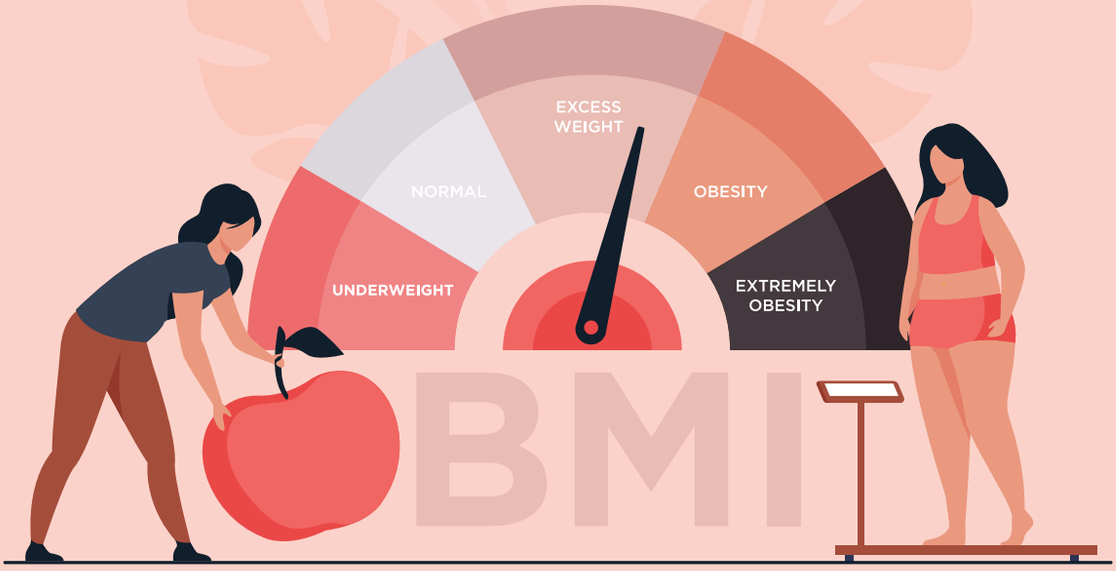
PREVENTIVE genomics



Hereditary
Metabolic Disorders Risk

PREVENTIVE GENOMICS

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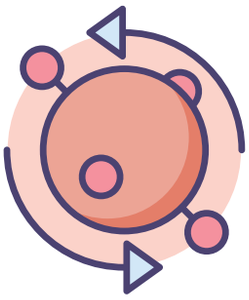
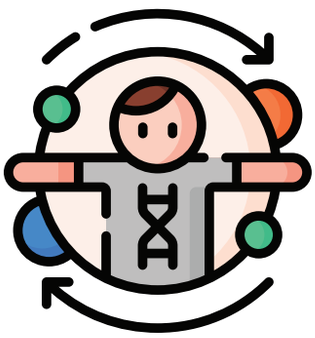
Personalized Health: Comprehensive Overview of Hereditary Metabolic Disorders Genetic Testing

Imagine a world where you can foresee and combat metabolic disorders before they take hold. With Hereditary Metabolic Disoders Genetic Testing, this vision becomes reality. This state-of-the-art diagnostic tool delves into your DNA, revealing the genetic bluprints that influence your metabolic health. Through sophisticated genetic sequencing, we decode your DNA to detect mutations that could lead to metabolic disorders.





The results illuminate your unique genetic landscape, identifying pathogenic or likely pathogenic variants that could impact your health. The magic of early detection lies in its power to transform lives. By identifying genetic risks before symptoms arise, healthcare providers can intervene with targeted strategies, preventing or delaying the onset of metabolic conditions. This proactive approach ensures that treatment is not just reactive, but preemptive.

Understanding Metabolic Disorders

Metabolic disorders encompass a wide range of conditions that affect the body's ability to process and convert nutrients into energy. These disorders can be inherited and often result from genetic mutations that affect enzyme function. Diagnosing metabolic disorders has historically relied on clinical observations and biochemical tests. While these methods have been valuable, they often fall short in uncovering the genetic components that play a significant role in many metabolic conditions.

