Optic atrophy

Gene panel

Gene panel information

Gene panel	**Optic atrophy**
Version	3
Total genes	49
Activation date	Friday 27 september 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ACO2	99.99 %	100850	Optic atrophy 9, 616289 (3), Autosomal dominant, Autosomal recessive; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive
AFG3L2	99.97 %	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
ATAD3A	99.62 %	612316	Harel-Yoon syndrome, 617183 (3), Autosomal dominant, Autosomal recessive; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
ATG7	99.9 %	608760	Spinocerebellar ataxia, autosomal recessive 31, 619422 (3), Autosomal recessive
ATP1A3	99.98 %	182350	Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Developmental and epileptic encephalopathy 99, 619606 (3), Autosomal dominant
AUH	99.95 %	600529	3-methylglutaconic aciduria, type I, 250950 (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
CACNA1F	99.94 %	300110	Cone-rod dystrophy, X-linked, 3, 300476 (3), X-linked recessive; Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 (3), X-linked; Aland Island eye disease, 300600 (3), X-linked
CISD2	96.27 %	611507	Wolfram syndrome 2, 604928 (3), Autosomal recessive
COQ2	99.9 %	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
DHX16	99.98 %	603405	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733 (3), Autosomal dominant
DNAJC19	99.76 %	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
DNAJC30	100 %	618202	Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382 (3), Autosomal recessive
DNM1L	99.4 %	603850	Optic atrophy 5, 610708 (3), Autosomal dominant; Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 (3), Autosomal dominant, Autosomal recessive
EPRS1	99.53 %	138295	Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive
FDXR	99.99 %	103270	Multiple mitochondrial dysfunctions syndrome 9B, 620887 (3); Auditory neuropathy and optic atrophy, 617717 (3), Autosomal recessive
ISCA2	100 %	615317	Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive
KLC2	100 %	611729	Spastic paraplegia, optic atrophy, and neuropathy, 609541 (3), Autosomal recessive

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MCAT	99.99 %	614479	Optic atrophy 15, 620583 (3), Autosomal recessive
MECR	99.63 %	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive; Optic atrophy 16, 620629 (3), Autosomal recessive
MFF	99.97 %	614785	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 (3), Autosomal recessive
MFN2	99.98 %	608507	Lipomatosis, multiple symmetric, with or without peripheral neuropathy, 151800 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 (3), Autosomal recessive; Hereditary motor and sensory neuropathy VIA, 601152 (3), Autosomal dominant
MIEF1	99.99 %	615497	Optic atrophy 14, 620550 (3), Autosomal dominant
MTPAP	99.97 %	613669	?Spastic ataxia 4, autosomal recessive, 613672 (3), Autosomal recessive
MTRFR	99.87 %	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
NBAS	99.86 %	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive
NDUFA12	99.21 %	614530	Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive
NDUFS1	99.79 %	157655	Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive
NDUFS2	99.66 %	602985	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 6, 618228 (3), Autosomal recessive
NR2F1	99.99 %	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant
OPA1	99.95 %	605290	Optic atrophy plus syndrome, 125250 (3), Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3); Optic atrophy 1, 165500 (3), Autosomal dominant; Behr syndrome, 210000 (3), Autosomal recessive; ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3), Autosomal recessive
OPA3	100 %	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
PDSS1	95.7 %	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
POLG	100 %	174763	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
RTN4IP1	99.98 %	610502	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732 (3), Autosomal recessive
SLC25A46	99.88 %	610826	Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 1E, 619303 (3), Autosomal recessive
SLC44A1	99.9 %	606105	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 (3), Autosomal recessive
SLC52A2	100 %	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive
SNF8	99.78 %	610904	Developmental and epileptic encephalopathy 115, 620783 (3), Autosomal recessive; Neurodevelopmental disorder plus optic atrophy, 620784 (3), Autosomal recessive
SPG7	99.99 %	602783	Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal dominant, Autosomal recessive

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SSBP1	100 %	600439	Optic atrophy 13 with retinal and foveal abnormalities, 165510 (3), Autosomal dominant
TFG	98.68 %	602498	?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive; Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant
TIMM8A	100 %	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
TMEM126A	99.88 %	612988	Optic atrophy 7, 612989 (3), Autosomal recessive
TSFM	100 %	604723	Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive
UCHL1	99.99 %	191342	{?Parkinson disease 5, susceptibility to}, 613643 (3), Autosomal dominant; Spastic paraplegia 79A, autosomal dominant, 620221 (3), Autosomal dominant; Spastic paraplegia 79B, autosomal recessive, 615491 (3), Autosomal recessive
WFS1	99.99 %	606201	Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive
YME1L1	99.87 %	607472	?Optic atrophy 11, 617302 (3), Autosomal recessive
ZNHIT3	62.89 %	604500	PEHO syndrome, 260565 (3), Autosomal recessive

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.