**SHIPPING LOGISTICS:**

For each test, NEONATAL-ONE kit is available with the necessary material for performing the test and sending it to our laboratory.

The envelope contains:

- Application form (two copies).
- Specific card filter paper.
- Shipment and payment information.
- Procedure for healthcare personnel to collect the sample.
- A shipment envelope with the mailing data.

The doctor (or the mother) will send the sample using our courier service giving our client number.

The results are sent by post and/or e-mail to the address indicated on the application form. ( Optionally, they can also be downloaded from our website/encrypted.)

---

**SCIENTIFIC BIBLIOGRAPHY**


---

**GENETADI BIOTECH, S.L.**

Parque Tecnológico de Bizkaia  
Edificio 502  
48160 - Derio (Vizcaya)  
SPAIN

Tel.: (+34) 944.044.343  
Fax: (+34) 946.566.628  
www.genetadi.com
### Analysis of 56 congenital metabolic diseases

- Expanded Dry Blood Neonatal Test: Expanded Heel Test
- Additional screening for the standard performed by the States.
- Analysis of 56 congenital metabolic diseases. (Inborn errors of metabolism)

#### LIST OF DISEASES

- **Congenital metabolic diseases** (also known as inborn errors of metabolism (IEM) or congenital metabolic disorders) are a group of rare genetic disorders with which the body is not able to breakdown the nutrients. One out of every four thousand newborns has IEM.

  **Metabolic alterations** can appear in the form of disease from a very early age, either due to the lack of necessary substances for the body’s operation or due to the accumulation of intermediate products that cannot be processed correctly.

  If an individual substrate correctly and transform them into sub-products and energy, the following consequences may result:
  - Intermediate compounds that when accumulated are toxic may appear.
  - The individual is in physiological need of an essential metabolite that cannot be obtained because of altered metabolic pathway.
  - The person may lack the energy necessary for the correct functioning of the body.

  The diseases that can be detected with this test are severe, some even mortal in some cases; their early identification makes dietetic treatments possible, either by substitution of metabolites that is missing the necessary nutrient or by eliminating the substance that cannot be metabolized by the affected person.

#### What is NEONATAL-ONE?

**NEONATAL-ONE** is an expanded neonatal screening device (expanded heel test) based on tandem mass spectroscopy technology (MS/MS) that allows detection of 56 congenital metabolic diseases in a dry blood sample.

This test is in addition to the official screenings offered by the health services of each state or region, and so it must always be done jointly. The MS/MS detects the levels of amino acids and acylcarnitines derived from alterations in the metabolism of amino acids, organic acids and fatty acids.

### What are metabolic disorders?

Metabolic disorders are a group of rare genetic disorders in which the body is not able to breakdown the nutrients. One out of every four thousand newborns has IEM.

**Metabolic alterations** can appear in the form of disease from a very early age, either due to the lack of necessary substances for the body’s operation or due to the accumulation of intermediate products that cannot be processed correctly.

If an individual substrate correctly and transform them into sub-products and energy, the following consequences may result:
- Intermediate compounds that when accumulated are toxic may appear.
- The individual is in physiological need of an essential metabolite that cannot be obtained because of altered metabolic pathway.
- The person may lack the energy necessary for the correct functioning of the body.

### How does NEONATAL-ONE work?

**NEONATAL-ONE** identifies and quantifies metabolites that are characteristic of a large number of genetic diseases known as “inborn errors of metabolism.” These compounds are identified on a filter paper soaked with a few drops of the baby’s blood obtained by a puncture on the baby’s heel. The filter with the dry blood is sent at room temperature in an urgent postal envelope. Once the laboratory receives the sample, it is analysed within 24 hours and the results are confidentially delivered to the doctor who requested the test.

### Who should undergo the NEONATAL-ONE test?

All newborns within 15 days of life.

### Is the test painful for the baby?

No. The procedure of collecting a few drops of blood from the newborn’s heel is a well implemented painless procedure that can be considered as a routine.

### How are the detected diseases treated?

Each disorder is different. Some are treated with a special diet and other with medications. If they are treated early, many children grow up and live a normal healthy life, preventing mental retardation or other serious secondary effects of the accumulation of substrates. In very few cases, it will not be possible to treat the complications completely.

Early diagnosis and treatment gives babies the opportunity to have the best development and growth.

### How can I obtain additional information?

By sending an e-mail to neonatal@genetadi.com

By calling +34-944044343

---

#### Other potentially detectable diseases

- **8-carboxilasa, deficiencia**
- **2,4-Dienoil-CoA reductasa, deficiencia**
- **Malat y aspartato reductasa, deficiencia**
- **Malat y aspartato reductasa, deficiencia**
- **Malat y aspartato reductasa, deficiencia**
- **Malat y aspartato reductasa, deficiencia**
- **Malat y aspartato reductasa, deficiencia**
- **Malat y aspartato reductasa, deficiencia**
- **Malat y aspartato reductasa, deficiencia**