

# Diabetes and Obesity Genetic Panel

## Genetic testing for associated diseases and syndromes

Alarming statistics report that one in 10 Americans has diabetes, and 90% of those 34 million has type 2 diabetes. Obesity prevalence is equally startling, with the obesity rate of the U.S. population age 20 and older surpassing 40%. If current trends continue, one in three people in the United States will have type 2 diabetes by 2050, and the obesity rate will climb even higher.\*

While diabetes often goes hand in hand with obesity, the underlying reason for the disease can often be found in a person's genomic DNA. Diabetes and Obesity Genetic Panel specifically tests for genetic types of diabetes.

### The Extensive Benefits of Genetic Testing

Although there's no cure for diabetes and obesity disorders, there are many advantages to understanding a patient's genetic risk factors.

#### Lab's Diabetes and Obesity Genetic Panel can:

- Detect potential underlying genetic causes for diabetes
- Provide or lead to an accurate diagnosis

#### As a result, the patient can:

- Gain insight into potential risks and conditions
- Make timely lifestyle adjustments
- Seek necessary treatment to minimize long-term complications
- Share risk factors with family members who might have the same genetic variant
- Participate in clinical trials
- Obtain gene therapy, if available
- Explore options for family planning and embryo selection

#### And the physician can:

- Personalize a patient's treatment plan
- Connect the patient with relevant resources and support

### Under the Microscope

Acceptable sample requirements:

- Blood, two 4-mL EDTA tubes, lavender top
- Extracted DNA, 3 µg in EB buffer
- Buccal swab or saliva

**Positive** A pathogenic/likely pathogenic variant is detected in the patient's DNA  
This type of variant is known to increase the patient's risk of a genetic condition

**Negative** No variant detected in the patient's DNA using this panel  
Patient may still be at risk for the tested condition, particularly if a family history exists

**VUS** Variant of Uncertain Significance (VUS) is detected  
Not enough information is known to link the variant to diabetes

### Who Should Be Tested?

Our Diabetes and Obesity Genetic Panel is uniquely designed for patients who have symptoms associated with diabetes, an obesity disorder and/or a family history of diabetes. It is also appropriate for family members – siblings, parents, children – of patients who have a genetic variant for diabetes or obesity disorders.

### Diabetes and Obesity Disorders Tested

**Diabetes and Obesity Genetic Panel tests for associated forms of diabetes and obesity, including:**

- Type 1 diabetes mellitus (T1DM)
- Type 2 diabetes mellitus (T2DM)
- Permanent neonatal diabetes mellitus (PNDM, onset within the first six months of life)
- Bardet Biedl syndrome (BBS, a rare genetic obesity disease)
- Prader Willi syndrome (PWS, a complex medical condition)
- Maturity-onset diabetes of the young (MODY)
- Alström syndrome (a rare genetic disorder affecting organ systems)
- Wolfram syndrome (a rare genetic endocrine disorder)
- Renal cysts and diabetes syndrome (MODY 5, maturity-onset diabetes of the young)
- Insulin-dependent diabetes mellitus secretory diarrhea syndrome (IPEX, systemic autoimmunity that typically begins in infancy)
- Familial partial lipodystrophy (FPL, a genetic disorder of progressive body fat loss)

L A B S

\* Centers for Disease Control and Prevention

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## Beyond the Test

The importance of an accurate, timely diagnosis cannot be overstated.

- Diabetes impacts many parts of the body beyond blood: eyes, kidneys, nerves, heart and blood vessels, feet and gums.
- The risk factors associated with diabetes are also risk factors for other serious chronic conditions and diseases, including sleep apnea, dementia, bacterial and fungal infections, stroke, heart disease and high blood pressure.
- An accurate diagnosis allows a patient to address a wide range of environmental factors, including nutrition and physical activity affecting health and health behavior.
- A diagnosis is also important to family members who might be at similar risk of having inherited the same genetic variant and make necessary lifestyle changes to delay the onset of diabetes.

