INVITAE IMMUNOLOGY GENE PANEL TESTS

Our testing menu covers the common causes of severe combined immunodeficiency (SCID) and periodic fever syndromes with curated panels designed by medical genetics experts.

CLINICAL AREA: IMMUNOLOGY

Test name	# gene(s)	Gene list
Primary Immunodeficiency		
Invitae Primary Immunodeficiency Panel	207	ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, AK2, AP3B1, ATM, B2M, BCL10, BLNK, BLOC1S6, BTK, CARD11, CARD14, CARD9, CASP10, CASP8, CD247, CD27, CD3D, CD3E, CD3G, CD40LG, CD79A, CD79B, CD8A, CEBPE, CHD7, CIITA, CLPB, COPA, CORO1A, CR2, CSF2RA, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DCLRE1B, DCLRE1C, DKC1, DNMT3B, DOCK2, DOCK8, ELANE, EPG5, FADD, FAS, FASLG, FERMT3, FOXN1, FOXP3, FPR1, G6PC3, GATA2, GFI1, HAX1, ICOS, IFIH1, IFNGR1, IFNGR2, IGLL1, IKBKB, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL2RA, IL2RG, IL36RN, IL7R, IRAK4, IRF7, IRF8, ISG15, ITCH, ITGB2, ITK, JAGN1, JAK3, LAMTOR2, LCK, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MAP3K14, MEFV, MOGS, MVK, MYD88, NBN, NCF2, NCF4, NFAT5, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP12, NLRP3, NOD2, NOP10, ORAI1, PARN, PGM3, PIK3CD, PIK3R1, PLCG2, PMM2, PNP, POLE, PRF1, PRKCD, PRKDC, PSMB8, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RBCK1, RFX5, RFXANK, RFXAP, RHOH, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RORC, RTEL1, SAMHD1, SEMA3E, SH2D1A, SH3BP2, SLC29A3, SLC35C1, SLC37A4, SLC7A7, SMARCAL1, SP110, SPINK5, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBX1, TCN2, TERC, TERT, TFNSF12, TICAM1, TINF2, TLR3, TMC6, TMC8, TMEM173, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TPP2, TRAF3, TRAF3IP2, TREX1, TRNT1, TTC7A, TYK2, UNC13D, UNC93B1, UNG, VPS13B, VPS45, WAS, WIPF1, XIAP, ZAP70, ZBTB24
Antibody Deficiencies		
Invitae Agammaglobulinemia Panel	6	BLNK, BTK, CD79A, CD79B, IGLL1, PIK3R1
Add-on hypogammaglobulinemia genes	5	GATA2, MOGS, SH2D1A, TRNT1, XIAP
Add-on common variable immunodeficiency genes	20	CD27, CR2, CTLA4, DCLRE1C, ICOS, IL21, IL21R, JAK3, LRBA, NFKB2, PIK3CD, PLCG2, PRKCD, RAC2, RAG1, STAT3, STXBP2, TNFRSF13B, TNFRSF13C, TNFSF12
Invitae Common Variable Immunodeficiency Panel	17	CD27, CR2, CTLA4, ICOS, IL21, IL21R, LRBA, NFKB2, PIK3CD, PIK3R1, PLCG2, PRKCD, RAC2, STAT3, TNFRSF13B, TNFRSF13C, TNFSF12
Add-on primary immunodeficiencies that can mimic common variable immunodeficiency	6	DCLRE1C, GATA2, JAK3, RAG1, RAG2, STXBP2
