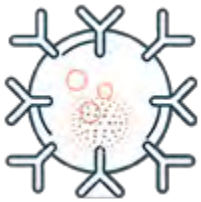
Primary Immunodeficiency

Precision Panel



Overview

Primary Immunodeficiencies are a growing group of over 400 inborn errors of immunity that range in severity from life-threatening disorders presenting in infancy to less severe disorders diagnosed in adulthood. Most patients with primary immunodeficiencies present with recurrent or chronic infections. Some disorders impact essential immunologic pathways and result in susceptibility to opportunistic organisms, whereas other disorders may cause susceptibility to a very narrow number of pathogens with a broad age of presentation. The clinical presentation is variable and includes severe or unusual infections, autoimmune diseases and malignanciesPatients with many forms of primary immunodeficiencies are at increased risk for malignancies secondary to a number of different factors, including immune dysregulation, genetic predisposition, radiation sensitivity and impaired viral clearance.

The Igenomix Primary Immunodeficiency 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 Primary Immunodeficiency Precision Panel is used for patients with a clinical diagnosis or suspicion with or without the following symptoms:

- Frequent and recurrent pneumonia, bronchitis, sinus infections, ear infections, meningitis or skin infections
- Inflammation and infection of internal organs
- Blood disorders
- Digestive problems such as cramping, loss of appetite, nausea and diarrhea
- Delayed growth and development
- Autoimmune disorders
- Family history of primary immunodeficiency

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, antifungals, rapid recognition and treatment of infections as well as aggressive management of infectious complications. Early continuous surveillance due to increased risk of malignancies.
- Possibility of early immunologic reconstitution in the form of hematopoietic cell transplantation (HCT), enzyme replacement, thymic transplantation or gene therapies.
- 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, Familial Melanoma, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.89	14 of 14
ACP5	Immunodeficiency, Autoimmunity, Spondyloenchondrodysplasia	AR	100	27 of 28
ACTB	Baraitser-Winter Syndrome, Dystonia, Becker Nevus Syndrome, Developmental Malformations, Deafness	AD	100	40 of 40
ADA	Immunodeficiency, Omenn Syndrome	AR	100	97 of 98
ADA2	Polyarteritis Nodosa, Sneddon Syndrome, Blackfan-Diamond Anemia	AR	100	-
AGL	Glycogen Storage Disease	AR	100	253 of 253
AICDA	Immunodeficiency	AR	99.94	48 of 48
AK2	Reticular Dysgenesis	AR	100	21 of 21
AKT1	Breast Cancer, Colorectal Cancer, Cowden Syndrome, Proteus Syndrome, Meningioma	AD	100	6 of 6
ANTXR2	Systemic Hyalinosis	AR	99.96	53 of 53
AP3D1	Hermansky-Pudlak Syndrome, Ocular Albinism	AR	99.69	5 of 5
ARHGEF1	Immunodeficiency	AR	90.23	2 of 2
ARVCF	22q11.2 Deletion Syndrome	-	99.95	2 of 2
ATM	Ataxia-Telangiectasia, Breast Cancer, Mantle Cell Lymphoma	AD,AR	99.93	1608 of 1632
ATP6AP1	Immunodeficiency	X,XR,G	99.2	-
ATRX	Mental Retardation, Alpha-Thalassemia-Myelodysplastic Syndrome, Carpenter-Waziri Syndrome, Chudley-Lowry-Hoar Syndrome, Holmes-Gang Syndrome, Juberg-Marsidi Syndrome, Neuroendocrine Tumor Of Stomach, Smith-Fineman-Myers Syndrome	X,XR,XD,G	98.5	-
BACH2	Immunodeficiency	AD	99.89	2 of 2
BCL10	Immunodeficiency, Lymphoma Non-Hodgkin, Mesothelioma, Testicular Tumor, Malt Lymphoma	AR	99.47	1 of 1
BCL11B	Immunodeficiency, Intellectual Developmental Disorder, Speech Delay, Dysmorphic Facies, T-Cell Abnormalities	AD	96.06	12 of 12
BCR	Chronic Myeloid Leukemia, 22q11.2 Microdeletion Syndrome	MU,P	97.78	-
BLNK	Agammaglobulinemia	AR	97.97	6 of 6
BTK	Agammaglobulinemia, Hypogammaglobulinemia, Growth Hormone Deficiency	X,XR,G	100	-
BUB1B	Colorectal Cancer, Mosaic Variegated Aneuploidy Syndrome	AD,AR	99.84	30 of 31
CARD11	Immunodeficiency, Atopic Dermatitis	AD,AR	100	30 of 31
CARD9	Candidiasis, Immunodeficiency	AR	99.86	34 of 34

