PreDNA Test

ΚΑΤΑΛΟΓΟΣ ΓΟΝΙΔΙΩΝ ΚΑΙ ΑΝΤΙΣΤΟΙΧΩΝ ΓΕΝΕΤΙΚΩΝ ΝΟΣΩΝ

ΓΟΝΙΔΙΟ	ΚΩΔΙΚΟΣ NLM	ΝΟΣΟΣ	ΣΥΧΝΟΤΗΤΑ ΕΜΦΑΝΙΣΗΣ ΝΟΣΟΥ
ABCA4	NM_000350.2	**Cone-rod dystrophy type 3, Retinitis pigmentosa type 19, Stargardt disease type 1**	1:3.500 - 1:30.000
ACAD9	NM_014049.4	**Acyl-CoA dehydrogenase type 9 deficiency**	1-9:1.000.000
ACADM	NM_000016.5	**Acyl-CoA dehydrogenase deficiency, medium-chain**	1:17.000
ACADS	NM_000017.2	**Acyl-CoA dehydrogenase deficiency, short-chain**	1:35.000 - 1:50.000
ACADSB	NM_001609.3	**2-Methylbutyryl-CoA dehydrogenase deficiency**	1:250 - 1:500
ACADVL	NM_000018.3	**Very long chain acyl-CoA dehydrogenase deficiency**	1:40.000 - 1:120.000
ACAT1	NM_000019	**beta-ketothiolase deficiency**	1:1.000.000
ACOX1	NM_001185039	**peroxisomal acyl-CoA oxidase deficiency**	< 1: 1.000.000
ACTN4	NM_004924.4	**Glomerulosclerosis, focal segmental, type 1**	2-4:100.000
ADA	NM_000022.2	**Adenosine deaminase deficiency**	1:200.000 - 1:1.000.000
ADAMTS2	NM_014244.4	**Ehlers-Danlos syndrome type 7C**	1:5.000
AFG3L2	NM_006796	**Progressive external ophthalmoplegia, Spastic ataxia, Spinocerebellar ataxia**	Unknown
AGA	NM_000027.3	**Aspartylglucosaminuria**	1:85.000
AGL	NM_000642.2	**Glycogen storage disease type 3**	1:100.000
AGPS	NM_003659.3	**Rhizomelic chondrodysplasia punctata type 3**	1:100.000
AGRN	NM_198576	**Congenital myasthenic syndrome**	Unknown
AGXT	NM_000030.2	**Primary hyperoxaluria type 1**	1:58.000
AHI1	NM_017651.4	**Joubert syndrome type 3**	1:80.000 - 1:100.000

AIPL1	NM_014336.4	**Cone-rod dystrophy, Leber congenital amaurosis type 4**	1:30.000 - 1:40.000
AKR1D1	NM_005989	**Congenital bile acid synthesis defect**	1-9:1.000.000
ALAS2	NM_000032.4	**Erythropoietic protoporphyria, Sideroblastic anemia, X-linked**	1:500 - 1:50.000
ALDOA	NM_000034.3	**Glycogen storage disease type 12**	1:100.000
ALDOB	NM_000035.3	**Hereditary fructose intolerance**	1:20.000 - 1:30.000
ALPL	NM_000478.4	**Hypophosphatasia**	1:2.500 - 1:100.000
AMACR	NM_014324	**Alpha-methylacyl-CoA racemase (AMACR) deficiency**	Unknown
AMT	NM_000481.3	**Glycine encephalopathy**	1:55.000 - 1:63.000
ANK1	NM_020475	**Hereditary spherocytosis**	1:2.000
ANO5	NM_213599.2	**Limb-girdle muscular dystrophy type 2L, autosomal recessive**	1:14.500 - 1:123.000
APOB	NM_000384	**Familial hypobetalipoproteinemia, Hypercholesterolemia**	1:500 – 1:1.000
APOC2	NM_000483	**Apolipoprotein C2 deficiency, Hyperlipoproteinemia**	1:1.000
APOE	NM_000041	**Age-related macular degeneration, Alzheimer disease**	1:2.000
APTX	NM_175073.2	**Ataxia with oculomotor apraxia type 1**	>1:100.000
AR	NM_000044.3	**Androgen insensitivity syndrome**	2-5:100.000
ARG1	NM_000045	**Arginase deficiency**	1:300.000
ARL13B	NM_182896.2	**Joubert syndrome type 8**	<1:80.000 - 1:100.000
ARL6	NM_177976.2	**Retinitis pigmentosa type 55, Bardet-Biedl syndrome type 3**	1:3.500 - 1:160.000
ARSA	NM_000487.5	**Metachromatic leukodystrophy**	1:75 - 1:160.000
ARSB	NM_000046.3	**Mucopolysaccharidosis type 6**	1:100.000 - 1:250.000
ARX	NM_139058.2	**Epileptic encephalopathy, early infantile, type 1, Lissencephaly with abnormal genitalia, X-linked**	1 - 9:1.000.000
ASL	NM_000048.3	**Argininosuccinic aciduria**	1:70.000

ASPA	NM_000049.2	**Canavan disease**	1:6.400 - 1:13.500
ASPM	NM_018136.4	**Microcephaly primary, type 5, autosomal recessive**	1:30.000 - 1:250.000
ASS1	NM_000050.4	**Citrullinemia type 1**	1:57.000
ATP2A1	NM_004320	**Brody myopathy**	Unknown
ATP7A	NM_000052.6	**Menkes disease, Occipital horn syndrome, Spinal muscular atrophy, distal, X-linked**	1:50.000 - 100.000
ATP7B	NM_000053.3	**Wilson disease**	1:40.000
ATXN1	NM_000332	**Spinocerebellar ataxia type 1**	1:50.000 – 1:100.000
ATXN2	NM_002973	**Amyotrophic lateral sclerosis, Spinocerebellar ataxia type 2**	1:20.000 – 1:50.000
ATNX3	NM_004993	**Spinocerebellar ataxia type 3**	Unknown
ATNX7	NM_000333	**Spinocerebellar ataxia 7**	Unknown
B9D1	NM_015681	**Meckel syndrome, Joubert syndrome**	1:3.000 - 1:13.250
B9D2	NM_030578.3	**Meckel syndrome type 10**	1:3.000 - 1:140.000
BBS1	NM_024649	**Bardet-Biedl syndrome**	1:13.500 – 1:160.000
BBS10	NM_024685	**Bardet-Biedl syndrome**	1:13.500 – 1:160.000
BBS12	NM_152618	**Bardet-Biedl syndrome**	1:13.500 – 1:160.000
BBS2	NM_031885	**Bardet-Biedl syndrome, Retinitis pigmentosa**	1:13.500 – 1:160.000
BBS4	NM_033028	**Bardet-Biedl syndrome**	1:13.500 – 1:160.000
BBS5	NM_152384	**Bardet-Biedl syndrome**	1:13.500 – 1:160.000
BBS7	NM_018190	**Bardet-Biedl syndrome**	1:13.500 – 1:160.000
BBS9	NM_001033604	**Bardet-Biedl syndrome**	1:13.500 – 1:160.000
BCKDHA	NM_000709.3	**Maple syrup urine disease type 1A**	1 - 9:1.000.000
BCKDHB	NM_183050.2	**Maple syrup urine disease type 1B**	1 - 9:1.000.000

