokalemic Periodic Paralysis	SCN4A	Autosomal Dominant	NM_000334.4
North American Indian Childhood Cirrhosis	CIRH1A	Autosomal Recessive	NM_032830.2
Obesity	MC4R	Autosomal Dominant	NM_005912.2
Obesity	PPARG	Autosomal Dominant	NM_015869.4
Occult Macular Dystrophy	RP1L1	Autosomal Dominant	NM_178857.5
Oculocutaneous Albinism	OCA2	Autosomal Recessive	NM_000275.2
Oculocutaneous Albinism	SLC45A2	Autosomal Recessive	NM_016180.3
Oculocutaneous Albinism	TYRP1	Autosomal Recessive	NM_000550.2
Oculodentodigital Dysplasia	GJA1	Autosomal Dominant	NM_000165.3
Odontoonychodermal Dysplasia	WNT10A	Autosomal Recessive	NM_025216.2
Oguchi Disease	GRK1	Autosomal Recessive	NM_002929.2
Oguchi Disease	SAG	Autosomal Recessive	NM_000541.4
Oligodontia-Colorectal Cancer Syndrome	AXIN2	Autosomal Dominant	NM_004655.3
Omenn Syndrome	DCLRE1C	Autosomal Recessive	NM_001033855.1
Omenn Syndrome	RAG1	Autosomal Recessive	NM_000448.2
Omenn Syndrome	RAG2	Autosomal Recessive	NM_000536.3
Omodysplasia	GPC6	Autosomal Dominant	NM_005708.3
Optic Atrophy, Dominant	OPA3	Autosomal Dominant	NM_025136.3
Optic Atrophy, Recessive	OPA1	Autosomal Dominant	NM_015560.2
Optic Atrophy, Recessive	TMEM126A	Autosomal Dominant	NM_032273.3
Ornithine Aminotransferase Deficiency	OAT	Autosomal Recessive	NM_000274.3
Ornithine Transcarbamylase Deficiency	OTC	X-Linked	NM_000531.5
Orofacial Cleft	BMP4	Autosomal Dominant	NM_001202.3
Orotic Aciduria	UMPS	Autosomal Recessive	NM_000373.3
Osteogenesis Imperfecta, Dominant	COL1A1	Autosomal Dominant	NM_000088.3

Osteogenesis Imperfecta, Dominant	COL1A2	Autosomal Dominant	NM_000089.3
Osteogenesis Imperfecta, Recessive	CRTAP	Autosomal Recessive	NM_006371.4
Osteogenesis Imperfecta, Recessive	FKBP10	Autosomal Recessive	NM_021939.3
Osteogenesis Imperfecta, Recessive	LEPRE1	Autosomal Recessive	NM_022356.3
Osteogenesis Imperfecta, Recessive	PPIB	Autosomal Recessive	NM_000942.4
Osteogenesis Imperfecta, Recessive	SERPINF1	Autosomal Recessive	NM_002615.5
Osteogenesis Imperfecta, Recessive	SERPINH1	Autosomal Recessive	NM_001235.3
Osteoglophonic Dysplasia	FGFR1	Autosomal Dominant	NM_023110.2
Osteopetrosis	CLCN7	Mixed	NM_001287.5
Osteopetrosis	OSTM1	Mixed	NM_014028.3
Osteopetrosis	TCIRG1	Mixed	NM_006019.3
Osteopetrosis	TNFRSF11A	Mixed	NM_003839.3
Osteopetrosis	TNFSF11	Mixed	NM_003701.3
Osteopetrosis with Renal Tubular Acidosis	CA2	Autosomal Recessive	NM_000067.2
Otofaciocervical Syndrome	EYA1	Autosomal Dominant	NM_000503.4
Otospondylomegaepiphyseal Dysplasia	COL11A2	Autosomal Recessive	NM_080680.2
OTX2-Related Syndromic Microphthalmia	OTX2	Autosomal Dominant	NM_172337.2
Ovarian Dysgenesis	BMP15	X-Linked	NM_005448.2
Ovarian Dysgenesis	FSHR	X-Linked	NM_000145.3
Ovarian Hyperstimulation Syndrome	FSHR	Autosomal Dominant	NM_000145.3
Paget Disease of Bone	SQSTM1	Autosomal Recessive	NM_003900.4
Paget Disease of Bone	TNFRSF11A	Autosomal Recessive	NM_003839.3
PALB2-Related Cancer Susceptibility	PALB2	Autosomal Dominant	NM_024675.3
Pallister-Hall Syndrome	GLI3	Autosomal Dominant	NM_000168.5
Palmoplantar Keratoderma, Mutilating, with Periorificial Keratotic Plaques	TRPV3	Autosomal Dominant	NM_145068.3
Pancreatic Cancer	PALLD	Autosomal Dominant	NM_001166108.1
Pantothenate Kinase-Associated Neurodegeneration	PANK2	Autosomal Recessive	NM_153638.2
Papillary Renal Carcinoma	MET	Autosomal Dominant	NM_001127500.1

Papillon-Lefevre Syndrome	CTSC	Autosomal Recessive	NM_001814.4
Paraganglioma and Gastric Stromal Sarcoma	SDHB	Autosomal Dominant	NM_003000.2
Paramyotonia Congenita of Von Eulenburg	SCN4A	Autosomal Dominant	NM_000334.4
Parathyroid Carcinoma	CDC73	Autosomal Dominant	NM_024529.4
Parkes Weber Syndrome	RASA1	Autosomal Dominant	NM_002890.2
Parkinson Disease ,Recessive	FBXO7	Autosomal Recessive	NM_012179.3
Parkinson Disease, Dominant	HTRA2	Autosomal Dominant	NM_013247.4
Parkinson Disease, Dominant	LRRK2	Autosomal Dominant	NM_198578.3
Parkinson Disease, Dominant	SNCA	Autosomal Dominant	NM_000345.3
Parkinson Disease, Dominant	VPS35	Autosomal Dominant	NM_018206.4
Parkinson Disease, Dominant/Recessive	MAPT	Mixed	NM_005910.5
Parkinson Disease, Dominant/Recessive	NR4A2	Mixed	NM_006186.3
Parkinson Disease, Dominant/Recessive	SNCAIP	Mixed	NM_005460.2
Parkinson Disease, Dominant/Recessive	UCHL1	Mixed	NM_004181.4
Parkinson Disease, Juvenile	PARK2	Mixed	NM_004562.2
Parkinson Disease, Recessive	PARK7	Autosomal Recessive	NM_007262.4
Parkinson Disease, Recessive	PINK1	Autosomal Recessive	NM_032409.2
Paroxysmal Extreme Pain Disorder	SCN9A	Autosomal Dominant	NM_002977.3
Paroxysmal Familial Ventricular Fibrillation	SCN5A	Autosomal Dominant	NM_198056.2
Partial Epilepsy with Auditory Features	LGI1	Autosomal Dominant	NM_005097.2
Partial Isolated Growth Hormone Deficiency	GHSR	Autosomal Dominant	NM_198407.2
Patterned Dystrophy of Retinal Pigment Epithelium	PRPH2	Autosomal Dominant	NM_000322.4
PCWH Syndrome	SOX10	Autosomal Dominant	NM_006941.3
Peeling Skin Syndrome	TGM5	Autosomal Recessive	NM_201631.3
Pendred Syndrome	FOXI1	Autosomal Recessive	NM_012188.4
Pendred Syndrome	KCNJ10	Autosomal Recessive	NM_002241.4
Pendred Syndrome	SLC26A4	Autosomal Recessive	NM_000441.1
Periventricular Heterotopia	ARFGEF2	Autosomal Recessive	NM_006420.2

