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HEREDITARY OPTIC NEUROPATHIES PANEL DG-5.1.0 (34 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
ACO2	90.7%	86.8%	100%	100%	99.7%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559
AFG3L2	94.2%	92.2%	100%	100%	99.5%	Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246
CISD2	100%	100%	100%	99.9%	99.4%	Wolfram syndrome 2, 604928
DNAJC30	100%	100%	100%	100%	99.7%	Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382
DNM1L	100%	100%	100%	100%	99.9%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNMT1	100%	99.3%	100%	100%	99.4%	Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
FDXR	100%	100%	100%	100%	99.4%	Multiple mitochondrial dysfunctions syndrome 9B, 620887;Auditory neuropathy and optic atrophy, 617717

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IBA57	100%	100%	100%	100%	99.5%	Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451
MCAT	95.9%	95.9%	100%	100%	99.6%	Optic atrophy 15, 620583
MECR	93.5%	93.5%	100%	100%	99.6%	?Optic atrophy 16, 620629;Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MIEF1	100%	100%	100%	100%	99.3%	Optic atrophy 14, 620550
NBAS	100%	100%	100%	100%	99.7%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483
NDUFA10	82.9%	80.5%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA12	80.4%	80.4%	100%	99.9%	99.7%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFAF2	67.4%	67.4%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF4	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	90.9%	90.9%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF8	100%	100%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFS1	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 5, 618226

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NDUFS2	99.9%	98.3%	100%	100%	99.6%	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS7	89.1%	85%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFV1	100%	100%	100%	100%	98.7%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100%	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NSUN3	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 48, 619012
OPA1	94.7%	94.7%	100%	100%	99.7%	Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;Mitochondrial DNA depletion syndrome 14A (encephalomyopathic type), 621481;?Mitochondrial DNA depletion syndrome 14B (cardioencephalomyopathic type), 616896
OPA3	100%	100%	100%	100%	99.6%	3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300
PPIB	100%	100%	100%	100%	99.6%	Osteogenesis imperfecta, type IX, 259440

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RTN4IP1	100%	100%	100%	100%	99.8%	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732
SNX10	89.3%	89.3%	100%	100%	99.6%	Osteopetrosis, autosomal recessive 8, 615085
SPG7	100%	100%	100%	99.9%	99.3%	Spastic paraplegia 7, autosomal recessive, 607259
SSBP1	100%	100%	100%	100%	100%	Optic atrophy 13 with retinal and foveal abnormalities, 165510
TMEM126A	100%	100%	100%	100%	100%	Optic atrophy 7, 612989
WFS1	91.5%	90.5%	100%	100%	99.4%	Deafness, autosomal dominant 6/14/38, 600965;?Cataract 41, 116400;Wolfram-like syndrome, autosomal dominant, 614296;{Diabetes mellitus, noninsulin-dependent, association with}, 125853;Wolfram syndrome 1, 222300
YME1L1	96.7%	96.7%	100%	100%	99.7%	?Optic atrophy 11, 617302

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors