METABOLIC DISORDERS GENE PANEL

Gene symbol	Depth (reads)	Coverage (avg %)	OMIM disease	Description
AASS	104	95	238700	Hyperlysinemia
ABAT	82	86	613163	GABA-transaminase deficiency
ABCD1	59	84	300100	Adrenoleukodystrophy
ABCD4	121	88	614857	Methylmalonic aciduria and homocystinuria cblJ type
ABCG5	132	86	210250	Sitosterolemia
ABCG8	99	85	611465	Gallbladder disease 4
ABHD12	72	98	612674	Polyneuropathy hearing loss ataxia retinitis pigmentosa and cataract
ABHD5	125	91	275630	Chanarin-Dorfman syndrome
ACACA	106	87	613933	Acetyl-CoA carboxylase deficiency
ACAD8	101	80	611283	Isobutyryl-CoA dehydrogenase deficiency
ACAD9	105	89	611126	ACAD9 deficiency
ACADM	146	99	201450	Acyl-CoA dehydrogenase medium chain deficiency of
ACADS	106	95	201470	Acyl-CoA dehydrogenase short-chain deficiency of
ACADSB	89	95	610006	2-methylbutyrylglycinuria
ACADVL	100	89	201475	VLCAD deficiency
ACAT1	101	96	203750	Alpha-methylacetoacetic aciduria
ACAT2	96	95	614055	ACAT2 deficiency
ACO2	76	67	614559	Infantile cerebellar-retinal degeneration
ACOX1	90	73	264470	Peroxisomal acyl-CoA oxidase deficiency
ACSF3	79	89	614265	Combined malonic and methylmalonic aciduria
ACSL4	98	95	300387	Mental retardation X-linked 63
ACY1	85	88	609924	Aminoacylase 1 deficiency
ADA	82	90	102700	Adenosine deaminase deficiency partial
ADCY5	80	89	606703	Dyskinesia familial with facial myokymia
ADK	98	94	614300	Hypermethioninemia due to adenosine kinase deficiency
ADSL	124	83	103050	Adenylosuccinase deficiency
AGA	121	94	208400	Aspartylglucosaminuria
AGK	121	84	614691	Cataract 38 autosomal recessive

AGL	134	97	232400	Glycogen storage disease IIIa
AGPAT2	53	98	608594	Lipodystrophy congenital generalized type 1
AGPS	101	98	600121	Rhizomelic chondrodysplasia punctata type 3
AGXT	88	95	259900	Hyperoxaluria primary type 1
AHCY	93	77	613752	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
AK1	84	91	612631	Hemolytic anemia due to adenylate kinase deficiency
AK2	107	44	267500	Reticular dysgenesis
AKR1D1	116	79	235555	Bile acid synthesis defect congenital 2
ALAD	87	94	612740	Porphyria acute hepatic
ALAS2	84	77	300751	Anemia sideroblastic X-linked
ALDH18A1	103	82	219150	Cutis laxa autosomal recessive type IIIA
ALDH1A3	109	84	615113	Microphthalmia isolated 8
ALDH2	94	86	610251	Alcohol sensitivity acute
ALDH3A2	103	85	270200	Sjogren-Larsson syndrome
ALDH4A1	65	90	239510	Hyperprolinemia type II
ALDH5A1	71	88	271980	Succinic semialdehyde dehydrogenase deficiency
ALDH6A1	118	88	614105	Methylmalonate semialdehyde dehydrogenase deficiency
ALDH7A1	92	79	266100	Epilepsy pyridoxine-dependent
ALDOA	110	75	611881	Glycogen storage disease XII
ALDOB	115	79	229600	Fructose intolerance
ALG1	104	87	608540	Congenital disorder of glycosylation type Ik
ALG10	139	16	613688	{Acquired long QT syndrome, reduced susceptibility to}
ALG11	157	95	613661	Congenital disorder of glycosylation type Ip
ALG12	105	88	607143	Congenital disorder of glycosylation type Ig
ALG13	101	85	300884	Congenital disorder of glycosylation type Is
ALG2	109	97	607906	Congenital disorder of glycosylation type Ii
ALG3	117	86	601110	Congenital disorder of glycosylation type Id
ALG6	87	100	603147	Congenital disorder of glycosylation type Ic
ALG8	91	96	608104	Congenital disorder of glycosylation type Ih
ALG9	107	87	608776	Congenital disorder of glycosylation type Il
ALOX12B	94	91	242100	Ichthyosis congenital autosomal recessive 2
ALPL	88	81	146300	Hypophosphatasia adult
AMACR	85	91	614307	Alpha-methylacyl-CoA racemase deficiency