CARMIL2	Immunodeficiency	AR	96.16	-
CCDC47	Trichohepatoneurodevelopmental Syndrome	AR	99.94	5 of 5
CD19	Immunodeficiency	AD,AR	99.99	7 of 7
CD247	Immunodeficiency, Oligoarticular Idiopathic Arthritis	AR	100	4 of 4
CD28	Mycosis Fungoides, Sézary Syndrome	-	99.03	-
CD3D	Immunodeficiency	AR	100	7 of 7
CD3E	Immunodeficiency	AR	99.95	9 of 9
CD3G	Immunodeficiency	AR	100	3 of 3
CD40	Immunodeficiency	AR	100	11 of 11
CD40LG	Immunodeficiency	X,XR,G	100	-
CD79A	Agammaglobulinemia	AR	99.99	8 of 8
CD79B	Agammaglobulinemia	AR	100	3 of 3
CD81	Immunodeficiency	AR	100	2 of 2
CDC42	Takenouchi-Kosaki Syndrome, Macrothrombocytopenia, Lymphedema	AD	99.97	10 of 10
CDCA7	Immunodeficiency, Centromeric Instability, Icf Syndrome	AR	99.89	6 of 6
CDH23	Deafness, Pituitary Adenoma, Usher Syndrome, Cushing Disease, Prolactinoma	AD,AR	98	400 of 403
CFTR	Bronchiectasis, Cystic Fibrosis, Pancreatitis, Aplasia Of Vas Deferens, Cystic Fibrosis, Male Infertility	AD,AR	95.45	1615 of 1730
CHD1	Pilarowski-Bjornsson Syndrome, Intellectual Disability, Autism, Speech Apraxia, Craniofacial Dysmorphism	AD	99.06	8 of 8
CHD7	Charge Syndrome, Hypogonadotropic Hypogonadism, Kallmann Syndrome, Omenn Syndrome	AD	96.25	823 of 896
CIITA	Bare Lymphocyte Syndrome, Rheumatoid Arthritis, Immunodeficiency	AR	98.51	15 of 16
CLCA4	Cystic Fibrosis	-	97.66	-
COMT	Panic Disorder, Schizophrenia, 22q11.2 Deletion Syndrome	AD	99.98	5 of 5
CORO1A	Immunodeficiency	AR	93	9 of 9
CPLX1	Myoclonic Epilepsy, Epileptic Encephalopathy, Wolf-Hirschhorn Syndrome	AD,AR	99.81	3 of 3
CR2	Immunodeficiency	AD,AR	99.92	19 of 19
CREBBP	Menke-Hennekam Syndrome, Rubinstein-Taybi Syndrome	AD	100	318 of 318
CRKL	22q11.2 Microdeletion Syndrome	-	99.93	5 of 6
CTBP1	Hypotonia, Ataxia, Developmental Delay, Tooth Enamel Defect, Wolf-Hirschhorn Syndrome	AD	98.45	1 of 1
CTC1	Cerebroretinal Microangiopathy, Dyskeratosis Congenita	AR	99.73	43 of 44
CTLA4	Autoimmune Lymphoproliferative Syndrome, Hashimoto Thyroiditis, Lupus Erythematosus, Mycosis Fungoides, Granulomatosis, Polyangiitis, Sézary Syndrome	AD	99.97	60 of 60
CTPS1	Immunodeficiency	AR	100	4 of 4
CUL4B	Mental Retardation, Short Stature, Small Testes, Musclewasting, Tremor	X,XR,G	99.77	-
CYBA	Granulomatous Disease	AR	99.98	67 of 67
CYBB	Mycobacteriosis, Chronic Granulomatous Disease	X,XR,G	100	-
DCLRE1C	Omenn Syndrome, Immunodeficiency	AR	99.99	72 of 73
DCTN4	Cystic Fibrosis	-	100	1 of 1
DKC1	Dyskeratosis Congenita, Hoyeraal-Hreidarsson Syndrome	X,XR,G	100	-
DNAJC21	Bone Marrow Failure Syndrome, Shwachman-Diamond Syndrome	AR	99.83	12 of 12
DNMT3B	Immunodeficiency, Centromeric Instability, Facioscapulohumeral Dystrophy, Icf Syndrome	AR	100	59 of 59
DOCK2	Immunodeficiency	AR	100	11 of 11

