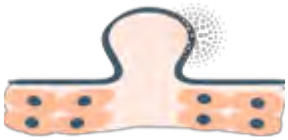
Oculocutaneous Albinism

Precision Panel



Overview

Albinism is a group of inherited abnormalities of melanin synthesis and are characterized by a decrease or absence of melanin pigment. There are several types of albinism, one of them being oculocutaneous albinism (OCA). OCA is an autosomal recessive disease of melanin biosynthesis which leads to complete or partial loss of melanin in the skin, hair follicles and eyes. This is due to mutations in genes encoding for enzymes responsible for melanin synthesis. The clinical symptoms and the course of the disease show a pronounced variability, this is due to different gene mutations affecting various points along the melanin pathway. Some of the manifestations include nystagmus, iris hypopigmentation and translucency, reduced pigmentation of the retina, reduced visual acuity etc. They are prone to deleterious effects of ultraviolet light, and therefore increased susceptibility to develop actinic keratosis, squamous cell carcinoma and basal cell carcinoma.

The Igenomix Oculocutaneous Albinism Precision Panel can be used to make an accurate and directed diagnosis 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 Oculocutaneous Albinism Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Pinkish-coloured skin
- White hair
- Blue-gray irides
- Prominent red reflex
- Poor visual acuity
- Photophobia
- Nystagmus
- Foveal hypoplasia

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient. Patient education on preventive sun exposure measures.

- Early initiation of protection from sunlight in form of sunscreen and sun-avoidance methods. Surveillance for early detection of neoplasms and ophthalmology consultations.
- 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**
AP3B1	Hermansky-Pudlak Syndrome	AR	100	34 of 35
AP3D1	Hermansky-Pudlak Syndrome, Ocular Albinism	AR	99.69	5 of 5
BLOC1S3	Hermansky-Pudlak Syndrome	AR	97.87	2 of 4
BLOC1S6	Hermansky-Pudlak Syndrome	AR	99.48	2 of 2
DTNBP1	Hermansky-Pudlak Syndrome	AR	99.96	4 of 4
EPG5	Immunodeficiency With Cleft Lip/Palate, Cataract, Hypopigmentation, And Absent Corpus Callosum, Vici Syndrome	AR	98.98	73 of 73
GPR143	Ocular Albinism, Nystagmus, X-linked Recessive Ocular Albinism	X,XR,G	96.52	-
HPS1	Hermansky-Pudlak Syndrome	AR	99.98	68 of 68
HPS3	Hermansky-Pudlak Syndrome	AR	99.92	20 of 21
HPS4	Hermansky-Pudlak Syndrome	AR	99.7	30 of 30
HPS5	Hermansky-Pudlak Syndrome	AR	99.88	32 of 32
HPS6	Hermansky-Pudlak Syndrome	AR	99.87	39 of 41
LRMDA	Oculocutaneous Albinism	AR	-	-
LYST	Chediak-Higashi Syndrome	AR	99.98	117 of 117
MC1R	Oculocutaneous Albinism, Familial Melanoma, Large Congenital Melanocytic Nevus	AR	-	-
MITF	Coloboma, Osteopetrosis, Microphthalmia, Macrocephaly, Albinism, And Deafness, Tietz Syndrome, Waardenburg Syndrome	AD,AR	100	72 of 72
MLPH	Griscelli Syndrome	AR	100	7 of 7
MYO5A	Griscelli Syndrome, Neuroectodermal Melanolysosomal Disease	AR	100	10 of 10
OCA2	Oculocutaneous Albinism	AR	100	310 of 312
RAB27A	Griscelli Syndrome	AR	100	54 of 55
SLC24A5	Oculocutaneous Albinism	AR	99.75	28 of 28
SLC38A8	Foveal Hypoplasia	AR	100	16 of 16
SLC45A2	Oculocutaneous Albinism	AR	100	157 of 159
TYR	Oculocutaneous Albinism	AR	99.77	437 of 455
TYRP1	Oculocutaneous Albinism	AR	100	64 of 64

*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

Contact us

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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