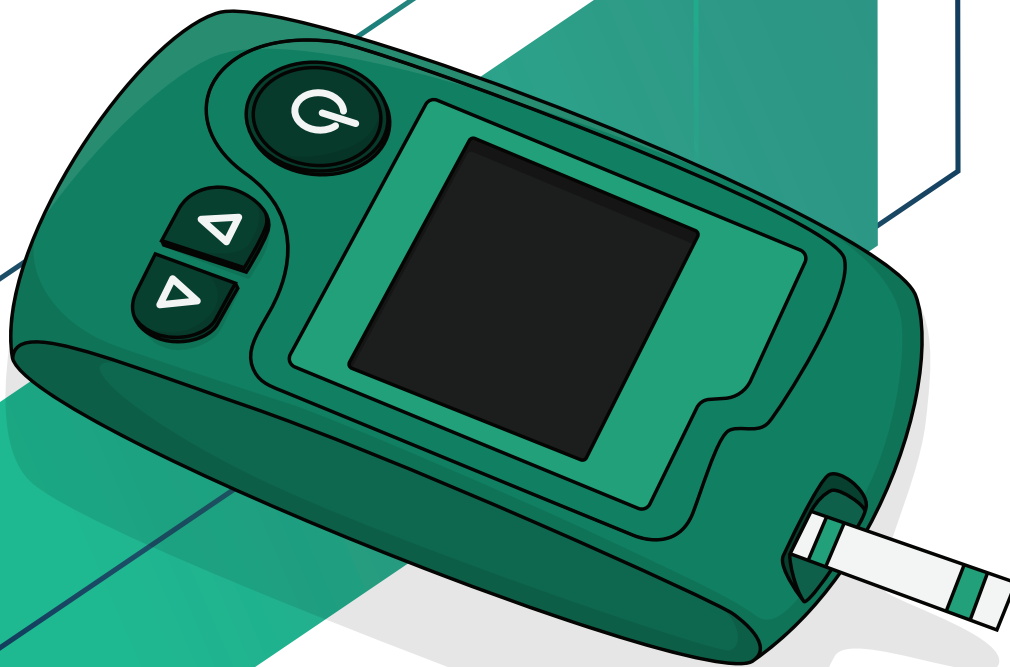




PREVENTIVE  
genomics



# ***Diabetes (MODY) Testing***

**PREVENTIVE GENOMICS**

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# MODY (MATURITY-ONSET DIABETES OF THE YOUNG) TESTING

Maturity-onset diabetes of the young (MODY) is a type of monogenic diabetes characterized by non-insulin-dependent diabetes and early onset (usually before age35). Diabetes affects 29.1 million people in the United States, or 9.3% of the population. The most common types of diabetes are type 1 and type 2. The genetic basis of these types of diabetes is largely unknown. The disease is thought to be the result of a combination of multiple genetic and environmental risk factors. Monogenic forms of diabetes are rare, accounting for approximately 2% of all diabetes cases.

Genetic testing (NGS) can establish an accurate diagnosis and identify a genetic etiology, which has important implications for individualized management of symptoms and prognostic information for family members.

## SYMPTOMS AND CAUSE:

Diabetes is a disorder that results in elevated blood glucose. Over time, the disorder can cause various health problems, including diseases of the heart, kidneys, eyes, and nervous system. Monogenic forms of diabetes are caused by a mutation in a single gene. There are 14 known MODY genes, and three account for the majority of cases:

### MODY 3:

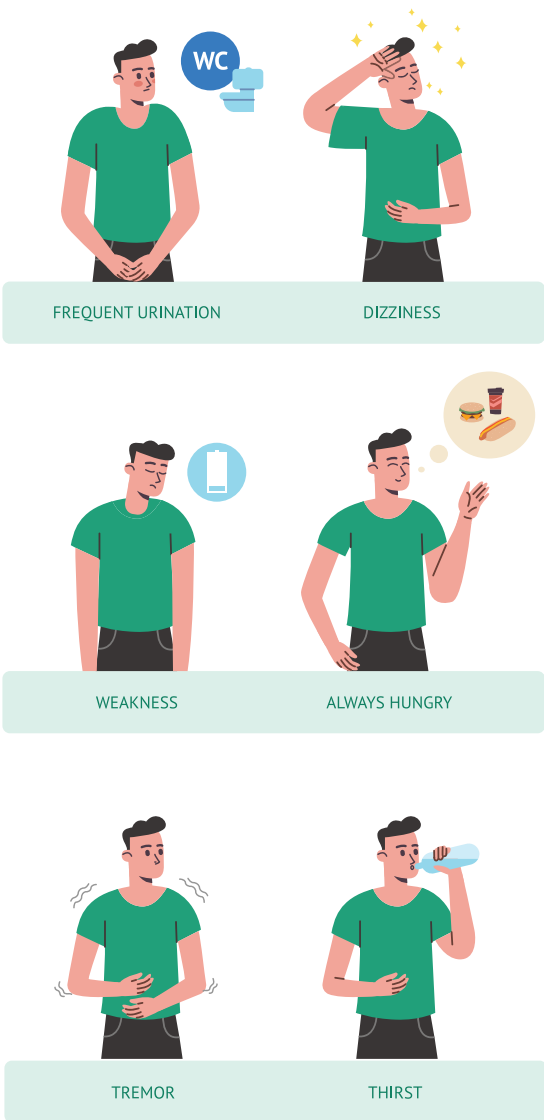
Mutations in the hepatocyte nuclear factor-1 alpha (HNF1A) gene are the most common cause of MODY, accounting for about half of cases. This type is characterized by a progressive insulin secretory defect due to beta-cell failure.

