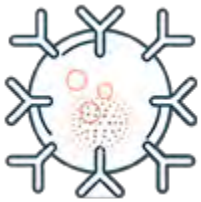
Severe Combined Immunodeficiency

Precision Panel



Overview

Severe Combined Immunodeficiency (SCID) is a genetically heterogeneous group of disorders resulting from genetic defects in both cellular and humoral immunity where there is an impaired lymphocyte development and function. Aside from lymphocytes other components of the innate and adaptive immune system such as neutrophils, macrophages, dendritic cells, complement proteins and natural killer cells are affected. Children present with bacterial, viral and fungal infections that begin during infancy and result in fatal outcome in the first few years of life if untreated. Early, accurate and precise diagnosis have enabled major advances in the care of infants with SCID, including better outcomes of allogeneic hematopoietic stem cell transplantation. The mode of inheritance of SCID determines the severity of the disease as different modes of inheritance will determine the immune cells that will be affected. Autosomal, sporadic or X-linked patterns may affect the neonate.

The Igenomix Severe Combined Immunodeficiency Precision Panel can be used for an accurate and directed diagnosis as well as differential diagnosis of early 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 Severe Combined Immunodeficiency Precision Panel is used for patients with a clinical diagnosis or suspicion with or without the following symptoms:

- Neonate with a family history of a known immunologic disorder
- Failure to thrive
- Recurrent upper and lower respiratory tract infections that do not respond to antibiotics
- Recurrent skin infections and delayed wound healing
- Underdeveloped lymphoid tissue

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 haematopoietic stem cell transplantation, gene therapy and enzyme replacement.
- Risk assessment of asymptomatic family members according to the mode of inheritance via genetic counselling.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ADA	Autosomal Recessive Severe Combined Immunodeficiency, Omenn Syndrome	AR	100	97 of 98
AK2	Reticular Dysgenesia	AR	100	21 of 21
ATM	Ataxia-Telangiectasia	AD,AR	99.93	1608 of 1632
BCL11B	Immunodeficiency, Intellectual Developmental Disorder With Speech Delay, Dysmorphic Facies, And T-Cell Abnormalities	AD	96.06	12 of 12
CARD11	B-Cell Expansion With Nfkb And T-Cell Anergy, Card11 Immunodeficiency	AD,AR	100	30 of 31
CD247	Immunodeficiency Due To Defect In Cd3-zeta, Oligoarticular Juvenile Idiopathic Arthritis	AR	100	4 of 4
CD3D	Immunodeficiency, Severe Combined Immunodeficiency Due To Cd3delta/Cd3epsilon/Cd3zeta	AR	100	7 of 7
CD3E	Immunodeficiency, Severe Combined Immunodeficiency Due To Cd3delta/Cd3epsilon/Cd3zeta	AR	99.95	9 of 9
CHD7	Charge Syndrome, Kallmann Syndrome, Normosmic Congenital Hypogonadotropic Hypogonadism, Omenn Syndrome	AD	96.25	823 of 896
CORO1A	Immunodeficiency	AR	93	9 of 9
DCLRE1C	Omenn Syndrome, Severe Combined Immunodeficiency With Sensitivity To Ionizing Radiation, Omenn Syndrome, Severe Combined Immunodeficiency Due To Dclre1c Deficiency	AR	99.99	72 of 73
DOCK8	Hyperimmunoglobulin E-Recurrent Infection Syndrome, Combined Immunodeficiency Due To Dock8 Deficiency	AR	99.92	106 of 114
EXTL3	Skeletal Dysplasia-T-Cell Immunodeficiency-Developmental Delay Syndrome	AR	99.99	10 of 10
FOXN1	T-cell Immunodeficiency, Congenital Alopecia, And Nail Dystrophy, Infantile T-Cell Lymphopenia, Infantile, Severe Combined Immunodeficiency Due To Foxn1 Deficiency	AD,AR	100	30 of 30
IKBKB	Immunodeficiency	AD,AR	100	9 of 9
IL2RG	X-linked Combined Immunodeficiency, Omenn Syndrome, Severe Combined Immunodeficiency Due To Gamma Chain Deficiency	X,XR,G	99.86	-
IL7R	Autosomal Recessive Severe Combined Immunodeficiency, Omenn Syndrome	AR	100	54 of 55

JAK3	Autosomal Recessive Severe Combined Immunodeficiency, Severe Combined Immunodeficiency Due To Jak3 Deficiency	AR	99.98	86 of 88
LAT	Immunodeficiency	AR	100	3 of 3
LIG4	Lig4 Syndrome, Multiple Myeloma, Dubowitz Syndrome	AR	99.48	46 of 46
MTHFD1	Combined Immunodeficiency And Megaloblastic Anemia With Or Without Hyperhomocysteinemia	AR	99.94	11 of 12
NHEJ1	Cernunnos-Xlf Deficiency	-	100	12 of 14
ORAI1	Immunodeficiency, Tubular Aggregate Myopathy, Stormorken-Sjaastad-Langslet Syndrome	AD,AR	91.93	20 of 22
PGM3	Immunodeficiency	AR	99.99	17 of 17
PNP	Immunodeficiency Due To Purine Nucleoside Phosphorylase Deficiency	AR	99.73	39 of 39
PRKDC	Immunodeficiency With Or Without Neurologic Abnormalities	AR	99.74	9 of 10
PTPRC	Autosomal Recessive Severe Combined Immunodeficiency	AR	99.98	7 of 7
RAC2	Immunodeficiency With Defective Neutrophil Chemotaxis And Lymphopenia, Neutrophil Immunodeficiency Syndrome	AD,AR	100	5 of 5
RAG1	Combined Cellular And Humoral Immune Defects With Granulomas, Omenn Syndrome, Autosomal Recessive Severe Combined Immunodeficiency, Combined Immunodeficiency Due To Partial Rag1 Deficiency	AR	100	193 of 193
RAG2	Combined Cellular And Humoral Immune Defects With Granulomas, Omenn Syndrome, Autosomal Recessive Severe Combined Immunodeficiency, Severe Combined Immunodeficiency Due To Complete Rag1/2 Deficiency	AR	100	90 of 91
RMRP	Anauxetic Dysplasia, Cartilage-Hair Hypoplasia, Metaphyseal Dysplasia Without Hypotrichosis, Omenn Syndrome	AR	-	-
STAT1	Immunodeficiency, Mycobacterial And Viral Infections, Autoimmune Enteropathy And Endocrinopathy-Susceptibility To Chronic Infections Syndrome	AD,AR	100	138 of 138
STIM1	Immune Dysfunction With T-Cell Inactivation Due To Calcium Entry Defect,Tubular Aggregate Myopathy, Stormorken Syndrome	AD,AR	100	28 of 28
TBX1	Digeorge Syndrome, Velocardiofacial Syndrome	AD,AR	88.7	35 of 42
TTC7A	Gastrointestinal Defects And Immunodeficiency Syndrome, Combined Immunodeficiency-Enteropathy Spectrum	AR	100	44 of 45
XRCC4	Short Stature, Microcephaly, And Endocrine Dysfunction, Lig4 Syndrome, Microcephalic Primordial Dwarfism-Insulin Resistance Syndrome	AR	99.73	10 of 10
ZAP70	Severe Combined Immunodeficiency, Combined Immunodeficiency Due To Zap70 Deficiency	AR	99.99	30 of 30

*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

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Methodology

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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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