



Pathway Diagnostic
Laboratories

Hereditary Thyroid Disorders Risk Panel



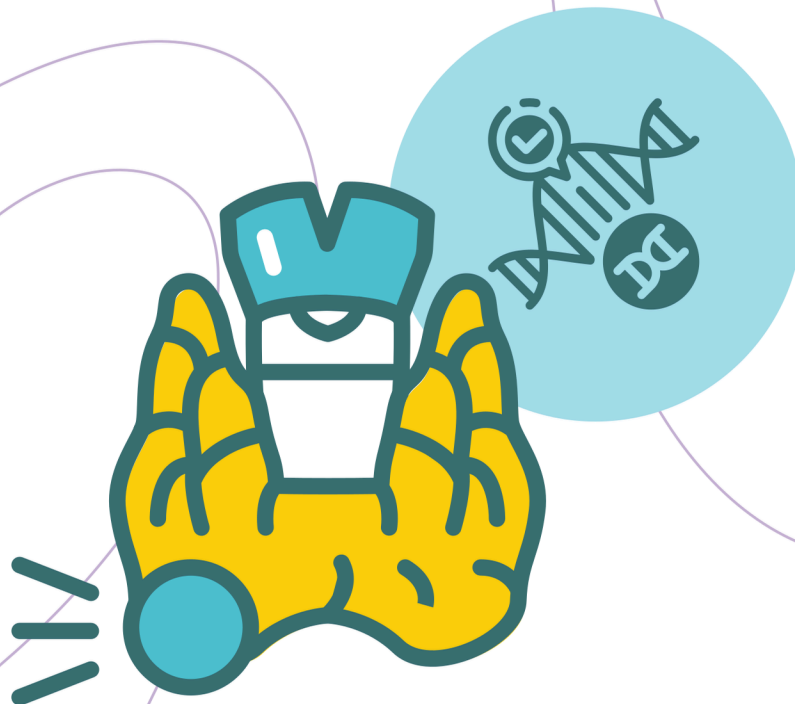
PATHWAY DIAGNOSTICS LABORATORIES

Hereditary Thyroid Disorders Risk Panel

Genetic testing for thyroid disease is a type of medical testing that looks for changes or mutations in a person's DNA that may indicate an increased risk of developing thyroid disorders. Thyroid disorders are conditions that affect the thyroid gland, which is located in the neck and produces hormones that regulate metabolism and other bodily functions.

Genetic testing for thyroid disease can help identify individuals who may be at risk for developing thyroid disorders, such as thyroid cancer, thyroid nodules, hyperthyroidism, or hypothyroidism. It can also help guide treatment decisions and identify family members who may be at risk for developing these conditions.

It is important to note that genetic testing for thyroid disease is not appropriate for everyone. It should only be conducted under the guidance of a healthcare provider with expertise in genetic testing and thyroid disease.



Genetic testing for thyroid conditions can provide several benefits for doctors, which can ultimately lead to better patient health outcomes. Here are some of the key benefits:

Early detection:

Genetic testing can identify individuals who are at increased risk for developing thyroid conditions before any symptoms appear. Early detection can enable doctors to initiate appropriate monitoring or treatment strategies before the disease progresses.

Personalized treatment:

Genetic testing can provide doctors with information about a patient's genetic profile that can help tailor treatment options to their specific needs. For example, some genetic mutations may indicate a higher likelihood of response to certain medications or therapies.

Improved accuracy:

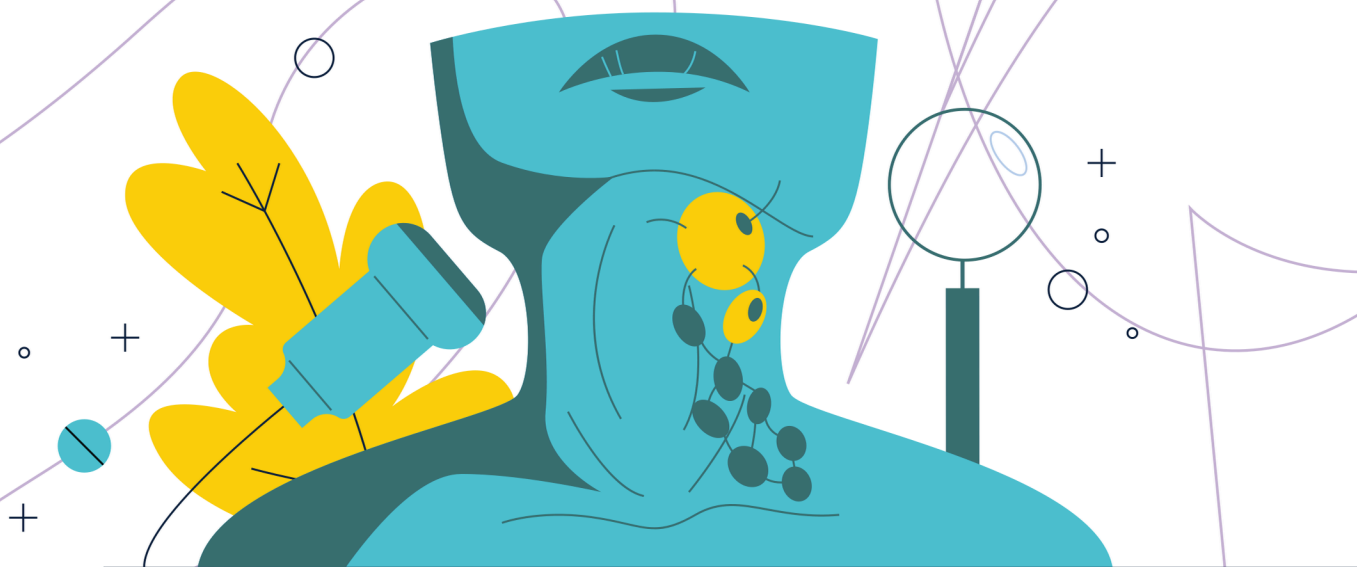
Genetic testing can help doctors distinguish between different types of thyroid conditions that may have similar symptoms but require different treatments. This can improve accuracy in diagnosis and avoid unnecessary treatment.

