

WHAT ARE INHERITED METABOLIC DISEASES?

Metabolic pathways have pivotal roles in keeping our bodies healthy. Their many actions include producing energy and taking the necessary nutrients from the foods we consume. When a mutation (genetic change) occurs in a metabolic pathway, it can lead to either accumulation of toxic substances in our body, or insufficient production of required products that keep us healthy and functional. As a result, metabolic diseases can **affect our health and development**, and cause **acute** and **chronic complications**.

Symptoms of inherited metabolic diseases usually appear shortly after birth. However, depending on the mutation, the metabolic pathway involved and the severity of the condition, some people with inherited metabolic diseases can develop symptoms in **early or late adulthood**. There is **great variability** in the symptoms and the severity of symptoms caused by metabolic diseases. Importantly, adults and children with the same metabolic disorder may have **different symptoms**. In addition, the misconception that metabolic diseases only affect babies and young children, leaves many adults suffering from metabolic diseases **unrecognized** and **undetected**.

Symptoms of metabolic diseases include:



Vomiting



Abdominal pain



Lethargy



Psychological or neurological symptoms



Weight loss



Seizures

Additional symptoms may include vision disturbances, impaired kidney function, heart problems, abnormal movements, behavioral or learning issues, distinctive facial features and recurrent infections.

HOW IS **Evartia** ADMINISTERED?



ASK YOUR DOCTOR ABOUT Evartia



YOUR DOCTOR WILL COLLECT A SAMPLE USING A BUCCAL SWAB



THE SAMPLE WILL BE SENT TO NIPD Genetics



THE SAMPLE WILL BE ANALYZED IN OUR LABORATORY



RESULTS WILL BE SENT TO YOUR DOCTOR WITHIN 2-4 WEEKS FROM SAMPLE RECEIPT

MORE **QUESTIONS?**

If you have additional questions or concerns, please ask your doctor. You can also contact us:



NIPD Genetics Public Company Ltd
www.nipd.com
info@nipd.com



NIPD GENETICS

Evartia
metabolic test

Answers through genetic
metabolic testing

SAFE | SENSITIVE | RELIABLE

WHAT IS **Evertia**?

Evertia is a new genetic test that detects genetic mutations (changes) that cause **inherited metabolic diseases** in people.

People with inherited metabolic diseases can have a **range of symptoms**, with **variable expressivity** and **age of onset**. Symptoms can manifest shortly after birth or at infancy, childhood, adolescence or adulthood. Symptoms can also occur suddenly due to specific foods or medications, dehydration, illnesses or other factors. With Evertia, if you have an inherited metabolic disorder, detecting and managing it early can **prevent or reduce symptoms, avoid chronic health consequences** and **improve your quality of life**.

HOW DOES **Evertia** HELP ME?

Evertia can help you identify:



What disease you have



What is your prognosis (*how the disease will progress based on the specific mutation you have*)



What is the best treatment and clinical management for you



What complications you should be aware of



How it will affect your life



Who in your family should be tested



Who can help you (*specialized doctors, dieticians, support groups*)



WHO IS **Evertia** FOR?



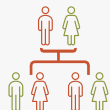
Individuals with common symptoms of a metabolic disease



Individuals with a spectrum of overlapping symptoms that vary in age of onset and severity



Individuals with neurological symptoms that haven't improved with routine therapies



Individuals with a family history of a metabolic disease

WHY ASK MY DOCTOR ABOUT **Evertia**?

Inherited metabolic diseases may be individually rare, but collectively they are numerous. As they have **variable** symptoms, detecting an inherited metabolic disorder in an individual is not always straightforward.

The usual pathway of identifying a metabolic disease involves lengthy and complicated biochemical or enzymatic tests. This may include testing a variety of specimen types, such as blood, urine, sweat, and even undergoing invasive brain or muscle tissue biopsy. Such tests rely on reference intervals which may not always be suitable for the individual's age, gender or current state of health - whether they are in the middle of a metabolic episode or not. Often, they also require a genetic test to confirm the mutation.

With Evertia, you are one **simple, painless** and **reliable** test away from taking **informed** and **accurate decisions** on the **best clinical management for you**.

WHY CHOOSE **Evertia**?

223
GENES

Tests for disease-causing genetic changes that cause serious health problems



Identifies the best therapy for you that can reduce or improve symptoms and chronic complications



Painless and non-invasive collection method



Reduces the need for lengthy, complicated and invasive procedures



Utilizes superior technology that yields accurate results

WHAT DOES **Evertia** TEST FOR?

Evertia metabolic test covers the major categories of inherited metabolic diseases and is offered as a single, detailed panel of **223 genes** involved in metabolic pathways.

Evertia tests for autosomal recessive, autosomal dominant and X-linked disorders. These may be inherited from carrier parents, who are unaware of their carrier status, to their children, or without a known or prior family history of that disorder. Additionally, this means that if you are affected by a metabolic disease, some of your family members may also benefit from metabolic testing. **Early detection** has a higher potential to reduce symptoms or minimize their severity, **improving the quality of life** of the person with the metabolic disease.

Disease categories tested by **Evertia** metabolic panel

3-Methylglutaconic aciduria

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