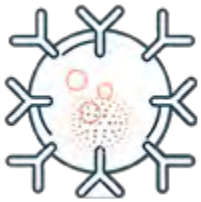
Congenital Neutropenia

Precision Panel



Overview

Neutropenia is a dangerous and potentially fatal condition that exposes patients to recurrent infections. Primary causes constitute a small portion of the whole and are mostly unknown. Congenital neutropenia is a primary immunodeficiency disorder associated with recurrent bacterial infections, auto-inflammatory and auto-immune phenomena, hematologic malignancy and neuro-psychiatric manifestations. It results from impaired maturation of neutrophil granulocytes and is associated with a variety of syndromic diseases including: oculocutaneous albinism, metabolic diseases and bone marrow failure syndromes. Congenital neutropenia is a genetically heterogeneous group of related disorders. It demonstrates several modes of inheritance, including autosomal recessive, autosomal dominant, sporadic and X-linked forms.

The Igenomix Congenital Neutropenia Precision Panel can be as a tool for an accurate and directed diagnosis as well as differential diagnosis of recurrent bacterial 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 Congenital Neutropenia Precision Panel is used for patients with a clinical diagnosis or suspicion with or without the following symptoms:

- Oral ulcers
- Gingivitis
- Pharyngitis
- Sinusitis, otitis media
- Lymphadenopathy, lymphadenitis
- Bronchitis, pneumonia
- Cellulitis
- Cutaneous abscess
- Abscesses
- Bacteremia and/or septicemia
- Urinary tract infection

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 focusing on preventive care of infections and other complications, symptomatic medical care for neurologic symptoms alongside early surveillance for cancer detection.
- Risk assessment of asymptomatic family members according to the mode of inheritance via genetic counselling and explanation of the multisystem nature of the disease.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AK2	Reticular Dysgenesia	AR	100	21 of 21
AP3B1	Hermansky-Pudlak Syndrome	AR	100	34 of 35
CD40LG	X-linked Immunodeficiency With Hyper-IgM	X,XR,G	100	-
CLPB	3-a Methylglutaconic Aciduria With Cataracts, Neurologic Involvement, And Neutropenia	AR	96	26 of 26
CSF3R	Severe Congenital Neutropenia	AR	99.99	19 of 19
CXCR2	Severe Congenital Neutropenia, Human Granulocytic Anaplasmosis	-	99.94	1 of 1
CXCR4	Whim Syndrome	AD	100	19 of 19
DNAJC21	Bone Marrow Failure Syndrome, Shwachman-Diamond Syndrome	AR	99.83	12 of 12
EFL1	Shwachman-Diamond Syndrome	AR	99.94	-
EIF2AK3	Multiple Epiphyseal Dysplasia With Early-Onset Diabetes Mellitus, Wolcott-Rallison Syndrome	AR	99.3	89 of 89
ELANE	Cyclic Hematopoiesis, Severe Congenital Neutropenia, Cyclic Neutropenia	AD	100	227 of 227
G6PC3	Severe Congenital Neutropenia	AR	100	45 of 45
GATA1	X-linked Anemia With Or Without Neutropenia And/Or Platelet Abnormalities, Down Syndrome Trisomy 21, Dyserythropoietic Anemia With Thrombocytopenia, Blackfan-Diamond Anemia, Congenital Erythropoietic Porphyria	X,XR,G	99.93	-
GATA2	Dendritic Cell, Monocyte, B Lymphocyte, And Natural Killer Lymphocyte Deficiency, Acute Myeloid Leukemia, Myelodysplastic Syndrome	AD	100	137 of 142
GFI1	Nonimmune Chronic Idiopathic Neutropenia, Severe Congenital Neutropenia	AD	98.77	4 of 4
HAX1	Severe Congenital Neutropenia	AR	100	22 of 23
HYOU1	Immunodeficiency And Hypoglycemia	AR	99.94	2 of 2
JAGN1	Severe Congenital Neutropenia	AR	99.95	10 of 10
LAMTOR2	Immunodeficiency Due To Defect In Mapbp-Interacting Protein, Primary Immunodeficiency Syndrome Due To Lamtor2 Deficiency	AR	100	1 of 1
LYST	Chediak-Higashi Syndrome	AR	99.98	117 of 117
MRTFA	Immunodeficiency	AR	99.8	-
RAB27A	Griscelli Syndrome	AR	100	54 of 55
RAC2	Immunodeficiency With Defective Neutrophil Chemotaxis And Lymphopenia, Neutrophil Immunodeficiency Syndrome	AD,AR	100	5 of 5
RMRP	Anauxetic Dysplasia, Cartilage-Hair Hypoplasia, Omenn Syndrome	AR	-	-
RUNX1	Acute Myeloid Leukemia, Platelet Disorder, Familial, With Associated Myeloid Malignancy, Aggressive Systemic Mastocytosis, Chronic Myeloid Leukemia	AD	99.83	90 of 90
SBDS	Aplastic Anemia, Shwachman-Diamond Syndrome, Idiopathic Aplastic Anemia	AR	100	77 of 79

SLC37A4	Glycogen Storage Disease	AR	99.97	112 of 112
SMARCD2	Specific Granule Deficiency	AR	91.58	1 of 1
SRP54	Severe Congenital Neutropenia, Shwachman-Diamond Syndrome	AD,AR	99.95	8 of 8
STK4	T-Cell Immunodeficiency, Recurrent Infections, And Autoimmunity With Or Without Cardiac Malformations	AR	99.88	10 of 10
TAZ	Barth Syndrome	X,XR,G	100	-
TCIRG1	Autosomal Dominant Severe Congenital Neutropenia	AR	100	140 of 146
TCN2	Transcobalamin Deficiency	AR	100	25 of 27
TP53	Bone Marrow Failure Syndrome, Li-Fraumeni Syndrome	AD,MU,P	98.92	557 of 563
USB1	Poikiloderma With Neutropenia, Dyskeratosis Congenita	AR	100	24 of 24
VPS13B	Cohen Syndrome	AR	99.98	182 of 190
VPS45	Severe Congenital Neutropenia	AR	100	4 of 4
WAS	Severe Congenital Neutropenia, Wiskott-Aldrich Syndrome	X,XR,G	100	-
WDR1	Periodic Fever, Immunodeficiency, And Thrombocytopenia Syndrome	AR	100	9 of 9
WIPF1	Wiskott-Aldrich Syndrome	AR	99.79	3 of 3

*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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