Perlman Syndrome	DIS3L2	Autosomal Recessive	NM_152383.4
Permanent Neonatal Diabetes Mellitus	ABCC8	Mixed	NM_000352.3
Permanent Neonatal Diabetes Mellitus	GCK	Mixed	NM_000162.3
Peroxisomal Bifunctional Enzyme Deficiency	HSD17B4	Autosomal Recessive	NM_000414.3
Perrault Syndrome	HSD17B4	Autosomal Recessive	NM_000414.3
Perry Syndrome	DCTN1	Autosomal Dominant	NM_004082.4
Peters Anomaly	CYP1B1	Autosomal Dominant	NM_000104.3
Peters Anomaly	PAX6	Autosomal Dominant	NM_000280.4
Peters Anomaly	PITX2	Autosomal Dominant	NM_153427.2
Peters Plus Syndrome	B3GALTL	Autosomal Recessive	NM_194318.3
Peutz-Jeghers Syndrome	STK11	Autosomal Dominant	NM_000455.4
Pfeiffer Syndrome	FGFR1	Autosomal Dominant	NM_023110.2
Pfeiffer Syndrome	FGFR2	Autosomal Dominant	NM_000141.4
Phenylalanine Hydroxylase Deficiency	PAH	Autosomal Recessive	NM_000277.1
Pheochromocytoma	KIF1B	Autosomal Dominant	NM_015074.3
Pheochromocytoma	RET	Autosomal Dominant	NM_020975.4
Pheochromocytoma	TMEM127	Autosomal Dominant	NM_017849.3
Phosphoenolpyruvate Carboxykinase Deficiency	PCK1	Autosomal Recessive	NM_002591.3
Phosphoglycerate Dehydrogenase Deficiency	PHGDH	Autosomal Recessive	NM_006623.3
Phosphoribosylpyrophosphate Synthetase Superactivity	PRPS1	X-Linked	NM_002764.3
Phosphorylase Kinase Deficiency	PHKB	Autosomal Recessive	NM_000293.2
Phosphorylase Kinase Deficiency	PHKG2	Autosomal Recessive	NM_000294.2
Phosphoserine Aminotransferase Deficiency	PSAT1	Autosomal Recessive	NM_058179.2
Phosphoserine Phosphatase Deficiency	PSPH	Autosomal Recessive	NM_004577.3
Piebald Trait	KIT	Autosomal Dominant	NM_000222.2
Piebald Trait	SNAI2	Autosomal Dominant	NM_003068.4
Pierson Syndrome	LAMB2	Autosomal Recessive	NM_002292.3
Pigmented Paravenous Chorioretinal Atrophy	CRB1	Autosomal Dominant	NM_201253.2

Pineal Hyperplasia, Insulin-Resistant Diabetes Mellitus, and Somatic Abnormalities	INSR	Autosomal Dominant	NM_000208.2
Pitt-Hopkins Syndrome	TCF4	Autosomal Dominant	NM_001083962.1
Pitt-Hopkins-Like Syndrome	CNTNAP2	Autosomal Recessive	NM_014141.5
Pitt-Hopkins-Like Syndrome	NRXN1	Autosomal Recessive	NM_001135659.1
Pituitary Dwarfism II	GHR	Autosomal Recessive	NM_000163.4
PITX2-Related Eye Abnormalities	PITX2	Autosomal Dominant	NM_153427.2
Plasminogen Activator Inhibitor-1 Deficiency	SERPINE1	Autosomal Dominant	NM_000602.3
Platelet Glycoprotein IV Deficiency	CD36	Autosomal Recessive	NM_001001547.2
Pleuropulmonary Blastoma	DICER1	Autosomal Dominant	NM_177438.2
Pol III-Related Leukodystrophy	POLR3A	Autosomal Recessive	NM_007055.3
Pol III-Related Leukodystrophy	POLR3B	Autosomal Recessive	NM_018082.5
POLG-Related Spectrum Disorders	POLG	Autosomal Recessive	NM_002693.2
Polycystic Kidney Disease, Autosomal Dominant	PKD2	Autosomal Dominant	NM_000297.3
Polycystic Kidney Disease, Autosomal Recessive	PKHD1	Autosomal Recessive	NM_138694.3
Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy	TREM2	Autosomal Recessive	NM_018965.3
Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy	TYROBP	Autosomal Recessive	NM_003332.3
Polycystic Liver Disease	PRKCSH	Autosomal Dominant	NM_002743.2
Polycystic Liver Disease	SEC63	Autosomal Dominant	NM_007214.4
Polydactyly	GLI3	Autosomal Dominant	NM_000168.5
Polymicrogyria	GPR56	Autosomal Recessive	NM_005682.5
Pontocerebellar Hypoplasia	RARS2	Autosomal Recessive	NM_020320.3
Pontocerebellar Hypoplasia	TSEN2	Autosomal Recessive	NM_025265.3
Pontocerebellar Hypoplasia	TSEN34	Autosomal Recessive	NM_024075.3
Pontocerebellar Hypoplasia	TSEN54	Autosomal Recessive	NM_207346.2
Pontocerebellar Hypoplasia	VRK1	Autosomal Recessive	NM_003384.2

Popliteal Pterygium Syndrome	IRF6	Autosomal Dominant	NM_006147.3
Porencephaly	COL4A1	Autosomal Dominant	NM_001845.4
Porencephaly	COL4A2	Autosomal Dominant	NM_001846.2
Porphyria Cutanea Tarda	UROD	Autosomal Dominant	NM_000374.4
Postaxial Acrofacial Dysostosis	DHODH	Autosomal Recessive	NM_001361.4
Posterior Column Ataxia with Retinitis Pigmentosa	FLVCR1	Autosomal Recessive	NM_014053.3
Posterior Polar Cataract	CRYAB	Autosomal Dominant	NM_001885.1
Posterior Polymorphous Corneal Dystrophy	VSX1	Autosomal Dominant	NM_014588.5
Potassium-Aggravated Myotonia	SCN4A	Autosomal Dominant	NM_000334.4
Prader-Willi-Like Syndrome	SIM1	Autosomal Dominant	NM_005068.2
Premature Ovarian Failure	FIGLA	Autosomal Dominant	NM_001004311.3
Premature Ovarian Failure	NOBOX	Autosomal Dominant	NM_001080413.3
Primary Ciliary Dyskinesia	CCDC103	Autosomal Recessive	NM_213607.2
Primary Ciliary Dyskinesia	CCDC39	Autosomal Recessive	NM_181426.1
Primary Ciliary Dyskinesia	CCDC40	Autosomal Recessive	NM_017950.3
Primary Ciliary Dyskinesia	DNAAF1	Autosomal Recessive	NM_178452.4
Primary Ciliary Dyskinesia	DNAAF2	Autosomal Recessive	NM_018139.2
Primary Ciliary Dyskinesia	DNAAF3	Autosomal Recessive	NM_001256714.1
Primary Ciliary Dyskinesia	DNAH11	Autosomal Recessive	NM_003777.3
Primary Ciliary Dyskinesia	DNAH5	Autosomal Recessive	NM_001369.2
Primary Ciliary Dyskinesia	DNAI1	Autosomal Recessive	NM_012144.2
Primary Ciliary Dyskinesia	DNAI2	Autosomal Recessive	NM_023036.4
Primary Ciliary Dyskinesia	DNAL1	Autosomal Recessive	NM_031427.3
Primary Ciliary Dyskinesia	RSPH4A	Autosomal Recessive	NM_001010892.2
Primary Coenzyme Q10 Deficiency	COQ2	Autosomal Recessive	NM_015697.7
Primary Coenzyme Q10 Deficiency	COQ9	Autosomal Recessive	NM_020312.3
Primary Coenzyme Q10 Deficiency	PDSS1	Autosomal Recessive	NM_014317.3
Primary Coenzyme Q10 Deficiency	PDSS2	Autosomal Recessive	NM_020381.3