BCS1L	NM_004328.4	**Björnstad syndrome, GRACILE syndrome, Mitochondrial complex III deficiency, nuclear type 1**	1:47.000 (FINLAND)
BEST1	NM_004183.3	**Retinitis pigmentosa type 50, Bestrophinopathy, Vitelliform macular dystrophy type 2**	1:3.500 - UNKNOWN
BICC1	NM_025044	**Renal dysplasia**	Unknown
BRCA1	NM_007294.3	**Breast Cancer gene 1**	1:33
BRCA2	NM_000059.3	**Breast Cancer gene 2, Fanconi anemia, complementation group D1**	1:33
BRIP1	NM_032043.2	**Fanconi anemia, complementation group J**	1 - 9:1.000.000
BSND	NM_057176.2	**Bartter syndrome type 4A**	< 1:1.000.000
BTD	NM_000060.3	**Biotinidase deficiency**	1:60.000
BTK	NM_000061.2	**Agammaglobulinemia, X-linked**	1:200.000
C3	NM_000064.2	**C3 deficiency, Atypical hemolytic-uremic syndrome with C3 anomaly**	1:10.000 - 1:1.000.000
C50RF42	NM_023073	**Joubert syndrome**	1:80.000 – 1:100.000
CA2	NM_000067.2	**Osteopetrosis, autosomal recessive, type 3**	1:250.000
CACNA1A	NM_000068	**19p13.13 deletion syndrome, Episodic ataxia, Familial hemiplegic migraine, Spinocerebellar ataxia type 6**	1:10.000 – 1:100.000
CAPN3	NM_000070.2	**Limb-girdle muscular dystrophy type 2A**	1:150 - 1:178.000
CAV3	NM_033337	**distal myopathy, hyperCKemia, Limb-girdle muscular dystrophy, Rippling muscle disease**	1:14.500 – 1:123.000
CBS	NM_000071.2	**Homocystinuria**	1:6.400 - 1:344.000
CC2D2A	NM_001080522.2	**Joubert syndrome type 9, Meckel syndrome type 6**	1:3.000 - 1:100.000
CD2AP	NM_012120.2	**Focal segmental glomerulosclerosis type 3**	5-24:1.000.000
CD40LG	NM_000074.2	**Hyper IgM syndrome, X-linked**	2:1.000.000
CDH23	NM_022124.5	**Usher syndrome type 1D, Deafness type 12, autosomal recessive**	1/500
CDH3	NM_001793.4	**Ectodermal dysplasia - ectrodactyly - macular dystrophy**	7:10.000
CDHR1	NM_033100.3	**Retinitis pigmentosa type 65**	1:3.500

CDK5RAP2	NM_018249.5	**Microcephaly, primary, type 3, autosomal recessive**	1:10.000 - 1:150.000
CDKL5	NM_003159	**X-linked infantile spasm syndrome**	>1:100.000
CDKN1C	NM_000076	**Beckwith-Wiedemann syndrome**	1:13.700
CENPJ	NM_018451.4	**Microcephaly primary, type 6, autosomal recessive**	1:10.000 - 1:150.000
CEP152	NM_014985.3	**Microcephaly, primary, type 9, autosomal recessive, Seckel syndrome type 5**	1:10.000 - 1:150.000
CEP164	NM_014956	**Nephronophthisis**	1:50.000
CEP290	NM_025114.3	**Joubert syndrome, Senior-Loken type, Leber congenital amaurosis type 1, Meckel syndrome type 4**	<1:80.000 -1:100.000
CEP41	NM_018718	**Joubert syndrome, autism**	<1:80.000 -1:100.000
CERKL	NM_201548.4	**Retinitis pigmentosa type 26**	1:4.000
CFH	NM_000186.3	**Complement factor H deficiency**	1:10.000
CFTR	NM_000492.3	**Cystic fibrosis**	1:2.500 - 1:3.500
CHAT	NM_020549	**Congenital myasthenic syndrome**	Unknown
CHKB	NM_005198	**Narcolepsy, Muscular dystrophy**	1:600 – 1:2.000
CHRNA1	NM_000079	**Congenital myasthenic syndrome, Multiple pterygium syndrome**	Unknown
CHRNB1	NM_000747	**Congenital myasthenic syndrome**	Unknown
CHRND	NM_000751	**Congenital myasthenic syndrome, Multiple pterygium syndrome**	Unknown
CHRNE	NM_000080	**Congenital myasthenic syndrome**	Unknown
CHRNG	NM_005199	**Escobar syndrome, Multiple pterygium syndrome**	Unknown
CHST6	NM_021615.4	**Macular corneal dystrophy**	1-9:100.000
CLCN1	NM_000083.2	**Myotonia congenital, autosomal recessive**	1:8.000
CLCN7	NM_001287.5	**Osteopetrosis type 4, autosomal recessive**	1:250.000
CLDN14	NM_144492.2	**Deafness type 29, autosomal recessive**	1:500 - 1:2.500

CLDN19	NM_148960.2	**Hypomagnesemia type 5, renal failure with severe ocular abnormalities**	1:40.000
CLN3	NM_001042432.1	**Ceroid-lipofuscinoses neuronal type 3**	1:12.500
CLN5	NM_006493.2	**Neuronal ceroid lipofuscinosis type 5**	1:12.500
CLN6	NM_017882.2	**Ceroid lipofuscinosis, neuronal, type 6**	1:12.500
CLN8	NM_018941.3	**Ceroid lipofuscinosis, neuronal, type 8**	1:12.500
CLRN1	NM_174878.2	**Retinitis pigmentosa type 61, Usher syndrome type 3A**	1:4.000 - 1:15.000
CNBP	NM_003418	**Myotonic dystrophy**	1:8.000
CNGA1	NM_000087.3	**Retinitis pigmentosa type 49**	1:4.000
CNGB1	NM_001297.4	**Retinitis pigmentosa type 45**	1:4.000
CNGB3	NM_019098.4	**Achromatopsia type 3, Macular degeneration, juvenile**	1:5.000 - 1:67.000
COL11A1	NM_001854.3	**Stickler syndrome type 2**	1:7.000 - 1:9.000
COL17A1	NM_000494.3	**Epidermolysis bullosa, junctional, non-Herlitz type**	1:30.000 - 1:50.000
COL1A2	NM_000089.3	**Ehlers-Danlos syndrome, cardiac valvular type**	1:5.000 - 1:250.000
COL4A3	NM_000091.4	**Alport syndrome, autosomal recessive**	1:50.000
COL4A4	NM_000092.4	**Alport syndrome, autosomal recessive**	1:50.000
COL6A1	NM_001848	**Bethlem myopathy, Ullrich congenital muscular dystrophy**	1:100.000
COL6A2	NM_001849	**Bethlem myopathy, Ullrich congenital muscular dystrophy**	1:100.000
COL6A3	NM_004369	**Bethlem myopathy, Ullrich congenital muscular dystrophy**	1:100.000
COL7A1	NM_000094.3	**Epidermolysis bullosa dystrophica, Hallopeau-Siemens type**	1:30.000 - 1:50.000
COL9A1	NM_001851.4	**Stickler syndrome type 4**	1:7.000 - 1:9.000
COL9A2	NM_001852.3	**Stickler syndrome type 5**	1:7.000 - 1:9.000
COLQ	NM_005677	**Congenital myasthenic syndrome**	Unknown