Harnessing Genetic Insights:
Transformative Benefits for Metabolic Disorders

Genetic testing for Hereditary Metabolic Disorders can provide numerous benefits for physicians and patients, including:

Early Detection: Genetic testing allows for the early identification of metabolic disorders, even before symptoms manifest. This early detection empowers healthcare professionals to intervene proactively, potentially preventing or mitigating the effects of the disorder.

Accurate Diagnosis: Metabolic disorders can present with overlapping symptoms, making it challenging to achieve an accurate diagnosis based on clinical presentation alone. Genetic testing helps differentiate between similar conditions, enabling tailored treatment plans and improved patient outcomes.

Tailored Treatment Strategies: Genetic testing provides critical information about the specific genetic mutations associated with an individual's metabolic disorder. Armed with this knowledge, healthcare professionals canpersonalize treatment plans, selecting interventions that target the underlying causes rather than merely managing symptoms.

Familial Screening: Many metabolic disorders have a hereditary component, meaning they can be passed down through generations. Genetic testing enables the identification of at-risk family members, facilitating early screening and preventive measures to protect their health.

Diseases and conditions related to Hereditary Metabolic Disorders genes:

Here are a few diseases and conditions related to hereditary metabolic disorders associated with some of the specified genes:

Familial Hypercholesterolemia (Related Gene: ABCA1) - A genetic disorder characterized by high cholesterol levels, specifically low-density lipoprotein (LDL) cholesterol, leading to an increased risk of cardiovascular disease.

Wilson Disease (Related Gene: ATP7B) - An inherited disorder that causes excessive accumulation of copper in the liver, brain, and other vital organs, leading to liver disease and neurological symptoms.

Cystic Fibrosis (Related Gene: CFTR) - A genetic disorder affecting the lungs and digestive system, causing severe respiratory and gastrointestinal complications due to thick mucus production.

Medium-Chain Acyl-CoA Dehydrogenase Deficiency (Related Gene: ACADM) - A metabolic disorder that impairs the body's ability to convert certain fats to energy, particularly during fasting, leading to hypoglycemia and other complications.

Phenylketonuria (Related Gene: PAH) - An inherited metabolic disorder characterized by an inability to break down the amino acid phenylalanine, leading to intellectual disability and other health issues if untreated.

Maple Syrup Urine Disease (Related Gene: BCKDHA, BCKDHB) - A metabolic disorder characterized by the inability to break down certain amino acids, leading to the accumulation of toxic substances in the blood and urine with a distinctive sweet odor.

Gaucher Disease (Related Gene: GBA) - A genetic disorder where fatty substances accumulate in cells and certain organs, causing enlargement of the liver and spleen, bone pain, and other symptoms.

Galactosemia (Related Gene: GALT) - An inherited metabolic disorder that affects the body's ability to process the sugar galactose properly, leading to serious complications including liver damage, intellectual disability, and cataracts if untreated.

Gene panel for Hereditary Metabolic Disorders

(199 - GENES PANEL)

ABCA1, ABCB4, ABCC2, ABCD1, ABCD3, ABCD4, ABCG5, ABCG8, ACACA, ACADM, ACADS, ACADVL, APOE, ARSA, ASPA, ASS1, ATP7B, BCKDHA, BCKDHB, BSCL2, BTD, CACNA1A, CBS, CFTR, CLCNKB, COX10, COX15, CPOX, CPT1A, CSTB, DGUOK, DHCR7, DLD, DPYD, F9, FAH, FH, G6PC, G6PD, GAA, GALNS, GALT, GALC, GAMT, GATM, GBA, GBE1, GLUD1, GCDH, GCK, GLA, GNE, GYS1, GYS2, HADH, HADHA, HADHB, HEXA, HFE, HMGCL, HPRT1, HSD17B10, HYAL1, IDH2, IDS, LCT, LIPA, LPL, MAN2B1, ISCU, MCCC1, MCCC2, MCOLN1, MECP2, MMAB, MMACHC, MMAA, MMADHC, MTHFR, MUT, NAGA, NAGLU, OTC, PAH, NHEG1, PCCA, PC, PCCB, PCK1, PCK2, PDHA1, PDHX, PEPD, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHKA1, PHKA2, PHKB, PHKG1, PHKG2, PHYH, POLG, PPARG, PRKAG2, PRPS1, PTS, PYGL, PYGM, QDPR, RBCK1, RFT1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SEC23B, SERPINA1, SGSH, SLC12A3, SLC16A1, SLC17A5, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A26, SLC25A4, SLC2A1, SLC2A2, SLC30A10, SLC35A1, SLC35A2, SLC35C1, SLC39A4, SLC3A1, SLC40A1, SLC41A2, SLC41A3, SLC46A1, SLC5A1, SLC6A19, SLC6A8, SLC6A9, SLC7A7, SLCO1B1, SMPD1, SSR4, STT3A, STT3B, SUCLA2, SUCLG1, SUOX, SURF1, TALDO1, TAT, TBC1D4, TCN2, TFR2, TIMM8A, TMEM126A, TMEM165, TNPO3, TMEM70, TPMT, TPP1, TREX1, TRIM32, TRIM37, TRMU, TRPM6, TRPM7, TSFM, TTC19, TUFM, TUSC3, TYMP, UGT1A1, UMPS, UCP2, UPB1, UQCRB, UQCRQ, UROD, UROS, WFS1, XDH, YARS2, ZMPSTE24

