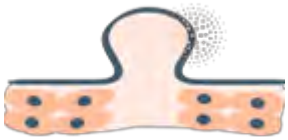
Xeroderma Pigmentosum

Precision Panel



Overview

Xeroderma Pigmentosum (XP) is a rare autosomal recessive disorder characterized by photosensitivity with easy skin burning following minimal sun exposure, pigmentary changes, premature skin aging and predisposition to malignant tumor development. These features result from a defect in DNA repair mechanisms after exposure to ultraviolet (UV) radiation. In addition to cutaneous complications, patients are prone to suffer from eye conditions, neurodegenerative processes and central nervous system tumors. Less than 40% of patients survive beyond age 20 years.

The Igenomix Xeroderma Pigmentosum Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of persistent sunburns 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 Xeroderma Pigmentosum Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Persistent sunburn
- Diffuse erythema
- Scaling of the skin
- Freckle-like areas of increased pigmentation
- A decreased appearance of sunburns, scaling and freckles during winter
- Skin atrophy

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**
DDB2	Xeroderma Pigmentosum, Complementation Group E	AR	100	17 of 17
ERCC1	Cerebrooculofacioskeletal Syndrome, Cockayne Syndrome Type 2	AR	93.12	6 of 6
ERCC2	Cerebrooculofacioskeletal Syndrome, Photosensitive Trichothiodystrophy, Xeroderma Pigmentosum Complementation Group D Trichothiodystrophy, Xeroderma Pigmentosum, Xeroderma Pigmentosum-Cockayne Syndrome Complex	AR	100	102 of 102
ERCC3	Photosensitive Trichothiodystrophy, Xeroderma Pigmentosum Complementation Group B, Xeroderma Pigmentosum-Cockayne Syndrome Complex	AR	99.98	24 of 24
ERCC4	Fanconi Anemia Complementation Group Q, Xeroderma Pigmentosum Complementation Group, Progeroid Syndrome, Cockayne Syndrome Type 1	AR	99.68	69 of 72
ERCC5	Cerebrooculofacioskeletal Syndrome, Xeroderma Pigmentosum Complementation Group G, Xeroderma Pigmentosum-Cockayne Syndrome Complex	AR	99.94	58 of 58
POLH	Xeroderma Pigmentosum Variant	AR	99.49	73 of 76
XPA	Xeroderma Pigmentosum Complementation Group A	AR	99.91	49 of 49
XPC	Xeroderma Pigmentosum Complementation Group C	AR	99.83	86 of 87

*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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