### MODY 2:

Mutations in the glucokinase gene (GCK) are the next most common cause of MODY, accounting for about 20-25% of cases. GCK encodes the glucokinase enzyme, which acts as the pancreatic glucose sensor. Mutations result in lifelong, stable, mild fasting hyperglycemia. HbA1C values are usually just above the high normal range. People with GCK mutations rarely require treatment. This type of MODY may be detected during pregnancy, when glucose tolerance testing is routinely performed.

### MODY 1:

Mutations in the hepatocyte nuclear factor-4 alpha (HNF4A) gene cause a clinical presentation similar to HNF1A. However, mutations in this gene are much less common (less than 10% of MODY). Age of onset may be later, and there is not a low renal threshold. HNF4A mutations can also cause high birth weight in newborns and transient neonatal hypoglycemia. These patients are also more sensitive to sulfonylurea treatment.routinely performed.



## WHY DOES MODY RUN IN FAMILIES?

MODY is typically inherited in an autosomal dominant manner. When a parent has a MODY mutation, each of her/his offspring have a 50% risk of inheriting the mutation. Mutations that occur de novo in an affected individual, reduced penetrance, and variable expressivity have been reported. Thus, the absence of a family history does not, by itself, rule out a diagnosis of MODY.

## WHY IS IT IMPORTANT TO RECOGNISE IT?

There are different types of MODY. By finding out which type of MODY a person has the most appropriate treatment for them can be determined. Knowing the type of MODY a person has also means we can advise them about how their diabetes will progress in the future. As it runs in families, it is important to advise other family members of their risk of inheriting it.



Are overweight or obese  
(BMI of 23.0 kg/m<sup>2</sup> or higher)



Lead an inactive lifestyle



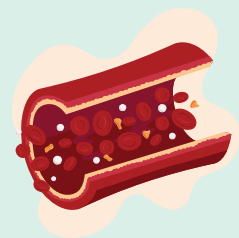
Are 40 years old  
and above



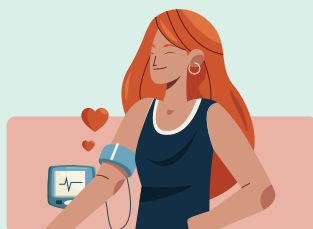
Have a parent or  
sibling with diabetes



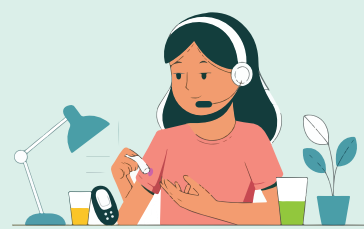
Have a history of  
gestational diabetes



Have abnormal  
blood cholesterol  
or lipid levels



Have high  
blood pressure



Have impaired glucose  
tolerance or impaired  
fasting glucose

## WHY SHOULD A PHYSICIAN ORDER THIS TEST?

Diabetes syndrome is a Multi chromosomal or multi-gene mutation disposed metabolic disease. Most of the genes are inherited in autosomal dominant fashion. Germline Variations in the Single nucleotide variants on these genes are shown to cause diabetes syndrome. The classical example is the Mature Onset of Diabetes in Young.

Preventive Genomics Services Diabetes Syndrome NGS panel is meticulously designed to capture the germ-line variations in the genes responsible for all types of Diabetes giving the physician a complete picture of future disposition to it. This will help in managing the lifestyle with Reuter check-up to the doctor and have a blissful and stress-free life.