AMN	46	92	261100	Megaloblastic anemia-1 Norwegian type
AMPD3	90	91	612874	[AMP deaminase deficiency, erythrocytic]
AMT	135	94	605899	Glycine encephalopathy
AP1S1	83	86	609313	MEDNIK syndrome
APOC2	168	100	207750	Hyperlipoproteinemia type Ib
APRT	55	89	614723	Adenine phosphoribosyltransferase deficiency
ARG1	110	88	207800	Argininemia
ARSA	83	94	250100	Metachromatic leukodystrophy
ARSB	91	85	253200	Mucopolysaccharidosis type VI (Maroteaux-Lamy)
ASAH1	103	92	228000	Farber lipogranulomatosis
ASL	79	88	207900	Argininosuccinic aciduria
ASPA	118	96	271900	Canavan disease
ASS1	68	37	215700	Citrullinemia
ATIC	116	87	608688	AICA-ribosiduria due to ATIC deficiency
ATP6V0A2	110	93	219200	Cutis laxa autosomal recessive type IIA
ATP7A	101	96	309400	Menkes disease
ATP7B	130	86	277900	Wilson disease
ATP8B1	122	87	243300	Cholestasis benign recurrent intrahepatic
AUH	111	87	250950	3-methylglutaconic aciduria type I
B3GALNT1	120	92	615021	[Blood group, globoside system]
B3GALNT2	99	79	615181	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies type A 11
B3GALTL	109	92	261540	Peters-plus syndrome
B3GAT3	87	61	245600	Multiple joint dislocations short stature craniofacial dysmorphism and congenital heart defects
B3GNT2	136	97	200	-
B4GALT1	86	95	607091	Congenital disorder of glycosylation type IId
B4GALT7	74	95	130070	Ehlers-Danlos syndrome progeroid type 1
BAAT	142	90	607748	Hypercholanemia familial
BCKDHA	109	92	248600	Maple syrup urine disease type Ia
BCKDHB	89	98	248600	Maple syrup urine disease type Ib
BCMO1	129	84	115300	Hypercarotenemia and vitamin A deficiency autosomal dominant
BLVRA	91	88	614156	Hyperbiliverdinemia
BMP2	119	100	112600	Brachydactyly type A2
BPGM	145	100	222800	Erythrocytosis due to bisphosphoglycerate mutase deficiency

BTD	146	94	253260	Biotinidase deficiency
C1GALT1C1	118	100	300622	Tn polyagglutination syndrome somatic
C7orf10	102	95	231690	[Glutaric aciduria III]
CANT1	90	92	251450	Desbuquois dysplasia
CAT	99	88	614097	Acatalasemia
CBS	77	86	236200	Homocystinuria B6-responsive and nonresponsive types
CEL	84	46	609812	Maturity-onset diabetes of the young type VIII
CERKL	127	93	608380	Retinitis pigmentosa 26
CERS3	88	93	615023	Ichthyosis congenital autosomal recessive 9
CFTR	136	94	277180	Congenital bilateral absence of vas deferens
CHIT1	82	77	614122	[Chitotriosidase deficiency]
CHKB	89	90	602541	Muscular dystrophy congenital megaconial type
CHST14	107	100	601776	Ehlers-Danlos syndrome musculocontractural type
CHST3	71	100	143095	Spondyloepiphyseal dysplasia with congenital joint dislocations
CHST6	109	48	217800	Macular corneal dystrophy
CHSY1	154	100	605282	Temtamy preaxial brachydactyly syndrome
CLN3	89	87	204200	Ceroid lipofuscinosis neuronal 3
CLN5	124	98	256731	Ceroid lipofuscinosis neuronal 5
CLN6	64	88	601780	Ceroid lipofuscinosis neuronal 6
CLN8	131	96	600143	Ceroid lipofuscinosis neuronal 8
CLPB	112	89	128100	Dystonia-1, torsion
COG1	125	90	611209	Congenital disorder of glycosylation type IIg
COG4	96	84	613489	Congenital disorder of glycosylation type IIj
COG5	105	95	613612	Congenital disorder of glycosylation type IIi
COG6	91	96	615328	Shaheen syndrome
COG7	99	76	608779	Congenital disorder of glycosylation type IIe
COG8	119	79	611182	Congenital disorder of glycosylation type IIh
COMT	64	94	181500	{Schizophrenia, susceptibility to}
CP	97	72	604290	Cerebellar ataxia
CPOX	84	93	121300	Coproporphyria
CPS1	107	93	237300	Carbamoylphosphate synthetase I deficiency
CPT1A	111	81	255120	CPT deficiency hepatic type IA
CPT2	116	96	600649	CPT deficiency hepatic type II