Primary Congenital Glaucoma	CYP1B1	Autosomal Recessive	NM_000104.3
Primary Congenital Glaucoma	LTBP2	Autosomal Recessive	NM_000428.2
Primary Hyperoxaluria	AGXT	Autosomal Recessive	NM_000030.2
Primary Hyperoxaluria	GRHPR	Autosomal Recessive	NM_012203.1
Primary Hyperoxaluria	HOGA1	Autosomal Recessive	NM_138413.3
Primary Hypertrophic Osteoarthropathy	HPGD	Autosomal Recessive	NM_000860.5
Primary Hypomagnesemia	CLDN16	Autosomal Recessive	NM_006580.3
Primary Lymphedema with Myelodysplasia	GATA2	Autosomal Dominant	NM_032638.4
Primary Microcephaly, Recessive	ASPM	Autosomal Recessive	NM_018136.4
Primary Microcephaly, Recessive	CDK5RAP2	Autosomal Recessive	NM_018249.5
Primary Microcephaly, Recessive	CENPJ	Autosomal Recessive	NM_018451.4
Primary Microcephaly, Recessive	CEP152	Autosomal Recessive	NM_014985.3
Primary Microcephaly, Recessive	MCPH1	Autosomal Recessive	NM_024596.3
Primary Microcephaly, Recessive	STIL	Autosomal Recessive	NM_003035.2
Primary Microcephaly, Recessive	WDR62	Autosomal Recessive	NM_001083961.1
Primary Open Angle Glaucoma	MYOC	Autosomal Dominant	NM_000261.1
Primary Open Angle Glaucoma	OPTN	Autosomal Dominant	NM_021980.4
Primary Open Angle Glaucoma	WDR36	Autosomal Dominant	NM_139281.2
Progeroid Laminopathies	LMNA	Mixed	NM_005572.3
Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	C10orf2	Autosomal Dominant	NM_021830.4
Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	POLG2	Autosomal Dominant	NM_007215.3
Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	RRM2B	Autosomal Dominant	NM_015713.4
Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	SLC25A4	Autosomal Dominant	NM_001151.3
Progressive Familial Heart Block	SCN5A	Autosomal Dominant	NM_198056.2
Progressive Familial Heart Block	TRPM4	Autosomal Dominant	NM_017636.3
Progressive Familial Intrahepatic Cholestasis	ABCB4	Autosomal Dominant	NM_000443.3
Progressive Myoclonic Epilepsy	GOSR2	Autosomal Recessive	NM_004287.3
Progressive Myoclonic Epilepsy	KCTD7	Autosomal Recessive	NM_153033.4

Progressive Myoclonic Epilepsy	PRICKLE2	Autosomal Recessive	NM_198859.3
Progressive Myoclonus Epilepsy with Ataxia	PRICKLE1	Autosomal Recessive	NM_153026.2
Progressive Myoclonus Epilepsy, Lafora type	NHLRC1	Autosomal Recessive	NM_198586.2
Progressive Pseudorheumatoid Arthropathy of Childhood	WISP3	Autosomal Recessive	NM_003880.3
Prolidase Deficiency	PEPD	Autosomal Recessive	NM_000285.3
Proliferative Vasculopathy And Hydranencephaly-Hydrocephaly Syndrome	FLVCR2	Autosomal Recessive	NM_017791.2
Proopiomelanocortin Deficiency	POMC	Autosomal Recessive	NM_001035256.1
Propionic Acidemia	PCCA	Autosomal Recessive	NM_000282.3
Propionic Acidemia	PCCB	Autosomal Recessive	NM_000532.4
Proprotein Convertase-1 Deficiency	PCSK1	Autosomal Recessive	NM_000439.4
Protein C Deficiency	PROC	Autosomal Dominant	NM_000312.3
Protein S Deficiency	PROS1	Autosomal Dominant	NM_000313.3
Prothrombin Deficiency	F2	Autosomal Recessive	NM_000506.3
Prothrombin-Related Thrombophilia	F2	Autosomal Dominant	NM_000506.3
Proximal Renal Tubular Acidosis with Ocular Abnormalities	SLC4A4	Autosomal Recessive	NM_003759.3
Pseudoachondroplasia	COMP	Autosomal Dominant	NM_000095.2
Pseudohypoaldosteronism, Type I, Dominant	CUL3	Autosomal Dominant	NM_003590.4
Pseudohypoaldosteronism, Type I, Dominant	KLHL3	Autosomal Dominant	NM_017415.2
Pseudohypoaldosteronism, Type I, Dominant	NR3C2	Autosomal Dominant	NM_000901.4
Pseudohypoaldosteronism, Type I, Dominant	STX16	Autosomal Dominant	NM_001001433.2
Pseudohypoaldosteronism, Type I, Recessive	SCNN1A	Autosomal Recessive	NM_001038.5
Pseudohypoaldosteronism, Type I, Recessive	SCNN1B	Autosomal Recessive	NM_000336.2
Pseudohypoaldosteronism, Type II	WNK1	Autosomal Dominant	NM_018979.3
Pseudohypoaldosteronism, Type II	WNK4	Autosomal Dominant	NM_032387.4
Pseudoinflammatory Fundus Dystrophy	TIMP3	Autosomal Recessive	NM_000362.4
Pseudoneonatal Adrenoleukodystrophy	ACOX1	Autosomal Recessive	NM_004035.6
Pulmonary Surfactant Metabolism Dysfunction, Dominant	SFTPC	Autosomal Dominant	NM_003018.3
Pulmonary Surfactant Metabolism Dysfunction, Recessive	ABCA3	Autosomal Recessive	NM_001089.2

