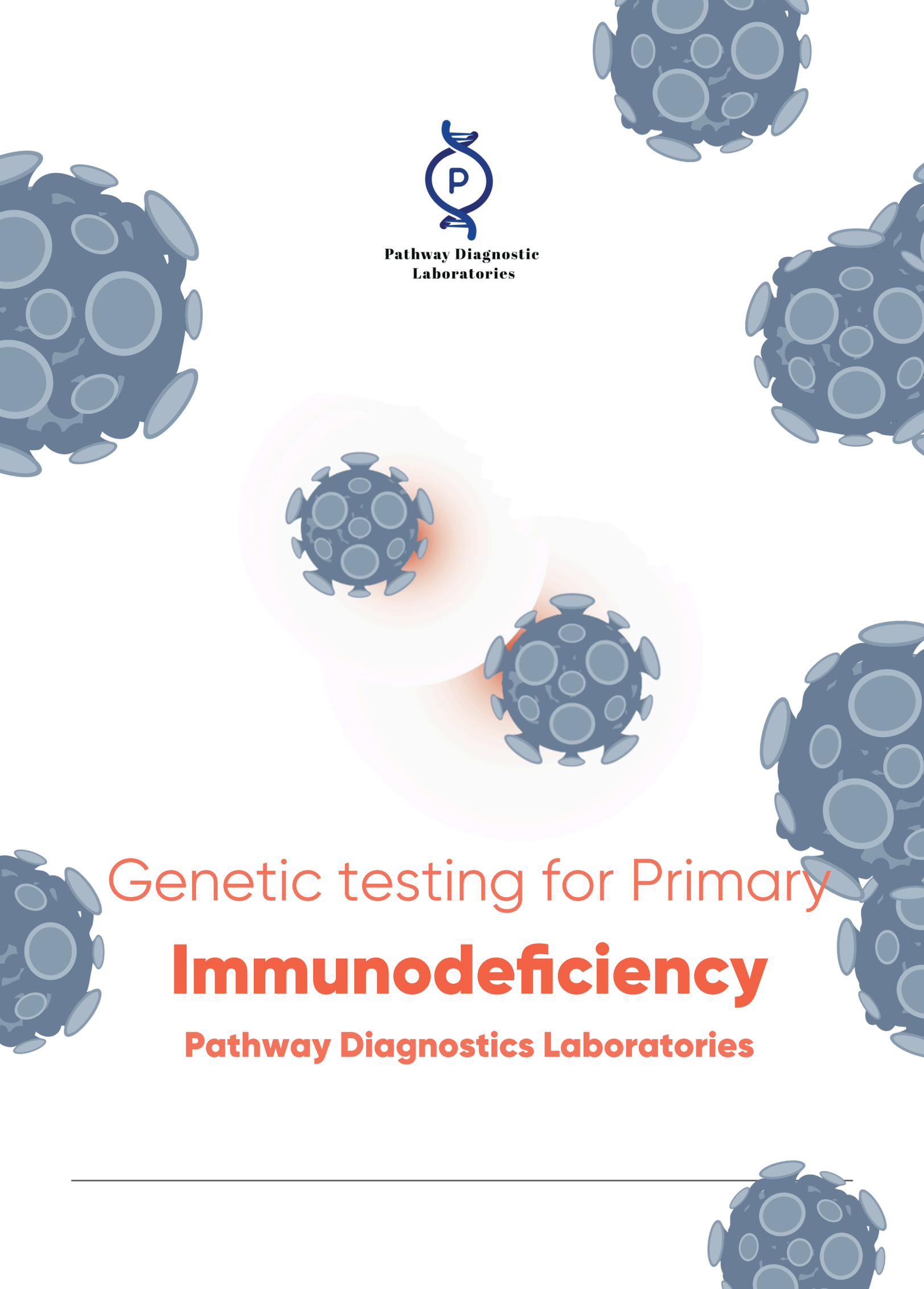


Pathway Diagnostic Laboratories

Genetic testing for Primary

Immunodeficiency

Pathway Diagnostics Laboratories

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 panel for Primary Immunodeficiency
(47 - Genes Panel)

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

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

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.

