Metabolic and Endocrine Genetics

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

What is ELAB of Flordida Metabolic and Endrorine Genetics?

Metabolic diseases span a vast range of conditions. Inherited metabolic disorders have a defective gene that results in an enzyme deficiency. Metabolic disorders include cystic fibrosis, phenylketonuria (PKU), hyperlipidemia, etc.Most metabolic disorders are genetic in origin. While metabolic disorders are individually rare, collectively they affect 1% to 3% of the world population. The onset of symptoms could be sudden or could progress slowly. Symptoms may be brought on by foods, medications, dehydration, minor illnesses, or other factors. Symptoms of several metabolic conditions appear within a few weeks of birth. Other inherited metabolic disorders might take years to develop.

Genome sequencing continues to reveal the functions and dysfunctions of particular genes. Therefore, genetic tests are better especially when hormone tests provide ambiguous results. These reveal inheritable components and treatment plans for common health issues such as thyroid disease and rare adrenal tumors.

Generality

Metabolic

Metabolic disorders are common in the United States. About 1 in 3 adults have either metabolic or endocrine syndrome. Among US adults aged 18 years or older, the prevalence of metabolic and endocrine syndrome rose by more than 35% from 1988–1994 to 2007–2012, increasing from 25.3% to 34.2%.

Endrocrine

Endocrine disorders with U.S. prevalence estimates of at least 5% in adults included diabetes mellitus, impaired fasting glucose, impaired glucose tolerance, obesity, metabolic syndrome, osteoporosis, osteopenia, mild-moderate hypovitaminosis D, dyslipidemia, and thyroiditis.

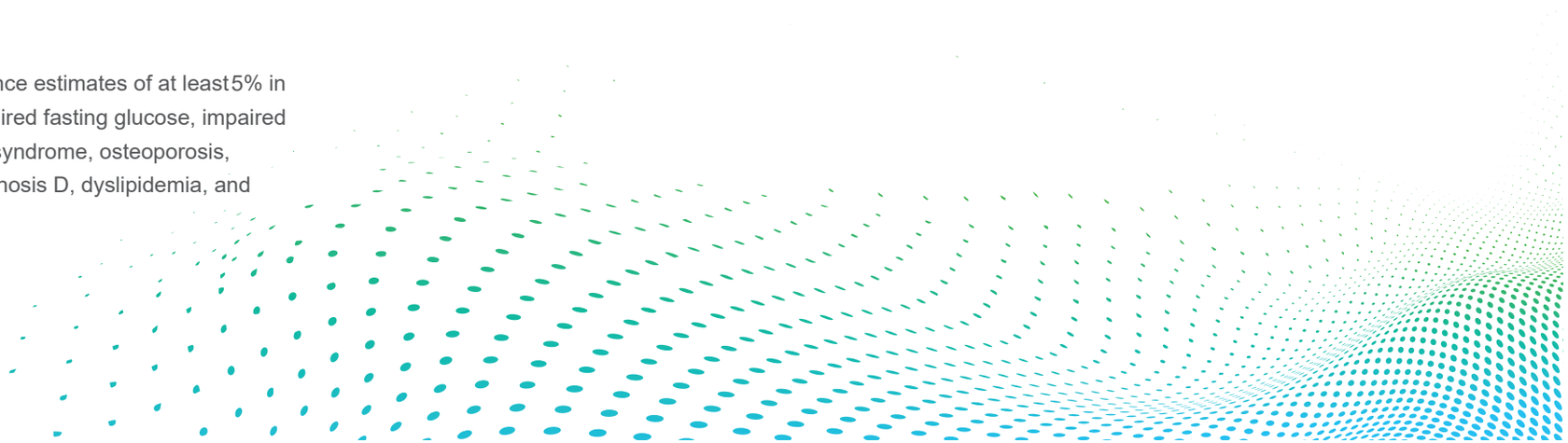
What are the 6 common Metabolic and Endocrine diseases?

Metabolic disorders

- Isovaleric acidaemia (IVA)
- Glutaric aciduria type 1 (GA1)
- Homocystinuria (pyridoxine unresponsive) (HCU)
- Hemochromatosis
- Type 1 diabetes
- Type 2 diabetes

Endocrine disorders

- Congenital Hypothyroidism
- Obesity
- Diabetes
- Congenital Adrenal Hyperplasia(CAH)
- Androgen receptor deficiency
- Kallmann syndrome
- A familial lipid disorder
- Prolactinoma
- Cancers of the endocrine glands



When do you need to get tested for Genetic Metabolic and Endocrine Disorders?

