



**PREVENTIVE**  
genomics

# Genetic Testing for **HEREDITARY EYE DISORDERS**



## **PREVENTIVE GENOMICS**



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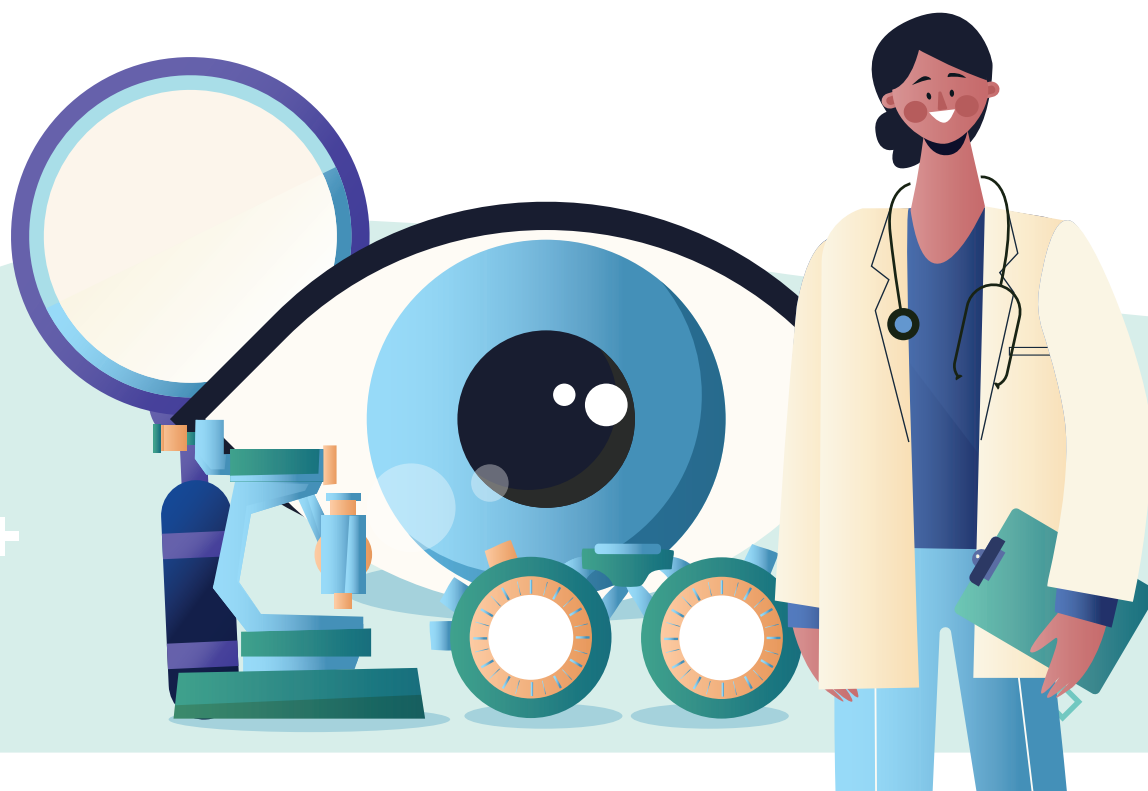
## REVOLUTIONIZING VISION CARE:

### THE POWER OF GENETIC TESTING IN UNDERSTANDING AND TREATING EYE DISORDERS

In the realm of healthcare, advancements in genetic testing have revolutionized the way we diagnose and treat various medical conditions. When it comes to eye disorders, genetic testing has emerged as a powerful tool, offering new insights into the underlying causes and paving the way for personalized treatment strategies. This article explores the significance of genetic testing in the context of eye disorders and its potential to transform the field of vision care.

### UNDERSTANDING EYE DISORDERS:

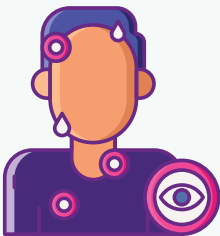
Eye disorders encompass a wide range of conditions, including inherited diseases, age-related degeneration, refractive errors, and more. Historically, diagnosing eye disorders relied on clinical observations and physical examinations. While these methods have been valuable, they often fall short in uncovering the genetic components that play a significant role in many ocular conditions.