## Diabetes Mody PANEL GENE LIST ( 30 - Gene Panel ) :

ABCC8, BLK, CAPN10, EIF2AK3, FOXP3, GATA6, GCK, GLIS3, GLUD1, HADH, HNF1A, HNF1B, HNF4A, INS, INSR, KCNJ11, KLF11, NEUROD1, NEUROG3, PAX4, PDX1, PPARG, PTF1A, RFX6, SLC16A1, SLC2A2, TCF7L2, UCP2, WFS1, ZFP57

**Specimen Requirements:**  
Buccal swab (Wet/Dry) or  
Extracted DNA (5ug)

**Turnaround Time:**  
7-10 Business days

# UNLOCKING THE SECRETS OF YOUR GENETIC HEALTH

## COMPREHENSIVE GENETIC TESTING AT

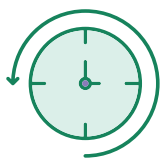
### PREVENTIVE GENOMICS

#### 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.



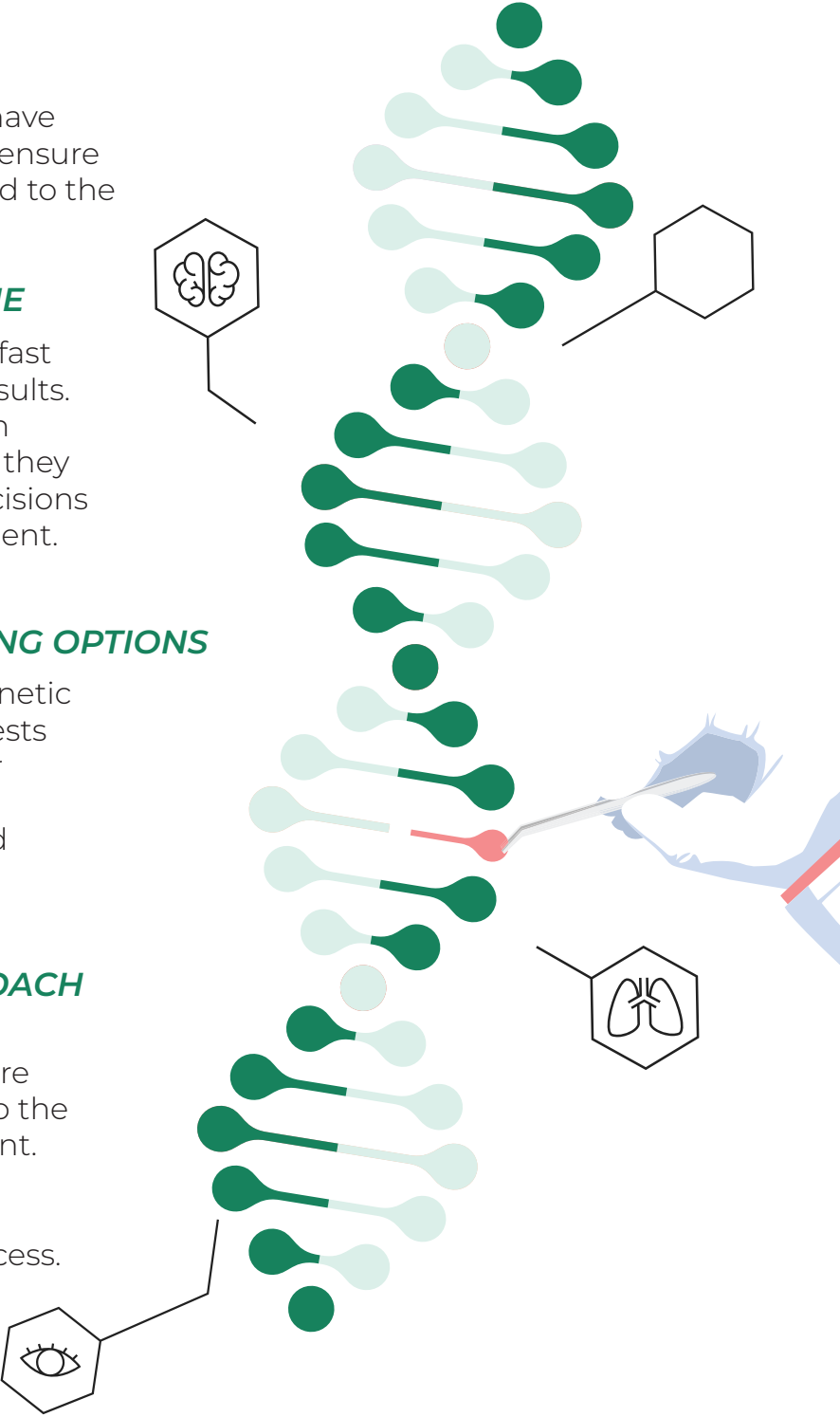
##### COMPREHENSIVE TESTING OPTIONS

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.