TEST SPECIFICATIONS

Acceptable sample requirements
Buccal Swab or Saliva

Turnaround time - 7-10 Business days
Coverage _>96% at 20x

Reporting
Likely pathogenic and Pathogenic variants

Customization
Customizable Gene List

What To Do When Hereditary Neurological Risks Genetic Testing Comes Back Positive: Understanding The Implications And Taking Action

If genetic testing for hereditary metabolic disorders, yields a positive result, it is important to proceed as follows:

Inform the Patient and Family: The doctor will inform the patient and their family members of the diagnosis and the implications of the genetic test results. They will also discuss the possibility of genetic counseling.

Develop a Treatment Plan: The doctor will develop a treatment plan tailored to the specific disorder and the patient's individual needs. This may include medications, dietary modifications, and lifestyle changes.

Monitor the Patient: The doctor will monitor the patient's condition over time to ensure that the treatment plan is effective and to adjust it as necessary.

Offer Genetic Counseling: If a genetic mutation is identified, doctors may confirm the diagnosis through further testing or clinical evaluation to determine the extent of the disease or condition.

UNLOCKING THE SECRETS OF YOUR GENENTIC HEALTH
COMPREHENSIVE GENETIC TESTING AT

PREVENTIVE GENOMICS

PREVENTIVE GENOMICS MAY BE A GOOD CHOICE FOR GENETIC TESTING FOR SEVERAL REASONS:

HIGH-QUALITY TESTING

Preventive Genomics uses advanced technology and have experienced technicians to ensure that the testing is performed to the highest standards