Pulmonary Surfactant Metabolism Dysfunction, Recessive	SFTPB	Autosomal Recessive	NM_198843.2
Purine Nucleoside Phosphorylase Deficiency	PNP	Autosomal Recessive	NM_000270.3
Pycnodysostosis	CTSK	Autosomal Recessive	NM_000396.3
Pyogenic Sterile Arthritis, Pyoderma Gangrenosum, and Acne	PSTPIP1	Autosomal Dominant	NM_003978.3
Pyridoxamine 5-Prime-Phosphate Oxidase Deficiency	PNPO	Autosomal Recessive	NM_018129.3
Pyridoxine-Dependent Epilepsy	ALDH7A1	Autosomal Recessive	NM_001182.4
Pyridoxine-Refractory Sideroblastic Anemia	SLC25A38	Autosomal Recessive	NM_017875.2
Pyruvate Carboxylase Deficiency	PC	Autosomal Recessive	NM_000920.3
Pyruvate Dehydrogenase Complex Deficiency	DLAT	Autosomal Recessive	NM_001931.4
Pyruvate Dehydrogenase Complex Deficiency	DLD	Autosomal Recessive	NM_000108.3
Pyruvate Dehydrogenase Complex Deficiency	PDHB	Autosomal Recessive	NM_000925.3
Pyruvate Dehydrogenase Complex Deficiency	PDHX	Autosomal Recessive	NM_003477.2
Pyruvate Dehydrogenase Phosphatase Deficiency	PDP1	Autosomal Recessive	NM_018444.3
Pyruvate Kinase Deficiency	PKLR	Autosomal Recessive	NM_000298.5
Quebec Platelet Disorder	PLAU	Autosomal Dominant	NM_002658.3
Rapid-Onset Dystonia-Parkinsonism	ATP1A3	Autosomal Dominant	NM_152296.4
Recurrent Hydatidiform Mole	NLRP7	Autosomal Recessive	NM_206828.3
Refsum Disease	PEX7	Autosomal Recessive	NM_000288.3
Refsum Disease	PHYH	Autosomal Recessive	NM_006214.3
Renal Adysplasia	RET	Autosomal Dominant	NM_020975.4
Renal Adysplasia	UPK3A	Autosomal Dominant	NM_006953.3
Renal Cysts and Diabetes Syndrome	HNF1B	Autosomal Dominant	NM_000458.2
Renal Glucosuria	SLC5A2	Autosomal Recessive	NM_003041.3
Renal Hypomagnesemia, Dominant	CNNM2	Autosomal Dominant	NM_017649.4
Renal Hypomagnesemia, Dominant	FXYD2	Autosomal Dominant	NM_001680.4
Renal Hypomagnesemia, Recessive	CLDN19	Autosomal Recessive	NM_148960.2
Renal Hypomagnesemia, Recessive	EGF	Autosomal Recessive	NM_001963.4
Renal Hypouricemia	SLC22A12	Autosomal Recessive	NM_144585.2

Renal Hypouricemia	SLC2A9	Autosomal Recessive	NM_020041.2
Renal Tubular Dysgenesis	ACE	Autosomal Recessive	NM_000789.3
Renal Tubular Dysgenesis	AGT	Autosomal Recessive	NM_000029.3
Renal Tubular Dysgenesis	AGTR1	Autosomal Recessive	NM_031850.3
Renal Tubular Dysgenesis	REN	Autosomal Recessive	NM_000537.3
Renal-Hepatic-Pancreatic Dysplasia	NPHP3	Autosomal Recessive	NM_153240.4
Retinal Cone Dystrophy	CACNA2D4	Autosomal Recessive	NM_172364.4
Retinal Cone Dystrophy	PDE6H	Autosomal Recessive	NM_006205.2
Retinal Degeneration	C1QTNF5	Autosomal Dominant	NM_015645.3
Retinal Dystrophy	LRAT	Autosomal Recessive	NM_004744.3
Retinal Dystrophy	OTX2	Autosomal Recessive	NM_172337.2
Retinal Macular Dystrophy	PROM1	Autosomal Dominant	NM_006017.2
Retinitis Pigmentosa, Dominant	ABCA4	Autosomal Dominant	NM_000350.2
Retinitis Pigmentosa, Dominant	AIPL1	Autosomal Dominant	NM_014336.3
Retinitis Pigmentosa, Dominant	CLRN1	Autosomal Dominant	NM_174878.2
Retinitis Pigmentosa, Dominant	CNGA1	Autosomal Dominant	NM_000087.3
Retinitis Pigmentosa, Dominant	CNGB1	Autosomal Dominant	NM_001297.4
Retinitis Pigmentosa, Dominant	CRX	Autosomal Dominant	NM_000554.4
Retinitis Pigmentosa, Dominant	GUCA1B	Autosomal Dominant	NM_002098.5
Retinitis Pigmentosa, Dominant	IMPDH1	Autosomal Dominant	NM_000883.3
Retinitis Pigmentosa, Dominant	KLHL7	Autosomal Dominant	NM_001031710.2
Retinitis Pigmentosa, Dominant	NRL	Autosomal Dominant	NM_006177.3
Retinitis Pigmentosa, Dominant	PRPF3	Autosomal Dominant	NM_004698.2
Retinitis Pigmentosa, Dominant	PRPF31	Autosomal Dominant	NM_015629.3
Retinitis Pigmentosa, Dominant	PRPF6	Autosomal Dominant	NM_012469.3
Retinitis Pigmentosa, Dominant	PRPF8	Autosomal Dominant	NM_006445.3
Retinitis Pigmentosa, Dominant	PRPH2	Autosomal Dominant	NM_000322.4
Retinitis Pigmentosa, Dominant	ROM1	Autosomal Dominant	NM_000327.3

Retinitis Pigmentosa, Dominant	RP1	Autosomal Dominant	NM_006269.1
Retinitis Pigmentosa, Dominant	SNRNP200	Autosomal Dominant	NM_014014.4
Retinitis Pigmentosa, Dominant	TOPORS	Autosomal Dominant	NM_005802.4
Retinitis Pigmentosa, Dominant	ZNF513	Autosomal Dominant	NM_144631.5
Retinitis Pigmentosa, Dominant/Recessive	RHO	Mixed	NM_000539.3
Retinitis Pigmentosa, Recessive	AIPL1	Autosomal Recessive	NM_014336.3
Retinitis Pigmentosa, Recessive	ARL6	Autosomal Recessive	NM_177976.1
Retinitis Pigmentosa, Recessive	BEST1	Autosomal Recessive	NM_004183.3
Retinitis Pigmentosa, Recessive	C2orf71	Autosomal Recessive	NM_001029883.2
Retinitis Pigmentosa, Recessive	C8orf37	Autosomal Recessive	NM_177965.3
Retinitis Pigmentosa, Recessive	CERKL	Autosomal Recessive	NM_201548.4
Retinitis Pigmentosa, Recessive	CRB1	Autosomal Recessive	NM_201253.2
Retinitis Pigmentosa, Recessive	DHDDS	Autosomal Recessive	NM_024887.3
Retinitis Pigmentosa, Recessive	EYS	Autosomal Recessive	NM_001142800.1
Retinitis Pigmentosa, Recessive	FAM161A	Autosomal Recessive	NM_032180.2
Retinitis Pigmentosa, Recessive	IDH3B	Autosomal Recessive	NM_006899.3
Retinitis Pigmentosa, Recessive	IMPG2	Autosomal Recessive	NM_016247.3
Retinitis Pigmentosa, Recessive	LRAT	Autosomal Recessive	NM_004744.3
Retinitis Pigmentosa, Recessive	MAK	Autosomal Recessive	NM_001242957.1
Retinitis Pigmentosa, Recessive	MERTK	Autosomal Recessive	NM_006343.2
Retinitis Pigmentosa, Recessive	NR2E3	Autosomal Recessive	NM_014249.2
Retinitis Pigmentosa, Recessive	PDE6A	Autosomal Recessive	NM_000440.2
Retinitis Pigmentosa, Recessive	PDE6B	Autosomal Recessive	NM_000283.3
Retinitis Pigmentosa, Recessive	PDE6G	Autosomal Recessive	NM_002602.3
Retinitis Pigmentosa, Recessive	PRCD	Autosomal Recessive	NM_001077620.2
Retinitis Pigmentosa, Recessive	PROM1	Autosomal Recessive	NM_006017.2
Retinitis Pigmentosa, Recessive	RBP3	Autosomal Recessive	NM_002900.2
Retinitis Pigmentosa, Recessive	RDH12	Autosomal Recessive	NM_152443.2