CTH	120	95	219500	Cystathioninuria
CTNS	116	93	219800	Cystinosis atypical nephropathic
CTSA	99	90	256540	Galactosialidosis
CTSC	99	96	245010	Haim-Munk syndrome
CTSD	84	97	610127	Ceroid lipofuscinosis neuronal 10
CTSK	128	85	265800	Pycnodysostosis
CUBN	100	78	261100	Megaloblastic anemia-1 Finnish type
CYB5R3	66	91	250800	Methemoglobinemia type I
CYP11A1	86	94	613743	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete
CYP11B1	124	64	202010	Adrenal hyperplasia congenital due to 11-beta-hydroxylase deficiency
CYP11B2	112	53	200	-
CYP17A1	112	87	202110	1720-lyase deficiency isolated
CYP19A1	140	92	613546	Aromatase deficiency
CYP1B1	108	98	231300	Glaucoma 3A primary open angle congenital juvenile or adult onset
CYP21A2	19	82	201910	Adrenal hyperplasia congenital due to 21-hydroxylase deficiency
CYP27A1	126	86	213700	Cerebrotendinous xanthomatosis
CYP27B1	119	85	264700	Vitamin D-dependent rickets type I
CYP2R1	108	93	600081	Rickets due to defect in vitamin D 25-hydroxylation
CYP2U1	109	94	615030	Spastic paraplegia 56 autosomal recessive
CYP7B1	98	88	613812	Bile acid synthesis defect congenital 3
D2HGDH	58	84	600721	D-2-hydroxyglutaric aciduria
DAO	119	88	181500	{Schizophrenia}
DBH	99	89	223360	Dopamine beta-hydroxylase deficiency
DBT	100	93	248600	Maple syrup urine disease type II
DCXR	87	91	260800	Pentosuria
DDC	102	93	608643	Aromatic L-amino acid decarboxylase deficiency
DDHD1	117	99	609340	Spastic paraplegia 28 autosomal recessive
DDOST	113	89	614507	Congenital disorder of glycosylation type Ir
DGAT1	71	100	200	-
DGKE	109	91	615008	Nephrotic syndrome type 7
DGUOK	102	90	251880	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)
DHCR24	95	79	602398	Desmosterolosis
DHCR7	118	88	270400	Smith-Lemli-Opitz syndrome

DHFR	57	32	613839	Megaloblastic anemia due to dihydrofolate reductase deficiency
DHODH	107	81	263750	Miller syndrome
DLD	141	94	246900	Dihydrolipoamide dehydrogenase deficiency
DMGDH	124	88	605850	Dimethylglycine dehydrogenase deficiency
DNAJC19	101	83	610198	3-methylglutaconic aciduria type V
DNM1L	93	98	614388	Encephalopahty lethal due to defective mitochondrial peroxisomal fission
DNM2	85	89	606482	Charcot-Marie-Tooth disease axonal type 2M
DNMT1	105	89	604121	Cerebellar ataxia deafness and narcolepsy autosomal dominant
DNMT3B	93	82	242860	Immunodeficiency-centromeric instability-facial anomalies syndrome 1
DOLK	142	100	610768	Congenital disorder of glycosylation, type Im
DPAGT1	116	89	608093	Congenital disorder of glycosylation type Ij
DPM1	145	100	608799	Congenital disorder of glycosylation type Ie
DPM2	72	90	615042	Congenital disorder of glycosylation type Iu
DPM3	87	100	612937	Congenital disorder of glycosylation type Io
DPYD	122	88	274270	5-fluorouracil toxicity
DPYS	90	78	222748	Dihydropyrimidinuria
EBP	71	78	302960	Chondrodysplasia punctata X-linked dominant
ECHS1	77	80	200	-
ELOVL4	111	92	614457	Ichthyosis spastic quadriplegia and mental retardation
ENO3	116	88	612932	Glycogen storage disease XIII
EPHX1	92	90	607748	Hypercholanemia familial
EPHX2	91	84	143890	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}
ETFA	109	97	231680	Glutaric acidemia IIA
ETFB	101	86	231680	Glutaric acidemia IIB
ETFDH	123	96	231680	Glutaric acidemia IIC
ETHE1	65	93	602473	Ethylmalonic encephalopathy
EXT1	111	90	215300	Chondrosarcoma
EXT2	127	79	133701	Exostoses multiple type 2
FA2H	61	88	612319	Spastic paraplegia 35 autosomal recessive
FAH	119	88	276700	Tyrosinemia type I
FBP1	88	90	229700	Fructose-16-bidphosphatase deficiency
FECH	109	84	177000	Protoporphyria erythropoietic autosomal recessive
FH	88	80	606812	Fumarase deficiency

