Evartia®
metabolic test

Answers through genetic metabolic testing



MEDICOVER GENETICS

WHAT IS **Evartia**?

Evartia is a genetic test which screens for genetic mutations in people suspected of having an **Inherited Metabolic Disease** (IMD). IMDs exhibit a **wide variety of characteristics**, both in the age of symptoms onset and in the clinical presentation and severity of symptoms. Through **genetic testing**, Evartia can identify the disease-causing variant accurately, quickly, and reliably, signaling the start of beneficial clinical management that can prevent or reduce acute, life-threatening symptoms and chronic complications, benefiting your patient's quality of life.

CLINICAL UTILITY

Defects in metabolic genes, which cause IMDs, disrupt the actions of metabolic pathways leading to either toxic accumulation of substances or deficient production of important enzymes and proteins. These can cause a **variety of symptoms**, that usually manifest shortly after or within weeks of birth; but they could also appear in infancy, childhood, adolescence, or adulthood. Symptoms and metabolic episodes, which necessitate urgent, appropriate medical action, can also be triggered by specific foods or medications, dehydration, minor illness, sweat or other factors. Without appropriate clinical management, symptoms can get **progressively worse**, cause acute pain, and **chronic**, **irreversible complications**.

The misconception that IMDs are solely 'childhood diseases' has kept many adults with metabolic diseases undiagnosed. **Adult manifestation** of symptoms differs from the classic presentation of signs and symptoms seen in children for the same disease, and may also include **cardiovascular or neurological symptoms**. This makes identifying a metabolic disorder in an adult a complex, lengthy, and extensive process, with patients being undiagnosed or misdiagnosed and receiving wrong and unnecessary treatments.

Through genetic testing, Evartia could provide a simple, straightforward answer for your patients, regardless of their age and how complicated their clinical presentation is.

WHO IS **Evartia** FOR?

- Patients with common symptoms of a metabolic disease (abdominal pain, vomiting, weight loss, vision disturbances, etc.)
- Patients with a spectrum of overlapping symptoms that vary in age of onset and severity
- Patients with neurological symptoms that haven't improved with routine therapies
- Individuals with a family history of a metabolic disease

WHAT DOES **Evartia** TEST FOR?

To facilitate the detection process, Evartia metabolic test is offered as **a single**, **detailed panel of 223 genes** involved in metabolic pathways.

223 genes

DISEASE CATEGORIES TESTED BY Evartia METABOLIC PANEL

3-Methylglutaconic aciduria disorders
Cerebral creatine deficiency
Congenital disorders of glycosylation
Fatty acid oxidation disorders
Glycine encephalopathy
Glycogen storage diseases
Hyperinsulinemic hypoglycemia
Hyperphenylalaninemia
Lysosomal storage disorders
Maple syrup urine disease and DLD deficiency
Methylmalonic acidemia
Peroxisomal disorders
Urea cycle disorders

For a complete list of the 223 genes tested by Evartia, please visit www.medicover-genetics.com

WHY RECOMMEND **Evartia** TO MY PATIENTS?

The diagnostic path for an IMD hasn't been straightforward. Many IMDs are included in national newborn screening programs; however, patients may not be identified due to:

- the IMD not being tested in their country, or not tested on the year the patient was born
- technological limitations
- detection depending on the time of symptom onset

Often, patients have to undergo lengthy biochemical testing which includes a variety of specimen types, or invasive biopsies from muscle or cerebrospinal fluid. Evartia can help you take informed and accurate decisions on the best clinical management for your patient through a simple, non-invasive procedure.

CASE STUDY

7-year-old female with rhabdomyolysis attack, high levels of acylcarnitine, and retinopathy

Medical history shows

- sibling who passed away from Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase (LCHAD) deficiency
- no irregular heart function, which is an indicator of LCHAD deficiency

Genetic testing was performed to confirm the suspected clinical diagnosis.

Evartia metabolic testing results

A **pathogenic**, **homozygous variant** at exon 15 of the HADHA gene was detected, which is associated with **LCHAD** and **Mitochondrial Trifunctional Protein** (MTP) deficiency.

Evartia facilitated the patient's diagnosis, along with the need for an appropriate clinical management plan. The latter included a specialized diet with decreasing feeding intervals, monitoring and management of symptoms by specialists, and a list of triggers, such as fasting, for the patient to avoid, prevent or control acute episodes. Genetic testing and genetic counseling was also recommended for family members.

TECHNOLOGICAL ADVANTAGES

TARGETED TECHNOLOGY

Evartia metabolic test is based on a novel, **Target Capture Enrichment Technology** which utilizes Next Generation Sequencing, and has been thoroughly validated for its accuracy and precision.

NOVEL BIOINFORMATICS

Innovative bioinformatic pipelines analyze the sequencing data produced from each sample, increasing the sensitivity and specificity of Evartia.

FULL COVERAGE

Evartia screens for **all coding regions*** of the genes tested, and targets adjacent non-coding sequence, increasing the chances of identifying any pathogenic or likely pathogenic mutations:

- single nucleotide variants (SNVs)
- small insertions and deletions (INDELs)
- copy number variants (CNVs)

**Exceptions on regions containing repeats, sequences of high homology (pseudogenes and segmental duplications) or high GC-content*

BENEFITS OF **Evartia**

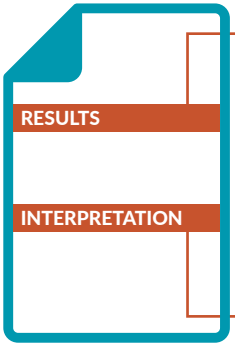
SAFE
Non-invasive sample collection

VALUABLE
Leads to the most applicable clinical management

EXPANDED
Tests an extensive list of clinically actionable mutations

THOROUGH
Full exonic coverage*

WHAT WILL THE **REPORT** TELL ME?



CLINICALLY SIGNIFICANT VARIANT DETECTED
A pathogenic or likely pathogenic variant has been identified in a gene tested, which is associated with a metabolic disease.

NO CLINICALLY SIGNIFICANT VARIANT DETECTED
No disease-causing genetic variant has been identified in the genes tested.

VARIANT OF UNCERTAIN SIGNIFICANCE
A genetic change has been detected, but it is currently unknown whether that change is associated with a genetic disorder.

Summary of the results and recommendations

Upon request, carrier status can be reported for recessive conditions.
Variant re-evaluation can be requested by healthcare providers at defined timeframes.

WHAT CAN I DO AFTER **Evartia**?

Evartia could detect the genetic variant that is responsible for causing your patient's symptoms and clinical condition.
The comprehensive report will help you to:

- Identify your patient's disease
- Determine an accurate prognosis
- Provide the most optimal treatment and clinical management
- Consider applicable investigational therapies, gene therapies or clinical trials
- Be aware of specific complications that may arise and how to prevent or manage them
- Inform your patient of how this disease will affect their lives and that of their families
- Recommend testing and genetic counseling to family members, if applicable

**Exceptions apply*

HOW TO ADMINISTER **Evartia**?

Recommend **Evartia** to your patient

Collect a buccal swab from your patient



Send the sample to **Medicover Genetics**

The sample will be analyzed at **Medicover Genetics** laboratories

Results will be sent to you within 2-4 weeks from sample receipt

MORE **QUESTIONS**?

If you have additional questions or concerns, please contact us at **info.genetics@medicover.com**

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