CONDITION NAME	GENE (and Refseq)	CARRIER RATES
3-METHYLCROTONYL-CoA CARBOXYLASE 1 DEFICIENCY	MCCC1 (NM_ 020166.4)	1 in 137
3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY	MCCC2 (NM_ 022132.4)	1 in 112
ACHROMATOPSIA	CNGB3 (NM_ 019098.4)	1 in 91
ALPHA THALASSEMIA	HBA1/HBA2 (NM_ 000553.4; NM_ 000517.4)	1 in 25
ATAXIA-TELANGIECTASIA	ATM (NM_ 000051.3)	1 in 100
BATTEN DISEASE (NEURONAL CEROID LIPOFUSCINOSIS, CLN3-RELATED)	CLN3 (NM_ 000086.2	1 in 138
BETA-HEMOGLOBINOPATHIES	HBB (NM_ 000518.4)	2-5%
BIOTINIDASE DEFICIENCY	BTD (NM_ 000060.3)	1 in 12
BLOOM SYNDROME	BLM (NM_ 000057.3)	1 in 134
CANAVAN DISEASE	ASPA (NM_000049.2)	1 in 158
CEREBROTENDINOUS XANTHOMATOSIS	CYP27A1 (NM_ 000784.3)	1 in 112
CITRULLINEMIA, TYPE I	ASS1 (NM_ 000050.4)	1 in 195
COMBINED MALONIC AND METHYLMALONI ACIDURIA	ACSF3 (NM_001127214.3)	1 in 86
COMBINED PITUITARY HORMONE DEFICIENCY-2	PROP1 (NM_ 006261.4)	1 in 141
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, PMM2-RELATED	PMM2 (NM_ 000303.2)	1 in 42
CONGENITAL MYASTHENIC SYNDROME, RAPSN-RELATED	RAPSN (NM_ 005055.4)	1 in 176
CRB1-RELATED RETINAL DYSTROPHIES	CRB1 (NM_ 201253.2)	1 in 112
CYSTIC FIBROSIS	CFTR (NM_ 000492.3)	1 in 25
DUCHENNE/BECKER MUSCULAR DYSTROPHY	DMD (NM_ 004006.2)	1 in 500
ELLIS-VAN CREVELD SYNDROME (WEYERS ACROFACIAL DYSOSTOSIS)	EYS (NM_ 001142800.1)	1 in 129
FACTOR XI DEFICIENCY	F11 (NM_ 000128.3)	1 in 101
FAMILIAL HYPERCHOLESTEROLEMIA, LDLRAP1-RELATED	LDLRAP1 (NM_ 015627.2)	1 in 143
FAMILIAL HYPERINSULINISM	ABCC8 (NM_000352.4)	1 in 100
FAMILIAL MEDITERRANEAN FEVER	MEFV (NM_ 000243.2)	1 in 115
FANCONI ANEMIA, GROUP A	FANCA (NM_ 000135.2)	1 in 64
FRAGILE X SYNDROME	FMR1 (NM_ 001185076.1)	1 in 178
GALACTOKINASE DEFICIENCY (GALACTOSEMIA, TYPE II)	GALK1 (NM_ 000154.1)	1 in 47
GALACTOSEMIA	GALT (NM_ 000155.3)	1 in 152
GAUCHER DISEASE	GBA (NM_ 000157.3)	1 in 164
GLUTARYL-CoA DEHYDROGENASE DEFICIENCY (GLUTARIC ACIDEMIA, TYPE 1)	GCDH (NM_ 000159.3)	1 in 172
GLYCINE ENCEPHALOPATHY	GLDC (NM_ 000170.2)	1 in 140
GLYCOGEN STORAGE DISEASE, TYPE 1a	G6PC (NM_ 000151.3)	1 in 177
GLYCOGEN STORAGE DISEASE, TYPE 2 (POMPE DISEASE)	GAA (NM_ 000152.3)	1 in 100
GLYCOGEN STORAGE DISEASE, TYPE III	AGL (NM_000028.2)	1 in 158
GLYCOGEN STORAGE DISEASE, TYPE IV	GBE1 (NM_ 000158.3)	1 in 144
