Inherited Thrombocytopenia

Precision Panel

Overview

Inherited thrombocytopenia (IT) is a group of hereditary disorders characterized by a low platelet count typically less than 150,000/uL with variations depending on age, gender and ethnic background. The main feature of these disorders results from a reduced platelet count often associated with an abnormal platelet function subsequently leading to impaired homeostasis. Severe inherited thrombocytopenias can present in the newborn period, whereas mild thrombocytopenias can remain undiagnosed until incidental detection on routine blood test during adulthood. Certain types of inherited thrombocytopenias carry a predisposition to acute myelogenous leukemia and/or myelodysplastic syndromes.

The Igenomix Inherited Thrombocytopenia Telangiectasia Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of recurrent bleeding 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 Inherited Thrombocytopenia Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Purpura
- Petechiae
- Prolonged bleeding from cuts
- Nosebleeds
- Gum bleeding
- Excessive bleeding after surgery
- Hemoptysis
- Hematuria
- Menorrhagia

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 with a multidisciplinary team in the form of routine screening for malignancies, medical therapy with antifibrinolytic agents, platelet transfusion and prevention of bleeding complications.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.
- Identification of causative gene of inherited thrombocytopenias given the high degree of heterogeneity with various clinical presentations and prognoses.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCA1	HDL Deficiency, Apolipoprotein A-I Deficiency, Tangier Disease	AD,AR	100	238 of 241
ABCD4	Methylmalonic Aciduria And Homocystinuria	AR	100	8 of 8
ABCG5	Homozygous Familial Hypercholesterolemia	-	99.81	57 of 57
ABCG8	Sitosterolemia, Hypercholesterolemia	AR,MU,P	100	64 of 64
ABL1	Congenital Heart Defects And Skeletal Malformations Syndrome, Chronic Myeloid Leukemia	AD	99.93	8 of 8
ACAD9	Acyl-Coa Dehydrogenase 9 Deficiency	AR	100	62 of 62
ACD	Dyskeratosis Congenita, Familial Melanoma, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.89	14 of 14
ACP5	Combined Immunodeficiency With Autoimmunity, Spondyloenchondrodysplasia	AR	100	27 of 28
ACTN1	Bleeding Disorder	AD	99.94	40 of 40
ADA	Severe Immunodeficiency, Omenn Syndrome	AR	100	97 of 98
ADA2	Polyarteritis Nodosa, Sneddon Syndrome, Blackfan-Diamond Anemia	AR	100	-
ADAMTS13	Thrombotic Thrombocytopenic Purpura	AR	99.59	195 of 213
ADAR	Aicardi-Goutieres Syndrome, Dyschromatosis Symmetrica Hereditaria, Familial Infantile Bilateral Striatal Necrosis	AD,AR	99.93	252 of 252
AGK	Cataract, Cardiomyopathy	AR	99.98	33 of 33
ALG8	Congenital Disorder Of Glycosylation, Polycystic Liver Disease	AD,AR	99.5	22 of 22
ANKRD11	16q24.3 Microdeletion Syndrome, Kbg Syndrome	AD	99.6	119 of 124
ANKRD26	Thrombocytopenia	AD	98.76	3 of 23
AP3B1	Hermansky-Pudlak Syndrome	AR	100	34 of 35
APOE	Alzheimer Disease, Lipoprotein Glomerulopathy, Macular Degeneration, Sea-Blue Histiocyte Disease, Dysbetalipoproteinemia	AD,AR	99.53	65 of 68
ARHGAP31	Adams-Oliver Syndrome	AD	100	6 of 6
ARHGEF1	Immunodeficiency	AR	90.23	2 of 2
ARPC1B	Platelet Abnormalities With Eosinophilia And Immune-Mediated Inflammatory Disease	AR	100	15 of 15
ARVCF	22q11.2 Deletion Syndrome		99.95	2 of 2
ASAH1	Farber Lipogranulomatosis, Spinal Muscular Atrophy With Progressive Myoclonic Epilepsy	AR	99.98	69 of 70
ATP7B	Wilson Disease	AR	99.97	989 of 1000
ATRX	Alpha-Thalassemia, 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	-
BCOR	Microphthalmia, Acute Promyelocytic Leukemia, Oculofaciocardiodental Syndrome	X,XD,G	99.87	-
BCR	Chronic Myeloid Leukemia, 22q11.2 Microdeletion Syndrome	MU,P	97.78	-