FKRP	67	98	613153	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 5
FKTN	112	91	611615	Cardiomyopathy dilated 1X
FMO3	127	84	602079	Trimethylaminuria
FOLR1	99	81	613068	Neurodegeneration due to cerebral folate transport deficiency
FTCD	49	84	229100	Glutamate formiminotransferase deficiency
FUCA1	81	88	230000	Fucosidosis
FUT2	191	47	612542	{Vitamin B12 plasma level QTL1}
FUT6	119	26	613852	Fucosyltransferase 6 deficiency
G6PC	137	91	232200	Glycogen storage disease Ia
G6PC3	123	86	612541	Dursun syndrome
G6PD	77	93	134700	Favism
GAA	98	91	232300	Glycogen storage disease II
GAD1	112	87	603513	Cerebral palsy spastic quadriplegic 1
GALC	100	96	245200	Krabbe disease
GALE	107	97	230350	Galactose epimerase deficiency
GALK1	84	90	230200	Galactokinase deficiency with cataracts
GALNS	63	92	253000	Mucopolysaccharidosis IVA
GALT	128	90	230400	Galactosemia
GAMT	77	92	612736	Cerebral creatine deficiency syndrome 2
GATM	91	87	612718	Cerebral creatine deficiency syndrome 3
GBA	145	76	608013	Gaucher disease perinatal lethal
GBA2	142	96	614409	Spastic paraplegia 46 autosomal recessive
GBE1	109	96	232500	Glycogen storage disease IV
GCDH	89	86	231670	Glutaricaciduria type I
GCH1	85	98	128230	Dystonia DOPA-responsive with or without hyperphenylalaninemia
GCK	77	87	125851	Diabetes mellitus gestational
GCLC	124	93	230450	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency
GCLM	92	89		
GCSH	50	31	605899	Glycine encephalopathy
GFPT1	109	96	610542	Myasthenia congenital with tubular aggregates 1
GK	59	29	307030	Glycerol kinase deficiency
GLA	82	80	301500	Fabry disease
GLB1	88	84	230500	GM1-gangliosidosis type I

GLDC	74	67	605899	Glycine encephalopathy
GLRA1	116	85	149400	Hyperekplexia hereditary 1 autosomal dominant or recessive
GLRX5	37	76	205950	Anemia sideroblastic pyridoxine-refractory autosomal recessive
GLUD1	130	67	606762	Hyperinsulinism-hyperammonemia syndrome
GLUL	66	35	610015	Glutamine deficiency congenital
GLYCTK	93	91	220120	D-glyceric aciduria
GM2A	107	93	272750	GM2-gangliosidosis AB variant
GMPPB	106	100	615350	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 14
GMPS	128	87	601626	Leukemia acute myelogenous
GNE	112	84	600737	Inclusion body myopathy autosomal recessive
GNMT	77	95	606664	Glycine N-methyltransferase deficiency
GNPAT	139	83	222765	Chondrodysplasia punctata rhizomelic type 2
GNPTAB	138	93	252500	Mucolipidosis II alpha/beta
GNPTG	69	100	252605	Mucolipidosis III gamma
GNS	86	85	252940	Mucopolysaccharidosis type IIID
GOT1	110	85	614419	Aspartate aminotransferase serum level of QTL1
GPD1	77	88	614480	Hypertriglyceridemia transient infantile
GPD1L	111	85	611777	Brugada syndrome 2
GPHN	121	95	252150	Molybdenum cofactor deficiency type C
GPI	103	92	613470	Hemolytic anemia nonspherocytic due to glucose phosphate isomerase deficiency
GPX1	62	16	614164	Hemolytic anemia due to glutathione peroxidase deficiency
GRHPR	99	86	260000	Hyperoxaluria primary type II
GSS	104	77	266130	Glutathione synthetase deficiency
GUSB	79	70	253220	Mucopolysaccharidosis VII
GYG1	72	39	613507	Glycogen storage disease XV
GYS1	74	86	611556	Glycogen storage disease 0 muscle
GYS2	93	96	240600	Glycogen storage disease type 0
H6PD	128	98	604931	Cortisone reductase deficiency 1
HADH	91	94	609975	Hyperinsulinemic hypoglycemia, familial, 4
HADHA	110	68	609016	Fatty liver acute of pregnancy
HADHB	90	76	609015	Trifunctional protein deficiency
HAGH	81	61	614033	[Glyoxalase II deficiency]
HEXA	101	88	272800	GM2-gangliosidosis several forms

HEXB	106	97	268800	Sandhoff disease infantile juvenile and adult forms
HFE	127	85	200	Hemochromatosis
HGD	117	86	203500	Alkaptonuria
HGSNAT	112	91	252930	Mucopolysaccharidosis type IIIC (Sanfilippo C)
HIBADH	130	93	200	-
HIBCH	75	77	250620	3-hydroxyisobutryl-CoA hydrolase deficiency
HK1	117	87	235700	Hemolytic anemia due to hexokinase deficiency
HLCS	143	89	253270	Holocarboxylase synthetase deficiency
HMBS	122	81	176000	Porphyria acute intermittent
HMGCL	123	83	246450	HMG-CoA lyase deficiency
HMGCS2	129	85	605911	HMG-CoA synthase-2 deficiency
HMOX1	66	86	614034	Heme oxygenase-1 deficiency
HOGA1	65	96	613616	Hyperoxaluria primary type III
HPD	95	97	140350	Hawkinsinuria
HPRT1	75	80	300323	HPRT-related gout
HS6ST1	11	4	614880	{Hypogonadotropic hypogonadism 15 with or without anosmia}
HSD11B1	105	87	614662	Cortisone reductase deficiency 2
HSD11B2	105	89	218030	Apparent mineralocorticoid excess
HSD17B10	87	89	300438	17-beta-hydroxysteroid dehydrogenase X deficiency
HSD17B3	102	92	264300	Pseudohermaphroditism male with gynecomastia
HSD17B4	103	89	261515	D-bifunctional protein deficiency
HSD3B2	98	21	201810	3-beta-hydroxysteroid dehydrogenase type II deficiency
HSD3B7	50	77	607765	Bile acid synthesis defect congenital 1
HYAL1	85	96	601492	Mucopolysaccharidosis type IX
IDH2	109	94	613657	D-2-hydrosyglutaric aciduria 2
IDH3B	141	92	612572	Retinitis pigmentosa 46
IDS	92	89	309900	Mucopolysaccharidosis II
IDUA	83	88	607014	Mucopolysaccharidosis Ih
IMPAD1	97	99	614078	Chondrodysplasia with joint dislocations GRAPP type
IMPDH1	54	35	613837	Leber congenital amaurosis 11
INPP5E	67	95	213300	Joubert syndrome 1
INPPL1	90	90	258480	Opsismodysplasia
ISPD	100	89	614643	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 7

