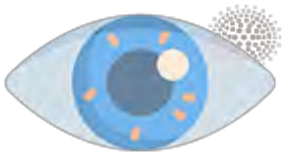
Optic Atrophy

Precision Panel



Overview

Optic atrophy is the clinical manifestation of any disease process causing axon degeneration in the retinogeniculate pathway. Particularly, these diseases affect the retinal ganglion cells and their axons forming the optic nerve, which are in charge of transferring the visual information from the photoreceptors to the lateral geniculus in the brain. Optic atrophy is clinically seen as changes in the color and the structure of the optic disc associated with variable degrees of visual dysfunction starting typically in the first decade of life. It is a relative common form of inherited optic neuropathy and it may be syndromic which can include extra-ocular symptoms mostly neuromuscular. The mode of inheritance varies from mitochondrial, autosomal dominant and recessive.

The Igenomix Optic Atrophy Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of progressive blindness ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Optic Atrophy Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Blurred vision
- Difficulties with peripheral vision
- Difficulties with color vision
- Reduction in the sharpness of vision

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team in the form of regular ophthalmologic examination, low vision aids and preventive measures such as avoidance of alcohol, tobacco and some medications.

- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

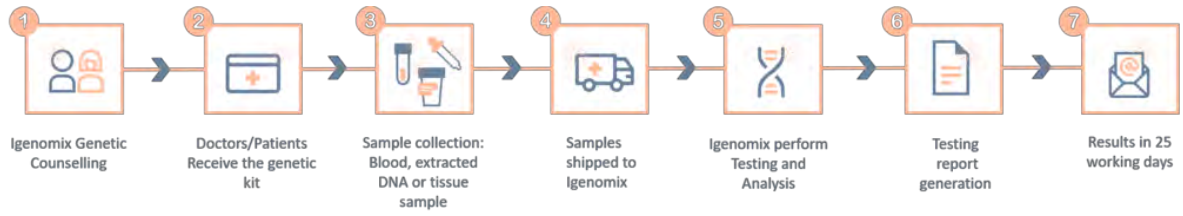
Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ACO2	Cerebellar-Retinal Degeneration, Optic Atrophy	AR	100	33 of 33
AFG3L2	Optic Atrophy, Spinocerebellar And Spastic Ataxia, Myoclonic Epilepsy, Neuropathy	AD,AR	99.74	42 of 42
ATP6	Leber Optic Atrophy, Neuropathy, Ataxia, Retinitis Pigmentosa, Striatal Necrosis, Leigh Syndrome, Spastic Paraplegia, Narp Syndrome	MI	-	-
ATP8	Kearns-Sayre Syndrome	-	98.02	-
AUH	3-Methylglutaconic Aciduria	AR	99.99	11 of 11
C12ORF65	Oxidative Phosphorylation Deficiency, Spastic Paraplegia	AR	-	-
C19ORF12	Neurodegeneration, Brain Iron Accumulation, Spastic Paraplegia, Mitochondrial Membrane Protein-Associated Neurodegeneration	AD,AR	-	-
CISD2	Wolfram Syndrome	AR	92.92	5 of 5
COX3	Leber Optic Atrophy, Leber Hereditary Optic Neuropathy, MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes)	MI	-	-
CYTB	Leber Optic Atrophy, MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), Histiocytoid Cardiomyopathy	MI	98.8	-
DNAJC19	3-Methylglutaconic Aciduria, Dilated Cardiomyopathy, Ataxia	AR	100	6 of 6
DNM1L	Encephalopathy, Optic Atrophy	AD,AR	100	29 of 29
FDXR	Neuropathy, Optic Atrophy, Ataxia, Global Developmental Delay	AR	99.93	23 of 23
MECR	Dystonia, Optic Atrophy, Basal Ganglia Abnormalities	AR	100	8 of 8
MFN2	Charcot-Marie-Tooth Disease, Motor And Sensory Neuropathy, Multiple Symmetric Lipomatosis	AD,AR	100	233 of 233
MT-CO1	Myoglobinuria, Mitochondrial Complex Iv Deficiency, Sideroblastic Anemia, Deafness	-	97.64	-
MT-CO2	Colorectal Cancer, Tetralogy Of Fallot, MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes)	-	99.19	-
MT-ND1	MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), Optic Neuropathy, Alzheimer, Sudden Infant Death Syndrome	-	98.8	-
MTPAP	Spastic Ataxia, Optic Atrophy, Dysarthria Syndrome	AR	99.99	2 of 2
ND2	Leber Optic Atrophy, Complex I Deficiency, Leigh Syndrome	MI	85.56	-
ND3	Complex I Deficiency, Leigh Syndrome	-	99.99	-
ND4	Leber Optic Atrophy, MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), Leigh Syndrome	MI	-	-
ND4L	Leber Optic Atrophy	MI	99.83	-
ND5	Leber Optic Atrophy, MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), MERRF (Mioclonic Epilepsy With Ragged-Red Fibres), Leigh Syndrome	MI	99.89	-
ND6	Leber Optic Atrophy, MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), Leigh Syndrome	MI	100	-
NDUFS1	Complex I Deficiency, Leigh Syndrome, Leukodystrophy	AR	99.98	30 of 30
NR2F1	Bosch-Boonstra Optic Atrophy Syndrome, Intellectual Disability	AD	89.78	26 of 31
OPA1	Behr Syndrome, DNA Depletion Syndrome, Optic Atrophy, Deafness, Ophthalmoplegia, Myopathy, Ataxia, Neuropathy	AD,AR	99.98	397 of 402