Family screening:

Genetic testing can identify family members who may also be at risk for developing thyroid conditions, which can lead to earlier detection and treatment.

To utilize genetic testing for better patient health outcomes, doctors may integrate the results of genetic testing with other clinical and laboratory tests to make a diagnosis and determine the best course of treatment.

They may also provide counseling and support to patients and their families before and after testing to address emotional and psychological concerns. Additionally, doctors may use genetic testing to inform screening and surveillance recommendations for patients and their family members who may be at increased risk for developing thyroid conditions. Overall, genetic testing can provide valuable insights into a patient's risk for thyroid conditions and can help doctors provide personalized and effective care.



Gene panel for **Hereditary Thyroid Disorders**

(47-Gene Panel)

PIK3CA, TRH, THRB, CTNNB1, KRAS, DUOX1, DUOX2, SLC5A5, CACNA1A, PRKCG, HAMP, SLC40A1, TPO, PAX8, GLIS3, FOXE1, SECISBP2, GNAQ, PLCG2, TGFBI, TG, THRA, TP53, TSHB, NRAS, ATP1A2, HRAS, TTR, IYD, HFE, ESR1, PLN, TFR2, SLC26A4, TSHR, NKX2-1, MECP2, IRAK1, G6PD, SLC16A2, IGSF1, TBLIX, IRS4, CST3, CST1, CSTB, RET

Mutations in thyroid-related genes can affect the function of the thyroid gland, leading to a variety of disorders. The thyroid gland is responsible for producing hormones that regulate metabolism, growth, and development in the body.

Here are different types of diseases associated with mutations in thyroid-related genes:

Congenital hypothyroidism:

Mutations in genes such as TSHR, PAX8, and TPO can lead to congenital hypothyroidism, which is a condition where the thyroid gland does not produce enough thyroid hormone.

Hyperthyroidism:

Mutations in the TSHR gene can lead to hyperthyroidism, which is a condition where the thyroid gland produces too much thyroid hormone.

Hypothyroidism:

Mutations in genes such as DUOX2 and SLC26A4 can lead to hypothyroidism, which is a condition where the thyroid gland does not produce enough thyroid hormone.

Pendred syndrome:

This is a rare genetic disorder that affects both the thyroid gland and the inner ear, causing hearing loss and an underactive thyroid gland. It is caused by mutations in the SLC26A4 gene.

Familial non-toxic goiter:

This is a condition in which the thyroid gland becomes enlarged, but there is no increase in thyroid hormone production. Mutations in genes such as TSHR and SLC5A5 have been linked to an increased risk of familial non-toxic goiter.

Thyroid hormone resistance:

This is a condition in which the body is resistant to the effects of thyroid hormones. Mutations in genes such as THRB have been associated with thyroid hormone resistance.

Overall, mutations in thyroid-related genes can have a significant impact on thyroid function and increase the risk of various thyroid-related diseases. Genetic testing may be recommended for individuals with a family history of thyroid disease or those with unexplained symptoms.



Test Specifications

Acceptable sample requirements

Buccal Swab or Saliva

Turnaround time - 2-3 weeks

Coverage $\geq 96\%$ at 20x

Reporting

Likely pathogenic and Pathogenic variants

Customization

Customizable Gene List



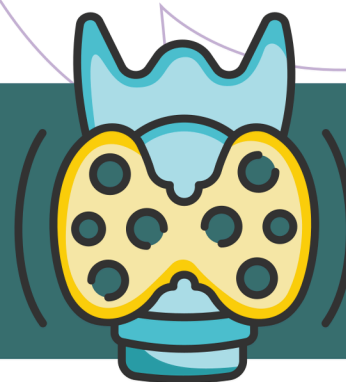
What To Do when thyroid genetic testing comes back positive: understanding the implications and taking action

Developing personalized treatment plans:

Positive test results can help doctors to develop personalized treatment plans for their patients. For example, if a patient is found to have a genetic mutation that increases their risk of developing thyroid cancer, their doctor may recommend more frequent monitoring or more aggressive treatment options.

Identifying family members at risk:

Positive test results can also help doctors to identify family members who may be at increased risk of developing thyroid disease. This information can be used to encourage these family members to undergo genetic testing and/or increased surveillance.



Improving patient education:

Positive test results can also improve patient education and understanding of their condition. By providing patients with information about their genetic risk factors, doctors can help patients to make informed decisions about their treatment options and lifestyle choices.

Offer genetic counseling:

The doctor may refer the patient and their family members to a genetic counselor to discuss the risk of passing on the disorder to future generations and options for family planning.

Overall, genetic testing for thyroid conditions can provide important benefits for doctors and their patients, leading to earlier detection, more personalized treatment plans, and better health outcomes.

UNLOCKING THE SECRETS OF YOUR GENETIC HEALTH

COMPREHENSIVE GENETIC TESTING AT

PATHWAY DIAGNOSTICS LABORATORIES

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

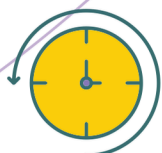


HIGH-QUALITY TESTING

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.



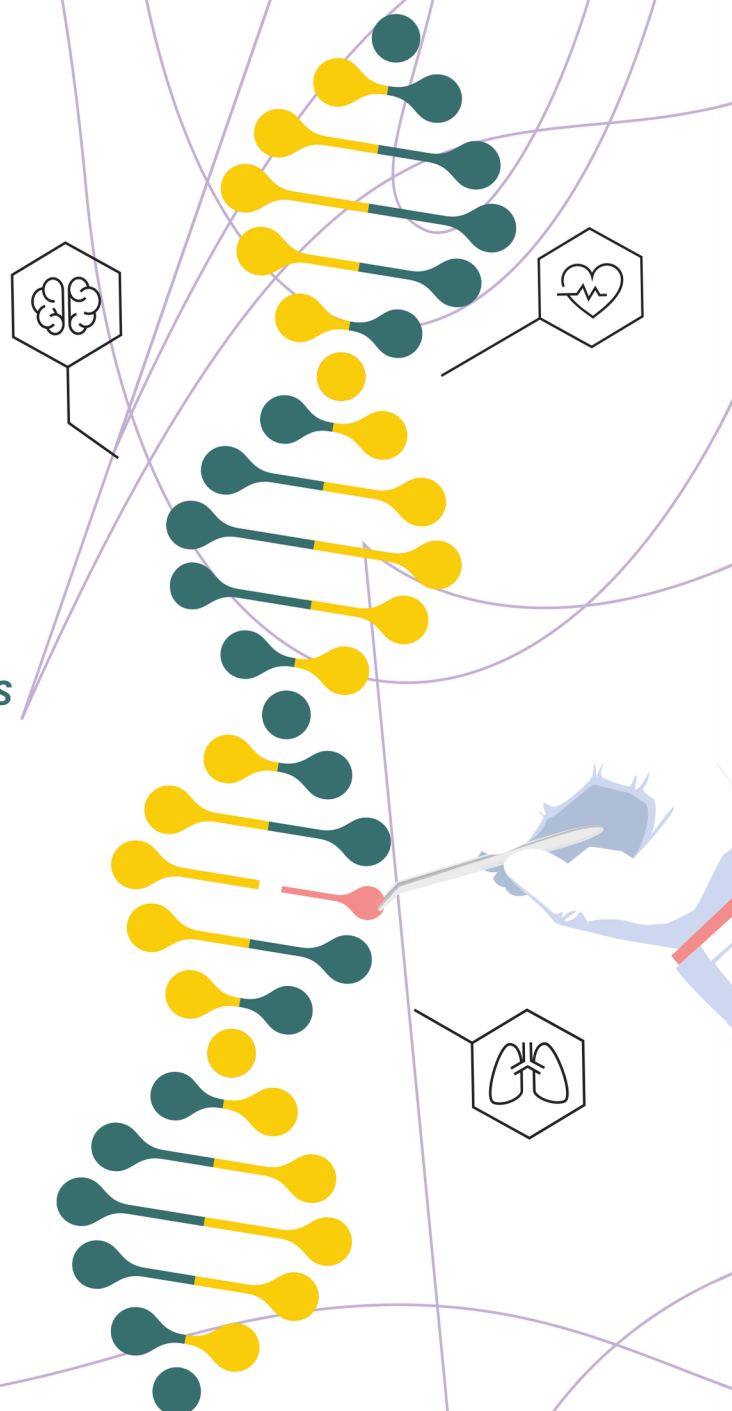
COMPREHENSIVE TESTING OPTIONS

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 be associated 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 the development, 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 better 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.

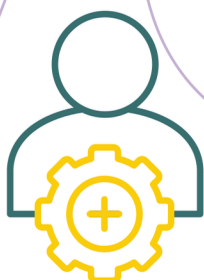


FAMILY PLANNING:

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.