DOCK8	Hyperimmunoglobulin-E Recurrent Infection Syndrome, Combined Immunodeficiency	AR	99.92	106 of 114
EFL1	Shwachman-Diamond Syndrome	AR	99.94	-
EP300	Colorectal Cancer, Menke-Hennekam Syndrome, Rubinstein-Taybi Syndrome	AD	100	109 of 109
EPG5	Immunodeficiency, Cleft Lip/Palate, Cataract, Hypopigmentation, Absent Corpus Callosum, Vici Syndrome	AR	98.98	73 of 73
EXTL3	Immunoskeletal Dysplasia, Neurodevelopmental Abnormalities, T-Cell Immunodeficiency	AR	99.99	10 of 10
FADD	Infections, Encephalopathy, Hepatic Dysfunction, Cardiovascular Malformations, Fadd-Related Immunodeficiency	AR	95.19	3 of 3
FCGR3A	Immunodeficiency	AR	99.63	1 of 1
FCN3	Immunodeficiency	AR	99.98	1 of 1
FGFRL1	Wolf-Hirschhorn Syndrome	AD	99.94	1 of 1
FOXN1	T-Cell Immunodeficiency, T-Cell Lymphopenia, Nail Dystrophy	AD,AR	100	30 of 30
FRAS1	Fraser Syndrome	AR	98.73	57 of 58
GATA1	Anemia, Neutropenia, Platelet Abnormalities, Down Syndrome, Dyserythropoietic Anemia, Thrombocytopenia, Hemolysis, Beta-Thalassemia, Blackfan-Diamond Anemia, Erythropoietic Porphyria	X,XR,G	99.93	-
GATA2	Dendritic Cell, Monocyte, B Lymphocyte, And Natural Killer Lymphocyte Deficiency, Leukemia, Lymphedema, Myelodysplastic Syndrome	AD	100	137 of 142
GINS1	Immunodeficiency	AR	99.87	5 of 5
GP1BB	Bernard-Soulier Syndrome, 22q11.2 Deletion Syndrome, Fetal And Neonatal Alloimmune Thrombocytopenia	AR	74.08	26 of 50
HBB	Alpha-Thalassemia, Beta-Thalassemia, Fetal Hemoglobin Quantitative Trait Locus, Heinz Body Anemias, Sickle Cell Anemia	AD,AR	100	753 of 789
HELLS	Immunodeficiency, Centromeric Instability, Facial Anomalies, Icf Syndrome	AR	99.48	7 of 7
HIRA	22q11.2 Deletion Syndrome	-	99.99	5 of 5
HYOU1	Immunodeficiency, Hypoglycemia	AR	99.94	2 of 2
ICOS	Immunodeficiency	AD,AR	100	4 of 5
IFNAR2	Immunodeficiency	AR	99.78	2 of 2
IFNG	Aplastic Anemia, Immunodeficiency, Mycobacteriosis, Tuberous Sclerosis	AD,AR	99.77	-
IFNGR1	Helicobacter Pylori Infection, Immunodeficiency	AD,AR	99.99	46 of 46
IFNGR2	Immunodeficiency	AR	96.6	18 of 20
IGHM	Agammaglobulinemia	AR	100	-
IGLL1	Agammaglobulinemia	AR	100	2 of 2
IKBKB	Immunodeficiency	AD,AR	100	9 of 9
IKBKG	Ectodermal Dysplasia, Immunodeficiency, Incontinentia Pigmenti	X,XR,XD,G	38.16	-
IKZF1	Immunodeficiency, Stevens-Johnson Syndrome	AD	99.98	43 of 43
IL12B	Immunodeficiency, Takayasu Arteritis	AR	100	12 of 12
IL12RB1	Immunodeficiency, Biliary Cholangitis	AR	99.94	92 of 96
IL21	Immunodeficiency	AR	99.73	1 of 1
IL21R	Immunodeficiency	AR	99.97	10 of 10
IL2RA	Diabetes Mellitus, Interleukin 2 Receptor Deficiency, Oligoarticular Idiopathic Arthritis	AR	100	9 of 9
IL2RB	Immunodeficiency, Lymphoproliferation, Autoimmunity, Oligoarticular Idiopathic Arthritis	AR	94.56	6 of 6
IL2RG	Immunodeficiency, Omenn Syndrome	X,XR,G	99.86	-
IL7R	Immunodeficiency, Omenn Syndrome	AR	100	54 of 55
IRAK4	Immunodeficiency	AR	99.96	27 of 28