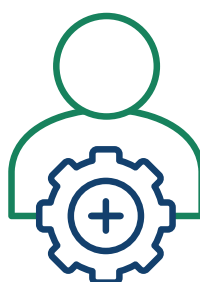


### **FAMILY PLANNING:**

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.

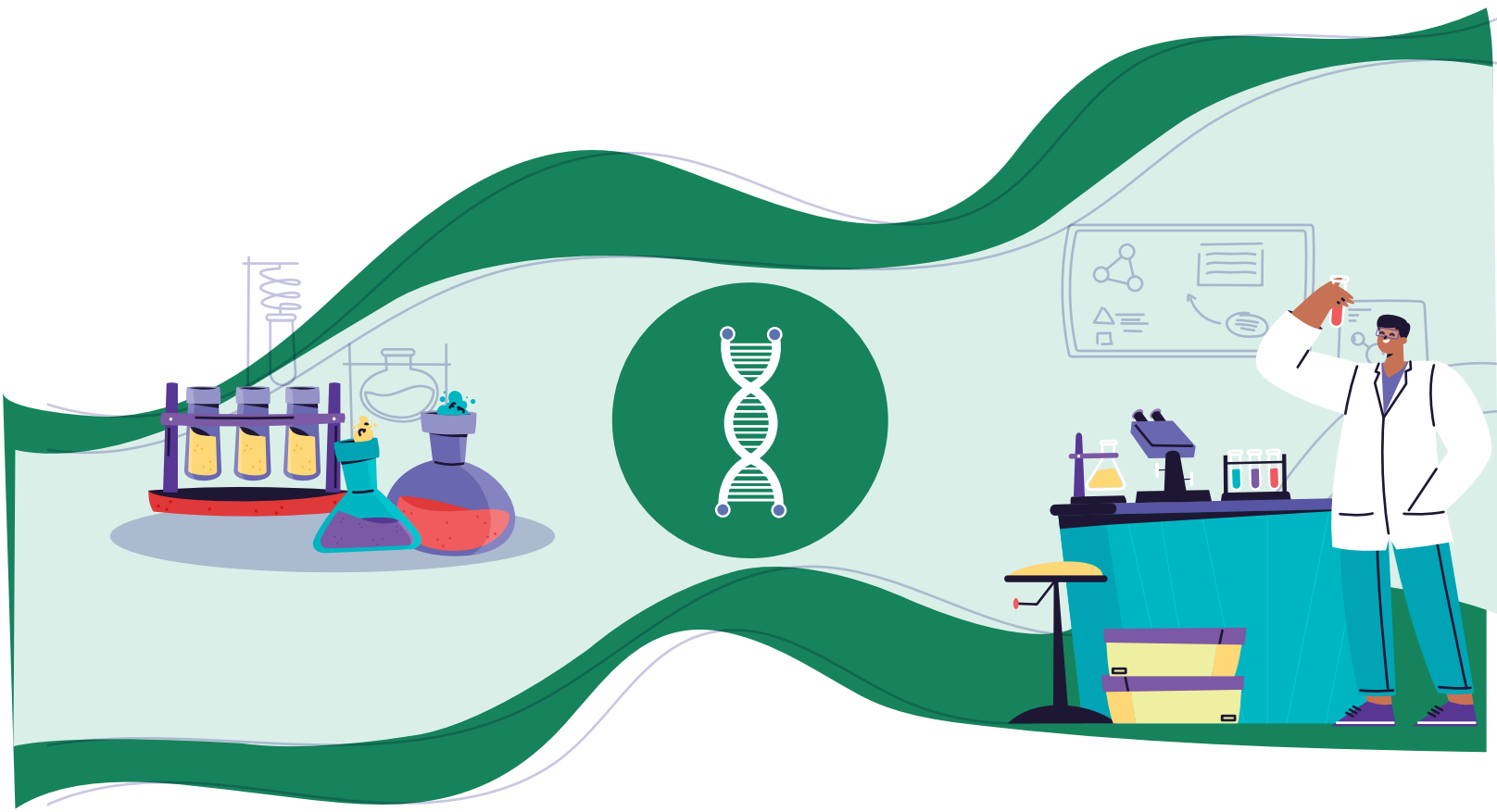
### **RISK REDUCTION:**

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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