COQ2	NM_015697	**Multiple system atrophy**	2-5:100.000
CPS1	NM_001122633	**Carbamoyl phosphate synthetase I deficiency**	1:800.000
CPT1A	NM_001876	**Carnitine palmitoyltransferase I (CPT I) deficiency**	Rare
CPT2	NM_000098	**Carnitine palmitoyltransferase II (CPT II) deficiency**	Rare
CRB1	NM_201253.2	**Leber congenital amaurosis type 8, Retinitis pigmentosa type 12, Pigmented paravenous chorioretinal atrophy**	1:4.000 - 1:50.000
CRTAP	NM_006371.4	**Osteogenesis imperfecta type 7**	<1:15.000
CRX	NM_000554.4	**Leber congenital amaurosis type 7**	1:50.000
CSTB	NM_000100.3	**Progressive myoclonic epilepsy type 1A**	UNKNOWN
CTNS	NM_004937.2	**Cystinosis, ocular nonnephropathic, Nephropathic cystinosis**	1-9:100.000
CTSD	NM_001909.4	**Ceroid lipofuscinosis, neuronal, type 10**	1:12.500 - 1:200.000
CTSK	NM_000396.3	**Pycnodysostosis**	<1:100.000
CYP21A2	NM_000500.7	**Congenital adrenal hyperplasia due to 21-hydroxylase deficiency**	1:10.000
CYP27A1	NM_000784	**Cerebrotendinous xanthomatosis**	1:1.000.000
CYP4V2	NM_207352.3	**Bietti crystalline corneoretinal dystrophy**	1:67.000
CYP7A1	NM_000780	**hypercholesterolemia**	1:500
CYP7B1	NM_004820.3	**Congenital bile acid synthesis defect type 3, Spastic paraplegia type 5A, autosomal recessive**	1-9:100.000
DBT	NM_001918.3	**Maple syrup urine disease type 2**	1:380 - 1:185.000
DECR1	NM_001359	**2,4-dienoyl-CoA reductase deficiency**	Rare
DFNB31	NM_015404.3	**Deafness type 31, autosomal recessive**	<1:333
DFNB59	NM_001042702.3	**Deafness type 59, autosomal recessive**	<1:333
DHCR24	NM_014762	**Desmosterolosis**	Unknown

DHCR7	NM_001360.2	**Smith-Lemli-Opitz syndrome**	1:20.000 - 1:60.000
DHDDS	NM_024887.3	**Retinitis pigmentosa type 59**	1:4.000
DLD	NM_000108.4	**Dihydrolipoamide dehydrogenase deficiency E3**	1:35.000 - 1:48.000
DMD	NM_004006.2	**Becker/Duchenne muscular dystrophy, Dilated cardiomyopathy type 3B**	1:2.500 - 1:3.300
DMP1	NM_004407.3	**Hypophosphatemic rickets type 1, autosomal recessive**	1:20.000
DMPK	NM_004409	**Myotonic dystrophy**	1:8.000
DNAJB6	NM_005494	**Limb-girdle muscular dystrophy**	1:14.500 – 1:123.000
DNM2	NM_004945	**Centronuclear myopathy, Charcot-Marie-Tooth disease**	1:2.500
DOK7	NM_173660	**Congenital myasthenic syndrome**	Unknown
DPAGT1	NM_001382	**Congenital disorder of glycosylation, Myasthenic syndrome**	Unknown
DPM2	NM_003863	**Congenital disorder of glycosylation**	Unknown
DPM3	NM_153741	**Congenital disorder of glycosylation**	Unknown
DSP	NM_004415.2	**Cardiomyopathy, arrhythmogenic, dilated with woolly hair and keratoderma, Lethal acantholytic epidermolysis bullosa**	1:1.000 - 1:30.000
DYSF	NM_003494.3	**Dysferlinopathy, Miyoshi myopathy, Muscular dystrophy, limb girdle type 2B**	1:1.300 - 1:14.500
EBP	NM_006579	**X-linked chondrodysplasia punctata**	1:400.000
EDA	NM_001399.4	**Hypohidrotic ectodermal dysplasia, X-linked**	1:15.000
EDN3	NM_207034.1	**Shah-Waardenburg syndrome type 4B**	1:40.000
EDNRB	NM_000115.3	**Shah-Waardenburg syndrome type 4A**	1:40.000
EGR2	NM_000399.3	**Charcot-Marie-Tooth disease type 4E**	1:2.500
EMD	NM_000117.2	**Emery-Dreifuss muscular dystrophy type 1, X-linked**	1:100.000
ENO3	NM_053013.3	**Glycogen storage disease type 13**	1:100.000

ENPP1	NM_006208.2	**Generalized arterial calcification of infancy and pseudoxanthoma elasticum, Hypophosphatemic rickets type 2, autosomal recessive, Idiopathic infantile arterial calcification**	1:20.000 - 391.000
ERCC4	NM_005236	**Xeroderma pigmentosum, Fanconi anemia, Tracheoesophageal fistula, XFE progeroid syndrome**	1:1.000.000
ERCC6	NM_000124.3	**Cerebrooculofacioskeletal syndrome type 1, Cockayne syndrome type B**	1:200.000 - 1:1.000.000
ERCC8	NM_000082.3	**Cockayne syndrome type A**	1:200.000
ESCO2	NM_001017420.2	**Roberts syndrome, SC Phocomelia syndrome**	<1:1.000.000
ESPN	NM_031475.2	**Deafness type 36, autosomal recessive**	<1:333
ESRRB	NM_004452.3	**Deafness type 35, autosomal recessive**	<1:333
ETFA	NM_ 001014763	**Glutaric acidemia**	Rare
ETFB	NM_ 001014763	**Glutaric acidemia**	Rare
ETFDH	NM_001281737	**Glutaric acidemia**	Rare
EYS	NM_001142800.1	**Retinitis pigmentosa type 25**	1:4.000
F11	NM_000128.3	**Factor 11 deficiency**	1:7 - 1:1.000.000
F5	NM_000130.4	**Factor 5 deficiency, Thrombosis**	1:1.000 - 1:100.000
F8	NM_000132.3	**Hemophilia A**	1:4.000
F9	NM_000133.3	**Hemophilia B**	1:20.000
FAH	NM_000137.2	**Tyrosinemia type 1**	1:60.000 - 1:100.000
FAM126A	NM_032581.3	**Hypomyelination and congenital cataract**	<1:1.000.000
FAM20C	NM_020223.3	**Osteosclerotic bone dysplasia**	<1:1.000.000
FANCA	NM_000135.2	**Fanconi anemia, complementation group A**	1:160.000
FANCB	NM_152633	**Fanconi anemia, complementation group B**	1:160.000
FANCC	NM_000136.2	**Fanconi anemia, complementation group C**	1:160.000

FANCD2	NM_033084.3	**Fanconi anemia, complementation group D2**	1:160.000
FANCE	NM_021922.2	**Fanconi anemia, complementation group E**	1:160.000
FANCG	NM_004629.1	**Fanconi anemia, complementation group G**	1:160.000
FANCI	NM_001113378.1	**Fanconi anemia, complementation group I**	1:160.000
FANCL	NM_018062.3	**Fanconi anemia, complementation group L**	1:160.000
FANCM	NM_020937.2	**Fanconi anemia, complementation group M**	1:160.000
FBP1	NM_000507	**Fructose-biphosphatase deficiency**	Rare
FGD4	NM_139241.2	**Charcot-Marie-Tooth disease type 4H**	1:2.500
FHL1	NM_001449.4	**Emery-Dreifuss muscular dystrophy type 6, Myopathy, reducing body**	1:100.000
FIG4	NM_014845.5	**Charcot-Marie-Tooth disease type 4J, Yunis-Varon syndrome**	1:2.500
FKRP	NM_024301.4	**Congenital muscular dystrophy type 5B, Limb-girdle muscular dystrophy type 2I, autosomal recessive**	UNKNOWN
FKTN	NM_001079802.1	**Fukuyama congenital muscular dystrophy, Muscular dystrophy, limb girdle, type 2M**	1:25.000
FMR1	NM_002024	**Fragile X syndrome, Premature ovarian failure**	1:3600 - 1:6.000
FOXG1	NM_005249	**Rett syndrome**	Rare
FOXN1	NM_003593.2	**T-cell immunodeficiency, congenital alopecia, and nail dystrophy**	<1:200.000
FUCA1	NM_000147.4	**Fucosidosis**	<1:200.000
FXN	NM_000144.4	**Friedreich ataxia**	1:25.000 - 1:50.000
G6PC	NM_000151.3	**Glycogen storage disease type 1a**	1:100.000
G6PC3	NM_138387.3	**Severe congenital neutropenia type 4**	1:200.000
GAA	NM_000152.3	**Glycogen storage disease type 2**	1:100.000
GALC	NM_000153.3	**Krabbe disease**	1:150 - 1:100.000
GALE	NM_000403	**Galactosemia**	1:3.500 - 1:50.000

