Disorder	Inheritance	Gene/Locus	Detection Rate	Carrier frequency	Residual risk of being a carrier	Residual risk of having an affected child *If both parents test negative	Residual risk of having an affected child *If one of the parents tests positive and one tests negative
Metabolic System							
Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	AR	*CYP11B1*	99	1/158	1/15800	1/998560000	1/63200
17-alpha-hydroxylase	AR	*CYP17A1*	99	<1/500	1/50000	1/10000000000	1/200000
Pseudohermaphroditism, male, with gynecomastia	AR	*HSD17B3*	99	1/135	1/13500	1/729000000	1/54000
Peroxisome biogenesis disorder 1A (Zellweger)	AR	*PEX1*	99	1/500	1/50000	1/10000000000	1/200000
2,4-dienoyl-CoA reductase deficiency	AR	*NADK2*	99	<1/500	1/50000	1/10000000000	1/200000
Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	AR	*CYP21A2*	63	1/61	1/165	1/108722	1/659
2-methylbutyrylglycinuria	AR	*ACADSB*	99	<1/500	1/50000	1/10000000000	1/200000
Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR	*HSD3B2*	99	<1/500	1/50000	1/10000000000	1/200000
3-Methylcrotonyl-CoA carboxylase 1 deficiency	AR	*MCCC1*	98	1/95	1/4750	1/90250000	1/19000
3-Methylcrotonyl-CoA carboxylase 2 deficiency	AR	*MCCC2*	94	1/95	1/1583	1/10027778	1/6333
3-methylglutaconic aciduria, type III	AR	*OPA3*	99	<1/500	1/50000	1/10000000000	1/200000
3-methylglutaconic aciduria, type IX	AR	*TIMM50*	89	3/500	1/1515	1/9182736	1/6061
3-methylglutaconic aciduria, type I	AR	*AUH*	99	3/500	1/16667	1/1111111111	1/66667
3-methylglutaconic aciduria, type VIII	AR	*HTRA2*	99	3/500	1/16667	1/1111111111	1/66667
3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia	AR	*CLPB*	99	3/500	1/16667	1/1111111111	1/66667
3-methylglutaconic aciduria, type V	AR	*DNAJC19*	99	3/500	1/16667	1/1111111111	1/66667
3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	AR	*SERAC1*	93	3/500	1/2381	1/22675737	1/9524
HMG-CoA synthase-2 deficiency	AR	*HMGCS2*	99	<1/250	1/25000	1/2500000000	1/100000
3-hydroxyacyl-CoA dehydrogenase deficiency	AR	*HADH*	92	1/240	1/6250	1/156250000	1/25000
Peroxisome biogenesis disorder 4A (Zellweger)	AR	*PEX6*	99	1/280	1/28000	1/3136000000	1/112000
Glycine encephalopathy	AR	*AMT*	99	1/373	1/37300	1/5565160000	1/149200
Hyperphenylalaninemia, BH4-deficient, A	AR	*PTS*	98	<1/500	1/25000	1/2500000000	1/100000
Hyperphenylalaninemia, BH4-deficient, B	AR	*GCH1*	83	<1/500	1/2941	1/34602076	1/11765
Hyperphenylalaninemia, BH4-deficient, C	AR	*QDPR*	99	<1/500	1/50000	1/10000000000	1/200000
Hyperphenylalaninemia, BH4-deficient, D	AR	*PCBD1*	99	1/200	1/20000	1/1600000000	1/80000
D-bifunctional protein deficiency	AR	*HSD17B4*	93	1/158	1/2257	1/20378776	1/9029
HMG-CoA lyase deficiency	AR	*HMGCL*	99	<1/500	1/50000	1/10000000000	1/200000
Glycogen storage disease IIIa	AR	*AGL*	99	1/158	1/15800	1/998560000	1/63200
Glycogen storage disease IV	AR	*GBE1*	92	1/387	1/4838	1/93605625	1/19350
N-acetylglutamate synthase deficiency	AR	*NAGS*	99	<1/500	1/50000	1/10000000000	1/200000
Peroxisome biogenesis disorder 6A (Zellweger)	AR	*PEX10*	86	1/500	1/3571	1/51020408	1/14286
Peroxisome biogenesis disorder 3A (Zellweger)	AR	*PEX12*	99	1/373	1/37300	1/5565160000	1/149200
Peroxisome biogenesis disorder 5A (Zellweger)	AR	*PEX2*	99	1/500	1/50000	1/10000000000	1/200000
Combined oxidative phosphorylation deficiency 3	AR	*TSFM*	99	<1/500	1/50000	1/10000000000	1/200000
Adrenal hypoplasia, congenital	XL	*NR0B1*	99	1/12500	1/1250000	1/6250000000000	1/5000000
Emphysema due to AAT deficiency	AR	*SERPINA1*	99	1/33	1/3300	1/43560000	1/13200
Mannosidosis, alpha-, types I and II	AR	*MAN2B1*	99	1/354	1/35400	1/5012640000	1/141600
Alpha-methylacetoacetic aciduria	AR	*ACAT1*	89	<1/500	1/4545	1/82644628	1/18182
Carbamoylphosphate synthetase I deficiency	AR	*CPS1*	99	1/570	1/57000	1/12996000000	1/228000
Galactosemia	AR	*GALT*	99	1/110	1/11000	1/484000000	1/44000
Galactose epimerase deficiency	AR	*GALE*	99	1/100	1/10000	1/400000000	1/40000
Galactokinase deficiency with cataracts	AR	*GALK1*	99	1/110	1/11000	1/484000000	1/44000
Galactosemia IV	AR	*GALM*	99	1/250	1/25000	1/2500000000	1/100000
Pyruvate carboxylase deficiency	AR	*PC*	99	1/250	1/25000	1/2500000000	1/100000
Phenylketonuria	AR	*PAH*	99	1/99	1/9900	1/392040000	1/39600
Propionicacidemia	AR	*PCCB*	96	1/224	1/5600	1/125440000	1/22400

Propionic acidemia	AR	*PCCA*	82	1/224	1/1244	1/6194568	1/4978
Pyruvate dehydrogenase E1-beta deficiency	AR	*PDHB*	96	<1/500	1/12500	1/625000000	1/50000
Cerebral creatine deficiency syndrome 1	XL	*SLC6A8*	94	<1/500	1/8333	1/277777778	1/33333
Cholesteryl ester storage disease	AR	*LIPA*	99	<1/500	1/50000	1/10000000000	1/200000
Hypophosphatasia, infantile	AR	*ALPL*	99	1/158	1/15800	1/998560000	1/63200
Butyrylcholinesterase deficiency	AR	*BCHE*	99	1/28	1/2800	1/31360000	1/11200
Multiple sulfatase deficiency	AR	*SUMF1*	99	1/500	1/50000	1/10000000000	1/200000
Dihydropyrimidine dehydrogenase deficiency	AR	*DPYD*	99	<1/500	1/50000	1/10000000000	1/200000
Fabry disease	XL	*GLA*	93	<1/500	1/7143	1/204081633	1/28571
Aromatase deficiency	AR	*CYP19A1*	99	<1/500	1/50000	1/10000000000	1/200000
Hyperphenylalaninemia, mild, non-BH4-deficient	AR	*DNAJC12*	94	<1/200	1/3333	1/44444444	1/13333
Glycine encephalopathy	AR	*GLDC*	99	1/193	1/19300	1/1489960000	1/77200
Maple syrup urine disease, type Ia	AR	*BCKDHA*	99	1/321	1/32100	1/4121640000	1/128400
Maple syrup urine disease, type Ib	AR	*BCKDHB*	90	1/364	1/3640	1/52998400	1/14560
Dihydrolipoamide dehydrogenase deficiency	AR	*DLD*	99	1/500	1/50000	1/10000000000	1/200000
Maple syrup urine disease, type II	AR	*DBT*	69	1/481	1/1552	1/9630010	1/6206
Wilson disease	AR	*ATP7B*	99	1/90	1/9000	1/324000000	1/36000
Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	AR	*MPV17*	99	<1/500	1/50000	1/10000000000	1/200000
Methionine adenosyltransferase deficiency, autosomal recessive	AR/AD	*MAT1A*	99	3/500	1/16667	1/1111111111	1/66667
Homocystinuria-megaloblastic anemia, cbl E type	AR	*MTRR*	93	<1/500	1/7143	1/204081633	1/28571
Homocystinuria-megaloblastic anemia, cblG complementation type	AR	*MTR*	94	<1/2000	1/33333	1/4444444444	1/133333
Homocystinuria due to MTHFR deficiency	AR	*MTHFR*	99	1/2	1/200	1/160000	1/800
Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	AR	*SLC25A15*	92	<1/500	1/6250	1/156250000	1/25000
Thrombosis, hyperhomocysteinemic	AR	*CBS*	10	1/224	1/249	1/247783	1/996
Gaucher disease	AR	*GBA1*	64	3/500	1/463	1/857339	1/1852
Sitosterolemia 1	AR	*ABCG8*	99	9/1000	1/11111	1/493827160	1/44444
Sitosterolemia 2	AR	*ABCG5*	93	9/1000	1/1587	1/10078105	1/6349
Citrullinemia, type II, neonatal-onset	AR	*SLC25A13*	99	1/65	1/6500	1/169000000	1/26000
Citrullinemia	AR	*ASS1*	41	1/119	1/202	1/162723	1/807
Cerebral creatine deficiency syndrome 2	AR	*GAMT*	72	1/371	1/1325	1/7022500	1/5300
Cystinosis, ocular nonnephropathic	AR	*CTNS*	93	1/158	1/2257	1/20378776	1/9029
Peroxisomal acyl-CoA oxidase deficiency	AR	*ACOX1*	99	<1/500	1/50000	1/10000000000	1/200000
VLCAD deficiency	AR	*ACADVL*	99	1/200	1/20000	1/1600000000	1/80000
Lipoprotein lipase deficiency	AR	*LPL*	99	1/500	1/50000	1/10000000000	1/200000
Methylmalonyl-CoA epimerase deficiency	AR	*MCEE*	99	3/500	1/16667	1/1111111111	1/66667
Methylmalonic aciduria, vitamin B12-responsive, cblA type	AR	*MMAA*	99	1/301	1/30100	1/3624040000	1/120400
Methylmalonic aciduria, vitamin B12-responsive, cblB type	AR	*MMAB*	99	1/435	1/43500	1/7569000000	1/174000
Methylmalonic aciduria and homocystinuria, cblF type	AR	*LMBRD1*	90	1/25	1/250	1/250000	1/1000
Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	XL	*HCFC1*	86	1/25	1/179	1/127551	1/714
Methylmalonic aciduria, mut(0) type	AR	*MMUT*	96	<9/1000	1/2778	1/30864198	1/11111
Methylmalonic aciduria and homocystinuria, cblC type	AR	*MMACHC*	99	1/134	1/13400	1/718240000	1/53600
Methylmalonic aciduria and homocystinuria, cblC type, digenic	AR	*PRDX1*	99	1/25	1/2500	1/25000000	1/10000
Methylmalonic aciduria and homocystinuria, cblD type	AR	*MMADHC*	83	<1/500	1/2941	1/34602076	1/11765
Argininosuccinic aciduria	AR	*ASL*	97	1/200	1/6667	1/177777778	1/26667
Argininemia	AR	*ARG1*	98	<4/1000	1/12500	1/625000000	1/50000
Lysinuric protein intolerance	AR	*SLC7A7*	99	<1/125	1/12500	1/625000000	1/50000
Tyrosinemia, type III	AR	*HPD*	99	<1/500	1/50000	1/10000000000	1/200000
Tyrosinemia, type II	AR	*TAT*	99	1/200	1/20000	1/1600000000	1/80000
Tyrosinemia, type I	AR	*FAH*	99	1/99	1/9900	1/392040000	1/39600

Lipoid adrenal hyperplasia	AR	*STAR*	99	<1/500	1/50000	1/10000000000	1/200000
Combined malonic and methylmalonic aciduria	AR	*ACSF3*	99	<1/500	1/50000	1/10000000000	1/200000
Combined oxidative phosphorylation deficiency 1	AR	*GFM1*	96	<1/500	1/12500	1/625000000	1/50000
Phosphoglycerate dehydrogenase deficiency	AR	*PHGDH*	99	<1/500	1/50000	1/10000000000	1/200000
Menkes disease	XL	*ATP7A*	93	<1/500	1/7143	1/204081633	1/28571
Molybdenum cofactor deficiency A	AR	*MOCS1*	88	<3/500	1/1389	1/7716049	1/5556
Cerebrotendinous xanthomatosis	AR	*CYP27A1*	99	1/500	1/50000	1/10000000000	1/200000
Niemann-Pick disease, type B	AR	*SMPD1*	98	<9/1000	1/5556	1/123456790	1/22222
Niemann-Pick disease, type C1	AR	*NPC1*	99	<3/500	1/16667	1/1111111111	1/66667
Niemann-pick disease, type C2	AR	*NPC2*	99	<3/500	1/16667	1/1111111111	1/66667
Mucopolysaccharidosis type IIIC (Sanfilippo C)	AR	*HGSNAT*	96	1/434	1/10850	1/470890000	1/43400
Mucopolysaccharidosis Ih	AR	*IDUA*	67	<1/500	1/1515	1/9182736	1/6061
Mucopolysaccharidosis type IIIA (Sanfilippo A)	AR	*SGSH*	98	1/454	1/22700	1/2061160000	1/90800
Mucopolysaccharidosis type IIIB (Sanfilippo B)	AR	*NAGLU*	89	<1/500	1/4545	1/82644628	1/18182
Mucopolysaccharidosis II	XL	*IDS*	87	<1/500	1/3846	1/59171598	1/15385
Mucopolysaccharidosis IVA	AR	*GALNS*	96	1/224	1/5600	1/125440000	1/22400
Mucopolysaccharidosis type IVB (Morquio)	AR	*GLB1*	99	1/134	1/13400	1/718240000	1/53600
Mucopolysaccharidosis type IX	AR	*HYAL1*	99	<1/500	1/50000	1/10000000000	1/200000
Mucopolysaccharidosis VII	AR	*GUSB*	95	1/250	1/5000	1/100000000	1/20000
Mucopolysaccharidosis type VI (Maroteaux-Lamy)	AR	*ARSB*	92	1/250	1/3125	1/39062500	1/12500
Mucolipidosis II alpha;;beta	AR	*GNPTAB*	98	<1/500	1/25000	1/2500000000	1/100000
Mucolipidosis IV	AR	*MCOLN1*	94	1/300	1/5000	1/100000000	1/20000
Ornithine transcarbamylase deficiency	XL	*OTC*	99	<1/500	1/50000	1/10000000000	1/200000
Hypoaldosteronism, congenital, due to CMO I deficiency	AR	*CYP11B2*	99	<1/500	1/50000	1/10000000000	1/200000
Holocarboxylase synthetase deficiency	AR	*HLCS*	99	1/500	1/50000	1/10000000000	1/200000
Carnitine-acylcarnitine translocase deficiency	AR	*SLC25A20*	98	<1/500	1/25000	1/2500000000	1/100000
CPT II deficiency, myopathic, stress-induced	AR/AD	*CPT2*	99	<1/500	1/50000	1/10000000000	1/200000
CPT deficiency, hepatic, type IA	AR	*CPT1A*	98	1/354	1/17700	1/1253160000	1/70800
Trifunctional protein deficiency	AR	*HADHB*	74	1/500	1/1923	1/14792899	1/7692
Biotinidase deficiency	AR	*BTD*	99	1/125	1/12500	1/625000000	1/50000
Glycogen storage disease II	AR	*GAA*	37	1/100	1/159	1/100781	1/635
Glycogen storage disease Ia	AR	*G6PC1*	99	1/177	1/17700	1/1253160000	1/70800
Glycogen storage disease Ib	AR	*SLC37A4*	97	1/158	1/5267	1/110951111	1/21067
Glycogen storage disease VII	AR	*PFKM*	99	<1/500	1/50000	1/10000000000	1/200000
McArdle disease	AR	*PYGM*	99	<1/500	1/50000	1/10000000000	1/200000
Aspartylglucosaminuria	AR	*AGA*	99	<1/500	1/50000	1/10000000000	1/200000
Asparagine synthetase deficiency	AR	*ASNS*	53	<1/500	1/1064	1/4526935	1/4255
Glutaricaciduria, type I	AR	*GCDH*	99	1/87	1/8700	1/302760000	1/34800
Glutaric acidemia IIA	AR	*ETFA*	99	1/500	1/50000	1/10000000000	1/200000
Glutaric acidemia IIB	AR	*ETFB*	99	1/500	1/50000	1/10000000000	1/200000
Glutaric acidemia IIC	AR	*ETFDH*	99	1/500	1/50000	1/10000000000	1/200000
Bile acid synthesis defect, congenital, 1	AR	*HSD3B7*	61	1/500	1/1282	1/6574622	1/5128
Bile acid synthesis defect, congenital, 2	AR	*AKR1D1*	99	<3/500	1/16667	1/1111111111	1/66667
Bile acid synthesis defect, congenital, 3	AR	*CYP7B1*	94	<1/200	1/3333	1/44444444	1/13333
Bile acid synthesis defect, congenital, 4	AR	*AMACR*	99	<1/200	1/20000	1/1600000000	1/80000
Bile acid synthesis defect, congenital, 5	AR	*ABCD3*	99	<1/200	1/20000	1/1600000000	1/80000
Bile acid synthesis defect, congenital, 6	AR	*ACOX2*	99	<1/200	1/20000	1/1600000000	1/80000
Hyperbilirubinemia, familial transient neonatal	AR	*UGT1A1*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type IIb	AR	*MOGS*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type IIc	AR	*SLC35C1*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type IId	AR	*B4GALT1*	99	<1/500	1/50000	1/10000000000	1/200000

Congenital disorder of glycosylation, type IIf	AR	*SLC35A1*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type Im	AR	*DOLK*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type Ia	AR	*PMM2*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type Ib	AR	*MPI*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type Ic	AR	*ALG6*	63	<1/500	1/1351	1/7304602	1/5405
Congenital disorder of glycosylation, type Id	AR	*ALG3*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type Ie	AR	*DPM1*	92	<1/500	1/6250	1/156250000	1/25000
Congenital disorder of glycosylation, type If	AR	*MPDU1*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type Ig	AR	*ALG12*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type Ih	AR	*ALG8*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type Ij	AR	*DPAGT1*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type Ik	AR	*ALG1*	25	<1/500	1/667	1/1777778	1/2667
Congenital disorder of glycosylation, type In	AR	*RFT1*	99	<1/500	1/50000	1/10000000000	1/200000
Congenital disorder of glycosylation, type Iq	AR	*SRD5A3*	99	<1/500	1/50000	1/10000000000	1/200000
Mitochondrial complex I deficiency, nuclear type 20	AR	*ACAD9*	99	<1/500	1/50000	1/10000000000	1/200000
Mitochondrial complex I deficiency, nuclear type 16	AR	*NDUFAF5*	67	1/447	1/1355	1/7339174	1/5418
Mitochondrial complex I deficiency, nuclear type 9	AR	*NDUFS6*	99	<1/500	1/50000	1/10000000000	1/200000
Mitochondrial trifunctional protein deficiency	AR	*HADHA*	93	3/200	1/952	1/3628118	1/3810
Adenosine deaminase deficiency, partial	AR	*ADA*	98	1/224	1/11200	1/501760000	1/44800
Fumarase deficiency	AR	*FH*	88	<1/500	1/4167	1/69444444	1/16667
Fructose intolerance, hereditary	AR	*ALDOB*	99	1/122	1/12200	1/595360000	1/48800
Ethylmalonic encephalopathy	AR	*ETHE1*	99	<1/500	1/50000	1/10000000000	1/200000
Isobutyryl-CoA dehydrogenase deficiency	AR	*ACAD8*	99	1/125	1/12500	1/625000000	1/50000
Isovaleric acidemia	AR	*IVD*	99	1/167	1/16700	1/1115560000	1/66800
Hyperoxaluria, primary, type II	AR	*GRHPR*	95	<1/500	1/10000	1/400000000	1/40000
Hyperoxaluria, primary, type III	AR	*HOGA1*	99	1/184	1/18400	1/1354240000	1/73600
Hyperoxaluria, primary, type 1	AR	*AGXT*	99	1/120	1/12000	1/576000000	1/48000