Most genetic metabolic disorders have no specific clinical appearances, and their clinical manifestations are highly complex and variable. Some of the commonly seen symptoms are listed below.

- Weight loss
- Failure to gain weight or grow
- Lethargy
- Poor appetite
- Abdominal pain
- Vomiting
- Jaundice
- Seizures
- Developmental delay
- Coma
- Abnormal odour of urine, breath, sweat, or saliva

Endrocrine

- Ambiguous genitalia
- Dysgenic gonads
- Short stature
- Obesity with or without associated developmental delay
- Hyperglycemia with a strong family history

Mitochondrial Disoders

- A clinically heterogeneous group of genetic disorders arising due to mutations in mitochondrial DNA, or nuclear DNA coding for mitochondrial components
- A group of disorders that are difficult to diagnose, because it affects each individual differently
- Symptoms can include seizures, strokes, severe developmental delays, inability to walk, talk, see, and digest food, combined with a host of other complications

Who should be screened for Metabolic and Endocrine disorders?

- Individuals with family history of metabolic or endorcine disorders.
- Individuals with a history of high blood pressure, high triglycerides and being overweight or obese.
- In the United States,The most common endocrine disease is diabetes in both men and women. Women who have had diabetes during pregnancy (gestational diabetes) or people who have a family member with type 2 diabetes are at greater risk for metabolic syndrome.
- PCOS is a combination of metabolic and reproductive endocrine disorders, so there are a large number of changes in metabolites involved.
- If your hormone levels are too high or too low, you may have an endocrine disease or disorder. Endocrine diseases and disorders also occur if your body does not respond to hormones the way it is supposed to.

Advantages of using ELAB of Flordia

- Expertise in genetic testing with large in -house database
- High coverage with more than 22000 genes
- Dedicated in-house lab
- High throughput NGS machines
- Screening on world-class Illumina platform
- Best-in-class methods - NGS (X10, HiSeq, MiSeq) Sanger

Why do we Recommend ELAB of Florida custom testing for patients with Genetic Metabolic and Endocrine Disorders?

ELAB/LABDX offers a wide and flexible range of custom designed gene mutation tests which have been developed with in-depth insightful understanding of the genetic disorder with the latest research in that particular domain. Updated technologies, world-class customer service, and clear result interpretation along with counselling sessions with our expert genetic counsellors make us equipped to provide you the best available support for your patients and families with Genetic Metabolic and Endocrine Disorders.

What is ***OUR*** test methodology?

1. Next-generation Sequencing (NGS)

Using genomic DNA extracted from blood, the coding regions of all the genes are captured and sequenced simultaneously by NGS technology on an Illumina platform. The sequence data that is generated is aligned and analyzed for sequence variants.

2. *Multiplex Ligation-Dependent Probe Amplification (MLPA)*

Deletion and duplication analysis of genomic DNA is carried out by MLPA. This method allows for the amplification of multiple targets with only a single primer pair.

3. *Fragment Analysis PCR for Repeat Expansion Analysis*

These rely on the detection of changes in the length of a specific DNA sequence toindicate the presence of repeat expansions.

Test sample requirements

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Required forms

- Relevant clinical information including all the clinical presentations and symptoms
- Test request form

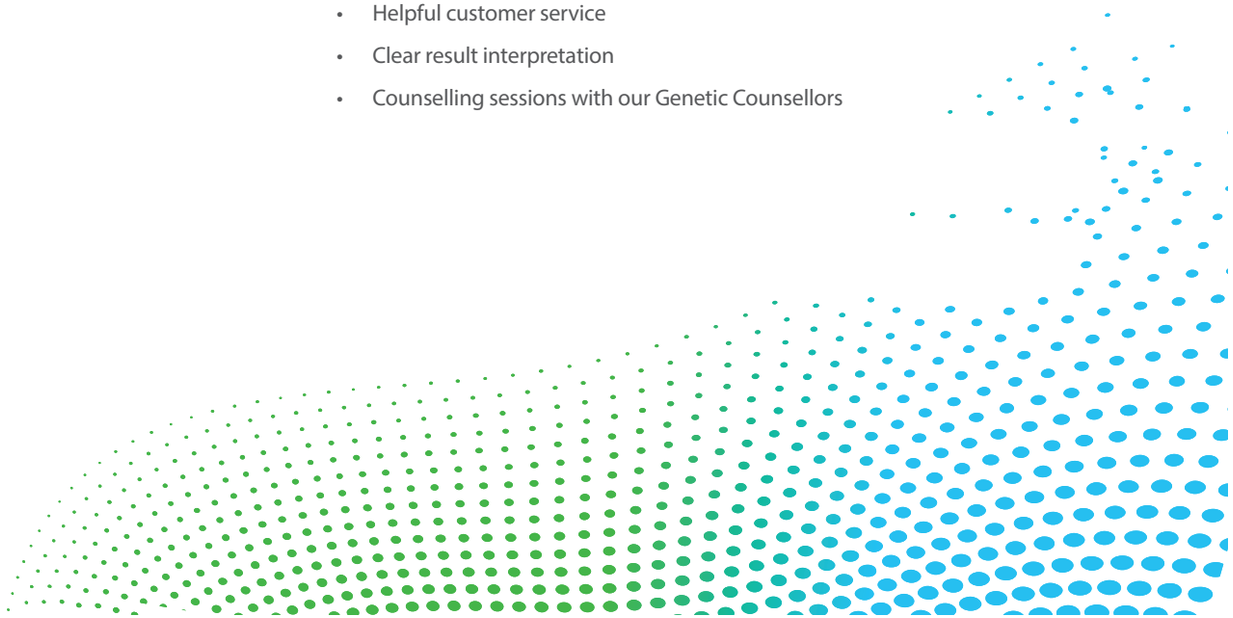
Turn Around Time (TAT)

- 21 working days for NGS
- 21 working days for Sanger Sequencing

Free Genetic Counselling

E-lab of Florida offers all your patients FREE pre & post-test genetic counselling with our expert *and certified genetic counsellors.*
Best available support for your patients and families via

- Latest technologies
- Helpful customer service
- Clear result interpretation
- Counselling sessions with our Genetic Counsellors



ELAB of Florida offers the following custom gene tests for Metabolic and Endocrine disorders

Metabolic

- *Aicardi-Goutières Syndrome Panel*
- *Comprehensive Metabolism Panel*
- *Congenital and Familial Lipodystrophy Panel*
- *Congenital Disorders of Glycosylation Panel*
- *Congenital Mono- and Disaccharide Disorders Panel*
- *Creatine Metabolism Deficiency Panel*
- *Cystinuria Panel*
- *Fatty Acid Oxidation Syndrome Panel*
- *Glycogen Storage Disorder Panel*
- *Hereditary Hemochromatosis Panel*
- *Homocystinuria Core Panel*
- *Hyperammonemia and Urea Cycle Disorder Panel*
- *Hyperphenylalaninemia Panel*
- *Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel*
- *Hypomagnesemia Panel*
- *Lysosomal Disorders and Mucopolysaccharidosis Panel*
- *Metabolic Liver Failure Panel*
- *Metabolic Myopathy and Rhabdomyolysis Panel*
- *Mitochondrial DNA Depletion Syndrome Panel*
- *Monogenic Obesity Panel*
- *Nephrolithiasis Panel*
- *Nonketotic Hyperglycinemia / Glycine Encephalopathy Panel*
- *Organic Acidemia/Aciduria & Cobalamin Deficiency Panel*
- *Periodic Paralysis Panel*
- *Peroxisomal Disorders Panel*
- *Porphyria Panel*
- *Purine and Pyrimidine Metabolism Disorders Panel*
- *Tyrosinemia Panel*

Endocrine

- *Comprehensive Monogenic Diabetes Panel*
- *Congenital Adrenal Hyperplasia Panel*
- *Glucocorticoid Deficiency Panel*
- *Hyperlipidemia Panel*
- *Hyperparathyroidism Panel*
- *Hypothyroidism and Resistance to Thyroid Hormone Panel*
- *Kallmann Syndrome Panel*
- *Monogenic Obesity Panel*

e-lab

6100 Hollywood Blvd STE 306
Hollywood, FL 33024
Phone: +1 954-530-8332

CLIA – 10D2126070

For more information
www.elabfl.com