BLOC1S6	Hermansky-Pudlak Syndrome	AR	99.48	2 of 2
BRAF	Cardiofaciocutaneous Syndrome, Leopard Syndrome, Lung Cancer, Craniopharyngioma, Noonan Syndrome	AD	100	80 of 80
BRCA1	Breast Cancer, Fanconi Anemia, Familial Pancreatic Carcinoma, Primary Peritoneal Carcinoma	AD,AR,MU	98.97	2783 of 2894
BRCA2	Breast Cancer, Fanconi Anemia, Glioma Susceptibility, Medulloblastoma, Pancreatic Cancer, Prostate Cancer, Wilms Tumor, Hereditary Breast And Ovarian Cancer Syndrome, Nephroblastoma	AD,AR,MU	98.51	3343 of 3451
BRIP1	Breast Cancer, Ovarian Cancer, Fanconi Anemia	AD,AR	94.97	235 of 237
BTK	Agammaglobulinemia, Hypogammaglobulinemia And Isolated Growth Hormone Deficiency	X,XR,G	100	-
BTNL2	Sarcoidosis	AD	99.98	1 of 1
C3	Complement Component 3 Deficiency, Hemolytic Uremic Syndrome	AD,AR	100	123 of 124
CA2	Osteopetrosis With Renal Tubular Acidosis	AR	100	36 of 36
CALR	Myelofibrosis With Myeloid Metaplasia, Thrombocythemia	AD	100	3 of 4
CASP10	Autoimmune Lymphoproliferative Syndrome, Gastric Cancer, Lymphoma Non-Hodgkin	AD	99.86	6 of 6
CD109	Fetal And Neonatal Alloimmune Thrombocytopenia		99.94	-
CD19	Immunodeficiency	AD,AR	99.99	7 of 7
CD36	Platelet Glycoprotein Iv Deficiency	AR	100	39 of 39
CD40LG	Immunodeficiency	X,XR,G	100	-
CD46	Hemolytic Uremic Syndrome, Hellp Syndrome	AD,AR	100	83 of 84
CD81	Immunodeficiency	AR	100	2 of 2
CDC42	Takenouchi-Kosaki Syndrome, Macrothrombocytopenia	AD	99.97	10 of 10
CFB	Complement Factor B Deficiency, Hemolytic Uremic Syndrome	AD,AR	100	26 of 26
CFH	Basal Laminar Drusen, Complement Factor H Deficiency, Hemolytic Uremic Syndrome, Hellp Syndrome	AD,AR,MU,P	99.94	340 of 342
CFHR1	Hemolytic Uremic Syndrome, Macular Degeneration	AD,AR	88.29	0 of 9
CFHR3	Hemolytic Uremic Syndrome, Macular Degeneration	AD,AR	89.89	0 of 7
CFI	Complement Factor I Deficiency, Hemolytic Uremic Syndrome, Macular Degeneration, Hellp Syndrome	AD,AR	99.93	156 of 158
CIITA	Bare Lymphocyte Syndrome, Rheumatoid Arthritis, Immunodeficiency By Defective Expression Of Mhc Class Ii	AR	98.51	15 of 16
CLCN7	Hypopigmentationand Delayed Myelination, Osteopetrosis	AD,AR	99.85	109 of 111
COG1	Congenital Disorder Of Glycosylation	AR	99.91	3 of 3
COG4	Congenital Disorder Of Glycosylation, Saul-Wilson Syndrome, Microcephalic Osteodysplastic Dysplasia	AD,AR	100	5 of 5
COG6	Congenital Disorder Of Glycosylation, Shaheen Syndrome, Hypohidrosis-Enamel Hypoplasia-Palmoplantar Keratoderma-Intellectual Disability Syndrome	AR	100	13 of 13
COL4A5	Alport Syndrome	X,XD,G	99.88	-
COMT	22q11.2 Deletion Syndrome	AD	99.98	5 of 5
CORIN	Preeclampsia	AD	99.7	5 of 5
CR2	Immunodeficiency	AD,AR	99.92	19 of 19
CTC1	Cerebroretinal Microangiopathy With Calcifications And Cysts, Dyskeratosis Congenita	AR	99.73	43 of 44
CTLA4	Autoimmune Lymphoproliferative Syndrome, Hashimoto Thyroiditis, Systemic Lupus Erythematosus, Classic Mycosis Fungoides, Granulomatosis With Polyanginitis , Sézary Syndrom	AD	99.97	60 of 60
CYCS	Thrombocytopenia	AD	100	4 of 4
DCLRE1C	Omenn Syndrome, Severe Combined Immunodeficiency With Sensitivity To Ionizing Radiation	AR	99.99	72 of 73
DGKE	Nephrotic Syndrome	AR	99.67	41 of 42

DGUOK	Mitochondrial Dna Depletion Syndrome, Portal Hypertension, Progressive External Ophthalmoplegia	AR	100	68 of 70
DHFR	Megaloblastic Anemia	AR	99.7	4 of 4
DIAPH1	Deafness, Seizures	AD,AR	99.94	15 of 15
DKC1	Dyskeratosis Congenita, Hoyeraal-Hreidarsson Syndrome	X,XR,G	100	-
DLL4	Adams-Oliver Syndrome, Cutis Aplasia	AD	99.98	21 of 21
DNAJC21	Bone Marrow Failure Syndrome, Shwachman-Diamond Syndrome	AR	99.83	12 of 12
DNASE1	Systemic Lupus Erythematosus	AD	100	5 of 5
DOCK6	Adams-Oliver Syndrome	AR	98.06	37 of 37
DZIP1L	Polycystic Kidney Disease	AR	99.83	5 of 5
EFL1	Shwachman-Diamond Syndrome	AR	99.94	-
ELANE	Cyclic Hematopoiesis, Neutropenia	AD	100	227 of 227
EOGT	Adams-Oliver Syndrome	AR	100	11 of 11
ERBB3	Erythroleukemia, Lethal Congenital Contracture Syndrome	AD,AR	99.91	6 of 6
ERCC4	Fanconi Anemia, Xeroderma Pigmentosum, Cockayne Syndrome	AR	99.68	69 of 72
ERCC6L2	Bone Marrow Failure Syndrome	AR	97.82	13 of 14
ESCO2	Roberts Syndrome	AR	99.69	32 of 32
ETV6	Leukemia, Thrombocytopenia	AD	100	41 of 41
FANCA	Fanconi Anemia	AR	95.17	497 of 502
FANCB	Fanconi Anemia, Vacterl Association With Hydrocephalus	X,XR,G	95.53	-
FANCC	Fanconi Anemia	AR	100	75 of 75
FANCD2	Fanconi Anemia	AR	100	62 of 63
FANCE	Fanconi Anemia	AR	97	17 of 18
FANCF	Fanconi Anemia	AR	99.31	17 of 18
FANCG	Fanconi Anemia		100	94 of 94
FANCI	Fanconi Anemia	AR	100	53 of 54
FANCL	Fanconi Anemia	AR	100	25 of 26
FANCM	Spermatogenic Failure, Fanconi Anemia, Male Infertility	AR	99.73	59 of 61
FARS2	Oxidative Phosphorylation Deficiency, Spastic Paraplegia	AR	99.98	23 of 23
FAS	Autoimmune Lymphoproliferative Syndrome, Behçet Disease, Vogt-Koyanagi-Harada Disease	AD	100	135 of 135
FASLG	Autoimmune Lymphoproliferative Syndrome, Lung Cancer	AD	99.98	8 of 9
FCGR2A	Cystic Fibrosis, Systemic Lupus Erythematosus	AD,AR	93.97	-
FCGR2B	Systemic Lupus Erythematosus	AD	73.78	1 of 1
FCGR2C	Immune Thrombocytopenia		81.45	-
FIP1L1	Acute Promyelocytic Leukemia		99.92	0 of 1
FLI1	Bleeding Disorder, Jacobsen Syndrome, Paris-Trousseau Thrombocytopenia, Peripheral Primitive Neuroectodermal Tumor	AD,AR	100	7 of 7
FLNA	Cardiac Valvular Dysplasia, Fg Syndrome, Frontometaphyseal & Terminal Osseous Dysplasia, Otopalatodigital Syndrome, Melnick-Needles Syndrome, Neuronal Intestinal Pseudoobstruction, Periventricular Nodular Heterotopia, Ehlers-Danlos Syndrome	X,XR,XD,G	100	-
FLT1	Preeclampsia		100	6 of 6
FOXP3	Immunodysregulation, Polyendocrinopathy, Enteropathy	X,XR,G	99.86	-
FYB1	Thrombocytopenia	AR	99.68	-
G6PC3	Neutropenia	AR	100	45 of 45
GALC	Krabbe Disease	AR	99.38	252 of 254

GATA1	Down Syndrome, Dyserythropoietic Anemia, Thrombocytopenia, Beta-Thalassemia, Blackfan-Diamond Anemia, Congenital Erythropoietic Porphyria	X,XR,G	99.93	-
GATA2	Dendritic Cell, Monocyte, B Lymphocyte And Natural Killer Lymphocytedeficiency, Leukemia, Lymphedema, Myelodysplastic Syndrome	AD	100	137 of 142
GBA	Dementia, Gaucher Disease, Parkinson Disease	AD,AR	100	469 of 471
GFI1B	Thrombasthenia, Thrombocytopenia, Gray Platelet Syndrome	AD,AR	100	13 of 15
GNA14	Tufted Angioma		99.96	2 of 2
GP1BA	Bernard-Soulier Syndrome, Nonarteritic Anterior Ischemic Optic Neuropathy, Pseudo-Von Willebrand Disease, Fetal And Neonatal Alloimmune Thrombocytopenia	AD,AR	99.98	73 of 73
GP1BB	Bernard-Soulier Syndrome, 22q11.2 Deletion Syndrome, Fetal And Neonatal Alloimmune Thrombocytopenia	AR	74.08	26 of 50
GP9	Bernard-Soulier Syndrome	AR	99.96	41 of 41
GUCY1A1	Moyamoya Disease 6 With Achalasia; Mymy6, Moyamoya Disease	AR	99.91	-
HELLPAR	Hellp Syndrome		-	-
HIRA	22q11.2 Deletion Syndrome		99.99	5 of 5
HLA-B	Spondyloarthropathy, Behçet Disease, Stevens-Johnson Syndrome, Takayasu Arteriti	MU	99.55	1 of 1
HLA-DRB1	Multiple Sclerosis, Sarcoidosis, Bullous Pemphigoid, Cutaneous Systemic Sclerosis, Follicular Lymphoma, Narcolepsy, Systemic-Onset Juvenile Idiopathic Arthritis	AD,MU	97.19	2 of 2
HLCS	Holocarboxylase Synthetase Deficiency	AR	100	47 of 47
HOXA11	Radioulnar Synostosis-Amegakaryocytic Thrombocytopenia Syndrome	AD	99.92	3 of 3
HPS5	Hermansky-Pudlak Syndrome	AR	99.88	32 of 32
HYOU1	Immunodeficiency, Hypoglycemia	AR	99.94	2 of 2
ICOS	Common Variable Immunodeficiency, Icos Deficiency	AD,AR	100	4 of 5
IFIH1	Aicardi-Goutieres Syndrome, Singleton-Merten Syndrome	AD	99.62	26 of 27
IFNG	Aplastic Anemia, Immunodeficiency, Tuberous Sclerosis	AD,AR	99.77	-
IKZF1	Immunodeficiency, Stevens-Johnson Syndrome	AD	99.98	43 of 43
IL7R	Severe Combined Immunodeficiency, Omenn Syndrome	AR	100	54 of 55
IRAK1	Pediatric Systemic Lupus Erythematosus		96.2	-
IRF2BP2	Immunodeficiency, Acute Promyelocytic Leukemia	AD	86.22	1 of 2
ITGA2	Fetal And Neonatal Alloimmune Thrombocytopenia		100	7 of 7
ITGA2B	Glanzmann Thrombasthenia, Fetal And Neonatal Alloimmune Thrombocytopenia	AD,AR	100	237 of 239
ITGB3	Glanzmann Thrombasthenia, Fetal And Neonatal Alloimmune Thrombocytopenia	AD,AR	99.44	178 of 179
ITK	Lymphoproliferative Syndrome	AR	100	19 of 19
IVD	Isovaleric Acidemia	AR	100	105 of 105
JAK2	Erythrocytosis, Leukemia, Myelofibrosismyelofibrosis, Polycythemia Vera, Thrombocythemia, Budd-Chiari Syndrome, Essential Thrombocythemia, Familial Thrombocytosis, Polycythemia Vera, Primary Myelofibrosis	AD,AR	99.63	25 of 27
JAM2	Basal Ganglia Calcification, Bilateral Striopallidodentate Calcinosis	AR	99.98	-
JMJD1C	22q11.2 Deletion Syndrome		99.09	27 of 27
KDM6A	Kabuki Syndrome	AD,X,XD,G	99.98	-
KIF15	21q22.11q22.12 Microdeletion Syndrome		99.85	3 of 3
KMT2D	Kabuki Syndrome	AD	99.71	839 of 847
KRAS	Leukemia, Autoimmune Lymphoproliferative Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Noonan Syndrome, Toriello-Lacassie-Droste Syndrome	AD	100	38 of 38