OPA3	Optic Atrophy, 3-Methylglutaconic Aciduria, Cataract	AD,AR	100	18 of 18
POLG	DNA Depletion Syndrome, Ophthalmoplegia, Sensory Ataxic Neuropathy, Dysarthria, Alpers-Huttenlocher Syndrome, Neurogastrointestinal Encephalomyopathy	AD,AR	99.92	325 of 326
RTN4IP1	Optic Atrophy, Ataxia, Mental Retardation, Seizures	AR	99.91	14 of 14
SLC24A1	Night Blindness	AR	99.23	26 of 26
SLC25A46	Neuropathy	AR	99.79	16 of 17
SLC52A2	Brown-Vialetto-Van Laere Syndrome, Spinocerebellar Ataxia, Blindness, Deafness	AR	100	31 of 32
SNX10	Osteopetrosis	AR	100	14 of 14
SPG7	Spastic Paraplegia, Primary Lateral Sclerosis	AD,AR	99.94	125 of 126
TIMM8A	Mohr-Tranebjaerg Syndrome	X,XR,G	100	-
TMEM126A	Optic Atrophy, Auditory Neuropathy	AR	100	4 of 4
TRNC	MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes)	MI	-	-
TRNE	Diabetes, Deafness, Myopathy, Cytochrome C Oxidase Deficiency	-	-	-
TRNF	MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), MERRF (Mioclonic Epilepsy With Ragged-Red Fibres)	MI	-	-
TRNH	MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), MERRF (Mioclonic Epilepsy With Ragged-Red Fibres)	-	-	-
TRNI	MERRF (Mioclonic Epilepsy With Ragged-Red Fibres)	MI	-	-
TRNK	Maternally-Inherited Diabetes And Deafness, Leigh Syndrome , Cardiomyopathy, Hearing Loss, MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), MERRF (Mioclonic Epilepsy With Ragged-Red Fibres)	MI	-	-
TRNL1	MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), MERRF (Mioclonic Epilepsy With Ragged-Red Fibres), Kearns-Sayre Syndrome, Diabetes, Deafness, Leigh Syndrome, Ophthalmoplegia	MI	-	-
TRNL2	Ophthalmoplegia	-	-	-
TRNN	Mitochondrial Complex Iv Deficiency, Ophthalmoplegia	AR,MI	-	-
TRNQ	MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), MERRF (Mioclonic Epilepsy With Ragged-Red Fibres)	MI	-	-
TRNS1	Deafness, Mitochondrial Complex Iv Deficiency, Ophthalmoplegia, Palmoplantar Keratoderma, MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), MERRF (Mioclonic Epilepsy With Ragged-Red Fibres)	AR,MI	-	-
TRNS2	MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), MERRF (Mioclonic Epilepsy With Ragged-Red Fibres), Usher Syndrome	MI	-	-
TRNT	Lethal Infantile Mitochondrial Myopathy	MI	-	-
TRNV	MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes), Leigh Syndrome	MI	-	-
TRNW	Optic Atrophy, Leigh Syndrome, MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke-Like Episodes)	AR,MI	-	-
TSFM	Oxidative Phosphorylation Deficiency	AR	93.35	11 of 14
UCHL1	Neurodegeneration, Optic Atrophy, Parkinson Disease	AD,AR	100	5 of 5
WFS1	Cataract, Deafness, Diabetes Mellitus, Wolfram Syndrome	AD,AR	99.97	390 of 395
YME1L1	Optic Atrophy	AR	99.98	1 of 1
ZNHIT3	Peho Syndrome	AR	73.96	1 of 1

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology



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Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

References

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