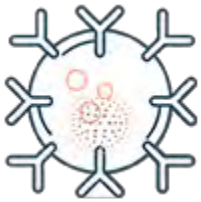
Chronic Granulomatous Disease

Precision Panel



Overview

Chronic Granulomatous Disease (CGD) is a genetically heterogeneous condition featuring recurrent, life-threatening bacterial and fungal infections as well as granuloma formation. CGD is caused by defects in the oxidative mechanisms found in phagocytes that are responsible for the destruction of certain microbes. Thus, these genetic defects result in a primary immunodeficiency caused by the inability of phagocytes to destroy foreign microbes. The characteristic infections of patients with CGD are those caused by catalase positive microorganism in the lung, skin, lymph nodes and liver. Symptoms typically appear during the first year of life in the form of infections, dermatitis, gastrointestinal complications and failure to thrive. This disease primarily affects males, and the mode of inheritance is X-linked.

The Igenomix Chronic Granulomatous Disease Precision Panel can be used for an accurate and directed diagnosis as well as differential diagnosis of recurrent infections 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 Chronic Granulomatous Disease Precision Panel is used for patients with a clinical diagnosis or suspicion with or without the following symptoms:

- Recurrent bacterial infections
- Recurrent fungal infections
- Growth failure
- Abnormal wound healing
- Diarrhea
- Granulomatous dermatitis
- Hepatomegaly
- Splenomegaly
- Lymphadenitis

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 antimicrobial prophylaxis with antibacterial, antifungal and immunomodulators, rapid recognition and treatment of infections as well as aggressive management of infectious complications. In case of multidrug refractoriness, life-threatening infections, hematopoietic stem cell transplantation (HSCT) represents a valid curative option.
- 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**
CYBA	Granulomatous Disease	AR	99.98	67 of 67
CYBB	Atypical Mycobacteriosis, Granulomatous Disease	X,XR,G	100	-
CYBC1	Granulomatous Disease	AR	-	-
G6PD	Anemia	X,XR,XD,G	100	-
NCF1	Granulomatous Disease	AR	74.19	31 of 39
NCF2	Granulomatous Disease	AR	100	72 of 73
NCF4	Granulomatous Disease	AR	100	14 of 14
NOD2	Inflammatory Bowel Disease, Granulomatous Synovitis, Cranial Neuropathies, Yao Syndrome, Blau Syndrome	AD,MU	100	97 of 97
XK	Mcleod Syndrome	X,G	99.97	-

*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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