GALNS	NM_000512	**Mucopolysaccharidosis**	1:200.000 – 1:300.000
GALT	NM_000155.3	**Galactosemia**	1:3.500 - 1:50.000
GAMT	NM_138924	**Guanidinoacetate methyltransferase deficiency**	Rare
GATM	NM_001482	**Arginine:glycine amidinotransferase deficiency**	Unknown
GBA	NM_001005741.2	**Gaucher disease**	1:855 - 1:57.000
GBE1	NM_000158.3	**Glycogen storage disease type 4, Polyglucosan body disease, adult**	1:100.000
GCDH	NM_000159.3	**Glutaric acidemia type 1**	1:300 - 1:40.000
GCH1	NM_000161	**Dopa-responsive dystonia, Tetrahydrobiopterin deficiency**	1:500.000 – 1:1.000.000
GCSH	NM_004483.4	**Glycine encephalopathy (GCSH)**	1:12.000 - 1:63.000
GDAP1	NM_018972.2	**Charcot-Marie-Tooth disease type 4A**	1:2.500
GFPT1	NM_001244710	**Congenital myasthenic syndrome**	Unknown
GJB2	NM_004004.5	**Deafness type 1A, autosomal recessive**	<1:333
GJB3	NM_024009.2	**Deafness type 1A, autosomal recessive**	<1:333
GJB6	NM_006783.4	**Deafness type 1B, autosomal recessive**	<1:333
GJC2	NM_020435.3	**Pelizaeus-Merzbacher-like disease type 1**	1:100.000 - 1:500.000
GLA	NM_000169	**Fabry disease**	1:40.000 – 1:60.000
GLB1	NM_000404.2	**GM1 Gangliosidosis, Mucopolysaccharidosis type 4B**	1:100.000 - 1:200.000
GLDC	NM_000170.2	**Glycine encephalopathy**	1:12.000 - 1:63.000
GLE1	NM_001003722.1	**Lethal arthrogryposis with anterior horn cell disease**	1:3.000
GM2A	NM_000405.4	**GM2 Gangliosidosis (Tay-Sachs disease)**	1:3.500 - 1:320.000
GNE	NM_005476.5	**Distal myopathy Nonaka type**	1:5.000 - 1:300.000
GNPAT	NM_001316350	**Rhizomelic chondrodysplasia punctata**	1:100.000

GNPTAB	NM_024312.4	**Mucolipidosis type 2/type 3**	1:100.000 - 1:400.000
GNPTG	NM_032520	**Mucolipidosis III gamma**	1:100.000 – 1:400.000
GNS	NM_002076.3	**Mucopolysaccharidosis type 3D**	1:100.000 - 1:200.000
GPR143	NM_000273.2	**Ocular albinism, X-linked**	1:60.000
GPR98	NM_032119.3	**Usher syndrome type 2C**	1:6.000
GRHPR	NM_012203.1	**Primary hyperoxaluria type 2**	1:58.000
GRXCR1	NM_001080476.2	**Deafness type 25, autosomal recessive**	<1:333
GUCY2D	NM_000180.3	**Leber congenital amaurosis type 1**	1:30.000
GUSB	NM_000181	**Mucopolysaccharidosis type VII (Sly syndrome)**	1:250.000
GYS2	NM_021957	**Glycogen storage disease**	Unknown
HADHA	NM_000182	**Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency, Mitochondrial trifunctional protein deficiency**	Rare
HADHB	NM_000183	**Mitochondrial trifunctional protein deficiency**	Rare
HAL	NM_002108.3	**Histidinemia**	1:20.000
HBA	NM_000517/000558	**Alpha-thalassemia**	1:100 - 1:2.272
HBB	NM_000518.4	**Beta-thalassemia, Sickle cell anaemia**	1:50 - 1:1.500
HESX1	NM_003865.2	**Combined pituitary hormone deficiencies, genetic forms**	1:8.000
HEXA	NM_000520.4	**Tay-Sachs disease**	1:2.900 - 1:360.000
HEXB	NM_000521.3	**Sandhoff disease**	1:130.000
HFE	NM_000410.3	**Haemochromatosis**	1:200 - 1:1.000
HGD	NM_000187.3	**Alkaptonuria**	1:19.000 - 1:250.000
HGF	NM_000601.4	**Deafness type 39, autosomal recessive**	<1:333
HGSNAT	NM_152419.2	**Mucopolysaccharidosis type 3C**	1:100.000 - 1:500.000

HIBCH	NM_014362.3	**3-Hydroxyisobutryl-CoA hydrolase deficiency**	1:130.000
HLCS	NM_000411	**Holocarboxylase synthetase deficiency**	1:87.000
HMGCS2	NM_005518	**mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency**	Unknown
HPD	NM_002150.2	**Tyrosinemia type 3**	1:1.800 - 1:100.000
HPRT1	NM_000194.2	**Lesch-Nyhan syndrome**	1:380.000
HPS1	NM_000195.3	**Hermansky-Pudlak syndrome type 1**	1:1.800 - 1:500.000
HSD17B4	NM_000414.3	**D-bifunctional protein deficiency, Perrault syndrome**	1:100.000
HSD3B7	NM_001142777	**Congenital bile acid synthesis defect**	1-9:1.000.000
HSPG2	NM_005529	**Schwartz-Jampel syndrome**	Rare
HYLS1	NM_145014.2	**Hydrolethalus syndrome type 1**	1:20.000
IDH3B	NM_006899.3	**Retinitis pigmentosa type 46**	1:4.000
IDS	NM_000202.6	**Mucopolysaccharidosis type 2**	1:100.000 - 1:500.000
IDUA	NM_000203	**Mucopolysaccharidosis type I**	1:100.000 - 1:500.000
IFT80	NM_020800.2	**Short-rib thoracic dysplasia type 2 with or without polydactyly**	1:100.000 - 1:130.000
IGF1	NM_000618.3	**Growth delay due to insulin-like growth factor type 1 deficiency**	<1.000.000
IGHMBP2	NM_002180.2	**Spinal muscular atrophy, distal, type 1, autosomal recessive**	1:6.000 - 1:10.000
IKBKAP	NM_003640.3	**Familial dysautonomia**	1:3.700 (Ashkenazi)
IL2RG	NM_000206.2	**Severe combined immunodeficiency T-B+; X-linked**	1:2.500 - 1:100.000
IMPDH1	NM_000883.3	**Retinitis pigmentosa type 10**	1:4.000
IMPG2	NM_016247.3	**Retinitis pigmentosa type 56**	1:4.000
INF2	NM_022489	**Charcot-Marie-Tooth disease**	1:2.500
INPP5E	NM_019892.4	**Joubert syndrome type 1, MORM syndrome**	1:100.000

