OMNIHEALTH
DIAGNOSTICS

PRIMARY IMMUNODEFICIENCY

Primary Immunodeficiency

Immunodeficiency is when a part of the immune system does not work correctly. Genetic, or inherited, immunodeficiencies are called primary immunodeficiencies, whereas secondary immunodeficiencies are caused by environmental factors, such as use of certain medications or poor nutrition. People with immunodeficiency tend to get sick more often with ear infections, sinus infections, pneumonia, and skin infections. They also have longer infections that are hard to treat with regular antibiotics and may result in hospitalization. Infants may have poor weight gain and digestive problems like diarrhea.

WHAT CAUSES PRIMARY IMMUNODEFICIENCY?

Primary immunodeficiency is caused by pathogenic (disease-causing) variants in genes that help develop the immune system and keep it working. These variants may make it easier for germs to enter the body, make it more difficult for the body to identify germs, or make it so the body cannot "remember" how to fight off germs it has encountered before.

ASSOCIATED CONDITIONS

Primary immunodeficiency disorders may be isolated (occurring with no other symptoms) or as one of several features of a more complex genetic syndrome. Conditions associated with primary immunodeficiency include but are not limited to:

- Adenosine deaminase deficiency
- Agammaglobulinemia (X-linked and autosomal recessive)
- Ataxia telangiectasia
- Chronic granulomatous disease
- Immunoglobulin A deficiency
- Wiskott-Aldrich syndrome
- Hyper-IgE syndrome
- X-linked SCID (severe combined immunodeficiency)

GENE LIST

BLM, BRCA2, CFTR, F9, F5, FANCC, G6PD, G6PC, JAK2, MSH6, MYD88, PALB2, NRAS, PMS2, PLCG2, PTEN, RUNX1, MPL, TERT, F13B, F7, FGB, STAT1, STAT3, MEFV, CYBB, JAGN1, STK4, CYBA, NFKB2, CDX1, PIK3CD, MSH2, VPS13B, BRCA1, ATM, RFXANK, PTPRC, NCF1, TNFRSF13B, ITGB2, IFNGR1, IFNGR2, RAG1, RAG2, SPINK5, BTK

WHO IS THIS TEST FOR?

This panel may be appropriate for anyone who has a personal or family history of frequent infections, fevers, or rash, particularly if infections do not completely clear up or keep coming back, require hospitalization or IV antibiotics, or are caused by an uncommon organism.

BENEFITS OF GENETIC TESTING

Genetic testing for Primary Immunodeficiency can:

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

TEST SPECIFICATIONS

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Acceptable Sample Requirements
• Buccal swab or saliva

Turnaround Time 2-3 weeks

Coverage ≥96% at 20x

Reporting
likely pathogenic, and pathogenic variants

Customization
Customizable gene list

GET CONNECTED

Primary Immunodeficiency (PI) | CDC - cdc.gov/genomics/disease/primary_immunodeficiency.htm

Immune Deficiency Foundation - primaryimmune.org/about-primary-immunodeficiencies

Primary Immunodeficiency Disease Overview - aaaai.org/Conditions-Treatments/Primary-Immunodeficiency-Disease/Primary-Immunodeficiency-Disease-Overview

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?

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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, informscreening 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.

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

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