LARS2	Sideroblastic Anemia, Perrault Syndrome	AR	99.99	20 of 20
LAT	Immunodeficiency	AR	100	3 of 3
LBR	Pelger-Huet Anomaly, Reynolds Syndrome, Greenberg Dysplasia	AD,AR	99.98	34 of 34
LIG4	Lig4 Syndrome, Multiple Myeloma, Dubowitz Syndrome, Omenn Syndrome	AR	99.48	46 of 46
LMBRD1	Methylmalonic Aciduria And Homocystinuria	AR	99.88	8 of 8
LRBA	Immunodeficiency With Autoimmunity	AR	99.91	79 of 81
LYST	Chediak-Higashi Syndrome	AR	99.98	117 of 117
MAD2L2	Fanconi Anemia	AR	99.91	1 of 1
MAP2K1	Cardiofaciocutaneous Syndrome, Melorheostosis, Noonan Syndrome	AD	100	31 of 31
MASTL	Thrombocytopenia	-	99.95	5 of 5
MECOM	Radioulnar Synostosis With Amegakaryocytic Thrombocytopenia	AD	99.97	26 of 27
MMAA	Methylmalonic Aciduria	AR	99.98	77 of 77
MMAB	Methylmalonic Aciduria	AR	99.52	43 of 43
MMACHC	Methylmalonic Aciduria And Homocystinuria	AR	99.97	105 of 105
MMUT	Vitamin B12-Unresponsive Methylmalonic Acidemia	AR	99.97	-
MPIG6B	Thrombocytopenia, Anemia, Myelofibrosis	AR	-	-
MPL	Thrombocytopenia, Myelofibrosismyelofibrosis With Myeloid Metaplasia, Thrombocytosis, Polycythemia Vera	AD,AR	100	55 of 55
MS4A1	Immunodeficiency	AR	100	2 of 2
MTOR	Taylor Dysplasia, Smith-Kingsmore Syndrome, Macrocephaly-Intellectual Disability-Neurodevelopmental Disorder-Small Thorax Syndrome	AD	99.98	39 of 39
MVK	Hyper-IgD Syndrome, Porokeratosis, Mevalonic Aciduria	AD,AR	100	180 of 181
MYH9	Deafness, May-Hegglin Anomaly	AD	100	144 of 145
MYORG	Basal Ganglia Calcification, Bilateral Striopallidodentate Calcinosis	AR	100	-
MYSM1	Bone Marrow Failure Syndrome	AR	98.5	4 of 4
NABP1	Promyelocytic Leukemia	-	100	-
NBEAL2	Gray Platelet Syndrome	AR	99.74	51 of 51
NBN	Aplastic Anemia, Leukemia, Nijmegen Breakage Syndrome, Hereditary Breast And Ovarian Cancer Syndrome	AR,MU,P	100	200 of 200
NFKB1	Immunodeficiency	AD	99.98	38 of 41
NFKB2	Immunodeficiency	AD	100	22 of 22
NHEJ1	Cernunnos-Xlf Deficiency		100	12 of 14
NHP2	Dyskeratosis Congenital	AR	100	3 of 3
NIPBL	Cornelia De Lange Syndrome	AD	99.32	409 of 426
NLRC4	Autoinflammation With Infantile Enterocolitis	AD	99.54	15 of 15
NOP10	Dyskeratosis Congenital	AR	100	1 of 1
NOS3	Alzheimer Disease, Hypertension, Ischemic Stroke, Eclampsia	AD,MU	99.98	13 of 13
NOTCH1	Adams-Oliver Syndrome, Aortic Valve Disease	AD	99.83	178 of 179
NPM1	Leukemia, Dyskeratosis Congenital	AD	99.89	2 of 2
NRAS	Colorectal Cancer, Epidermal And Sebaceus Nevus, Neurocutaneous Melanosis, Noonan Syndrome, Autoimmune Lymphoproliferative Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Thyroid Cancer	AD	100	15 of 15
NSUN2	Dubowitz Syndrome	AR	99.99	8 of 8
NUMA1	Acute Promyelocytic Leukemia	-	99.99	5 of 5
OCLN	Pseudo-Torch Syndrome, Congenital Intrauterine Infection-Like Syndrome	AR	86.89	15 of 17

OCRL	Dent Disease, Lowe Oculocerebrorenal Syndrome	X,XR,G	100	-
OSTM1	Osteopetrosis With Neuroaxonal Dysplasia	AR	100	8 of 9
PALB2	Breast Cancer, Ovarian Cancer, Fanconi Anemia, Familial Pancreatic Carcinoma	AD,AR	98.78	601 of 617
PARN	Dyskeratosis Congenital, Pulmonary Fibrosis, Bone Marrow Failure, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.98	33 of 33
PCCA	Propionic Acidemia	AR	100	137 of 137
PCCB	Propionic Acidemia	AR	99.95	136 of 138
PDGFB	Basal Ganglia Calcification, Meningioma, Bilateral Striopallidodentate Calcinosis, Dermatofibrosarcoma Protuberans	AD	100	22 of 22
PDGFRB	Basal Ganglia Calcification, Kosaki Overgrowth Syndrome, Myeloproliferative Disorder, Myofibromatosis, Premature Aging Syndrome, Bilateral Striopallidodentate Calcinosis	AD	99.64	28 of 28
PEPD	Prolidase Deficiency	AR	95	34 of 34
PHGDH	Neu-Laxova Syndrome, Phosphoglycerate Dehydrogenase Deficiency	AR	100	26 of 26
PKHD1	Polycystic Kidney Disease	AR	99.97	582 of 585
PLAU	Alzheimer Disease, Quebec Platelet Disorder	AD	100	5 of 5
PML	Acute Promyelocytic Leukemia	-	100	4 of 4
PNP	Purine Nucleoside Phosphorylase Deficiency	AR	99.73	39 of 39
POMP	Proteasome-Associated Autoinflammatory Syndrome, Keratosis Linearis-Ichthyosis Congenita-Sclerosing Keratoderma Syndrome	AD,AR	99.99	4 of 4
PRDX1	Methylmalonic Aciduria And Homocystinuria	AR	100	3 of 3
PRF1	Aplastic Anemia, Hemophagocytic Lymphohistiocytosis, Lymphoma, Non-Hodgkin	AR	99.99	196 of 196
PRKACG	Bleeding Disorder	AR	99.88	1 of 1
PRKAR1A	Acrodysostosis, Carney Complex, Pigmented Nodular Adrenocortical Disease, Promyelocytic Leukemia, Familial Atrial Myxoma	AD	95.93	165 of 171
PRKCD	Immunodeficiency, Autoimmune Lymphoproliferative Syndrome	AR	100	9 of 9
PSAP	Saposin Deficiency, Gaucher Disease, Krabbe Disease, Metachromatic Leukodystrophy, Encephalopathy	AR	100	33 of 33
PSMB4	Proteasome-Associated Autoinflammatory Syndrome	AR	100	4 of 4
PSMB8	Proteasome-Associated Autoinflammatory Syndrome	AR	100	11 of 11
PSMB9	Proteasome-Associated Autoinflammatory Syndrome	AR	100	2 of 2
PTPN11	Juvenile Myelomonocytic Leukemia, Leopard Syndrome, Metachondromatosis, Noonan Syndrome	AD	100	150 of 151
PTPN22	Diabetes Mellitus, Rheumatoid An Idiopathic Arthritis, Systemic Lupus Erythematosus, Giant Cell Arteritis, Granulomatosis With Polyangiitis, Vogt-Koyanagi-Harada Disease	AD	99.67	5 of 5
RAD51	Hereditary Breast And Ovarian Cancer Syndrome, Fanconi Anemia, Mirror Movements	AD	99.98	16 of 16
RAD51C	Fanconi Anemia, Hereditary Breast And Ovarian Cancer Syndrome	AR	100	130 of 130
RAG1	Omenn Syndrome, Severe Combined Immunodeficiency	AR	100	193 of 193
RAG2	Severe Combined Immunodeficiency With Granulomas, Omenn Syndrome	AR	100	90 of 91
RARA	Acute Promyelocytic Leukemia	-	97.16	2 of 2
RASGRP1	Immunodeficiency, Autoimmune Lymphoproliferative Syndrome	AR	98.41	8 of 9
RBM8A	Thrombocytopenia	AR	100	4 of 4
RBPJ	Adams-Oliver Syndrome	AD	99.98	8 of 8
RFWD3	Fanconi Anemia	AR	99.99	2 of 2
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
RNASEH2A	Aicardi-Goutieres Syndrome	AR	100	23 of 23