Carnitine deficiency, systemic primary	AR	*SLC22A5*	99	1/129	1/12900	1/665640000	1/51600
Mucolipidosis III gamma	AR	*GNPTG*	94	<1/500	1/8333	1/277777778	1/33333
Mucopolysaccharidosis type IIID	AR	*GNS*	93	1/500	1/7143	1/204081633	1/28571
Acyl-CoA dehydrogenase, medium chain, deficiency of	AR	*ACADM*	99	<3/500	1/16667	1/1111111111	1/66667
Acyl-CoA dehydrogenase, short-chain, deficiency of	AR	*ACADS*	99	1/85	1/8500	1/289000000	1/34000

Musculoskeletal System

Achondrogenesis Ib	AR	*SLC26A2*	99	1/158	1/15800	1/998560000	1/63200
Myasthenic syndrome, congenital	AR	*CHRNE*	99	1/408	1/40800	1/6658560000	1/163200
Emery-Dreifuss muscular dystrophy 1, X-linked	XL	*EMD*	87	<1/500	1/3846	1/59171598	1/15385
Emery-Dreifuss muscular dystrophy 3, autosomal recessive	AR	*LMNA*	93	<1/500	1/7143	1/204081633	1/28571
Emery-Dreifuss muscular dystrophy 6, X-linked	XL	*FHL1*	99	<1/500	1/50000	1/10000000000	1/200000
Muscular dystrophy-dystroglycanopathy	AR	*FKTN*	99	<1/500	1/50000	1/10000000000	1/200000
Muscular dystrophy, limb-girdle, autosomal recessive 23	AR	*LAMA2*	99	<1/500	1/50000	1/10000000000	1/200000
Schimke immunoosseous dysplasia	AR	*SMARCAL1*	99	1/500	1/50000	1/10000000000	1/200000
Nonaka myopathy	AR	*GNE*	99	<1/500	1/50000	1/10000000000	1/200000
Osteopetrosis, autosomal recessive 1	AR	*TCIRG1*	80	1/250	1/1250	1/6250000	1/5000
Osteopetrosis, autosomal recessive 4	AR	*CLCN7*	93	1/250	1/3571	1/51020408	1/14286
Duchenne muscular dystrophy	XL	*DMD*	80	<1/500	1/2500	1/25000000	1/10000
Nemaline myopathy 2, autosomal recessive	AR	*NEB*	94	1/112	1/1867	1/13937778	1/7467
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6	AR	*LARGE1*	99	1/125	1/12500	1/625000000	1/50000
Spinal muscular atrophy-2	AR	*SMN1*	10	1/50	1/56	1/12346	1/222
Spinal and bulbar muscular atrophy of Kennedy	XL	*AR*	54	<1/500	1/1087	1/4725898	1/4348
Spondylocostal dysostosis 2, autosomal recessive	AR	*MESP2*	99	<1/500	1/50000	1/10000000000	1/200000
Carpenter syndrome	AR	*RAB23*	95	<1/500	1/10000	1/400000000	1/40000

Congenital arthrogryposis with anterior horn cell disease	AR	*GLE1*	99	<1/500	1/50000	1/10000000000	1/200000
Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	AR	*RAPSN*	99	<1/500	1/50000	1/10000000000	1/200000
Myasthenic syndrome, congenital, 10	AR	*DOK7*	63	1/163	1/441	1/776304	1/1762
Myasthenic syndrome, congenital, 1B, fast-channel	AR/AD	*CHRNA1*	99	1/577	1/57700	1/13317160000	1/230800
Myasthenic syndrome, congenital, 5	AR	*COLQ*	99	1/163	1/16300	1/1062760000	1/65200
Myasthenic syndrome, congenital, 6, presynaptic	AR	*CHAT*	99	1/272	1/27200	1/2959360000	1/108800
Charcot-Marie-Tooth disease, type 4D	AR	*NDRG1*	99	1/22	1/2200	1/19360000	1/8800
Muscular dystrophy, limb-girdle, autosomal recessive 8	AR	*TRIM32*	99	<1/500	1/50000	1/10000000000	1/200000
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1	AR	*POMT1*	99	1/843	1/84300	1/28425960000	1/337200
Muscular dystrophy, limb-girdle, autosomal recessive 12	AR	*ANO5*	97	<1/500	1/16667	1/1111111111	1/66667
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2	AR	*POMT2*	99	<1/500	1/50000	1/10000000000	1/200000
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9	AR	*DAG1*	99	<1/500	1/50000	1/10000000000	1/200000
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14	AR	*GMPPB*	99	<1/500	1/50000	1/10000000000	1/200000
Muscular dystrophy, limb-girdle, autosomal recessive 1	AR	*CAPN3*	99	<1/500	1/50000	1/10000000000	1/200000
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3	AR	*POMGNT1*	99	1/462	1/46200	1/8537760000	1/184800
Muscular dystrophy, limb-girdle, autosomal recessive 2	AR	*DYSF*	99	<1/500	1/50000	1/10000000000	1/200000
Muscular dystrophy, limb-girdle, autosomal recessive 3	AR	*SGCA*	98	<1/500	1/25000	1/2500000000	1/100000
Muscular dystrophy, limb-girdle, autosomal recessive 4	AR	*SGCB*	78	1/500	1/2273	1/20661157	1/9091
Muscular dystrophy, limb-girdle, autosomal recessive 5	AR	*SGCG*	99	1/381	1/38100	1/5806440000	1/152400
Muscular dystrophy, limb-girdle, autosomal recessive 6	AR	*SGCD*	74	<1/500	1/1923	1/14792899	1/7692
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	AR	*FKRP*	95	<1/500	1/10000	1/400000000	1/40000
Rhizomelic chondrodysplasia punctata, type 1	AR	*PEX7*	66	1/158	1/465	1/863806	1/1859
Rhizomelic chondrodysplasia punctata, type 3	AR	*AGPS*	99	<1/500	1/50000	1/10000000000	1/200000
Chondrodysplasia punctata, X-linked recessive	XL	*ARSL*	99	<1/500	1/50000	1/10000000000	1/200000
Myotubular myopathy, X-linked	XL	*MTM1*	83	<1/500	1/2941	1/34602076	1/11765
Pycnodysostosis	AR	*CTSK*	99	<1/500	1/50000	1/10000000000	1/200000
Respiratory System							
Ciliary dyskinesia, primary, 1, with or without situs inversus	AR	*DNAI1*	99	1/230	1/23000	1/2116000000	1/92000
Ciliary dyskinesia, primary, 9, with or without situs inversus	AR	*DNAI2*	99	1/447	1/44700	1/7992360000	1/178800
Ciliary dyskinesia, primary, 16	AR	*DNAL1*	99	<1/500	1/50000	1/10000000000	1/200000
