

# DIABETES (MODY) TESTING

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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 age 35). 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.



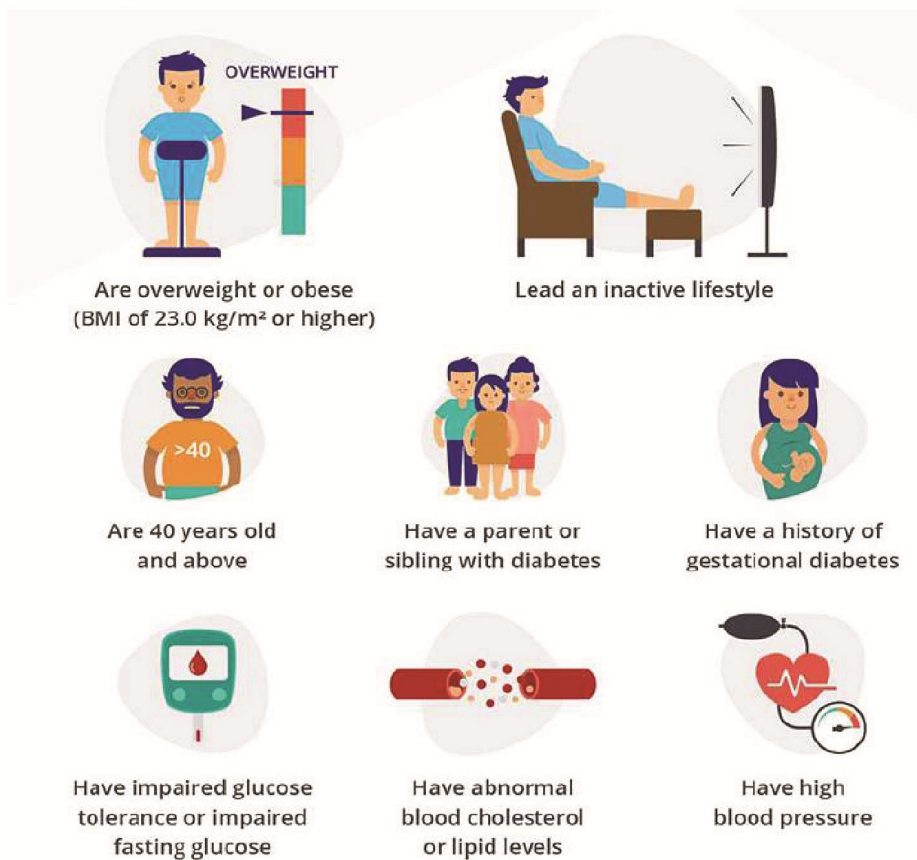
## WHY DOES MODY RUN IN FAMILIES?

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.

## RISK FACTORS



## WHY SHOULD A PHYSICIAN ORDER THIS TEST?

Diabetes syndrome is a Multi chromosomal or multi-gene mutation disposed of metabolic disease. Most of the genes are inherited in an 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.

Redwood Lab 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-ups to the doctor and have a blissful and stress-free life.

## PANEL GENE LIST:

ABCCB, 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:**  
3-4 weeks

## TEST INFORMATION

Description	Hereditary Diabetes Disease Panel
Method	Next Generation Sequencing
Specimen	Buccal swab shipped at room temperature
Requirements	temperature
Turnaround Time	3 to 4 Weeks
Shipping	Pickup/FedEx Service Available Monday - Friday
Testing Performed	Monday - Saturday

Medex Laboratories is a full service, national diagnostic testing laboratory headquartered in Houston, Texas with concentrations in clinical diagnostics, toxicology, genetic sequencing and molecular testing. Medex Laboratories is devoted to redefining diagnostic services by providing medical practitioners and their patients with exceptional customer service paired with the most advanced and informative medical analytics to assist them in making effective treatment decisions.

Medex Laboratories fully automated laboratory utilizes state-of-the-art technologies to deliver high quality test results and service while exceeding the turnaround time requirements and demands of our physician clients. Medex Laboratories currently analyzes samples for hundreds of thousands of patients per year from providers and healthcare facilities all across the nation.

As our clients have trusted our laboratory with being an analytical and integral part of their patients' diagnosis and treatment process, we believe in respecting that trust with continuous dedication to customer satisfaction and support. We join our clients and physicians in their belief that patient care is and always will be the number one priority. Medex Laboratories' personalized support and professional service continue to exceed the expectations of our valued clients, providers and facilities. More healthcare facilities and providers, in private practices, in hospitals and in long term care facilities, are placing their trust in Medex Laboratories; and, together we are transforming advanced diagnostic information into knowledge and superior treatment options for more and more patients every day.



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