PRIMARY IMMUNODEFICIENCIES PANEL DG-4.2.0 (78 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
AP1S3	90.6%	90.6%	100%	99.9%	99%	{Psoriasis 15, pustular, susceptibility to}, 616106
AP3B1	100%	100%	100%	100%	99.5%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	100%	100%	100%	99.9%	99.1%	?Hermansky-Pudlak syndrome 10, 617050
APOL1	100%	100%	100%	99.9%	99.2%	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
ARHGEF1	100%	99.6%	100%	99.8%	97.5%	?Immunodeficiency 62, 618459
ARPC1B	100%	100%	100%	100%	98.7%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718

ARPC5	100%	100%	100%	100%	99%	Immunodeficiency 133 with autoimmunity and autoinflammation, 620565
ASXL1	100%	100%	100%	99.9%	99.2%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ATAD3A	100%	100%	100%	99.9%	98.6%	Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATG4A	100%	100%	99.4%	90.6%	71.1%	
ATM	100%	100%	100%	99.9%	99.4%	Lymphoma, B-cell non-Hodgkin, somatic;Ataxia-telangiectasia, 208900;{Breast cancer, susceptibility to}, 114480;T-cell prolymphocytic leukemia, somatic;Lymphoma, mantle cell, somatic
ATP6AP1	100%	99.8%	99%	89.7%	67.3%	Immunodeficiency 47, 300972

CASP10	100%	99.7%	100%	100%	99.5%	Autoimmune lymphoproliferative syndrome, type II, 603909;Gastric cancer, somatic, 613659;Lymphoma, non-Hodgkin, somatic, 605027
CASP8	97%	97%	100%	100%	99.5%	{Breast cancer, protection against}, 114480;?Caspase 8 lymphadenopathy syndrome, 607271;Hepatocellular carcinoma, somatic, 114550;{Lung cancer, protection against}, 211980
CAVIN1	100%	100%	100%	99.8%	97.9%	Lipodystrophy, congenital generalized, type 4, 613327
CBLB	100%	100%	100%	99.9%	99.4%	Autoimmune disease, multisystem, infantile-onset, 3, 620430
CCBE1	100%	100%	100%	100%	99.3%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCR2	100%	100%	100%	99.8%	99.3%	{HIV infection, susceptibility/resistance to}, 609423;Polycystic lung disease, 219600

CD19	100%	100%	100%	99.9%	98.5%	Immunodeficiency, common variable, 3, 613493
CD247	77.1%	71.6%	100%	100%	99.2%	?Immunodeficiency 25, 610163
CD27	100%	100%	100%	100%	98.6%	Lymphoproliferative syndrome 2, 615122
CD28	100%	100%	100%	100%	99.1%	?Immunodeficiency 123 with HPV-related verrucosis, 620901
CD3D	100%	100%	100%	100%	99%	Immunodeficiency 19, severe combined, 615617
CD3E	100%	100%	100%	99.8%	99.4%	Immunodeficiency 18, 615615;Immunodeficiency 18, SCID variant, 615615
CD3G	100%	100%	100%	100%	99.7%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	100%	100%	100%	100%	98.8%	Immunodeficiency 79, 619238;OKT4 epitope deficiency, 613949
CD40	100%	100%	100%	100%	99.4%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	100%	100%	99.4%	90.1%	69.2%	Immunodeficiency, X-linked, with hyper-IgM, 308230

CD46	100%	100%	100%	99.9%	99.6%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD48	100%	99.9%	100%	100%	99.6%	
CD55	96.1%	92.5%	100%	100%	99.3%	[Blood group Cromer], 613793;Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	100%	99.9%	100%	100%	99.9%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	100%	100%	100%	99.7%	98%	Lymphoproliferative syndrome 3, 618261
CD79A	100%	99.5%	100%	99.8%	97.5%	Agammaglobulinemia 3, 613501
CD79B	100%	100%	100%	99.8%	98.5%	Agammaglobulinemia 6, 612692
CD81	99.9%	98.7%	100%	99.8%	97.7%	Immunodeficiency, common variable, 6, 613496
CD8A	100%	100%	100%	100%	98.7%	Immunodeficiency 116, 608957
CDC42	100%	100%	100%	100%	99.5%	Takenouchi-Kosaki syndrome, 616737

CDCA7	100%	100%	100%	99.9%	99.1%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CEBPE	100%	100%	100%	100%	97.8%	?Immunodeficiency 108 with autoinflammation, 260570;Specific granule deficiency, 245480
CFB	100%	100%	100%	100%	99.4%	?Complement factor B deficiency, 615561;{Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924;{Macular degeneration, age-related, 14, reduced risk of}, 615489
CFD	100%	100%	99.9%	99.2%	94.8%	Complement factor D deficiency, 613912
CFH	97.5%	97.5%	100%	100%	99.6%	{Macular degeneration, age-related, 4}, 610698;Basal laminar drusen, 126700;Complement factor H deficiency, 609814;{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400