Ciliary dyskinesia, primary, 3, with or without situs inversus	AR	*DNAH5*	99	1/142	1/14200	1/806560000	1/56800
Ciliary dyskinesia, primary, 17	AR	*DNAAF19*	99	1/316	1/31600	1/3994240000	1/126400
Ciliary dyskinesia, primary, 30	AR	*ODAD3*	99	1/365	1/36500	1/5329000000	1/146000
Ciliary dyskinesia, primary, 14	AR	*CCDC39*	62	1/211	1/555	1/1233269	1/2221
Urinary System							
Alkaptonuria	AR	*HGD*	99	<1/500	1/50000	1/10000000000	1/200000
Polycystic kidney disease 5	AR	*DZIP1L*	99	<1/100	1/10000	1/400000000	1/40000
Polycystic kidney disease 4, with or without hepatic disease	AR	*PKHD1*	99	<1/100	1/10000	1/400000000	1/40000
Nephrotic syndrome, type 1	AR	*NPHS1*	98	1/289	1/14450	1/835210000	1/57800
Nephrotic syndrome, type 2	AR	*NPHS2*	99	3/500	1/16667	1/1111111111	1/66667
Nephrotic syndrome, type 3	AR	*PLCE1*	99	3/500	1/16667	1/1111111111	1/66667
Nephrotic syndrome, type 6	AR	*PTPRO*	99	3/500	1/16667	1/1111111111	1/66667
Alport syndrome 2, autosomal recessive	AR	*COL4A3*	99	1/267	1/26700	1/2851560000	1/106800
Alport syndrome 2, autosomal recessive	AR	*COL4A4*	64	1/267	1/742	1/2200278	1/2967
Dent disease 2	XL	*OCRL*	95	<1/500	1/10000	1/400000000	1/40000
Diabetes insipidus, nephrogenic	AR/AD	*AQP2*	89	<1/500	1/4545	1/82644628	1/18182
Nephronophthisis 3	AR	*NPHP3*	71	<3/1000	1/1149	1/5284714	1/4598
Immune System							

Chronic granulomatous disease 2, autosomal recessive	AR	*NCF2*	99	1/224	1/22400	1/2007040000	1/89600
Chronic granulomatous disease 4, autosomal recessive	AR	*CYBA*	89	1/224	1/2036	1/16587107	1/8145
Chronic granulomatous disease 5, autosomal recessive	AR	*CYBC1*	99	1/224	1/22400	1/2007040000	1/89600
Chronic granulomatous disease, X-linked	XL	*CYBB*	84	<1/500	1/3125	1/39062500	1/12500
Familial Mediterranean fever, AR	AR	*MEFV*	99	1/20	1/2000	1/16000000	1/8000
Wiskott-Aldrich syndrome	XL	*WAS*	27	<1/500	1/685	1/1876525	1/2740
Agammaglobulinemia, X-linked 1	XL	*BTK*	99	<1/500	1/50000	1/10000000000	1/200000
Lymphoproliferative syndrome, X-linked, 2	XL	*XIAP*	80	<1/500	1/2500	1/25000000	1/10000
Lymphoproliferative syndrome, X-linked, 1	XL	*SH2D1A*	93	<1/500	1/7143	1/204081633	1/28571
Combined cellular and humoral immune defects with granulomas	AR	*RAG1*	99	1/137	1/13700	1/750760000	1/54800
Combined cellular and humoral immune defects with granulomas	AR	*RAG2*	99	1/137	1/13700	1/750760000	1/54800
Severe combined immunodeficiency, X-linked	XL	*IL2RG*	99	<1/500	1/50000	1/10000000000	1/200000
Severe combined immunodeficiency, Athabascan type	AR	*DCLRE1C*	99	<1/500	1/50000	1/10000000000	1/200000
Immunodeficiency, X-linked, with hyper-IgM	XL	*CD40LG*	97	<1/500	1/16667	1/1111111111	1/66667
Bare lymphocyte syndrome, type II, complementation group A	AR	*CIITA*	99	<1/500	1/50000	1/10000000000	1/200000
Hemophagocytic lymphohistiocytosis, familial, 2	AR	*PRF1*	99	3/500	1/16667	1/1111111111	1/66667
Hemophagocytic lymphohistiocytosis, familial, 3	AR	*UNC13D*	99	3/500	1/16667	1/1111111111	1/66667
Hemophagocytic lymphohistiocytosis, familial, 4	AR	*STX11*	99	3/500	1/16667	1/1111111111	1/66667
Hemophagocytic lymphohistiocytosis, familial, 5	AR	*STXBP2*	99	3/500	1/16667	1/1111111111	1/66667
Endocrine System							
Hyperinsulinemic hypoglycemia, familial, 1	AR/AD	*ABCC8*	99	1/112	1/11200	1/501760000	1/44800
Hyperinsulinemic hypoglycemia, familial, 2	AR/AD	*KCNJ11*	99	1/423	1/42300	1/7157160000	1/169200
Hyperinsulinemic hypoglycemia, familial, 4	AR	*HADH*	92	<1/500	1/6250	1/156250000	1/25000
Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	AR/AD	*AIRE*	99	1/150	1/15000	1/900000000	1/60000
Pituitary hormone deficiency, combined, 2	AR	*PROP1*	99	1/45	1/4500	1/81000000	1/18000
Hypothyroidism, congenital, nongoitrous, 1	AR	*TSHR*	99	1/50	1/5000	1/100000000	1/20000
Hypothyroidism, congenital, nongoitrous 4	AR	*TSHB*	99	1/50	1/5000	1/100000000	1/20000
Pituitary hormone deficiency, combined, 3	AR	*LHX3*	99	1/45	1/4500	1/81000000	1/18000
Pituitary hormone deficiency, combined, 1	AR/AD	*POU1F1*	94	1/267	1/4450	1/79210000	1/17800
Hypercholesterolemia, familial, 4	AR	*LDLRAP1*	99	1/8	1/800	1/2560000	1/3200
Integumentary System							
Ehlers-Danlos syndrome, dermatosparaxis type	AR	*ADAMTS2*	90	<1/500	1/5000	1/100000000	1/20000
Ocular albinism, type I, Nettleship-Falls type	XL	*GPR143*	83	1/25000	1/147059	1/86505190311	1/588235
Acrodermatitis enteropathica	AR	*SLC39A4*	99	<1/500	1/50000	1/10000000000	1/200000
Ichthyosis, congenital, autosomal recessive 4A	AR	*ABCA12*	99	1/284	1/28400	1/3226240000	1/113600
Epidermolysis bullosa, junctional, localisata variant	AR	*COL17A1*	96	<1/2000	1/50000	1/10000000000	1/200000
Ataxia-telangiectasia	AR	*ATM*	89	1/100	1/909	1/3305785	1/3636
Epidermolysis bullosa	AR	*LAMA3*	99	1/781	1/78100	1/24398440000	1/312400
Epidermolysis bullosa, junctional, Herlitz type	AR	*LAMB3*	99	1/781	1/78100	1/24398440000	1/312400
Epidermolysis bullosa, junctional, Herlitz type	AR	*LAMC2*	99	1/781	1/78100	1/24398440000	1/312400
Epidermolysis bullosa dystrophica	AR	*COL7A1*	99	1/196	1/19600	1/1536640000	1/78400
Dyskeratosis congenita, autosomal recessive 5	AR/AD	*RTEL1*	98	1/500	1/25000	1/2500000000	1/100000
Ichthyosis, congenital, autosomal recessive 1	AR	*TGM1*	99	1/224	1/22400	1/2007040000	1/89600
Albinism, oculocutaneous, type IB	AR	*TYR*	99	3/200	1/6667	1/177777778	1/26667
Albinism, oculocutaneous, type II, modifier of	AR	*MC1R*	99	3/200	1/6667	1/177777778	1/26667
Albinism, oculocutaneous, type III	AR	*TYRP1*	99	3/200	1/6667	1/177777778	1/26667
Albinism, oculocutaneous, type IV	AR	*SLC45A2*	99	3/200	1/6667	1/177777778	1/26667
Skin;;hair;;eye pigmentation 4, fair	AR	*SLC24A5*	38	3/200	1/108	1/46248	1/430
Albinism, oculocutaneous, type VII	AR	*LRMDA*	99	3/200	1/6667	1/177777778	1/26667

Oculocutaneous albinism, type VIII	AR	*DCT*	99	3/200	1/6667	1/177777778	1/26667
Xeroderma pigmentosum, group A	AR	*XPA*	69	1/500	1/1613	1/10405827	1/6452
Xeroderma pigmentosum, group C	AR	*XPC*	99	1/500	1/50000	1/10000000000	1/200000
Albinism, brown oculocutaneous	AR	*OCA2*	99	3/200	1/6667	1/177777778	1/26667
Nervous System							
Salt and pepper developmental regression syndrome	AR	*ST3GAL5*	92	9/50	1/69	1/19290	1/278
Ceroid lipofuscinosis, neuronal, 3	AR	*CLN3*	93	1/230	1/3286	1/43183673	1/13143
CRASH syndrome Corpus callosum, partial agenesis of;;CRASH syndrome;;Hydrocephalus due to aqueductal stenosis;;Hydrocephalus with congenital idiopathic intestinal pseudoobstruction;;Hydrocephalus with Hirschsprung disease;;MASA syndrome	XL	*L1CAM*	99	<1/500	1/50000	1/10000000000	1/200000
Segawa syndrome, recessive	AR	*TH*	97	1/224	1/7467	1/223004444	1/29867
Tay-Sachs disease	AR	*HEXA*	99	1/300	1/30000	1/3600000000	1/120000
Lissencephaly, X-linked	XL	*DCX*	99	<1/500	1/50000	1/10000000000	1/200000
Leukoencephalopathy with vanishing white matter	AR	*EIF2B5*	99	<1/500	1/50000	1/10000000000	1/200000
Spastic ataxia, Charlevoix-Saguenay type	AR	*SACS*	99	<1/500	1/50000	1/10000000000	1/200000
Ataxia with isolated vitamin E deficiency	AR	*TTPA*	83	<1/500	1/2941	1/34602076	1/11765
Arthrogryposis, mental retardation, and seizures	AR	*SLC35A3*	41	<1/500	1/847	1/2872738	1/3390
Dysautonomia, familial	AR	*ELP1*	99	1/300	1/30000	1/3600000000	1/120000
Microcephaly, postnatal progressive, with seizures and brain atrophy	AR	*MED17*	99	<1/500	1/50000	1/10000000000	1/200000
Spastic paraplegia 15, autosomal recessive	AR	*ZFYVE26*	99	<1/500	1/50000	1/10000000000	1/200000
Spastic paraplegia 49, autosomal recessive	AR	*TECPR2*	99	<1/500	1/50000	1/10000000000	1/200000
Megalencephalic leukoencephalopathy with subcortical cysts	AR	*MLC1*	99	<1/500	1/50000	1/10000000000	1/200000
Canavan disease	AR	*ASPA*	99	1/57	1/5700	1/129960000	1/22800
Hydrolethalus syndrome	AR	*HYLS1*	99	7/500	1/7143	1/204081633	1/28571
Pontocerebellar hypoplasia type 1A	AR	*VRK1*	99	<1/500	1/50000	1/10000000000	1/200000
Pontocerebellar hypoplasia, type 1B	AR	*EXOSC3*	77	<1/500	1/2174	1/18903592	1/8696
Pontocerebellar hypoplasia type 2D	AR	*SEPSECS*	89	<1/500	1/4545	1/82644628	1/18182
Pontocerebellar hypoplasia, type 6	AR	*RARS2*	97	<1/500	1/16667	1/1111111111	1/66667
Agenesis of the corpus callosum with peripheral neuropathy	AR	*SLC12A6*	99	<1/500	1/50000	1/10000000000	1/200000
Sandhoff disease, infantile, juvenile, and adult forms	AR	*HEXB*	97	1/600	1/20000	1/1600000000	1/80000
Ceroid lipofuscinosis, neuronal, 10	AR	*CTSD*	99	1/1761	1/176100	1/124044840000	1/704400
Ceroid lipofuscinosis, neuronal, 1	AR	*PPT1*	99	1/368	1/36800	1/5416960000	1/147200
Ceroid lipofuscinosis, neuronal, 2	AR	*TPP1*	99	1/252	1/25200	1/2540160000	1/100800
Ceroid lipofuscinosis, neuronal, 5	AR	*CLN5*	66	<1/500	1/1471	1/8650519	1/5882
Ceroid lipofuscinosis, neuronal, 6	AR	*CLN6*	86	<1/500	1/3571	1/51020408	1/14286
Ceroid lipofuscinosis, neuronal, 7	AR	*MFSD8*	95	<1/500	1/10000	1/400000000	1/40000
Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant	AR	*CLN8*	99	<1/500	1/50000	1/10000000000	1/200000
Adrenoleukodystrophy	XL	*ABCD1*	86	1/100	1/714	1/2040816	1/2857
Polymicrogyria, bilateral frontoparietal	AR	*ADGRG1*	98	<1/500	1/25000	1/2500000000	1/100000
Choreoacanthocytosis	AR	*VPS13A*	80	<1/500	1/2500	1/25000000	1/10000
Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)	AR	*LRPPRC*	91	1/447	1/4967	1/98671111	1/19867
Metachromatic leukodystrophy	AR	*ARSA*	99	1/100	1/10000	1/400000000	1/40000
Infantile neuroaxonal dystrophy 1	AR	*PLA2G6*	97	1/500	1/16667	1/1111111111	1/66667
Krabbe disease	AR	*GALC*	97	1/158	1/5267	1/110951111	1/21067
Metachromatic leukodystrophy due to SAP-b deficiency	AR	*PSAP*	99	<1/500	1/50000	1/10000000000	1/200000
Visual System							
Achromatopsia 3	AR	*CNGB3*	99	1/87	1/8700	1/302760000	1/34800
Progressive external ophthalmoplegia, autosomal recessive 1;;Mitochondrial DNA depletion syndrome 4	AR	*POLG*	99	1/113	1/11300	1/510760000	1/45200
Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset	AR	*CYP1B1*	99	1/50	1/5000	1/100000000	1/20000

