Maturity Onset Diabetes of the Young and Neonatal Diabetes

Precision Panel

Overview

Maturity-onset diabetes of the young (MODY) and Neonatal Diabetes Mellitus are a heterogeneous group of inherited monogenic and polygenic disorders that are present during adolescence or young adulthood and affect an individual's blood sugar level due to beta cell dysfunction. Genetic defects in the pancreatic beta cells result in the decrease of insulin production required for glucose utilization thereby leading to very high blood sugar levels. MODY is inherited in an autosomal dominant manner and cases can also arise de-novo (for the first time) in the affected individual.

The Igenomix Maturity Onset Diabetes of the Young and Neonatal Diabetes Precision Panel can be used to make a directed and accurate differential diagnosis of neonatal and paediatric hyperglycemia ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix MODY and Neonatal Diabetes Mellitus Precision Panel is indicated for those patients with clinical suspicion of diabetes presenting with the following manifestations:

- Diabetes under the age of 35
- Family history
- Frequent urination
- Thirst
- Dehydration
- Blurry vision
- Recurrent skin infections
- Recurrent yeast infections

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.

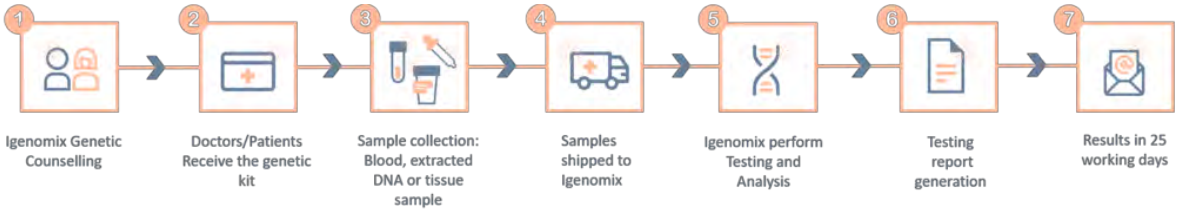
- Early initiation of treatment with a multidisciplinary team for early insulin therapy, dietary modifications, and primary prevention to reduce comorbidities associated with diabetes.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCC8	Autosomal Dominant Hyperinsulinism Due To Sur1 Deficiency, DEND Syndrome, Noninsulin-Dependent Diabetes Mellitus, Transient Neonatal Diabetes Mellitus, Transient Neonatal Hyperinsulinemic Hypoglycemia, Familial Hypoglycemia Of Infancy, Leucine-Sensitive MODY Permanent Neonatal Diabetes Mellitus, Transient Neonatal Diabetes Mellitus	AD,AR	99.98%	710 of 712
APPL1	Maturity-Onset Diabetes Of The Young, Type 14	AD	99.91%	4 of 4
BLK	Maturity-Onset Diabetes Of The Young, Type 11	AD	100%	21 of 21
CEL	Maturity-Onset Diabetes Of The Young Type 8 With Exocrine Dysfunction	AD	91.14%	30 of 31
GCK	Diabetes Mellitus, Noninsulin-Dependent Diabetes Mellitus, Permanent Neonatal Hyperinsulinemic Hypoglycemia, Familial Hyperinsulinism Due To Glucokinase Deficiency, Maturity-Onset Diabetes Of The Young Type II, Permanent Neonatal Diabetes Mellitus	AD,AR	100%	905 of 909
HNF1A	Insulin-Dependent Diabetes Mellitus, Noninsulin-Dependent Diabetes Mellitus, Hepatic Adenomas, Familial Hyperinsulinism Due To HNF1a Deficiency, Maturity-Onset Diabetes Of The Young Type III, Nonpapillary Renal Cell Carcinoma	AD	100%	529 of 538
HNF1B	17q12 Microdeletion Syndrome, Noninsulin-Dependent Diabetes Mellitus, HNF1b-related Autosomal Dominant Tubulointerstitial Kidney Disease, Nonpapillary Renal Cell Carcinoma, Nonpapillary Renal Cysts And Diabetes Syndrome	AD	100%	219 of 220
HNF4A	Noninsulin-Dependent Diabetes Mellitus, Fanconi Renotubular Syndrome With Maturity-Onset Diabetes Of The Young, HNF1b-related Autosomal Dominant Tubulointerstitial Kidney Disease, Hyperinsulinism Due To HNF4a Deficiency, Maturity-Onset Diabetes Of The Young Type 1	AD	100%	172 of 174
INS	Insulin Diabetes Mellitus, Permanent Neonatal Diabetes Mellitus, Hyperproinsulinemia, Maturity-Onset Diabetes Of The Young, Permanent Neonatal Diabetes Mellitus	AD,AR	100%	78 of 84
KCNJ11	Autosomal Dominant Hyperinsulinism Due To KIR6.2 Deficiency, DEND Syndrome, Noninsulin-Dependent Diabetes Mellitus, Permanent Neonatal Diabetes Mellitus	AD,AR	100%	190 of 191
KLF11	Maturity-Onset Diabetes Of The Young Type VII	AD	99.87%	10 of 10
NEUROD1	Noninsulin-Dependent Diabetes Mellitus, Maturity-Onset Diabetes Of The Young	AD	99.94%	23 of 24
PAX4	Ketosis-Prone Diabetes Mellitus, Noninsulin-Dependent Diabetes Mellitus, Maturity-Onset Diabetes Of The Young Type IX	AD,AR,MU	100%	21 of 21
PDX1	Noninsulin-Dependent Diabetes Mellitus, Permanent Neonatal Diabetes Mellitus, Maturity-Onset Diabetes Of The Young Type 4, Pancreatic Agenesis, Congenital Permanent Neonatal Diabetes Mellitus	AD,AR	98.02%	32 of 36
RFX6	Mitchell-Riley Syndrome	AR	100%	34 of 34

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology



Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

References

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