Retinitis Pigmentosa, Recessive	RGR	Autosomal Recessive	NM_001012720.1
Retinitis Pigmentosa, Recessive	RLBP1	Autosomal Recessive	NM_000326.4
Retinitis Pigmentosa, Recessive	RPE65	Autosomal Recessive	NM_000329.2
Retinitis Pigmentosa, Recessive	SAG	Autosomal Recessive	NM_000541.4
Retinitis Pigmentosa, Recessive	SEMA4A	Autosomal Recessive	NM_022367.3
Retinitis Pigmentosa, Recessive	SPATA7	Autosomal Recessive	NM_018418.4
Retinitis Pigmentosa, Recessive	TTC8	Autosomal Recessive	NM_198309.2
Retinitis Pigmentosa, Recessive	TULP1	Autosomal Recessive	NM_003322.3
Retinitis Pigmentosa, Recessive	USH2A	Autosomal Recessive	NM_206933.2
Retinitis Pigmentosa, X-Linked	CA4	X-Linked	NM_000717.3
Retinitis Pigmentosa, X-Linked	RP2	X-Linked	NM_006915.2
Retinoblastoma	RB1	Autosomal Dominant	NM_000321.2
Revesz Syndrome	TINF2	Autosomal Dominant	NM_001099274.1
Rhabdoid Tumor Predisposition Syndrome	SMARCB1	Autosomal Dominant	NM_003073.3
Rhizomelic Chondrodysplasia Punctata	AGPS	Autosomal Recessive	NM_003659.3
Rhizomelic Chondrodysplasia Punctata	GNPAT	Autosomal Recessive	NM_014236.3
Rhizomelic Chondrodysplasia Punctata	PEX7	Autosomal Recessive	NM_000288.3
Ribose 5-Phosphate Isomerase Deficiency	RPIA	Autosomal Dominant	NM_144563.2
Ring Dermoid of Cornea	PITX2	Autosomal Dominant	NM_153427.2
Roberts Syndrome	ESCO2	Autosomal Recessive	NM_001017420.2
Robinow Syndrome	ROR2	Autosomal Dominant	NM_004560.3
Robinow Syndrome	WNT5A	Autosomal Dominant	NM_003392.4
Romano-Ward Syndrome	AKAP9	Autosomal Dominant	NM_005751.4
Romano-Ward Syndrome	CAV3	Autosomal Dominant	NM_033337.2
Romano-Ward Syndrome	KCNE1	Autosomal Dominant	NM_000219.3
Romano-Ward Syndrome	KCNE2	Autosomal Dominant	NM_172201.1
Romano-Ward Syndrome	KCNJ5	Autosomal Dominant	NM_000890.3
Romano-Ward Syndrome	KCNQ1	Autosomal Dominant	NM_000218.2

Romano-Ward Syndrome	SCN4B	Autosomal Dominant	NM_174934.3
Romano-Ward Syndrome	SCN5A	Autosomal Dominant	NM_198056.2
Romano-Ward Syndrome	SNTA1	Autosomal Dominant	NM_003098.2
Rotor Syndrome	SLCO1B1	Autosomal Recessive	NM_006446.4
Rotor Syndrome	SLCO1B3	Autosomal Recessive	NM_019844.3
Roussy-Levy Syndrome	MPZ	Autosomal Dominant	NM_000530.6
Rubinstein-Taybi Syndrome	EP300	Autosomal Dominant	NM_001429.3
Saethre-Chotzen Syndrome	FGFR2	Autosomal Dominant	NM_000141.4
Salih Myopathy	TTN	Autosomal Recessive	NM_133378.4
SALL4-Related Spectrum Disorders	SALL4	Autosomal Dominant	NM_020436.3
Salla Disease	SLC17A5	Autosomal Recessive	NM_012434.4
Sandhoff Disease	HEXB	Autosomal Recessive	NM_000521.3
SCAD Deficiency	ACADS	Autosomal Recessive	NM_000017.2
Scapuloperoneal Myopathy	MYH7	Autosomal Dominant	NM_000257.2
Scapuloperoneal Spinal Muscular Atrophy	TRPV4	Autosomal Dominant	NM_021625.4
Schimke Immunoosseous Dysplasia	SMARCAL1	Autosomal Recessive	NM_014140.3
Schindler Disease	NAGA	Autosomal Recessive	NM_000262.2
Schinzel-Giedion Midface Retraction Syndrome	SETBP1	Autosomal Dominant	NM_015559.2
Schnyder Crystalline Corneal Dystrophy	UBIAD1	Autosomal Dominant	NM_013319.2
Schopf-Schulz-Passarge Syndrome	WNT10A	Autosomal Recessive	NM_025216.2
Schwannomatosis	SMARCB1	Autosomal Dominant	NM_003073.3
Schwartz-Jampel Syndrome	HSPG2	Autosomal Recessive	NM_005529.5
Sclerosing Bone Dysplasias	SOST	Autosomal Recessive	NM_025237.2
Sebastian Syndrome	MYH9	Autosomal Dominant	NM_002473.4
Seckel Syndrome	ATR	Autosomal Recessive	NM_001184.3
Seckel Syndrome	CENPJ	Autosomal Recessive	NM_018451.4
Seckel Syndrome	CEP152	Autosomal Recessive	NM_014985.3
Seckel Syndrome	RBBP8	Autosomal Recessive	NM_002894.2

Seizure Disorders	SCN1A	Autosomal Dominant	NM_001165963.1
Seizures, Sensorineural Deafness, Ataxia, Mental Retardation, and Electrolyte Imbalance Syndrome	KCNJ10	Autosomal Recessive	NM_002241.4
Selective Tooth Agenesis	PAX9	Autosomal Dominant	NM_006194.3
Selective Tooth Agenesis	WNT10A	Autosomal Dominant	NM_025216.2
Sengers Syndrome	AGK	Autosomal Recessive	NM_018238.3
Senior-Loken Syndrome	CEP290	Autosomal Recessive	NM_025114.3
Senior-Loken Syndrome	IQCB1	Autosomal Recessive	NM_001023570.2
Senior-Loken Syndrome	NPHP1	Autosomal Recessive	NM_000272.3
Senior-Loken Syndrome	NPHP4	Autosomal Recessive	NM_015102.3
Senior-Loken Syndrome	SDCCAG8	Autosomal Recessive	NM_006642.3
Sensory Neuropathy with Spastic Paraplegia	CCT5	Autosomal Recessive	NM_012073.3
Septooptic Dysplasia	HESX1	Autosomal Recessive	NM_003865.2
Severe Combined Immune Deficiency	CD3D	Autosomal Recessive	NM_000732.4
Severe Combined Immune Deficiency	CD3E	Autosomal Recessive	NM_000733.3
Severe Combined Immune Deficiency	IL7R	Autosomal Recessive	NM_002185.3
Severe Combined Immune Deficiency	JAK3	Autosomal Recessive	NM_000215.3
Severe Combined Immune Deficiency	RAG1	Autosomal Recessive	NM_000448.2
Severe Combined Immune Deficiency	RAG2	Autosomal Recessive	NM_000536.3
Severe Combined Immunodeficiency	IL2RG	Autosomal Recessive	NM_000206.2
Severe Combined Immunodeficiency	ZAP70	Autosomal Recessive	NM_001079.3
Severe Combined Immunodeficiency with Sensitivity to Ionizing Radiation	LIG4	Autosomal Recessive	NM_002312.3
Severe Congenital Neutropenia	G6PC3	Autosomal Dominant	NM_138387.3
Severe Congenital Neutropenia	GFI1	Autosomal Dominant	NM_005263.3
Severe Congenital Neutropenia	HAX1	Autosomal Dominant	NM_006118.3
Short QT Syndrome	KCNJ2	Autosomal Dominant	NM_000891.2
Short QT Syndrome	KCNQ1	Autosomal Dominant	NM_000218.2
Short Rib Polydactyly Syndrome	DYNC2H1	Autosomal Recessive	NM_001080463.1

Short Rib Polydactyly Syndrome	NEK1	Autosomal Recessive	NM_012224.2
Short Rib Polydactyly Syndrome	WDR35	Autosomal Recessive	NM_001006657.1
Sialuria	GNE	Autosomal Dominant	NM_005476.5
Sick Sinus Syndrome	SCN5A	Autosomal Dominant	NM_198056.2
Sickle Cell Disease	HBB	Autosomal Recessive	NM_000518.4
Sideroblastic Anemia and Ataxia	ABCB7	X-Linked	NM_004299.4
Sitosterolemia	ABCG5	Autosomal Recessive	NM_022436.2
Sitosterolemia	ABCG8	Autosomal Recessive	NM_022437.2
Sjogren-Larsson Syndrome	ALDH3A2	Autosomal Recessive	NM_000382.2
Skeletal Dysplasia	CHST3	Autosomal Recessive	NM_004273.4
Skin Fragility-Woolly Hair Syndrome	DSP	Autosomal Recessive	NM_004415.2
SLC6A4-Related Behavior Disorders	SLC6A4	Autosomal Dominant	NM_001045.5
Small Fiber Neuropathy	SCN9A	Autosomal Dominant	NM_002977.3
Small Patella Syndrome	TBX4	Autosomal Dominant	NM_018488.2
Smith-Lemli-Opitz Syndrome	DHCR7	Autosomal Recessive	NM_001360.2
Smith-McCort Dysplasia	DYM	Autosomal Recessive	NM_017653.3
Sotos Syndrome	NSD1	Autosomal Dominant	NM_022455.4
Spastic Paraplegia, Dominant	ATL1	Autosomal Dominant	NM_015915.4
Spastic Paraplegia, Dominant	HSPD1	Autosomal Dominant	NM_002156.4
Spastic Paraplegia, Dominant	KIAA0196	Autosomal Dominant	NM_014846.3
Spastic Paraplegia, Dominant	KIF5A	Autosomal Dominant	NM_004984.2
Spastic Paraplegia, Dominant	NIPA1	Autosomal Dominant	NM_144599.4
Spastic Paraplegia, Dominant	SLC33A1	Autosomal Dominant	NM_004733.3
Spastic Paraplegia, Dominant	SPAST	Autosomal Dominant	NM_014946.3
Spastic Paraplegia, Dominant	ZFYVE27	Autosomal Dominant	NM_001002261.3
Spastic Paraplegia, Recessive	CYP7B1	Autosomal Recessive	NM_004820.3
Spastic Paraplegia, Recessive	KIF1A	Autosomal Recessive	NM_004321.6
Spastic Paraplegia, Recessive	PNPLA6	Autosomal Recessive	NM_006702.4

Spastic Paraplegia, Recessive	SPG11	Autosomal Recessive	NM_025137.3
Spastic Paraplegia, Recessive	SPG7	Autosomal Recessive	NM_003119.2
Spastic Paraplegia, Recessive	ZFYVE26	Autosomal Recessive	NM_015346.3
Spastic Quadriplegic Cerebral Palsy	GAD1	Autosomal Recessive	NM_000817.2
Speech-Language Disorder	FOXP2	Autosomal Dominant	NM_014491.3
Spermatogenic Failure	AURKC	Autosomal Recessive	NM_001015878.1
Spermatogenic Failure	SPATA16	Autosomal Recessive	NM_031955.5
Spherocytosis, Dominant	ANK1	Autosomal Dominant	NM_000037.3
Spherocytosis, Dominant	SLC4A1	Autosomal Dominant	NM_000342.3
Spherocytosis, Dominant	SPTB	Autosomal Dominant	NM_000347.5
Spherocytosis, Recessive	EPB42	Autosomal Recessive	NM_000119.2
Spherocytosis, Recessive	SPTA1	Autosomal Recessive	NM_003126.2
Spheroid Body Myopathy	MYOT	Autosomal Dominant	NM_006790.2
Spinocerebellar Ataxia, Dominant	AFG3L2	Autosomal Dominant	NM_006796.2
Spinocerebellar Ataxia, Dominant	DYNC1H1	Autosomal Dominant	NM_001376.4
Spinocerebellar Ataxia, Dominant	FGF14	Autosomal Dominant	NM_004115.3
Spinocerebellar Ataxia, Dominant	ITPR1	Autosomal Dominant	NM_002222.5
Spinocerebellar Ataxia, Dominant	PDYN	Autosomal Dominant	NM_024411.4
Spinocerebellar Ataxia, Dominant	PRKCG	Autosomal Dominant	NM_002739.3
Spinocerebellar Ataxia, Dominant	SPTBN2	Autosomal Dominant	NM_006946.2
Spinocerebellar Ataxia, Dominant	TGM6	Autosomal Dominant	NM_198994.2
Spinocerebellar Ataxia, Dominant	VAPB	Autosomal Dominant	NM_004738.4
Spinocerebellar Ataxia, Recessive	ADCK3	Autosomal Recessive	NM_020247.4
Spinocerebellar Ataxia, Recessive	ANO10	Autosomal Recessive	NM_018075.3
Spinocerebellar Ataxia, Recessive	C10orf2	Autosomal Recessive	NM_021830.4
Spinocerebellar Ataxia, Recessive	IGHMBP2	Autosomal Recessive	NM_002180.2
Spinocerebellar Ataxia, Recessive	TDP1	Autosomal Recessive	NM_018319.3
Spinocerebellar Ataxia, X-Linked	UBA1	X-Linked	NM_003334.3

Split-Hand/Foot Malformation	FBXW4	Autosomal Dominant	NM_022039.3
Spondylocostal Dysostosis	DLL3	Autosomal Recessive	NM_016941.3
Spondyloepimetaphyseal Dysplasia	DDR2	Autosomal Dominant	NM_006182.2
Spondyloepimetaphyseal Dysplasia	MMP13	Autosomal Dominant	NM_002427.3
Spondyloepiphyseal Dysplasia	CHST3	Autosomal Recessive	NM_004273.4
Spondylometaphyseal Dysplasia	TRPV4	Autosomal Dominant	NM_021625.4
Spontaneous Pneumothorax	FLCN	Autosomal Dominant	NM_144997.5
Stargardt Disease, Dominant	ELOVL4	Autosomal Dominant	NM_022726.3
Stargardt Disease, Dominant	PROM1	Autosomal Dominant	NM_006017.2
Stargardt Disease, Recessive	ABCA4	Autosomal Recessive	NM_000350.2
Stargardt Disease, Recessive	CNGB3	Autosomal Recessive	NM_019098.4
Steroid 5-Alpha-Reductase Deficiency	SRD5A2	Autosomal Recessive	NM_000348.3
Steroid-Resistant Nephrotic Syndrome	NPHS2	Autosomal Recessive	NM_014625.2
Stickler Syndrome, Dominant	COL11A1	Autosomal Dominant	NM_001854.3
Stickler Syndrome, Dominant	COL11A2	Autosomal Dominant	NM_080680.2
Stickler Syndrome, Dominant	COL2A1	Autosomal Dominant	NM_001844.4
Stickler Syndrome, Recessive	COL9A1	Autosomal Recessive	NM_001851.4
Stickler Syndrome, Recessive	COL9A2	Autosomal Recessive	NM_001852.3
Stiff Skin Syndrome	FBN1	Autosomal Dominant	NM_000138.4
Striatal Degeneration	PDE8B	Autosomal Dominant	NM_003719.3
Stuve-Wiedemann Syndrome	LIFR	Autosomal Recessive	NM_002310.5
Succinic Semialdehyde Dehydrogenase Deficiency	ALDH5A1	Autosomal Recessive	NM_001080.3
Succinyl-CoA:3-Oxoacid CoA Transferase Deficiency	OXCT1	Autosomal Recessive	NM_000436.3
Sulfate Transporter-Related Osteochondrodysplasia	SLC26A2	Autosomal Recessive	NM_000112.3
Sulfocysteinuria	SUOX	Autosomal Recessive	NM_000456.2
Supravalvular Aortic Stenosis	ELN	Autosomal Dominant	NM_000501.2
Syndactyly	DLL3	Autosomal Dominant	NM_016941.3
Syndactyly	GJA1	Autosomal Dominant	NM_000165.3