Retinitis pigmentosa 2	XL	*RP2*	99	3/100	1/3333	1/44444444	1/13333
Retinitis pigmentosa 3	XL	*RPGR*	22	1/259	1/332	1/441032	1/1328
Retinitis pigmentosa-12	AR	*CRB1*	99	1/104	1/10400	1/432640000	1/41600
Retinitis pigmentosa 19	AR	*ABCA4*	99	3/100	1/3333	1/44444444	1/13333
Retinitis pigmentosa 20	AR	*RPE65*	84	1/228	1/1425	1/8122500	1/5700
Retinitis pigmentosa 25	AR	*EYS*	73	1/66	1/244	1/239012	1/978
Retinitis pigmentosa 26	AR	*CERKL*	97	1/148	1/4933	1/97351111	1/19733
Retinitis pigmentosa 28	AR	*FAM161A*	99	1/296	1/29600	1/3504640000	1/118400
Retinitis pigmentosa-40	AR	*PDE6B*	99	3/100	1/3333	1/44444444	1/13333
Retinitis pigmentosa 43	AR	*PDE6A*	99	3/100	1/3333	1/44444444	1/13333
Retinitis pigmentosa 47	AR	*SAG*	99	3/100	1/3333	1/44444444	1/13333
Retinitis pigmentosa 59	AR	*DHDDS*	99	1/296	1/29600	1/3504640000	1/118400
Microphthalmia with coloboma 3	AR	*VSX2*	99	1/91	1/9100	1/331240000	1/36400
Corneal endothelial dystrophy, autosomal recessive	AR	*SLC4A11*	99	<1/500	1/50000	1/10000000000	1/200000
Retinoschisis	XL	*RS1*	85	<1/500	1/3333	1/44444444	1/13333
Leber congenital amaurosis 13	AR/AD	*RDH12*	60	<1/500	1/1250	1/6250000	1/5000
Gyrate atrophy of choroid and retina with or without ornithinemia	AR	*OAT*	87	<1/500	1/3846	1/59171598	1/15385
Retinitis pigmentosa 37	AR/AD	*NR2E3*	99	1/209	1/20900	1/1747240000	1/83600
Leber congenital amaurosis 5	AR	*LCA5*	93	1/500	1/7143	1/204081633	1/28571
Auditory System							
Deafness, autosomal recessive 2	AR	*MYO7A*	99	1/206	1/20600	1/1697440000	1/82400
Deafness, autosomal recessive 1A	AR	*GJB2*	99	1/80	1/8000	1/256000000	1/32000
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	AR	*SLC26A4*	99	1/80	1/8000	1/256000000	1/32000
Deafness, autosomal recessive 77	AR	*LOXHD1*	99	1/500	1/50000	1/10000000000	1/200000
Deafness, autosomal recessive 1B	AR	*GJB6*	99	1/423	1/42300	1/7157160000	1/169200
Digestive System							
Cholestasis, progressive familial intrahepatic 2	AR	*ABCB11*	98	<1/500	1/25000	1/2500000000	1/100000
Cholestasis, progressive familial intrahepatic 3	AR/AD	*ABCB4*	99	<1/500	1/50000	1/10000000000	1/200000
Cholestasis, progressive familial intrahepatic 4	AR	*TJP2*	99	<1/502	1/50200	1/10080160000	1/200800
Cholestasis, progressive familial intrahepatic, 5	AR	*NR1H4*	99	<1/500	1/50000	1/10000000000	1/200000
Cholestasis, progressive familial intrahepatic 1	AR	*ATP8B1*	99	<1/500	1/50000	1/10000000000	1/200000
Cystic fibrosis	AR	*CFTR*	95	<1/20	1/400	1/640000	1/1600
Liver failure, transient infantile	AR	*TRMU*	99	<1/500	1/50000	1/10000000000	1/200000
Diarrhea 1, secretory chloride, congenital	AR	*SLC26A3*	99	<1/500	1/50000	1/10000000000	1/200000
Hematologic System							
Factor XI deficiency, autosomal recessive	AR/AD	*F11*	99	1/500	1/50000	1/10000000000	1/200000
Hemophilia A	XL	*F8*	98	<1/500	1/25000	1/2500000000	1/100000
Hemophilia B	XL	*F9*	94	<1/500	1/8333	1/277777778	1/33333
Thalassemia, alpha-	AR	*HBA1/HBA2*	33/31	1/20	1/30；1/29	1/3564；1/3361	1/119；1/116
Neutropenia, severe congenital 4, autosomal recessive	AR	*G6PC3*	99	1/341	1/34100	1/4651240000	1/136400
Fanconi anemia, complementation group A	AR	*FANCA*	93	1/239	1/3414	1/46629388	1/13657
Fanconi anemia, complementation group C	AR	*FANCC*	99	1/535	1/53500	1/11449000000	1/214000
Fanconi anemia, complementation group D2	AR	*FANCD2*	95	<1/200	1/4000	1/64000000	1/16000
Fanconi anemia, complementation group G	AR	*FANCG*	99	1/239	1/23900	1/2284840000	1/95600
Fanconi anemia, complementation group I	AR	*FANCI*	97	<1/200	1/6667	1/177777778	1/26667
Bernard-Soulier syndrome, type A1 (recessive)	AR	*GP1BA*	99	1/500	1/50000	1/10000000000	1/200000
Bernard-Soulier syndrome, type C	AR	*GP9*	99	1/500	1/50000	1/10000000000	1/200000
Thalassemia, beta	AR	*HBB*	99	1/158	1/15800	1/998560000	1/63200
Hypoprothrombinemia	AR	*F2*	99	1/33	1/3300	1/43560000	1/13200
Factor V deficiency	AR	*F5*	99	1/36	1/3600	1/51840000	1/14400
Abetalipoproteinemia	AR	*MTTP*	99	<1/500	1/50000	1/10000000000	1/200000

Thrombocytopenia, congenital amegakaryocytic	AR	*MPL*	99	1/102	1/10200	1/416160000	1/40800
Myopathy, lactic acidosis, and sideroblastic anemia 1	AR	*PUS1*	93	<1/500	1/7143	1/204081633	1/28571
Neutropenia, severe congenital 3, autosomal recessive	AR	*HAX1*	99	1/224	1/22400	1/2007040000	1/89600
Neutropenia, severe congenital, 5, autosomal recessive	AR	*VPS45*	99	1/224	1/22400	1/2007040000	1/89600
Multiple Systems							
Usher syndrome, type 1B	AR	*MYO7A*	99	1/206	1/20600	1/1697440000	1/82400
Usher syndrome, type 1C	AR	*USH1C*	99	1/353	1/35300	1/4984360000	1/141200
Usher syndrome, type 1D	AR	*CDH23*	99	1/285	1/28500	1/3249000000	1/114000
Usher syndrome, type 1F	AR	*PCDH15*	97	1/395	1/13167	1/693444444	1/52667
Usher syndrome, type 2A	AR	*USH2A*	99	1/126	1/12600	1/635040000	1/50400
Usher syndrome, type 2C	AR	*ADGRV1*	99	3/250	1/8333	1/277777778	1/33333
Usher syndrome, type 3A	AR	*CLRN1*	99	1/500	1/50000	1/10000000000	1/200000
