What are Inherited Metabolic Diseases?

Inherited Metabolic Diseases, also known as Inborn Errors of Metabolism, are genetic conditions that affect a person's metabolism. They can lead to either accumulation of toxic substances in our body, or insufficient production of required products that keep us healthy and functional.

How common are Inherited Metabolic Diseases?

Inherited Metabolic Diseases are individually rare but collectively numerous. They occur in about 1 in 2500 infants.

What are the complications of Inherited Metabolic Diseases?

Complications of Inherited Metabolic Diseases include abdominal pain, lethargy, vomiting, psychological or neurological symptoms, vision Disturbances, impaired kidney function, heart problems, abnormal movements, behavioral or learning issues, distinctive facial features, and recurrent infections.

When do the symptoms of Inherited Metabolic Diseases appear?

Symptoms of inherited metabolic diseases usually appear shortly after birth, and sometimes 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.

Why ask my healthcare provider about Evartia?

As Inherited Metabolic Diseases 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. 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. Additionally, invasive brain or muscle tissue biopsy might be needed. Often, they also require a genetic test to confirm the mutation. With Evartia, you are one simple, painless and reliable test away from taking informed and accurate decisions on the best clinical management for you.

I have other questions. Where can I find additional information?

Please contact your healthcare provider for additional information. You can also have a look at www.nipd.com/products/postnatal/evartia/