Acceptable sample requirements
Buccal Swab or Saliva

Turnaround time - 2-3 weeks
Coverage _>96% at 20x

Reporting
Likelypathogenic 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

UNLOCKING THE SECRETS OF YOUR GENENTIC HEALTH
COMPREHENSIVE GENETIC TESTING AT
PATHWAY DIAGNOSTICS LABORATORIES

PATHWAY DIAGNOSTICS LABORATORIES MAY BE A GOOD CHOICE FOR GENETIC TESTING FOR SEVERAL REASONS:

HIGH-QUALITY TESTING

Pathway Diagnostics Laboratories uses advanced technology and have experienced technicians to ensure that the testing is performed to the highest standards

FAST TURNAROUND TIME

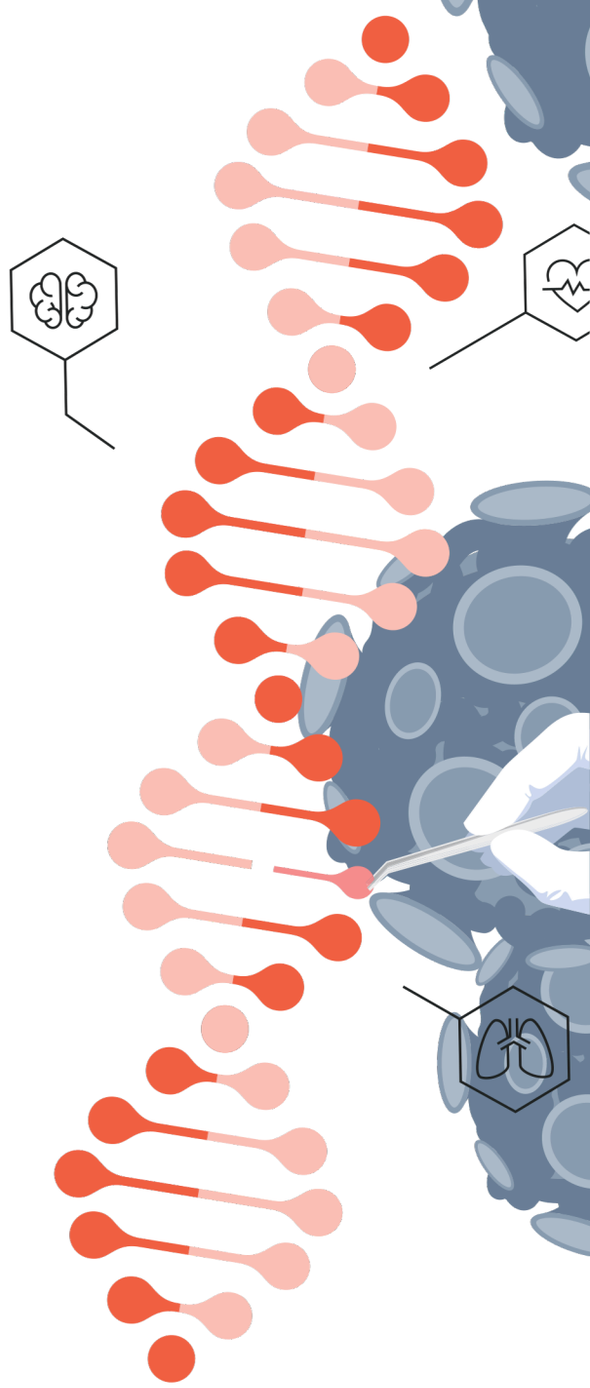
Pathway Diagnostics Laboratories has a fast turn around time for test results. This means that doctors can quickly get the information they need to make informed decisions about their patients' treatment.

COMPREHENSIVE TESTING OPTIONS

We offer a wide range of genetic testing options, including tests for Diabetes predict, Cancer genetics, Cardiovascular, Drug-Gene Interactions and Neurological disorders.

COLLABORATIVE APPROACH

Pathway Diagnostics Laboratories works closely with doctors to ensure that the testing is tailored to the specific needs of each patient. They provide personalized support and guidance throughout the testing process.



DECODING THE MYSTERY OF GENETIC TESTING: A PATIENT'S ROADMAP TO BETTER HEALTH

Genetic testing is a medical test that examines your DNA to identify any changes or mutations that may beassociated with a specific disease or condition. This guide aims to provide you with an overview of genetic testing, its significance, testing outcomes, and how it could be beneficial to you and your family members.