# GENETIC TESTING FOR HEREDITARY EYE DISORDERS

CAN PROVIDE SEVERAL BENEFITS FOR PHYSICIANS AND PATIENTS, INCLUDING:

**Early Detection:** Genetic testing allows for the early identification of eye disorders, even before symptoms manifest. This early detection empowers healthcare professionals to intervene proactively, potentially preventing or delaying the onset of vision loss.



**Accurate Diagnosis:** Eye disorders can exhibit overlapping symptoms, making it challenging to arrive at 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 eye disorder. Armed with this knowledge, healthcare professionals can personalize treatment plans, selecting interventions that target the underlying causes rather than merely managing symptoms.



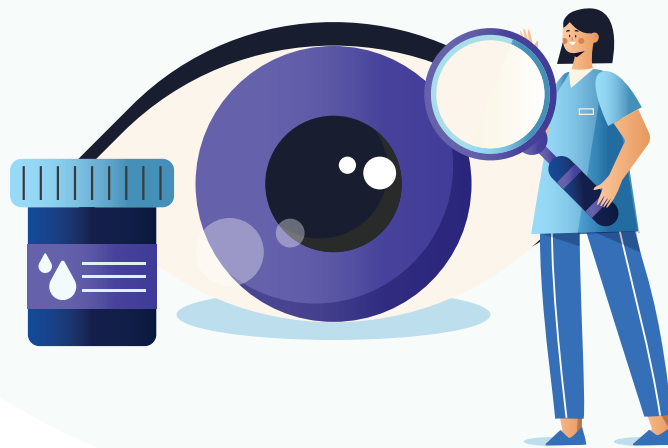
**Familial Screening:** Many eye disorders have a hereditary component, meaning they can be passed down through generations. Genetic testing enables the identification of at-risk family members, facilitating early screening and preventive measures to protect their vision.

**Research and Development:** Genetic testing plays a vital role in advancing our understanding of eye disorders. By collecting genetic data from individuals with various eye conditions, scientists can uncover new genetic markers, paving the way for the development of novel treatments and interventions.



## ETHICAL CONSIDERATIONS

While the benefits of genetic testing for eye disorders are undeniable, it is crucial to address the ethical implications associated with this practice. Protecting patient privacy, ensuring informed consent, and providing counseling services to help individuals understand and cope with their results are essential aspects of responsible genetic testing.



## DISEASES AND CONDITIONS RELATED TO HEREDITARY EYE DISORDERS GENES:

**Retinitis Pigmentosa (RP):** RP is a group of inherited retinal disorders characterized by progressive degeneration of the retina. Genetic mutations in several genes, including RPGR, RHO and PRPF31, among others, have been associated with different forms of RP.

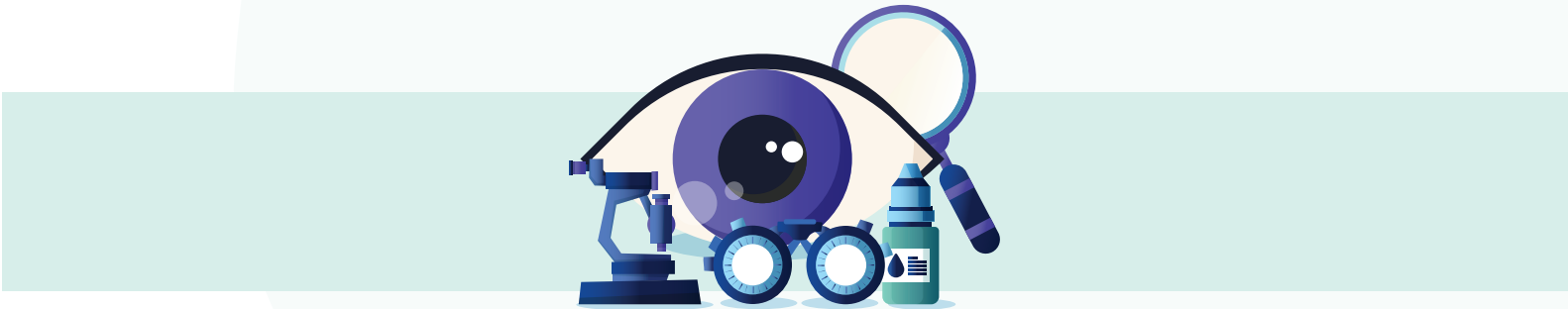
**Leber Congenital Amaurosis (LCA):** LCA is a rare inherited disorder that affects the retina, leading to severe visual impairment or blindness in infancy. Mutations in genes such as RPE65 have been implicated in LCA.

**Age-Related Macular Degeneration (AMD):** AMD is a common cause of vision loss in older adults. While age and environmental factors contribute to AMD, genetic variations in genes like CFH have been identified as significant risk factors for the disease.

**Glaucoma:** Glaucoma encompasses a group of eye conditions that damage the optic nerve, leading to progressive vision loss. Mutations in genes such as MYOC and CYP1B1 have been linked to specific forms of glaucoma.

**Congenital Cataracts:** Congenital cataracts refer to the presence of clouding in the lens of the eye at birth or early infancy. Several genetic mutations, including those in genes like CRYAA and HSF4 have been associated with different types of congenital cataracts.

**Usher Syndrome:** Usher syndrome is a genetic disorder characterized by both hearing loss and visual impairment. Multiple genes, including MYO7A, USH2A, and CDH23, have been implicated in different subtypes of Usher syndrome.



GENE PANEL FOR **HEREDITARY EYE DISORDERS RISK**  
(86-GENE PANEL)

FOXC1, ALDH7A1, ATXN7, CACNA1A, CDH23, CDKL5, CFH, CHD2, CLRN, CNGA1, CTSD, EYS, FTL, GABRG2, GJB2, GJB6, GPR98, GRIN2A, KCNQ2, MECP2, MTRNR1, MYO15A, MYO7A, MYOC, OTOF, PAX2, PCDH15, PCDH19, PDE6A, PDE6B, POLG, PRPF31, PRRT2, RDH12, RP2, RPGR, SCN1A, SCN1B, SCN2A, SCN8A, SLC26A4, SLC2A1, SLC9A6, STXBP1, SYNGAP1, TCF4, TGFBI, TMC1, TMPRSS3, TPP1, TSC1, TSC2, USH1C, USH1G, USH2A, WFS1, ZEB2, HSF4, BFSP2, GALK1, BFSP1, CRYAA, CRYAB, CRYGC, FOXE3, BEST1, NR2E3, NRL, RHO, RP1, RPE65, CAV1, CAV2, SIX1, SIX6, CDKN2B-AS, TMCO1, CYP1B1, LTBP2, PITX2, PAX6, OPN1LW, OPN1MW, ABC4, CRB1, PRPH2

TEST SPECIFICATIONS

**Acceptable sample requirements**  
Buccal Swab or Saliva

**Reporting**  
Likely pathogenic and Pathogenic variants

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

**Customization**  
Customizable Gene List



## WHAT TO DO WHEN HEREDITARY EYE DISORDERS GENETIC TESTING COMES BACK POSITIVE : UNDERSTANDING THE IMPLICATIONS AND TAKING ACTION

IF A DOCTOR RECEIVES A POSITIVE REPORT FOR THE GENETIC TESTING OF HEREDITARY EYE DISORDERS, THERE ARE SEVERAL NEXT STEPS THEY MAY TAKE:

**Confirming the diagnosis:** 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.

**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, therapies, 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:** 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.