INSR	NM_000208.2	**Diabetes mellitus, insulin-resistant, Leprechaunism**	1:6 - 1:33
INVS	NM_014425	**Nephronophthisis**	1:50.000 – 1:1.000.000
IQCB1	NM_014642	**Senior-Løken syndrome**	1:1.000.000
ISCU	NM_213595.3	**Myopathy with deficiency of ISCU**	1:35.000
ISPD	NM_001101426	**Limb-girdle muscular dystrophy, Walker-Warburg syndrome**	1:14.500 – 1:123.000
ITGA3	NM_005501	**Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital**	Unknown
ITGA7	NM_002206	**Muscular dystrophy congenital due to integrin alpha-7 deficiency**	Unknown
IVD	NM_002225.3	**Isovaleric acidemia**	1:250.000
JAK3	NM_000215.3	**Severe combined immunodeficiency T-B+NK-**	1:2.500 - 1:100.000
KCNC3	NM_004977	**Spinocerebellar ataxia 13**	Unknown
KCNJ13	NM_002242.4	**Leber congenital amaurosis type 16**	1:30.000
KCNQ2	NM_172109	**Benign familial neonatal seizures**	1:100.000
KCNQ3	NM_004519	**Benign familial neonatal seizures**	1:100.000
KCNV2	NM_133497.3	**Retinal cone dystrophy type 3B**	1:40.000
KIF7	NM_198525	**Acrocallosal syndrome, Joubert syndrome**	1:80.000 – 1:100.000
L1CAM	NM_000425.4	**MASA syndrome/hydrocephalus**	1:30.000
LAMA2	NM_000426.3	**Congenital muscular dystrophy type 1A**	1:30.000
LAMB2	NM_002292	**Congenital nephrotic syndrome**	1:10.000 – 1:100.000
LARGE	NM_004737.4	**Muscular dystrophy-dystroglycanopathy type 6**	1:30.000
LBR	NM_002296	**Greenberg dysplasia**	Rare
LDHA	NM_005566.3	**Glycogen storage disease type 11**	1:100.000
LDLR	NM_000527	**Hypercholesterolemia**	1:500

LEPRE1	NM_022356.3	**Osteogenesis imperfecta type 8**	1:10.000 - 1:20.000
LHFPL5	NM_182548.3	**Deafness type 67, autosomal recessive**	<1:333
LHX3	NM_014564.3	**Combined pituitary hormone deficiency type 3**	1:8.000
LIFR	NM_002310.5	**Stuve-Wiedemann syndrome**	1:20.000
LMNA	NM_170707.3	**Hutchinson-Gilford progeria syndrome, Lipodystrophy, familial partial, type 2, Mandibuloacral dysplasia, Muscular dystrophy, Emery-Dreifuss type 3, Cardiomyopathy, dilated type 1A**	1:2.500 - 1:100.000
LMX1B	NM_001174146	**Nail-patella syndrome**	1:50.000
LOXHD1	NM_144612.6	**Deafness type 77, autosomal recessive**	<1:333
LPL	NM_000237	**Familial lipoprotein lipase deficiency**	<1:1.000.000
LRAT	NM_004744.3	**Leber congenital amaurosis type 14, Retinal dystrophy, early-onset severe**	1:5.000 - 1:67.000
LRP5	NM_002335.3	**Exudative vitreoretinopathy type 4, Isolated polycystic liver disease, Osteoporosis-pseudoglioma syndrome**	1:100.000
LRPPRC	NM_133259.3	**Leigh syndrome, French-Canadian type**	1:36.000
LRTOMT	NM_001145308.4	**Deafness type 63, utosomal recessive**	<1:333
MAK	NM_001242957.1	**Retinitis pigmentosa type 62**	1:4.000
MAN2B1	NM_000528.3	**Alpha-mannosidosis**	1:300.000
MANBA	NM_005908	**Beta-mannosidosis**	Rare
MARVELD2	NM_001038603.2	**Deafness type 49, autosomal recessive**	<1:333
MAT1A	NM_000429.2	**Methionine adenosyltransferase deficiency**	1:20.000
MATN3	NM_002381.4	**Multiple epiphyseal dysplasia type 5**	1:10.000
MBTPS2	NM_015884.3	**Ichthyosis follicularis-atrichia-photophobia**	1:2.000 - 1:10.000
MCCC1	NM_020166.4	**3-Methylcrotonyl-CoA carboxylase type 1 deficiency**	1:36.000
MCCC2	NM_022132.4	**3-Methylcrotonyl-CoA carboxylase 2 deficiency, type 2**	1:36.000

MCEE	NM_032601.3	**Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency**	1:50.000 - 1:80.000
MCOLN1	NM_020533.2	**Mucolipidosis type 4**	1:100.000 - 1:400.000
MCPH1	NM_024596.3	**Microcephaly, primary, type 1, autosomal recessive**	1:10.000 - 1:250.000
MECP2	NM_004992.3	**Rett syndrome**	1:8.500
MED25	NM_030973.3	**Charcot-Marie-Tooth disease type 2B2**	1:2.500 - 1:250.000
MEFV	NM_000243.2	**Familial mediterranean fever**	1:200 - 1:1.000
MERTK	NM_006343.2	**Retinitis pigmentosa type 38**	1:4.000
MFRP	NM_031433.3	**Microphthalmia - Retinitis pigmentosa - foveoschisis - optic disc drusen**	1:4.000
MFSD8	NM_152778.2	**Ceroid lipofuscinosis, neuronal, type 7**	1:385.000
MGAT2	NM_002408.3	**Congenital disorders of glycosylation 2a type**	1:20.000
MKKS	NM_018848.3	**Bardet-Biedl/McKusick-Kaufman syndrome, Bardet-Biedl syndrome type 6, McKusick-Kaufman syndrome**	1:100.000 - 1:175.000
MKS1	NM_017777.3	**Bardet-Biedl syndrome type 13, Meckel type 1/Bardet-Biedl syndrome**	1:13.500 - 1:140.000
MMAA	NM_172250.2	**Vitamin B12-responsive methylmalonic acidemia type cblA**	1:48.000 - 1:61.000
MMAB	NM_052845.3	**Vitamin B12-responsive methylmalonic acidemia type cblB**	1:48.000 - 1:61.000
MMACHC	NM_015506.2	**Methylmalonic aciduria cblC type, with homocystinuria**	1:37.000 - 1:100.000
MMADHC	NM_015702.2	**methylmalonic aciduria cblD type, with homocystinuria**	1:37.000 - 1:100.000
MOCS1	NM_005943.5	**Molybdenum cofactor deficiency type A**	1:100.000 - 1:200.000
MOCS2	NM_176806.3	**Molybdenum cofactor deficiency type B**	1:100.000 - 1:200.000
MPI	NM_002435.2	**Congenital disorders of glycosylation type 1b**	1:20.000
MPZ	NM_000530.6	**Dejerine-Sottas syndrome (MPZ), Neuropathy, congenital hypomyelinating or amyelinating**	1:3.500
MTHFR	NM_005957.4	**Homocystinuria due to MTHFR deficiency**	1:65.000
MTM1	NM_000252.2	**Myotubular myopathy, X-linked**	1:50.000

MTMR2	NM_016156.5	**Charcot-Marie-Tooth disease type 4B1**	1:2.500 - 1:250.000
MUSK	NM_001166280	**Congenital myasthenic syndrome**	Unknown
MUT	NM_000255.3	**Methylmalonic acidemia**	1:25.000 - 1:48.000
MVK	NM_000431	**Mevalonate kinase deficiency**	Rare
MYO15A	NM_016239.3	**Deafness type 3, autosomal recessive**	<1:333
MYO3A	NM_017433.4	**Deafness type 30, autosomal recessive**	<1:333
MYO6	NM_004999.3	**Deafness type 37, autosomal recessive**	<1:333
MYO7A	NM_000260.3	**Deafness type 2, autosomal recessive, Usher syndrome type 1B**	<1:333
NAGLU	NM_000263	**Mucopolysaccharidosis type III**	1:70.000
NAGS	NM_153006	**N-acetylglutamate synthase deficiency**	Rare
NDRG1	NM_006096.3	**Charcot-Marie-Tooth disease, type 4D**	1:2.500 - 1:250.000
NEB	NM_004543.4	**Nemaline myopathy type 2**	1:50.000
NEFL	NM_006158.3	**Charcot-Marie-Tooth disease type 1F**	1:2.500 - 1:250.000
NEK8	NM_178170	**Nephronophthisis**	1:50.000 – 1:1.000.000
NMNAT1	NM_022787.3	**Leber congenital amaurosis type 9**	1:50.000 - 1:100.000
NOTCH2	NM_024408	**Alagille syndrome, Hajdu-Cheney syndrome, cancers**	1 :70.000
NPC1	NM_000271.4	**Niemann-Pick disease type C1**	1:120.000
NPC2	NM_006432.3	**Niemann-Pick disease type C2**	1:120.000
NPHP1	NM_000272.3	**Nephronophthisis type 1**	1:50.000 - 1:100.000
NPHP3	NM_153240.4	**Nephronophthisis type 3**	1:50.000 - 1:100.000
NPHP4	NM_015102.4	**Nephronophthisis type 4**	1:50.000 - 1:100.000
NPHS1	NM_004646.3	**Nephrotic syndrome type 1**	1:6.000

NPHS2	NM_001297575	**Nephrotic syndrome**	1:10.000
NR0B1	NM_000475.4	**Cytomegalic congenital adrenal hypoplasia**	1:10.000
NSDHL	NM_015922	**Congenital hemidysplasia with ichthyosiform erythroderma and limb defects (CHILD syndrome)**	Rare
NTRK1	NM_001012331.1	**Hereditary sensory and autonomic neuropathy type 4**	1:25.000
NYX	NM_022567.2	**Night blindness, congenital stationary , type 1A, X-linked**	1:100
OAT	NM_000274.3	**Gyrate atrophy of choroid and retina with or without ornithinemia**	1:50.000
OCA2	NM_000275.2	**Oculocutaneous albinism type 2**	1:1.400 - 1:20.000
OCRL	NM_000276.3	**Lowe syndrome**	1:500.000
OFD1	NM_003611.2	**Joubert syndrome type 10, Orofaciodigital syndrome type 1**	1:100.000
OPA3	NM_025136.3	**3-methylglutaconic aciduria type 3**	1:10.000
OSTM1	NM_014028.3	**Osteopetrosis type 5, autosomal recessive**	1:100.000 - 1:500.000
OTC	NM_000531.5	**Ornithine transcarbamylase deficiency**	1:56.500 - 1:77.000
OTOA	NM_144672.3	**Deafness type 22, autosomal recessive**	<1:333
OTOF	NM_194248.2	**Deaffness type 9, autosomal recessive**	<1:333
OXCT1	NM_000436	**Succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency**	Unknown
PAH	NM_000277.1	**Phenylketonuria**	1:10.000
PALB2	NM_024675.3	**Fanconi anemia, complementation group N**	1:30.000 - 1:40.000
PAX3	NM_181457.3	**Waardenburg syndrome type 3**	1:40.000
PAX6	NM_000280.4	**Aniridia**	1:40.000 - 1:100.000
PC	NM_000920.3	**Pyruvate carboxylase deficiency**	1:250.000
PCCA	NM_000282.3	**Propionic acidemia type 1**	1:35.000
PCCB	NM_000532.4	**Propionic acidemia type 2**	1:35.000

PCDH15	NM_033056.3	**Usher syndrome type 1F**	1:6.000
PDE6A	NM_000440.2	**Retinitis pigmentosa type 43**	1:4.000
PDE6B	NM_000283.3	**Retinitis pigmentosa type 43**	1:4.000
PDE6C	NM_006204.3	**Cone dystrophy type 4**	1:40.000
PDE6G	NM_002602.3	**Retinitis pigmentosa type 57**	1:4.000
PDSS1	NM_001321978	**Coenzyme Q10 deficiency**	Unknown
PDSS2	NM_020381	**Leigh syndrome**	1:2.000 – 1:40.000
PDYN	NM_001190892	**Spinocerebellar ataxia 23**	Unknown
PDZD7	NM_001195263.1	**Usher syndrome type 2C**	1:6.000
PEX1	NM_000466.2	**Peroxisome biogenesis disorder type 1A, Peroxisome biogenesis disorder type 1B**	1:50.000
PEX12	NM_000286.2	**Peroxisome biogenesis disorder complementation group 6**	1:50.000
PEX13	NM_002618	**Zellweger spectrum disorder**	1:50.000
PEX14	NM_004565	**Zellweger spectrum disorder**	1:50.000
PEX2	NM_000318.2	**Peroxisome biogenesis disorder complementation group 5**	1:50.000
PEX26	NM_017929.5	**Peroxisome biogenesis disorder type 7**	1:50.000
PEX3	NM_003630	**Zellweger spectrum disorder**	1:50.000
PEX5	NM_001131025.1	**Peroxisome biogenesis disorder type 2**	1:50.000
PEX6	NM_000287	**Zellweger spectrum disorder**	1:50.000
PEX7	NM_000288.3	**Rhizomelic chondrodysplasia punctata type 1**	1:100.000
PFKM	NM_000289	**Glycogen storage disease type VII**	Rare
PGM1	NM_002633.2	**Congenital disorder of glycosylation, type 1T**	1:20.000
PHKA2	NM_000292	**Glycogen storage disease type IX**	1:100.000

PHKG2	NM_000294.2	**Glycogen storage disease type 9C**	1:100.000
PHYH	NM_001037537	**Refsum disease**	Unknown
PKD1	NM_000296	**Polycystic kidney disease**	1:500
PKD2	NM_000297	**Polycystic kidney disease**	1:500
PKHD1	NM_138694.3	**Polycystic kidney disease, autosomal recessive**	1:20.000
PKLR	NM_000298.5	**Hemolytic anemia due to red cell pyruvate kinase deficiency**	1:20.000
PLCE1	NM_016341.3	**Nephrotic syndrome type 3**	2-17:100.000
PLEC	NM_000445.4	**Epidermolysis bullosa simplex with muscular dystrophy, Epidermolysis bullosa simplex with pyloric atresia**	1:53.000
PLEKHG5	NM_020631.4	**Charcot-Marie-Tooth disease, intermediate type C**	1:2.500 - 1:250.000
PLG	NM_000301.3	**Congenital plasminogen deficiency type 1**	1,6:1.000.000
PLOD1	NM_000302.3	**Ehlers-Danlos syndrome, type 6**	1:5.000
PLP1	NM_000533.3	**Pelizaeus-Merzbacher disease, Spastic paraplegia type 2, X-linked**	1:100.000 - 200.000
PNPO	NM_018129.3	**PNPO deficiency**	1-9:1.000.000
POLG	NM_002693.2	**Mitochondrial DNA depletion syndrome, Alpers type, Progressive external ophthalmoplegia**	UNKNOWN
POMGNT1	NM_017739.3	**Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A3**	1:10.000
POMGNT2	NM_032806	**Walker-Warburg syndrome**	1:65.000
POMT1	NM_007171.3	**Congenital muscular dystrophy with intellectual disability type B1, Walker-Warburg syndrome**	1:60.000
POMT2	NM_013382.5	**Congenital muscular dystrophy with intellectual disability type A2, Walker-Warburg syndrome**	1:60.000
POR	NM_000941	**Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis**	Unknown
POU1F1	NM_000306.3	**Pituitary hormone deficiency, combined, type 1**	1:8.000

