Comprehensive Primary Immunodeficiency

Primary immunodeficiency is the term used to describe immune system disorders caused by genetic factors. Patients with these conditions are highly susceptible to illness and may experience recurring infections and susceptibility to autoimmunity, autoinflammatory diseases, and/or allergies. Individuals may also have a difficult time recovering from these conditions and require more intensive care than other patients.

WHAT CAUSES PRIMARY IMMUNODEFICIENCY?

The discovery of new genes within the last 10 years has influenced our understanding of immunodeficiencies. The definition of "primary immunodeficiencies" as fundamental defects in the immune response has shifted toward the broader concept of "inborn errors of immunity" as a comprehensive group of different symptoms, including infection, autoinflammation, autoimmunity, allergy, and malignancy. Inborn errors of immunity are caused by genes that influence the immune system and related pathways.

TYPES OF IMMUNODEFICIENCY

Inborn errors of immunity are used to describe over 400 conditions. Highlighting some examples are:

- **Severe combined immunodeficiency**
- **Primary immunodeficiency**
- **Fanconi anemia**
- **Primary ciliary dyskinesia**

WHO IS THIS TEST FOR?

This panel may be appropriate for anyone who has a personal or family history of frequent infections, fevers, allergies, certain malignancies or rash, particularly if infections are recurrent and difficult to treat, require hospitalization or IV antibiotics, or are caused by an uncommon organism. Also if the person has suspected hereditary immunodeficiency, antibody deficiencies, autoinflammatory disorders, combined immunodeficiencies, or immune dysregulation disorders.

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BENEFITS OF GENETIC TESTING

This panel can help confirm a diagnosis and guide the course of treatment. Patients with immunodeficiency can take precautions to prevent infection. Diagnosis through genetic testing can help with the development of a management plan.

Genetic testing for primary immunodeficiency can:

- Establish or confirm the appropriate diagnosis
- Identify risks for additional health-related symptoms
- Assist in modifying lifestyle changes
- Result in more personalized treatment and symptom management
- Inform family members about their own risk factors
- Connect patients to relevant resources & support
- Provide options for family planning

RELATED PANELS

Visit Fulgent website for most up-to-date list

- **Agammaglobulinemia NGS Panel**
- **Primary Antibody Deficiency NGS Panel**
- **Severe Combined Immunodeficiency NGS Panel**
- **Periodic Fever and Autoinflammatory Disorders NGS Panel**

TEST SPECIFICATIONS

Acceptable Sample Requirements (Kits available upon request)

- Blood, two 4-mL EDTA tubes, lavender top
- Extracted DNA, 3 µg in EB buffer
- Buccal swab or saliva

Turnaround Time 2-3 weeks

Coverage ≥96% at 20x

Reporting

VUS, likely pathogenic, and pathogenic variants

Customization

Customizable gene list, VUS opt-out

TEST DESCRIPTION

Sequencing | Del/Dup | Rush/STAT | Exclude VUS | Duo/Trio | MCC

REFERENCES

PMID: 35020168 & PMID: 32226610

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GENES (592)