# UNLOCKING THE SECRETS OF YOUR GENETIC HEALTH

## COMPREHENSIVE GENETIC TESTING AT

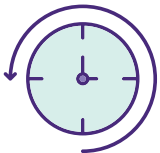
### PREVENTIVE GENOMICS

**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.



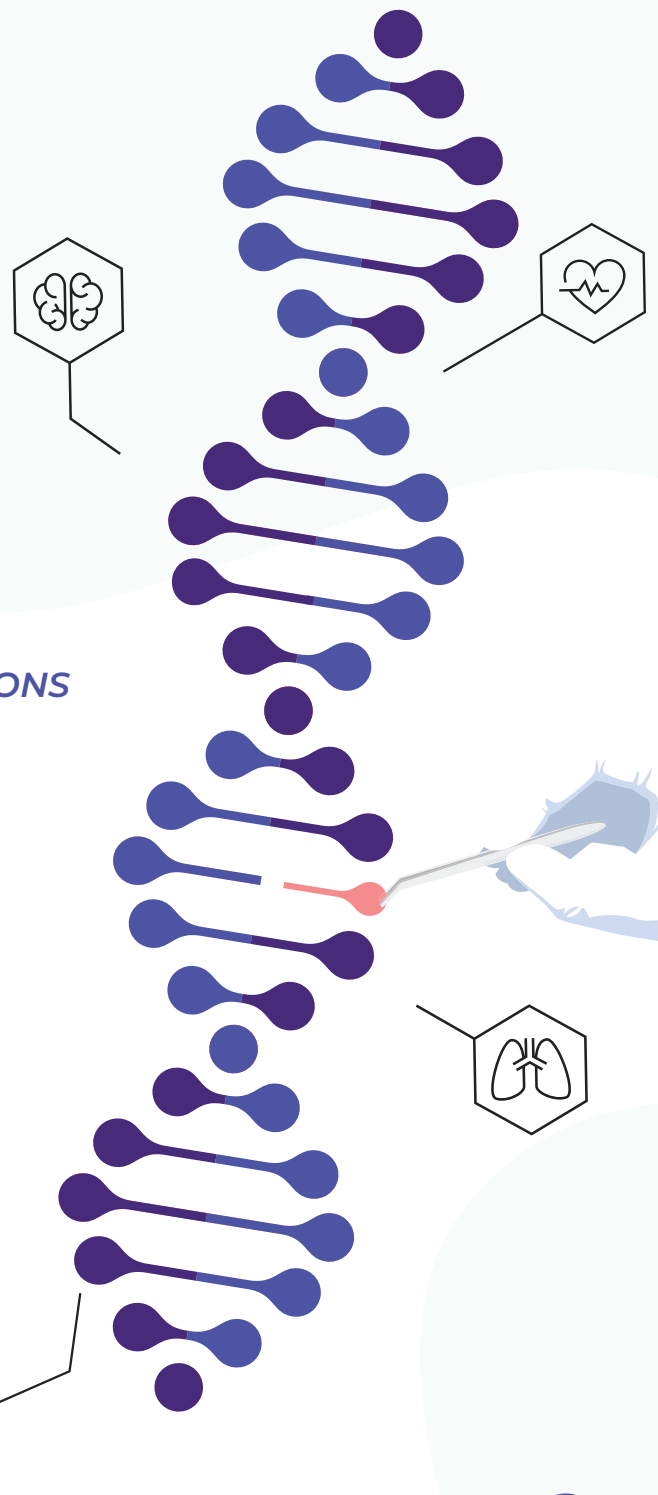
#### **COMPREHENSIVE TESTING OPTIONS**

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 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.

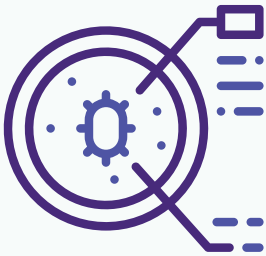


## HOW CAN GENETIC TESTING BE BENEFICIAL TO YOU AND YOUR FAMILY MEMBERS?

*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.

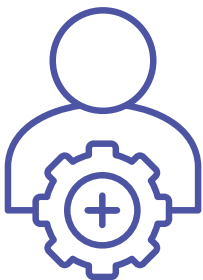


### FAMILY PLANNING:

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.

