

Invitae has added 26 new panels and expanded three panels in immunology. The panels are based on the latest research—one more way in which we're working to provide you and your patients the answers that you need.

## PRIMARY IMMUNODEFICIENCY PANEL - NEW

As the availability of molecular testing expands for primary immunodeficiencies (PIDs), studies are showing that the phenotypic spectrum of individual PIDs are much greater than previously thought and often overlap with other PIDs (PMID: 27577878, 27931534).

The Invitae Primary Immunodeficiency Panel provides our most comprehensive coverage of primary immunodeficiencies, including over 200 genes, to help precisely identify the underlying defect and help you deliver the most appropriate treatment.

## COMBINED T/B CELL DEFICIENCIES PANELS - NEW & UPDATED

Nine new tests and panels are included in our combined T/B cell deficiencies tests, including the Invitae Comprehensive Severe Combined Immunodeficiency (SCID) and Combined Immunodeficiency (CID) Panel.

In addition, you can choose more focused panels based on flow cytometry results, the presence of radiosensitivity, or the presence of syndromic features.

## ANTIBODY DEFICIENCIES PANELS - NEW

We have designed four new panels such as the Invitae Agammaglobulinemia Panel, Invitae Hyper IgE Panel, Invitae Hyper IgM Panel, and Invitae Common Variable Immunodeficiency Panel to cover testing for antibody deficiencies.

## AUTOINFLAMMATORY SYNDROMES PANELS - NEW & UPDATED

The new Invitae Autoinflammatory Syndromes Panel and Invitae Monogenic Inflammatory Bowel Disease Panel as well as the Invitae Familial Mediterranean Fever Test and expanded Invitae Periodic Fever Syndromes Panel cover a wider range of autoinflammatory syndromes.

## PHAGOCYTE DEFECTS PANEL - NEW

The new Invitae Phagocyte Defects Panel, includes testing for severe congenital neutropenia, leukocyte adhesion deficiency, chronic granulomatous disease, and syndromic causes of neutropenia.

## IMMUNE DYSREGULATION PANELS - NEW

We have designed three new dysregulation panels, including the 73-gene Invitae Autoimmunity Panel, the Invitae Autoimmune Lymphoproliferative Disorders Panel, and the Invitae Hereditary Hemophagocytic Lymphohistiocytosis (HLH) Disorders Panel for immune dysregulation.

## DISORDERS OF INTRINSIC AND INNATE IMMUNITY PANELS - NEW

We have designed four new panels to test for intrinsic and innate immunity diseases including the Invitae Chronic Mucocutaneous Candidiasis Panel and the Invitae Herpes Simplex Encephalitis Panel.

All new and updated panels are now available. Visit [www.invitae.com](http://www.invitae.com) to order online or download a paper order form. If you have questions, please do not hesitate to contact Client Services at 800-436-3037 or [clientservices@invitae.com](mailto:clientservices@invitae.com), or reach out to your regional manager.

## DID YOU KNOW?

Pathogenic copy number and large indel variants make up 10% of all pathogenic variants, yet are traditionally considered difficult to identify using next-generation sequencing. Invitae's methods have been shown to routinely identify these variants, in large part because we include deletion/duplication analysis with all testing at no additional charge.

Learn more in Rebecca Truty's ACMG presentation, Tracing the dark matter: Prevalence of copy number and structural variants across Mendelian disorders, available at: [www.invitae.com/presentations](http://www.invitae.com/presentations)

## 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: METABOLIC DISORDERS, NEWBORN SCREENING & IMMUNOLOGY

#### PRIMARY IMMUNODEFICIENCY

##### Invitae Primary Immunodeficiency Panel (207 genes)

###### Primary panel (207 genes)

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	GF11	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	PMS2	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	TBK1	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 (up to 32 genes)

###### Primary panel (6 genes)

BLNK	BTK	CD79A	CD79B	IGLL1	PIK3R1
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###### Add-on hypogammaglobulinemia genes (5 genes)

GATA2	MOGS	SH2D1A	TRNT1	XIAP
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###### Add-on common variable immunodeficiency genes (21 genes)

CD27	CR2	CTLA4	DCLRE1C	ICOS	IL21	IL21R	JAK3	LRBA	NFKB2	PIK3CD	PIK3R1
PLCG2	PRKCD	RAC2	RAG1	STAT3	STXBP2	TNFRSF13B	TNFRSF13C	TNFSF12			

##### Invitae Common Variable Immunodeficiency Panel (up to 32 genes)

###### Primary panel (17 genes)

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 (5 genes)

DCLRE1C	GATA2	JAK3	RAG1	STXBP2
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###### Add-on agammaglobulinemia/hypogammaglobulinemia genes (10 genes)

BLNK	BTK	CD79A	CD79B	GATA2	IGLL1	MOGS	SH2D1A	TRNT1	XIAP
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##### Invitae Hyper IgE Syndrome Panel (4 genes)

###### Primary panel (4 genes)

DOCK8	PGM3	SPINK5	STAT3
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##### Invitae Hyper IgM Syndrome Panel (up to 7 genes)

###### Primary panel (4 genes)

AICDA	CD40	CD40LG	UNG
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###### Add-on clinically overlapping genes (3 genes)

BTK	IL2RG	SH2D1A
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#### AUTOINFLAMMATORY SYNDROMES

##### Invitae Autoinflammatory Syndromes Panel (26 genes)

###### Primary panel (26 genes)

ADA2	ADAM17	CARD14	COPA	ELANE	IL10	IL10RA	IL10RB	IL1RN	IL36RN	LPIN2	MEFV
MVK	NFAT5	NLRC4	NLRP12	NLRP3	NOD2	PLCG2	PSMB8	PSTPIP1	RBCK1	SH3BP2	SLC29A3
TNFRSF1A	TRNT1										

##### Invitae Familial Cold Autoinflammatory Syndrome Panel (4 genes)

###### Primary panel (4 genes)

NLRC4	NLRP12	NLRP3	PLCG2
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##### Invitae Familial Mediterranean Fever Panel (up to 12 genes)

###### Primary panel (1 gene)

MEFV
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###### Add-on additional syndromes genes (11 genes)

ADA2	ELANE	LPIN2	MVK	NLRC4	NLRP12	NLRP3	PSMB8	PSTPIP1	TNFRSF1A	TRNT1
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##### Invitae Monogenic Inflammatory Bowel Disease Panel (46 genes)

###### Primary panel (46 genes)

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		

##### Invitae Periodic Fever Syndromes Panel (12 genes)

###### Primary panel (12 genes)

ADA2	ELANE	LPIN2	MEFV	MVK	NLRC4	NLRP12	NLRP3	PSMB8	PSTPIP1	TNFRSF1A	TRNT1
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## INVITAE IMMUNOLOGY GENE PANEL TESTS (continued)

### COMBINED T/B CELL DEFICIENCIES

**Invitae Comprehensive Severe Combined Immunodeficiency (SCID) and Combined Immunodeficiency (CID) Panel**  
(up to 85 genes)

**Primary panel (49 genes)**

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	STK4	TAP1	TAP2	TAPBP	TNFRSF4
ZAP70											

**Add-on combined immunodeficiency (CID) with syndromic features genes (36 genes)**

ACD	ATM	CHD7	CTC1	DCLRE1B	DKC1	DNMT3B	EPG5	FOXN1	NBN	NFKBIA	NHP2
NOPI0	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**  
(up to 86 genes)

**Primary panel (18 genes)**

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 genes)**

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	STK4	TAP1	TAP2	TAPBP	TNFRSF4	ZAP70				

**Add-on combined immunodeficiency (CID) with syndromic features genes (36 genes)**

ACD	ATM	CHD7	CTC1	DCLRE1B	DKC1	DNMT3B	EPG5	FOXN1	NBN	NFKBIA	NHP2
NOPI0	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 genes)

**Primary panel (2 genes)**

ADA	AK2
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**Invitae T-B-NK+ Severe Combined Immunodeficiency (SCID) Panel**  
(6 genes)

**Primary panel (6 genes)**

DCLRE1C	LIG4	NHEJ1	PRKDC	RAG1	RAG2
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**Invitae T-B+NK- Severe Combined Immunodeficiency (SCID) Panel**  
(2 genes)

**Primary panel (2 genes)**

IL2RG	JAK3
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**Invitae T-B+NK+ Severe Combined Immunodeficiency (SCID) Panel**  
(6 genes)

**Primary panel (6 genes)**

CD247	CD3D	CD3E	CORO1A	IL7R	PTPRC
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**Invitae X-Linked Severe Combined Immunodeficiency (SCID) Test**  
(1 gene)

**Primary panel (1 gene)**

IL2RG
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**Invitae Radiation-Sensitive Severe Combined Immunodeficiency (SCID) Panel**  
(4 genes)

**Primary panel (4 genes)**

DCLRE1C	LIG4	NHEJ1	PRKDC
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**Invitae Combined Immunodeficiency (CID) Panel**  
(up to 68 genes)

**Primary panel (32 genes)**

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	STK4	TAP1	TAP2	TAPBP	TNFRSF4	ZAP70				

**Add-on combined immunodeficiencies (CID) with syndromic features genes (36 genes)**

ACD	ATM	CHD7	CTC1	DCLRE1B	DKC1	DNMT3B	EPG5	FOXN1	NBN	NFKBIA	NHP2
NOPI0	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 genes)

**Primary panel (37 genes)**

ACD	ATM	CHD7	CTC1	DCLRE1B	DKC1	DNMT3B	EPG5	FOXN1	NBN	NFKBIA	NHP2
NOPI0	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**  
(up to 11 genes)

**Primary panel (4 genes)**

IL17F	IL17RA	IL17RC	TRAF3IP2
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**Add-on syndromic chronic mucocutaneous candidiasis genes (7 genes)**

AIRE	CARD9	IL12B	IL12RB1	RORC	STAT1	STAT3
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**Invitae Epidermodysplasia Verruciformis Panel**  
(4 genes)

**Primary panel (4 genes)**

CXCR4	RHOH	TMC6	TMC8
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**Invitae Herpes Simplex Encephalitis Panel**  
(up to 7 genes)

**Primary panel (5 genes)**

TBK1	TICAM1	TLR3	TRAF3	UNC93B1
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**Add-on predisposition to multiple viral infections genes (2 genes)**

STAT1	TYK2
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**Invitae Mendelian Susceptibility to Mycobacterial Disease Panel**  
(16 genes)

**Primary panel (16 genes)**

ACP5	ADAR	CYBB	GATA2	IFNGR1	IFNGR2	IL12B	IL12RB1	IRAK4	IRF8	ISG15	MYD88
SAMHD1	STAT1	STAT2	TYK2								

## INVITAE IMMUNOLOGY GENE PANEL TESTS (continued)

### IMMUNE DYSREGULATION

#### Invitae Monogenic Autoimmunity Panel (73 genes)

##### Primary panel (73 genes)

ACPS5	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	LRBA	LYST	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											

#### Invitae Autoimmune Lymphoproliferative Disorders (ALPS) Panel (up to 10 genes)

##### Primary panel (8 genes)

CASP8	CTLA4	FAS	FASLG	ITK	MAGT1	PIK3CD	PRKCD
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##### Add-on preliminary evidence genes (2 genes)

CASP10	FADD
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#### Invitae Hereditary Hemophagocytic Lymphohistiocytosis (HLH) Disorders Panel (21 genes)

##### Primary panel (21 genes)

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 genes)

##### Primary panel (27 genes)

CEBPE	CLPB	CSF2RA	CSF3R	CTSC	CYBA	CYBB	ELANE	FERMT3	FPR1	G6PC3	G6PD
GF11	HAX1	ITGB2	JAGN1	LAMTOR2	NCF2	NCF4	PMM2	SLC35C1	SLC37A4	SPINK5	TAZ
VPS13B	VPS45	WAS									

### WELL-DEFINED SYNDROMES

#### Invitae Dyskeratosis Congenita Panel (7 genes)

##### Primary panel (7 genes)

CTC1	DKC1	NHP2	NOP10	TERC	TERT	TINF2
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#### Invitae Immunodeficiency, Centromeric Instability, Facial Anomalies Syndrome Panel (2 genes)

##### Primary panel (2 genes)

DNMT3B	ZBTB24
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