IRF2BP2	Immunodeficiency, Promyelocytic Leukemia	AD	86.22	1 of 2
IRF7	Immunodeficiency	AR	99.97	3 of 3
IRF8	Immunodeficiency, Mendelian Susceptibility To Mycobacterial Diseases	AD,AR	100	9 of 9
IRF9	Immunodeficiency	AR	100	5 of 5
ISG15	Immunodeficiency	AR	100	3 of 3
IVNS1ABP	Immunodeficiency	AD	99.83	-
JAK3	Immunodeficiency	AR	99.98	86 of 88
JMJD1C	22q11.2 Deletion Syndrome	-	99.09	27 of 27
KLLN	Cowden Syndrome	-	97.52	9 of 9
KNSTRN	Immunodeficiency, Faciooculoskeletal Anomalies	-	99.98	-
LAMTOR2	Immunodeficiency	AR	100	1 of 1
LAT	Immunodeficiency	AR	100	3 of 3
LCK	Immunodeficiency	AR	99.99	4 of 4
LETM1	Wolf-Hirschhorn Syndrome	AD	98.2	2 of 2
LIG4	Lig4 Syndrome, Multiple Myeloma, Dubowitz Syndrome, Omenn Syndrome	AR	99.48	46 of 46
LMNB2	Barraquer-Simons Syndrome, Epilepsy, Lipodystrophy	AD,AR	95.03	5 of 5
LRBA	Immunodeficiency, Autoimmunity	AR	99.91	79 of 81
LRRC8A	Agammaglobulinemia	AD	100	2 of 2
LYST	Chediak-Higashi Syndrome	AR	99.98	117 of 117
MAGT1	Congenital Disorder Of Glycosylation, Immunodeficiency	X,XR,G	100	-
MALT1	Immunodeficiency, Malt Lymphoma	AR	95.76	9 of 9
MAN2B1	Mannosidosis, Alpha-Mannosidosis	AR	100	149 of 149
MAPK1	Distal 22q11.2 Microdeletion Syndrome	-	96.91	1 of 1
MBTPS2	Ichthyosis Follicularis, Atrichia, Photophobia Syndrome, Keratosis Follicularis Spinulosa Decalvans, Osteogenesis Imperfecta, Palmoplantar Keratoderma, Bresek Syndrome	X,XR,G	100	-
MEIS2	Cleft Palate, Mental Retardation, 15q14 Microdeletion Syndrome	AD	92	18 of 20
MMUT	Methylmalonic Aciduria, Vitamin B12-Unresponsive Methylmalonic Acidemia	AR	99.97	-
MRTFA	Immunodeficiency	AR	99.8	-
MS4A1	Immunodeficiency	AR	100	2 of 2
MSN	Immunodeficiency	X,XR,G	99.98	-
MTHFD1	Immunodeficiency, Megaloblastic Anemia, Hyperhomocysteinemia, Neural Tube Defects	AR	99.94	11 of 12
MYC	Burkitt Lymphoma	-	99.3	2 of 2
MYD88	Immunodeficiency, Macroglobulinemia, Waldenstrom Macroglobulinemia	AR	99.55	7 of 7
MYSM1	Bone Marrow Failure Syndrome, B-Cell Immunodeficiency, Skeletal Dysplasia	AR	98.5	4 of 4
NBN	Aplastic Anemia, Leukemia, Nijmegen Breakage Syndrome, Breast And Ovarian Cancer	AR,MU,P	100	200 of 200
NCF1	Granulomatous Disease	AR	74.19	31 of 39
NCF2	Granulomatous Disease	AR	100	72 of 73
NCKAP1L	Immunodeficiency	AR	100	-
NFE2L2	Immunodeficiency, Developmental Delay, Hypohomocysteinemia	AD	97.24	7 of 7
NFKB1	Immunodeficiency	AD	99.98	38 of 41
NFKB2	Immunodeficiency, Deficiency In Anterior Pituitary Function	AD	100	22 of 22