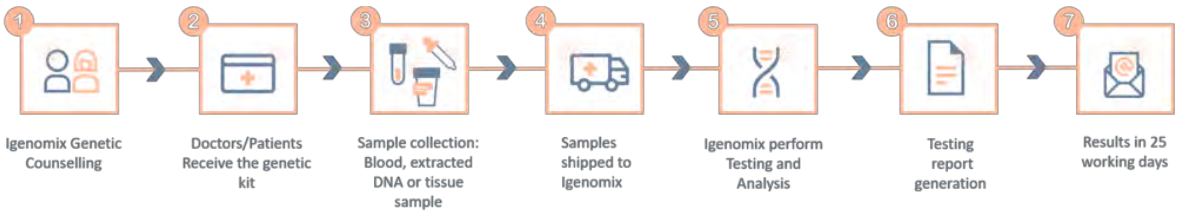
RNASEH2B	Aicardi-Goutieres Syndrome	AR	99.95	41 of 41
RNASEH2C	Aicardi-Goutieres Syndrome	AR	100	14 of 14
RPL11	Diamond-Blackfan Anemia	AD	100	52 of 52
RPL15	Diamond-Blackfan Anemia	AD	99.74	8 of 9
RPL18	Diamond-Blackfan Anemia	AD	100	1 of 1
RPL26	Diamond-Blackfan Anemia	AD	92.97	1 of 1
RPL27	Diamond-Blackfan Anemia	AD	100	2 of 2
RPL31	Diamond-Blackfan Anemia	AD	100	0 of 1
RPL35	Diamond-Blackfan Anemia	AD	100	1 of 1
RPL35A	Diamond-Blackfan Anemia	AD	100	12 of 12
RPL5	Diamond-Blackfan Anemia	AD	100	95 of 95
RPS10	Diamond-Blackfan Anemia	AD	100	7 of 7
RPS15A	Diamond-Blackfan Anemia	AD	98.74	1 of 1
RPS17	Diamond-Blackfan Anemia	AD	0	0 of 7
RPS19	Diamond-Blackfan Anemia	AD	78	159 of 165
RPS24	Diamond-Blackfan Anemia	AD	90.17	11 of 14
RPS26	Diamond-Blackfan Anemia	AD	100	28 of 29
RPS27	Diamond-Blackfan Anemia	AD	99.85	1 of 1
RPS28	Diamond-Blackfan Anemia	AD	100	1 of 1
RPS29	Diamond-Blackfan Anemia	AD	100	4 of 4
RPS7	Diamond-Blackfan Anemia	AD	100	7 of 10
RRAS2	Noonan Syndrome	AD	99.8	6 of 6
RREB1	22q11.2 Deletion Syndrome	-	99.92	8 of 8
RTEL1	Congenital Dyskeratosis, Hoyeraal-Hreidarsson Syndrome, Idiopathic Pulmonary Fibrosis	AD,AR	99.73	127 of 131
RUNX1	Platelet Disorder, Aggressive Systemic Mastocytosis, Chronic Myeloid Leukemia	AD	99.83	90 of 90
SALL4	Ivic Syndrome, Acro-Renal-Ocular Syndrome, Duane Retraction Syndrome	AD	100	54 of 54
SAMD9	Mirage Syndrome, Tumoral Calcinosis	AD,AR	99.72	45 of 46
SAMD9L	Ataxia Pancytopenia Syndrome	AD	99.81	39 of 39
SAMHD1	Aicardi-Goutieres Syndrome, Chilblain Lupus	AD,AR	100	51 of 51
SARS2	Hyperuricemia, Pulmonary Hypertension, Renal Failure, Alkalosis	AR	97.5	6 of 6
SBDS	Aplastic Anemia, Shwachman-Diamond Syndrome	AR	100	77 of 79
SC5D	Lathosterolosis	AR	99.62	6 of 6
SCARB2	Action Myoclonus-Renal Failure Syndrome, Gaucher Disease, Unverricht-Lundborg Disease	AR	99.95	29 of 29
SEC24C	22q11.2 Deletion Syndrome	-	99.98	-
SF3B1	Myelodysplastic Syndrome, Sideroblastic Anemia, Uveal Melanoma	-	99.95	4 of 4
SH2D1A	Lymphoproliferative Syndrome	X,XR,G	99.94	-
SLC19A2	Thiamine-Responsive Megaloblastic Anemia Syndrome	AR	99.99	67 of 68
SLC20A2	Basal Ganglia Calcification, Bilateral Striopallidodentate Calcinosis	AD	99.96	123 of 127
SLC35A1	Congenital Disorder Of Glycosylation, Type Iif; Cdg2f, Slc35a1-CDG	AR	100	6 of 6
SLC46A1	Hereditary Folate Malabsorption	AR	99.8	21 of 21
SLC7A7	Lysinuric Protein Intolerance	AR	100	61 of 61
SLFN14	Bleeding Disorder	AD	99.86	5 of 5
SLX4	Fanconi Anemia	AR	99.92	76 of 76