WHAT IS GENETIC TESTING?

DNA is the genetic material that contains the instructions for thedevel-opment, growth, and function of all living organisms. Genetic testing can provide information about inherited diseases or conditions, predispositions to certain diseases, and the likelihood of passing these conditions onto your children.

WHY IS GENETIC TESTING SIGNIFICANT?

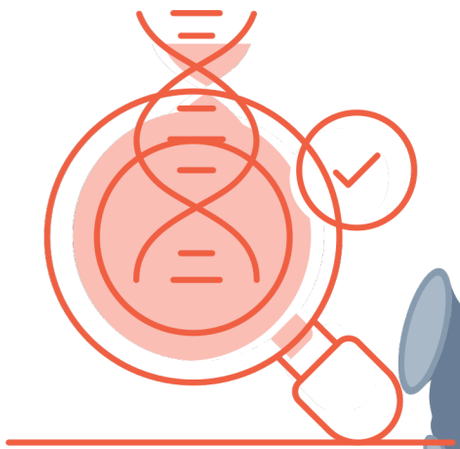
Genetic testing can provide valuable information that can help you and your healthcare provider make informed decisions about your health. It can help to:

DIAGNOSE GENETIC DISEASES:

Genetic testing can identify changes or mutations in genes that are associated with specific diseases or conditions. This information can help healthcare providers diagnose genetic diseases and develop appropriate treatment plans.

INFORM REPRODUCTIVE DECISIONS:

Genetic testing can help individuals and couples make informed decisions about family planning. If a genetic mutation is identified, it can be used to determine the likelihood of passing the condition onto future children.



DETERMINE THE RISK OF DEVELOPING A DISEASE:

Some genetic mutations are associated with an increased risk of developing certain diseases or conditions. Genetic testing can help identify these mutations, allowing for early intervention or prevention.

PERSONALIZE TREATMENT PLANS:

Genetic testing can provide information about how an individual may respond to certain medications or treatments. This information can help healthcare providers personalize treatment plans for betier outcomes.

TESTING OUTCOMES

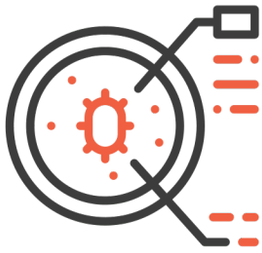
The results of genetic testing can be positive, negative, or uncertain. A positive result indicates that a genetic mutation associated with a specific disease or condition was identified. A negative result means that no mutations were identified. An uncertain result means that the test did not provide a definitive answer and further testing may be necessary.

HOW CAN GENETIC TESTING BE BENEFICIAL TO YOU AND YOUR FAMILY MEMBERS?

Genetic testing can be beneficial to you and your family members in several ways, including:

EARLY DETECTION AND TREATMENT:

Genetic testing can help identify conditions at an early stage, allowing for earlier treatment and better outcomes.



FAMILYPLANNING:

Genetic testing can help individuals and couples make informed decisions about family planning, such as whether to have children or how to manage the risk of passing on a genetic condition.

RISK REDUCTION:

Genetic testing can help identify individuals at increased risk for certain diseases or conditions, allowing for early intervention and lifestyle changes to reduce the risk.

PERSONALIZED TREATMENT:

Genetic testing can provide information about how an individual may respond to certain medications or treatments, allowing for personalized treatment plans for better outcomes.

IF YOU ARE CONSIDERING GENETIC TESTING, IT IS IMPORTANT TO CHECK WITH YOUR HEALTH INSURANCE PROVIDER TO DETERMINE YOUR COVERAGE. YOU CAN DO THIS BY:

REVIEWING YOUR INSURANCE POLICY:

Check your insurance policy or contact your insurance provider to see if genetic testing is covered and under what circumstances.

CONSULTING WITH YOUR HEALTHCARE PROVIDER:

Talk to your healthcare provider about whether genetic testing is medically necessary and covered by your insurance.

SEEKING PRIOR AUTHORIZATION:

Some insurance plans may require prior authorization for genetic testing, which means your healthcare provider will need to submit a request to your insurance provider for approval.