NFKBIA	Ectodermal Dysplasia, T-Cell Immunodeficiency	AD	99.98	13 of 13
NHEJ1	Cernunnos-Xlf Deficiency	-	100	12 of 14
NHP2	Dyskeratosis Congenita	AR	100	3 of 3
NOP10	Dyskeratosis Congenita	AR	100	1 of 1
NPM1	Leukemia, Dyskeratosis Congenita	AD	99.89	2 of 2
NSD2	Wolf-Hirschhorn Syndrome	AD	99.91	-
NSMCE3	Lung Disease, Immunodeficiency	AR	99.8	-
ORAI1	Immunodeficiency, Stormorken-Sjaastad-Langslet Syndrome, Tubular Aggregate Myopathy	AD,AR	91.93	20 of 22
PARN	Dyskeratosis Congenita, Pulmonary Fibrosis, Bone Marrow Failure, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.98	33 of 33
PGM3	Immunodeficiency, Congenital Disorder Of Glycosylation	AR	99.99	17 of 17
PIK3CA	Multiple Cancer Types, Capillary Malformation Of The Lower Lip, Lymphatic Malformation Of Face And Neck, Partial Or Generalized Overgrowth, Cowden Syndrome, Epidermal Nevus, Keratosis, Macrocephaly, Megalodactyly, Hemihyperplasia, Lynch Syndrome, Megalencephaly	AD	99.58	54 of 58
PIK3CD	Immunodeficiency, Faciooculoskeletal Anomalies	AD	100	23 of 23
PIK3R1	Agammaglobulinemia, Immunodeficiency, Short Syndrome	AD,AR	99.89	29 of 29
PKP1	Ectodermal Dysplasia, Skin Fragility, Epidermolysis Bullosa Simplex	AR	100	18 of 18
PNP	Immunodeficiency, Purine Nucleoside Phosphorylase Deficiency	AR	99.73	39 of 39
POLE	Colorectal Cancer, Facial Dysmorphism, Immunodeficiency, Intrauterine Growth Retardation, Metaphyseal Dysplasia, Adrenal Hypoplasia Congenita, Genital Anomalies, Image Syndrome	AD,AR	100	100 of 100
PRKCD	Immunodeficiency, Autoimmune Lymphoproliferative Syndrome	AR	100	9 of 9
PRKDC	Immunodeficiency, Neurologic Abnormalities	AR	99.74	9 of 10
PRPS1	Arts Syndrome, Charcot-Marie-Tooth Disease, Deafness, Phosphoribosylpyrophosphate Synthetase Superactivity, Lethal Ataxia, Deafness, Optic Atrophy	X,XR,G	100	-
PTEN	Cowden Disease, Macrocephaly, Autism Syndrome, Meningioma, Prostate Cancer, Bannayan-Riley-Ruvalcaba Syndrome, Breast And Ovarian Cancer, Polyposis, Lhermitte-Duclos Disease, Proteus Syndrome, Segmental Outgrowth, Lipomatosis, Epidermal Nevus	AD	99.97	609 of 629
PTPRC	Immunodeficiency	AR	99.98	7 of 7
RAB27A	Griscelli Syndrome	AR	100	54 of 55
RAC2	Immunodeficiency, Lymphopenia, Defective Neutrophil Chemotaxis, Hypogammaglobulinemia	AD,AR	100	5 of 5
RAG1	Cellular And Humoral Immune Defects, Omenn Syndrome, Immunodeficiency	AR	100	193 of 193
RAG2	Omenn Syndrome, Immunodeficiency	AR	100	90 of 91
RASGRP1	Immunodeficiency, Autoimmune Lymphoproliferative Syndrome	AR	98.41	8 of 9
RBCK1	Polyglucosan Body Myopathy, Immunodeficiency	AR	100	13 of 13
RELB	Immunodeficiency	AR	99.47	1 of 1
RFX5	Bare Lymphocyte Syndrome, Immunodeficiency	AR	99.98	13 of 13
RFXANK	Bare Lymphocyte Syndrome, Immunodeficiency	AR	95.14	24 of 24
RFXAP	Bare Lymphocyte Syndrome, Immunodeficiency	AR	94.32	8 of 9
RIPK1	Autoinflammation, Episodic Fever, Lymphadenopathy, Immunodeficiency	AD,AR	98.03	12 of 14
RMRP	Anauxetic And Metaphyseal Dysplasia, Cartilage-Hair Hypoplasia, Hypotrichosis, Omenn Syndrome	AR	-	-
RNF168	Riddle Syndrome	AR	99.91	6 of 6