Syndromic Microphthalmia, Dominant	BMP4	Autosomal Dominant	NM_001202.3
Syndromic Microphthalmia, Dominant	OTX2	Autosomal Dominant	NM_172337.2
Syndromic Microphthalmia, Recessive	STRA6	Autosomal Recessive	NM_022369.3
Systemic Primary Carnitine Deficiency	SLC22A5	Autosomal Recessive	NM_003060.3
Tangier Disease	ABCA1	Autosomal Recessive	NM_005502.3
Tetra-Amelia Syndrome	WNT3	Autosomal Recessive	NM_030753.3
Thiamine Metabolism Dysfunction Syndrome	SLC19A3	Autosomal Recessive	NM_025243.3
Thiamine-Responsive Megaloblastic Anemia	SLC19A2	Autosomal Recessive	NM_006996.2
Thiamine-Responsive Megaloblastic Anemia Syndrome	SLC19A2	Autosomal Recessive	NM_006996.2
Thiopurine S-Methyltransferase Deficiency	TPMT	Autosomal Recessive	NM_000367.2
Thoracic Aortic Aneurysms and Aortic Dissections	ACTA2	Autosomal Dominant	NM_001613.2
Thoracic Aortic Aneurysms and Aortic Dissections	FBN1	Autosomal Dominant	NM_000138.4
Thoracic Aortic Aneurysms and Aortic Dissections	MYH11	Autosomal Dominant	NM_002474.2
Thoracic Aortic Aneurysms and Aortic Dissections	MYLK	Autosomal Dominant	NM_053025.3
Thoracic Aortic Aneurysms and Aortic Dissections	SMAD3	Autosomal Dominant	NM_005902.3
Thoracic Aortic Aneurysms and Aortic Dissections	TGFBR1	Autosomal Dominant	NM_004612.2
Thoracic Aortic Aneurysms and Aortic Dissections	TGFBR2	Autosomal Dominant	NM_003242.5
Thrombasthenia of Glanzmann and Naegeli	ITGA2B	Autosomal Recessive	NM_000419.3
Thrombasthenia of Glanzmann and Naegeli	ITGB3	Autosomal Recessive	NM_000212.2
Thrombocytopenia	ANKRD26	Autosomal Dominant	NM_014915.2
Thrombocytopenia	CYCS	Autosomal Dominant	NM_018947.5
Thrombocytopenia	MASTL	Autosomal Dominant	NM_032844.3
Thyroid Dyshormonogenesis	TG	Autosomal Recessive	NM_003235.4
Thyroid Hormone Resistance	THRB	Autosomal Dominant	NM_000461.4
Thyroid Hormonogenesis Defect	SLC5A5	Autosomal Recessive	NM_000453.2
Tietz Syndrome	MITF	Autosomal Dominant	NM_000248.3
Timothy Syndrome	CACNA1C	Autosomal Dominant	NM_000719.6
Tourette Syndrome	SLITRK1	Autosomal Dominant	NM_052910.1

Townes-Brocks Syndrome	SALL1	Autosomal Dominant	NM_002968.2
TP63-Related Spectrum Disorders	TP63	Autosomal Dominant	NM_003722.4
Transaldolase Deficiency	TALDO1	Autosomal Recessive	NM_006755.1
Transcobalamin II Deficiency	TCN2	Autosomal Recessive	NM_000355.3
Transient Familial Neonatal Hyperbilirubinemia	UGT1A1	Autosomal Dominant	NM_000463.2
Transient Neonatal Diabetes, Dominant	KCNJ11	Autosomal Dominant	NM_000525.3
Transient Neonatal Diabetes, Dominant/Recessive	ABCC8	Mixed	NM_000352.3
Transient Neonatal Diabetes, Dominant/Recessive	INS	Mixed	NM_000207.2
Transient Neonatal Diabetes, Recessive	GCK	Autosomal Recessive	NM_000162.3
Treacher Collins Syndrome, Dominant	POLR1D	Autosomal Dominant	NM_015972.3
Treacher Collins Syndrome, Dominant	TCOF1	Autosomal Dominant	NM_001135243.1
Treacher Collins Syndrome, Recessive	POLR1C	Autosomal Recessive	NM_203290.2
Trichohepatoenteric Syndrome	SKIV2L	Autosomal Recessive	NM_006929.4
Trichorhinophalangeal Syndrome	TRPS1	Autosomal Dominant	NM_014112.2
Trichorhinophalangeal Syndrome	TRPS1	Autosomal Dominant	NM_014112.2
Trifunctional Protein Deficiency	HADHA	Autosomal Recessive	NM_000182.4
Trifunctional Protein Deficiency	HADHB	Autosomal Recessive	NM_000183.2
Trimethylaminuria	FMO3	Autosomal Recessive	NM_006894.5
Triosephosphate Isomerase Deficiency	TPI1	Autosomal Dominant	NM_000365.5
Triphalangeal Thumb-Polysyndactyly Syndrome	LMBR1	Autosomal Dominant	NM_022458.3
Trismus-Pseudocamptodactyly Syndrome	MYH8	Autosomal Dominant	NM_002472.2
Tryptophan Hydroxylase Deficiency	TPH2	Autosomal Dominant	NM_173353.3
Tuberous Sclerosis	TSC1	Autosomal Dominant	NM_000368.4
Tuberous Sclerosis	TSC2	Autosomal Dominant	NM_000548.3
Tumor Predisposition Syndrome	BAP1	Autosomal Dominant	NM_004656.2
Tylosis with Esophageal Cancer	RHBDF2	Autosomal Dominant	NM_024599.5
Type II Collagenopathies	COL2A1	Autosomal Dominant	NM_001844.4
Tyrosine Hydroxylase Deficiency	TH	Autosomal Recessive	NM_000360.3