CR2	100%	100%	100%	100%	99.4%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927;?Immunodeficiency, common variable, 7, 614699
CRACR2A	100%	100%	100%	99.9%	99.1%	
CREBBP	100%	100%	100%	99.9%	98.6%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849
CSF2RA	48.9%	48.3%	50%	50%	49.5%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	100%	100%	100%	100%	99%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	100%	100%	100%	100%	99%	Neutropenia, severe congenital, 7, autosomal recessive, 617014;?Neutrophilia, hereditary, 162830
CTC1	100%	100%	100%	99.9%	98.8%	Cerebroretinal microangiopathy with calcifications and cysts, 612199

CTLA4	93.2%	93.2%	100%	100%	99.3%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100;{Diabetes mellitus, insulin-dependent, 12}, 601388;{Celiac disease, susceptibility to, 3}, 609755;{Hashimoto thyroiditis}, 140300;{Systemic lupus erythematosus, susceptibility to}, 152700
CTNNBL1	100%	100%	100%	100%	99.6%	?Immunodeficiency 99 with hypogammaglobulinemia and autoimmune cytopenias, 619846
CTPS1	100%	100%	100%	100%	99.5%	Immunodeficiency 24, 615897
CTSC	94.8%	94.5%	100%	99.9%	99.3%	Periodontitis 1, juvenile, 170650;Haim-Munk syndrome, 245010;Papillon-Lefevre syndrome, 245000
CXCR2	100%	100%	100%	99.6%	97.8%	?WHIM syndrome 2, 619407

CXCR4	99%	99%	100%	100%	99.3%	WHIM syndrome 1, 193670;Myelokathexis, isolated, 193670
CYBA	70.8%	69.7%	100%	99.9%	98.2%	Chronic granulomatous disease 4, autosomal recessive, 233690
CYBB	100%	100%	99%	89.2%	68.7%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645;Chronic granulomatous disease, X-linked, 306400
CYBC1	100%	100%	100%	100%	99.3%	Chronic granulomatous disease 5, autosomal recessive, 618935
DBF4	100%	100%	100%	99.9%	98.5%	
DBR1	100%	100%	100%	100%	99.2%	Xerosis and growth failure with immune and pulmonary dysfunction syndrome, 620510;{Encephalitis, acute, infection (viral)-induced, susceptibility to, 11}, 619441
DCLRE1B	100%	100%	100%	100%	99%	Dyskeratosis congenita, autosomal recessive 8, 620133
DCLRE1C	97.1%	97.1%	100%	100%	99.4%	Severe combined immunodeficiency, Athabascan type, 602450;Omenn syndrome, 603554

DDX41	100%	100%	100%	100%	98.8%	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871
DEF6	100%	100%	100%	99.6%	98.1%	Immunodeficiency 87 and autoimmunity, 619573
DHFR	100%	100%	100%	99.9%	99.1%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
ELANE	100%	100%	100%	100%	99.2%	Neutropenia, cyclic, 162800;Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	100%	99.4%	98.3%	85.4%	65.5%	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074
EPG5	100%	100%	100%	100%	99.4%	Vici syndrome, 242840
ERBIN	100%	100%	100%	99.9%	99.5%	

FAS	100%	99.9%	100%	99.9%	99.5%	Squamous cell carcinoma, burn scar-related, somatic;Autoimmune lymphoproliferative syndrome, type IA, 601859;{Autoimmune lymphoproliferative syndrome}, 601859
FASLG	100%	100%	100%	99.9%	99.2%	Autoimmune lymphoproliferative syndrome, type IB, 601859;{Lung cancer, susceptibility to}, 211980
FAT4	100%	100%	100%	99.9%	99.2%	Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FLT3LG	100%	100%	100%	99.8%	98%	?Immunodeficiency 125, 620926
FNIP1	100%	100%	100%	99.9%	99.3%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOXI3	97%	92.7%	99.8%	98.7%	94.4%	Craniofacial microsomia 2, 620444

GATA1	100%	99.8%	97%	86.4%	64.8%	Anemia, congenital, nonspherocytic hemolytic, 9, 301083;Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 159595;Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367;Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835;Thrombocytopenia with beta-thalassemia, X-linked, 314050
GATA2	85.7%	85.7%	100%	100%	99.2%	{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.

srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 4.2.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors