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

Disease	Associated Gene	Mode of Inheritance	Transcript ID	Callability (%)
17-Beta-Hydroxysteroid Dehydrogenase III Deficiency	HSD17B3	RECESSIVE	NM_000197.1	99.98%
3-Hydroxy-3-Methylglutaryl-CoA Synthase 2 Deficiency	HMGCS2	RECESSIVE	NM_005518.3	99.90%
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	HMGCL	RECESSIVE	NM_000191.2	98.43%
3-Hydroxyacyl-Coenzyme A Dehydrogenase Deficiency	HADH	RECESSIVE	NM_005327.4	99.54%
3-M Syndrome	CUL7	RECESSIVE	NM_014780.4	99.22%
3-MCC Deficiency	MCCC1	RECESSIVE	NM_020166.3	99.59%
3-MCC Deficiency	MCCC2	RECESSIVE	NM_022132.4	99.67%
3-Methylglutaconic Aciduria, Type 1	AUH	RECESSIVE	NM_001698.2	99.03%
3-Methylglutaconic Aciduria, Type 2	TAZ	X_LINKED	NM_000116.3	90.55%
3-Methylglutaconic Aciduria, Type 3	OPA3	RECESSIVE	NM_025136.3	97.52%
3-Methylglutaconic Aciduria, Type 5	DNAJC19	RECESSIVE	NM_145261.3	99.80%
46,XY DSD/46,XY CGD	DHH	RECESSIVE	NM_021044.2	99.37%
6-Pyruvoyltetrahydropterin Synthase Deficiency	PTS	DOMINANT	NM_000317.2	96.48%
Abetalipoproteinemia	MTTP	RECESSIVE	NM_000253.2	99.20%
ACAD9 Deficiency	ACAD9	RECESSIVE	NM_014049.4	97.67%
Aceruloplasminemia	CP	RECESSIVE	NM_000096.3	98.64%
Achalasia-Addisonianism-Alacrima Syndrome	AAAS	RECESSIVE	NM_015665.5	99.82%
Achondrogenesis	SLC26A2	DOMINANT	NM_000112.3	99.63%
Achondrogenesis	TRIP11	DOMINANT	NM_004239.3	99.76%
Achromatopsia	CNGA3	RECESSIVE	NM_001298.2	99.74%
Achromatopsia	CNGB3	RECESSIVE	NM_019098.4	99.76%

Achromatopsia	GNAT2	RECESSIVE	NM_005272.3	99.98%
Achromatopsia	PDE6C	RECESSIVE	NM_006204.3	99.96%
Acid Sphingomyelinase Deficiency	SMPD1	RECESSIVE	NM_000543.4	97.05%
Acid-Labile Subunit Deficiency	IGFALS	RECESSIVE	NM_004970.2	97.51%
Acrodermatitis Enteropathica	SLC39A4	RECESSIVE	NM_130849.2	98.53%
Acrodysostosis	PDE4D	DOMINANT	NM_001104631.1	98.42%
Acrodysostosis	PRKAR1A	DOMINANT	NM_002734.3	99.91%
Acromesomelic Dysplasia	GDF5	RECESSIVE	NM_000557.2	98.65%
Acromesomelic Dysplasia	NPR2	RECESSIVE	NM_003995.3	99.04%
Acromicric Dysplasia	FBN1	DOMINANT	NM_000138.4	99.59%
ACTH Deficiency	TBX19	RECESSIVE	NM_005149.2	99.72%
Acute Hepatic Porphyria	ALAD	DOMINANT	NM_000031.5	99.37%
Acute Infantile Liver Failure	TRMU	RECESSIVE	NM_018006.4	97.62%
Acute Recurrent Myoglobinuria	LPIN1	DOMINANT	NM_145693.2	98.10%
Acyl-CoA Dehydrogenase, Short/Branched Chain Deficiency	ACADSB	RECESSIVE	NM_001609.3	99.29%
Adams-Oliver Syndrome	ARHGAP31	DOMINANT	NM_020754.2	99.53%
Adenine Phosphoribosyltransferase Deficiency	APRT	RECESSIVE	NM_000485.2	99.73%
Adenosine Deaminase Deficiency	ADA	RECESSIVE	NM_000022.2	96.79%
Adenylosuccinase Deficiency	ADSL	RECESSIVE	NM_000026.2	97.88%
Adult Polyglucosan Body Disease	GBE1	RECESSIVE	NM_000158.3	98.20%
Age-Related Cortical Cataract	EPHA2	DOMINANT	NM_004431.3	96.56%
Aicardi-Goutieres Syndrome	RNASEH2A	RECESSIVE	NM_006397.2	98.76%
Aicardi-Goutieres Syndrome	RNASEH2B	RECESSIVE	NM_024570.3	97.14%
Aicardi-Goutieres Syndrome	RNASEH2C	RECESSIVE	NM_032193.3	99.13%
Aicardi-Goutieres Syndrome	SAMHD1	RECESSIVE	NM_015474.3	99.80%
Alagille Syndrome	JAG1	DOMINANT	NM_000214.2	99.22%
Aldolase A Deficiency	ALDOA	RECESSIVE	NM_000034.3	99.48%
Alexander Disease	GFAP	DOMINANT	NM_002055.4	98.03%
Alkaptonuria	HGD	RECESSIVE	NM_000187.3	100.00%

Alopecia and T-Cell Immunodeficiency	FOXN1	RECESSIVE	NM_003593.2	98.70%
Alopecia Universalis	HR	RECESSIVE	NM_005144.4	98.05%
Alpha-1 Antitrypsin Deficiency	SERPINA1	RECESSIVE	NM_000295.4	99.77%
Alpha-B Crystallinopathy	CRYAB	RECESSIVE	NM_001885.1	99.76%
Alpha-Mannosidosis	MAN2B1	RECESSIVE	NM_000528.3	97.46%
Alpha-Methylacyl-CoA Racemase Deficiency	AMACR	RECESSIVE	NM_014324.5	98.70%
Alpha-Sarcoglycanopathy	SGCA	RECESSIVE	NM_000023.2	99.02%
Alport Syndrome	COL4A3	DOMINANT	NM_000091.4	99.27%
Alport Syndrome	COL4A4	DOMINANT	NM_000092.4	99.38%
ALS2-Related Spectrum Disorders	ALS2	RECESSIVE	NM_020919.3	99.92%
Alstrom Syndrome	ALMS1	RECESSIVE	NM_015120.4	98.67%
Alternating Hemiplegia of Childhood	ATP1A2	DOMINANT	NM_000702.3	98.69%
Alternating Hemiplegia of Childhood	ATP1A3	DOMINANT	NM_152296.4	97.26%
Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins	FOXF1	DOMINANT	NM_001451.2	95.48%
Alzheimer Disease	APP	DOMINANT	NM_000484.3	99.28%
Alzheimer Disease	PSEN1	DOMINANT	NM_000021.3	99.77%
Alzheimer Disease	PSEN2	DOMINANT	NM_000447.2	98.87%
Amelogenesis Imperfecta, Dominant	DLX3	DOMINANT	NM_005220.2	96.40%
Amelogenesis Imperfecta, Dominant	ENAM	DOMINANT	NM_031889.2	99.93%
Amelogenesis Imperfecta, Recessive	MMP20	RECESSIVE	NM_004771.3	98.80%
Amelogenesis Imperfecta, Recessive	WDR72	RECESSIVE	NM_182758.2	99.30%
Amish Infantile Epilepsy Syndrome	ST3GAL5	RECESSIVE	NM_003896.3	96.43%
Amish Lethal Microcephaly	SLC25A19	RECESSIVE	NM_021734.4	97.88%
Amyloidosis	GSN	DOMINANT	NM_000177.4	95.19%
Amyotrophic Lateral Sclerosis, Dominant	ANG	DOMINANT	NM_001145.4	98.69%
Amyotrophic Lateral Sclerosis, Dominant	FIG4	DOMINANT	NM_014845.5	99.84%
Amyotrophic Lateral Sclerosis, Dominant	SETX	DOMINANT	NM_015046.5	99.83%
Amyotrophic Lateral Sclerosis, Dominant	SOD1	DOMINANT	NM_000454.4	99.92%

Amyotrophic Lateral Sclerosis, Dominant	TARDBP	DOMINANT	NM_007375.3	98.20%
Amyotrophic Lateral Sclerosis, Dominant	VAPB	DOMINANT	NM_004738.4	99.01%
Amyotrophic Lateral Sclerosis, Dominant	VCP	DOMINANT	NM_007126.3	99.01%
Amyotrophic Lateral Sclerosis, Recessive	ALS2	RECESSIVE	NM_020919.3	99.92%
Amyotrophic Lateral Sclerosis, Recessive	FUS	RECESSIVE	NM_004960.3	98.25%
Amyotrophic Lateral Sclerosis, Recessive	OPTN	RECESSIVE	NM_021980.4	99.01%
Amyotrophic Lateral Sclerosis/Frontotemporal Dementia	C9orf72	DOMINANT	NM_001256054.1	99.39%
Andersen-Tawil Syndrome	KCNJ2	DOMINANT	NM_000891.2	98.32%
Angiokeratoma Corporis Diffusum with Arteriovenous Fistulas	KRIT1	DOMINANT	NM_194456.1	99.20%
Aniridia	PAX6	DOMINANT	NM_000280.4	99.26%
Aniridia, Cerebellar Ataxia, And Mental Retardation	PAX6	RECESSIVE	NM_000280.4	99.26%
Anophthalmia	PAX6	DOMINANT	NM_000280.4	99.26%
Anophthalmia/Microphthalmia	SIX6	DOMINANT	NM_007374.2	95.65%
Antenatal Bartter Syndrome	KCNJ1	RECESSIVE	NM_000220.3	100.00%
Antenatal Bartter Syndrome	SLC12A1	RECESSIVE	NM_000338.2	99.97%
Anterior Segment Mesenchymal Dysgenesis	PITX2	DOMINANT	NM_153427.2	99.12%
Antithrombin-III Deficiency	SERPINC1	DOMINANT	NM_000488.3	99.98%
APC-Associated Polyposis Disorders	APC	DOMINANT	NM_000038.5	99.94%
Apert Syndrome	FGFR2	DOMINANT	NM_000141.4	97.52%
Aplasia of Lacrimal and Salivary Glands	FGF10	DOMINANT	NM_004465.1	99.92%
Aplastic Anemia	IFNG	RECESSIVE	NM_000619.2	99.90%
Apolipoprotein C-II Deficiency	APOC2	RECESSIVE	NM_000483.4	99.25%
Arginase Deficiency	ARG1	RECESSIVE	NM_000045.2	99.91%
Argininosuccinate Lyase Deficiency	ASL	RECESSIVE	NM_000048.3	96.32%
Aromatase Deficiency	CYP19A1	RECESSIVE	NM_031226.2	99.10%
Aromatic L-Amino Acid Decarboxylase Deficiency	DDC	RECESSIVE	NM_000790.3	99.93%
ARSACS	SACS	RECESSIVE	NM_014363.4	99.89%
Arterial Tortuosity Syndrome	SLC2A10	RECESSIVE	NM_030777.3	97.77%

Arthrogryposis Multiplex Congenita	MYH3	DOMINANT	NM_002470.3	99.45%
Arthrogryposis Multiplex Congenita	TNNI2	DOMINANT	NM_003282.3	96.15%
Arthrogryposis Multiplex Congenita	TNNT3	DOMINANT	NM_006757.3	99.33%
Arthrogryposis Multiplex Congenita	TPM2	DOMINANT	NM_003289.3	97.92%
Arthrogryposis, Renal Dysfunction, and Cholestasis Syndrome	VPS33B	RECESSIVE	NM_018668.3	99.37%
Arts Syndrome	PRPS1	X_LINKED	NM_002764.3	88.28%
Arylsulfatase A Deficiency	ARSA	RECESSIVE	NM_000487.5	96.29%
Aspartylglycosaminuria	AGA	RECESSIVE	NM_000027.3	99.92%
Asphyxiating Thoracic Dystrophy	DYNC2H1	RECESSIVE	NM_001080463.1	99.44%
Asphyxiating Thoracic Dystrophy	IFT80	RECESSIVE	NM_020800.2	99.65%
Asphyxiating Thoracic Dystrophy	TTC21B	RECESSIVE	NM_024753.4	98.51%
Asphyxiating Thoracic Dystrophy	WDR19	RECESSIVE	NM_025132.3	99.31%
Ataxia Neuropathy Spectrum Disorders	C10orf2	DOMINANT	NM_021830.4	98.46%
Ataxia with Oculomotor Apraxia	APTX	RECESSIVE	NM_175073.2	99.02%
Ataxia with Oculomotor Apraxia	SETX	RECESSIVE	NM_015046.5	99.83%
Ataxia with Vitamin E Deficiency	TTPA	RECESSIVE	NM_000370.3	96.78%
Ataxia-Telangiectasia	ATM	RECESSIVE	NM_000051.3	98.79%
Ataxia-Telangiectasia-Like Disorder	MRE11A	RECESSIVE	NM_005591.3	99.83%
Atelosteogenesis	SLC26A2	RECESSIVE	NM_000112.3	99.63%
Athabaskan Brainstem Dysgenesis Syndrome	HOXA1	RECESSIVE	NM_005522.4	99.19%
Atransferrinemia	TF	RECESSIVE	NM_001063.3	99.67%
Atrial Septal Defect	ACTC1	DOMINANT	NM_005159.4	98.49%
Atrial Septal Defect	MYH6	DOMINANT	NM_002471.3	98.59%
Atrichia with Papular Lesions	HR	RECESSIVE	NM_005144.4	98.05%
Atypical Gaucher Disease	PSAP	DOMINANT	NM_002778.2	99.84%
Atypical Hemolytic-Uremic Syndrome	C3	DOMINANT	NM_000064.2	98.50%
Atypical Hemolytic-Uremic Syndrome	CD46	DOMINANT	NM_002389.4	99.27%
Atypical Hemolytic-Uremic Syndrome	CFB	DOMINANT	NM_001710.5	98.11%

Atypical Hemolytic-Uremic Syndrome	CFH	DOMINANT	NM_000186.3	98.32%
Atypical Hemolytic-Uremic Syndrome	CFI	DOMINANT	NM_000204.3	99.88%
Atypical Hemolytic-Uremic Syndrome	THBD	DOMINANT	NM_000361.2	97.43%
Atypical Werner Syndrome	LMNA	DOMINANT	NM_005572.3	96.33%
Auriculocondylar Syndrome	PLCB4	DOMINANT	NM_000933.3	99.62%
Autism Spectrum Disorders	SHANK2	DOMINANT	NM_012309.3	99.40%
Autism Spectrum Disorders	SNRPN	DOMINANT	NM_022807.2	99.94%
Autoimmune Lymphoproliferative Syndrome	CASP10	DOMINANT	NM_032977.3	99.28%
Autoimmune Lymphoproliferative Syndrome	FAS	DOMINANT	NM_000043.4	99.88%
Autoimmune Lymphoproliferative Syndrome	FASLG	DOMINANT	NM_000639.1	99.62%
Autoinflammation, Lipodystrophy, and Dermatosis Syndrome	PSMB8	RECESSIVE	NM_148919.3	99.96%
Axenfeld-Rieger Syndrome	PITX2	DOMINANT	NM_153427.2	99.12%
Axonal Neuropathy	GARS	DOMINANT	NM_002047.2	99.52%
Bardet-Biedl Syndrome	ARL6	RECESSIVE	NM_177976.1	99.79%
Bardet-Biedl Syndrome	BBS1	RECESSIVE	NM_024649.4	99.74%
Bardet-Biedl Syndrome	BBS10	RECESSIVE	NM_024685.3	99.71%
Bardet-Biedl Syndrome	BBS12	RECESSIVE	NM_152618.2	99.89%
Bardet-Biedl Syndrome	BBS2	RECESSIVE	NM_031885.3	99.70%
Bardet-Biedl Syndrome	BBS4	RECESSIVE	NM_033028.4	99.07%
Bardet-Biedl Syndrome	BBS7	RECESSIVE	NM_176824.2	99.15%
Bardet-Biedl Syndrome	BBS9	RECESSIVE	NM_198428.2	99.50%
Bardet-Biedl Syndrome	CEP290	RECESSIVE	NM_025114.3	99.36%
Bardet-Biedl Syndrome	MKKS	RECESSIVE	NM_018848.3	99.99%
Bardet-Biedl Syndrome	MKS1	RECESSIVE	NM_017777.3	99.30%
Bardet-Biedl Syndrome	SDCCAG8	RECESSIVE	NM_006642.3	98.99%
Bardet-Biedl Syndrome	TRIM32	RECESSIVE	NM_012210.3	99.49%
Bardet-Biedl Syndrome	TTC8	RECESSIVE	NM_198309.2	99.65%
Bardet-Biedl Syndrome	WDPCP	RECESSIVE	NM_015910.5	98.91%

Bare Lymphocyte Syndrome, Type II	CIITA	RECESSIVE	NM_000246.3	99.20%
Bare Lymphocyte Syndrome, Type II	RFX5	RECESSIVE	NM_000449.3	99.56%
Bare Lymphocyte Syndrome, Type II	RFXAP	RECESSIVE	NM_000538.3	98.26%
Bartter Syndrome	BSND	RECESSIVE	NM_057176.2	99.77%
Basal Laminar Drusen	CFH	RECESSIVE	NM_000186.3	98.32%
Beare-Stevenson Syndrome	FGFR2	DOMINANT	NM_000141.4	97.52%
Benign Chronic Pemphigus	ATP2C1	DOMINANT	NM_014382.3	99.61%
Benign Familial Neonatal Infantile Seizures	SCN2A	DOMINANT	NM_021007.2	98.70%
Benign Familial Neonatal Seizures	KCNQ3	DOMINANT	NM_004519.3	97.84%
Benign Hereditary Chorea	NKX2-1	DOMINANT	NM_001079668.2	95.23%
Benign Neonatal Epilepsy	KCNQ3	DOMINANT	NM_004519.3	97.84%
Berardinelli-Seip Congenital Lipodystrophy	AGPAT2	RECESSIVE	NM_006412.3	95.16%
Berardinelli-Seip Congenital Lipodystrophy	BSCL2	RECESSIVE	NM_032667.6	98.94%
Bernard-Soulier Syndrome	GP9	DOMINANT	NM_000174.3	96.48%
Best Vitelliform Macular Dystrophy	BEST1	DOMINANT	NM_004183.3	99.21%
Beta-Mannosidosis	MANBA	RECESSIVE	NM_005908.3	98.72%
Beta-Sarcoglycanopathy	SGCB	RECESSIVE	NM_000232.4	98.84%
Beta-Thalassemia	HBB	RECESSIVE	NM_000518.4	99.96%
Beta-Ureidopropionase Deficiency	UPB1	RECESSIVE	NM_016327.2	99.14%
BH4-Deficient Hyperphenylalaninemia	PCBD1	RECESSIVE	NM_000281.2	96.44%
BH4-Deficient Hyperphenylalaninemia	QDPR	RECESSIVE	NM_000320.2	95.78%
Bietti Crystalline Dystrophy	CYP4V2	RECESSIVE	NM_207352.3	98.99%
Biotinidase Deficiency	BTD	RECESSIVE	NM_000060.2	99.73%
Birk-Barel Mental Retardation Dysmorphism Syndrome	KCNK9	DOMINANT	NM_016601.2	96.95%
Birt-Hogg-Dube Syndrome	FLCN	DOMINANT	NM_144997.5	96.89%
Blau Syndrome	NOD2	DOMINANT	NM_022162.1	99.39%
Bloom Syndrome	BLM	RECESSIVE	NM_000057.2	99.93%
BMP4-Related Syndromic Microphthalmia	BMP4	DOMINANT	NM_001202.3	98.31%
Bohring-Opitz Syndrome	ASXL1	DOMINANT	NM_015338.5	95.70%

Bone Mineral Density Variation	LRP4	RECESSIVE	NM_002334.3	97.69%
Bosley-Salih-Alorainy Syndrome	HOXA1	RECESSIVE	NM_005522.4	99.19%
Brachydactyly	BMPR1B	DOMINANT	NM_001203.2	97.27%
Brachydactyly	GDF5	DOMINANT	NM_000557.2	98.65%
Brachydactyly	IHH	DOMINANT	NM_002181.3	97.64%
Brachydactyly	ROR2	DOMINANT	NM_004560.3	98.00%
Brachyolmia	TRPV4	RECESSIVE	NM_021625.4	97.17%
Brain Small Vessel Disease with Hemorrhage	COL4A1	DOMINANT	NM_001845.4	99.41%
Branchiootorenal Spectrum Disorders	EYA1	DOMINANT	NM_000503.4	98.57%
Branchiootorenal Spectrum Disorders	SIX1	DOMINANT	NM_005982.3	98.47%
Breast and Ovarian Cancer Susceptibility	RAD51C	DOMINANT	NM_058216.1	98.27%
Breast and Ovarian Cancer Susceptibility	RAD51D	DOMINANT	NM_002878.3	99.29%
Breast Cancer	BARD1	RECESSIVE	NM_000465.2	98.53%
Breast Cancer	BRIP1	DOMINANT	NM_032043.2	98.05%
Breast Cancer	CHEK2	DOMINANT	NM_007194.3	96.19%
Brittle Cornea Syndrome	PRDM5	RECESSIVE	NM_018699.2	99.18%
Brittle Cornea Syndrome	ZNF469	RECESSIVE	NM_001127464.1	97.33%
Brody Myopathy	ATP2A1	DOMINANT	NM_173201.3	99.40%
Brooke-Spiegler Syndrome	CYLD	DOMINANT	NM_015247.2	99.42%
Bruck Syndrome	PLOD2	RECESSIVE	NM_000935.2	99.05%
Brugada Syndrome	CACNA1C	DOMINANT	NM_000719.6	98.44%
Brugada Syndrome	CACNB2	DOMINANT	NM_201590.2	98.72%
Brugada Syndrome	GPD1L	DOMINANT	NM_015141.3	99.56%
Brugada Syndrome	KCNE3	DOMINANT	NM_005472.4	94.16%
Brugada Syndrome	SCN1B	DOMINANT	NM_001037.4	90.69%
Brugada Syndrome	SCN3B	DOMINANT	NM_018400.3	99.72%
Brugada Syndrome	SCN5A	DOMINANT	NM_198056.2	98.54%
Budd-Chiari Syndrome	F5	DOMINANT	NM_000130.4	99.15%
Budd-Chiari Syndrome	JAK2	DOMINANT	NM_004972.3	99.18%

Buschke-Ollendorff Syndrome	LEMD3	DOMINANT	NM_014319.4	98.25%
Butyrylcholinesterase Deficiency	BCHE	RECESSIVE	NM_000055.2	99.95%
C Syndrome	CD96	DOMINANT	NM_198196.2	99.48%
C3 Deficiency	C3	RECESSIVE	NM_000064.2	98.50%
Caffey Disease	COL1A1	DOMINANT	NM_000088.3	96.26%
Calpainopathy	CAPN3	RECESSIVE	NM_000070.2	99.52%
Campomelic Dysplasia	SOX9	DOMINANT	NM_000346.3	97.58%
Canavan Disease	ASPA	RECESSIVE	NM_000049.2	99.98%
Capillary Malformation-Arteriovenous Malformation Syndrome	RASA1	DOMINANT	NM_002890.2	97.65%
Carbamoylphosphate Synthetase I Deficiency	CPS1	RECESSIVE	NM_001875.4	99.64%
Cardiofaciocutaneous Syndrome	BRAF	DOMINANT	NM_004333.4	97.81%
Cardiofaciocutaneous Syndrome	KRAS	DOMINANT	NM_004985.3	98.29%
Cardiofaciocutaneous Syndrome	MAP2K1	DOMINANT	NM_002755.3	98.72%
Cardiomyopathy, ARVC	DSC2	DOMINANT	NM_024422.3	99.67%
Cardiomyopathy, ARVC	DSG2	DOMINANT	NM_001943.3	98.52%
Cardiomyopathy, ARVC	DSP	DOMINANT	NM_004415.2	99.77%
Cardiomyopathy, ARVC	JUP	DOMINANT	NM_002230.2	98.34%
Cardiomyopathy, ARVC	PKP2	DOMINANT	NM_004572.3	97.02%
Cardiomyopathy, ARVC	RYR2	DOMINANT	NM_001035.2	99.18%
Cardiomyopathy, ARVC	TGFB3	DOMINANT	NM_003239.2	99.65%
Cardiomyopathy, ARVC	TMEM43	DOMINANT	NM_024334.2	99.41%
Carney Complex	PRKAR1A	DOMINANT	NM_002734.3	99.91%
Carnitine Palmitoyltransferase II Deficiency	CPT2	RECESSIVE	NM_000098.2	99.14%
Carnitine-Acylcarnitine Translocase Deficiency	SLC25A20	RECESSIVE	NM_000387.5	99.82%
Carpenter Syndrome	RAB23	RECESSIVE	NM_183227.1	99.98%
Caspase 8 Deficiency	CASP8	RECESSIVE	NM_001228.4	99.73%
Cataract-Microcornea Syndrome	GJA8	DOMINANT	NM_005267.4	99.35%
Cataracts	BFSP2	DOMINANT	NM_003571.2	99.85%

Cataracts	CRYGD	DOMINANT	NM_006891.3	97.96%
Cataracts	PITX2	DOMINANT	NM_153427.2	99.12%
Catecholaminergic Polymorphic Ventricular Tachycardia	CASQ2	DOMINANT	NM_001232.3	99.12%
Catecholaminergic Polymorphic Ventricular Tachycardia	RYR2	DOMINANT	NM_001035.2	99.18%
Catecholaminergic Polymorphic Ventricular Tachycardia	TRDN	DOMINANT	NM_006073.3	99.51%
Caudal Dysgenesis Syndrome	VANGL1	DOMINANT	NM_138959.2	98.53%
Caveolinopathies	CAV3	DOMINANT	NM_033337.2	99.33%
Central Core Disease	RYR1	DOMINANT	NM_000540.2	97.36%
Centronuclear Myopathy, Dominant	DNM2	DOMINANT	NM_001005360.2	97.58%
Centronuclear Myopathy, Dominant	MYF6	DOMINANT	NM_002469.2	99.81%
Centronuclear Myopathy, Recessive	BIN1	RECESSIVE	NM_139343.2	97.70%
Cerebellar Ataxia	SYNE1	RECESSIVE	NM_033071.3	99.70%
Cerebellar Ataxia, Cayman type	ATCAY	RECESSIVE	NM_033064.4	98.58%
Cerebellar Hypoplasia	VLDLR	RECESSIVE	NM_003383.3	95.99%
Cerebral Dysgenesis, Neuropathy, Ichthyosis, and Palmoplantar Keratoderma Syndrome	SNAP29	RECESSIVE	NM_004782.3	96.18%
Cerebrooculofacioskeletal Syndrome	ERCC6	RECESSIVE	NM_000124.2	99.64%
Cerebrotendinous Xanthomatosis	CYP27A1	RECESSIVE	NM_000784.3	99.77%
Chanarin-Dorfman Syndrome	ABHD5	RECESSIVE	NM_016006.4	99.29%
Char Syndrome	TFAP2B	DOMINANT	NM_003221.3	99.08%
Charcot-Marie-Tooth with Vocal Cord Paresis	GDAP1	RECESSIVE	NM_018972.2	99.26%
Charcot-Marie-Tooth, Intermediate	DNM2	DOMINANT	NM_001005360.2	97.58%
Charcot-Marie-Tooth, Intermediate	GDAP1	DOMINANT	NM_018972.2	99.26%
Charcot-Marie-Tooth, Intermediate	KARS	DOMINANT	NM_001130089.1	99.41%
Charcot-Marie-Tooth, Intermediate	MPZ	DOMINANT	NM_000530.6	98.87%
Charcot-Marie-Tooth, Intermediate	YARS	DOMINANT	NM_003680.3	98.09%
Charcot-Marie-Tooth, Type 1	EGR2	DOMINANT	NM_000399.3	99.33%
Charcot-Marie-Tooth, Type 1	LITAF	DOMINANT	NM_004862.3	99.74%
Charcot-Marie-Tooth, Type 1	MPZ	DOMINANT	NM_000530.6	98.87%