FAST TURNAROUND TIME

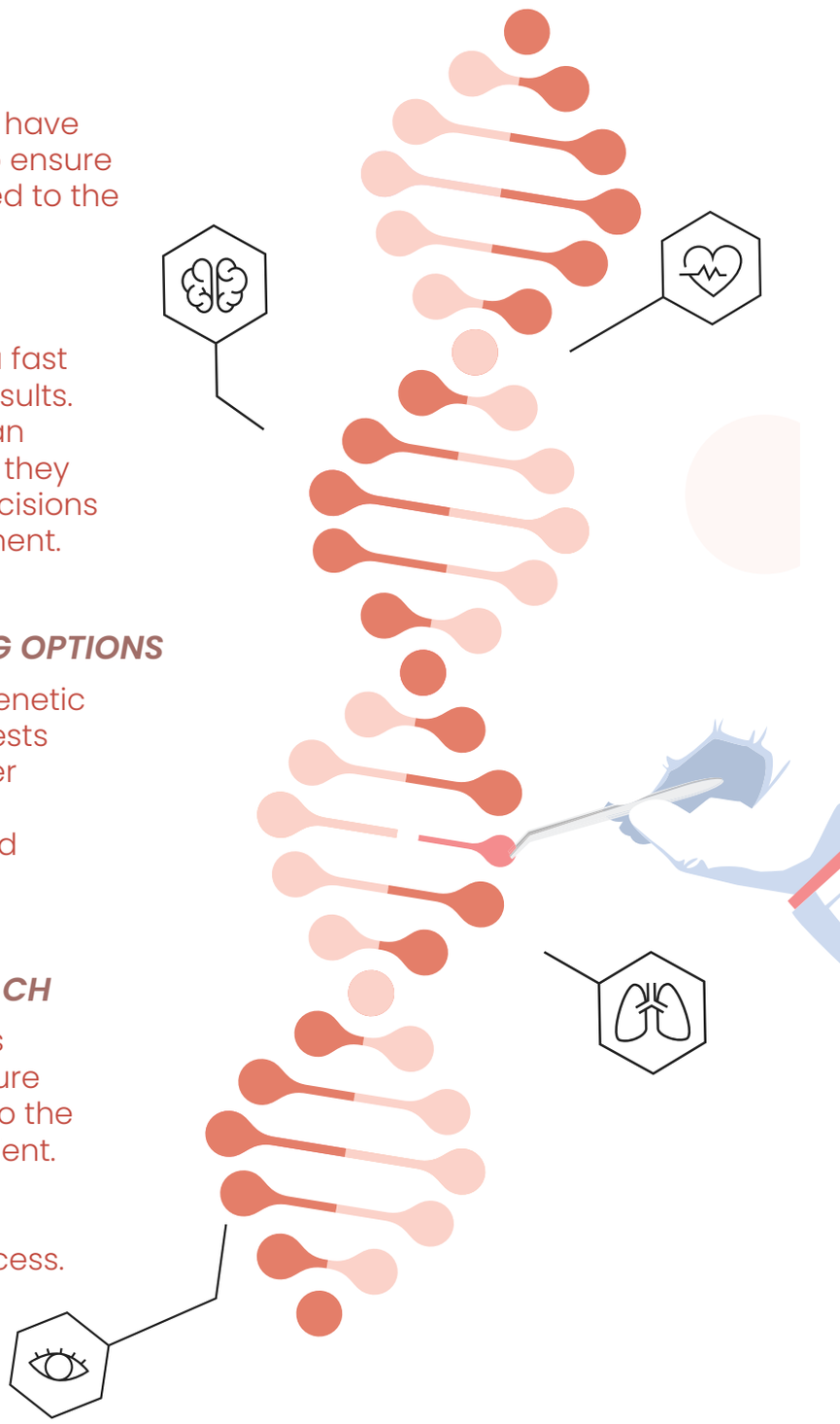
Preventive Genomics has a fast turn around time for test results. This means that doctors can quickly get the information they need to make informed decisions about their patients' treatment.

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

Preventive Genomics 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?**

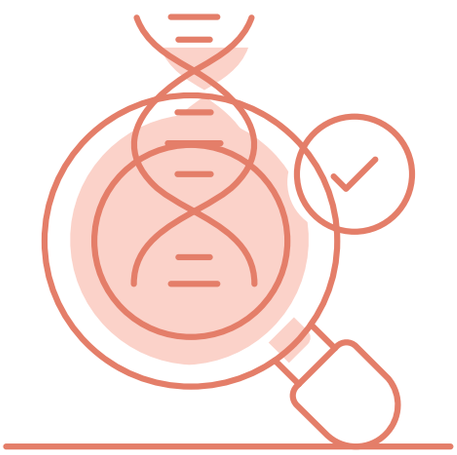
DNA is the genetic material that contains the instructions for thedevel-opment, growth, and function of all living organisms. Genetic testing can provide information about inherited diseases or conditions, predispositions to certain diseases, and the likelihood of passing these conditions onto your children.

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

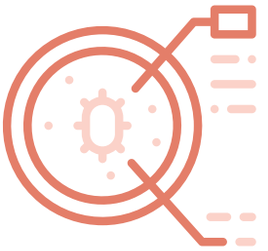
The results of genetic testing can be positive, negative, or uncertain. A positive result indicates that a genetic mutation associated with a specific disease or condition was identified. A negative result means that no mutations were identified. An uncertain result means that the test did not provide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

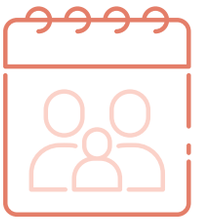
HOW CAN GENETIC TESTING BE BENEFICIAL TO **YOU AND YOUR FAMILY MEMBERS?**

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.