IVD	94	82	243500	Isovaleric acidemia
KMT2A	158	97	200	-
KMT2D	111	96	147920	Kabuki syndrome 1
L2HGDH	81	97	236792	L-2-hydroxyglutaric aciduria
LAMP2	90	87	300257	Danon disease
LARGE	116	82	613154	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 6
LCAT	98	91	136120	Fish-eye disease
LCT	137	92	223000	Lactase deficiency congenital
LDHA	78	22	612933	Glycogen storage disease XI
LDHB	102	66	614128	Lactate dehydrogenase-B deficiency
LFNG	83	81	609813	Spondylocostal dysostosis autosomal recessive 3
LIPA	108	93	278000	Cholesteryl ester storage disease
LIPC	95	86	614025	Hepatic lipase deficiency
LMBRD1	103	98	277380	Methylmalonic aciduria and homocystinuria cblF type
LPIN1	112	87	268200	Myoglobinuria acute recurrent autosomal recessive
LPIN2	89	85	609628	Majeed syndrome
LPL	123	80	144250	Combined hyperlipidemia familial
LRAT	182	100	613341	Leber congenital amaurosis 14
LTC4S	48	74	614037	Leukotriene C4 synthase deficiency
LYST	124	94	214500	Chediak-Higashi syndrome
MAN1B1	97	85	614202	Mental retardation autosomal recessive 15
MAN2B1	89	85	248500	Mannosidosis alpha- types I and II
MANBA	98	92	248510	Mannosidosis beta
MAOA	81	90	300615	Brunner syndrome
MAT1A	105	87	250850	Hypermethioninemia persistent autosomal dominant due to methionine adenosyltransferase I/III deficiency
MCCC1	117	91	210200	3-Methylcrotonyl-CoA carboxylase 1 deficiency
MCCC2	118	91	210210	3-Methylcrotonyl-CoA carboxylase 2 deficiency
MCEE	84	97	251120	Methylmalonyl-CoA epimerase deficiency
MCOLN1	97	90	252650	Mucolipidosis IV
MFSD8	115	93	610951	Ceroid lipofuscinosis neuronal 7
MGAT2	207	100	212066	Congenital disorder of glycosylation type IIa
MINPP1	111	98	188470	Thyroid carcinoma follicular
MLYCD	90	86	248360	Malonyl-CoA decarboxylase deficiency

MMAA	172	98	251100	Methylmalonic aciduria vitamin B12-responsive
MMAB	90	92	251110	Methylmalonic aciduria vitamin B12-responsive due to defect in synthesis of adenosylcobalamin cblB complementation type
MMACHC	166	89	277400	Methylmalonic aciduria and homocystinuria cblC type
MMADHC	79	80	277410	Methylmalonic aciduria and homocystinuria, cblD type
MMADHC	79	80	277410	Methylmalonic aciduria, cblD type, variant 2
MOCS1	125	19	252150	Molybdenum cofactor deficiency type A
MOCS2	85	70	252150	Molybdenum cofactor deficiency type B
MOGS	129	96	606056	Congenital disorder of glycosylation, type IIb
MPDU1	134	96	609180	Congenital disorder of glycosylation type If
MPI	110	82	602579	Congenital disorder of glycosylation type Ib
MSMO1	108	78	200	-
MTHFD1	115	76	200	{Abruptio placentae, susceptibility to}
MTHFD1	115	76	601634	{Spina bifida, folate-sensitive, susceptibility to}
MTHFR	98	91	236250	Homocystinuria due to MTHFR deficiency
MTM1	89	89	310400	Myotubular myopathy X-linked
MTMR2	107	84	601382	Charcot-Marie-Tooth disease type 4B1
MTR	119	86	250940	Homocystinuria-megaloblastic anemia cblG complementation type
MTRR	113	93	236270	Homocystinuria-megaloblastic anemia cbl E type
MUT	125	94	251000	Methylmalonic aciduria mut(0) type
MVK	101	78	260920	Hyper-IgD syndrome
NAGA	93	84	609242	Kanzaki disease
NAGLU	62	92	252920	Mucopolysaccharidosis type IIIB (Sanfilippo B)
NAGS	53	88	237310	N-acetylglutamate synthase deficiency
NEU1	16	87	256550	Sialidosis type I
NMNAT1	144	52	608553	Leber congenital amaurosis 9
NNT	107	91	614736	Glucocorticoid deficiency 4
NPC1	110	84	257220	Niemann-Pick disease type C1
NPC2	76	86	607625	Niemann-pick disease type C2
NSD1	138	92	130650	Beckwith-Wiedemann syndrome
NSDHL	77	91	308050	CHILD syndrome
NT5C3A	85	66	266120	Anemia, hemolytic, due to UMPH1 deficiency
NT5E	115	93	211800	Calcification of joints and arteries