Tyrosinemia	FAH	Autosomal Recessive	NM_000137.2
Tyrosinemia	HPD	Autosomal Recessive	NM_002150.2
Tyrosinemia	TAT	Autosomal Recessive	NM_000353.2
Udd Distal Myopathy	TTN	Autosomal Dominant	NM_133378.4
Ulnar-Mammary Syndrome	TBX3	Autosomal Dominant	NM_005996.3
Unverricht-Lundborg Disease	CSTB	Autosomal Recessive	NM_000100.3
Usher Syndrome	CDH23	Autosomal Recessive	NM_022124.5
Usher Syndrome	CLRN1	Autosomal Recessive	NM_174878.2
Usher Syndrome	DFNB31	Autosomal Recessive	NM_015404.3
Usher Syndrome	GPR98	Autosomal Recessive	NM_032119.3
Usher Syndrome	HARS	Autosomal Recessive	NM_002109.4
Usher Syndrome	MYO7A	Autosomal Recessive	NM_000260.3
Usher Syndrome	PCDH15	Autosomal Recessive	NM_033056.3
Usher Syndrome	USH1C	Autosomal Recessive	NM_005709.3
Usher Syndrome	USH2A	Autosomal Recessive	NM_206933.2
VACTERL Association with Hydrocephalus	FANCB	X-Linked	NM_001018113.1
van der Woude Syndrome	IRF6	Autosomal Dominant	NM_006147.3
Variegate Porphyria	PPOX	Autosomal Dominant	NM_000309.3
Vesicoureteral Reflux	ROBO2	Autosomal Dominant	NM_002942.4
Vitamin D-Dependent Rickets	CYP27B1	Autosomal Recessive	NM_000785.3
Vitamin D-Dependent Rickets	VDR	Autosomal Recessive	NM_001017535.1
Vitamin K-Dependent Clotting Factors	GGCX	Autosomal Recessive	NM_000821.5
Vitamin K-Dependent Clotting Factors	VKORC1	Autosomal Recessive	NM_024006.4
Vitelliform Macular Dystrophy	PRPH2	Autosomal Dominant	NM_000322.4
Vitreoretinochoroidopathy	BEST1	Autosomal Dominant	NM_004183.3
Vitreoretinopathy	VCAN	Autosomal Dominant	NM_004385.4
VLCAD Deficiency	ACADVL	Autosomal Recessive	NM_000018.2
Vohwinkel Syndrome	GJB2	Autosomal Dominant	NM_004004.5

von Willebrand Disease	VWF	Autosomal Dominant	NM_000552.3
VSX2-related Microphthalmia	VSX2	Autosomal Recessive	NM_182894.2
Waardenburg Syndrome	EDN3	Autosomal Dominant	NM_207034.1
Waardenburg Syndrome	EDNRB	Autosomal Dominant	NM_000115.3
Waardenburg Syndrome	MITF	Autosomal Dominant	NM_000248.3
Waardenburg Syndrome	PAX3	Autosomal Dominant	NM_181457.3
Waardenburg Syndrome	SNAI2	Autosomal Dominant	NM_003068.4
Waardenburg Syndrome	SOX10	Autosomal Dominant	NM_006941.3
Wagner Syndrome	VCAN	Autosomal Dominant	NM_004385.4
WAGR Syndrome	PAX6	Autosomal Dominant	NM_000280.4
WAGR Syndrome	WT1	Autosomal Dominant	NM_024426.4
Walker-Warburg Syndrome	LARGE	Autosomal Recessive	NM_004737.4
Warburg Micro Syndrome	RAB18	Autosomal Recessive	NM_021252.4
Warburg Micro Syndrome	RAB3GAP1	Autosomal Recessive	NM_012233.2
Warburg Micro Syndrome	RAB3GAP2	Autosomal Recessive	NM_012414.3
Watson Syndrome	NF1	Autosomal Dominant	NM_000267.3
Weaver Syndrome	EZH2	Autosomal Dominant	NM_004456.4
Weaver Syndrome	NSD1	Autosomal Dominant	NM_022455.4
Weill-Marchesani Syndrome	ADAMTS10	Autosomal Dominant	NM_030957.2
Weill-Marchesani Syndrome	FBN1	Autosomal Dominant	NM_000138.4
Weill-Marchesani Syndrome	LTBP2	Autosomal Dominant	NM_000428.2
Weill-Marchesani-Like Syndrome	ADAMTS17	Autosomal Recessive	NM_139057.2
Weissenbacher-Zweymuller Syndrome	COL11A2	Autosomal Dominant	NM_080680.2
Werner Syndrome	WRN	Autosomal Recessive	NM_000553.4
Weyers Acrofacial Dysostosis	EVC	Autosomal Dominant	NM_153717.2
WFS1-Related Spectrum Disorders	WFS1	Mixed	NM_006005.3
White Sponge Nevus of Cannon	KRT13	Autosomal Dominant	NM_002274.3
White Sponge Nevus of Cannon	KRT4	Autosomal Dominant	NM_002272.3

Wilms Tumor	WT1	Autosomal Dominant	NM_024426.4
Wilson Disease	ATP7B	Autosomal Recessive	NM_000053.2
Winchester Syndrome	MMP2	Autosomal Recessive	NM_004530.4
Wolff-Parkinson-White Syndrome	PRKAG2	Autosomal Dominant	NM_016203.3
Wolf-Hirschhorn Syndrome	WHSC1	Autosomal Dominant	NM_133330.2
Wolman Disease	LIPA	Autosomal Recessive	NM_000235.2
Woodhouse-Sakati Syndrome	DCAF17	Autosomal Recessive	NM_025000.3
Xanthinuria	XDH	Autosomal Dominant	NM_000379.3
Xeroderma Pigmentosum	DDB2	Autosomal Recessive	NM_000107.2
Xeroderma Pigmentosum	ERCC1	Autosomal Recessive	NM_202001.2
Xeroderma Pigmentosum	ERCC2	Autosomal Recessive	NM_000400.3
Xeroderma Pigmentosum	ERCC3	Autosomal Recessive	NM_000122.1
Xeroderma Pigmentosum	ERCC4	Autosomal Recessive	NM_005236.2
Xeroderma Pigmentosum	ERCC5	Autosomal Recessive	NM_000123.3
Xeroderma Pigmentosum	POLH	Autosomal Recessive	NM_006502.2
Xeroderma Pigmentosum	XPA	Autosomal Recessive	NM_000380.3
Xeroderma Pigmentosum	XPC	Autosomal Recessive	NM_004628.4
X-Linked Hypophosphatemia	PHEX	X-Linked	NM_000444.4
Zaspopathy	LDB3	Autosomal Dominant	NM_001080116.1
Zellweger Syndrome	PEX1	Autosomal Recessive	NM_000466.2
Zellweger Syndrome	PEX10	Autosomal Recessive	NM_153818.1
Zellweger Syndrome	PEX12	Autosomal Recessive	NM_000286.2
Zellweger Syndrome	PEX13	Autosomal Recessive	NM_002618.3
Zellweger Syndrome	PEX14	Autosomal Recessive	NM_004565.2
Zellweger Syndrome	PEX16	Autosomal Recessive	NM_004813.2
Zellweger Syndrome	PEX19	Autosomal Recessive	NM_002857.3
Zellweger Syndrome	PEX2	Autosomal Recessive	NM_000318.2
Zellweger Syndrome	PEX26	Autosomal Recessive	NM_017929.5

Zellweger Syndrome	PEX3	Autosomal Recessive	NM_003630.2
Zellweger Syndrome	PEX5	Autosomal Recessive	NM_001131025.1
Zellweger Syndrome	PEX6	Autosomal Recessive	NM_000287.3
Zonular Pulverulent Cataract	GJA3	Autosomal Dominant	NM_021954.3
Zonular Pulverulent Cataract	GJA8	Autosomal Dominant	NM_005267.4