Treatable ID

Gene panel

Gene panel information

Gene panel	Treatable ID
Version	2
Total genes	130
Activation date	Monday 23 october 2023
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
ABCC8	99.98 %	600509	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 (3), Autosomal recessive, Autosomal dominant; Diabetes mellitus, transient neonatal 2, 610374 (3); Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal recessive, Autosomal dominant
ABCD1	99.98 %	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
ABCD4	100 %	603214	Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3), Autosomal recessive
ACAD9	100 %	611103	Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive
ACAT1	99.81 %	607809	Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive
AGA	99.92 %	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
AHCY	100 %	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive
ALDH5A1	96.19 %	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
ALDH7A1	99.49 %	107323	Epilepsy, pyridoxine-dependent, 266100 (3), Autosomal recessive
AMT	100 %	238310	Glycine encephalopathy 2, 620398 (3)
AP1S1	99.49 %	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
ARG1	99.95 %	608313	Argininemia, 207800 (3), Autosomal recessive
ARSA	99.99 %	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ASL	99.98 %	608310	Argininosuccinic aciduria, 207900 (3), Autosomal recessive
ASS1	77.52 %	603470	Citrullinemia, 215700 (3), Autosomal recessive
ATP1A2	99.85 %	182340	Developmental and epileptic encephalopathy 98, 619605 (3), Autosomal dominant; Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 (3), Autosomal recessive; Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant
ATP7A	99.87 %	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive
ATP7B	100 %	606882	Wilson disease, 277900 (3), Autosomal recessive
BCKDHA	99.97 %	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
BCKDHB	99.73 %	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
BCKDK	99.99 %	614901	Branched-chain keto acid dehydrogenase kinase deficiency, 614923 (3)
BTD	100 %	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
CA5A	99.99 %	114761	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3), Autosomal recessive

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CAD	99.86 %	114010	Developmental and epileptic encephalopathy 50, 616457 (3), Autosomal recessive
CBS	17.79 %	613381	Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive; Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive
COQ5	99.95 %	616359	?Coenzyme Q10 deficiency, primary, 9, 619028 (3), Autosomal recessive
COQ8A	100 %	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
CP	99.95 %	117700	Cerebellar ataxia, 604290 (3), Autosomal recessive; [Hypoceruloplasminemia, hereditary], 604290 (3), Autosomal recessive; Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3), Autosomal recessive
CPS1	99.91 %	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3)
CYP27A1	100 %	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
DBT	94.51 %	248610	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
DDC	99.67 %	107930	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
DHCR7	99.97 %	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DHFR	98.89 %	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
DLAT	99.65 %	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
DLD	99.89 %	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DNAJC12	99.72 %	606060	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive
ECHS1	100 %	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
ETFA	99.88 %	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
ETFB	100 %	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
ETFDH	99.82 %	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive
ETHE1	84.97 %	608451	Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive
FARSA	100 %	602918	?Rajab interstitial lung disease with brain calcifications 2, 619013 (3), Autosomal recessive
FARSB	99.64 %	609690	Rajab interstitial lung disease with brain calcifications 1, 613658 (3), Autosomal recessive
FOLR1	100 %	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
FUCA1	98.72 %	612280	Fucosidosis, 230000 (3), Autosomal recessive
GAMT	100 %	601240	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
GATM	99.92 %	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive; Fanconi renotubular syndrome 1, 134600 (3), Autosomal dominant
GCDH	100 %	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
GCH1	99.94 %	600225	Dystonia, DOPA-responsive, 128230 (3), Autosomal recessive, Autosomal dominant; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
GLDC	99.99 %	238300	Glycine encephalopathy1, 605899 (3), Autosomal recessive
GLUL	99.88 %	138290	Glutamine deficiency, congenital, 610015 (3), Autosomal recessive
GOT2	99.78 %	138150	Developmental and epileptic encephalopathy 82, 618721 (3), Autosomal recessive
GRIN1	100 %	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive; Developmental and epileptic encephalopathy 101, 619814 (3), Autosomal recessive; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
GRIN2A	100 %	138253	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570 (3), Autosomal dominant
GRIN2B	99.99 %	138252	Developmental and epileptic encephalopathy 27, 616139 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 (3), Autosomal dominant
GRIN2D	99.97 %	602717	Developmental and epileptic encephalopathy 46, 617162 (3), Autosomal dominant
GUK1	99.99 %	139270	*No OMIM phenotypes*
GUSB	95.07 %	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
HIBCH	99.7 %	610690	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive
HLCS	99.97 %	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
HMGCL	99.31 %	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
HMGCS2	98.37 %	600234	HMG-CoA synthase-2 deficiency, 605911 (3), Autosomal recessive
IARS1	99.89 %	600709	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive
IDS	99.82 %	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
IDUA	99.99 %	252800	Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive
IVD	100 %	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
KCNJ11	100 %	600937	Diabetes, permanent neonatal 2, with or without neurologic features, 618856 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant; Diabetes mellitus, transient neonatal 3, 610582 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal recessive, Autosomal dominant
LARS1	99.87 %	151350	?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive
LMBRD1	99.67 %	612625	Methylmalonic aciduria and homocystinuria, cblF type, 277380 (3), Autosomal recessive
MAN2B1	99.99 %	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MARS1	99.97 %	156560	Spastic paraplegia 70, autosomal recessive, 620323 (3), Autosomal recessive; Interstitial lung and liver disease, 615486 (3), Autosomal recessive; ?Trichothiodystrophy 9, nonphotosensitive, 619692 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant
MFSD8	99.7 %	611124	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
MMAA	99.95 %	607481	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100 (3), Autosomal recessive
MMAB	99.99 %	607568	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110 (3), Autosomal recessive
MMACHC	99.98 %	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MMADHC	99.76 %	611935	Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive
MMUT	99.68 %	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
MOCS1	99.95 %	603707	Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive
MPI	99.95 %	154550	Congenital disorder of glycosylation, type Ib, 602579 (3), Autosomal recessive

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
MTHFR	99.97 %	607093	{Vascular disease, susceptibility to} (3); Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive
MTHFS	100 %	604197	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367 (3), Autosomal recessive
MTR	99.95 %	156570	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive
MTRR	99.98 %	602568	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
NAGS	99.99 %	608300	N-acetylglutamate synthase deficiency, 237310 (3), Autosomal recessive
NAXE	99.99 %	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive
NFE2L2	99.97 %	600492	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744 (3), Autosomal dominant
NPC1	99.99 %	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NPC2	100 %	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
OTC	99.42 %	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked
PAH	99.96 %	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
PCCA	99.9 %	232000	Propionicacidemia, 606054 (3), Autosomal recessive
PCCB	99.97 %	232050	Propionicacidemia, 606054 (3), Autosomal recessive
PDHA1	99.04 %	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PDHB	99.94 %	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3), Autosomal recessive
PDHX	99.64 %	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
PDP1	100 %	605993	Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive
PHGDH	99.79 %	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
PIGA	99.81 %	311770	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3); Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072 (3)
PIGM	99.97 %	610273	Glycosylphosphatidylinositol deficiency, 610293 (3), Autosomal recessive
PIGO	99.99 %	614730	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749 (3), Autosomal recessive
PLPBP	99.99 %	604436	Epilepsy, early-onset, vitamin B6-dependent, 617290 (3), Autosomal recessive
PMM2	99.93 %	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
PNPO	99.9 %	603287	Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive
PRPS1	99.95 %	311850	Arts syndrome, 301835 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive
PSAT1	99.98 %	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
PSPH	99.09 %	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
PTS	99.93 %	612719	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive

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QDPR	99.92 %	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
SARS1	98.53 %	607529	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 (3), Autosomal recessive
SLC18A2	100 %	193001	?Parkinsonism-dystonia, infantile, 2, 618049 (3), Autosomal recessive
SLC19A3	99.95 %	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive
SLC25A12	99.72 %	603667	Developmental and epileptic encephalopathy 39, 612949 (3), Autosomal recessive
SLC25A13	99.67 %	603859	Citrullinemia, type II, neonatal-onset, 605814 (3), Autosomal recessive; Citrullinemia, adult-onset type II, 603471 (3), Autosomal recessive
SLC25A15	100 %	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
SLC25A19	99.99 %	606521	Microcephaly, Amish type, 607196 (3), Autosomal recessive; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3), Autosomal recessive
SLC2A1	99.93 %	138140	Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal recessive, Autosomal dominant; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant
SLC35A2	99.97 %	314375	Congenital disorder of glycosylation, type IIm, 300896 (3), X-linked dominant, Somatic mosaicism
SLC35C1	100 %	605881	Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive
SLC39A8	99.95 %	608732	Congenital disorder of glycosylation, type IIn, 616721 (3), Autosomal recessive
SLC46A1	100 %	611672	Folate malabsorption, hereditary, 229050 (3), Autosomal recessive
SLC5A6	100 %	604024	Sodium-dependent multivitamin transporter deficiency, 618973 (3), Autosomal recessive; Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903 (3), Autosomal recessive
SLC6A8	99.99 %	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
SPR	99.99 %	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), Autosomal recessive, ?Autosomal dominant
TAT	99.99 %	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
TCN2	100 %	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
TH	99.99 %	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive
TMLHE	77.23 %	300777	{Autism, susceptibility to, X-linked 6}, 300872 (3), X-linked recessive
TPK1	99.96 %	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive
TPP1	99.99 %	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2023-07-31**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known

(2) the disorder has been placed on the map by linkage; no mutation has been found

(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* Exome panels: >=20x, HyperCap panels: >=30x