NBIA

Gene panel

Gene panel information

Gene panel	**NBIA**
Version	4
Total genes	25
Activation date	Friday 30 october 2026
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
AP1S2	99.72 %	300629	Pettigrew syndrome, 304340 (3), X-linked recessive
AP4M1	98.54 %	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
ATP13A2	99.27 %	610513	Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive
ATP7B	99.95 %	606882	Wilson disease, 277900 (3), Autosomal recessive
BCAS3	98.22 %	607470	Hengel-Maroofian-Schols syndrome, 619641 (3), Autosomal recessive
C19orf12	99.83 %	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal recessive, Autosomal dominant; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
COASY	99.73 %	609855	Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
CP	99.82 %	117700	Aceruloplasminemia, 604290 (3), Autosomal recessive
CRAT	99.92 %	600184	?Neurodegeneration with brain iron accumulation 8, 617917 (3), Autosomal recessive
DCAF17	99.65 %	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
FA2H	99.5 %	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
FTH1	99.97 %	134770	Neurodegeneration with brain iron accumulation 9, 620669 (3), Autosomal dominant; ?Hemochromatosis, type 5, 615517 (3), Autosomal dominant
FTL	99.52 %	134790	Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal recessive, Autosomal dominant; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant
FUCA1	98.82 %	612280	Fucosidosis, 230000 (3), Autosomal recessive
GLB1	99.98 %	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive
GTPBP2	99.76 %	607434	Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive
KMT2B	99.65 %	606834	Intellectual developmental disorder, autosomal dominant 68, 619934 (3), Autosomal dominant; Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant
PANK2	99.86 %	606157	Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
PLA2G6	99.79 %	603604	Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive
REPS1	99.84 %	614825	?Neurodegeneration with brain iron accumulation 7, 617916 (3), Autosomal recessive
SCP2	96.89 %	184755	?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TBCE	99.8 %	604934	Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive
THAP1	99.85 %	609520	Dystonia 6, torsion, 602629 (3), Autosomal dominant
VAC14	99.53 %	604632	Striatonigral degeneration, childhood-onset, 617054 (3), Autosomal recessive
WDR45	99.91 %	300526	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

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.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.