# NBIA

Gene panel

## Gene panel information

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| **Gene panel** | **NBIA** |
| **Version** | 3 |
| **Total genes** | 23 |
| **Activation date** | Friday 21 march 2025 |
| **Publisher** | Center for Medical Genetics, Ghent |

## Genes

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **AP4M1** | 99.98 % | 602296 | Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive |
| **ATP13A2** | 99.96 % | 610513 | Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive |
| **C19orf12** | 99.99 % | 614297 | Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal dominant, Autosomal recessive; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive |
| **COASY** | 99.98 % | 609855 | Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive |
| **CP** | 99.95 % | 117700 | Aceruloplasminemia, 604290 (3), Autosomal recessive |
| **CRAT** | 99.99 % | 600184 | ?Neurodegeneration with brain iron accumulation 8, 617917 (3), Autosomal recessive |
| **DCAF17** | 99.84 % | 612515 | Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive |
| **DDHD1** | 99.93 % | 614603 | Spastic paraplegia 28, autosomal recessive, 609340 (3), Autosomal recessive |
| **FA2H** | 99.98 % | 611026 | Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive |
| **FTH1** | 22.62 % | 134770 | Neurodegeneration with brain iron accumulation 9, 620669 (3), Autosomal dominant; ?Hemochromatosis, type 5, 615517 (3), Autosomal dominant |
| **FTL** | 99.99 % | 134790 | Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal dominant, Autosomal recessive; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant |
| **FUCA1** | 98.72 % | 612280 | Fucosidosis, 230000 (3), Autosomal recessive |
| **GLB1** | 100 % | 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** | 99.98 % | 607434 | Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive |
| **IRF2BPL** | 99.21 % | 611720 | Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant |
| **KMT2B** | 99.99 % | 606834 | Intellectual developmental disorder, autosomal dominant 68, 619934 (3), Autosomal dominant; Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant |
| **PANK2** | 99.99 % | 606157 | Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive |
| **PLA2G6** | 99.98 % | 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** | 99.93 % | 614825 | ?Neurodegeneration with brain iron accumulation 7, 617916 (3), Autosomal recessive |
| **SCP2** | 94.94 % | 184755 | ?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive |

| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **TBCE** | 99.91 % | 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 |
| **VAC14** | 99.91 % | 604632 | Striatonigral degeneration, childhood-onset, 617054 (3), Autosomal recessive |
| **WDR45** | 99.99 % | 300526 | Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant |

## Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2024-09-05**

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.