POU3F4	NM_000307.4	**Deafness type 2, X-linked**	<1:333
PPP2R2B	NM_181678	**Spinocerebellar ataxia 12**	Unknown
PPT1	NM_000310.3	**Neuronal ceroid-lipofuscinoses type 1**	1:20.000 -1:100.000
PRCD	NM_001077620.2	**Retinitis pigmentosa 36**	1:4.000
PRKCG	NM_002739	**Spinocerebellar ataxia 14**	Unknown
PRKRA	NM_003690.4	**Dystonia tipo 16**	1:1.000 - 1:50.000
PROM1	NM_006017.2	**Retinitis pigmentosa type 41**	1:4.000
PROP1	NM_006261.4	**Pituitary hormone deficiency, combined, type 2**	1:8.000
PRPS1	NM_002764.3	**Charcot-Marie-Tooth disease type 5, X-linked recessive, Deafness, X-linked, Phosphoribosylpyrophosphate synthetase superactivity, Sensorineural deafness, nonsyndromic, X-linked**	<1:333
PRRT2	NM_145239	**Familial hemiplegic migraine, Familial paroxysmal kinesigenic dyskinesia**	1:10.000 – 1:150.000
PRX	NM_181882.2	**Charcot-Marie-Tooth disease type 4F, Dejerine-Sottas syndrome (PRX)**	1:2.500 - 1:250.000
PSAP	NM_002778	**Metachromatic leukodystrophy**	1:8.000 – 1:160.000
PTPRO	NM_002848	**Nephrotic syndrome 6**	Unknown
PTRF	NM_012232	**Congenital generalized lipodystrophy (Berardinelli-Seip congenital lipodystrophy)**	1:10.000.000
PTS	NM_000317	**Tetrahydrobiopterin deficiency**	Rare
PYGL	NM_002863	**Glycogen storage disease type VI**	Unknown
PYGM	NM_005609.2	**McArdle disease**	1:100.000
RAD51	NM_002875	**Breast cancer**	1:33
RAD51C	NM_058216.2	**Fanconi anemia, complementation group O**	1:160.000
RAG1	NM_000448.2	**Immunodeficiency severe combined B cell-negative, Omenn syndrome**	1:200.000 -1:1.000.000

RAG2	NM_000536.3	**Combined immunodeficiency with skin granulomas, Omenn syndrome**	1:200.000 -1:1.000.000
RAPSN	NM_005055.4	**Congenital myasthenic syndrome, Fetal akinesia deformation sequence**	1:500.000
RAX	NM_013435.2	**Isolated microphthalmia type 3**	1:10.000 - 1:100.000
RDH12	NM_152443.2	**Leber congenital amaurosis type 13**	1:30.000 - 1:50.000
RDX	NM_002906.3	**Deafness type 24, autosomal recessive**	<1:333
RELN	NM_005045.3	**Lissencephaly syndrome, Norman-Roberts type**	1:100.000
RGR	NM_001012720.1	**Retinitis pigmentosa type 44**	1:3.500
REX5	NM_000319	**Peroxisome biogenesis disorder (Zellweger spectrum disorder)**	1:50.000
RHO	NM_000539.3	**Retinitis pigmentosa type 4**	1:3.500
RLBP1	NM_000326.4	**Retinitis punctata albescens**	1:800.000
RP2	NM_006915.2	**Retinitis pigmentosa type 2**	1:3.500
RPE65	NM_000329.2	**Leber congenital amaurosis type 2, Retinitis pigmentosa type 20**	1:3.500 - 1:50.000
RPGR	NM_001034853.1	**Retinitis pigmentosa type 3**	1:3.500
RPGRIP1L	NM_015272.2	**Joubert syndrome type 7, Meckel syndrome type 5**	1:1.300 - 1:100.000
SACS	NM_014363.5	**Spastic ataxia, Charlevoix-Saguenay type**	1:20.000 - 1:100.000
SBDS	NM_016038.2	**Shwachman-Diamond syndrome**	1:77.000
SBF2	NM_030962.3	**Charcot-Marie-Tooth disease type 4B2**	1:2.500 - 1:250.000
SC5D	NM_006918.4	**Lathosterolosis**	1:10.000 - 1:60.000
SCN1A	NM_006920	**Familial hemiplegic migraine, Malignant migrating partial seizures of infancy**	1:10.000
SCN2A	NM_021007	**Benign familial neonatal-infantile seizures, Early infantile epileptic encephalopathy 11**	Unknown
SCN4A	NM_000334	**Congenital myasthenic syndrome, Hyperkalemic periodic paralysis, Hypokalemic periodic paralysis, Paramyotonia congenita**	1:100.000

SDCCAG8	NM_006642	**Senior-Løken syndrome**	Rare
SEC63	NM_007214	**Polycystic liver disease**	Unknown
SEMA4A	NM_022367.3	**Retinitis pigmentosa type 35**	1:3.500
SEPN1	NM_020451.2	**Muscular dystrophy, rigid spine, type 1**	UNKNOWN
SERPINA1	NM_000295.4	**Alpha1-antitrypsin deficiency**	1:1.500 - 1:3.500
SETX	NM_015046.5	**Spinocerebellar ataxia with axonal neuropathy type 2**	1:10.000
SGCA	NM_000023.2	**Limb-girdle muscular dystrophy type 2D**	1:14.500 - 1:143.000
SGCB	NM_000232.4	**Muscular dystrophy, limb-girdle, type 2E**	1:14.500 - 1:143.000
SGCD	NM_000337	**Familial dilated cardiomyopathy, Limb-girdle muscular dystrophy**	1:14.500
SGCG	NM_000231.2	**Limb-girdle muscular dystrophy type 2C**	1:14.500 - 1:143.000
SGSH	NM_000199.3	**Mucopolysaccharidosis type 3A (Sanfilippo disease type A)**	1:10.000
SH3TC2	NM_024577.3	**Charcot-Marie-Tooth disease type 4C**	1:2.500 - 1:250.000
SIX6	NM_007374.2	**Anophthalmia or microphthalmia, isolated**	1:4.000
SLC22A5	NM_003060	**Primary carnitine deficiency**	1:40.000
SLC25A13	NM_014251.2	**Citrullinemia type 2**	1:57.000 - 1:100.000
SLC25A15	NM_014252	**Ornithine translocase deficiency**	Rare
SLC25A20	NM_000387	**Carnitine-acylcarnitine translocase (CACT) deficiency**	Rare
SLC25A22	NM_024698.5	**Epileptic encephalopathy, early infantile, type 3**	1:50.000 - 1:100.000
SLC26A2	NM_000112.3	**Atelosteogenesis type 2, Diastrophic dysplasia**	1:100.000
SLC26A4	NM_000441.1	**Deafness type 4, autosomal recessive, Pendred syndrome**	<1:333
SLC26A5	NM_198999.2	**Deafness type 61, autosomal recessive**	<1:333
SLC27A1	NM_198580	**Futty acid transportation defect**	Unknown

SLC2A1	NM_006516	**GLUT1 deficiency syndrome**	1:90.000
SLC2A2	NM_000340	**Diabetes mellitus type 2, Fanconi-Bickel syndrome**	Unknown
SLC37A4	NM_001164278.1	**Glycogen storage disease types 1b, 1c and 1d**	1:100.000
SLC45A2	NM_016180.3	**Albinism, oculocutaneous, type 4**	1:20.000
SLC4A11	NM_032034.3	**Congenital hereditary endothelial dystrophy type 2, Corneal dystrophy and perceptive deafness**	1:64.500
SLX4	NM_032444.2	**Fanconi anemia, complementation group P**	1:160.000
SMN1	NM_000344	**Spinal muscular atrophy**	1:10.000
SMPD1	NM_000543.4	**Niemann-Pick disease**	1:40.000 - 1:250.000
SNAI2	NM_003068.4	**Waardenburg syndrome type 2**	1:40.000
SPG11	NM_025137.3	**Spastic paraplegia type 11**	1:12.000 - 1:80.000
SPG20	NM_015087.4	**Spastic paraplegia type 20, autosomal recessive**	1:12.000 - 1:80.000
SPG7	NM_003119.3	**Spastic paraplegia type 7**	1:12.000 - 1:80.000
STAR	NM_000349.2	**Lipoid adrenal hyperplasia**	1:10.000
STIL	NM_003035.2	**Microcephaly primary, type 7, autosomal recessive**	1:30.000 - 1:250.000
STRA6	NM_022369.3	**Syndromic microphthalmia type 9**	1:5.000 - 1:50.000
STRC	NM_153700.2	**Deafness type 16, autosomal recessive**	<1:333
SUMF1	NM_182760	**Multiple sulfatase deficiency**	1:1.000.000
SUOX	NM_000456.2	**Sulfocysteinuria**	1:20.000 - 1:64.000
SYNE1	NM_182961	**Autosomal recessive cerebellar ataxia type 1**	Unknown
SYNE2	NM_182914	**Emery-Dreifuss muscular dystrophy 5**	Unknown
TAF1	NM_004606.4	**Dystonia-Parkinsonism, X-linked**	1:20.000
TAT	NM_000353.2	**Tyrosinemia type 2**	1:250.000

TBC1D24	NM_020705	**DOORS syndrome, Malignant migrating partial seizures of infancy , Nonsyndromic hearing loss**	1:500
TBP	NM_003194	**Huntington disease-like (HDL) syndrome**	Rare
TCAP	NM_003673.3	**Cardiomyopathy, hypertrophic, type 25, Limb-girdle muscular dystrophy type 2G**	1:500 - 1:143.000
TCIRG1	NM_006019.3	**Osteopetrosis type 1, autosomal recessive**	1:250.000
TCTN1	NM_024549	**Joubert syndrome 13**	Unknown
TCTN3	NM_015631	**Joubert syndrome 13, Orofacial-digital syndrome IV**	Unknown
TECTA	NM_005422.2	**Deafness type 21, autosomal recessive**	<1:333
TFR2	NM_003227.3	**Hemochromatosis, type 3**	1:200 - 1:400
TH	NM_000360.3	**Segawa syndrome, autosomal recessive**	1:200.000 - 1:1.000.000
TMC1	NM_138691.2	**Deafness type 7, autosomal recessive**	<1:333
TMEM138	NM_016464	**Joubert syndrome 16**	Unknown
TMEM216	NM_001173990.2	**Joubert syndrome type 2, Meckel syndrome type 2**	1:80.000 - 1:100.000
TMEM231	NM_001077416	**Meckel syndrome, type 11, Joubert syndrome 20**	Unknown
TMEM237	NM_152388	**Joubert syndrome 14**	Unknown
TMEM43	NM_024334	**Arrhythmogenic right ventricular cardiomyopathy**	1:1.000 – 1:1.250
TMEM67	NM_153704.5	**COACH syndrome, Joubert syndrome type 6, Meckel syndrome type 3**	1:80.000 - 1:100.000
TMIE	NM_147196.2	**Deafness type 6, autosomal recessive**	<1:333
TMPRSS3	NM_024022.2	**Deafness types 8/10, autosomal recessive**	<1:333
TNNT1	NM_003283.5	**Nemaline myopathy type 5**	1:50.000
TPP1	NM_000391.3	**Neuronal ceroid-lipofuscinoses type 2**	1:200.000
TPRN	NM_001128228.2	**Deafness type 79, autosomal recessive**	<1:333
TRIM32	NM_012210.3	**Limb-girdle muscular dystrophy type 2H**	1:14.500 - 1:143.000

TRIOBP	NM_001039141.2	**Deafness type 28, autosomal recessive**	<1:333
TRPC6	NM_004621	**Focal segmental glomerulosclerosis 2**	Unknown
TSHR	NM_000369.2	**Hypothyroidism**	1:4.000
TTC21B	NM_024753	**Joubert syndrome 11, Nephronophthisis 12, Bardet-Biedl syndrome**	Unknown
TTN	NM_133378	**Centronuclear myopathy, Early-onset myopathy with fatal cardiomyopathy, familial dilated cardiomyopathy, familial hypertrophic cardiomyopathy, Hereditary myopathy with early respiratory failure, Limb-girdle muscular dystrophy Tibial muscular dystrophy**	>1:500
TULP1	NM_003322.4	**Leber congenital amaurosis type 15, Retinitis pigmentosa type 14**	1:3.500 - 1:50.000
TYR	NM_000372.4	**Oculocutaneous albinism type 1**	1:20.000
TYRP1	NM_000550.2	**Oculocutaneous albinism type 3**	1:20.000
UBA1	NM_003334.3	**Spinal muscular atrophy type 2, X-linked**	1:12.000
UBE2T	NM_014176	**Fanconi anemia complementation group T**	Unknown
UBR1	NM_174916.2	**Johanson-Blizzard syndrome**	1:250.000
UGT1A1	NM_000463.2	**Crigler-Najjar syndrome type 1, Crigler-Najjar syndrome type 2, Gilbert syndrome**	1:10 - 1:10.000
UMOD	NM_001008389	**Uromodulin-associated kidney disease**	Unknown
UQCRB	NM_006294.4	**Mitochondrial complex III deficiency, nuclear type 3**	UNKNOWN
UQCRQ	NM_014402.4	**Mitochondrial complex III deficiency, nuclear type 4**	UNKNOWN
USH1C	NM_153676.3	**Usher syndrome type 1C**	1:6.000 - 1:15.000
USH1G	NM_173477.4	**Usher syndrome type 1G**	1:6.000 - 1:15.000
USH2A	NM_206933.2	**Retinitis pigmentosa type 39, Usher syndrome type 2A**	1:3.500 - 1:15.000
VDR	NM_001017535.1	**Rickets, vitamin D-resistant, type 2A**	1:200.000
WAS	NM_000377.2	**Neutropenia, severe congenital, X-linked, Thrombocytopaenia type 1,Wiskott-Aldrich syndrome**	1:100.000 - 1:200.000
WDR19	NM_001317924	**Asphyxiating thoracic dystrophy, cranioectodermal dysplasia, Nephronophthisis, Retinitis pigmentosa, Senior-Løken syndrome**	1:3.500 - 1:15.000

WDR62	NM_001083961.1	**Microcephaly primary, type 2, autosomal recessive**	1:30.000 - 1:250.000
WFS1	NM_006005.3	**Wolfram syndrome**	1:500.000
WNT10A	NM_025216.2	**Hypohidrotic ectodermal dysplasia, autosomal recessive, Odontoonychodermal dysplasia**	1:50.000 - 1:100.000
WT1	NM_000378	**Congenital nephrotic syndrome, Denys-Drash syndrome, Frasier syndrome**	1:100.000
ZFYVE26	NM_015346.3	**Spastic paraplegia type 15, autosomal recessive**	1:10.000 - 1:100.000
ZNF423	NM_015069	**Joubert syndrome 19, Nephronophthisis 14**	Unknown

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