GLYCOGEN STORAGE DISEASE, TYPE V (MCARDLE DISEASE)	PYGM (NM_ 005609.2)	1 in 191
HEREDITARY FRUCTOSE INTOLERANCE	ALDOB (NM_000035.3)	1 in 80
HOMOCYSTINURIA	CBS (NM_ 000071.2)	1 in 52
ISOVALERIC ACIDEMIA	IVD (NM_ 002225.3)	1 in 144
KRABBE DISEASE	GALC (NM_ 000153.3)	1 in 158
LEBER CONGENITAL AMAUROSIS, TYPE CEP290	CEP290 (NM_ 025114.3)	1 in 185
LIMB GIRDLE MUSCULAR DYSTROPHY, TYPE 2A	CAPN3 (NM_ 000070.2)	1 in 130
LIMB GIRDLE MUSCULAR DYSTROPHY, TYPE 2I	FKRP (NM_ 024301.4)	1 in 158
MEDIUM CHAIN ACYL_CoA DEHYDROGENASE DEFICIENCY	ACADM (NM_000016.5)	1 in 55
METACHROMATIC LEUKODYSTROPHY, ARSA-RELATED	ARSA (NM_000487.5)	1 in 100
METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE cblC	MMACHC (NM_ 015506.2)	1 in 135
MUCOLIPIDOSIS, TYPE IV	MCOLN1 (NM_ 020533.2)	1 in 89
MUCOPOLYSACCHARIDOSIS, TYPE IIIC (SANFILIPPO C)	IDUA (NM_ 000203.4)	1 in 144
NIEMANN PICK DISEASE, TYPE A/B	SMPD1 (NM_ 000543.4)	General population: 1 in 190; Caucasian: 1 in 244
NIEMANN PICK DISEASE, TYPE C1/D	NPC1 (NM_ 000271.4)	1 in 185
NIJMEGEN BREAKAGE SYNDROME	NBN (NM_ 002485.4)	1 in 155
NON SYNDROMIC HEARING LOSS	GJB2 (NM_ 004004.5)	1 in 42
PENDRED SYNDROME	SLC26A4 (NM_ 000441.1)	1 in 88
PHENYLKETONURIA	PAH (NM_ 000277.1)	Caucasian: 1 in 50; Sicily: 1 in 26
POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE	PKHD1 (NM_ 138694.3)	1 in 100
POLYGLANDULAR AUTOIMMUNE SYNDROME	AIRE (NM_000383.3)	1 in 60
PRIMARY CILIARY DYSKINESIA, DNAH5-RELATED	DNAH5 (NM_ 001369.2)	1 in 120
PRIMARY HYPEROXALURIA, TYPE 1	AGXT (NM_000030.2)	1 in 158
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 2	ABCB11 (NM_003742.2)	1 in 158
RETINITIS PIGMENTOSA 26	CERKL (NM_ 001030311.2)	1 in 137
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE I	PEX7 (NM_ 000288.3)	1 in 158
SMITH LEMLI-OPITZ SYNDROME	DHCR7 (NM_ 001360.2)	1 in 48
SPINAL MUSCULAR ATROPHY	SMN1 (NM_ 000344.3)	1 IN 40
TAY-SACHS DISEASE	HEXA (NM_ 000520.4)	1 in 182
USHER SYNDROME, TYPE 1B	MYO7A (NM_ 000260.3)	1 in 145
USHER SYNDROME, TYPE 2A	USH2A (NM_ 206933.2	1 in 73
VERY LONG CHAIN ACYL_CoA DEHYDROGENASE DEFICIENCY	ACADVL (NM_000018.3)	1 in 88
WILSON DISEASE	ATP7B (NM_ 000053.3)	1 in 90
WOLMAN DISEASE	LIPA (NM_ 000235.3)	1 in 145
ZELLWEGER SPECTRUM DISORDERS, PEX1-RELATED	PEX1 (NM_ 000466.2)	1 in 147