OAT	58	58	258870	Gyrate atrophy of choroid and retina with or without ornithinemia
OCRL	107	89	300555	Dent disease 2
OPA3	85	87	258501	3-methylglutaconic aciduria type III
OPLAH	92	95	260005	5-oxoprolinase deficiency
OTC	85	93	311250	Ornithine transcarbamylase deficiency
OXCT1	107	97	245050	Succinyl CoA:3-oxoacid CoA transferase deficiency
PAH	93	91	261600	Phenylketonuria
PANK2	152	93	607236	HARP syndrome
PC	98	91	266150	Pyruvate carboxylase deficiency
PCBD1	80	85	264070	Hyperphenylalaninemia, BH4-deficient, D
PCCA	96	90	606054	Propionicacidemia
PCCB	116	84	606054	Propionicacidemia
PEPD	78	89	170100	Prolidase deficiency
PEX1	122	93	214100	Peroxisome biogenesis disorder 1A (Zellweger)
PEX10	76	90	614870	Peroxisome biogenesis disorder 6A (Zellweger)
PEX11B	184	88	614920	Peroxisome biogenesis disorder 14B
PEX12	131	90	614859	Peroxisome biogenesis disorder 3A (Zellweger)
PEX13	131	95	614883	Peroxisome biogenesis disorder 11A (Zellweger)
PEX14	98	85	614887	Peroxisome biogenesis disorder 13A (Zellweger)
PEX16	101	90	614876	Peroxisome biogenesis disorder 8A (Zellweger)
PEX19	112	86	614886	Peroxisome biogenesis disorder 12A (Zellweger)
PEX2	148	87	614867	Peroxisome biogenesis disorder 5B
PEX2	148	87	614866	Peroxisome biogenesis disorder 5A (Zellweger)
PEX26	118	91	614872	Peroxisome biogenesis disorder 7A (Zellweger)
PEX3	124	100	614882	Peroxisome biogenesis disorder 10A (Zellweger)
PEX5	98	74	214110	Peroxisome biogenesis disorder 2A (Zellweger)
PEX6	113	88	614862	Peroxisome biogenesis disorder 4A (Zellweger)
PEX7	112	91	614879	Peroxisome biogenesis disorder 9B
PFKM	130	80	232800	Glycogen storage disease VII
PGAM2	77	100	261670	Glycogen storage disease X
PGAP2	116	97	614207	Hyperphosphatasia with mental retardation syndrome 3
PGK1	82	43	300653	Phosphoglycerate kinase 1 deficiency
PGM1	109	76	614921	Congenital disorder of glycosylation type It

PHGDH	103	86	601815	Phosphoglycerate dehydrogenase deficiency
PHKA1	80	90	300559	Muscle glycogenosis
PHKA2	84	87	306000	Glycogen storage disease type IXa1
PHYH	90	93	266500	Refsum disease
PIGA	110	89	300868	Multiple congenital anomalies-hypotonia-seizures syndrome 2
PIGL	96	85	280000	CHIME syndrome
PIGM	128	100	610293	Glycosylphosphatidylinositol deficiency
PIGN	101	97	614080	Multiple congenital anomalies-hypotonia-seizures syndrome 1
PIGO	114	93	614749	Hyperphosphatasia with mental retardation syndrome 2
PIGT	135	91	615398	Multiple congenital anomalies-hypotonia-seizures syndrome 3
PIGV	191	97	239300	Hyperphosphatasia with mental retardation syndrome 1
PIK3CA	133	93	114480	Breast cancer somatic
PIK3R1	162	93	615214	Agammaglobulinemia 7 autosomal recessive
PIK3R2	84	89	603387	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome
PIK3R5	82	91	615217	Ataxia-oculomotor apraxia 3
PIKFYVE	144	96	121850	Corneal fleck dystrophy
PIP5K1C	61	90	611369	Lethal congenital contractural syndrome 3
PKLR	113	86	102900	Adenosine triphosphate elevated of erythrocytes
PLA2G5	123	74	228980	Fleck retina familial benign
PLA2G6	79	85	256600	Infantile neuroaxonal dystrophy 1
PLA2G7	114	92	614278	Platelet-activating factor acetylhydrolase deficiency
PLCB1	117	93	613722	Epileptic encephalopathy early infantile 12
PLCB4	98	90	614669	Auriculocondylar syndrome 2
PLCD1	98	93	151600	Nail disorder nonsyndromic congenital 3 (leukonychia)
PLCE1	132	86	610725	Nephrotic syndrome type 3
PLCG2	125	83	614878	Autoinflammation antibody deficiency and immune dysregulation syndrome
PLIN1	56	87	613877	Lipodystrophy familial partial type 4
PLOD1	84	88	225400	Ehlers-Danlos syndrome type VI
PLOD2	124	96	609220	Bruck syndrome 2
PLOD3	82	89	612394	Lysyl hydroxylase 3 deficiency
PMM2	118	89	212065	Congenital disorder of glycosylation type Ia
PNLIP	99	90	614338	Pancreatic lipase deficiency
PNMT	40	88	145500	Hypertension, essential

