Why is Newborn Screening important for my newborn?

Approximately the

3%-4%

of newborns will have a **genetic disease***.

- Some genes are responsible for inherited genetic diseases hat can result in metabolic and develop mental diseases that **cause serious health problems** from a nearly age.

- **Early detection** is key in order to be able to act and improve the patient's quality of life
  - Delaying or preventing the **appearance of symptoms** such as physical and intellectual disabilities
  - Helping to **improve the natural** evolution of the disease

*https://www.eurordis.org/IMG/pdf/princeps_document-EN.pdf

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Newborn Screening

by Igenomix®

In newborn health, early detection is key

Igenomix®
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We care about your baby's health

At Igenomix, we have taken neonatal screening one step further by developing Newborn Screening, an expanded genetic test that complements the heel prick test, with the aim of continuing to take care of your baby's health at birth.

The standard Newborn screening heel prick test is a mandatory public health program to check for certain disorders that may appear at birth.

Newborn Screening is indicated for all newborns. It is performed in healthy babies during the first days of life.

Newborn Screening is an expanded genetic test that analyzes **237 genes** linked to more than **200 genetic diseases** that **can be treated** at birth, or in the early stages of life.

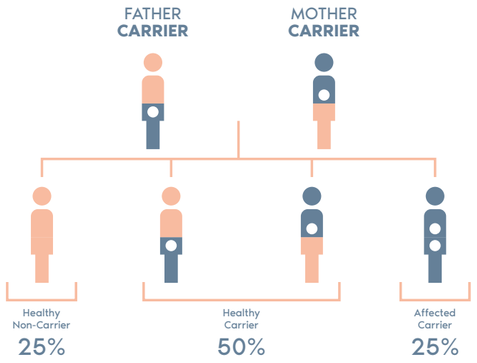
What information can I get from this test?

8 out of 10 children with **genetic disorders** are born **without a family history.** How is this possible?

This occurs because many of these diseases are transmitted in an **autosomal recessive inheritance pattern.**

Genes are responsible for giving **instructions** to our bodies and all of us have **two copies** of each gene. In these cases, when one of these copies suffers an **alteration,** the other continues to function normally. This individual would be considered a **healthy carrier.** When two carriers of the same condition have a baby, the baby has a 25% chance of being **affected,** 50% of **being a carrier** or 25% of being **free** of this alteration.

Thanks to **Newborn Screening,** it is possible to detect if your baby has inherited two altered copies of the same gene, which could lead to the development of diseases and therefore be able to anticipate treatment, hereby improving their quality of life.



Why have we selected these 237 genes?

The genes selected for Newborn Screening allow us to keep the focus on what is important, **prioritizing those serious diseases** that occur during **childhood** and for which we have sufficient **medical knowledge** and scientific evidence to **treat them and improve the prognosis** of the newborn.

Comparison of Newborn Screening vs. heel prick test

Disease Group	**Newborn Screening** +200 conditions	**Conventional screening** (Heel-Prick Test)*
Congenital errors of metabolism	✓	✓
Inmunodeficiencies	✓	✗
Endocrine diseases	✓	✓
Hemoglobinopathies	✓	✓
Neuromuscular diseases	✓	✗
Deafness of genetic origin	✓	✗
Lung diseases	✓	✓

*8-40 diseases, depending on the region

Referra Workflow

1. **Igenomix genetic counseling**
2. **You receive the kit at home. It should be brought to the hospital at the time of delivery.**
3. **The sample is taken at the time of the heel test.**
4. **Sending of samples to Igenomix**
5. **Igenomix performs the test and the analysis**
6. **Generation and review of the report**
7. **Results in 25 calendar days**
8. **Igenomix* Genetic Counseling**

*In case of positive result or doubts about the result.