Charcot-Marie-Tooth, Type 1	NEFL	DOMINANT	NM_006158.3	99.21%
Charcot-Marie-Tooth, Type 1	PMP22	DOMINANT	NM_000304.2	99.96%
Charcot-Marie-Tooth, Type 2	AARS	DOMINANT	NM_001605.2	99.25%
Charcot-Marie-Tooth, Type 2	DYNC1H1	DOMINANT	NM_001376.4	99.09%
Charcot-Marie-Tooth, Type 2	GARS	DOMINANT	NM_002047.2	99.52%
Charcot-Marie-Tooth, Type 2	HSPB1	DOMINANT	NM_001540.3	97.06%
Charcot-Marie-Tooth, Type 2	HSPB8	DOMINANT	NM_014365.2	98.37%
Charcot-Marie-Tooth, Type 2	KIF1B	DOMINANT	NM_015074.3	98.68%
Charcot-Marie-Tooth, Type 2	LMNA	DOMINANT	NM_005572.3	96.33%
Charcot-Marie-Tooth, Type 2	LRSAM1	DOMINANT	NM_138361.5	98.65%
Charcot-Marie-Tooth, Type 2	MED25	DOMINANT	NM_030973.3	97.67%
Charcot-Marie-Tooth, Type 2	MFN2	DOMINANT	NM_014874.3	98.90%
Charcot-Marie-Tooth, Type 2	RAB7A	DOMINANT	NM_004637.5	98.96%
Charcot-Marie-Tooth, Type 2	TRPV4	DOMINANT	NM_021625.4	97.17%
Charcot-Marie-Tooth, Type 4	FGD4	RECESSIVE	NM_139241.2	99.68%
Charcot-Marie-Tooth, Type 4	FIG4	RECESSIVE	NM_014845.5	99.84%
Charcot-Marie-Tooth, Type 4	MTMR2	RECESSIVE	NM_016156.5	99.95%
Charcot-Marie-Tooth, Type 4	NDRG1	RECESSIVE	NM_006096.3	98.98%
Charcot-Marie-Tooth, Type 4	PRX	RECESSIVE	NM_181882.2	97.48%
Charcot-Marie-Tooth, Type 4	SBF2	RECESSIVE	NM_030962.3	99.40%
Charcot-Marie-Tooth, Type 4	SH3TC2	RECESSIVE	NM_024577.3	99.34%
Charcot-Marie-Tooth, X-linked	PRPS1	X_LINKED	NM_002764.3	88.28%
CHARGE Syndrome	CHD7	DOMINANT	NM_017780.3	98.90%
Chediak-Higashi Syndrome	LYST	RECESSIVE	NM_000081.2	99.72%
Cherubism	SH3BP2	DOMINANT	NM_003023.4	97.69%
Chilblain Lupus	SAMHD1	DOMINANT	NM_015474.3	99.80%
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter	EIF2B1	RECESSIVE	NM_001414.3	98.86%
Childhood Ataxia with Central Nervous System	EIF2B2	RECESSIVE	NM_014239.3	98.62%

Hypomyelination/Vanishing White Matter				
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter	EIF2B3	RECESSIVE	NM_020365.4	99.87%
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter	EIF2B4	RECESSIVE	NM_015636.3	97.88%
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter	EIF2B5	RECESSIVE	NM_003907.2	99.33%
Childhood Restrictive Cardiomyopathy	ACTA1	DOMINANT	NM_001100.3	96.57%
Childhood Restrictive Cardiomyopathy	ACTC1	DOMINANT	NM_005159.4	98.49%
Chitotriosidase Deficiency	CHIT1	RECESSIVE	NM_003465.2	99.81%
Cholesterol Ester Storage Disease	LIPA	RECESSIVE	NM_000235.2	99.79%
Chondrocalcinosis	ANKH	DOMINANT	NM_054027.4	97.18%
Chondrodysplasia	GDF5	RECESSIVE	NM_000557.2	98.65%
Chondrodysplasia	IMPAD1	RECESSIVE	NM_017813.4	97.50%
Chondrodysplasia	PTH1R	RECESSIVE	NM_000316.2	98.57%
Chorea-acanthocytosis	VPS13A	RECESSIVE	NM_033305.2	99.48%
Choreoathetosis, Hypothyroidism, and Neonatal Respiratory Distress	NKX2-1	DOMINANT	NM_001079668.2	95.23%
Choroidal Dystrophy	PRPH2	DOMINANT	NM_000322.4	99.36%
Chronic Granulomatous Disease	NCF2	RECESSIVE	NM_000433.3	99.98%
Chronic Granulomatous Disease	NCF4	RECESSIVE	NM_013416.3	99.28%
Chronic Infantile Neurological Cutaneous and Articular Syndrome	NLRP3	DOMINANT	NM_004895.4	99.48%
Citrin Deficiency	SLC25A13	RECESSIVE	NM_014251.2	98.88%
Citrullinemia	ASS1	RECESSIVE	NM_054012.3	93.61%
Citrullinemia	SLC25A13	RECESSIVE	NM_014251.2	98.88%
Cleft Lip +/- Cleft Palate, Autosomal Dominant	BMP4	DOMINANT	NM_001202.3	98.31%
Cleft Lip +/- Cleft Palate, Autosomal Dominant	IRF6	DOMINANT	NM_006147.3	99.06%
Cleft Lip +/- Cleft Palate, Autosomal Dominant	SUMO1	DOMINANT	NM_001005781.1	98.86%
Cleft Lip +/- Cleft Palate, Autosomal Dominant	TP63	DOMINANT	NM_003722.4	99.84%

Cleft Lip +/- Cleft Palate, Autosomal Recessive	PVRL1	RECESSIVE	NM_002855.4	96.36%
Cleft Palate, X-Linked	TBX22	X_LINKED	NM_001109878.1	95.09%
Cleidocranial Dysplasia	RUNX2	DOMINANT	NM_001024630.3	98.42%
Cockayne Syndrome	ERCC6	RECESSIVE	NM_000124.2	99.64%
Cockayne Syndrome	ERCC8	RECESSIVE	NM_000082.3	98.97%
Coenzyme Q10 deficiency, Oculomotor Apraxia Type	APTX	RECESSIVE	NM_175073.2	99.02%
Coenzyme Q10 deficiency, Spinocerebellar Ataxia Type	ADCK3	RECESSIVE	NM_020247.4	97.82%
Coffin-Siris Syndrome	SMARCA4	DOMINANT	NM_001128849.1	97.97%
Coffin-Siris Syndrome	SMARCB1	DOMINANT	NM_003073.3	95.58%
Cohen Syndrome	VPS13B	RECESSIVE	NM_017890.4	99.40%
Collagen Type VI-Related Disorders	COL6A1	DOMINANT	NM_001848.2	95.68%
Collagen Type VI-Related Disorders	COL6A2	DOMINANT	NM_001849.3	93.40%
Collagen Type VI-Related Disorders	COL6A3	DOMINANT	NM_004369.3	99.74%
Coloboma, Congenital Heart Disease, Ichthyosiform Dermatosis, Mental Retardation, and Ear Anomalies Syndrome	PIGL	RECESSIVE	NM_004278.3	99.98%
Colorectal Cancer	CHEK2	DOMINANT	NM_007194.3	96.19%
Combined Deficiency of Factor V and Factor VIII	LMAN1	RECESSIVE	NM_005570.3	99.80%
Combined Deficiency of Factor V and Factor VIII	MCFD2	RECESSIVE	NM_139279.5	98.27%
Combined Oxidative Phosphorylation Deficiency	AARS2	RECESSIVE	NM_020745.3	98.04%
Combined Oxidative Phosphorylation Deficiency	EARS2	RECESSIVE	NM_001083614.1	98.92%
Combined Oxidative Phosphorylation Deficiency	GFM1	RECESSIVE	NM_024996.5	99.89%
Combined Oxidative Phosphorylation Deficiency	MRPS16	RECESSIVE	NM_016065.3	96.03%
Combined Oxidative Phosphorylation Deficiency	MRPS22	RECESSIVE	NM_020191.2	99.86%
Combined Oxidative Phosphorylation Deficiency	TSFM	RECESSIVE	NM_001172696.1	98.80%
Combined Oxidative Phosphorylation Deficiency	TUFM	RECESSIVE	NM_003321.4	98.25%
Combined Pituitary Hormone Deficiency, Dominant	LHX4	DOMINANT	NM_033343.3	96.63%
Combined Pituitary Hormone Deficiency, Dominant	OTX2	DOMINANT	NM_172337.2	99.73%
Combined Pituitary Hormone Deficiency,	HESX1	DOMINANT	NM_003865.2	99.85%

Dominant/Recessive				
Combined Pituitary Hormone Deficiency, Recessive	LHX3	RECESSIVE	NM_014564.3	95.82%
Combined Pituitary Hormone Deficiency, Recessive	POU1F1	RECESSIVE	NM_000306.2	99.78%
Combined Pituitary Hormone Deficiency, Recessive	PROP1	RECESSIVE	NM_006261.4	97.69%
Combined Saposin Deficiency	PSAP	RECESSIVE	NM_002778.2	99.84%
Common Variable Immune Deficiency, Dominant	TNFRSF13B	DOMINANT	NM_012452.2	97.39%
Common Variable Immune Deficiency, Recessive	CD19	RECESSIVE	NM_001770.5	99.48%
Common Variable Immune Deficiency, Recessive	ICOS	RECESSIVE	NM_012092.3	99.99%
Common Variable Immune Deficiency, Recessive	TNFRSF13C	RECESSIVE	NM_052945.3	97.46%
Complement Component C2 Deficiency	C2	RECESSIVE	NM_000063.4	99.90%
Cone Dystrophy	GUCA1A	DOMINANT	NM_000409.3	99.36%
Cone-Rod Dystrophy, Dominant	CRX	DOMINANT	NM_000554.4	96.57%
Cone-Rod Dystrophy, Dominant	PITPNM3	DOMINANT	NM_031220.3	95.78%
Cone-Rod Dystrophy, Dominant	PROM1	DOMINANT	NM_006017.2	99.87%
Cone-Rod Dystrophy, Dominant	PRPH2	DOMINANT	NM_000322.4	99.36%
Cone-Rod Dystrophy, Dominant	RAX2	DOMINANT	NM_032753.3	98.22%
Cone-Rod Dystrophy, Dominant	RIMS1	DOMINANT	NM_014989.5	99.23%
Cone-Rod Dystrophy, Dominant	UNC119	DOMINANT	NM_005148.3	97.50%
Cone-Rod Dystrophy, Recessive	ABCA4	RECESSIVE	NM_000350.2	99.79%
Cone-Rod Dystrophy, Recessive	ADAM9	RECESSIVE	NM_003816.2	99.19%
Cone-Rod Dystrophy, Recessive	C8orf37	RECESSIVE	NM_177965.3	98.57%
Cone-Rod Dystrophy, Recessive	CDHR1	RECESSIVE	NM_033100.2	99.14%
Cone-Rod Dystrophy, Recessive	PDE6C	RECESSIVE	NM_006204.3	99.96%
Cone-Rod Dystrophy, Recessive	RPGRIP1	RECESSIVE	NM_020366.3	99.78%
Cone-Rod Dystrophy, Recessive	SEMA4A	RECESSIVE	NM_022367.3	99.00%
Congenital Adrenal Hyperplasia	CYP11B1	RECESSIVE	NM_000497.3	96.74%
Congenital Adrenal Hyperplasia	CYP17A1	RECESSIVE	NM_000102.3	99.04%
Congenital Adrenal Hyperplasia	HSD3B2	RECESSIVE	NM_000198.3	99.87%
Congenital Adrenal Hyperplasia	STAR	RECESSIVE	NM_000349.2	98.67%

Congenital Adrenal Insufficiency	CYP11A1	DOMINANT	NM_000781.2	99.90%
Congenital Afibrinogenemia	FGA	RECESSIVE	NM_021871.2	99.96%
Congenital Afibrinogenemia	FGB	RECESSIVE	NM_005141.4	99.19%
Congenital Afibrinogenemia	FGG	RECESSIVE	NM_000509.4	99.98%
Congenital Amegakaryocytic Thrombocytopenia	MPL	RECESSIVE	NM_005373.2	98.41%
Congenital Aural Atresia	TSHZ1	DOMINANT	NM_005786.5	97.11%
Congenital Bile Acid Synthesis Defect	AKR1D1	RECESSIVE	NM_005989.3	99.57%
Congenital Bile Acid Synthesis Defect	CYP7B1	RECESSIVE	NM_004820.3	99.48%
Congenital Cataract	AGK	RECESSIVE	NM_018238.3	99.36%
Congenital Cataract	CRYAA	RECESSIVE	NM_000394.2	98.73%
Congenital Cataract	FYCO1	RECESSIVE	NM_024513.3	98.95%
Congenital Cataract	TDRD7	RECESSIVE	NM_014290.2	97.26%
Congenital Central Hypoventilation Syndrome	PHOX2B	DOMINANT	NM_003924.3	94.04%
Congenital Contractural Arachnodactyly	FBN2	DOMINANT	NM_001999.3	99.56%
Congenital Disorders of Glycosylation	ALG11	RECESSIVE	NM_001004127.2	99.96%
Congenital Disorders of Glycosylation	ALG12	RECESSIVE	NM_024105.3	99.55%
Congenital Disorders of Glycosylation	ALG2	RECESSIVE	NM_033087.3	99.72%
Congenital Disorders of Glycosylation	ALG3	RECESSIVE	NM_005787.5	99.73%
Congenital Disorders of Glycosylation	ALG6	RECESSIVE	NM_013339.3	99.15%
Congenital Disorders of Glycosylation	ALG8	RECESSIVE	NM_024079.4	99.71%
Congenital Disorders of Glycosylation	ALG9	RECESSIVE	NM_024740.2	98.56%
Congenital Disorders of Glycosylation	B4GALT1	RECESSIVE	NM_001497.3	97.53%
Congenital Disorders of Glycosylation	COG1	RECESSIVE	NM_018714.2	96.57%
Congenital Disorders of Glycosylation	COG4	RECESSIVE	NM_015386.2	99.62%
Congenital Disorders of Glycosylation	COG5	RECESSIVE	NM_006348.3	99.75%
Congenital Disorders of Glycosylation	COG6	RECESSIVE	NM_020751.2	99.25%
Congenital Disorders of Glycosylation	COG7	RECESSIVE	NM_153603.3	97.08%
Congenital Disorders of Glycosylation	COG8	RECESSIVE	NM_032382.4	98.67%
Congenital Disorders of Glycosylation	DDOST	RECESSIVE	NM_005216.4	97.10%

Congenital Disorders of Glycosylation	DOLK	RECESSIVE	NM_014908.3	98.59%
Congenital Disorders of Glycosylation	DPAGT1	RECESSIVE	NM_001382.3	99.06%
Congenital Disorders of Glycosylation	DPM1	RECESSIVE	NM_003859.1	99.60%
Congenital Disorders of Glycosylation	DPM3	RECESSIVE	NM_153741.1	99.97%
Congenital Disorders of Glycosylation	MGAT2	RECESSIVE	NM_002408.3	98.19%
Congenital Disorders of Glycosylation	MPDU1	RECESSIVE	NM_004870.3	98.85%
Congenital Disorders of Glycosylation	MPI	RECESSIVE	NM_002435.1	99.90%
Congenital Disorders of Glycosylation	PGM1	RECESSIVE	NM_002633.2	98.32%
Congenital Disorders of Glycosylation	PMM2	RECESSIVE	NM_000303.2	99.34%
Congenital Disorders of Glycosylation	RFT1	RECESSIVE	NM_052859.3	99.19%
Congenital Disorders of Glycosylation	SLC35C1	RECESSIVE	NM_018389.4	95.77%
Congenital Disorders of Glycosylation	SRD5A3	RECESSIVE	NM_024592.4	99.03%
Congenital Disorders of Glycosylation	TUSC3	RECESSIVE	NM_006765.3	99.84%
Congenital Dyserythropoietic Anemia	CDAN1	DOMINANT	NM_138477.2	98.35%
Congenital Dyserythropoietic Anemia	KLF1	DOMINANT	NM_006563.3	96.25%
Congenital Dyserythropoietic Anemia	SEC23B	DOMINANT	NM_006363.4	99.75%
Congenital Erythropoietic Porphyria	UROS	RECESSIVE	NM_000375.2	99.35%
Congenital Fiber-Type Disproportion	ACTA1	DOMINANT	NM_001100.3	96.57%
Congenital Fiber-Type Disproportion	TPM3	DOMINANT	NM_152263.2	98.81%
Congenital Fibrosis of the Extraocular Muscles	KIF21A	DOMINANT	NM_017641.3	99.12%
Congenital Finnish Nephrosis	NPHS1	RECESSIVE	NM_004646.3	98.02%
Congenital Glutamine Deficiency	GLUL	RECESSIVE	NM_002065.5	98.72%
Congenital Hypomyelinating Neuropathy	MPZ	DOMINANT	NM_000530.6	98.87%
Congenital Hypomyelination	MPZ	DOMINANT	NM_000530.6	98.87%
Congenital Hypothyroidism	DUOX2	RECESSIVE	NM_014080.4	96.64%
Congenital Hypothyroidism	IYD	RECESSIVE	NM_203395.2	99.82%
Congenital Hypothyroidism	PAX8	RECESSIVE	NM_003466.3	98.95%
Congenital Hypothyroidism	TPO	RECESSIVE	NM_000547.5	99.12%
Congenital Hypothyroidism	TSHB	RECESSIVE	NM_000549.3	99.97%

Congenital Hypothyroidism	TSHR	RECESSIVE	NM_000369.2	99.89%
Congenital Ichthyosis	ABCA12	RECESSIVE	NM_173076.2	99.82%
Congenital Ichthyosis	ALOX12B	RECESSIVE	NM_001139.2	99.28%
Congenital Ichthyosis	ALOXE3	RECESSIVE	NM_021628.2	99.85%
Congenital Ichthyosis	CYP4F22	RECESSIVE	NM_173483.3	99.02%
Congenital Ichthyosis	NIPAL4	RECESSIVE	NM_001099287.1	99.79%
Congenital Ichthyosis	PNPLA1	RECESSIVE	NM_001145717.1	99.44%
Congenital Ichthyosis	TGM1	RECESSIVE	NM_000359.2	98.49%
Congenital Indifference to Pain	SCN9A	RECESSIVE	NM_002977.3	99.43%
Congenital Insensitivity to Pain with Anhidrosis	NTRK1	RECESSIVE	NM_001012331.1	96.95%
Congenital Lactase Deficiency	LCT	RECESSIVE	NM_002299.2	99.86%
Congenital Muscular Dystrophy, alpha-dystroglycan related	ISPD	RECESSIVE	NM_001101426.3	97.40%
Congenital Muscular Dystrophy, alpha-dystroglycan related	LARGE	RECESSIVE	NM_004737.4	98.32%
Congenital Muscular Dystrophy, alpha-dystroglycan related	POMGNT1	RECESSIVE	NM_017739.3	97.69%
Congenital Muscular Dystrophy, alpha-dystroglycan related	POMT2	RECESSIVE	NM_013382.5	98.20%
Congenital Muscular Dystrophy, CKHB-related	CHKB	RECESSIVE	NM_005198.4	99.30%
Congenital Muscular Dystrophy, ITGA7-related	ITGA7	RECESSIVE	NM_002206.2	96.93%
Congenital Muscular Dystrophy, LAMA2-related	LAMA2	RECESSIVE	NM_000426.3	99.86%
Congenital Muscular Dystrophy, LMNA-related	LMNA	DOMINANT	NM_005572.3	96.33%
Congenital Myasthenic Syndrome, Dominant/Recessive	CHRNA1	DOMINANT	NM_000079.3	99.50%
Congenital Myasthenic Syndrome, Dominant/Recessive	CHRNB1	DOMINANT	NM_000747.2	98.50%
Congenital Myasthenic Syndrome, Dominant/Recessive	CHRND	DOMINANT	NM_000751.2	98.75%
Congenital Myasthenic Syndrome, Dominant/Recessive	CHRNE	DOMINANT	NM_000080.3	97.70%
Congenital Myasthenic Syndrome, Recessive	COLQ	RECESSIVE	NM_005677.3	99.52%
Congenital Myasthenic Syndrome, Recessive	GFPT1	RECESSIVE	NM_002056.3	98.71%
Congenital Myasthenic Syndrome, Recessive	MUSK	RECESSIVE	NM_005592.3	99.98%
Congenital Myasthenic Syndrome, Recessive	RAPSN	RECESSIVE	NM_005055.4	98.74%
Congenital Myasthenic Syndrome, Recessive	SCN4A	RECESSIVE	NM_000334.4	97.03%
Congenital Neuromuscular Disease with Uniform Type 1	RYR1	DOMINANT	NM_000540.2	97.36%

Fiber				
Congenital Nuclear Cataract	CRYBB1	RECESSIVE	NM_001887.3	99.01%
Congenital Nuclear Cataract	CRYBB3	RECESSIVE	NM_004076.3	97.42%
Congenital Stationary Night Blindness, Dominant	GNAT1	DOMINANT	NM_144499.2	99.54%
Congenital Stationary Night Blindness, Dominant	PDE6B	DOMINANT	NM_000283.3	97.01%
Congenital Stationary Night Blindness, Dominant	RHO	DOMINANT	NM_000539.3	99.45%
Congenital Stationary Night Blindness, Recessive	CABP4	RECESSIVE	NM_145200.3	97.64%
Congenital Stationary Night Blindness, Recessive	GPR179	RECESSIVE	NM_001004334.2	99.21%
Congenital Stationary Night Blindness, Recessive	SLC24A1	RECESSIVE	NM_004727.2	98.79%
Congenital Stationary Night Blindness, Recessive	TRPM1	RECESSIVE	NM_002420.5	99.73%
Congenital Stromal Corneal Dystrophy	DCN	DOMINANT	NM_001920.3	99.96%
Congenital Sucrase-Isomaltase Deficiency	SI	RECESSIVE	NM_001041.3	99.21%
Congenital Vertical Talus	HOXD10	DOMINANT	NM_002148.3	95.31%
Corneal Dystrophy, Dominant	TGFBI	DOMINANT	NM_000358.2	98.44%
Corneal Dystrophy, Dominant/Recessive	TACSTD2	RECESSIVE	NM_002353.2	97.25%
Corneal Dystrophy, Recessive	CYP4V2	RECESSIVE	NM_207352.3	98.99%
Corneal Dystrophy, Recessive	SLC4A11	RECESSIVE	NM_032034.3	97.53%
Corneal Fleck Dystrophy	PIKFYVE	DOMINANT	NM_015040.3	99.79%
Cornelia de Lange Syndrome	NIPBL	DOMINANT	NM_133433.3	98.16%
Cornelia de Lange Syndrome	SMC1A	DOMINANT	NM_006306.2	95.69%
Cornelia de Lange Syndrome	SMC3	DOMINANT	NM_005445.3	99.68%
Cortical Dysplasia-Focal Epilepsy Syndrome	CNTNAP2	RECESSIVE	NM_014141.5	99.23%
Cortical Pulverulent Cataract	LIM2	RECESSIVE	NM_030657.3	99.81%
Corticosterone Methyloxidase Type I Deficiency	CYP11B2	RECESSIVE	NM_000498.3	95.83%
Corticosterone Methyloxidase Type II Deficiency	CYP11B2	RECESSIVE	NM_000498.3	95.83%
Cranioectodermal Dysplasia	IFT122	RECESSIVE	NM_052985.2	96.37%
Cranioectodermal Dysplasia	IFT43	RECESSIVE	NM_052873.2	99.69%
Cranioectodermal Dysplasia	WDR19	RECESSIVE	NM_025132.3	99.31%
Cranioectodermal Dysplasia	WDR35	RECESSIVE	NM_001006657.1	99.96%

Craniofacial-Deafness-Hand Syndrome	PAX3	DOMINANT	NM_181457.3	97.63%
Craniometaphyseal Dysplasia	ANKH	DOMINANT	NM_054027.4	97.18%
Craniosynostosis	FGFR1	DOMINANT	NM_023110.2	98.79%
Craniosynostosis	FGFR2	DOMINANT	NM_000141.4	97.52%
Craniosynostosis	MSX2	DOMINANT	NM_002449.4	97.31%
Creatine Deficiency Syndromes	GAMT	RECESSIVE	NM_000156.4	97.97%
Creatine Deficiency Syndromes	GATM	RECESSIVE	NM_001482.2	99.69%
Crigler-Najjar Syndrome	UGT1A1	RECESSIVE	NM_000463.2	99.91%
Crohn Disease	NOD2	DOMINANT	NM_022162.1	99.39%
Crouzon Syndrome	FGFR2	DOMINANT	NM_000141.4	97.52%
Cutaneous Malignant Melanoma, Dominant	CDK4	DOMINANT	NM_000075.2	99.37%
Cutaneous Malignant Melanoma, Recessive	MC1R	RECESSIVE	NM_002386.3	95.94%
Cutis Laxa with Severe Pulmonary, Gastrointestinal, and Urinary Abnormalities	LTBP4	RECESSIVE	NM_003573.2	95.27%
Cutis Laxa, Dominant	ELN	DOMINANT	NM_000501.2	97.00%
Cutis Laxa, Dominant/Recessive	FBLN5	DOMINANT	NM_006329.3	98.82%
Cutis Laxa, Recessive	ALDH18A1	RECESSIVE	NM_002860.3	99.75%
Cutis Laxa, Recessive	ATP6V0A2	RECESSIVE	NM_012463.3	98.78%
Cutis Laxa, Recessive	PYCR1	RECESSIVE	NM_006907.2	99.38%
Cystathioninuria	CTH	RECESSIVE	NM_001902.5	98.95%
Cystic Fibrosis	CFTR	RECESSIVE	NM_000492.3	99.85%
Cystic Fibrosis-Like Syndrome	SCNN1A	DOMINANT	NM_001038.5	97.55%
Cystinosis	CTNS	RECESSIVE	NM_004937.2	97.66%
Cystinuria	SLC3A1	RECESSIVE	NM_000341.3	99.94%
Cystinuria	SLC7A9	RECESSIVE	NM_014270.4	99.62%
Cytochrome P450 Oxidoreductase Deficiency	POR	RECESSIVE	NM_000941.2	96.88%
D-2-Hydroxyglutaric Aciduria	D2HGDH	DOMINANT	NM_152783.3	95.88%
Danon Disease	LAMP2	X_LINKED	NM_002294.2	94.75%
Darier-White Disease	ATP2A2	DOMINANT	NM_001681.3	96.99%

Delta-Sarcoglycanopathy	SGCD	RECESSIVE	NM_000337.5	99.33%
Dementia, Deafness, and Sensory Neuropathy	DNMT1	DOMINANT	NM_001130823.1	99.26%
Dense Deposit Disease / Membranoproliferative Glomerulonephritis Type II	CFH	RECESSIVE	NM_000186.3	98.32%
Dense Deposit Disease / Membranoproliferative Glomerulonephritis Type II	CFHR5	RECESSIVE	NM_030787.3	98.77%
Dent Disease	CLCN5	X_LINKED	NM_000084.2	96.78%
Desbuquois Dysplasia	CANT1	RECESSIVE	NM_138793.3	96.43%
Desminopathy	DES	DOMINANT	NM_001927.3	96.15%
Desmosterolosis	DHCR24	RECESSIVE	NM_014762.3	99.41%
Diabetes Mellitus, Insulin-Resistant, with Acanthosis Nigricans	INSR	DOMINANT	NM_000208.2	97.67%
Diabetes Mellitus, Neonatal, with Congenital Hypothyroidism	GLIS3	RECESSIVE	NM_001042413.1	98.52%
Diabetes Mellitus, Noninsulin-Dependent, with Acanthosis Nigricans and Hypertension	PPARG	DOMINANT	NM_015869.4	99.99%
Diabetes, Transient Neonatal	ZFP57	RECESSIVE	NM_001109809.2	99.81%
Diamond-Blackfan Anemia	RPL11	DOMINANT	NM_000975.3	99.99%
Diamond-Blackfan Anemia	RPL35A	DOMINANT	NM_000996.2	100.00%
Diamond-Blackfan Anemia	RPL5	DOMINANT	NM_000969.3	99.52%
Diamond-Blackfan Anemia	RPS10	DOMINANT	NM_001014.4	99.53%
Diamond-Blackfan Anemia	RPS19	DOMINANT	NM_001022.3	99.07%
Diamond-Blackfan Anemia	RPS24	DOMINANT	NM_033022.3	99.33%
Diamond-Blackfan Anemia	RPS26	DOMINANT	NM_001029.3	99.82%
Diamond-Blackfan Anemia	RPS7	DOMINANT	NM_001011.3	99.76%
Diaphanospondylodysostosis	BMPER	RECESSIVE	NM_133468.4	97.30%
Diaphyseal Medullary Stenosis with Malignant Fibrous Histiocytoma	MTAP	DOMINANT	NM_002451.3	98.90%
Diarrhea with Microvillus Atrophy	MYO5B	RECESSIVE	NM_001080467.2	97.26%
Diastrophic Dysplasia	SLC26A2	RECESSIVE	NM_000112.3	99.63%

Dicarboxylicaminoaciduria	SLC1A1	RECESSIVE	NM_004170.5	99.15%
Diffuse Mesangial Sclerosis Syndromes (DMS)	WT1	DOMINANT	NM_024426.4	95.44%
Dihydropyrimidinase Deficiency	DPYS	RECESSIVE	NM_001385.2	99.14%
Dihydropyrimidine Dehydrogenase Deficiency	DPYD	RECESSIVE	NM_000110.3	99.68%
Dilated Cardiomyopathy with Quadriceps Myopathy	LMNA	DOMINANT	NM_005572.3	96.33%
Dilated Cardiomyopathy, Dominant	ABCC9	DOMINANT	NM_005691.2	99.66%
Dilated Cardiomyopathy, Dominant	ACTC1	DOMINANT	NM_005159.4	98.49%
Dilated Cardiomyopathy, Dominant	ACTN2	DOMINANT	NM_001103.2	97.96%
Dilated Cardiomyopathy, Dominant	ANKRD1	DOMINANT	NM_014391.2	97.05%
Dilated Cardiomyopathy, Dominant	BAG3	DOMINANT	NM_004281.3	97.40%
Dilated Cardiomyopathy, Dominant	CSRP3	DOMINANT	NM_003476.4	97.72%
Dilated Cardiomyopathy, Dominant	CTF1	DOMINANT	NM_001330.3	88.41%
Dilated Cardiomyopathy, Dominant	DES	DOMINANT	NM_001927.3	96.15%
Dilated Cardiomyopathy, Dominant	DSG2	DOMINANT	NM_001943.3	98.52%
Dilated Cardiomyopathy, Dominant	EYA4	DOMINANT	NM_172105.3	99.82%
Dilated Cardiomyopathy, Dominant	LDB3	DOMINANT	NM_001080116.1	99.33%
Dilated Cardiomyopathy, Dominant	LMNA	DOMINANT	NM_005572.3	96.33%
Dilated Cardiomyopathy, Dominant	MYBPC3	DOMINANT	NM_000256.3	98.60%
Dilated Cardiomyopathy, Dominant	MYH6	DOMINANT	NM_002471.3	98.59%
Dilated Cardiomyopathy, Dominant	MYH7	DOMINANT	NM_000257.2	99.12%
Dilated Cardiomyopathy, Dominant	NEXN	DOMINANT	NM_144573.3	99.09%
Dilated Cardiomyopathy, Dominant	PLN	DOMINANT	NM_002667.3	99.99%
Dilated Cardiomyopathy, Dominant	PSEN1	DOMINANT	NM_000021.3	99.77%
Dilated Cardiomyopathy, Dominant	PSEN2	DOMINANT	NM_000447.2	98.87%
Dilated Cardiomyopathy, Dominant	RBM20	DOMINANT	NM_001134363.1	99.49%
Dilated Cardiomyopathy, Dominant	SCN5A	DOMINANT	NM_198056.2	98.54%
Dilated Cardiomyopathy, Dominant	SGCD	DOMINANT	NM_000337.5	99.33%
Dilated Cardiomyopathy, Dominant	TCAP	DOMINANT	NM_003673.3	98.58%
Dilated Cardiomyopathy, Dominant	TMPO	DOMINANT	NM_003276.2	98.51%

Dilated Cardiomyopathy, Dominant	TNNC1	DOMINANT	NM_003280.2	98.67%
Dilated Cardiomyopathy, Dominant	TNNT2	DOMINANT	NM_001001430.1	99.40%
Dilated Cardiomyopathy, Dominant	TPM1	DOMINANT	NM_001018005.1	97.66%
Dilated Cardiomyopathy, Dominant	TTN	DOMINANT	NM_133378.4	99.55%
Dilated Cardiomyopathy, Dominant	VCL	DOMINANT	NM_014000.2	99.09%
Dilated Cardiomyopathy, Recessive	FKTN	RECESSIVE	NM_001079802.1	99.86%
Dilated Cardiomyopathy, Recessive	TNNI3	RECESSIVE	NM_000363.4	98.74%
Dilated Cardiomyopathy, X-Linked	DMD	X_LINKED	NM_004006.2	94.87%
Dilated Cardiomyopathy, X-Linked	TAZ	X_LINKED	NM_000116.3	90.55%
Disorders of Intracellular Cobalamin Metabolism	LMBRD1	RECESSIVE	NM_018368.3	98.54%
Disorders of Intracellular Cobalamin Metabolism	MMACHC	RECESSIVE	NM_015506.2	96.82%
Disorders of Intracellular Cobalamin Metabolism	MMADHC	RECESSIVE	NM_015702.2	99.95%
Disorders of Intracellular Cobalamin Metabolism	MTR	RECESSIVE	NM_000254.2	99.40%
Disorders of Intracellular Cobalamin Metabolism	MTRR	RECESSIVE	NM_002454.2	99.86%
Distal Arthrogryposis	MYBPC1	DOMINANT	NM_002465.3	99.80%
Distal Arthrogryposis	MYH3	DOMINANT	NM_002470.3	99.45%
Distal Arthrogryposis Multiplex Congenita	TNNI2	DOMINANT	NM_003282.3	96.15%
Distal Arthrogryposis Multiplex Congenita	TNNT3	DOMINANT	NM_006757.3	99.33%
Distal Congenital Nonprogressive Spinal Muscular Atrophy	TRPV4	DOMINANT	NM_021625.4	97.17%
Distal Hereditary Motor Neuronopathy	DCTN1	DOMINANT	NM_004082.4	99.86%
Distal Hereditary Motor Neuronopathy	HSPB1	DOMINANT	NM_001540.3	97.06%
Distal Hereditary Motor Neuronopathy	HSPB3	DOMINANT	NM_006308.2	97.81%
Distal Hereditary Motor Neuronopathy	HSPB8	DOMINANT	NM_014365.2	98.37%
Distal Myopathy	MATR3	DOMINANT	NM_199189.2	98.61%
Distal Renal Tubular Acidosis with Progressive Nerve Deafness	ATP6V1B1	RECESSIVE	NM_001692.3	98.98%
Distal Renal Tubular Acidosis, Dominant	SLC4A1	DOMINANT	NM_000342.3	97.69%
Distal Renal Tubular Acidosis, Recessive	ATP6V0A4	RECESSIVE	NM_020632.2	99.74%
Distal Spinal Muscular Atrophy	GARS	DOMINANT	NM_002047.2	99.52%

Distal Spinal Muscular Atrophy	PLEKHG5	RECESSIVE	NM_020631.4	95.76%
Donnai-Barrow Syndrome	LRP2	RECESSIVE	NM_004525.2	99.52%
Donohue Syndrome	INSR	RECESSIVE	NM_000208.2	97.67%
Dopamine Beta-Hydroxylase Deficiency	DBH	RECESSIVE	NM_000787.3	97.73%
Dopa-Responsive Dystonia	GCH1	DOMINANT	NM_000161.2	98.40%
Dopa-Responsive Dystonia	SPR	DOMINANT	NM_003124.4	99.62%
Doyne Honeycomb Retinal Dystrophy	EFEMP1	DOMINANT	NM_001039348.2	99.35%
Dravet Syndrome	GABRG2	DOMINANT	NM_000816.3	99.93%
Dravet Syndrome	SCN9A	DOMINANT	NM_002977.3	99.43%
DRPLA	ATN1	DOMINANT	NM_001007026.1	96.76%
Duane Syndrome	CHN1	DOMINANT	NM_001822.5	96.75%
Dubin-Johnson Syndrome	ABCC2	RECESSIVE	NM_000392.3	99.58%
Dyggve-Melchior-Clausen Syndrome	DYM	RECESSIVE	NM_017653.3	99.04%
Dysalbuminemic Hyperthyroxinemia	ALB	DOMINANT	NM_000477.5	99.99%
Dyschromatosis Symmetrica Hereditaria	ADAR	DOMINANT	NM_001111.4	99.10%
Dyskeratosis Congenita, Dominant	TINF2	DOMINANT	NM_001099274.1	99.77%
Dyskeratosis Congenita, Recessive	CTC1	RECESSIVE	NM_025099.5	99.22%
Dyskeratosis Congenita, Recessive	NHP2	RECESSIVE	NM_017838.3	97.99%
Dyskeratosis Congenita, Recessive	NOP10	RECESSIVE	NM_018648.3	98.96%
Dyskeratosis Congenita, Recessive	WRAP53	RECESSIVE	NM_018081.2	99.09%
Dyssegmental Dysplasia	HSPG2	RECESSIVE	NM_005529.5	96.57%
Dystonia	PRKRA	DOMINANT	NM_003690.4	99.45%
Dystonia	SLC2A1	DOMINANT	NM_006516.2	98.39%
Dystonia	THAP1	DOMINANT	NM_018105.2	98.59%
Dystonia/Parkinsonism, Hypermanganesemia, Polycythemia, and Chronic Liver Disease	SLC30A10	RECESSIVE	NM_018713.2	97.84%
Dystrophic Epidermolysis Bullosa	COL7A1	DOMINANT	NM_000094.3	98.71%
Early Infantile Epileptic Encephalopathy, Autosomal Dominant	SCN2A	DOMINANT	NM_021007.2	98.70%

Early Infantile Epileptic Encephalopathy, Autosomal Dominant	SCN8A	DOMINANT	NM_014191.3	98.58%
Early Infantile Epileptic Encephalopathy, Autosomal Dominant	SPTAN1	DOMINANT	NM_001130438.2	99.32%
Early Infantile Epileptic Encephalopathy, Autosomal Dominant	STXBP1	DOMINANT	NM_003165.3	98.02%
Early Infantile Epileptic Encephalopathy, Autosomal Recessive	PLCB1	RECESSIVE	NM_015192.2	99.85%
Early Infantile Epileptic Encephalopathy, Autosomal Recessive	SLC25A22	RECESSIVE	NM_024698.5	97.80%
Early-Onset Primary Dystonia	TOR1A	DOMINANT	NM_000113.2	98.82%
Ectodermal Dysplasia, Anhidrotic, with T-cell Immunodeficiency	NFKBIA	DOMINANT	NM_020529.2	98.11%
Ectodermal Dysplasia/Skin Fragility Syndrome	DSP	RECESSIVE	NM_004415.2	99.77%
Ectodermal Dysplasia/Skin Fragility Syndrome	PKP1	RECESSIVE	NM_001005337.2	99.04%
Ectopia Lentis	ADAMTSL4	RECESSIVE	NM_019032.4	98.41%
Ectopia Lentis	FBN1	RECESSIVE	NM_000138.4	99.59%
Ectrodactyly	TP63	DOMINANT	NM_003722.4	99.84%
EEM Syndrome	CDH3	RECESSIVE	NM_001793.4	98.64%
Ehlers-Danlos Syndrome, Arthrochalasia Type	ADAMTS2	DOMINANT	NM_014244.4	95.31%
Ehlers-Danlos Syndrome, Arthrochalasia Type	COL1A2	DOMINANT	NM_000089.3	98.95%
Ehlers-Danlos Syndrome, Classic	COL1A1	DOMINANT	NM_000088.3	96.26%
Ehlers-Danlos Syndrome, Classic	COL5A1	DOMINANT	NM_000093.3	97.68%
Ehlers-Danlos Syndrome, Classic	COL5A2	DOMINANT	NM_000393.3	99.52%
Ehlers-Danlos Syndrome, Kyphoscoliotic Form	PLOD1	RECESSIVE	NM_000302.3	97.78%
Ehlers-Danlos Syndrome, Vascular Type	COL3A1	DOMINANT	NM_000090.3	99.78%
Elliptocytosis	SPTA1	DOMINANT	NM_003126.2	99.65%
Elliptocytosis	SPTB	DOMINANT	NM_000347.5	98.26%
Ellis-van Creveld Syndrome	EVC	RECESSIVE	NM_153717.2	95.39%
Ellis-van Creveld Syndrome	EVC2	RECESSIVE	NM_147127.4	98.46%

Emery-Dreifuss Muscular Dystrophy	LMNA	DOMINANT	NM_005572.3	96.33%
Emery-Dreifuss Muscular Dystrophy	SYNE1	DOMINANT	NM_033071.3	99.70%
Emery-Dreifuss Muscular Dystrophy	SYNE2	DOMINANT	NM_182914.2	99.46%
Endocardial Fibroelastosis	TAZ	DOMINANT	NM_000116.3	90.55%
Enhanced S-Cone Syndrome	NR2E3	RECESSIVE	NM_014249.2	98.63%
Enlarged Parietal Foramina	ALX4	DOMINANT	NM_021926.3	98.09%
Enlarged Parietal Foramina	MSX2	DOMINANT	NM_002449.4	97.31%
Epidermolysis Bullosa Simplex	KRT5	DOMINANT	NM_000424.3	97.06%
Epidermolysis Bullosa with Pyloric Atresia	ITGA6	RECESSIVE	NM_000210.2	99.82%
Epidermolysis Bullosa with Pyloric Atresia	ITGB4	RECESSIVE	NM_001005731.1	96.66%
Epidermolysis Bullosa, Lethal Acantholytic	DSP	RECESSIVE	NM_004415.2	99.77%
Epidermolytic Hyperkeratosis	KRT1	DOMINANT	NM_006121.3	96.60%
Epidermolytic Palmoplantar Keratoderma	KRT9	DOMINANT	NM_000226.3	96.94%
Epilepsy with Neurodevelopmental Defects	GRIN2A	DOMINANT	NM_000833.3	99.38%
Epileptic Encephalopathy	MAPK10	DOMINANT	NM_138982.2	99.81%
Epimerase Deficiency Galactosemia	GALE	RECESSIVE	NM_000403.3	97.56%
Episodic Ataxia	CACNB4	DOMINANT	NM_000726.3	99.76%
Episodic Ataxia	KCNA1	DOMINANT	NM_000217.2	98.67%
Episodic Ataxia	SLC1A3	DOMINANT	NM_004172.4	99.88%
Epstein Syndrome	MYH9	DOMINANT	NM_002473.4	99.28%
Erosive Vitreoretinopathy	VCAN	DOMINANT	NM_004385.4	99.60%
Erythrocyte AMP Deaminase Deficiency	AMPD3	RECESSIVE	NM_001025389.1	98.97%
Erythrokeratodermia Variabilis	GJB3	DOMINANT	NM_024009.2	96.05%
Erythropoietic Protoporphyria	FECH	DOMINANT	NM_000140.3	98.21%
Escobar Syndrome	CHRNG	RECESSIVE	NM_005199.4	96.52%
Essential Fructosuria	KHK	RECESSIVE	NM_000221.2	98.46%
Essential Thrombocythemia	MPL	DOMINANT	NM_005373.2	98.41%
Essential Thrombocythemia	THPO	DOMINANT	NM_000460.2	99.93%
Ethylmalonic Encephalopathy	ETHE1	RECESSIVE	NM_014297.3	98.86%

Exocrine Pancreatic Insufficiency, Dyserythropoietic Anemia, and Calvarial Hyperostosis	COX4I2	RECESSIVE	NM_032609.2	99.59%
Fabry Disease	GLA	X_LINKED	NM_000169.2	96.24%
Factor V Cambridge Thrombophilia	F5	DOMINANT	NM_000130.4	99.15%
Factor V Deficiency	F5	RECESSIVE	NM_000130.4	99.15%
Factor V Leiden Thrombophilia	F5	DOMINANT	NM_000130.4	99.15%
Factor V R2 Mutation Thrombophilia	F5	DOMINANT	NM_000130.4	99.15%
Factor VII Deficiency	F7	RECESSIVE	NM_000131.3	90.75%
Factor VII Marburg I Variant Thrombophilia	HABP2	DOMINANT	NM_004132.3	99.73%
Factor X Deficiency	F10	RECESSIVE	NM_000504.3	98.91%
Factor XI Deficiency	F11	DOMINANT	NM_000128.3	99.50%
Factor XII Deficiency	F12	DOMINANT	NM_000505.3	98.74%
Factor XIII Subunit A Deficiency	F13A1	RECESSIVE	NM_000129.3	99.76%
Factor XIII Subunit B Deficiency	F13B	RECESSIVE	NM_001994.2	99.82%
Familial Atrial Fibrillation	ABCC9	DOMINANT	NM_005691.2	99.66%
Familial Atrial Fibrillation	GJA5	DOMINANT	NM_005266.5	99.89%
Familial Atrial Fibrillation	KCNA5	DOMINANT	NM_002234.3	97.79%
Familial Atrial Fibrillation	KCNE2	DOMINANT	NM_172201.1	100.00%
Familial Atrial Fibrillation	KCNJ2	DOMINANT	NM_000891.2	98.32%
Familial Atrial Fibrillation	KCNQ1	DOMINANT	NM_000218.2	92.10%
Familial Atypical Mycobacteriosis, Autosomal Dominant	STAT1	DOMINANT	NM_007315.3	99.81%
Familial Atypical Mycobacteriosis, Autosomal Recessive	IFNGR1	RECESSIVE	NM_000416.2	99.55%
Familial Atypical Mycobacteriosis, Autosomal Recessive	IL12B	RECESSIVE	NM_002187.2	99.64%
Familial Atypical Mycobacteriosis, Autosomal Recessive	IL12RB1	RECESSIVE	NM_005535.1	98.50%
Familial Atypical Mycobacteriosis, Autosomal Recessive	TYK2	RECESSIVE	NM_003331.4	98.05%
Familial Bone Marrow Failure	SRP72	DOMINANT	NM_006947.3	96.41%
Familial Candidiasis, Dominant	IL17F	DOMINANT	NM_052872.3	99.84%
Familial Candidiasis, Recessive	IL17RA	RECESSIVE	NM_014339.5	98.31%
Familial Cerebral Cavernous Malformation	KRIT1	DOMINANT	NM_194456.1	99.20%

Familial Cerebral Cavernous Malformation	PDCD10	DOMINANT	NM_145860.1	99.44%
Familial Chloride Diarrhea	SLC26A3	RECESSIVE	NM_000111.2	99.99%
Familial Cold Autoinflammatory Syndrome	NLRP12	DOMINANT	NM_144687.2	98.84%
Familial Cold Autoinflammatory Syndrome	NLRP3	DOMINANT	NM_004895.4	99.48%
Familial Cylindromatosis	CYLD	DOMINANT	NM_015247.2	99.42%
Familial Dysautonomia	IKBKAP	RECESSIVE	NM_003640.3	99.55%
Familial Encephalopathy with Neuroserpin Inclusion Bodies	SERPINI1	DOMINANT	NM_005025.4	99.98%
Familial Erythrocytosis	EGLN1	DOMINANT	NM_022051.2	97.05%
Familial Erythrocytosis	EPAS1	DOMINANT	NM_001430.4	98.07%
Familial Erythrocytosis	EPOR	DOMINANT	NM_000121.3	98.88%
Familial Exudative Vitreoretinopathy	FZD4	DOMINANT	NM_012193.3	99.42%
Familial Exudative Vitreoretinopathy	TSPAN12	DOMINANT	NM_012338.3	97.51%
Familial Febrile Seizures	SCN9A	DOMINANT	NM_002977.3	99.43%
Familial Hemiplegic Migraine	ATP1A2	DOMINANT	NM_000702.3	98.69%
Familial Hemiplegic Migraine	SCN1A	DOMINANT	NM_001165963.1	99.76%
Familial Hemophagocytic Lymphohistiocytosis	PRF1	RECESSIVE	NM_001083116.1	98.25%
Familial Hemophagocytic Lymphohistiocytosis	STX11	RECESSIVE	NM_003764.3	97.28%
Familial Hemophagocytic Lymphohistiocytosis	STXBP2	RECESSIVE	NM_006949.2	97.56%
Familial Hemophagocytic Lymphohistiocytosis	UNC13D	RECESSIVE	NM_199242.2	97.37%
Familial High Density Lipoprotein Deficiency	ABCA1	DOMINANT	NM_005502.3	99.11%
Familial High Density Lipoprotein Deficiency	APOA1	DOMINANT	NM_000039.1	99.23%
Familial Horizontal Gaze Palsy with Progressive Scoliosis	ROBO3	RECESSIVE	NM_022370.3	98.85%
Familial Hyperaldosteronism	KCNJ5	DOMINANT	NM_000890.3	95.90%
Familial Hypercholesterolemia	APOB	DOMINANT	NM_000384.2	99.52%
Familial Hypercholesterolemia	LDLR	DOMINANT	NM_000527.4	98.90%
Familial Hypercholesterolemia	LDLRAP1	DOMINANT	NM_015627.2	96.72%
Familial Hypercholesterolemia	PCSK9	DOMINANT	NM_174936.3	97.07%
Familial Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome	PRKAG2	DOMINANT	NM_016203.3	97.03%

Familial Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome	TNNI3	DOMINANT	NM_000363.4	98.74%
Familial Hypocalciuric Hypercalcemia	CASR	DOMINANT	NM_000388.3	99.26%
Familial Idiopathic Basal Ganglia Calcification	SLC20A2	DOMINANT	NM_006749.4	98.70%
Familial Infantile Myoclonic Epilepsy	TBC1D24	RECESSIVE	NM_020705.2	96.57%
Familial Intrahepatic Cholestasis	ABCB11	RECESSIVE	NM_003742.2	99.85%
Familial Intrahepatic Cholestasis	ABCB4	RECESSIVE	NM_000443.3	99.79%
Familial Intrahepatic Cholestasis	ATP8B1	RECESSIVE	NM_005603.4	99.49%
Familial Isolated Hypoparathyroidism	CASR	DOMINANT	NM_000388.3	99.26%
Familial Isolated Hypoparathyroidism	GCM2	DOMINANT	NM_004752.3	99.64%
Familial Isolated Hypoparathyroidism	PTH	DOMINANT	NM_000315.2	99.99%
Familial Isolated Pituitary Adenomas	AIP	DOMINANT	NM_003977.2	97.89%
Familial Juvenile Hyperuricemic Nephropathy	REN	DOMINANT	NM_000537.3	99.84%
Familial Lipoprotein Lipase Deficiency	LPL	RECESSIVE	NM_000237.2	99.94%
Familial Mediterranean Fever	MEFV	RECESSIVE	NM_000243.2	94.09%
Familial Paroxysmal Nonkinesigenic Dyskinesia	PNKD	DOMINANT	NM_015488.4	98.25%
Familial Partial Lipodystrophy	LMNA	DOMINANT	NM_005572.3	96.33%
Familial Partial Lipodystrophy	PPARG	DOMINANT	NM_015869.4	99.99%
Familial Periodic Fever	TNFRSF1A	DOMINANT	NM_001065.3	98.92%
Familial Platelet Disorder with Propensity to Acute Myelogenous Leukemia	RUNX1	DOMINANT	NM_001754.4	97.08%
Familial Pulmonary Fibrosis	SFTPC	DOMINANT	NM_003018.3	98.49%
Familial Restrictive Cardiomyopathy	TNNI3	DOMINANT	NM_000363.4	98.74%
Familial Restrictive Cardiomyopathy	TNNT2	DOMINANT	NM_001001430.1	99.40%
Familial Spinal Neurofibromatosis	NF1	DOMINANT	NM_000267.3	98.11%
Familial Temporal Lobe Epilepsy	CPA6	DOMINANT	NM_020361.4	99.98%
Familial Thrombotic Thrombocytopenia Purpura	ADAMTS13	RECESSIVE	NM_139025.3	96.40%
Familial Transthyretin Amyloidosis	TTR	DOMINANT	NM_000371.3	98.86%
Familial Visceral Amyloidosis	APOA1	DOMINANT	NM_000039.1	99.23%

Familial Visceral Amyloidosis	FGA	DOMINANT	NM_021871.2	99.96%
Familial Visceral Amyloidosis	LYZ	DOMINANT	NM_000239.2	98.33%
Fanconi Anemia	BRCA2	RECESSIVE	NM_000059.3	99.97%
Fanconi Anemia	BRIP1	RECESSIVE	NM_032043.2	98.05%
Fanconi Anemia	FANCA	RECESSIVE	NM_000135.2	99.09%
Fanconi Anemia	FANCC	RECESSIVE	NM_000136.2	99.79%
Fanconi Anemia	FANCD2	RECESSIVE	NM_033084.3	98.39%
Fanconi Anemia	FANCE	RECESSIVE	NM_021922.2	98.41%
Fanconi Anemia	FANCF	RECESSIVE	NM_022725.3	99.42%
Fanconi Anemia	FANCG	RECESSIVE	NM_004629.1	98.99%
Fanconi Anemia	FANCI	RECESSIVE	NM_001113378.1	99.80%
Fanconi Anemia	FANCL	RECESSIVE	NM_001114636.1	99.95%
Fanconi Anemia	FANCM	RECESSIVE	NM_020937.2	99.02%
Fanconi Anemia	PALB2	RECESSIVE	NM_024675.3	99.95%
Fanconi Anemia	RAD51C	RECESSIVE	NM_058216.1	98.27%
Fanconi Anemia	SLX4	RECESSIVE	NM_032444.2	99.31%
Fanconi Anemia, X-Linked	FANCB	X_LINKED	NM_001018113.1	94.19%
Fanconi-Bickel Syndrome	SLC2A2	RECESSIVE	NM_000340.1	99.96%
Farber Lipogranulomatosis	ASAH1	RECESSIVE	NM_177924.3	99.36%
Fatal Infantile Cardioencephalomyopathy	SCO2	RECESSIVE	NM_005138.2	97.71%
Fatal Infantile Cardioencephalomyopathy	SCO2	RECESSIVE	NM_005138.2	97.71%
Fatal Infantile Lactic Acidosis	SUCLG1	RECESSIVE	NM_003849.3	98.92%
Fatty Acid Hydroxylase-Associated Neurodegeneration	FA2H	RECESSIVE	NM_024306.4	97.26%
Fechtner Syndrome	MYH9	DOMINANT	NM_002473.4	99.28%
Fetal Akinesia Deformation Sequence	RAPSN	RECESSIVE	NM_005055.4	98.74%
Fibrochondrogenesis	COL11A1	RECESSIVE	NM_001854.3	98.66%
Fibrochondrogenesis	COL11A2	RECESSIVE	NM_080680.2	99.38%
Fibrodysplasia Ossificans Progressiva	ACVR1	DOMINANT	NM_001105.4	99.91%
Fibular Hypoplasia and Complex Brachydactyly	GDF5	RECESSIVE	NM_000557.2	98.65%

FLNB-Related Spectrum Disorders	FLNB	DOMINANT	NM_001457.3	98.53%
Floating-Harbor Syndrome	SRCAP	DOMINANT	NM_006662.2	99.46%
Focal Cortical Dysplasia of Taylor	TSC1	DOMINANT	NM_000368.4	99.02%
Focal Segmental Glomerulosclerosis	CD2AP	RECESSIVE	NM_012120.2	98.03%
Focal Segmental Glomerulosclerosis	TRPC6	RECESSIVE	NM_004621.5	98.55%
Formiminotransferase Deficiency	FTCD	RECESSIVE	NM_006657.2	95.57%
Foveal Hypoplasia and Presenile Cataract Syndrome	PAX6	DOMINANT	NM_000280.4	99.26%
Frank-ter Haar Syndrome	SH3PXD2B	RECESSIVE	NM_001017995.2	96.75%
Fraser Syndrome	FRAS1	RECESSIVE	NM_025074.6	99.89%
Fraser Syndrome	FREM2	RECESSIVE	NM_207361.4	99.77%
Fraser Syndrome	GRIP1	RECESSIVE	NM_021150.3	99.96%
Free Sialic Acid Storage Disorders	SLC17A5	RECESSIVE	NM_012434.4	98.00%
Freeman-Sheldon Syndrome	MYH3	RECESSIVE	NM_002470.3	99.45%
Frontotemporal Dementia	CHMP2B	DOMINANT	NM_014043.3	99.77%
Frontotemporal Dementia	GRN	DOMINANT	NM_002087.2	97.73%
Frontotemporal Dementia	TARDBP	DOMINANT	NM_007375.3	98.20%
Fructose 1,6 Bisphosphatase Deficiency	FBP1	DOMINANT	NM_000507.3	96.36%
Fucosidosis	FUCA1	RECESSIVE	NM_000147.4	99.22%
Fukuyama Congenital Muscular Dystrophy	FKTN	DOMINANT	NM_001079802.1	99.86%
Fumarate Hydratase Deficiency	FH	RECESSIVE	NM_000143.3	99.47%
Fundus Albipunctatus	PRPH2	DOMINANT	NM_000322.4	99.36%
Fundus Albipunctatus	RDH5	DOMINANT	NM_002905.3	99.28%
Fundus Albipunctatus	RLBP1	DOMINANT	NM_000326.4	97.56%
Furlong Syndrome	TGFBR1	DOMINANT	NM_004612.2	97.77%
GABA-Transaminase Deficiency	ABAT	RECESSIVE	NM_020686.5	99.67%
Galactokinase Deficiency	GALK1	RECESSIVE	NM_000154.1	97.56%
Galactosemia	GALT	RECESSIVE	NM_000155.2	99.38%
Galactosialidosis	CTSA	RECESSIVE	NM_000308.2	99.07%
Gamma-Sarcoglycanopathy	SGCG	RECESSIVE	NM_000231.2	99.52%