ACD
ACP5
ACTB
ADA
ADA2
ADAM17
ADAMTS13
ADAMTS3
ADAR
ADGRE2
ADIPOR1
ADIPOR2
AICDA
AIRE
AK2
ALG6
ALPI
ALPK1
ANGPT1
ANKZF1
AP1S3
AP3B1
AP3D1
APOL1
ARHGEF1
ARMC4
ARPC1B
ASAH1
ATG4A
ATM
ATP6AP1
B2M
BACH2
BCL10
BCL11B
BLM
BLNK
BLOC1S3
BLOC1S6
BRCA2
BRIP1
BTK
C1QA
C1QB
C1QC
C1R
C1S
C2
C3
C3AR1
C4BPA
C4BPB
C5
C5AR1
C6
C7
C8A
C8B
C8G
C9
CARD11
CARD14
CARD8
CARD9
CARMIL2
CASP10
CASP8
CAVIN1
CBL
CCBE1
CCDC103
CCDC114
CCDC151
CCDC39
CCDC40
CCDC65
CCNO
CD19
CD247
CD27
CD3D
CD3E
CD3G
CD4
CD40
CD40LG
CD46
CD55
CD59
CD70
CD79A
CD79B
CD81
CD8A
CDC42
CDCA7
CEBPE
CENPF
CFAP298
CFAP300
CFB
CFD
CFH
CFHR1
CFHR2
CFHR3
CFHR4
CFHR5
CFI
CFP
CFTR
CHD7
CIB1
CIITA
CLCN7
CLEC7A
CLPB
COG6
COL7A1
COLEC11
COPA
CORO1A
CR2
CREBBP
CSF2RA
CSF2RB
CSF3R
CTC1
CTLA4
CTNNBL1
CTPS1
CTSC
CXCR2
CXCR4
CYBA
CYBB
CYBC1
CYP27A1
DBR1
DCLRE1B
DCLRE1C
DDX58
DEF6
DGAT1
DGKE
DHFR
DIAPH1
DKC1
DNAAF1
DNAAF2
DNAAF3
DNAAF4
DNAAF5
DNAH1
DNAH11
DNAH5
DNAH9
DNAI1
DNAI2
DNAJB13
DNAJC21
DNAL1
DNASE1L3
DNASE2
DNMT3B
DOCK2
DOCK8
DRC1
DTNBP1
EFL1
ELANE
EPG5
ERBIN
ERCC2
ERCC3
ERCC4
ERCC6L2
ETV6
EXTL3
F11
F13A1
F13B
F5
F7
F8
F9
FAAP24
FADD
FANCA
FANCB
FANCC
FANCD2
FANCE
FANCF
FANCG
FANCI
FANCL
FANCM
FAS
FASLG
FAT4
FCGR3A
FCHO1
FCN3
FERMT3
FGA
FGB
FNIP1
FOXJ1
FOXN1
FOXP3
FPR1
G6PC
G6PC3
G6PD
GAS2L2
GAS8
GATA1
GATA2
GFI1
GINS1
GP1BA
GP1BB
GP9
GTF2H5
GUCY2C
HAVCR2
HAX1
HELLS
HMOX1
HPS1
HPS3
HPS4
HPS5
HPS6
HYDIN
HYOU1
ICOS
IFIH1
IFNAR1
IFNAR2
IFNG
IFNGR1
IFNGR2
IGHM
IGLL1
IKBKB
IKZF1
IL10
IL10RA
IL10RB
IL12B
IL12RB1
IL17F
IL17RA
IL17RC
IL18BP
IL1RN
IL2
IL21
IL21R
IL23R
IL2RA
IL2RB
IL2RG
IL36RN
IL6R
IL6ST
IL7
IL7R
INO80
INSR
INVS
IRAK1
IRAK4
IRF2BP2
IRF3
IRF4
IRF7
IRF8
IRF9
ISG15
ITCH
ITGAM
ITGB2
ITK
IVNS1ABP
JAGN1
JAK1
JAK2
JAK3
KDM6A
KIT
KMT2D
KRAS
LAMTOR2
LAT
LCK
LCP2
LIG1
LIG4
LPIN2
LRBA
LRRC6
LRRC8A
LSM11
LYST
MAGT1
MALT1
MAN2B1
MANBA
MAP3K14
MASP1
MASP2
MBL2
MC2R
MCIDAS
MCM10
MCM4
MEFV
MKL1
MLPH
MOGS
MPL
MPO
MRE11
MS4A1
MSH6
MSN
MTHFD1
MVK
MYD88
MYH9
MYO5A
MYSM1
NBAS
NBN
NCF1
NCF2
NCF4
NCKAP1L
NCSTN
NFAT5
NFE2L2
NFKB1
NFKB2
NFKBIA
NHEJ1
NHP2
NKX2-5
NLRC4
NLRP1
NLRP12
NLRP3
NME8
NOD2
NOP10
NRAS
NSMCE3
OAS1
OFD1
ORAI1
OSTM1
OTULIN
PALB2
PARN
PAX1
PCCA
PCCB
PEPD
PGM3
PI4KA
PIGA
PIH1D3
PIK3CD
PIK3CG
PIK3R1
PLCG2
PLEKHM1
PLG
PLVAP
PMM2
PMS2
PNP
POLA1
POLD1
POLD2
POLE
POLE2
POLR3A
POLR3C
POLR3F
POMP
PRF1
PRG4
PRKCD
PRKDC
PROC
PROS1
PSENEN
PSMA3
PSMB4
PSMB8
PSMB9
PSMG2
PSTPIP1
PTEN
PTPRC
RAB27A
RAC2
RAD50
RAD51C
RAG1
RAG2
RANBP2
RASGRP1
RBCK1
RBM8A
RECQL4
REL
RELA
RELB
RFWD3
RFX5
RFXANK
RFXAP
RHOH
RIPK1
RMRP
RNASEH2A
RNASEH2B
RNASEH2C
RNF113A
RNF168
RNF31
RNU4ATAC
RORC
RPGR
RPL11
RPL15
RPL26
RPL35A
RPL36
RPL5
RPS10
RPS15
RPS15A
RPS17
RPS19
RPS24
RPS26
RPS27A
RPS28
RPS29
RPS7
RPSA
RSPH1
RSPH3
RSPH4A
RSPH9
RTEL1
RUNX1
SAMD9
SAMD9L
SAMHD1
SAR1B
SBDS
SEC61A1
SEMA3E
SERPING1
SGPL1
SH2D1A
SH3BP2
SH3KBP1
SI
SKIV2L
SLC10A2
SLC26A3
SLC29A3
SLC35A1
SLC35C1
SLC37A4
SLC39A4
SLC39A7
SLC46A1
SLC51B
SLC5A1
SLC7A7
SLC9A3
SLX4
SMARCAL1
SMARCD2
SNORA31
SNX10
SOCS1
SP110
SPAG1
SPINK5
SPINT2
SPPL2A
SRP54
SRP72
STAT1
STAT2
STAT3
STAT4
STAT5B
STIM1
STK36
STK4
STN1
STX11
STX3
STXBP2
TAP1
TAP2
TAPBP
TAZ
TBK1
TBX1
TCF3
TCIRG1
TCN2
TERC
TERT
TET2
TFRC
TGFB1
TGFBR1
TGFBR2
THBD
TICAM1
TINF2
TIRAP
TLR2
TLR3
TLR7
TMC6
TMC8
TMEM173
TMPRSS15
TNFAIP3
TNFRSF11A
TNFRSF13B
TNFRSF13C
TNFRSF1A
TNFRSF4
TNFRSF9
TNFSF11
TNFSF12
TNFSF13
TONSL
TOP2B
TP63
TPP1
TPP2
TRADD
TRAF3
TRAF3IP2
TREX1
TRNT1
TTC12
TTC25
TTC37
TTC37
TTC7A
TYK2
UBA1
UBE2T
UNC119
UNC13D
UNC93B1
UNG
USB1
USP18
VAV1
VPS13B
VPS45
VSIG4
WAS
WDR1
WIPF1
WNT2B
WRAP53
XIAP
XK
XRCC2
ZAP70
ZBTB24
ZCCHC8
ZMYND10
ZNF341
ZNFX1

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A Patient's Guide to Genetic Testing

What does a genetic test check for?

Genetic testing checks the order of one's DNA sequence (coded by the letters A, T, G, C) in specific genes linked to genetic conditions. Letters that were added, missing, or changed, are known as variants and can sometimes be harmful to one's health, increasing the risk for a genetic condition.

What are the potential results?

There are three possible results from genetic testing:

Positive

A pathogenic/likely pathogenic variant is detected in one's DNA. This type of variant is known to increase one's risk of a genetic condition. Identifying the specific gene involved can help confirm a diagnosis, inform screening and management, and reveal risk factors for an individual and/or their family.

Negative

No variation known to be associated with a genetic condition was detected in one's DNA . While a result may not show an increased risk for the condition(s) tested for, one can still be at risk for disease, especially if there is a family history.

Variant of Uncertain Significance (VUS)

A variant was detected in one's DNA, however, not enough information is known about this variant to determine whether or not it is known cause the condition(s) tested for. More research is needed to better understand this variant.

What about family members?

Children, siblings, and parents of individuals who have a variant(s) identified in genetic testing could carry the same variant(s) and benefit from testing. Regardless of whether or not a variant was identified, individuals can still be at an increased risk for a genetic condition, especially with a family history.

Do genetic test results affect health insurance or employment?

No, the Genetic Information Nondiscrimination Act (GINA) was signed into law in 2008.
It protects individuals from discrimination by an employer or a health insurance company based on genetic testing results and genetic information. GINA does not protect against life and disability insurance discrimination. For more information on GINA, go to www.ginahelp.org.

Where can I learn more?

Medline Plus/Genetics Home Reference - medlineplus.gov/genetics/understanding
National Society of Genetic Counselors - nsgc.org
Fulgent Genetics - fulgentgenetics.com/products/carrierscreening/learning.html

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