PNP	138	97	613179	Immunodeficiency due to purine nucleoside phosphorylase deficiency
PNPLA2	69	91	610717	Neutral lipid storage disease with myopathy
PNPLA6	90	88	612020	Spastic paraplegia 39 autosomal recessive
PNPO	71	86	610090	Pyridoxamine 5-phosphate oxidase deficiency
POLR3A	107	83	607694	Leukodystrophy hypomyelinating 7 with or without oligodontia and/or hypogonadotropic hypogonadism
POLR3B	112	89	614381	Leukodystrophy hypomyelinating 8 with or without oligodontia and/or hypogonadotropic hypogonadism
POMGNT1	106	90	253280	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 3
POMGNT2	114	100	614830	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8
POMK	136	97		
POMT1	116	87	236670	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 1
POMT2	84	90	613150	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 2
PPM1K	98	97	615135	Maple syrup urine disease mild variant
PPOX	107	93	176200	Porphyria variegata
PPT1	77	91	200	Ceroid lipofuscinosis neuronal 1
PRODH	63	81	239500	Hyperprolinemia type I
PRPS1	95	64	301835	Arts syndrome
PSAP	105	84	611721	Combined SAP deficiency
PSAT1	83	40	610992	Phosphoserine aminotransferase deficiency
PSPH	46	74	614023	Phosphoserine phosphatase deficiency
PTEN	130	65	153480	Bannayan-Riley-Ruvalcaba syndrome
PTGIS	74	76	145500	Hypertension essential
PTPN11	90	25	151100	LEOPARD syndrome 1
PTS	116	93	261640	Hyperphenylalaninemia BH4-deficient A
PYCR1	78	86	612940	Cutis laxa autosomal recessive type IIB
PYGL	109	84	232700	Glycogen storage disease VI
PYGM	100	86	232600	McArdle disease
QDPR	89	82	261630	Hyperphenylalaninemia BH4-deficient C
RDH12	83	78	612712	Leber congenital amaurosis 13
RDH5	115	83	136880	Fundus albipunctatus
RFT1	91	84	612015	Congenital disorder of glycosylation type In
RPE65	129	88	204100	Leber congenital amaurosis 2
RPIA	83	85	608611	Ribose 5-phosphate isomerase deficiency
SARDH	71	85	268900	[Sarcosinemia]

SAT1	99	92	308800	Keratosis follicularis spinulosa decalvans
SC5D	145	90	607330	Lathosterolosis
SCARB2	85	87	254900	Epilepsy progressive myoclonic 4 with or without renal failure
SCP2	102	90	613724	Leukoencephalopathy with dystonia and motor neuropathy
SEPSECS	104	93	613811	Pontocerebellar hypoplasia type 2D
SERAC1	95	95	614739	3-methylglutaconic aciduria with deafness encephalopathy and Leigh-like syndrome
SGSH	80	93	252900	Mucopolysaccharidisis type IIIA (Sanfilippo A)
SI	104	97	222900	Sucrase-isomaltase deficiency congenital
SLC16A1	145	94	245340	Erythrocyte lactate transporter defect
SLC17A5	107	94	604369	Salla disease
SLC22A5	128	81	212140	Carnitine deficiency systemic primary
SLC25A1	66	89	615182	Combined D-2- and L-2-hydroxyglutaric aciduria
SLC25A13	101	91	603471	Citrullinemia adult-onset type II
SLC25A15	146	88	238970	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome
SLC25A19	94	83	607196	Microcephaly Amish type
SLC25A20	95	83	212138	Carnitine-acylcarnitine translocase deficiency
SLC25A38	93	65	205950	Anemia sideroblastic pyridoxine-refractory autosomal recessive
SLC2A1	90	94	601042	Dystonia 9
SLC2A2	139	96	227810	Fanconi-Bickel syndrome
SLC30A10	125	97	613280	Hypermanganesemia with dystonia polycythemia and cirrhosis
SLC33A1	93	96	614482	Congenital cataracts hearing loss and neurodegeneration
SLC35A1	106	97	603585	Congenital disorder of glycosylation type IIf
SLC35C1	83	100	266265	Congenital disorder of glycosylation type IIc
SLC37A4	94	91	232220	Glycogen storage disease Ib
SLC39A4	73	90	201100	Acrodermatitis enteropathica
SLC3A1	132	92	220100	Cystinuria
SLC46A1	94	91	229050	Folate malabsorption hereditary
SLC52A1	125	100	615026	Riboflavin deficiency
SLC52A2	105	92	614707	Brown-Vialetto-Van Laere syndrome 2
SLC52A3	69	97	211530	Brown-Vialetto-Van Laere syndrome 1
SLC5A1	109	89	606824	Glucose/galactose malabsorption
SLC5A2	69	90	233100	Renal glucosuria
SLC6A8	15	27	300352	Cerebral creatine deficiency syndrome 1

SLC7A7	106	89	222700	Lysinuric protein intolerance
SLC7A9	83	92	220100	Cystinuria
SLCO1B1	107	80	237450	Hyperbilirubinemia Rotor type digenic
SLCO1B3	97	77	237450	Hyperbilirubinemia Rotor type digenic
SMPD1	95	94	257200	Niemann-Pick disease type A
SMS	52	23	309583	Mental retardation X-linked Snyder-Robinson type
SOD1	119	87	105400	Amyotrophic lateral sclerosis 1
SPR	69	90	612716	Dystonia dopa-responsive due to sepiapterin reductase deficiency
SPTLC1	96	91	162400	Neuropathy hereditary sensory and autonomic type IA
SPTLC2	104	94	613640	Neuropathy hereditary sensory and autonomic type IC
SRD5A2	71	96	264600	Pseudovaginal perineoscrotal hypospadias
SRD5A3	107	84	612379	Congenital disorder of glycosylation type Iq
ST3GAL3	134	97	615006	Epileptic encephalopathy early infantile 15
ST3GAL3	134	97	615006	Epileptic encephalopathy early infantile 15
ST3GAL3	134	97	615006	Epileptic encephalopathy early infantile 15
ST3GAL5	107	86	609056	Amish infantile epilepsy syndrome
STAR	120	81	201710	Lipoid adrenal hyperplasia
STS	119	87	308100	Ichthyosis X-linked
SUCLA2	93	57	612073	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with methylmalonic aciduria)
SUCLG1	120	86	245400	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)
SUCLG2	94	74	200	-
SUMF1	88	90	272200	Multiple sulfatase deficiency
SUOX	174	99	272300	Sulfite oxidase deficiency
TALDO1	101	86	606003	Transaldolase deficiency
TAT	122	85	276600	Tyrosinemia type II
TAZ	68	97	302060	Barth syndrome
TBXAS1	123	86	231095	Ghosal hematodiaphyseal syndrome
TCIRG1	76	91	259700	Osteopetrosis autosomal recessive 1
TCN2	112	85	275350	Transcobalamin II deficiency
TECR	77	77	614020	Mental retardation autosomal recessive 14
TH	86	91	605407	Segawa syndrome recessive
TK2	110	85	609560	Mitochondrial DNA depletion syndrome 2 (myopathic type)
TMEM165	83	91	614727	Congenital disorder of glycosylation type IIk

TMEM5	151	88	615041	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 10
TMLHE	68	81	300872	Epsilon-trimethyllysine hydroxylase deficiency
TPI1	63	39	200	-
TPMT	91	82	610460	6-mercaptopurine sensitivity
TPP1	137	95	204500	Ceroid lipofuscinosis neuronal 2
TREH	108	90	612119	Trehalase deficiency
TUSC3	73	33	611093	Mental retardation autosomal recessive 7
TYMP	78	95	603041	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
TYR	196	100	203100	Albinism oculocutaneous type IA
TYRP1	114	89	203290	Albinism oculocutaneous type III
UGT1A1	147	97	218800	Crigler-Najjar syndrome type I
UMPS	118	92	258900	Orotic aciduria
UPB1	124	81	613161	Beta-ureidopropionase deficiency
UROC1	76	85	276880	Urocanase deficiency
UROD	104	82	176100	Porphyria cutanea tarda
UROS	86	92	263700	Porphyria congenital erythropoietic
XDH	103	86	278300	Xanthinuria type I

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Depth describes the average number of reads seen across 50 exomes
Coverage describes the average coverage of a gene across 50 exomes in percentiles
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013

Ad 1. OMIM identifier 200 signifies a gene without a current OMIM association

Ad 2. OMIM phenotype descriptions between {} signify risk factors