Gastrointestinal Stromal Tumor	KIT	DOMINANT	NM_000222.2	99.98%
Gastrointestinal Stromal Tumor	PDGFRA	DOMINANT	NM_006206.4	99.88%
Geleophysic Dysplasia	ADAMTSL2	DOMINANT	NM_014694.3	98.32%
Geleophysic Dysplasia	FBN1	DOMINANT	NM_000138.4	99.59%
Generalized Arterial Calcification of Infancy	ENPP1	RECESSIVE	NM_006208.2	96.75%
Generalized Epilepsy and Paroxysmal Dyskinesia	KCNMA1	DOMINANT	NM_002247.3	97.50%
Generalized Epilepsy with Febrile Seizures Plus	GABRG2	DOMINANT	NM_000816.3	99.93%
Generalized Epilepsy with Febrile Seizures Plus	SCN1B	DOMINANT	NM_001037.4	90.69%
Generalized Epilepsy with Febrile Seizures Plus	SCN9A	DOMINANT	NM_002977.3	99.43%
Generalized Pustular Psoriasis	IL36RN	RECESSIVE	NM_012275.2	99.76%
Genetic Prion Diseases	PRNP	DOMINANT	NM_000311.3	95.77%
Geroderma Osteodysplasticum	GORAB	RECESSIVE	NM_152281.2	99.74%
Giant Axonal Neuropathy	GAN	RECESSIVE	NM_022041.3	99.61%
Gilbert Syndrome	UGT1A1	RECESSIVE	NM_000463.2	99.91%
Gingival Fibromatosis	SOS1	DOMINANT	NM_005633.3	99.48%
Gitelman Syndrome	SLC12A3	RECESSIVE	NM_000339.2	99.14%
Glaucoma	MYOC	DOMINANT	NM_000261.1	99.85%
Global Cerebral Hypomyelination	SLC25A12	RECESSIVE	NM_003705.4	99.89%
Glomuvenous Malformation	GLMN	DOMINANT	NM_053274.2	99.21%
Glucocorticoid Deficiency	MC2R	RECESSIVE	NM_000529.2	97.40%
Glucocorticoid Deficiency	MRAP	RECESSIVE	NM_178817.3	99.61%
Glucocorticoid Resistance	NR3C1	DOMINANT	NM_001018077.1	99.34%
Glucocorticoid-Remediable Aldosteronism	CYP11B1	DOMINANT	NM_000497.3	96.74%
Glucocorticoid-Remediable Aldosteronism	CYP11B2	DOMINANT	NM_000498.3	95.83%
Glucose Transporter Type 1 Deficiency Syndrome	SLC2A1	DOMINANT	NM_006516.2	98.39%
Glucose-6-Phosphate Dehydrogenase Deficiency	G6PD	X_LINKED	NM_001042351.1	78.21%
Glucose-Galactose Malabsorption	SLC5A1	RECESSIVE	NM_000343.3	99.73%
Glutaricacidemia	GCDH	RECESSIVE	NM_000159.2	99.35%
Glutathione Synthetase Deficiency	GSS	RECESSIVE	NM_000178.2	99.94%

Glycine Encephalopathy	AMT	RECESSIVE	NM_000481.3	99.49%
Glycine Encephalopathy	GLDC	RECESSIVE	NM_000170.2	97.74%
Glycogen Storage Disease of Heart, Lethal Congenital	PRKAG2	DOMINANT	NM_016203.3	97.03%
Glycogen Storage Disease Type 0, Liver	GYS2	RECESSIVE	NM_021957.3	99.92%
Glycogen Storage Disease Type 0, Muscle	GYS1	RECESSIVE	NM_002103.4	98.85%
Glycogen Storage Disease Type I	G6PC	RECESSIVE	NM_000151.3	98.32%
Glycogen Storage Disease Type I	SLC37A4	RECESSIVE	NM_001164277.1	97.76%
Glycogen Storage Disease Type III	AGL	RECESSIVE	NM_000642.2	99.67%
Glycogen Storage Disease Type IV	GBE1	RECESSIVE	NM_000158.3	98.20%
Glycogen Storage Disease Type V	PYGM	RECESSIVE	NM_005609.2	99.07%
Glycogen Storage Disease Type VI	PYGL	RECESSIVE	NM_002863.4	99.30%
Glycogen Storage Disease Type VII	PFKM	RECESSIVE	NM_000289.5	99.93%
Glycogen Storage Disease Type X	PGAM2	RECESSIVE	NM_000290.3	97.42%
Glycogen Storage Disease Type XIII	ENO3	RECESSIVE	NM_053013.3	97.42%
Glycogen Storage Disease Type XIV	PGM1	RECESSIVE	NM_002633.2	98.32%
Glycogen Storage Disease XI	LDHA	RECESSIVE	NM_005566.3	98.98%
Glycogen Storage Disease, Type II	GAA	RECESSIVE	NM_000152.3	97.39%
Glycoprotein 1a Deficiency	ITGA2	DOMINANT	NM_002203.3	99.39%
GM1 Gangliosidosis	GLB1	RECESSIVE	NM_000404.2	99.90%
GM2 Activator Deficiency	GM2A	RECESSIVE	NM_000405.4	99.28%
Goldberg-Shprintzen Megacolon Syndrome	KIAA1279	RECESSIVE	NM_015634.3	99.83%
Gracile Syndrome	BCS1L	RECESSIVE	NM_004328.4	97.20%
Gray Platelet Syndrome	NBEAL2	RECESSIVE	NM_015175.2	96.43%
Greenberg Dysplasia	LBR	RECESSIVE	NM_002296.3	98.52%
Greig Cephalopolysyndactyly Syndrome	GLI3	DOMINANT	NM_000168.5	98.64%
Griscelli Syndrome	RAB27A	DOMINANT	NM_004580.4	97.45%
Growth Retardation, Developmental Delay, Coarse Facies, and Early Death	FTO	RECESSIVE	NM_001080432.2	99.53%
GTP Cyclohydrolase 1 Deficiency (GTPCH)	GCH1	RECESSIVE	NM_000161.2	98.40%

Guanidinoacetate Methyltransferase Deficiency	GAMT	RECESSIVE	NM_000156.4	97.97%
Haim-Munk Syndrome	CTSC	RECESSIVE	NM_001814.4	99.55%
Hawkinsinuria	HPD	DOMINANT	NM_002150.2	98.58%
Hemoglobin E	HBB	RECESSIVE	NM_000518.4	99.96%
Hemolytic Anemia	SLC4A1	RECESSIVE	NM_000342.3	97.69%
Hennekam Lymphangiectasia-Lymphedema Syndrome	CCBE1	RECESSIVE	NM_133459.3	97.38%
Hepatic Failure, Early-Onset, and Neurologic Disorder due to Cytochrome C Oxidase Deficiency	SCO1	RECESSIVE	NM_004589.2	97.37%
Hepatic Lipase Deficiency	LIPC	RECESSIVE	NM_000236.2	99.06%
Hepatic Veno-occlusive Disease with Immunodeficiency	SP110	RECESSIVE	NM_004509.3	99.14%
Hepatocerebral Mitochondrial DNA Depletion Syndrome	MPV17	RECESSIVE	NM_002437.4	97.52%
Hereditary Angioedema	F12	DOMINANT	NM_000505.3	98.74%
Hereditary Angioedema	SERPING1	DOMINANT	NM_000062.2	99.34%
Hereditary Angiopathy with Nephropathy, Aneurysms, and Muscle Cramps	COL4A1	DOMINANT	NM_001845.4	99.41%
Hereditary Breast and Ovarian Cancer	BRCA1	DOMINANT	NM_007294.3	99.41%
Hereditary Breast and Ovarian Cancer	BRCA2	DOMINANT	NM_000059.3	99.97%
Hereditary Coproporphyria	CPOX	DOMINANT	NM_000097.5	96.61%
Hereditary Diffuse Gastric Cancer	CDH1	DOMINANT	NM_004360.3	98.58%
Hereditary Diffuse Leukoencephalopathy with Spheroids	CSF1R	DOMINANT	NM_005211.3	98.27%
Hereditary Essential Tremor	DRD3	DOMINANT	NM_000796.3	99.52%
Hereditary Folate Malabsorption	SLC46A1	RECESSIVE	NM_080669.4	99.61%
Hereditary Fructose Intolerance	ALDOB	RECESSIVE	NM_000035.3	99.88%
Hereditary Hemochromatosis	HFE	RECESSIVE	NM_000410.3	99.97%
Hereditary Hemochromatosis	SLC40A1	RECESSIVE	NM_014585.5	99.81%
Hereditary Hemochromatosis	TFR2	RECESSIVE	NM_003227.3	96.67%
Hereditary Hemorrhagic Telangiectasia	ACVRL1	DOMINANT	NM_000020.2	97.64%
Hereditary Hemorrhagic Telangiectasia	ENG	DOMINANT	NM_000118.2	97.28%
Hereditary Hemorrhagic Telangiectasia	SMAD4	DOMINANT	NM_005359.5	98.23%

Hereditary Keratitis	PAX6	DOMINANT	NM_000280.4	99.26%
Hereditary Leiomyomatosis and Renal Cell Cancer	FH	DOMINANT	NM_000143.3	99.47%
Hereditary Motor and Sensory Neuropathy	MFN2	RECESSIVE	NM_014874.3	98.90%
Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum	SLC12A6	RECESSIVE	NM_133647.1	99.28%
Hereditary Multiple Osteochondromatosis	EXT1	DOMINANT	NM_000127.2	97.33%
Hereditary Multiple Osteochondromatosis	EXT2	DOMINANT	NM_207122.1	99.61%
Hereditary Myopathy with Early Respiratory Failure	TTN	DOMINANT	NM_133378.4	99.55%
Hereditary Neuropathy with Liability to Pressure Palsies	PMP22	DOMINANT	NM_000304.2	99.96%
Hereditary Pancreatitis	CFTR	DOMINANT	NM_000492.3	99.85%
Hereditary Pancreatitis	CTRC	DOMINANT	NM_007272.2	99.04%
Hereditary Pancreatitis	SPINK1	DOMINANT	NM_003122.3	99.99%
Hereditary Paraganglioma-Pheochromocytoma Syndrome	MAX	DOMINANT	NM_002382.3	96.97%
Hereditary Paraganglioma-Pheochromocytoma Syndrome	SDHAF2	DOMINANT	NM_017841.2	99.92%
Hereditary Paraganglioma-Pheochromocytoma Syndrome	SDHB	DOMINANT	NM_003000.2	99.74%
Hereditary Paraganglioma-Pheochromocytoma Syndrome	SDHC	DOMINANT	NM_003001.3	91.32%
Hereditary Paraganglioma-Pheochromocytoma Syndrome	SDHD	DOMINANT	NM_003002.2	89.67%
Hereditary Persistence of Fetal Hemoglobin	HBB	RECESSIVE	NM_000518.4	99.96%
Hereditary Persistence of Fetal Hemoglobin	HBD	RECESSIVE	NM_000519.3	99.99%
Hereditary Pyropoikilocytosis	SPTA1	RECESSIVE	NM_003126.2	99.65%
Hereditary Sensory and Autonomic Neuropathy, Type IC	SPTLC2	DOMINANT	NM_004863.3	99.08%
Hereditary Sensory and Autonomic Neuropathy, Type II	FAM134B	RECESSIVE	NM_001034850.2	98.17%
Hereditary Sensory and Autonomic Neuropathy, Type II	KIF1A	RECESSIVE	NM_004321.6	98.68%
Hereditary Sensory and Autonomic Neuropathy, Type II	SPTLC1	RECESSIVE	NM_006415.2	97.50%
Hereditary Sensory and Autonomic Neuropathy, Type II	WNK1	RECESSIVE	NM_018979.3	98.81%
Hereditary Sensory and Autonomic Neuropathy, Type IV	NTRK1	RECESSIVE	NM_001012331.1	96.95%
Hereditary Sensory and Autonomic Neuropathy, Type V	NGF	RECESSIVE	NM_002506.2	97.65%
Hereditary Sensory and Autonomic Neuropathy, Type VI	DST	RECESSIVE	NM_001723.5	99.98%
Hereditary Sideroblastic Anemia	SLC25A38	RECESSIVE	NM_017875.2	99.87%

Hereditary Sideroblastic Anemia with Myopathy and Lactic Acidosis	YARS2	RECESSIVE	NM_001040436.2	98.74%
Hereditary Sideroblastic Anemia with Spinocerebellar Ataxia	ABCB7	X_LINKED	NM_004299.3	96.52%
Heritable Pulmonary Arterial Hypertension	BMPR2	DOMINANT	NM_001204.6	99.21%
Heritable Pulmonary Arterial Hypertension	SMAD9	DOMINANT	NM_001127217.2	96.46%
Hermanskly-Pudlak Syndrome	AP3B1	RECESSIVE	NM_003664.3	99.53%
Hermanskly-Pudlak Syndrome	DTNBP1	RECESSIVE	NM_032122.4	97.80%
Hermanskly-Pudlak Syndrome	HPS1	RECESSIVE	NM_000195.3	98.66%
Hermanskly-Pudlak Syndrome	HPS3	RECESSIVE	NM_032383.3	97.75%
Hermanskly-Pudlak Syndrome	HPS4	RECESSIVE	NM_022081.4	98.69%
Hermanskly-Pudlak Syndrome	HPS5	RECESSIVE	NM_181507.1	99.96%
Hermanskly-Pudlak Syndrome	HPS6	RECESSIVE	NM_024747.5	98.02%
Heterotaxy Syndrome	ACVR2B	DOMINANT	NM_001106.3	98.59%
Heterotaxy Syndrome	NODAL	DOMINANT	NM_018055.4	99.55%
Hexosaminidase A Deficiency	HEXA	RECESSIVE	NM_000520.4	99.77%
Hidrotic Ectodermal Dysplasia	GJB6	DOMINANT	NM_006783.4	99.83%
Hirschsprung Disease, Dominant	EDN3	DOMINANT	NM_207034.1	98.17%
Hirschsprung Disease, Dominant	RET	DOMINANT	NM_020975.4	98.25%
Hirschsprung Disease, Recessive	EDNRB	RECESSIVE	NM_000115.3	99.44%
Histidinemia	HAL	RECESSIVE	NM_002108.3	99.73%
Holocarboxylase Synthetase Deficiency	HLCS	RECESSIVE	NM_000411.6	99.81%
Holoprosencephaly	CDON	DOMINANT	NM_016952.4	99.20%
Holoprosencephaly	FOXH1	DOMINANT	NM_003923.2	98.79%
Holoprosencephaly	NODAL	DOMINANT	NM_018055.4	99.55%
Holoprosencephaly	PTCH1	DOMINANT	NM_000264.3	97.68%
Holoprosencephaly	TGIF1	DOMINANT	NM_173208.1	96.95%
Holt-Oram Syndrome	TBX5	DOMINANT	NM_000192.3	98.45%
Homocystinuria	CBS	RECESSIVE	NM_000071.2	95.41%

Humerospinal Dysostosis	CHST3	RECESSIVE	NM_004273.4	98.22%
Hutchinson-Gilford Progeria Syndrome	LMNA	DOMINANT	NM_005572.3	96.33%
Hydrolethalus Syndrome	HYLS1	RECESSIVE	NM_145014.2	99.17%
Hydroxymethylbilane Synthase Deficiency	HMBS	DOMINANT	NM_000190.3	99.95%
Hyper IgD Syndrome	MVK	RECESSIVE	NM_000431.2	99.43%
Hyper IgE Syndrome	DOCK8	RECESSIVE	NM_203447.3	99.23%
Hyper IgE Syndrome	STAT3	DOMINANT	NM_139276.2	97.94%
Hyperalphalipoproteinemia	CETP	DOMINANT	NM_000078.2	99.77%
Hypercholanemia	BAAT	RECESSIVE	NM_001701.3	99.42%
Hyperekplexia	GLRA1	DOMINANT	NM_000171.3	99.53%
Hyperekplexia	GLRB	DOMINANT	NM_000824.4	98.71%
Hyperekplexia	SLC6A5	DOMINANT	NM_004211.3	99.69%
Hypereosinophilic Syndrome	PDGFRA	DOMINANT	NM_006206.4	99.88%
Hyperferritinemia Cataract Syndrome	FTL	DOMINANT	NM_000146.3	98.97%
Hyperglycinuria	SLC6A20	DOMINANT	NM_020208.3	98.69%
Hypergonadotropic Hypogonadism	LHCGR	RECESSIVE	NM_000233.3	98.17%
Hyperinsulinism, Dominant	GCK	DOMINANT	NM_000162.3	96.32%
Hyperinsulinism, Dominant/Recessive	ABCC8	DOMINANT	NM_000352.3	98.00%
Hyperinsulinism, Dominant/Recessive	GLUD1	DOMINANT	NM_005271.3	95.10%
Hyperinsulinism, Dominant/Recessive	HADH	DOMINANT	NM_005327.4	99.54%
Hyperinsulinism, Dominant/Recessive	HNF4A	DOMINANT	NM_000457.4	98.07%
Hyperinsulinism, Dominant/Recessive	KCNJ11	DOMINANT	NM_000525.3	91.82%
Hyperinsulinism, Dominant/Recessive	SLC16A1	DOMINANT	NM_003051.3	99.42%
Hyperkalemic Periodic Paralysis	SCN4A	DOMINANT	NM_000334.4	97.03%
Hypermethioninemia	ADK	RECESSIVE	NM_001123.3	98.55%
Hypermethioninemia	AHCY	RECESSIVE	NM_000687.2	99.72%
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	SLC25A15	RECESSIVE	NM_014252.3	96.55%
Hyperparathyroidism	MEN1	DOMINANT	NM_130799.2	96.55%

Hyperparathyroidism-Jaw Tumor Syndrome	CDC73	DOMINANT	NM_024529.4	99.19%
Hyperphosphatasia with Mental Retardation Syndrome	PIGV	RECESSIVE	NM_017837.3	99.12%
Hyperphosphatemic Familial Tumoral Calcinosis	FGF23	RECESSIVE	NM_020638.2	97.85%
Hyperphosphatemic Familial Tumoral Calcinosis	GALNT3	RECESSIVE	NM_004482.3	99.18%
Hyperphosphatemic Familial Tumoral Calcinosis	KL	RECESSIVE	NM_004795.3	98.02%
Hyperpigmentation, Cutaneous, with Hypertrichosis, Hepatosplenomegaly, Heart Anomalies, Hearing Loss, and Hypogonadism	SLC29A3	RECESSIVE	NM_018344.5	99.41%
Hyperprolinemia	ALDH4A1	RECESSIVE	NM_003748.3	98.47%
Hypertrichotic Osteochondrodysplasia	ABCC9	DOMINANT	NM_005691.2	99.66%
Hypertrophic Cardiomyopathy	GLA	X_LINKED	NM_000169.2	96.24%
Hypertrophic Cardiomyopathy	ACTC1	DOMINANT	NM_005159.4	98.49%
Hypertrophic Cardiomyopathy	ACTN2	DOMINANT	NM_001103.2	97.96%
Hypertrophic Cardiomyopathy	CAV3	DOMINANT	NM_033337.2	99.33%
Hypertrophic Cardiomyopathy	CSRP3	DOMINANT	NM_003476.4	97.72%
Hypertrophic Cardiomyopathy	LAMP2	DOMINANT	NM_002294.2	94.75%
Hypertrophic Cardiomyopathy	MYBPC3	DOMINANT	NM_000256.3	98.60%
Hypertrophic Cardiomyopathy	MYH6	DOMINANT	NM_002471.3	98.59%
Hypertrophic Cardiomyopathy	MYH7	DOMINANT	NM_000257.2	99.12%
Hypertrophic Cardiomyopathy	MYL2	DOMINANT	NM_000432.3	99.71%
Hypertrophic Cardiomyopathy	MYL3	DOMINANT	NM_000258.2	99.78%
Hypertrophic Cardiomyopathy	MYLK2	DOMINANT	NM_033118.3	99.29%
Hypertrophic Cardiomyopathy	MYOZ2	DOMINANT	NM_016599.3	99.23%
Hypertrophic Cardiomyopathy	NEXN	DOMINANT	NM_144573.3	99.09%
Hypertrophic Cardiomyopathy	PLN	DOMINANT	NM_002667.3	99.99%
Hypertrophic Cardiomyopathy	TCAP	DOMINANT	NM_003673.3	98.58%
Hypertrophic Cardiomyopathy	TNNC1	DOMINANT	NM_003280.2	98.67%
Hypertrophic Cardiomyopathy	TNNI3	DOMINANT	NM_000363.4	98.74%
Hypertrophic Cardiomyopathy	TNNT2	DOMINANT	NM_001001430.1	99.40%

Hypertrophic Cardiomyopathy	TPM1	DOMINANT	NM_001018005.1	97.66%
Hypertrophic Cardiomyopathy	TTN	DOMINANT	NM_133378.4	99.55%
Hyperuricemia, Pulmonary Hypertension, Renal Failure, and Alkalosis	SARS2	RECESSIVE	NM_017827.3	99.04%
Hypocalcemia	CASR	DOMINANT	NM_000388.3	99.26%
Hypochromic Microcytic Anemia with Iron Overload	SLC11A2	RECESSIVE	NM_000617.2	99.89%
Hypohidrotic Ectodermal Dysplasia, Dominant	EDAR	DOMINANT	NM_022336.3	99.85%
Hypohidrotic Ectodermal Dysplasia, Recessive	EDARADD	RECESSIVE	NM_145861.2	99.24%
Hypokalemic Periodic Paralysis	CACNA1S	DOMINANT	NM_000069.2	99.06%
Hypokalemic Periodic Paralysis	KCNE3	DOMINANT	NM_005472.4	94.16%
Hypokalemic Periodic Paralysis	SCN4A	DOMINANT	NM_000334.4	97.03%
Hypomagnesemia with Secondary Hypocalcemia	TRPM6	RECESSIVE	NM_017662.4	97.71%
Hypomyelination and Congenital Cataract	FAM126A	RECESSIVE	NM_032581.3	99.70%
Hypoparathyroidism, Sensorineural Deafness, and Renal Disease	GATA3	DOMINANT	NM_001002295.1	96.46%
Hypoparathyroidism-Retardation-Dysmorphism Syndrome	TBCE	RECESSIVE	NM_003193.3	99.79%
Hypophosphatasia	ALPL	DOMINANT	NM_000478.4	97.41%
Hypophosphatemic Nephrolithiasis/Osteoporosis	SLC34A1	RECESSIVE	NM_003052.4	97.04%
Hypophosphatemic Rickets, Dominant	FGF23	DOMINANT	NM_020638.2	97.85%
Hypophosphatemic Rickets, Recessive	DMP1	RECESSIVE	NM_004407.3	99.72%
Hypophosphatemic Rickets, Recessive	ENPP1	RECESSIVE	NM_006208.2	96.75%
Hypoplastic Left Heart Syndrome	GJA1	DOMINANT	NM_000165.3	95.58%
Ichthyosis Bullosa of Siemens	KRT2	DOMINANT	NM_000423.2	98.63%
Ichthyosis, Hystrix-like, with Deafness	GJB2	DOMINANT	NM_004004.5	97.20%
Immunodeficiency due to Defect in CD3-Gamma	CD3G	RECESSIVE	NM_000073.2	96.51%
Immunodeficiency with Hyper-IgM	AICDA	RECESSIVE	NM_020661.2	96.90%
Immunodeficiency with Hyper-IgM	CD40	RECESSIVE	NM_001250.4	98.46%
Immunodeficiency with Hyper-IgM	UNG	RECESSIVE	NM_080911.2	99.37%
Immunodeficiency-Centromeric Instability-Facial Anomalies	DNMT3B	RECESSIVE	NM_006892.3	95.41%

Syndrome				
Inclusion Body Myopathy, Dominant	MYH2	DOMINANT	NM_017534.5	99.85%
Inclusion Body Myopathy, Dominant	VCP	DOMINANT	NM_007126.3	99.01%
Inclusion Body Myopathy, Recessive	GNE	RECESSIVE	NM_005476.5	95.71%
Infantile Hypercalcemia	CYP24A1	DOMINANT	NM_000782.4	99.41%
Infantile Nystagmus	FRMD7	X_LINKED	NM_194277.2	97.16%
Infantile Spinal Muscular Atrophy, X-Linked	UBA1	X_LINKED	NM_003334.3	89.32%
Inflammatory Bowel Disease	IL10RA	RECESSIVE	NM_001558.3	99.82%
Inflammatory Bowel Disease	IL10RB	RECESSIVE	NM_000628.4	97.73%
Inherited Erythromelalgia	SCN9A	DOMINANT	NM_002977.3	99.43%
Inherited Systemic Hyalinosis	ANTXR2	RECESSIVE	NM_058172.5	99.56%
Insulin-Like Growth Factor I Deficiency	IGF1	RECESSIVE	NM_000618.3	99.11%
Insulin-Like Growth Factor I Deficiency	IGF1R	RECESSIVE	NM_000875.3	98.74%
Intellectual Disability Syndrome	KANSL1	DOMINANT	NM_001193466.1	99.22%
Interleukin 1 Receptor Antagonist Deficiency	IL1RN	RECESSIVE	NM_173841.2	99.91%
Interleukin 2 Receptor Alpha Chain Deficiency	IL2RA	RECESSIVE	NM_000417.2	99.74%
Intrinsic Factor Deficiency	GIF	RECESSIVE	NM_005142.2	99.94%
IRAK4 Deficiency	IRAK4	RECESSIVE	NM_016123.3	98.62%
Iris Hypoplasia	PITX2	DOMINANT	NM_153427.2	99.12%
Iron Overload	FTH1	DOMINANT	NM_002032.2	96.46%
Iron-Refractory Iron Deficiency Anemia	TMPRSS6	RECESSIVE	NM_153609.2	98.15%
Isobutyryl-CoA Dehydrogenase Deficiency	ACAD8	RECESSIVE	NM_014384.2	99.45%
Isolated Congenital Digital Clubbing	HPGD	RECESSIVE	NM_000860.5	99.68%
Isolated Coronal Synostosis	FGFR2	DOMINANT	NM_000141.4	97.52%
Isolated Follicle-Stimulating Hormone Deficiency	FSHB	RECESSIVE	NM_000510.2	99.97%
Isolated GnRH Deficiency	GNRH1	RECESSIVE	NM_000825.3	99.99%
Isolated GnRH Deficiency	GNRHR	RECESSIVE	NM_000406.2	99.65%
Isolated GnRH Deficiency	TACR3	RECESSIVE	NM_001059.2	99.07%
Isolated Growth Hormone Deficiency	GH1	RECESSIVE	NM_000515.3	97.24%

