NBIA panel

versie	16-Oct-2018 (16 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
AP4M1	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
ATP13A2	610513	Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive; Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive
C19orf12	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal recessive; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
COASY	609855	Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
CP	117700	Cerebellar ataxia, 604290 (3), Autosomal recessive; Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3), Autosomal recessive; [Hypoceruloplasminemia, hereditary], 604290 (3), Autosomal recessive
CRAT	600184	?Neurodegeneration with brain iron accumulation 8, 617917 (3), Autosomal recessive
DCAF17	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
DDHD1	614603	Spastic paraplegia 28, autosomal recessive, 609340 (3), Autosomal recessive
FA2H	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
FTL	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
GTPBP2	607434	Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive
PANK2	606157	HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
PLA2G6	603604	Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive
REPS1	614825	?Neurodegeneration with brain iron accumulation 7, 617916 (3), Autosomal recessive
SCP2	184755	?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive

WDR45	300526	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: July 04, 2018

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.