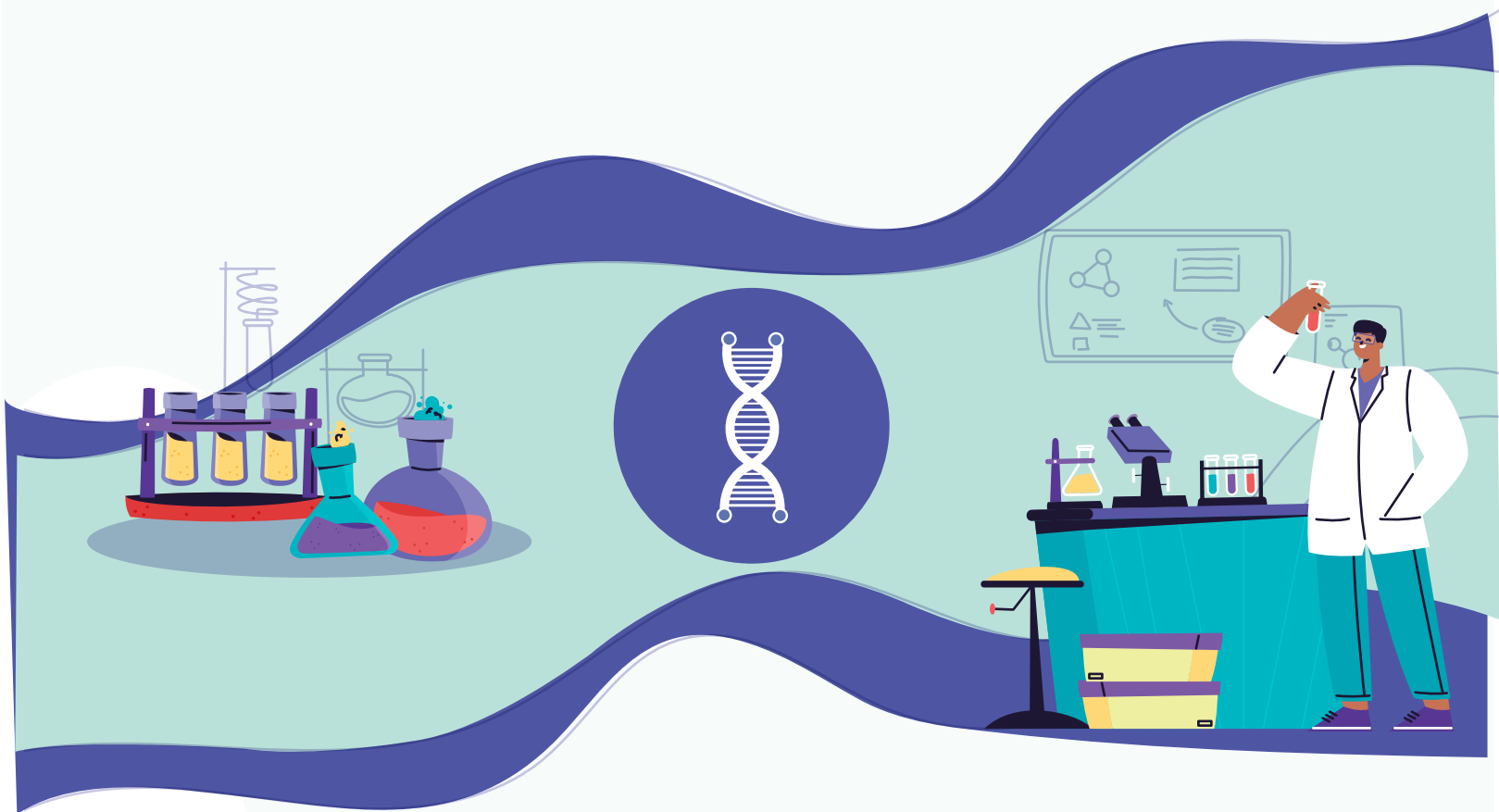
### RISK REDUCTION:

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.



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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