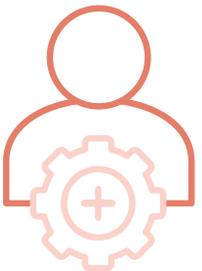


FAMILY PLANNING:

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.

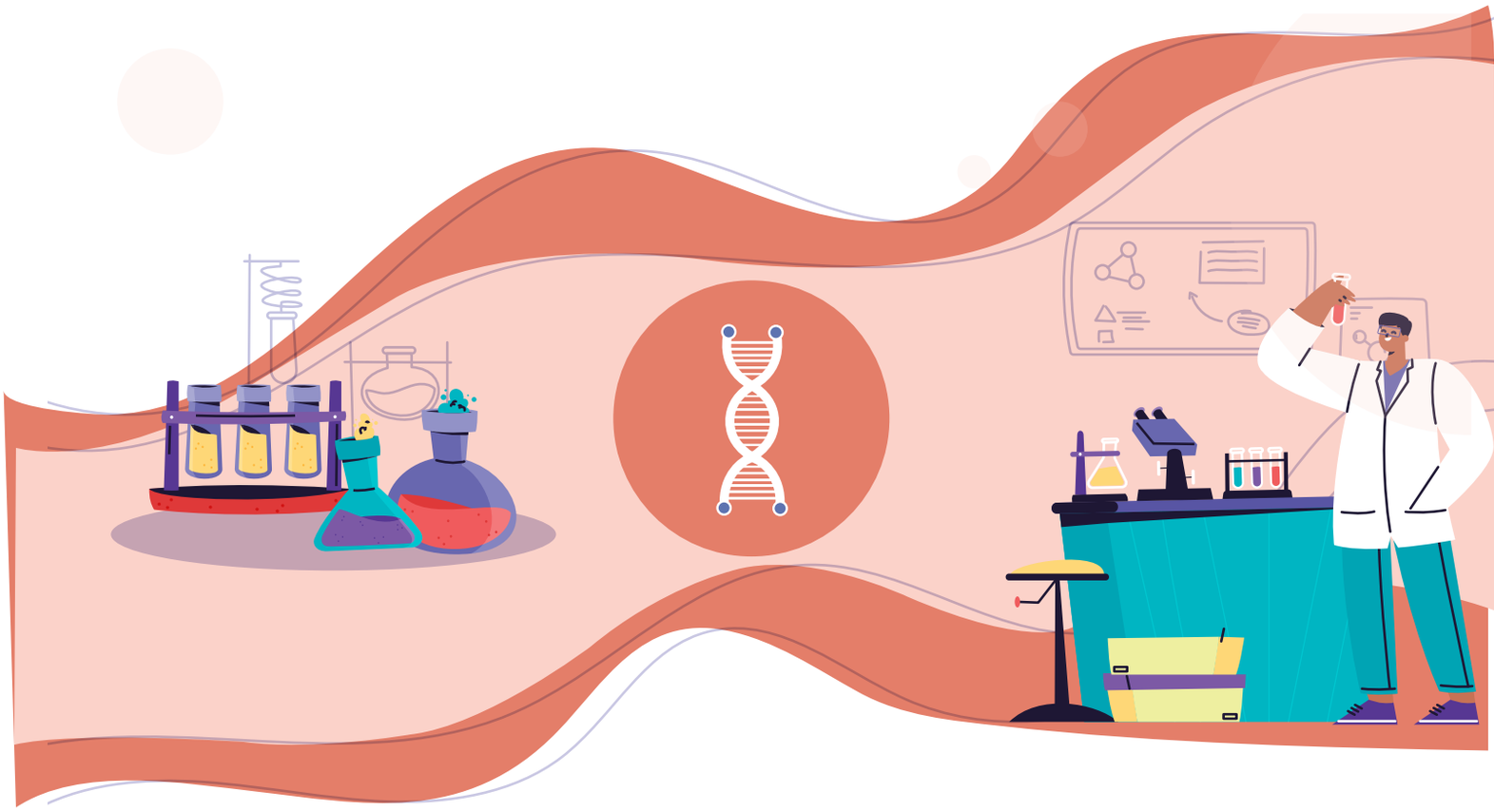
RISK REDUCTION:

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.



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.

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