SMARCAL1	Immunoosseous Dysplasia	AR	99.94	93 of 93
SMARCD2	Specific Granule Deficiency	AR	91.58	1 of 1
SMPD1	Niemann-Pick Disease	AR	99.98	258 of 258
SNX10	Osteopetrosis	AR	100	14 of 14
SP110	Hepatic Veno-Occlusive Disease-Immunodeficiency Syndrome	AR	99.94	8 of 8
SPATA5	Epilepsy, Hearing Loss, And Mental Retardation Syndrome	AR	99.83	30 of 30
SPP1	Pediatric Systemic Lupus Erythematosus	-	99.77	2 of 2
SRC	Colorectal Cancer, Thrombocytopenia	AD	99.98	3 of 3
SRP54	Neutropenia, Shwachman-Diamond Syndrome	AD,AR	99.95	8 of 8
STAT1	Immunodeficiency, Autoimmune Enteropathy And Endocrinopathy-Susceptibility To Chronic Infections Syndrome	AD,AR	100	138 of 138
STAT2	Immunodeficiency, Pseudo-Torch Syndrome	AR	100	9 of 9
STAT3	Autoimmune Disease, Acute Promyelocytic Leukemia, Hyper-Ige Syndrome, Permanent Neonatal Diabetes Mellitus	AD	100	171 of 171
STAT4	Behçet Disease, Idiopathic Arthritis, Systemic Lupus Erythematosus	-	99.98	4 of 4
STAT5B	Growth Hormone Insensitivity With Immunodeficiency, Acute Promyelocytic Leukemia	AD	99.94	12 of 12
STIM1	Immune Dysfunction With T-Cell Inactivation Due To Calcium Entry Defect, Myopathy, Stormorken Syndrome, Tubular Aggregate Myopathy	AD,AR	100	28 of 28
STOX1	Preeclampsia	MU,P	90.34	4 of 5
STT3B	Congenital Disorder Of Glycosylation	AR	98.71	5 of 5
STX11	Hemophagocytic Lymphohistiocytosis	AR	100	24 of 24
TALDO1	Transaldolase Deficiency	AR	95	13 of 14
TBL1XR1	Pierpont Syndrome, Acute Promyelocytic Leukemia	AD	99.78	23 of 23
TBX1	Conotruncal Heart Malformations, DiGeorge Syndrome, Tetralogy Of Fallot, Velocardiofacial Syndrome	AD,AR	88.7	35 of 42
TBXAS1	Ghosal Hematodiaphyseal Dysplasia	AR	100	6 of 6
TCN2	Transcobalamin Deficiency	AR	100	25 of 27
TERC	Dyskeratosis Congenita, Pulmonary Fibrosis And/Or Bone Marrow Failure, Idiopathic Aplastic Anemia, Idiopathic Pulmonary Fibrosis	AD	-	-
TERT	Aplastic Anemia, Dyskeratosis Congenita, Leukemia, Melanoma, Meningioma, Pulmonary Fibrosis And/Or Bone Marrow Failure, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.09	194 of 197
TET2	Myelodysplastic Syndrome, Mastocytosis, Thrombocythemia, Polycythemia Vera, Primary Myelofibrosis, Refractory Anemia	-	99.96	15 of 15
TFRC	Immunodeficiency	AR	100	2 of 2
THBD	Hemolytic Uremic Syndrome, Thrombophilia	AD	99.91	29 of 30
TINF2	Dyskeratosis Congenita, Revesz Syndrome, Hoyeraal-Hreidarsson Syndrome	AD	99.94	47 of 47
TMEM165	Congenital Disorder Of Glycosylation	AR	93.69	4 of 5
TNFAIP3	Autoinflammatory Syndrome	AD	99.91	30 of 30
TNFRSF11A	Osteopetrosis, Paget Disease, Polyostotic Osteolytic Dysplasia, Dysosteosclerosis	AD,AR	96.37	17 of 22
TNFRSF13B	Immunodeficiency	AD,AR	100	50 of 50
TNFRSF13C	Immunodeficiency	AD,AR	99.2	3 of 3
TNFSF11	Osteopetrosis	AR	99.84	4 of 4
TNFSF12	Immunodeficiency	-	95.06	1 of 1
TPP2	Autoimmune Hemolytic Anemia-Autoimmune Thrombocytopenia-Primary Immunodeficiency Syndrome-		99.84	11 of 11
TREX1	Chilblain Lupus, Vasculopathy, Aicardi-Goutières Syndrome	AD,AR	100	75 of 75
TSR2	Diamond-Blackfan Anemia	X,XR,G	99.96	-

TUBB1	Macrothrombocytopenia	AD	100	13 of 13
UBE2T	Fanconi Anemia	AR	100	4 of 4
UFD1	22q11.2 Deletion Syndrome	-	99.98	-
UQCRFS1	Mitochondrial Complex Iii Deficiency	AR	99.58	-
UROS	Congenital Erythropoietic Porphyria	AR	100	44 of 50
USB1	Poikiloderma With Neutropenia, Dyskeratosis Congenita	AR	100	24 of 24
USP18	Pseudo-Torch Syndrome	AR	95.84	1 of 1
VPS33A	Mucopolysaccharidosis-Plus Syndrome	AR	97.86	1 of 1
VPS45	Neutropenia	AR	100	4 of 4
VWF	Von Willebrand Disease	AD,AR	98	933 of 1001
WARS2	Neurodevelopmental Disorder, Wars2-Related Combined Oxidative Phosphorylation Defect	AR	99.95	14 of 15
WAS	Neutropenia, Thrombocytopenia, Wiskott-Aldrich Syndrome	X,XR,G	100	-
WDR1	Periodic Fever, Immunodeficiency, Thrombocytopenia	AR	100	9 of 9
WFS1	Cataract, Deafness, Diabetes Mellitus, Wolfram Syndrome	AD,AR	99.97	390 of 395
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	-
XPR1	Basal Ganglia Calcification, Bilateral Striopallidodentate Calcinosis	AD	99.88	14 of 14
XRCC2	Fanconi Anemia, Male Infertility With Azoospermia Or Oligozoospermia	AR	98.39	28 of 28
ZAP70	Autoimmune Disease, Severe Combined Immunodeficiency	AR	99.99	30 of 30
ZBTB16	Skeletal Defects, Genital Hypoplasia, Acute Promyelocytic Leukemia	AR	100	1 of 1

*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



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- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

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