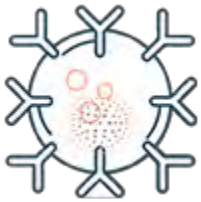
Dyskeratosis Congenita

Precision Panel



Overview

Dyskeratosis Congenita (DKC) is a rare, progressive bone marrow failure syndrome characterized by reticulated skin hyperpigmentation, nail dystrophy and oral leukoplakia. Patients usually present with symptoms of skin hyperpigmentation and nail changes during the first decade of life. It is caused by germline mutations in genes regulating telomere maintenance, resulting in very short telomeres. DKC is a genetically heterogeneous with X-linked recessive form being the most common, autosomal dominant and autosomal recessive subtypes based on different patters of inheritance. Early mortality is associated with bone marrow failure, infections, lung and pulmonary complications as well as malignancy.

The Igenomix Dyskeratosis Congenita Precision Panel can be used for an accurate and directed diagnosis as well as differential diagnosis of reticulate pigmentary disorders 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 Dyskeratosis Congenita Precision Panel is used for patients with a clinical diagnosis or suspicion with or without the following symptoms:

- Abnormal skin pigmentation (tan-to-gray hyperpigmented or hypopigmented macules and patches)
- Nail dystrophy
- Skin atrophy and telangiectasia
- Alopecia of the skin, eyebrows and eyelashes
- Mucosal leukoplakia
- Bone marrow failure
- Dental manifestations

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.

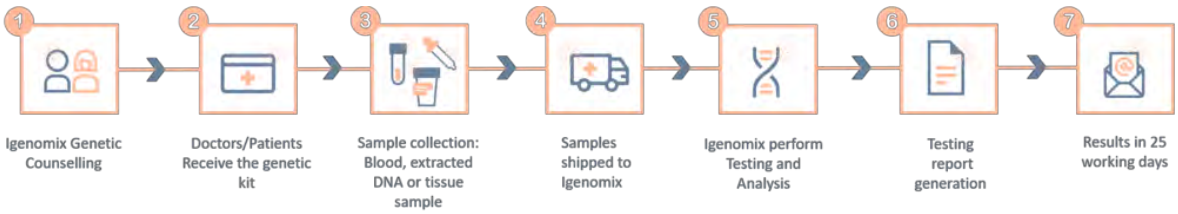
- Early initiation of treatment involving a multidisciplinary team in the form of hematopoietic stem cell transplantation as well as medical care to prevent complications and early surveillance of malignancy.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ACD	Dyskeratosis Congenita, Hoyeraal-Hreidarsson Syndrome, Familial Melanoma	AD,AR	99.89	14 of 14
CTC1	Cerebroretinal Microangiopathy With Calcifications And Cysts, Dyskeratosis Congenita	AR	99.73	43 of 44
DKC1	Dyskeratosis Congenita, Hoyeraal-Hreidarsson Syndrome	X,XR,G	100	NA of NA
NHP2	Dyskeratosis Congenita	AR	100	3 of 3
NOP10	Dyskeratosis Congenita	AR	100	1 of 1
NPM1	Acute Myeloid Leukemia, Acute Promyelocytic Leukemia, Dyskeratosis Congenita	AD	99.89	2 of 2
PARN	Dyskeratosis Congenita, Pulmonary Fibrosis And/Or Bone Marrow Failure, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.98	33 of 33
RTEL1	Dyskeratosis Congenita, Pulmonary Fibrosis And/Or Bone Marrow Failure, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.73	127 of 131
TERC	Dyskeratosis Congenita, Pulmonary Fibrosis And/Or Bone Marrow Failure, Idiopathic Aplastic Anemia	AD	na	na
TERT	Aplastic Anemia, Dyskeratosis Congenita, Acute Myeloid Leukemia, Cutaneous Malignant Melanoma, Pulmonary Fibrosis And/Or Bone Marrow Failure, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.09	194 of 197
TINF2	Dyskeratosis Congenita, Revesz Syndrome, Hoyeraal-Hreidarsson Syndrome	AD	99.94	47 of 47
USB1	Poikiloderma With Neutropenia, Dyskeratosis Congenita	AR	100	24 of 24
WRAP53	Dyskeratosis Congenita	AR	100	10 of 10

*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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