## 56 Genes Tested in Our Panel

Gene	Diseases-Related Conditions
<i>ABCC8</i>	Hyperinsulinemic Hypoglycemia, Diabetes Mellitus, Transient Neonatal
<i>ADRB2</i>	Pulmonary Disease, Body Mass Index Quantitative Trait Locus
<i>ADRB3</i>	Obesity Due to Melanocortin 4 Receptor Deficiency, Metabolic syndrome
<i>AGRP</i>	Body Mass Index Quantitative Trait Locus, Anorexia Nervosa
<i>ALMS1</i>	Alström Syndrome
<i>ARL6</i>	Bardet-Biedl Syndrome, Retinitis Pigmentosa
<i>BBS1</i>	Obesity, Bardet-Biedl Syndrome
<i>BBS10</i>	Obesity, Bardet-Biedl Syndrome
<i>BBS12</i>	Obesity, Bardet-Biedl Syndrome
<i>BBS2</i>	Obesity, Bardet-Biedl Syndrome
<i>BBS4</i>	Obesity, Bardet-Biedl Syndrome
<i>BBS5</i>	Obesity, Bardet-Biedl Syndrome
<i>BBS7</i>	Obesity, Bardet-Biedl Syndrome
<i>BBS9</i>	Obesity, Bardet-Biedl Syndrome
<i>BDNF</i>	Bipolar Disorder, Anxiety, Eating disorders
<i>CARTPT</i>	Body Mass Index Quantitative Trait Locus, Cocaine Abuse
<i>CEL</i>	Maturity-Onset Diabetes of the Young, Pancreatic Disease
<i>CEP290</i>	Joubert Syndrome, Senior-Loken Syndrome
<i>EIF2AK3</i>	Epiphyseal Dysplasia, Early-Onset Diabetes Mellitus, Permanent Neonatal Diabetes Mellitus
<i>ENPP1</i>	Hypopigmentation, Keratoderma
<i>FOXP3</i>	IPEX Syndrome
<i>GCK</i>	Maturity-Onset Diabetes of the Young
<i>GHRL</i>	Body Mass Index Quantitative Trait Locus, Bulimia Nervosa
<i>GLIS3</i>	Neonatal Diabetes Mellitus with Congenital Hypothyroidism
<i>GNAS</i>	Mccune-Albright Syndrome, Pseudohypoparathyroidism
<i>HNF1A</i>	Maturity-Onset Diabetes of the Young
<i>HNF1B</i>	Maturity-Onset Diabetes of the Young
<i>HNF4A</i>	Maturity-Onset Diabetes of the Young
<i>INS</i>	Permanent Neonatal Diabetes Mellitus
<i>KCNJ11</i>	Permanent Neonatal Diabetes Mellitus
<i>LEP</i>	Congenital Leptin Deficiency
<i>LEPR</i>	Excessive Hunger, Massive Weight Gain
<i>MAGEL2</i>	Arthrogryposis, Prader-Willi Syndrome, Autism
<i>MC4R</i>	Monogenic Obesity
<i>MKKS</i>	McKusick-Kaufman Syndrome, Bardet-Biedl Syndrome
<i>MKS1</i>	Meckel Syndrome, Type 1 Diabetes Mellitus, Bardet-Biedl Syndrome
<i>NEUROD1</i>	Maturity-Onset Diabetes of the Young, Type 6 and Type 2 Diabetes Mellitus
<i>NEUROG3</i>	Congenital Malabsorptive Diarrhea, Hyperglycemia
<i>NTRK2</i>	Obesity, Hyperphagia, Developmental Delay, Developmental and Epileptic Encephalopathy
<i>PCSK1</i>	Obesity, Malabsorptive Diarrhea, Hypogonadotropic Hypogonadism, Altered Thyroid and Adrenal Function, Impaired Regulation of Plasma
<i>PDX1</i>	Maturity-Onset Diabetes of the Young Type 4, Pancreatic Agenesis
<i>POMC</i>	Early-Onset Diabetes
<i>PPARG</i>	Lipodystrophy, Familial Partial, Type 3 Diabetes, Body Mass Index Quantitative Trait Locus
<i>PPARGC1B</i>	Type 2 Diabetes Mellitus
<i>PTF1A</i>	Pancreatic and Cerebellar Agenesis, Pancreatic Agenesis
<i>PYY</i>	Anorexia Nervosa, N-Acetylglutamate Synthase Deficiency
<i>RFX6</i>	Mitchell-Riley Syndrome, Diabetes Mellitus
<i>SDC3</i>	Body Mass Index Quantitative Trait Locus
<i>SDCCAG8</i>	Bardet-Biedl Syndrome, Senior-Loken Syndrome
<i>SIM1</i>	Obesity Due to SIM1 Deficiency, SIM1-Related Prader-Willi-Like Syndrome
<i>TRIM2</i>	Muscular Dystrophy, Limb-Girdle
<i>TTC8</i>	Bardet-Biedl Syndrome with Early-Onset Obesity
<i>UCP1</i>	Multiple Symmetric Lipomatosis, Lipomatosis
<i>UCP3</i>	Body Mass Index Quantitative Trait Locus, Leptin Deficiency or Dysfunction
<i>WDPCC</i>	Congenital Heart Defects, Hamartomas of Tongue, Polysyndactyly Syndrome, Bardet-Biedl Syndrome
<i>WFS1</i>	Wolfram Syndrome