Add-on agammaglobulinemia/hypogammaglobulinemia genes	10	BLNK, BTK, CD79A, CD79B, GATA2, IGLL1, MOGS, SH2D1A, TRNT1, XIAP
Invitae Hyper IgE Syndrome Panel	4	DOCK8, PGM3, SPINK5, STAT3
Invitae Hyper IgM Syndrome Panel	3	AICDA, CD40LG, UNG
Add-on clinically overlapping genes	3	BTK, IL2RG, SH2D1A
Autoinflammatory Syndromes		
Invitae Autoinflammatory Syndromes Panel	72	ACP5, ADA, ADA2, ADAR, ADAM17, AICDA, BTK, CARD14, CD3G, CD40LG, CTLA4, COPA, CYBA, CYBB, DCLRE1C, DKC1, DOCK8, ELANE, FOXP3, G6PC3, ICOS, IFIH1, IL10, IL10RA, IL10RB, IL1RN, IL21, IL2RA, IL2RG, IL36RN, ITGB2, LIG4, LPIN2, LRBA, MEFV, MVK, NCF2, NCF4, NFAT5, NLRC4, NLRP12, NLRP3, NOD2, PIK3CD, PIK3R1, PLCG2, PSMB8, PSTPIP1, RAG1, RAG2, RBCK1, RNASEH2A, RNASEH2B, RNASEH2C, RTEL1, SAMHD1, SH2D1A, SH3BP2, SLC29A3, SLC37A4, STAT1, STAT3, STIM1, STXBP2, TMEM173, TNFRSF1A, TREX1, TRNT1, TTC7A, WAS, XIAP, ZAP70
Add-on Autoimmunity Genes	37	AIRE, AP3B1, BLOC1S6, CASP10, CASP8, CD27, CR2, FADD, FAS, FASLG, IL21R, ITCH, ITK, LYST, MAGT1, NFKB2, NFKBIA, ORAI1, PNP, PRF1, PRKCD, RAB27A, RAC2, RFX5, RFXANK, RFXAP, RMRP, SLC7A7, STAT5B, STX11, TBX1, TNFRSF13B, TNFRSF13C, TNFSF12, TPP2, UNC13D, UNG

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Autoinflammatory Syndromes (continued)		
Invitae Familial Cold Autoinflammatory Syndrome Panel	4	NLRC4, NLRP12, NLRP3, PLCG2
Invitae Familial Mediterranean Fever Test	1	MEFV
Add-on additional periodic fever syndromes genes	11	ADA2, ELANE, LPIN2, MVK, NLRC4, NLRP12, NLRP3, PSMB8, PSTPIP1, TNFRSF1A, TRNT1
Invitae Monogenic Inflammatory Bowel Disease Panel	46	ADA, ADAM17, AICDA, BTK, CD3G, CD40LG, CTLA4, CYBA, CYBB, DCLRE1C, DKC1, DOCK8, FOXP3, G6PC3, ICOS, IL10, IL10RA, IL10RB, IL21, IL2RA, IL2RG, ITGB2, LIG4, LRBA, MEFV, MVK, NCF2, NCF4, NFAT5, NLRC4, PIK3CD, PIK3R1, PLCG2, RAG1, RAG2, RTEL1, SH2D1A, SLC37A4, STAT1, STAT3, STIM1, STXBP2, TTC7A, WAS, XIAP, ZAP70
Add-on Increased risk alleles in NOD2 associated with Crohn's disease	1	NOD2
Invitae Periodic Fever Syndromes Panel	12	ADA2, ELANE, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, PSMB8, PSTPIP1, TNFRSF1A, TRNT1
Combined T/B Cell Deficiencies		
Invitae Comprehensive Severe Combined Immunodeficiency (SCID) and Combined Immunodeficiency (CID) Panel	50	ADA, AK2, B2M, BCL10, CARD11, CD247, CD27, CD3D, CD3E, CD3G, CD40LG, CD8A, CIITA, CORO1A, CTPS1, DCLRE1C, DOCK2, DOCK8, ICOS, IKBKB, IL21, IL21R, IL2RG, IL7R, ITK, JAK3, LCK, LIG4, LRBA, MAGT1, MALT1, MAP3K14, NHEJ1, PNP, PRKDC, PTPRC, RAC2, RAG1, RAG2, RFX5, RFXANK, RFXAP, RHOH, SH2D1A, STK4, TAP1, TAP2, TAPBP, TNFRSF4, ZAP70
Add-on combined immunodeficiency (CID) with syndromic features genes	36	ACD, ATM, CHD7, CTC1, DCLRE1B, DKC1, DNMT3B, EPG5, FOXN1, NBN, NFKBIA, NHP2, NOP10, ORAI1, PARN, PGM3, PMS2, POLE, RMRP, RTEL1, SEMA3E, SMARCAL1, SP110, SPINK5, STAT3, STAT5B, STIM1, TBX1, TCN2, TERC, TERT, TINF2, TTC7A, WAS, WIPF1, ZBTB24
Invitae Comprehensive Severe Combined Immunodeficiency (SCID) Panel	18	ADA, AK2, CD247, CD3D, CD3E, CORO1A, DCLRE1C, IL2RG, IL7R, JAK3, LIG4, NHEJ1, PNP, PRKDC, PTPRC, RAG1, RAG2, ZAP70
Add-on combined immunodeficiency (CID) genes	32	B2M, BCL10, CARD11, CD27, CD3G, CD40LG, CD8A, CIITA, CTPS1, DOCK2, DOCK8, ICOS, IKBKB, IL21, IL21R, ITK, LCK, LRBA, MAGT1, MALT1, MAP3K14, RAC2, RFX5, RFXANK, RFXAP, RHOH, SH2D1A, STK4, TAP1, TAP2, TAPBP, TNFRSF4
Add-on combined immunodeficiency (CID) with syndromic features genes	36	ACD, ATM, CHD7, CTC1, DCLRE1B, DKC1, DNMT3B, EPG5, FOXN1, NBN, NFKBIA, NHP2, NOP10, ORAI1, PARN, PGM3, PMS2, POLE, RMRP, RTEL1, SEMA3E, SMARCAL1, SP110, SPINK5, STAT3, STAT5B, STIM1, TBX1, TCN2, TERC, TERT, TINF2, TTC7A, WAS, WIPF1, ZBTB24
Invitae T-B-NK- Severe Combined Immunodeficiency (SCID) Panel	2	ADA, AK2
Invitae T-B-NK+ Severe Combined Immunodeficiency (SCID) Panel	6	DCLRE1C, LIG4, NHEJ1, PRKDC, RAG1, RAG2
Invitae T-B+NK- Severe Combined Immunodeficiency (SCID) Panel	2	IL2RG, JAK3
Invitae T-B+NK+ Severe Combined Immunodeficiency (SCID) Panel	6	CD247, CD3D, CD3E, CORO1A, IL7R, PTPRC
Invitae X-Linked Severe Combined Immunodeficiency (SCID) Test	1	IL2RG
Invitae Radiation-Sensitive Severe Combined Immunodeficiency (SCID) Panel	4	DCLRE1C, LIG4, NHEJ1, PRKDC

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Combined T/B Cell Deficiencies (continued)		
Invitae Combined Immunodeficiency (CID) Panel	33	B2M, BCL10, CARD11, CD27, CD3G, CD40LG, CD8A, CIITA, CTPS1, DOCK2, DOCK8, ICOS, IKBKB, IL21, IL21R, ITK, LCK, LRBA, MAGT1, MALT1, MAP3K14, RAC2, RFX5, RFXANK, RFXAP, RHOH, SH2D1A, STK4, TAP1, TAP2, TAPBP, TNFRSF4, ZAP70
Add-on combined immunodeficiencies (CID) with syndromic features	36	ACD, ATM, CHD7, CTC1, DCLRE1B, DKC1, DNMT3B, EPG5, FOXN1, NBN, NFKBIA, NHP2, NOP10, ORAI1, PARN, PGM3, PMS2, POLE, RMRP, RTEL1, SEMA3E, SMARCAL1, SP110, SPINK5, STAT3, STAT5B, STIM1, TBX1, TCN2, TERC, TERT, TINF2, TTC7A, WAS, WIPF1, ZBTB24
Invitae Syndromic Combined Immunodeficiency (CID) Panel	37	ACD, ATM, CHD7, CTC1, DCLRE1B, DKC1, DNMT3B, EPG5, FOXN1, NBN, NFKBIA, NHP2, NOP10, ORAI1, PARN, PGM3, PMS2, PNP, POLE, RMRP, RTEL1, SEMA3E, SMARCAL1, SP110, SPINK5, STAT3, STAT5B, STIM1, TBX1, TCN2, TERC, TERT, TINF2, TTC7A, WAS, WIPF1, ZBTB24
Disorders of Intrinsic and Innate Immunity		
Invitae Chronic Mucocutaneous Candidiasis Panel	4	IL17F, IL17RA, IL17RC, TRAF3IP2
Add-on syndromic chronic mucocutaneous candidiasis genes	7	AIRE, CARD9, IL12B, IL12RB1, RORC, STAT1, STAT3
Invitae Epidermodysplasia Verruciformis Panel	4	CXCR4, RHOH, TMC6, TMC8
Invitae Herpes Simplex Encephalitis Panel	4	TICAM1, TLR3, TRAF3, UNC93B1
Add-on predisposition to multiple viral infections genes	2	STAT1, TYK2
Invitae Mendelian Susceptibility to Mycobacterial Disease Panel	11	CYBB, GATA2, IFNGR1, IFNGR2, IL12B, IL12RB1, IRF8, ISG15, STAT1, STAT2, TYK2
Immune Dysregulation		
Invitae Monogenic Autoimmunity Panel	75	ACP5, ADA2, ADAR, AICDA, AIRE, AP3B1, BLOC1S6, BTK, CASP10, CASP8, CD27, CD40LG, CR2, CTLA4, CYBA, CYBB, DOCK8, FADD, FAS, FASLG, FOXP3, ICOS, IFIH1, IL10, IL10RA, IL10RB, IL2RA, IL21, IL21R, ITCH, ITK, LRBA, LYST, MAGT1, NCF2, NCF4, NFAT5, NFKB2, NFKBIA, ORAI1, PIK3CD, PIK3R1, PLCG2, PNP, PRF1, PRKCD, RAB27A, RAC2, RFX5, RFXANK, RFXAP, RNASEH2A, RNASEH2B, RNASEH2C, RMRP, SAMHD1, SH2D1A, SLC7A7, STAT1, STAT3, STAT5B, STIM1, STX11, STXBP2, TBX1, TMEM173, TNFRSF13B, TNFRSF13C, TNFSF12, TPP2, TREX1, UNC13D, UNG, WAS, XIAP
Add on Autoinflammatory syndrome genes	34	ADA, ADAM17, CARD14, CD3G, COPA, DCLRE1C, DKC1, ELANE, G6PC3, IL1RN, IL2RG, IL36RN, ITGB2, LIG4, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, PSMB8, PSTPIP1, RAG1, RAG2, RBCK1, RTEL1, SH3BP2, SLC29A3, SLC37A4, TNFRSF1A, TRNT1, TTC7A, ZAP70
Invitae Autoimmune Lymphoproliferative Disorders (ALPS) Panel	9	CASP8, CTLA4, FAS, FASLG, ITK, MAGT1, PIK3CD, PRKCD, STAT3
Add-on preliminary-evidence genes	2	CASP10, FADD
Invitae Hereditary Hemophagocytic Lymphohistiocytosis (HLH) Disorders Panel	21	ADA, AP3B1, BLOC1S6, BTK, CD27, IL2RA, IL2RG, ITK, LYST, MAGT1, MVK, PNP, PRF1, RAB27A, SH2D1A, SLC7A7, STX11, STXBP2, UNC13D, WAS, XIAP
Phagocytic Defects		
Invitae Phagocyte Defects Panel	27	CEBPE, CLPB, CSF2RA, CSF3R, CTSC, CYBA, CYBB, ELANE, FERMT3, FPR1, G6PC3, G6PD, GFI1, HAX1, ITGB2, JAGN1, LAMTOR2, NCF2, NCF4, PMM2, SLC35C1, SLC37A4, SPINK5, TAZ, VPS13B, VPS45, WAS
Well-Defined Syndromes		
Invitae Dyskeratosis Congenita Panel	7	CTC1, DKC1, NHP2, NOP10, TERC, TERT, TINF2
Invitae Immunodeficiency, Centromeric Instability, Facial Anomalies Syndrome Panel	2	DNMT3B, ZBTB24