Isolated Growth Hormone Deficiency	GHRHR	RECESSIVE	NM_000823.3	99.72%
Isolated Hyperparathyroidism	CDC73	DOMINANT	NM_024529.4	99.19%
Isolated Microphthalmia	MFRP	RECESSIVE	NM_031433.2	98.05%
Isolated Microphthalmia	VSX2	RECESSIVE	NM_182894.2	99.45%
Isolated Nonsyndromic Congenital Heart Disease	JAG1	DOMINANT	NM_000214.2	99.22%
Isolated Persistent Hypermethioninemia	MAT1A	DOMINANT	NM_000429.2	99.55%
Isovaleric Acidemia	IVD	RECESSIVE	NM_002225.3	96.05%
Jackson-Weiss Syndrome	FGFR2	DOMINANT	NM_000141.4	97.52%
Jalili Syndrome	CNNM4	RECESSIVE	NM_020184.3	95.75%
Jawad Syndrome	RBBP8	RECESSIVE	NM_002894.2	99.22%
Jervell and Lange-Nielsen Syndrome	KCNE1	RECESSIVE	NM_000219.3	99.96%
Jervell and Lange-Nielsen Syndrome	KCNQ1	RECESSIVE	NM_000218.2	92.10%
Joubert Syndrome	AHI1	RECESSIVE	NM_017651.4	99.72%
Joubert Syndrome	ARL13B	RECESSIVE	NM_182896.2	99.13%
Joubert Syndrome	C5orf42	RECESSIVE	NM_023073.3	99.61%
Joubert Syndrome	CC2D2A	RECESSIVE	NM_001080522.2	99.94%
Joubert Syndrome	CEP290	RECESSIVE	NM_025114.3	99.36%
Joubert Syndrome	CEP41	RECESSIVE	NM_018718.2	98.88%
Joubert Syndrome	NPHP1	RECESSIVE	NM_000272.3	99.47%
Joubert Syndrome	RPGRIP1L	RECESSIVE	NM_015272.2	99.57%
Joubert Syndrome	TCTN1	RECESSIVE	NM_001082538.2	98.23%
Joubert Syndrome	TCTN2	RECESSIVE	NM_024809.4	97.94%
Joubert Syndrome	TMEM138	RECESSIVE	NM_016464.4	99.85%
Joubert Syndrome	TMEM216	RECESSIVE	NM_001173990.2	98.50%
Joubert Syndrome	TMEM237	RECESSIVE	NM_001044385.2	99.69%
Joubert Syndrome	TMEM67	RECESSIVE	NM_153704.5	99.40%
Joubert Syndrome	TTC21B	RECESSIVE	NM_024753.4	98.51%
Junctional Epidermolysis Bullosa	COL17A1	RECESSIVE	NM_000494.3	99.76%
Junctional Epidermolysis Bullosa	LAMA3	RECESSIVE	NM_000227.3	99.95%

Junctional Epidermolysis Bullosa	LAMB3	RECESSIVE	NM_000228.2	98.81%
Junctional Epidermolysis Bullosa	LAMC2	RECESSIVE	NM_005562.2	99.49%
Juvenile Hereditary Hemochromatosis	HAMP	RECESSIVE	NM_021175.2	99.69%
Juvenile Hereditary Hemochromatosis	HFE2	RECESSIVE	NM_213653.3	99.62%
Juvenile Myoclonic Epilepsy	CACNB4	DOMINANT	NM_000726.3	99.76%
Juvenile Myoclonic Epilepsy	EFHC1	DOMINANT	NM_018100.3	98.91%
Juvenile Myoclonic Epilepsy	GABRA1	DOMINANT	NM_000806.5	99.89%
Juvenile Paget Disease	TNFRSF11B	RECESSIVE	NM_002546.3	99.50%
Juvenile Polyposis	ENG	DOMINANT	NM_000118.2	97.28%
Juvenile Polyposis	SMAD4	DOMINANT	NM_005359.5	98.23%
Kabuki Syndrome	MLL2	DOMINANT	NM_003482.3	97.81%
Kallmann Syndrome	CHD7	DOMINANT	NM_017780.3	98.90%
Kallmann Syndrome	FGFR1	DOMINANT	NM_023110.2	98.79%
Kallmann Syndrome	PROKR2	DOMINANT	NM_144773.2	95.89%
Kanzaki disease	NAGA	RECESSIVE	NM_000262.2	99.76%
KAT6B-Related Spectrum Disorders	KAT6B	DOMINANT	NM_012330.3	99.14%
Keratitis-Ichthyosis-Deafness Syndrome	GJB2	DOMINANT	NM_004004.5	97.20%
Keratoconus	VSX1	DOMINANT	NM_014588.5	97.29%
Ketothiolase Deficiency	ACAT1	RECESSIVE	NM_000019.3	98.88%
Keutel Syndrome	MGP	RECESSIVE	NM_000900.3	99.83%
Kindler Syndrome	FERMT1	RECESSIVE	NM_017671.4	98.54%
Kleefstra Syndrome	EHMT1	DOMINANT	NM_024757.4	96.71%
Klippel-Feil Syndrome	GDF6	DOMINANT	NM_001001557.2	97.64%
Knobloch Syndrome	COL18A1	RECESSIVE	NM_130445.2	95.38%
Krabbe Disease	GALC	RECESSIVE	NM_000153.3	98.90%
Krabbe Disease	PSAP	DOMINANT	NM_002778.2	99.84%
Kufor-Rakeb Syndrome	ATP13A2	RECESSIVE	NM_022089.2	96.02%
Lacrimo-Auriculo-Dento-Digital Syndrome	FGF10	DOMINANT	NM_004465.1	99.92%
Lacrimo-Auriculo-Dento-Digital Syndrome	FGFR2	DOMINANT	NM_000141.4	97.52%

Lactate Dehydrogenase B Deficiency	LDHB	RECESSIVE	NM_002300.6	98.26%
Lactose Intolerance	LCT	RECESSIVE	NM_002299.2	99.86%
Laing Distal Myopathy	MYH7	DOMINANT	NM_000257.2	99.12%
Langer-Giedion Syndrome	EXT1	DOMINANT	NM_000127.2	97.33%
Langer-Giedion Syndrome	TRPS1	DOMINANT	NM_014112.2	97.47%
L-Arginine:Glycine Amidinotransferase Deficiency	GATM	RECESSIVE	NM_001482.2	99.69%
Larsen Syndrome	CHST3	RECESSIVE	NM_004273.4	98.22%
Laryngoonychocutaneous Syndrome	LAMA3	RECESSIVE	NM_000227.3	99.95%
Lathosterolosis	SC5DL	RECESSIVE	NM_006918.4	98.08%
LCHAD Deficiency	HADHA	RECESSIVE	NM_000182.4	99.95%
Leber Congenital Amaurosis	AIPL1	RECESSIVE	NM_014336.3	98.86%
Leber Congenital Amaurosis	CEP290	RECESSIVE	NM_025114.3	99.36%
Leber Congenital Amaurosis	CRB1	RECESSIVE	NM_201253.2	99.98%
Leber Congenital Amaurosis	CRX	RECESSIVE	NM_000554.4	96.57%
Leber Congenital Amaurosis	IMPDH1	RECESSIVE	NM_000883.3	96.28%
Leber Congenital Amaurosis	KCNJ13	RECESSIVE	NM_002242.4	99.96%
Leber Congenital Amaurosis	LCA5	RECESSIVE	NM_181714.3	99.68%
Leber Congenital Amaurosis	LRAT	RECESSIVE	NM_004744.3	98.57%
Leber Congenital Amaurosis	RD3	RECESSIVE	NM_183059.2	98.27%
Leber Congenital Amaurosis	RPE65	RECESSIVE	NM_000329.2	99.90%
Leber Congenital Amaurosis	RPGRIP1	RECESSIVE	NM_020366.3	99.78%
Leber Congenital Amaurosis	SPATA7	RECESSIVE	NM_018418.4	99.91%
Leber Congenital Amaurosis	TULP1	RECESSIVE	NM_003322.3	98.64%
Lecithin Cholesterol Acyltransferase Deficiency	LCAT	RECESSIVE	NM_000229.1	96.00%
Left Ventricular Noncompaction Cardiomyopathy	ACTC1	DOMINANT	NM_005159.4	98.49%
Left Ventricular Noncompaction Cardiomyopathy	DTNA	DOMINANT	NM_032978.6	99.49%
Left Ventricular Noncompaction Cardiomyopathy	LDB3	DOMINANT	NM_001080116.1	99.33%
Left Ventricular Noncompaction Cardiomyopathy	MYBPC3	DOMINANT	NM_000256.3	98.60%
Left Ventricular Noncompaction Cardiomyopathy	MYH7	DOMINANT	NM_000257.2	99.12%

Left Ventricular Noncompaction Cardiomyopathy	TAZ	DOMINANT	NM_000116.3	90.55%
Left Ventricular Noncompaction Cardiomyopathy	TNNT2	DOMINANT	NM_001001430.1	99.40%
Legius Syndrome	SPRED1	DOMINANT	NM_152594.2	99.24%
Leigh Syndrome	BCS1L	RECESSIVE	NM_004328.4	97.20%
Leigh Syndrome	COX10	RECESSIVE	NM_001303.3	95.53%
Leigh Syndrome	DLD	RECESSIVE	NM_000108.3	99.95%
Leigh Syndrome	LRPPRC	RECESSIVE	NM_133259.3	99.39%
Leigh Syndrome	NDUFA10	RECESSIVE	NM_004544.3	99.01%
Leigh Syndrome	NDUFAF2	RECESSIVE	NM_174889.4	99.27%
Leigh Syndrome	NDUFS1	RECESSIVE	NM_005006.6	99.43%
Leigh Syndrome	NDUFS3	RECESSIVE	NM_004551.2	99.19%
Leigh Syndrome	NDUFS4	RECESSIVE	NM_002495.2	99.85%
Leigh Syndrome	NDUFS7	RECESSIVE	NM_024407.4	95.70%
Leigh Syndrome	NDUFS8	RECESSIVE	NM_002496.3	97.48%
Leigh Syndrome	NDUFV1	RECESSIVE	NM_007103.3	96.78%
Leigh Syndrome	SCO1	RECESSIVE	NM_004589.2	97.37%
LEOPARD Syndrome	BRAF	DOMINANT	NM_004333.4	97.81%
LEOPARD Syndrome	PTPN11	DOMINANT	NM_002834.3	98.03%
LEOPARD Syndrome	RAF1	DOMINANT	NM_002880.3	99.39%
Leptin Deficiency	LEP	RECESSIVE	NM_000230.2	98.22%
Leptin Receptor Deficiency	LEPR	RECESSIVE	NM_002303.5	98.34%
Lethal Arthrogryposis With Anterior Horn Cell Disease	GLE1	DOMINANT	NM_001003722.1	99.49%
Lethal Congenital Contracture Syndrome	GLE1	RECESSIVE	NM_001003722.1	99.49%
Lethal Encephalopathy	DNM1L	DOMINANT	NM_012062.3	99.70%
Lethal Restrictive Dermopathy	LMNA	RECESSIVE	NM_005572.3	96.33%
Lethal Restrictive Dermopathy	ZMPSTE24	RECESSIVE	NM_005857.4	99.89%
Leukocyte Adhesion Deficiency	ITGB2	RECESSIVE	NM_000211.3	98.87%
Leukodystrophy, Adult-Onset	LMNB1	DOMINANT	NM_005573.3	95.66%
Leukoencephalopathy with Brainstem and Spinal Cord	DARS2	RECESSIVE	NM_018122.4	99.54%

Involvement and Lactate Elevation				
Leukoencephalopathy, Cystic, without Megalencephaly	RNASET2	RECESSIVE	NM_003730.4	99.14%
Leydig Cell Hypoplasia/Agenesis	LHCGR	RECESSIVE	NM_000233.3	98.17%
Liddle Syndrome	SCNN1B	DOMINANT	NM_000336.2	98.80%
Li-Fraumeni Syndrome	TP53	DOMINANT	NM_000546.4	97.63%
LIG4 Syndrome	LIG4	RECESSIVE	NM_002312.3	99.86%
Limb-Girdle Muscular Dystrophy, Dominant	CAV3	DOMINANT	NM_033337.2	99.33%
Limb-Girdle Muscular Dystrophy, Dominant	DNAJB6	DOMINANT	NM_058246.3	98.20%
Limb-Girdle Muscular Dystrophy, Dominant	MYOT	DOMINANT	NM_006790.2	99.03%
Limb-Girdle Muscular Dystrophy, Recessive	POMT1	DOMINANT	NM_007171.3	99.29%
Limb-Girdle Muscular Dystrophy, Recessive	ANO5	RECESSIVE	NM_213599.2	99.66%
Limb-Girdle Muscular Dystrophy, Recessive	CAPN3	RECESSIVE	NM_000070.2	99.52%
Limb-Girdle Muscular Dystrophy, Recessive	DYSF	RECESSIVE	NM_003494.3	98.83%
Limb-Girdle Muscular Dystrophy, Recessive	LMNA	RECESSIVE	NM_005572.3	96.33%
Limb-Girdle Muscular Dystrophy, Recessive	POMGNT1	RECESSIVE	NM_017739.3	97.69%
Limb-Girdle Muscular Dystrophy, Recessive	SGCA	RECESSIVE	NM_000023.2	99.02%
Limb-Girdle Muscular Dystrophy, Recessive	SGCB	RECESSIVE	NM_000232.4	98.84%
Limb-Girdle Muscular Dystrophy, Recessive	SGCD	RECESSIVE	NM_000337.5	99.33%
Limb-Girdle Muscular Dystrophy, Recessive	SGCG	RECESSIVE	NM_000231.2	99.52%
Limb-Girdle Muscular Dystrophy, Recessive	TCAP	RECESSIVE	NM_003673.3	98.58%
Limb-Girdle Muscular Dystrophy, Recessive	TRIM32	RECESSIVE	NM_012210.3	99.49%
Limb-Girdle Muscular Dystrophy, Recessive	TTN	RECESSIVE	NM_133378.4	99.55%
Limb-Girdle Myasthenia with Tubular Aggregates	GFPT1	RECESSIVE	NM_002056.3	98.71%
Lipoatrophy with Diabetes, Hepatic Steatosis, Hypertrophic Cardiomyopathy, and Leukomelanodermic Papules	LMNA	DOMINANT	NM_005572.3	96.33%
Lissencephaly, Dominant	TUBA1A	DOMINANT	NM_006009.3	96.65%
Lissencephaly, Recessive	NDE1	RECESSIVE	NM_001143979.1	98.77%
Lissencephaly, Recessive	RELN	RECESSIVE	NM_005045.3	99.30%
Lissencephaly/Subcortical Band Heterotopia	PAFAH1B1	DOMINANT	NM_000430.3	95.02%

Localized AR Hypotrichosis	DSG4	RECESSIVE	NM_177986.3	99.97%
Loeys-Dietz Syndrome	SMAD3	DOMINANT	NM_005902.3	97.72%
Loeys-Dietz Syndrome	TGFBR1	DOMINANT	NM_004612.2	97.77%
Loeys-Dietz Syndrome	TGFBR2	DOMINANT	NM_003242.5	99.28%
Long QT Syndrome	AKAP9	DOMINANT	NM_005751.4	99.71%
Long QT Syndrome	ANK2	DOMINANT	NM_001148.4	99.75%
Long QT Syndrome	CAV3	DOMINANT	NM_033337.2	99.33%
Long QT Syndrome	KCNE1	DOMINANT	NM_000219.3	99.96%
Long QT Syndrome	KCNE2	DOMINANT	NM_172201.1	100.00%
Long QT Syndrome	KCNH2	DOMINANT	NM_000238.3	88.25%
Long QT Syndrome	KCNJ5	DOMINANT	NM_000890.3	95.90%
Long QT Syndrome	KCNQ1	DOMINANT	NM_000218.2	92.10%
Long QT Syndrome	SCN4B	DOMINANT	NM_174934.3	98.40%
Long QT Syndrome	SCN5A	DOMINANT	NM_198056.2	98.54%
Long QT Syndrome	SNTA1	DOMINANT	NM_003098.2	89.35%
Long-Chain Acyl-CoA Dehydrogenase Deficiency	ACADL	RECESSIVE	NM_001608.3	99.87%
Lung Cancer	EGFR	DOMINANT	NM_005228.3	97.21%
Lymphoproliferative Syndrome	ITK	RECESSIVE	NM_005546.3	99.92%
Lynch Syndrome	EPCAM	DOMINANT	NM_002354.2	98.39%
Lynch Syndrome	MLH1	DOMINANT	NM_000249.3	98.91%
Lynch Syndrome	MLH3	DOMINANT	NM_001040108.1	99.59%
Lynch Syndrome	MSH2	DOMINANT	NM_000251.1	98.79%
Lynch Syndrome	MSH6	DOMINANT	NM_000179.2	99.46%
Lynch Syndrome	PMS1	DOMINANT	NM_000534.4	99.43%
Lynch Syndrome	PMS2	DOMINANT	NM_000535.5	75.95%
Lysinuric Protein Intolerance	SLC7A7	RECESSIVE	NM_001126106.2	99.91%
Macular Corneal Dystrophy	CHST6	RECESSIVE	NM_021615.4	96.00%
Macular Degeneration	ABCA4	DOMINANT	NM_000350.2	99.79%
Macular Degeneration	ARMS2	DOMINANT	NM_001099667.1	99.25%

Macular Degeneration	C2	DOMINANT	NM_000063.4	99.90%
Macular Degeneration	C3	DOMINANT	NM_000064.2	98.50%
Macular Degeneration	CFB	DOMINANT	NM_001710.5	98.11%
Macular Degeneration	CFH	DOMINANT	NM_000186.3	98.32%
Macular Degeneration	ERCC6	DOMINANT	NM_000124.2	99.64%
Macular Degeneration	FBLN5	DOMINANT	NM_006329.3	98.82%
Macular Degeneration	HMCN1	DOMINANT	NM_031935.2	99.84%
Macular Degeneration	RAX2	DOMINANT	NM_032753.3	98.22%
Mainzer-Saldino Syndrome	IFT140	RECESSIVE	NM_014714.3	97.76%
Majeed Syndrome	LPIN2	RECESSIVE	NM_014646.2	99.69%
Mal de Meleda	SLURP1	RECESSIVE	NM_020427.2	98.93%
Male Infertility	CATSPER1	RECESSIVE	NM_053054.3	98.60%
Male-Limited Precocious Puberty	LHCGR	DOMINANT	NM_000233.3	98.17%
Malignant Hyperthermia Susceptibility	CACNA1S	DOMINANT	NM_000069.2	99.06%
Malignant Hyperthermia Susceptibility	RYR1	DOMINANT	NM_000540.2	97.36%
Malonyl-CoA Decarboxylase Deficiency	MLYCD	RECESSIVE	NM_012213.2	95.49%
Mandibuloacral Dysplasia	LMNA	RECESSIVE	NM_005572.3	96.33%
Mandibuloacral Dysplasia	ZMPSTE24	RECESSIVE	NM_005857.4	99.89%
Manitoba Oculotrichoanal Syndrome	FREM1	RECESSIVE	NM_144966.5	99.81%
Mannose-Binding Protein Deficiency	MBL2	RECESSIVE	NM_000242.2	99.72%
Maple Syrup Urine Disease	BCKDHA	RECESSIVE	NM_000709.3	99.14%
Maple Syrup Urine Disease	BCKDHB	RECESSIVE	NM_183050.2	97.59%
Maple Syrup Urine Disease	DBT	RECESSIVE	NM_001918.2	98.69%
Maple Syrup Urine Disease	DLD	RECESSIVE	NM_000108.3	99.95%
MAPT-Related Spectrum Disorders	MAPT	DOMINANT	NM_005910.5	96.95%
Marfan Syndrome	FBN1	DOMINANT	NM_000138.4	99.59%
Marinesco-Sjogren Syndrome	SIL1	RECESSIVE	NM_022464.4	97.87%
Marshall Syndrome	COL11A1	DOMINANT	NM_001854.3	98.66%
Martsolf Syndrome	RAB3GAP2	RECESSIVE	NM_012414.3	99.31%

MASP2 Deficiency	MASP2	RECESSIVE	NM_006610.3	99.92%
MASS Syndrome	FBN1	DOMINANT	NM_000138.4	99.59%
Mast Cell Disease	KIT	DOMINANT	NM_000222.2	99.98%
Maturity Onset Diabetes of the Young	BLK	DOMINANT	NM_001715.2	98.65%
Maturity Onset Diabetes of the Young	GCK	DOMINANT	NM_000162.3	96.32%
Maturity Onset Diabetes of the Young	HNF1A	DOMINANT	NM_000545.5	99.22%
Maturity Onset Diabetes of the Young	HNF4A	DOMINANT	NM_000457.4	98.07%
Maturity Onset Diabetes of the Young	INS	DOMINANT	NM_000207.2	99.12%
Maturity Onset Diabetes of the Young	KCNJ11	DOMINANT	NM_000525.3	91.82%
Maturity Onset Diabetes of the Young	KLF11	DOMINANT	NM_003597.4	96.44%
Maturity Onset Diabetes of the Young	NEUROD1	DOMINANT	NM_002500.4	96.37%
Maturity Onset Diabetes of the Young	PAX4	DOMINANT	NM_006193.2	98.92%
May-Hegglin Anomaly	MYH9	DOMINANT	NM_002473.4	99.28%
MCAD Deficiency	ACADM	RECESSIVE	NM_000016.4	97.83%
McKusick-Kaufman Syndrome	MKKS	RECESSIVE	NM_018848.3	99.99%
Meacham Syndrome	WT1	DOMINANT	NM_024426.4	95.44%
Meckel Syndrome	B9D1	RECESSIVE	NM_015681.3	99.60%
Meckel Syndrome	B9D2	RECESSIVE	NM_030578.3	99.33%
Meckel Syndrome	CC2D2A	RECESSIVE	NM_001080522.2	99.94%
Meckel Syndrome	CEP290	RECESSIVE	NM_025114.3	99.36%
Meckel Syndrome	MKS1	RECESSIVE	NM_017777.3	99.30%
Meckel Syndrome	NPHP3	RECESSIVE	NM_153240.4	96.76%
Meckel Syndrome	RPGRIP1L	RECESSIVE	NM_015272.2	99.57%
Meckel Syndrome	TCTN2	RECESSIVE	NM_024809.4	97.94%
Meckel Syndrome	TMEM216	RECESSIVE	NM_001173990.2	98.50%
Meckel Syndrome	TMEM67	RECESSIVE	NM_153704.5	99.40%
Medulloblastoma	SUFU	DOMINANT	NM_016169.3	98.84%
Megalencephalic Leukoencephalopathy with Subcortical Cysts	HEPACAM	RECESSIVE	NM_152722.4	96.32%

Megalencephalic Leukoencephalopathy with Subcortical Cysts	MLC1	RECESSIVE	NM_015166.3	97.63%
Megaloblastic Anemia	CUBN	RECESSIVE	NM_001081.3	99.82%
Meier-Gorlin Syndrome	CDC6	RECESSIVE	NM_001254.3	99.92%
Meier-Gorlin Syndrome	ORC1	RECESSIVE	NM_004153.3	99.96%
Meier-Gorlin Syndrome	ORC6	RECESSIVE	NM_014321.3	99.85%
Mental Retardation with Language Impairment and Autistic Features	FOXP1	DOMINANT	NM_032682.5	96.66%
Mental Retardation, Dominant	DYNC1H1	DOMINANT	NM_001376.4	99.09%
Mental Retardation, Dominant	GRIN2B	DOMINANT	NM_000834.3	99.80%
Mental Retardation, Dominant	KIF1A	DOMINANT	NM_004321.6	98.68%
Mental Retardation, Dominant	MBD5	DOMINANT	NM_018328.4	99.86%
Mental Retardation, Recessive	MAN1B1	RECESSIVE	NM_016219.4	95.94%
Mental Retardation, Recessive	NSUN2	RECESSIVE	NM_017755.5	99.61%
Mental Retardation, Recessive	TRAPPC9	RECESSIVE	NM_031466.5	98.54%
Mental Retardation, Stereotypic Movements, Epilepsy, and/or Cerebral Malformations	MEF2C	DOMINANT	NM_002397.4	98.41%
Mental Retardation, X-linked	ARHGEF6	X_LINKED	NM_004840.2	97.27%
Mental Retardation, X-linked	IL1RAPL1	X_LINKED	NM_014271.3	96.18%
Mental Retardation, X-linked	RAB39B	X_LINKED	NM_171998.2	95.60%
Mental Retardation, X-linked	ZNF81	X_LINKED	NM_007137.3	97.19%
Metachondromatosis	PTPN11	DOMINANT	NM_002834.3	98.03%
Metachromatic Leukodystrophy	PSAP	RECESSIVE	NM_002778.2	99.84%
Metaphyseal Anadysplasia	MMP13	DOMINANT	NM_002427.3	99.91%
Metaphyseal Chondrodysplasia	COL10A1	DOMINANT	NM_000493.3	99.97%
Metaphyseal Chondrodysplasia	PTH1R	DOMINANT	NM_000316.2	98.57%
Metatropic Dysplasia	TRPV4	DOMINANT	NM_021625.4	97.17%
Methylmalonate Semialdehyde Dehydrogenase Deficiency	ALDH6A1	RECESSIVE	NM_005589.2	99.76%
Methylmalonic Acidemia	MCEE	RECESSIVE	NM_032601.3	96.22%

Methylmalonic Acidemia	MMAA	RECESSIVE	NM_172250.2	99.47%
Methylmalonic Acidemia	MMAB	RECESSIVE	NM_052845.3	96.68%
Methylmalonic Acidemia	MMADHC	RECESSIVE	NM_015702.2	99.95%
Methylmalonic Acidemia	MUT	RECESSIVE	NM_000255.3	99.54%
Mevalonicaciduria	MVK	RECESSIVE	NM_000431.2	99.43%
MHC Class II Deficiency	RFXANK	RECESSIVE	NM_003721.2	98.85%
Microcephalic Osteodysplastic Primordial Dwarfism	PCNT	RECESSIVE	NM_006031.5	98.70%
Microcephaly, Cortical Malformations, and Mental Retardation	WDR62	RECESSIVE	NM_001083961.1	98.54%
Microtia, Hearing Impairment, and Cleft Palate	HOXA2	RECESSIVE	NM_006735.3	97.57%
Mitochondrial Complex V (ATP Synthase) Deficiency, Nuclear Type	ATPAF2	RECESSIVE	NM_145691.3	97.54%
Mitochondrial Complex V (ATP Synthase) Deficiency, Nuclear Type	TMEM70	RECESSIVE	NM_017866.5	98.97%
Mitochondrial DNA Depletion Syndrome	C10orf2	RECESSIVE	NM_021830.4	98.46%
Mitochondrial DNA Depletion Syndrome	DGUOK	RECESSIVE	NM_080916.2	99.95%
Mitochondrial DNA Depletion Syndrome	MPV17	RECESSIVE	NM_002437.4	97.52%
Mitochondrial DNA Depletion Syndrome	RRM2B	RECESSIVE	NM_015713.4	97.04%
Mitochondrial DNA Depletion Syndrome	SUCLA2	RECESSIVE	NM_003850.2	99.43%
Mitochondrial DNA Depletion Syndrome	SUCLG1	RECESSIVE	NM_003849.3	98.92%
Mitochondrial DNA Depletion Syndrome	TK2	RECESSIVE	NM_004614.4	99.39%
Mitochondrial Membrane Protein-Associated Neurodegeneration	C19orf12	RECESSIVE	NM_001031726.3	97.99%
Mitochondrial Neurogastrointestinal Encephalopathy Disease	TYMP	RECESSIVE	NM_001953.4	96.63%
Mitochondrial Phosphate Carrier Deficiency	SLC25A3	RECESSIVE	NM_005888.3	99.76%
Mitochondrial Respiratory Chain Complex I Deficiency	FOXRED1	RECESSIVE	NM_017547.3	96.62%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFA10	RECESSIVE	NM_004544.3	99.01%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFA11	RECESSIVE	NM_175614.4	98.25%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFA2	RECESSIVE	NM_002488.4	99.53%

Mitochondrial Respiratory Chain Complex I Deficiency	NDUFAF1	RECESSIVE	NM_016013.3	98.43%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFAF2	RECESSIVE	NM_174889.4	99.27%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFAF3	RECESSIVE	NM_199069.1	99.32%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFAF4	RECESSIVE	NM_014165.3	99.72%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS1	RECESSIVE	NM_005006.6	99.43%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS2	RECESSIVE	NM_004550.4	99.69%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS3	RECESSIVE	NM_004551.2	99.19%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS4	RECESSIVE	NM_002495.2	99.85%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS6	RECESSIVE	NM_004553.4	98.73%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS7	RECESSIVE	NM_024407.4	95.70%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFS8	RECESSIVE	NM_002496.3	97.48%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFV1	RECESSIVE	NM_007103.3	96.78%
Mitochondrial Respiratory Chain Complex I Deficiency	NDUFV2	RECESSIVE	NM_021074.4	99.32%
Mitochondrial Respiratory Chain Complex I Deficiency	NUBPL	RECESSIVE	NM_025152.2	98.61%
Mitochondrial Respiratory Chain Complex II Deficiency	SDHAF1	RECESSIVE	NM_001042631.2	98.07%
Mitochondrial Respiratory Chain Complex III Deficiency	BCS1L	RECESSIVE	NM_004328.4	97.20%
Mitochondrial Respiratory Chain Complex III Deficiency	TTC19	RECESSIVE	NM_017775.3	97.21%
Mitochondrial Respiratory Chain Complex III Deficiency	UQCRQ	RECESSIVE	NM_014402.4	99.93%
Mitochondrial Respiratory Chain Complex IV Deficiency	COX10	RECESSIVE	NM_001303.3	95.53%
Mitochondrial Respiratory Chain Complex IV Deficiency	COX15	RECESSIVE	NM_004376.5	99.67%
Mitochondrial Respiratory Chain Complex IV Deficiency	COX6B1	RECESSIVE	NM_001863.4	99.85%
Mitochondrial Respiratory Chain Complex IV Deficiency	FASTKD2	RECESSIVE	NM_014929.3	99.32%
Mitochondrial Respiratory Chain Complex IV Deficiency	SCO1	RECESSIVE	NM_004589.2	97.37%
Mitochondrial Respiratory Chain Complex IV Deficiency	SCO2	RECESSIVE	NM_005138.2	97.71%
Miyoshi Muscular Dystrophy	ANO5	RECESSIVE	NM_213599.2	99.66%
Molybdenum Cofactor Deficiency	MOCS1	RECESSIVE	NM_005943.5	99.21%
Molybdenum Cofactor Deficiency	MOCS2	RECESSIVE	NM_176806.2	99.78%
Monilethrix	KRT83	DOMINANT	NM_002282.3	99.41%
Monogenic Non-Syndromic Obesity	LEP	RECESSIVE	NM_000230.2	98.22%

Monogenic Non-Syndromic Obesity	LEPR	RECESSIVE	NM_002303.5	98.34%
Monogenic Non-Syndromic Obesity	PCSK1	RECESSIVE	NM_000439.4	99.89%
Monogenic Non-Syndromic Obesity	POMC	RECESSIVE	NM_001035256.1	98.78%
Mononeuropathy of the Median Nerve	SH3TC2	RECESSIVE	NM_024577.3	99.34%
Mowat-Wilson Syndrome	ZEB2	DOMINANT	NM_014795.3	98.94%
Moyamoya Disease	ACTA2	DOMINANT	NM_001613.2	99.98%
MTHFR Deficiency	MTHFR	DOMINANT	NM_005957.4	98.66%
MTHFR Thermolabile Variant	MTHFR	DOMINANT	NM_005957.4	98.66%
Muckle-Wells Syndrome	NLRP3	DOMINANT	NM_004895.4	99.48%
Mucolipidosis, Type I	NEU1	RECESSIVE	NM_000434.3	99.66%
Mucolipidosis, Type II	GNPTAB	RECESSIVE	NM_024312.4	98.42%
Mucolipidosis, Type III Alpha/Beta	GNPTAB	RECESSIVE	NM_024312.4	98.42%
Mucolipidosis, Type III Gamma	GNPTG	X_LINKED	NM_032520.4	96.11%
Mucolipidosis, Type IV	MCOLN1	RECESSIVE	NM_020533.2	95.22%
Mucopolysaccharidosis, Type I	IDUA	RECESSIVE	NM_000203.3	92.99%
Mucopolysaccharidosis, Type III	GNS	RECESSIVE	NM_002076.3	99.69%
Mucopolysaccharidosis, Type III	HGSNAT	RECESSIVE	NM_152419.2	98.16%
Mucopolysaccharidosis, Type III	NAGLU	RECESSIVE	NM_000263.3	95.23%
Mucopolysaccharidosis, Type III	SGSH	RECESSIVE	NM_000199.3	95.57%
Mucopolysaccharidosis, Type IV	GALNS	RECESSIVE	NM_000512.4	96.52%
Mucopolysaccharidosis, Type IV	GLB1	RECESSIVE	NM_000404.2	99.90%
Mucopolysaccharidosis, Type IX	HYAL1	RECESSIVE	NM_153281.1	96.46%
Mucopolysaccharidosis, Type VI	ARSB	RECESSIVE	NM_000046.3	98.19%
Mucopolysaccharidosis, Type VII	GUSB	RECESSIVE	NM_000181.3	97.65%
Mulibrey Nanism	TRIM37	RECESSIVE	NM_015294.3	99.45%
Multicentric Carpotarsal Osteolysis Syndrome	MAFB	DOMINANT	NM_005461.3	98.66%
Multicentric Osteolysis of Torg	MMP2	RECESSIVE	NM_004530.4	97.78%
Multicentric Osteolysis, Nodulosis, and Arthropathy	MMP2	RECESSIVE	NM_004530.4	97.78%
Multiminicore Disease	RYR1	DOMINANT	NM_000540.2	97.36%

Multiple Acyl-CoA Dehydrogenase Deficiency	ETFA	RECESSIVE	NM_000126.3	99.46%
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFDH	RECESSIVE	NM_004453.2	99.69%
Multiple Cutaneous and Mucosal Venous Malformations	TEK	DOMINANT	NM_000459.3	99.92%
Multiple Cutaneous and Uterine Leiomyomas	FH	DOMINANT	NM_000143.3	99.47%
Multiple Endocrine Neoplasia	CDKN1B	DOMINANT	NM_004064.3	99.09%
Multiple Endocrine Neoplasia	MEN1	DOMINANT	NM_130799.2	96.55%
Multiple Endocrine Neoplasia	RET	DOMINANT	NM_020975.4	98.25%
Multiple Epiphyseal Dysplasia with Early-Onset Diabetes Mellitus	EIF2AK3	RECESSIVE	NM_004836.5	98.69%
Multiple Epiphyseal Dysplasia, Dominant	COL9A1	DOMINANT	NM_001851.4	99.62%
Multiple Epiphyseal Dysplasia, Dominant	COL9A2	DOMINANT	NM_001852.3	98.66%
Multiple Epiphyseal Dysplasia, Dominant	COL9A3	DOMINANT	NM_001853.3	95.56%
Multiple Epiphyseal Dysplasia, Dominant	COMP	DOMINANT	NM_000095.2	95.67%
Multiple Epiphyseal Dysplasia, Dominant	MATN3	DOMINANT	NM_002381.4	96.56%
Multiple Epiphyseal Dysplasia, Recessive	SLC26A2	DOMINANT	NM_000112.3	99.63%
Multiple Familial Trichoepithelioma	CYLD	DOMINANT	NM_015247.2	99.42%
Multiple Mitochondrial Dysfunctions Syndrome	BOLA3	RECESSIVE	NM_212552.2	98.99%
Multiple Mitochondrial Dysfunctions Syndrome	NFU1	RECESSIVE	NM_001002755.2	99.74%
Multiple Pterygium Syndrome	CHRNA1	RECESSIVE	NM_000079.3	99.50%
Multiple Pterygium Syndrome	CHRND	RECESSIVE	NM_000751.2	98.75%
Multiple Pterygium Syndrome	CHRNG	RECESSIVE	NM_005199.4	96.52%
Multiple Sulfatase Deficiency	SUMF1	RECESSIVE	NM_182760.3	99.06%
Multiple Synostoses Syndrome	FGF9	DOMINANT	NM_002010.2	97.21%
Multiple Synostoses Syndrome	GDF5	DOMINANT	NM_000557.2	98.65%
Multisystemic Smooth Muscle Dysfunction Syndrome	ACTA2	DOMINANT	NM_001613.2	99.98%
MYH9-related disorder	MYH9	DOMINANT	NM_002473.4	99.28%
MYH-Associated Polyposis	MUTYH	RECESSIVE	NM_001048171.1	99.52%
Myhre Syndrome	SMAD4	DOMINANT	NM_005359.5	98.23%
Myoadenylate Deaminase Deficiency	AMPD1	RECESSIVE	NM_000036.2	99.94%

Myoclonus-Dystonia	SGCE	DOMINANT	NM_003919.2	99.27%
Myofibrillar Myopathy, Dominant	BAG3	DOMINANT	NM_004281.3	97.40%
Myofibrillar Myopathy, Dominant	DNAJB6	DOMINANT	NM_058246.3	98.20%
Myofibrillar Myopathy, Dominant	LDB3	DOMINANT	NM_001080116.1	99.33%
Myofibrillar Myopathy, Dominant	MYOT	DOMINANT	NM_006790.2	99.03%
Myofibrillar Myopathy, Mixed	DES	DOMINANT	NM_001927.3	96.15%
Myofibrillar Myopathy, Recessive	CRYAB	DOMINANT	NM_001885.1	99.76%
Myokymia	KCNA1	DOMINANT	NM_000217.2	98.67%
Myopathy, Early-Onset, Areflexia, Respiratory Distress, and Dysphagia	MEGF10	RECESSIVE	NM_032446.2	99.87%
Myopathy, Lactic Acidosis, and Sideroblastic Anemia	YARS2	RECESSIVE	NM_001040436.2	98.74%
Myosclerosis	COL6A2	RECESSIVE	NM_001849.3	93.40%
Myosin Storage Myopathy	MYH7	DOMINANT	NM_000257.2	99.12%
Myostatin-Related Muscle Hypertrophy	MSTN	RECESSIVE	NM_005259.2	99.98%
Myotonia Congenita	CLCN1	DOMINANT	NM_000083.2	98.20%
N-Acetylglutamate Synthase Deficiency	NAGS	RECESSIVE	NM_153006.2	95.91%
Nager Syndrome	SF3B4	DOMINANT	NM_005850.4	99.66%
Nail-Patella Syndrome	LMX1B	DOMINANT	NM_002316.3	98.55%
Natural Killer Cell and Glucocorticoid Deficiency with DNA Repair Defect	MCM4	RECESSIVE	NM_005914.3	98.51%
Naxos Disease	JUP	RECESSIVE	NM_002230.2	98.34%
Nemaline Myopathy	TNNT1	DOMINANT	NM_003283.4	95.86%
Nemaline Myopathy	TPM2	DOMINANT	NM_003289.3	97.92%
Nemaline Myopathy	TPM3	DOMINANT	NM_152263.2	98.81%
Nemaline Myopathy, Dominant	ACTA1	DOMINANT	NM_001100.3	96.57%
Nemaline Myopathy, Dominant	KBTBD13	DOMINANT	NM_001101362.2	97.11%
Nemaline Myopathy, Recessive	CFL2	RECESSIVE	NM_021914.7	99.87%
Nemaline Myopathy, Recessive	NEB	RECESSIVE	NM_004543.4	99.75%
Neonatal Severe Primary Hyperparathyroidism	CASR	RECESSIVE	NM_000388.3	99.26%

Nephrogenic Diabetes Insipidus	AQP2	DOMINANT	NM_000486.5	99.39%
Nephronophthisis	GLIS2	RECESSIVE	NM_032575.2	97.92%
Nephronophthisis	INVS	RECESSIVE	NM_014425.3	99.52%
Nephronophthisis	NEK8	RECESSIVE	NM_178170.2	97.58%
Nephronophthisis	NPHP1	RECESSIVE	NM_000272.3	99.47%
Nephronophthisis	NPHP3	RECESSIVE	NM_153240.4	96.76%
Nephronophthisis	NPHP4	RECESSIVE	NM_015102.3	97.44%
Nephronophthisis	RPGRIP1L	RECESSIVE	NM_015272.2	99.57%
Nephronophthisis	TMEM67	RECESSIVE	NM_153704.5	99.40%
Nephronophthisis-Like Nephropathy	XPNPEP3	RECESSIVE	NM_022098.3	97.65%
Nephropathic Cystinosis	CTNS	RECESSIVE	NM_004937.2	97.66%
Nephrotic Syndrome	LAMB2	RECESSIVE	NM_002292.3	97.67%
Nephrotic Syndrome	PLCE1	RECESSIVE	NM_016341.3	99.31%
Nestor-Guillermo Progeria Syndrome	BANF1	RECESSIVE	NM_001143985.1	98.13%
Netherton Syndrome	SPINK5	RECESSIVE	NM_006846.3	99.82%
Neural Tube Defect	VANGL1	DOMINANT	NM_138959.2	98.53%
Neural Tube Defects, Folate-Sensitive	MTHFR	DOMINANT	NM_005957.4	98.66%
Neuroblastoma	KIF1B	DOMINANT	NM_015074.3	98.68%
Neuroblastoma	PHOX2B	DOMINANT	NM_003924.3	94.04%
Neuroblastoma Susceptibility	ALK	DOMINANT	NM_004304.4	97.38%
Neurodegeneration	PLA2G6	RECESSIVE	NM_003560.2	98.10%
Neurodegeneration due to Cerebral Folate Transport Deficiency	FOLR1	RECESSIVE	NM_016725.2	99.82%
Neuroferritinopathy	FTL	DOMINANT	NM_000146.3	98.97%
Neurofibromatosis, Type 1	NF1	DOMINANT	NM_000267.3	98.11%
Neurofibromatosis, Type 2	NF2	DOMINANT	NM_000268.3	96.91%
Neurofibromatosis-Noonan Syndrome	NF1	DOMINANT	NM_000267.3	98.11%
Neurogenic Scapuloperoneal Syndrome	DES	DOMINANT	NM_001927.3	96.15%
Neurologic Disorders/Seipinopathy	BSCL2	DOMINANT	NM_032667.6	98.94%

Neuronal Ceroid-Lipofuscinosis, Dominant/Recessive	CLN3	DOMINANT	NM_001042432.1	99.14%
Neuronal Ceroid-Lipofuscinosis, Dominant/Recessive	CLN5	DOMINANT	NM_006493.2	99.09%
Neuronal Ceroid-Lipofuscinosis, Dominant/Recessive	CTSD	DOMINANT	NM_001909.4	97.23%
Neuronal Ceroid-Lipofuscinosis, Recessive	CLN6	RECESSIVE	NM_017882.2	96.75%
Neuronal Ceroid-Lipofuscinosis, Recessive	DNAJC5	RECESSIVE	NM_025219.2	96.58%
Neuronal Ceroid-Lipofuscinosis, Recessive	MFSD8	RECESSIVE	NM_152778.2	98.86%
Neuronal Ceroid-Lipofuscinosis, Recessive	PPT1	RECESSIVE	NM_000310.3	98.74%
Neuronal Ceroid-Lipofuscinosis, Recessive	TPP1	RECESSIVE	NM_000391.3	99.30%
Neutral Lipid Storage Disease with Myopathy	PNPLA2	RECESSIVE	NM_020376.3	97.61%
Nevoid Basal Cell Carcinoma Syndrome	PTCH1	DOMINANT	NM_000264.3	97.68%
Newfoundland Rod-Cone Dystrophy	RLBP1	RECESSIVE	NM_000326.4	97.56%
Nicolaides-Baraitser Syndrome	SMARCA2	DOMINANT	NM_003070.3	97.97%
Niemann-Pick Disease	NPC1	RECESSIVE	NM_000271.4	99.29%
Niemann-Pick Disease	NPC2	RECESSIVE	NM_006432.3	96.54%
Nijmegen Breakage Syndrome	NBN	RECESSIVE	NM_002485.4	99.86%
Nocturnal Frontal Lobe Epilepsy	CHRNA2	DOMINANT	NM_000742.3	95.50%
Nonaka Myopathy	GNE	DOMINANT	NM_005476.5	95.71%
Nonautoimmune Hyperthyroidism	TSHR	DOMINANT	NM_000369.2	99.89%
Non-Classic Cystic Fibrosis-Like Syndrome	SCNN1B	DOMINANT	NM_000336.2	98.80%
Nonepidermolytic Palmoplantar Hyperkeratosis	KRT1	DOMINANT	NM_006121.3	96.60%
Nonspecific Cardiac Conduction Defect	SCN1B	DOMINANT	NM_001037.4	90.69%
Nonsyndromic Hearing Loss, Dominant	COCH	DOMINANT	NM_004086.2	99.71%
Nonsyndromic Hearing Loss, Dominant	COL11A2	DOMINANT	NM_080680.2	99.38%
Nonsyndromic Hearing Loss, Dominant	EYA4	DOMINANT	NM_172105.3	99.82%
Nonsyndromic Hearing Loss, Dominant	GJB2	DOMINANT	NM_004004.5	97.20%
Nonsyndromic Hearing Loss, Dominant	GJB3	DOMINANT	NM_024009.2	96.05%
Nonsyndromic Hearing Loss, Dominant	GJB6	DOMINANT	NM_006783.4	99.83%
Nonsyndromic Hearing Loss, Dominant	MYH14	DOMINANT	NM_024729.3	97.84%
Nonsyndromic Hearing Loss, Dominant	MYH9	DOMINANT	NM_002473.4	99.28%

Nonsyndromic Hearing Loss, Dominant	MYO1A	DOMINANT	NM_005379.2	99.92%
Nonsyndromic Hearing Loss, Dominant	MYO6	DOMINANT	NM_004999.3	99.22%
Nonsyndromic Hearing Loss, Dominant	MYO7A	DOMINANT	NM_000260.3	98.70%
Nonsyndromic Hearing Loss, Dominant	POU4F3	DOMINANT	NM_002700.2	94.81%
Nonsyndromic Hearing Loss, Dominant	SIX1	DOMINANT	NM_005982.3	98.47%
Nonsyndromic Hearing Loss, Dominant	SLC17A8	DOMINANT	NM_139319.2	98.94%
Nonsyndromic Hearing Loss, Dominant	TECTA	DOMINANT	NM_005422.2	98.28%
Nonsyndromic Hearing Loss, Dominant	TJP2	DOMINANT	NM_004817.3	99.24%
Nonsyndromic Hearing Loss, Dominant	TMC1	DOMINANT	NM_138691.2	99.22%
Nonsyndromic Hearing Loss, Dominant	WFS1	DOMINANT	NM_006005.3	86.69%
Nonsyndromic Hearing Loss, Mixed	DFNA5	DOMINANT	NM_004403.2	99.89%
Nonsyndromic Hearing Loss, Mixed	DIAPH1	DOMINANT	NM_005219.4	96.64%
Nonsyndromic Hearing Loss, Mixed	FOXI1	DOMINANT	NM_012188.4	98.39%
Nonsyndromic Hearing Loss, Mixed	HGF	DOMINANT	NM_000601.4	99.95%
Nonsyndromic Hearing Loss, Mixed	KCNJ10	DOMINANT	NM_002241.4	98.87%
Nonsyndromic Hearing Loss, Recessive	CDH23	RECESSIVE	NM_022124.5	97.08%
Nonsyndromic Hearing Loss, Recessive	CLDN14	RECESSIVE	NM_144492.2	96.37%
Nonsyndromic Hearing Loss, Recessive	DFNB31	RECESSIVE	NM_015404.3	96.78%
Nonsyndromic Hearing Loss, Recessive	DFNB59	RECESSIVE	NM_001042702.3	99.95%
Nonsyndromic Hearing Loss, Recessive	ESRRB	RECESSIVE	NM_004452.3	98.10%
Nonsyndromic Hearing Loss, Recessive	GJB2	RECESSIVE	NM_004004.5	97.20%
Nonsyndromic Hearing Loss, Recessive	GJB6	RECESSIVE	NM_006783.4	99.83%
Nonsyndromic Hearing Loss, Recessive	GPSM2	RECESSIVE	NM_013296.4	98.15%
Nonsyndromic Hearing Loss, Recessive	GRXCR1	RECESSIVE	NM_001080476.2	99.99%
Nonsyndromic Hearing Loss, Recessive	LHFPL5	RECESSIVE	NM_182548.3	94.21%
Nonsyndromic Hearing Loss, Recessive	LOXHD1	RECESSIVE	NM_144612.6	99.30%
Nonsyndromic Hearing Loss, Recessive	LRTOMT	RECESSIVE	NM_001145308.2	99.41%
Nonsyndromic Hearing Loss, Recessive	MARVELD2	RECESSIVE	NM_001038603.2	99.86%
Nonsyndromic Hearing Loss, Recessive	MYO15A	RECESSIVE	NM_016239.3	96.43%

Nonsyndromic Hearing Loss, Recessive	MYO3A	RECESSIVE	NM_017433.4	99.51%
Nonsyndromic Hearing Loss, Recessive	MYO6	RECESSIVE	NM_004999.3	99.22%
Nonsyndromic Hearing Loss, Recessive	MYO7A	RECESSIVE	NM_000260.3	98.70%
Nonsyndromic Hearing Loss, Recessive	OTOF	RECESSIVE	NM_194248.2	97.40%
Nonsyndromic Hearing Loss, Recessive	PCDH15	RECESSIVE	NM_033056.3	99.93%
Nonsyndromic Hearing Loss, Recessive	RDX	RECESSIVE	NM_002906.3	96.91%
Nonsyndromic Hearing Loss, Recessive	SLC26A4	RECESSIVE	NM_000441.1	98.71%
Nonsyndromic Hearing Loss, Recessive	SLC26A5	RECESSIVE	NM_198999.2	98.33%
Nonsyndromic Hearing Loss, Recessive	TECTA	RECESSIVE	NM_005422.2	98.28%
Nonsyndromic Hearing Loss, Recessive	TMC1	RECESSIVE	NM_138691.2	99.22%
Nonsyndromic Hearing Loss, Recessive	TMIE	RECESSIVE	NM_147196.2	98.19%
Nonsyndromic Hearing Loss, Recessive	TMPRSS3	RECESSIVE	NM_024022.2	99.81%
Nonsyndromic Hearing Loss, Recessive	USH1C	RECESSIVE	NM_005709.3	97.95%
Nonsyndromic Hearing Loss, X-Linked	POU3F4	X_LINKED	NM_000307.3	88.38%
Nonsyndromic Hearing Loss, X-Linked	PRPS1	X_LINKED	NM_002764.3	88.28%
Nonsyndromic Hearing Loss, X-Linked	SMPX	X_LINKED	NM_014332.2	95.35%
Nonsyndromic Trigonocephaly	FGFR1	DOMINANT	NM_023110.2	98.79%
Noonan Syndrome	BRAF	DOMINANT	NM_004333.4	97.81%
Noonan Syndrome	KRAS	DOMINANT	NM_004985.3	98.29%
Noonan Syndrome	MAP2K1	DOMINANT	NM_002755.3	98.72%
Noonan Syndrome	NRAS	DOMINANT	NM_002524.4	97.99%
Noonan Syndrome	PTPN11	DOMINANT	NM_002834.3	98.03%
Noonan Syndrome	RAF1	DOMINANT	NM_002880.3	99.39%
Noonan Syndrome	SOS1	DOMINANT	NM_005633.3	99.48%
Noonan-Like Syndrome Disorder	CBL	DOMINANT	NM_005188.3	99.04%
Noonan-Like Syndrome with Loose Anagen Hair	SHOC2	DOMINANT	NM_007373.3	97.64%
Norepinephrine Transporter Deficiency	SLC6A2	DOMINANT	NM_001043.3	98.92%
Normokalemic Periodic Paralysis	SCN4A	DOMINANT	NM_000334.4	97.03%
North American Indian Childhood Cirrhosis	CIRH1A	RECESSIVE	NM_032830.2	99.59%

Obesity	MC4R	DOMINANT	NM_005912.2	99.98%
Obesity	PPARG	DOMINANT	NM_015869.4	99.99%
Occult Macular Dystrophy	RP1L1	DOMINANT	NM_178857.5	97.29%
Oculocutaneous Albinism	OCA2	RECESSIVE	NM_000275.2	99.67%
Oculocutaneous Albinism	SLC45A2	RECESSIVE	NM_016180.3	98.35%
Oculocutaneous Albinism	TYRP1	RECESSIVE	NM_000550.2	99.81%
Oculodentodigital Dysplasia	GJA1	DOMINANT	NM_000165.3	95.58%
Odontoonychodermal Dysplasia	WNT10A	RECESSIVE	NM_025216.2	95.67%
Oguchi Disease	SAG	RECESSIVE	NM_000541.4	99.85%
Oligodontia-Colorectal Cancer Syndrome	AXIN2	DOMINANT	NM_004655.3	96.56%
Omenn Syndrome	DCLRE1C	RECESSIVE	NM_001033855.1	97.51%
Omenn Syndrome	RAG1	RECESSIVE	NM_000448.2	99.57%
Omenn Syndrome	RAG2	RECESSIVE	NM_000536.3	98.79%
Omodysplasia	GPC6	DOMINANT	NM_005708.3	99.89%
Optic Atrophy, Dominant	OPA3	DOMINANT	NM_025136.3	97.52%
Optic Atrophy, Recessive	OPA1	DOMINANT	NM_015560.2	99.89%
Optic Atrophy, Recessive	TMEM126A	RECESSIVE	NM_032273.3	99.99%
Ornithine Aminotransferase Deficiency	OAT	RECESSIVE	NM_000274.3	99.69%
Ornithine Transcarbamylase Deficiency	OTC	X_LINKED	NM_000531.5	95.17%
Orofacial Cleft	BMP4	DOMINANT	NM_001202.3	98.31%
Orotic Aciduria	UMPS	RECESSIVE	NM_000373.3	98.61%
Osteogenesis Imperfecta, Dominant	COL1A1	DOMINANT	NM_000088.3	96.26%
Osteogenesis Imperfecta, Dominant	COL1A2	DOMINANT	NM_000089.3	98.95%
Osteogenesis Imperfecta, Recessive	CRTAP	RECESSIVE	NM_006371.4	99.20%
Osteogenesis Imperfecta, Recessive	FKBP10	RECESSIVE	NM_021939.3	97.74%
Osteogenesis Imperfecta, Recessive	LEPRE1	RECESSIVE	NM_022356.3	99.60%
Osteogenesis Imperfecta, Recessive	PPIB	RECESSIVE	NM_000942.4	98.88%
Osteogenesis Imperfecta, Recessive	SERPINF1	RECESSIVE	NM_002615.5	99.56%
Osteogenesis Imperfecta, Recessive	SERPINH1	RECESSIVE	NM_001235.3	96.59%

Osteoglophonic Dysplasia	FGFR1	DOMINANT	NM_023110.2	98.79%
Osteopetrosis	CLCN7	DOMINANT	NM_001287.5	97.08%
Osteopetrosis	OSTM1	RECESSIVE	NM_014028.3	97.01%
Osteopetrosis	TCIRG1	RECESSIVE	NM_006019.3	95.51%
Osteopetrosis	TNFRSF11A	DOMINANT	NM_003839.2	96.20%
Osteopetrosis	TNFSF11	RECESSIVE	NM_003701.3	99.70%
Osteopetrosis with Renal Tubular Acidosis	CA2	RECESSIVE	NM_000067.2	95.07%
Otofaciocervical Syndrome	EYA1	DOMINANT	NM_000503.4	98.57%
Otospondylomegaepiphyseal Dysplasia	COL11A2	DOMINANT	NM_080680.2	99.38%
OTX2-Related Syndromic Microphthalmia	OTX2	DOMINANT	NM_172337.2	99.73%
Ovarian Dysgenesis	BMP15	X_LINKED	NM_005448.2	95.95%
Ovarian Dysgenesis	FSHR	RECESSIVE	NM_000145.3	98.20%
Ovarian Hyperstimulation Syndrome	FSHR	DOMINANT	NM_000145.3	98.20%
Paget Disease of Bone	SQSTM1	RECESSIVE	NM_003900.4	97.28%
Paget Disease of Bone	TNFRSF11A	DOMINANT	NM_003839.2	96.20%
PALB2-Related Cancer Susceptibility	PALB2	DOMINANT	NM_024675.3	99.95%
Pallister-Hall Syndrome	GLI3	DOMINANT	NM_000168.5	98.64%
Palmoplantar Keratoderma, Mutilating, with Periorificial Keratotic Plaques	TRPV3	DOMINANT	NM_145068.3	99.40%
Pancreatic Cancer	PALLD	DOMINANT	NM_001166108.1	99.67%
Pantothenate Kinase-Associated Neurodegeneration	PANK2	RECESSIVE	NM_153638.2	97.85%
Papillary Renal Carcinoma	MET	DOMINANT	NM_001127500.1	99.60%
Papillon-Lefevre Syndrome	CTSC	RECESSIVE	NM_001814.4	99.55%
Paraganglioma and Gastric Stromal Sarcoma	SDHB	DOMINANT	NM_003000.2	99.74%
Paramyotonia Congenita of Von Eulenburg	SCN4A	DOMINANT	NM_000334.4	97.03%
Parathyroid Carcinoma	CDC73	DOMINANT	NM_024529.4	99.19%
Parkes Weber Syndrome	RASA1	DOMINANT	NM_002890.2	97.65%
Parkinson Disease ,Recessive	FBXO7	RECESSIVE	NM_012179.3	97.36%
Parkinson Disease, Dominant	HTRA2	DOMINANT	NM_013247.4	98.65%

Parkinson Disease, Dominant	LRRK2	DOMINANT	NM_198578.3	99.67%
Parkinson Disease, Dominant	SNCA	DOMINANT	NM_000345.3	97.74%
Parkinson Disease, Dominant	VPS35	DOMINANT	NM_018206.4	97.74%
Parkinson Disease, Dominant/Recessive	MAPT	DOMINANT	NM_005910.5	96.95%
Parkinson Disease, Dominant/Recessive	NR4A2	DOMINANT	NM_006186.3	97.75%
Parkinson Disease, Dominant/Recessive	SNCAIP	DOMINANT	NM_005460.2	99.26%
Parkinson Disease, Dominant/Recessive	UCHL1	DOMINANT	NM_004181.4	99.21%
Parkinson Disease, Juvenile	PARK2	DOMINANT	NM_004562.2	98.57%
Parkinson Disease, Recessive	PARK7	RECESSIVE	NM_007262.4	97.37%
Parkinson Disease, Recessive	PINK1	RECESSIVE	NM_032409.2	94.52%
Paroxysmal Extreme Pain Disorder	SCN9A	DOMINANT	NM_002977.3	99.43%
Paroxysmal Familial Ventricular Fibrillation	SCN5A	DOMINANT	NM_198056.2	98.54%
Partial Epilepsy with Auditory Features	LGI1	DOMINANT	NM_005097.2	99.47%
Partial Isolated Growth Hormone Deficiency	GHSR	DOMINANT	NM_198407.2	95.92%
Patterned Dystrophy of Retinal Pigment Epithelium	PRPH2	DOMINANT	NM_000322.4	99.36%
PCWH Syndrome	SOX10	DOMINANT	NM_006941.3	96.64%
Peeling Skin Syndrome	TGM5	RECESSIVE	NM_201631.3	99.68%
Pendred Syndrome	FOXI1	RECESSIVE	NM_012188.4	98.39%
Pendred Syndrome	KCNJ10	RECESSIVE	NM_002241.4	98.87%
Pendred Syndrome	SLC26A4	RECESSIVE	NM_000441.1	98.71%
Periventricular Heterotopia	ARFGEF2	RECESSIVE	NM_006420.2	99.84%
Perlman Syndrome	DIS3L2	RECESSIVE	NM_152383.4	97.23%
Permanent Neonatal Diabetes Mellitus	ABCC8	DOMINANT	NM_000352.3	98.00%
Permanent Neonatal Diabetes Mellitus	GCK	DOMINANT	NM_000162.3	96.32%
Peroxisomal Bifunctional Enzyme Deficiency	HSD17B4	RECESSIVE	NM_000414.3	99.62%
Perrault Syndrome	HSD17B4	RECESSIVE	NM_000414.3	99.62%
Perry Syndrome	DCTN1	DOMINANT	NM_004082.4	99.86%
Peters Anomaly	CYP1B1	DOMINANT	NM_000104.3	98.08%
Peters Anomaly	PAX6	DOMINANT	NM_000280.4	99.26%

Peters Anomaly	PITX2	DOMINANT	NM_153427.2	99.12%
Peters Plus Syndrome	B3GALTL	RECESSIVE	NM_194318.3	96.61%
Peutz-Jeghers Syndrome	STK11	DOMINANT	NM_000455.4	88.22%
Pfeiffer Syndrome	FGFR1	DOMINANT	NM_023110.2	98.79%
Pfeiffer Syndrome	FGFR2	DOMINANT	NM_000141.4	97.52%
Phenylalanine Hydroxylase Deficiency	PAH	RECESSIVE	NM_000277.1	99.91%
Pheochromocytoma	KIF1B	DOMINANT	NM_015074.3	98.68%
Pheochromocytoma	RET	DOMINANT	NM_020975.4	98.25%
Pheochromocytoma	TMEM127	DOMINANT	NM_017849.3	99.48%
Phosphoenolpyruvate Carboxykinase Deficiency	PCK1	RECESSIVE	NM_002591.3	98.74%
Phosphoglycerate Dehydrogenase Deficiency	PHGDH	RECESSIVE	NM_006623.3	99.83%
Phosphoribosylpyrophosphate Synthetase Superactivity	PRPS1	X_LINKED	NM_002764.3	88.28%
Phosphorylase Kinase Deficiency	PHKB	RECESSIVE	NM_000293.2	99.20%
Phosphorylase Kinase Deficiency	PHKG2	X_LINKED	NM_000294.2	96.74%
Phosphoserine Aminotransferase Deficiency	PSAT1	RECESSIVE	NM_058179.2	99.64%
Phosphoserine Phosphatase Deficiency	PSPH	RECESSIVE	NM_004577.3	97.52%
Piebald Trait	KIT	DOMINANT	NM_000222.2	99.98%
Piebald Trait	SNAI2	DOMINANT	NM_003068.4	97.30%
Pierson Syndrome	LAMB2	RECESSIVE	NM_002292.3	97.67%
Pigmented Paravenous Chorioretinal Atrophy	CRB1	DOMINANT	NM_201253.2	99.98%
Pineal Hyperplasia, Insulin-Resistant Diabetes Mellitus, and Somatic Abnormalities	INSR	DOMINANT	NM_000208.2	97.67%
Pitt-Hopkins Syndrome	TCF4	DOMINANT	NM_001083962.1	95.46%
Pitt-Hopkins-Like Syndrome	CNTNAP2	RECESSIVE	NM_014141.5	99.23%
Pitt-Hopkins-Like Syndrome	NRXN1	RECESSIVE	NM_001135659.1	98.62%
Pituitary Dwarfism II	GHR	RECESSIVE	NM_000163.4	98.56%
PITX2-Related Eye Abnormalities	PITX2	DOMINANT	NM_153427.2	99.12%
Plasminogen Activator Inhibitor-1 Deficiency	SERPINE1	DOMINANT	NM_000602.3	97.56%
Platelet Glycoprotein IV Deficiency	CD36	RECESSIVE	NM_001001547.2	99.94%

Pleuropulmonary Blastoma	DICER1	DOMINANT	NM_177438.2	98.88%
Pol III-Related Leukodystrophy	POLR3A	RECESSIVE	NM_007055.3	99.59%
Pol III-Related Leukodystrophy	POLR3B	RECESSIVE	NM_018082.5	99.42%
POLG-Related Spectrum Disorders	POLG	RECESSIVE	NM_002693.2	99.36%
Polycystic Kidney Disease, Autosomal Dominant	PKD2	DOMINANT	NM_000297.3	95.67%
Polycystic Kidney Disease, Autosomal Recessive	PKHD1	RECESSIVE	NM_138694.3	99.59%
Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy	TREM2	RECESSIVE	NM_018965.2	98.73%
Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy	TYROBP	RECESSIVE	NM_003332.3	99.41%
Polycystic Liver Disease	PRKCSH	DOMINANT	NM_002743.2	97.12%
Polycystic Liver Disease	SEC63	DOMINANT	NM_007214.4	97.49%
Polydactyly	GLI3	DOMINANT	NM_000168.5	98.64%
Polymicrogyria	GPR56	RECESSIVE	NM_005682.5	98.00%
Pontocerebellar Hypoplasia	RARS2	RECESSIVE	NM_020320.3	99.04%
Pontocerebellar Hypoplasia	TSEN2	RECESSIVE	NM_025265.3	99.94%
Pontocerebellar Hypoplasia	TSEN34	RECESSIVE	NM_024075.3	98.60%
Pontocerebellar Hypoplasia	TSEN54	RECESSIVE	NM_207346.2	95.27%
Pontocerebellar Hypoplasia	VRK1	RECESSIVE	NM_003384.2	98.56%
Popliteal Pterygium Syndrome	IRF6	DOMINANT	NM_006147.3	99.06%
Porencephaly	COL4A1	DOMINANT	NM_001845.4	99.41%
Porencephaly	COL4A2	DOMINANT	NM_001846.2	98.18%
Porphyria Cutanea Tarda	UROD	DOMINANT	NM_000374.4	99.94%
Postaxial Acrofacial Dysostosis	DHODH	RECESSIVE	NM_001361.4	99.51%
Posterior Column Ataxia with Retinitis Pigmentosa	FLVCR1	RECESSIVE	NM_014053.3	97.09%
Posterior Polar Cataract	CRYAB	DOMINANT	NM_001885.1	99.76%
Posterior Polymorphous Corneal Dystrophy	VSX1	DOMINANT	NM_014588.5	97.29%
Potassium-Aggravated Myotonia	SCN4A	DOMINANT	NM_000334.4	97.03%
Prader-Willi-Like Syndrome	SIM1	DOMINANT	NM_005068.2	99.52%

Premature Ovarian Failure	FIGLA	DOMINANT	NM_001004311.3	98.80%
Premature Ovarian Failure	NOBOX	DOMINANT	NM_001080413.3	99.70%
Primary Ciliary Dyskinesia	CCDC103	RECESSIVE	NM_213607.2	99.77%
Primary Ciliary Dyskinesia	CCDC39	RECESSIVE	NM_181426.1	99.88%
Primary Ciliary Dyskinesia	CCDC40	RECESSIVE	NM_017950.3	98.95%
Primary Ciliary Dyskinesia	DNAAF1	RECESSIVE	NM_178452.4	98.00%
Primary Ciliary Dyskinesia	DNAAF2	RECESSIVE	NM_018139.2	99.04%
Primary Ciliary Dyskinesia	DNAAF3	RECESSIVE	NM_001256714.1	97.46%
Primary Ciliary Dyskinesia	DNAH11	RECESSIVE	NM_003777.3	99.69%
Primary Ciliary Dyskinesia	DNAH5	RECESSIVE	NM_001369.2	99.64%
Primary Ciliary Dyskinesia	DNAI1	RECESSIVE	NM_012144.2	99.59%
Primary Ciliary Dyskinesia	DNAI2	RECESSIVE	NM_023036.4	99.66%
Primary Ciliary Dyskinesia	DNAL1	RECESSIVE	NM_031427.3	98.71%
Primary Ciliary Dyskinesia	RSPH4A	RECESSIVE	NM_001010892.2	99.95%
Primary Coenzyme Q10 Deficiency	COQ2	RECESSIVE	NM_015697.7	95.98%
Primary Coenzyme Q10 Deficiency	COQ9	RECESSIVE	NM_020312.3	97.91%
Primary Coenzyme Q10 Deficiency	PDSS1	RECESSIVE	NM_014317.3	98.93%
Primary Coenzyme Q10 Deficiency	PDSS2	RECESSIVE	NM_020381.3	99.91%
Primary Congenital Glaucoma	CYP1B1	RECESSIVE	NM_000104.3	98.08%
Primary Congenital Glaucoma	LTBP2	RECESSIVE	NM_000428.2	99.04%
Primary Hyperoxaluria	AGXT	RECESSIVE	NM_000030.2	97.95%
Primary Hyperoxaluria	GRHPR	RECESSIVE	NM_012203.1	96.59%
Primary Hyperoxaluria	HOGA1	RECESSIVE	NM_138413.3	98.33%
Primary Hypertrophic Osteoarthropathy	HPGD	RECESSIVE	NM_000860.5	99.68%
Primary Hypomagnesemia	CLDN16	RECESSIVE	NM_006580.3	99.50%
Primary Lymphedema with Myelodysplasia	GATA2	DOMINANT	NM_032638.4	97.67%
Primary Microcephaly, Recessive	ASPM	RECESSIVE	NM_018136.4	99.32%
Primary Microcephaly, Recessive	CDK5RAP2	RECESSIVE	NM_018249.4	99.64%
Primary Microcephaly, Recessive	CENPJ	RECESSIVE	NM_018451.4	99.50%

Primary Microcephaly, Recessive	CEP152	RECESSIVE	NM_014985.3	99.51%
Primary Microcephaly, Recessive	MCPH1	RECESSIVE	NM_024596.3	99.74%
Primary Microcephaly, Recessive	STIL	RECESSIVE	NM_003035.2	99.35%
Primary Microcephaly, Recessive	WDR62	RECESSIVE	NM_001083961.1	98.54%
Primary Open Angle Glaucoma	MYOC	DOMINANT	NM_000261.1	99.85%
Primary Open Angle Glaucoma	OPTN	DOMINANT	NM_021980.4	99.01%
Primary Open Angle Glaucoma	WDR36	DOMINANT	NM_139281.2	99.26%
Progeroid Laminopathies	LMNA	DOMINANT	NM_005572.3	96.33%
Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	C10orf2	DOMINANT	NM_021830.4	98.46%
Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	POLG2	DOMINANT	NM_007215.3	99.53%
Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	RRM2B	DOMINANT	NM_015713.4	97.04%
Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions	SLC25A4	DOMINANT	NM_001151.3	99.43%
Progressive Familial Heart Block	SCN5A	DOMINANT	NM_198056.2	98.54%
Progressive Familial Heart Block	TRPM4	DOMINANT	NM_017636.3	97.92%
Progressive Familial Intrahepatic Cholestasis	ABCB4	DOMINANT	NM_000443.3	99.79%
Progressive Myoclonic Epilepsy	GOSR2	RECESSIVE	NM_004287.3	99.82%
Progressive Myoclonic Epilepsy	KCTD7	RECESSIVE	NM_153033.4	99.13%
Progressive Myoclonic Epilepsy	PRICKLE2	DOMINANT	NM_198859.3	99.37%
Progressive Myoclonus Epilepsy with Ataxia	PRICKLE1	RECESSIVE	NM_153026.2	99.11%
Progressive Myoclonus Epilepsy, Lafora type	NHLRC1	RECESSIVE	NM_198586.2	98.06%
Progressive Pseudorheumatoid Arthropathy of Childhood	WISP3	RECESSIVE	NM_003880.3	99.98%
Prolidase Deficiency	PEPD	RECESSIVE	NM_000285.3	98.19%
Proliferative Vasculopathy And Hydranencephaly-Hydrocephaly Syndrome	FLVCR2	RECESSIVE	NM_017791.2	98.91%
Proopiomelanocortin Deficiency	POMC	RECESSIVE	NM_001035256.1	98.78%
Propionic Acidemia	PCCA	RECESSIVE	NM_000282.3	99.35%

Propionic Acidemia	PCCB	RECESSIVE	NM_000532.4	99.74%
Proprotein Convertase-1 Deficiency	PCSK1	RECESSIVE	NM_000439.4	99.89%
Protein C Deficiency	PROC	DOMINANT	NM_000312.3	98.77%
Protein S Deficiency	PROS1	DOMINANT	NM_000313.3	95.46%
Prothrombin Deficiency	F2	RECESSIVE	NM_000506.3	99.50%
Prothrombin-Related Thrombophilia	F2	DOMINANT	NM_000506.3	99.50%
Proximal Renal Tubular Acidosis with Ocular Abnormalities	SLC4A4	RECESSIVE	NM_003759.3	99.83%
Pseudoachondroplasia	COMP	DOMINANT	NM_000095.2	95.67%
Pseudohypoaldosteronism, Type I, Dominant	CUL3	DOMINANT	NM_003590.4	96.27%
Pseudohypoaldosteronism, Type I, Dominant	KLHL3	DOMINANT	NM_017415.2	98.37%
Pseudohypoaldosteronism, Type I, Dominant	NR3C2	DOMINANT	NM_000901.4	98.94%
Pseudohypoaldosteronism, Type I, Dominant	STX16	DOMINANT	NM_001001433.2	99.21%
Pseudohypoaldosteronism, Type I, Recessive	SCNN1A	RECESSIVE	NM_001038.5	97.55%
Pseudohypoaldosteronism, Type I, Recessive	SCNN1B	DOMINANT	NM_000336.2	98.80%
Pseudohypoaldosteronism, Type II	WNK1	DOMINANT	NM_018979.3	98.81%
Pseudohypoaldosteronism, Type II	WNK4	DOMINANT	NM_032387.4	98.04%
Pseudoinflammatory Fundus Dystrophy	TIMP3	RECESSIVE	NM_000362.4	97.77%
Pseudoneonatal Adrenoleukodystrophy	ACOX1	RECESSIVE	NM_004035.6	98.76%
PTEN Hamartoma Tumor Syndrome	PTEN	DOMINANT	NM_000314.4	91.64%
Pulmonary Surfactant Metabolism Dysfunction, Dominant	SFTPC	DOMINANT	NM_003018.3	98.49%
Pulmonary Surfactant Metabolism Dysfunction, Recessive	ABCA3	RECESSIVE	NM_001089.2	96.99%
Pulmonary Surfactant Metabolism Dysfunction, Recessive	SFTPB	RECESSIVE	NM_198843.2	99.30%
Purine Nucleoside Phosphorylase Deficiency	PNP	RECESSIVE	NM_000270.3	99.77%
Pycnodysostosis	CTSK	RECESSIVE	NM_000396.3	99.97%
Pyogenic Sterile Arthritis, Pyoderma Gangrenosum, and Acne	PSTPIP1	DOMINANT	NM_003978.3	98.87%
Pyridoxamine 5-Prime-Phosphate Oxidase Deficiency	PNPO	RECESSIVE	NM_018129.3	98.35%
Pyridoxine-Dependent Epilepsy	ALDH7A1	RECESSIVE	NM_001182.4	97.97%
Pyridoxine-Refractory Sideroblastic Anemia	SLC25A38	RECESSIVE	NM_017875.2	99.87%

Pyruvate Carboxylase Deficiency	PC	RECESSIVE	NM_000920.3	98.59%
Pyruvate Dehydrogenase Complex Deficiency	DLAT	RECESSIVE	NM_001931.4	98.17%
Pyruvate Dehydrogenase Complex Deficiency	DLD	RECESSIVE	NM_000108.3	99.95%
Pyruvate Dehydrogenase Complex Deficiency	PDHB	RECESSIVE	NM_000925.3	98.96%
Pyruvate Dehydrogenase Complex Deficiency	PDHX	RECESSIVE	NM_003477.2	97.24%
Pyruvate Dehydrogenase Phosphatase Deficiency	PDP1	RECESSIVE	NM_018444.3	98.87%
Pyruvate Kinase Deficiency	PKLR	RECESSIVE	NM_000298.5	99.53%
Quebec Platelet Disorder	PLAU	DOMINANT	NM_002658.3	98.56%
Rapid-Onset Dystonia-Parkinsonism	ATP1A3	DOMINANT	NM_152296.4	97.26%
Recurrent Hydatidiform Mole	NLRP7	RECESSIVE	NM_206828.3	98.15%
Refsum Disease	PEX7	RECESSIVE	NM_000288.3	99.70%
Refsum Disease	PHYH	RECESSIVE	NM_006214.3	99.94%
Renal Adysplasia	RET	DOMINANT	NM_020975.4	98.25%
Renal Adysplasia	UPK3A	DOMINANT	NM_006953.3	99.60%
Renal Cysts and Diabetes Syndrome	HNF1B	DOMINANT	NM_000458.2	98.62%
Renal Glucosuria	SLC5A2	RECESSIVE	NM_003041.3	97.88%
Renal Hypomagnesemia, Dominant	CNNM2	DOMINANT	NM_017649.4	96.70%
Renal Hypomagnesemia, Dominant	FXYD2	DOMINANT	NM_001680.4	98.81%
Renal Hypomagnesemia, Recessive	CLDN19	RECESSIVE	NM_148960.2	98.04%
Renal Hypomagnesemia, Recessive	EGF	RECESSIVE	NM_001963.4	99.78%
Renal Hypouricemia	SLC22A12	RECESSIVE	NM_144585.2	97.52%
Renal Hypouricemia	SLC2A9	RECESSIVE	NM_020041.2	99.76%
Renal Tubular Dysgenesis	ACE	RECESSIVE	NM_000789.3	97.17%
Renal Tubular Dysgenesis	AGT	RECESSIVE	NM_000029.3	99.37%
Renal Tubular Dysgenesis	AGTR1	RECESSIVE	NM_031850.3	99.73%
Renal Tubular Dysgenesis	REN	RECESSIVE	NM_000537.3	99.84%
Renal-Hepatic-Pancreatic Dysplasia	NPHP3	RECESSIVE	NM_153240.4	96.76%
Retinal Cone Dystrophy	CACNA2D4	RECESSIVE	NM_172364.4	98.75%
Retinal Cone Dystrophy	PDE6H	DOMINANT	NM_006205.2	99.98%

Retinal Degeneration	C1QTNF5	DOMINANT	NM_015645.3	98.05%
Retinal Dystrophy	LRAT	RECESSIVE	NM_004744.3	98.57%
Retinal Dystrophy	OTX2	DOMINANT	NM_172337.2	99.73%
Retinal Macular Dystrophy	PROM1	DOMINANT	NM_006017.2	99.87%
Retinitis Pigmentosa, Dominant	ABCA4	DOMINANT	NM_000350.2	99.79%
Retinitis Pigmentosa, Dominant	AIPL1	DOMINANT	NM_014336.3	98.86%
Retinitis Pigmentosa, Dominant	CLRN1	DOMINANT	NM_174878.2	99.54%
Retinitis Pigmentosa, Dominant	CNGA1	DOMINANT	NM_000087.3	99.80%
Retinitis Pigmentosa, Dominant	CNGB1	DOMINANT	NM_001297.4	98.93%
Retinitis Pigmentosa, Dominant	CRX	DOMINANT	NM_000554.4	96.57%
Retinitis Pigmentosa, Dominant	GUCA1B	DOMINANT	NM_002098.5	98.92%
Retinitis Pigmentosa, Dominant	IMPDH1	DOMINANT	NM_000883.3	96.28%
Retinitis Pigmentosa, Dominant	KLHL7	DOMINANT	NM_001031710.2	99.86%
Retinitis Pigmentosa, Dominant	NRL	DOMINANT	NM_006177.3	99.13%
Retinitis Pigmentosa, Dominant	PRPF3	DOMINANT	NM_004698.2	98.97%
Retinitis Pigmentosa, Dominant	PRPF31	DOMINANT	NM_015629.3	97.25%
Retinitis Pigmentosa, Dominant	PRPF6	DOMINANT	NM_012469.3	99.48%
Retinitis Pigmentosa, Dominant	PRPF8	DOMINANT	NM_006445.3	99.46%
Retinitis Pigmentosa, Dominant	PRPH2	DOMINANT	NM_000322.4	99.36%
Retinitis Pigmentosa, Dominant	ROM1	DOMINANT	NM_000327.3	96.13%
Retinitis Pigmentosa, Dominant	RP1	DOMINANT	NM_006269.1	99.69%
Retinitis Pigmentosa, Dominant	SNRNP200	DOMINANT	NM_014014.4	99.30%
Retinitis Pigmentosa, Dominant	TOPORS	DOMINANT	NM_005802.4	99.19%
Retinitis Pigmentosa, Dominant	ZNF513	DOMINANT	NM_144631.5	95.17%
Retinitis Pigmentosa, Dominant/Recessive	RHO	DOMINANT	NM_000539.3	99.45%
Retinitis Pigmentosa, Recessive	AIPL1	RECESSIVE	NM_014336.3	98.86%
Retinitis Pigmentosa, Recessive	ARL6	RECESSIVE	NM_177976.1	99.79%
Retinitis Pigmentosa, Recessive	BEST1	RECESSIVE	NM_004183.3	99.21%
Retinitis Pigmentosa, Recessive	C2orf71	RECESSIVE	NM_001029883.2	98.48%

Retinitis Pigmentosa, Recessive	C8orf37	RECESSIVE	NM_177965.3	98.57%
Retinitis Pigmentosa, Recessive	CERKL	RECESSIVE	NM_201548.4	99.89%
Retinitis Pigmentosa, Recessive	CRB1	DOMINANT	NM_201253.2	99.98%
Retinitis Pigmentosa, Recessive	DHDDS	RECESSIVE	NM_024887.3	98.08%
Retinitis Pigmentosa, Recessive	EYS	RECESSIVE	NM_001142800.1	99.94%
Retinitis Pigmentosa, Recessive	FAM161A	RECESSIVE	NM_032180.2	98.75%
Retinitis Pigmentosa, Recessive	IDH3B	RECESSIVE	NM_006899.3	99.44%
Retinitis Pigmentosa, Recessive	IMPG2	RECESSIVE	NM_016247.3	98.78%
Retinitis Pigmentosa, Recessive	LRAT	RECESSIVE	NM_004744.3	98.57%
Retinitis Pigmentosa, Recessive	MAK	RECESSIVE	NM_001242957.1	98.36%
Retinitis Pigmentosa, Recessive	MERTK	RECESSIVE	NM_006343.2	99.67%
Retinitis Pigmentosa, Recessive	NR2E3	RECESSIVE	NM_014249.2	98.63%
Retinitis Pigmentosa, Recessive	PDE6A	RECESSIVE	NM_000440.2	98.76%
Retinitis Pigmentosa, Recessive	PDE6B	RECESSIVE	NM_000283.3	97.01%
Retinitis Pigmentosa, Recessive	PDE6G	RECESSIVE	NM_002602.3	99.18%
Retinitis Pigmentosa, Recessive	PRCD	RECESSIVE	NM_001077620.2	98.33%
Retinitis Pigmentosa, Recessive	PROM1	RECESSIVE	NM_006017.2	99.87%
Retinitis Pigmentosa, Recessive	RBP3	RECESSIVE	NM_002900.2	97.14%
Retinitis Pigmentosa, Recessive	RDH12	RECESSIVE	NM_152443.2	99.96%
Retinitis Pigmentosa, Recessive	RGR	RECESSIVE	NM_001012720.1	99.51%
Retinitis Pigmentosa, Recessive	RLBP1	DOMINANT	NM_000326.4	97.56%
Retinitis Pigmentosa, Recessive	RPE65	RECESSIVE	NM_000329.2	99.90%
Retinitis Pigmentosa, Recessive	SAG	RECESSIVE	NM_000541.4	99.85%
Retinitis Pigmentosa, Recessive	SEMA4A	RECESSIVE	NM_022367.3	99.00%
Retinitis Pigmentosa, Recessive	SPATA7	RECESSIVE	NM_018418.4	99.91%
Retinitis Pigmentosa, Recessive	TTC8	RECESSIVE	NM_198309.2	99.65%
Retinitis Pigmentosa, Recessive	TULP1	RECESSIVE	NM_003322.3	98.64%
Retinitis Pigmentosa, Recessive	USH2A	RECESSIVE	NM_007123.5	99.98%
Retinitis Pigmentosa, X-Linked	CA4	X_LINKED	NM_000717.3	99.36%

Retinitis Pigmentosa, X-Linked	RP2	X_LINKED	NM_006915.2	89.45%
Retinoblastoma	RB1	DOMINANT	NM_000321.2	99.08%
Revesz Syndrome	TINF2	DOMINANT	NM_001099274.1	99.77%
Rhabdoid Tumor Predisposition Syndrome	SMARCB1	DOMINANT	NM_003073.3	95.58%
Rhizomelic Chondrodysplasia Punctata	AGPS	RECESSIVE	NM_003659.3	97.98%
Rhizomelic Chondrodysplasia Punctata	GNPAT	RECESSIVE	NM_014236.3	99.82%
Rhizomelic Chondrodysplasia Punctata	PEX7	RECESSIVE	NM_000288.3	99.70%
Ribose 5-Phosphate Isomerase Deficiency	RPIA	DOMINANT	NM_144563.2	99.31%
Ring Dermoid of Cornea	PITX2	DOMINANT	NM_153427.2	99.12%
Roberts Syndrome	ESCO2	RECESSIVE	NM_001017420.2	97.69%
Robinow Syndrome	ROR2	DOMINANT	NM_004560.3	98.00%
Robinow Syndrome	WNT5A	DOMINANT	NM_003392.4	96.82%
Romano-Ward Syndrome	AKAP9	DOMINANT	NM_005751.4	99.71%
Romano-Ward Syndrome	CAV3	DOMINANT	NM_033337.2	99.33%
Romano-Ward Syndrome	KCNE1	DOMINANT	NM_000219.3	99.96%
Romano-Ward Syndrome	KCNE2	DOMINANT	NM_172201.1	100.00%
Romano-Ward Syndrome	KCNJ5	DOMINANT	NM_000890.3	95.90%
Romano-Ward Syndrome	KCNQ1	DOMINANT	NM_000218.2	92.10%
Romano-Ward Syndrome	SCN4B	DOMINANT	NM_174934.3	98.40%
Romano-Ward Syndrome	SCN5A	DOMINANT	NM_198056.2	98.54%
Romano-Ward Syndrome	SNTA1	DOMINANT	NM_003098.2	89.35%
Rotor Syndrome	SLCO1B1	RECESSIVE	NM_006446.4	98.23%
Rotor Syndrome	SLCO1B3	RECESSIVE	NM_019844.3	99.17%
Roussy-Levy Syndrome	MPZ	DOMINANT	NM_000530.6	98.87%
Rubinstein-Taybi Syndrome	EP300	DOMINANT	NM_001429.3	97.56%
Saethre-Chotzen Syndrome	FGFR2	DOMINANT	NM_000141.4	97.52%
Salih Myopathy	TTN	RECESSIVE	NM_133378.4	99.55%
SALL4-Related Spectrum Disorders	SALL4	DOMINANT	NM_020436.3	99.42%
Salla Disease	SLC17A5	RECESSIVE	NM_012434.4	98.00%

Sandhoff Disease	HEXB	RECESSIVE	NM_000521.3	96.82%
SCAD Deficiency	ACADS	RECESSIVE	NM_000017.2	98.96%
Scapuloperoneal Myopathy	MYH7	DOMINANT	NM_000257.2	99.12%
Scapuloperoneal Spinal Muscular Atrophy	TRPV4	DOMINANT	NM_021625.4	97.17%
Schimke Immunoosseous Dysplasia	SMARCAL1	RECESSIVE	NM_014140.3	99.41%
Schindler Disease	NAGA	RECESSIVE	NM_000262.2	99.76%
Schinzel-Giedion Midface Retraction Syndrome	SETBP1	DOMINANT	NM_015559.2	97.43%
Schnyder Crystalline Corneal Dystrophy	UBIAD1	DOMINANT	NM_013319.2	98.34%
Schopf-Schulz-Passarge Syndrome	WNT10A	RECESSIVE	NM_025216.2	95.67%
Schwannomatosis	SMARCB1	DOMINANT	NM_003073.3	95.58%
Schwartz-Jampel Syndrome	HSPG2	RECESSIVE	NM_005529.5	96.57%
Sclerosing Bone Dysplasias	SOST	RECESSIVE	NM_025237.2	98.71%
Sebastian Syndrome	MYH9	DOMINANT	NM_002473.4	99.28%
Seckel Syndrome	ATR	RECESSIVE	NM_001184.3	99.44%
Seckel Syndrome	CENPJ	RECESSIVE	NM_018451.4	99.50%
Seckel Syndrome	CEP152	RECESSIVE	NM_014985.3	99.51%
Seckel Syndrome	RBBP8	RECESSIVE	NM_002894.2	99.22%
Seizure Disorders	SCN1A	DOMINANT	NM_001165963.1	99.76%
Seizures, Sensorineural Deafness, Ataxia, Mental Retardation, and Electrolyte Imbalance Syndrome	KCNJ10	RECESSIVE	NM_002241.4	98.87%
Selective Tooth Agenesis	PAX9	DOMINANT	NM_006194.3	98.61%
Selective Tooth Agenesis	WNT10A	DOMINANT	NM_025216.2	95.67%
Sengers Syndrome	AGK	RECESSIVE	NM_018238.3	99.36%
Senior-Loken Syndrome	CEP290	RECESSIVE	NM_025114.3	99.36%
Senior-Loken Syndrome	IQCB1	RECESSIVE	NM_001023570.2	99.67%
Senior-Loken Syndrome	NPHP1	RECESSIVE	NM_000272.3	99.47%
Senior-Loken Syndrome	NPHP4	RECESSIVE	NM_015102.3	97.44%
Senior-Loken Syndrome	SDCCAG8	RECESSIVE	NM_006642.3	98.99%
Sensory Neuropathy with Spastic Paraplegia	CCT5	RECESSIVE	NM_012073.3	99.90%

Septooptic Dysplasia	HESX1	RECESSIVE	NM_003865.2	99.85%
Severe Combined Immune Deficiency	CD3D	RECESSIVE	NM_000732.4	99.88%
Severe Combined Immune Deficiency	CD3E	RECESSIVE	NM_000733.3	99.58%
Severe Combined Immune Deficiency	IL7R	RECESSIVE	NM_002185.3	99.99%
Severe Combined Immune Deficiency	JAK3	RECESSIVE	NM_000215.3	95.34%
Severe Combined Immune Deficiency	RAG1	RECESSIVE	NM_000448.2	99.57%
Severe Combined Immune Deficiency	RAG2	RECESSIVE	NM_000536.3	98.79%
Severe Combined Immunodeficiency	IL2RG	RECESSIVE	NM_000206.2	95.44%
Severe Combined Immunodeficiency	ZAP70	RECESSIVE	NM_001079.3	97.01%
Severe Combined Immunodeficiency with Sensitivity to Ionizing Radiation	LIG4	RECESSIVE	NM_002312.3	99.86%
Severe Congenital Neutropenia	G6PC3	DOMINANT	NM_138387.3	98.06%
Severe Congenital Neutropenia	GFI1	DOMINANT	NM_005263.3	95.47%
Severe Congenital Neutropenia	HAX1	DOMINANT	NM_006118.3	99.95%
Short QT Syndrome	KCNJ2	DOMINANT	NM_000891.2	98.32%
Short QT Syndrome	KCNQ1	DOMINANT	NM_000218.2	92.10%
Short Rib Polydactyly Syndrome	DYNC2H1	RECESSIVE	NM_001080463.1	99.44%
Short Rib Polydactyly Syndrome	NEK1	RECESSIVE	NM_012224.2	99.45%
Short Rib Polydactyly Syndrome	WDR35	RECESSIVE	NM_001006657.1	99.96%
Sialuria	GNE	DOMINANT	NM_005476.5	95.71%
Sick Sinus Syndrome	SCN5A	DOMINANT	NM_198056.2	98.54%
Sickle Cell Disease	HBB	RECESSIVE	NM_000518.4	99.96%
Sideroblastic Anemia and Ataxia	ABCB7	X_LINKED	NM_004299.3	96.52%
Sitosterolemia	ABCG5	RECESSIVE	NM_022436.2	99.52%
Sitosterolemia	ABCG8	RECESSIVE	NM_022437.2	99.82%
Sjogren-Larsson Syndrome	ALDH3A2	RECESSIVE	NM_000382.2	99.85%
Skeletal Dysplasia	CHST3	RECESSIVE	NM_004273.4	98.22%
Skin Fragility-Woolly Hair Syndrome	DSP	RECESSIVE	NM_004415.2	99.77%
SLC6A4-Related Behavior Disorders	SLC6A4	DOMINANT	NM_001045.4	99.17%

Small Fiber Neuropathy	SCN9A	DOMINANT	NM_002977.3	99.43%
Small Patella Syndrome	TBX4	DOMINANT	NM_018488.2	98.79%
Smith-Lemli-Opitz Syndrome	DHCR7	RECESSIVE	NM_001360.2	98.64%
Smith-McCort Dysplasia	DYM	RECESSIVE	NM_017653.3	99.04%
Sotos Syndrome	NSD1	DOMINANT	NM_022455.4	98.84%
Spastic Paraplegia, Dominant	ATL1	DOMINANT	NM_015915.4	99.91%
Spastic Paraplegia, Dominant	HSPD1	DOMINANT	NM_002156.4	96.58%
Spastic Paraplegia, Dominant	KIAA0196	DOMINANT	NM_014846.3	99.91%
Spastic Paraplegia, Dominant	KIF5A	DOMINANT	NM_004984.2	99.77%
Spastic Paraplegia, Dominant	NIPA1	DOMINANT	NM_144599.4	97.06%
Spastic Paraplegia, Dominant	SLC33A1	DOMINANT	NM_004733.3	97.03%
Spastic Paraplegia, Dominant	SPAST	DOMINANT	NM_014946.3	96.86%
Spastic Paraplegia, Dominant	ZFYVE27	DOMINANT	NM_001002261.3	99.72%
Spastic Paraplegia, Recessive	CYP7B1	RECESSIVE	NM_004820.3	99.48%
Spastic Paraplegia, Recessive	KIF1A	RECESSIVE	NM_004321.6	98.68%
Spastic Paraplegia, Recessive	PNPLA6	RECESSIVE	NM_006702.4	95.80%
Spastic Paraplegia, Recessive	SPG11	RECESSIVE	NM_025137.3	99.55%
Spastic Paraplegia, Recessive	SPG7	RECESSIVE	NM_003119.2	98.19%
Spastic Paraplegia, Recessive	ZFYVE26	RECESSIVE	NM_015346.3	99.35%
Spastic Quadriplegic Cerebral Palsy	GAD1	RECESSIVE	NM_000817.2	97.76%
Speech-Language Disorder	FOXP2	DOMINANT	NM_014491.3	99.15%
Spermatogenic Failure	AURKC	RECESSIVE	NM_001015878.1	96.35%
Spermatogenic Failure	SPATA16	RECESSIVE	NM_031955.5	99.96%
Spherocytosis, Dominant	ANK1	DOMINANT	NM_000037.3	97.51%
Spherocytosis, Dominant	SLC4A1	DOMINANT	NM_000342.3	97.69%
Spherocytosis, Dominant	SPTB	DOMINANT	NM_000347.5	98.26%
Spherocytosis, Recessive	EPB42	RECESSIVE	NM_000119.2	99.81%
Spherocytosis, Recessive	SPTA1	RECESSIVE	NM_003126.2	99.65%
Spheroid Body Myopathy	MYOT	DOMINANT	NM_006790.2	99.03%

Spinal Muscular Atrophy	IGHMBP2	X_LINKED	NM_002180.2	99.18%
Spinal Muscular Atrophy, Dominant	VAPB	X_LINKED	NM_004738.4	99.01%
Spinocerebellar Ataxia, Dominant	AFG3L2	DOMINANT	NM_006796.2	98.39%
Spinocerebellar Ataxia, Dominant	DYNC1H1	DOMINANT	NM_001376.4	99.09%
Spinocerebellar Ataxia, Dominant	FGF14	DOMINANT	NM_004115.3	99.45%
Spinocerebellar Ataxia, Dominant	ITPR1	DOMINANT	NM_002222.5	99.40%
Spinocerebellar Ataxia, Dominant	PDYN	DOMINANT	NM_024411.4	99.95%
Spinocerebellar Ataxia, Dominant	PRKCG	DOMINANT	NM_002739.3	97.49%
Spinocerebellar Ataxia, Dominant	SPTBN2	DOMINANT	NM_006946.2	97.26%
Spinocerebellar Ataxia, Dominant	TGM6	DOMINANT	NM_198994.2	98.26%
Spinocerebellar Ataxia, Recessive	ADCK3	RECESSIVE	NM_020247.4	97.82%
Spinocerebellar Ataxia, Recessive	ANO10	RECESSIVE	NM_018075.3	97.66%
Spinocerebellar Ataxia, Recessive	C10orf2	DOMINANT	NM_021830.4	98.46%
Spinocerebellar Ataxia, Recessive	TDP1	RECESSIVE	NM_018319.3	99.48%
Split-Hand/Foot Malformation	FBXW4	DOMINANT	NM_022039.3	98.61%
Spondylocostal Dysostosis	DLL3	RECESSIVE	NM_016941.3	97.41%
Spondyloepimetaphyseal Dysplasia	DDR2	DOMINANT	NM_006182.2	98.99%
Spondyloepimetaphyseal Dysplasia	MMP13	DOMINANT	NM_002427.3	99.91%
Spondyloepiphyseal Dysplasia	CHST3	RECESSIVE	NM_004273.4	98.22%
Spondylometaphyseal Dysplasia	TRPV4	DOMINANT	NM_021625.4	97.17%
Spontaneous Pneumothorax	FLCN	DOMINANT	NM_144997.5	96.89%
Stargardt Disease, Dominant	ELOVL4	DOMINANT	NM_022726.3	99.30%
Stargardt Disease, Dominant	PROM1	DOMINANT	NM_006017.2	99.87%
Stargardt Disease, Recessive	ABCA4	RECESSIVE	NM_000350.2	99.79%
Stargardt Disease, Recessive	CNGB3	RECESSIVE	NM_019098.4	99.76%
Steroid 5-Alpha-Reductase Deficiency	SRD5A2	RECESSIVE	NM_000348.3	99.71%
Steroid-Resistant Nephrotic Syndrome	NPHS2	RECESSIVE	NM_014625.2	96.76%
Stickler Syndrome, Dominant	COL11A1	DOMINANT	NM_001854.3	98.66%
Stickler Syndrome, Dominant	COL11A2	DOMINANT	NM_080680.2	99.38%

Stickler Syndrome, Dominant	COL2A1	DOMINANT	NM_001844.4	98.74%
Stickler Syndrome, Recessive	COL9A1	RECESSIVE	NM_001851.4	99.62%
Stickler Syndrome, Recessive	COL9A2	RECESSIVE	NM_001852.3	98.66%
Stiff Skin Syndrome	FBN1	DOMINANT	NM_000138.4	99.59%
Striatal Degeneration	PDE8B	DOMINANT	NM_003719.3	98.38%
Stuve-Wiedemann Syndrome	LIFR	RECESSIVE	NM_002310.5	99.64%
Succinic Semialdehyde Dehydrogenase Deficiency	ALDH5A1	RECESSIVE	NM_001080.3	98.94%
Succinyl-CoA:3-Oxoacid CoA Transferase Deficiency	OXCT1	RECESSIVE	NM_000436.3	99.61%
Sulfate Transporter-Related Osteochondrodysplasia	SLC26A2	RECESSIVE	NM_000112.3	99.63%
Sulfocysteinuria	SUOX	RECESSIVE	NM_000456.2	98.33%
Supravalvular Aortic Stenosis	ELN	DOMINANT	NM_000501.2	97.00%
Syndactyly	DLL3	DOMINANT	NM_016941.3	97.41%
Syndactyly	GJA1	DOMINANT	NM_000165.3	95.58%
Syndromic Microphthalmia, Dominant	BMP4	DOMINANT	NM_001202.3	98.31%
Syndromic Microphthalmia, Dominant	OTX2	DOMINANT	NM_172337.2	99.73%
Syndromic Microphthalmia, Recessive	STRA6	RECESSIVE	NM_022369.3	97.91%
Systemic Primary Carnitine Deficiency	SLC22A5	RECESSIVE	NM_003060.3	97.34%
Tangier Disease	ABCA1	RECESSIVE	NM_005502.3	99.11%
Tetra-Amelia Syndrome	WNT3	RECESSIVE	NM_030753.3	99.13%
Thiamine Metabolism Dysfunction Syndrome	SLC19A3	RECESSIVE	NM_025243.3	99.91%
Thiamine-Responsive Megaloblastic Anemia	SLC19A2	RECESSIVE	NM_006996.2	97.59%
Thiamine-Responsive Megaloblastic Anemia Syndrome	SLC19A2	RECESSIVE	NM_006996.2	97.59%
Thiopurine S-Methyltransferase Deficiency	TPMT	RECESSIVE	NM_000367.2	97.83%
Thoracic Aortic Aneurysms and Aortic Dissections	ACTA2	DOMINANT	NM_001613.2	99.98%
Thoracic Aortic Aneurysms and Aortic Dissections	FBN1	DOMINANT	NM_000138.4	99.59%
Thoracic Aortic Aneurysms and Aortic Dissections	MYH11	DOMINANT	NM_002474.2	99.77%
Thoracic Aortic Aneurysms and Aortic Dissections	MYLK	DOMINANT	NM_053025.3	98.34%
Thoracic Aortic Aneurysms and Aortic Dissections	SMAD3	DOMINANT	NM_005902.3	97.72%
Thoracic Aortic Aneurysms and Aortic Dissections	TGFBR1	DOMINANT	NM_004612.2	97.77%

Thoracic Aortic Aneurysms and Aortic Dissections	TGFBR2	DOMINANT	NM_003242.5	99.28%
Thrombasthenia of Glanzmann and Naegeli	ITGA2B	RECESSIVE	NM_000419.3	99.08%
Thrombasthenia of Glanzmann and Naegeli	ITGB3	RECESSIVE	NM_000212.2	99.34%
Thrombocytopenia	ANKRD26	DOMINANT	NM_014915.2	99.86%
Thrombocytopenia	CYCS	DOMINANT	NM_018947.5	95.94%
Thrombocytopenia	MASTL	DOMINANT	NM_032844.3	98.86%
Thyroid Dyshormonogenesis	TG	RECESSIVE	NM_003235.4	99.31%
Thyroid Hormone Resistance	THRB	DOMINANT	NM_000461.4	99.22%
Thyroid Hormonogenesis Defect	SLC5A5	RECESSIVE	NM_000453.2	97.72%
Tietz Syndrome	MITF	DOMINANT	NM_000248.3	99.88%
Timothy Syndrome	CACNA1C	DOMINANT	NM_000719.6	98.44%
Tourette Syndrome	SLITRK1	DOMINANT	NM_052910.1	97.73%
Townes-Brocks Syndrome	SALL1	DOMINANT	NM_002968.2	99.19%
TP63-Related Spectrum Disorders	TP63	DOMINANT	NM_003722.4	99.84%
Transaldolase Deficiency	TALDO1	RECESSIVE	NM_006755.1	95.39%
Transcobalamin II Deficiency	TCN2	RECESSIVE	NM_000355.3	99.54%
Transient Familial Neonatal Hyperbilirubinemia	UGT1A1	DOMINANT	NM_000463.2	99.91%
Transient Neonatal Diabetes, Dominant	KCNJ11	DOMINANT	NM_000525.3	91.82%
Transient Neonatal Diabetes, Dominant/Recessive	ABCC8	DOMINANT	NM_000352.3	98.00%
Transient Neonatal Diabetes, Dominant/Recessive	INS	DOMINANT	NM_000207.2	99.12%
Transient Neonatal Diabetes, Recessive	GCK	RECESSIVE	NM_000162.3	96.32%
Treacher Collins Syndrome, Dominant	POLR1D	DOMINANT	NM_015972.3	96.74%
Treacher Collins Syndrome, Dominant	TCOF1	DOMINANT	NM_001135243.1	98.70%
Treacher Collins Syndrome, Recessive	POLR1C	RECESSIVE	NM_203290.2	99.92%
Trichohepatoenteric Syndrome	SKIV2L	RECESSIVE	NM_006929.4	98.96%
Trichorhinophalangeal Syndrome	TRPS1	DOMINANT	NM_014112.2	97.47%
Trifunctional Protein Deficiency	HADHA	RECESSIVE	NM_000182.4	99.95%
Trifunctional Protein Deficiency	HADHB	RECESSIVE	NM_000183.2	99.35%
Trimethylaminuria	FMO3	RECESSIVE	NM_006894.5	99.96%

Triosephosphate Isomerase Deficiency	TPI1	DOMINANT	NM_000365.5	99.62%
Triphalangeal Thumb-Polysyndactyly Syndrome	LMBR1	DOMINANT	NM_022458.3	96.13%
Trismus-Pseudocamptodactyly Syndrome	MYH8	DOMINANT	NM_002472.2	99.74%
Tryptophan Hydroxylase Deficiency	TPH2	DOMINANT	NM_173353.3	99.88%
Tuberous Sclerosis	TSC1	DOMINANT	NM_000368.4	99.02%
Tuberous Sclerosis	TSC2	DOMINANT	NM_000548.3	97.93%
Tumor Predisposition Syndrome	BAP1	DOMINANT	NM_004656.2	97.07%
Tylosis with Esophageal Cancer	RHBDF2	DOMINANT	NM_024599.5	98.08%
Type II Collagenopathies	COL2A1	DOMINANT	NM_001844.4	98.74%
Tyrosine Hydroxylase Deficiency	TH	RECESSIVE	NM_000360.3	99.55%
Tyrosinemia	FAH	RECESSIVE	NM_000137.2	96.54%
Tyrosinemia	HPD	RECESSIVE	NM_002150.2	98.58%
Tyrosinemia	TAT	RECESSIVE	NM_000353.2	99.97%
Udd Distal Myopathy	TTN	DOMINANT	NM_133378.4	99.55%
Ulnar-Mammary Syndrome	TBX3	DOMINANT	NM_005996.3	98.86%
Unverricht-Lundborg Disease	CSTB	RECESSIVE	NM_000100.3	99.82%
Usher Syndrome	CDH23	RECESSIVE	NM_022124.5	97.08%
Usher Syndrome	CLRN1	RECESSIVE	NM_174878.2	99.54%
Usher Syndrome	DFNB31	RECESSIVE	NM_015404.3	96.78%
Usher Syndrome	GPR98	RECESSIVE	NM_032119.3	99.84%
Usher Syndrome	HARS	RECESSIVE	NM_002109.4	99.08%
Usher Syndrome	MYO7A	RECESSIVE	NM_000260.3	98.70%
Usher Syndrome	PCDH15	RECESSIVE	NM_033056.3	99.93%
Usher Syndrome	USH1C	RECESSIVE	NM_005709.3	97.95%
Usher Syndrome	USH2A	RECESSIVE	NM_007123.5	99.98%
VACTERL Association with Hydrocephalus	FANCB	X_LINKED	NM_001018113.1	94.19%
van der Woude Syndrome	IRF6	DOMINANT	NM_006147.3	99.06%
Variegate Porphyria	PPOX	DOMINANT	NM_000309.3	99.64%
Vesicoureteral Reflux	ROBO2	DOMINANT	NM_002942.4	99.78%

Vitamin D-Dependent Rickets	CYP27B1	RECESSIVE	NM_000785.3	98.41%
Vitamin D-Dependent Rickets	VDR	RECESSIVE	NM_001017535.1	98.22%
Vitamin K-Dependent Clotting Factors	GGCX	RECESSIVE	NM_000821.5	99.60%
Vitamin K-Dependent Clotting Factors	VKORC1	RECESSIVE	NM_024006.4	98.71%
Vitelliform Macular Dystrophy	PRPH2	DOMINANT	NM_000322.4	99.36%
Vitreoretinochoroidopathy	BEST1	DOMINANT	NM_004183.3	99.21%
Vitreoretinopathy	VCAN	DOMINANT	NM_004385.4	99.60%
VLCAD Deficiency	ACADVL	RECESSIVE	NM_000018.2	99.13%
Vohwinkel Syndrome	GJB2	DOMINANT	NM_004004.5	97.20%
Von Hippel-Lindau Syndrome	VHL	DOMINANT	NM_000551.3	94.77%
von Willebrand Disease	VWF	DOMINANT	NM_000552.3	95.17%
VSX2-related Microphthalmia	VSX2	RECESSIVE	NM_182894.2	99.45%
Waardenburg Syndrome	EDN3	DOMINANT	NM_207034.1	98.17%
Waardenburg Syndrome	EDNRB	DOMINANT	NM_000115.3	99.44%
Waardenburg Syndrome	MITF	DOMINANT	NM_000248.3	99.88%
Waardenburg Syndrome	PAX3	DOMINANT	NM_181457.3	97.63%
Waardenburg Syndrome	SNAI2	DOMINANT	NM_003068.4	97.30%
Waardenburg Syndrome	SOX10	DOMINANT	NM_006941.3	96.64%
Wagner Syndrome	VCAN	DOMINANT	NM_004385.4	99.60%
WAGR Syndrome	PAX6	DOMINANT	NM_000280.4	99.26%
WAGR Syndrome	WT1	DOMINANT	NM_024426.4	95.44%
Walker-Warburg Syndrome	LARGE	RECESSIVE	NM_004737.4	98.32%
Warburg Micro Syndrome	RAB18	RECESSIVE	NM_021252.4	99.27%
Warburg Micro Syndrome	RAB3GAP1	RECESSIVE	NM_012233.2	99.72%
Warburg Micro Syndrome	RAB3GAP2	RECESSIVE	NM_012414.3	99.31%
Watson Syndrome	NF1	DOMINANT	NM_000267.3	98.11%
Weaver Syndrome	EZH2	DOMINANT	NM_004456.4	97.77%
Weaver Syndrome	NSD1	DOMINANT	NM_022455.4	98.84%
Weill-Marchesani Syndrome	ADAMTS10	DOMINANT	NM_030957.2	97.80%

Weill-Marchesani Syndrome	FBN1	DOMINANT	NM_000138.4	99.59%
Weill-Marchesani Syndrome	LTBP2	DOMINANT	NM_000428.2	99.04%
Weill-Marchesani-Like Syndrome	ADAMTS17	RECESSIVE	NM_139057.2	95.08%
Weissenbacher-Zweymuller Syndrome	COL11A2	DOMINANT	NM_080680.2	99.38%
Werner Syndrome	WRN	RECESSIVE	NM_000553.4	96.11%
Weyers Acrofacial Dysostosis	EVC	DOMINANT	NM_153717.2	95.39%
WFS1-Related Spectrum Disorders	WFS1	DOMINANT	NM_006005.3	86.69%
White Sponge Nevus of Cannon	KRT13	DOMINANT	NM_002274.3	98.59%
White Sponge Nevus of Cannon	KRT4	DOMINANT	NM_002272.3	98.56%
Wilms Tumor	WT1	DOMINANT	NM_024426.4	95.44%
Wilson Disease	ATP7B	RECESSIVE	NM_000053.2	98.80%
Winchester Syndrome	MMP2	RECESSIVE	NM_004530.4	97.78%
Wolff-Parkinson-White Syndrome	PRKAG2	DOMINANT	NM_016203.3	97.03%
Wolf-Hirschhorn Syndrome	WHSC1	DOMINANT	NM_133330.2	99.58%
Wolman Disease	LIPA	RECESSIVE	NM_000235.2	99.79%
Woodhouse-Sakati Syndrome	DCAF17	RECESSIVE	NM_025000.3	99.77%
Xanthinuria	XDH	DOMINANT	NM_000379.3	99.95%
Xeroderma Pigmentosum	DDB2	RECESSIVE	NM_000107.2	99.69%
Xeroderma Pigmentosum	ERCC1	RECESSIVE	NM_202001.2	99.22%
Xeroderma Pigmentosum	ERCC2	RECESSIVE	NM_000400.3	97.35%
Xeroderma Pigmentosum	ERCC3	RECESSIVE	NM_000122.1	99.91%
Xeroderma Pigmentosum	ERCC4	RECESSIVE	NM_005236.2	99.89%
Xeroderma Pigmentosum	ERCC5	RECESSIVE	NM_000123.3	99.93%
Xeroderma Pigmentosum	POLH	RECESSIVE	NM_006502.2	96.06%
Xeroderma Pigmentosum	XPA	RECESSIVE	NM_000380.3	99.22%
Xeroderma Pigmentosum	XPC	RECESSIVE	NM_004628.4	98.81%
X-Linked Hypophosphatemia	PHEX	X_LINKED	NM_000444.4	96.84%
Zaspopathy	LDB3	DOMINANT	NM_001080116.1	99.33%
Zellweger Syndrome	PEX1	RECESSIVE	NM_000466.2	99.23%

Zellweger Syndrome	PEX10	RECESSIVE	NM_153818.1	95.56%
Zellweger Syndrome	PEX12	RECESSIVE	NM_000286.2	99.86%
Zellweger Syndrome	PEX13	RECESSIVE	NM_002618.3	99.37%
Zellweger Syndrome	PEX14	RECESSIVE	NM_004565.2	96.23%
Zellweger Syndrome	PEX16	RECESSIVE	NM_004813.2	98.64%
Zellweger Syndrome	PEX19	RECESSIVE	NM_002857.3	99.90%
Zellweger Syndrome	PEX2	RECESSIVE	NM_000318.2	99.44%
Zellweger Syndrome	PEX26	RECESSIVE	NM_017929.5	99.34%
Zellweger Syndrome	PEX3	RECESSIVE	NM_003630.2	97.54%
Zellweger Syndrome	PEX5	RECESSIVE	NM_001131025.1	97.44%
Zellweger Syndrome	PEX6	RECESSIVE	NM_000287.3	98.32%
Zonular Pulverulent Cataract	GJA3	DOMINANT	NM_021954.3	98.25%
Zonular Pulverulent Cataract	GJA8	DOMINANT	NM_005267.4	99.35%