RNU4ATAC	Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism, Roifman Syndrome	AR	-	-
RORC	Immunodeficiency	AR	99.99	6 of 6
RREB1	22q11.2 Deletion Syndrome	-	99.92	8 of 8
RTEL1	Dyskeratosis Congenita, Pulmonary Fibrosis, Marrow Failure, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.73	127 of 131
SBDS	Aplastic Anemia, Shwachman-Diamond Syndrome	AR	100	77 of 79
SDHB	Carney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paragangliomas, Pheochromocytoma, Cowden Syndrome, Succinate-Coq Reductase Deficiency	AD	100	261 of 264
SDHC	Carney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paragangliomas, Cowden Syndrome, Pheochromocytoma	AD	99.95	62 of 63
SDHD	Carney-Stratakis Syndrome, Mitochondrial Complex Ii Deficiency, Paragangliomas, Pheochromocytoma, Carcinoid Syndrome, Cowden Syndrome, Succinate-Coq Reductase Deficiency	AD,AR	99.98	164 of 166
SEC23B	Anemia, Cowden Syndrome	AD,AR	100	119 of 127
SEC24C	22q11.2 Deletion Syndrome	-	99.98	-
SH2D1A	Lymphoproliferative Syndrome	X,XR,G	99.94	-
SHANK3	Phelan-Mcdermid Syndrome, Schizophrenia, Monosomy 22q13.3	AD,MU,P	96.67	-
SIK3	Spondyloepimetaphyseal Dysplasia	AR	97.63	2 of 2
SKIV2L	Trichohepatoenteric Syndrome, Syndromic Diarrhea	AR	99.98	33 of 33
SLC46A1	Folate Malabsorption	AR	99.8	21 of 21
SMARCAL1	Schimke Immuno-Osseous Dysplasia	AR	99.94	93 of 93
SP110	Hepatic Venoocclusive Disease, Immunodeficiency, Veno-Occlusive Disease	AR	99.94	8 of 8
SPATA5	Epilepsy, Hearing Loss, Mental Retardation, Microcephaly	AR	99.83	30 of 30
SRP54	Neutropenia, Shwachman-Diamond Syndrome	AD,AR	99.95	8 of 8
STAT1	Immunodeficiency, Autoimmune Enteropathy, Endocrinopathy	AD,AR	100	138 of 138
STAT2	Immunodeficiency, Pseudo-Torch Syndrome	AR	100	9 of 9
STAT5B	Growth Hormone Insensitivity, Immunodeficiency, Promyelocytic Leukemia	AD	99.94	12 of 12
STIM1	Immune Dysfunction, T-Cell Inactivation, Myopathy, Stormorken-Sjaastad-Langslet Syndrome	AD,AR	100	28 of 28
STK4	T-Cell Immunodeficiency, Autoimmunity	AR	99.88	10 of 10
STX1A	Cystic Fibrosis	-	97	3 of 3
TBCE	Encephalopathy, Optic Atrophy, Hypoparathyroidism, Kenny-Caffey Syndrome, Spastic Ataxia, Sanjad-Sakati Syndrome	AR	100	8 of 8
TBK1	Encephalopathy, Frontotemporal Dementia, Amyotrophic Lateral Sclerosis, Herpes Simplex Virus Encephalitis	AD	99.91	141 of 142
TBX1	Conotruncal Heart Malformations, Digeorge Syndrome, Tetralogy Of Fallot, Velocardiofacial Syndrome, 22q11.2 Deletion Syndrome, 22q11.2 Microduplication Syndrome	AD,AR	88.7	35 of 42
TCF3	Agammaglobulinemia	AD	99.98	7 of 7
TERC	Dyskeratosis Congenita, Pulmonary Fibrosis, Bone Marrow Failure, Aplastic Anemia, Pulmonary Fibrosis	AD	-	-
TERT	Aplastic Anemia, Dyskeratosis Congenita, Leukemia, Melanoma, Pulmonary Fibrosis, Bone Marrow Failure, Hoyeraal-Hreidarsson Syndrome, Meningioma	AD,AR	99.09	194 of 197
TFRC	Immunodeficiency	AR	100	2 of 2
TGFB1	Camurati-Engelmann Disease, Cystic Fibrosis, Inflammatory Bowel Disease, Immunodeficiency, Encephalopathy	AD,AR	99.75	24 of 24
TICAM1	Encephalopathy, Herpes Simplex Virus Encephalitis	AD,AR	99.97	4 of 4
TINF2	Dyskeratosis Congenita, Revesz Syndrome, Hoyeraal-Hreidarsson Syndrome	AD	99.94	47 of 47
TLR3	Encephalopathy, Herpes Simplex Virus Encephalitis	AD,AR	100	16 of 16