Aicardi-Goutieres syndrome 5	AR	*SAMHD1*	95	<1/500	1/10000	1/400000000	1/40000
Arts syndrome	XL	*PRPS1*	99	<1/500	1/50000	1/10000000000	1/200000
Bardet-Biedl syndrome 10	AR	*BBS10*	99	1/395	1/39500	1/6241000000	1/158000
Bardet-Biedl syndrome 12	AR	*BBS12*	99	1/791	1/79100	1/25027240000	1/316400
Bardet-Biedl syndrome 14	AR	*CEP290*	25	1/190	1/629	1/1584242	1/2517
Bardet-Biedl syndrome 1	AR	*BBS1*	99	1/367	1/36700	1/5387560000	1/146800
Bardet-Biedl syndrome 2	AR	*BBS2*	97	1/621	1/20700	1/1713960000	1/82800
Bardet-Biedl syndrome 3	AR	*ARL6*	93	<1/500	1/7143	1/204081633	1/28571
Bardet-Biedl syndrome 4	AR	*BBS4*	91	<1/500	1/5556	1/123456790	1/22222
Bardet-Biedl syndrome 6	AR	*MKKS*	99	<1/500	1/50000	1/10000000000	1/200000
Barth syndrome	XL	*TAFAZZIN*	99	<3/1000	1/33333	1/4444444444	1/133333
Cohen syndrome	AR	*VPS13B*	96	<1/500	1/12500	1/625000000	1/50000
Ehlers-Danlos syndrome, kyphoscoliotic type, 1	AR	*PLOD1*	99	1/158	1/15800	1/998560000	1/63200
Ellis-van Creveld syndrome	AR	*EVC2*	98	1/240	1/12000	1/576000000	1/48000
Ellis-van Creveld syndrome	AR	*EVC*	93	1/142	1/2029	1/16460408	1/8114
Gitelman syndrome	AR	*SLC12A3*	99	1/100	1/10000	1/400000000	1/40000
Hermansky-Pudlak syndrome 1	AR	*HPS1*	91	<1/500	1/5556	1/123456790	1/22222
Hermansky-Pudlak syndrome 3	AR	*HPS3*	94	<1/500	1/8333	1/277777778	1/33333
Hermansky-Pudlak syndrome 4	AR	*HPS4*	99	<1/500	1/50000	1/10000000000	1/200000
Hermansky-Pudlak syndrome 5	AR	*HPS5*	99	<1/500	1/50000	1/10000000000	1/200000
Hermansky-Pudlak syndrome 6	AR	*HPS6*	85	<1/500	1/3333	1/44444444	1/13333
Joubert syndrome 17	AR	*CPLANE1*	81	1/472	1/2484	1/24685208	1/9937
Joubert syndrome 2	AR	*TMEM216*	99	1/472	1/47200	1/8911360000	1/188800
Joubert syndrome 3	AR	*AHI1*	57	1/472	1/1098	1/4819557	1/4391
Joubert syndrome 5	AR	*CEP290*	25	1/472	1/629	1/1584242	1/2517
Joubert syndrome 6	AR	*TMEM67*	70	1/472	1/1573	1/9901511	1/6293
Joubert syndrome 9	AR	*CC2D2A*	95	1/472	1/9440	1/356454400	1/37760
Laron dwarfism	AR	*GHR*	99	1/158	1/15800	1/998560000	1/63200
Meckel syndrome 1	AR	*MKS1*	99	1/260	1/26000	1/2704000000	1/104000
Netherton syndrome	AR	*SPINK5*	99	<2/100	1/5000	1/100000000	1/20000
Pendred syndrome	AR	*SLC26A4*	99	1/80	1/8000	1/256000000	1/32000
Shwachman-Diamond syndrome	AR	*SBDS*	99	1/146	1/14600	1/852640000	1/58400
Sjogren-Larsson syndrome	AR	*ALDH3A2*	99	1/250	1/25000	1/2500000000	1/100000
Smith-Lemli-Opitz syndrome	AR	*DHCR7*	99	1/30	1/3000	1/36000000	1/12000
Stuve-Wiedemann syndrome	AR	*LIFR*	80	<1/500	1/2500	1/25000000	1/10000
Usher syndrome, type 1G	AR	*USH1G*	99	1/600	1/60000	1/14400000000	1/240000
Wolcott-Rallison syndrome	AR	*EIF2AK3*	90	<1/500	1/5000	1/100000000	1/20000
Wolfram syndrome 1	AR	*WFS1*	99	<1/200	1/20000	1/1600000000	1/80000
Mental retardation-hypotonic facies syndrome, X-linked	XL	*ATRX*	58	<1/500	1/1190	1/5668934	1/4762

Bartter syndrome, type 4a	AR	*BSND*	99	1/500	1/50000	1/10000000000	1/200000
Mitochondrial complex III deficiency, nuclear type 1	AR	*BCS1L*	99	<1/500	1/50000	1/10000000000	1/200000
Bloom syndrome	AR	*BLM*	77	1/800	1/3478	1/48393195	1/13913
Multiple pterygium syndrome, lethal type	AR	*CHRNG*	99	<1/500	1/50000	1/10000000000	1/200000
Trichohepatoenteric syndrome 1	AR	*SKIC3*	99	1/500	1/50000	1/10000000000	1/200000
Osteoporosis-pseudoglioma syndrome	AR	*LRP5*	99	7/5000	1/71429	1/20408163265	1/285714
Mulibrey nanism	AR	*TRIM37*	80	1/100	1/500	1/1000000	1/2000
Cockayne syndrome, type A	AR	*ERCC8*	93	1/822	1/11743	1/551578776	1/46971
Cockayne syndrome, type B	AR	*ERCC6*	98	<1/500	1/25000	1/2500000000	1/100000
Roberts-SC phocomelia syndrome	AR	*ESCO2*	99	<1/500	1/50000	1/10000000000	1/200000
Joubert syndrome 7	AR	*RPGRIP1L*	87	1/259	1/1992	1/15877160	1/7969
Immunodeficiency-centromeric instability-facial anomalies syndrome 1	AR	*DNMT3B*	99	<1/500	1/50000	1/10000000000	1/200000
Nijmegen breakage syndrome	AR	*NBN*	86	1/158	1/1129	1/5094694	1/4514
Salla disease	AR	*SLC17A5*	95	<1/500	1/10000	1/400000000	1/40000
Ectodermal dysplasia 1, hypohidrotic, X-linked	XL	*EDA*	88	<1/500	1/4167	1/69444444	1/16667
Distal renal tubular acidosis 2 with progressive sensorineural hearing loss	AR	*ATP6V1B1*	99	<1/500	1/50000	1/10000000000	1/200000
Alstrom syndrome	AR	*ALMS1*	99	<1/500	1/50000	1/10000000000	1/200000
Steel syndrome	AR	*COL27A1*	99	<1/500	1/50000	1/10000000000	1/200000
Insensitivity to pain, congenital, with anhidrosis	AR	*NTRK1*	36	<1/500	1/781	1/2441406	1/3125
Mitochondrial DNA depletion syndrome 1 (MNGIE type)	AR	*TYMP*	99	<1/500	1/50000	1/10000000000	1/200000
Chediak-Higashi syndrome	AR	*LYST*	84	<1/500	1/3125	1/39062500	1/12500
Ehlers-Danlos syndrome, cardiac valvular type	AR	*COL1A2*	89	1/500	1/4545	1/82644628	1/18182
Schopf-Schulz-Passarge syndrome	AR	*WNT10A*	99	<1/500	1/50000	1/10000000000	1/200000
Joubert syndrome 4	AR	*NPHP1*	97	1/480	1/16000	1/1024000000	1/64000
Fragile X syndrome	XL	*FMR1*	99	1/250	1/25000	1/1250000000	1/33333