

# **MD35 MONOGENIC DIABETES TEST**

Do patients with monogenic diabetes require insulin therapy?





### **01** ABOUT MONOGENIC DIABETES

Monogenic diabetes (MD) is a subtype of diabetes caused by a single gene mutation. Maturity Onset Diabetes of the Young (MODY) is the main form of monogenic diabetes and is usually diagnosed during late childhood to adulthood, accounting for 1-5% of diabetic patients. 1 Mutations in the gene can reduce insulin production, causing the patient's blood sugar level to be persistently high. Since MODY is a genetic disease, family screening should be performed once a patient is diagnosed to understand the genetic risk to family members. <sup>2</sup>

The clinical presentation of MODY usually overlaps with other types of diabetes, making misdiagnosis common in children with MODY and young patients with diabetes.

### Who should get tested:



Diabetes patients diagnosed before the age of 25



Those with a family history of diabetes



Those with a negative antibodies test result, low to normal C-peptide level

#### Sources:

- 1. Kreider, K. E. (2019). The Diagnosis and Management of Atypical Types of Diabetes. The Journal for Nurse Practitioners, 15(2). doi:10.1016/j.nurpra.2018.09.022
- 2. Hattersley, A. T., & Patel, K. A. (2017). Precision diabetes: Learning from monogenic diabetes. Diabetologia, 60(5), 769-777. doi:10.1007/s00125-017-4226-2

## **92** DISTINGUISH YOUR MD SUBTYPE

Correct identification of the monogenic diabetes subtype will help doctors provide patients with the most suitable treatment plan. According to a report jointly published by two reputable diabetes organizations, the American Diabetes Association and the European Association for the Study of Diabetes, genetic testing is the best way to distinguish the different subtypes. 1

#### SOME COMMON GENES THAT CAUSE MD 2

Maturity-onset diabetes of the young 

Major clinical phenotype Neonatal diabetes mellitus Syndrome

Rare clinical phenotype

Gene Mutation	Clinical phenotype(s)	Recommended treatment option
ABCC8		High-dose oral sulfonylureas
GCK		No treatment needed for most patients
HNF1A		Low-dose oral sulfonylureas
HNF1B		Optimal treatment not well established
HNF4A		Low-dose oral sulfonylureas
INS	• •	Early intensive insulin treatment
KCNJII	• (	High-dose oral sulfonylureas

Note: Diabetes is caused by a variety of factors, and the presence of other genetic or non-genetic factors will also affect treatment strategies.



Test results can help doctors determine the medical needs of patients. such as recommending oral medications instead of insulin therapy for earlystage MODY patients.

#### Sources:

- 1.Chung, W.K., Erion, K., Florez, J.C. et al. (2020). Precision medicine in diabetes: a Consensus Report from the American Diabetes Association (ADA) and the European Association for the Study of Diabetes (EASD). Diabetologia 63, 1671–1693. https://doi.org/10.1007/s00125-020-05181-w.
- 2. Riddle, M. C., Philipson, L. H., Rich, S. S., Carlsson, A., Franks, P. W., Greeley, S., Nolan, J. J., Pearson, E. R., Zeitler, P. S. & Hattersley, A. T. (2020). Monogenic Diabetes: From Genetic Insights to Population-Based Precision in Care. Reflections From a Diabetes Care Editors' Expert Forum. Diabetes care, 43(12), 3117-3128 https://doi.org/10.2337/dci20-0065

### **03** GEMVCARE TEST



Tests 35 up-to-date genes related to MODY 34 sequenced genes + 1 hotspot (MtA3243G)



Double verification of results



Technical Accuracy - Validated by a local university research team (ranked No. 1 in Endocrinology and Metabolism in Asia)



Quality Assurance - periodical independent verification. GemVCare is a participant of the EMQN<sup>#</sup> programme



GemVCare has licensed the patent for monogenic diabetes testing

#EMQN (European Molecular Genetics Quality Network) is a reputable provider of molecular genetics quality assessment services in Europe.

### 94 SERVICE FLOW



### **95** ABOUT US

GemVCare - committed to helping the public prevent chronic diseases and improving the health of patients.

With professional genetic testing technology for chronic diseases, medical professionals can customize precise treatments and management models according to your personal genetic profile and health conditions so that you can live your best life.

### **OUR PARTNERS**

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#### Source

1. Best Global Universities for Endocrinology and Metabolism. U.S. News. https://www.usnews.com/education/best-global-universities/endocrinology-metabolism?region=asia. Published 2021.