TLR7	Immunodeficiency	X,XR,G	99.53	-
TNFRSF13B	Immunodeficiency, Immunoglobulin A Deficiency	AD,AR	100	50 of 50
TNFRSF13C	Immunodeficiency	AD,AR	99.2	3 of 3
TNFRSF1B	Classic Mycosis Fungoides, Sézary Syndrome	-	98.76	1 of 1
TNFRSF4	Immunodeficiency	AR	99.98	1 of 1
TNFSF12	Immunodeficiency	-	95.06	1 of 1
TPP2	Autoimmune Hemolytic Anemia, Autoimmune Thrombocytopenia, Immunodeficiency	-	99.84	11 of 11
TRAC	T-Cell Receptor Alpha/Beta Deficiency	AR	100	-
TRAF3	Herpes Simplex Virus Encephalitis	-	100	3 of 3
TRNT1	Retinitis Pigmentosa, Erythrocytic Microcytosis, Sideroblastic Anemia, B-Cell Immunodeficiency, Periodic Fevers, Developmental Delay	AR	99.47	22 of 27
TTC37	Trichohepatoenteric Syndrome, Syndromic Diarrhea	AR	100	66 of 66
TTC7A	Gastrointestinal Defects, Immunodeficiency Syndrome, Intestinal Atresia	AR	100	44 of 45
TYK2	Tyrosine Kinase 2 Deficiency	AR	97.56	9 of 9
UFD1	22q11.2 Deletion Syndrome	-	99.98	-
UNC119	Immunodeficiency, Cone Rod Dystrophy	AD	100	6 of 6
UNC93B1	Encephalopathy, Herpes Simplex Virus Encephalitis	AR	97.97	2 of 2
UNG	Immunodeficiency With Hiper Igm	AR	99.94	7 of 7
UROS	Erythropoietic Porphyria	AR	100	44 of 50
USB1	Poikiloderman Neutropenia, Dyskeratosis Congenita	AR	100	24 of 24
USF3	Cowden Syndrome	-	99.61	-
USP8	Pituitary Adenoma, Spastic Paraplegia, Cushing Disease	AD,AR	98.19	3 of 3
WAS	Neutropenia, Thrombocytopenia, Wiskott-Aldrich Syndrome	X,XR,G	100	-
WDR1	Periodic Fever, Immunodeficiency, Thrombocytopenia	AR	100	9 of 9
WHCR	Wolf-Hirschhorn Syndrome	AD	-	-
WIPF1	Wiskott-Aldrich Syndrome	AR	99.79	3 of 3
WRAP53	Dyskeratosis Congenita	AR	100	10 of 10
XIAP	Lymphoproliferative Syndrome	X,XR,G	99.94	-
XRCC4	Short Stature, Microcephaly, Endocrine Dysfunction, Lig4 Syndrome, Dwarfism, Insulin Resistance	AR	99.73	10 of 10
ZAP70	Autoimmune Disease, Immunodeficiency	AR	99.99	30 of 30
ZBTB24	Immunodeficiency, Icf Syndrome	AR	100	23 of 23

*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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