

# NBS

Newborn  
Screening

by **Igenomix**<sup>®</sup>

In newborn  
screening,  
early detection  
is key



**Igenomix**<sup>®</sup>  
WITH SCIENCE ON YOUR SIDE

# At Igenomix we care about the health of your newborn baby

Newborn screening test is a mandatory public health program provided by the national health system that provides all newborns with testing of certain diseases that appear after birth.

Igenomix has developed an advanced newborn screening test that uses genetic testing to detect a wider range of diseases.

Igenomix Newborn Screening test uses the latest technology (NGS) **that allows a precise analysis of a larger number of diseases,** to prevent the onset of diseases in the newborn.



## Why do a Newborn Screening Test?



Around

**3% - 4%**

of newborns are affected by a **genetic condition.\***

- These genes are responsible for developmental, genetic and metabolic disorders that cause serious health problems starting in early childhood.
- An **early intervention** could prevent intellectual and physical **disabilities** as well as life-threatening **illnesses**.

\*[https://www.eurordis.org/IMG/pdf/princeps\\_document-EN.pdf](https://www.eurordis.org/IMG/pdf/princeps_document-EN.pdf)

# What is included and why?

The diseases included in the Newborn Screening have been selected given the following criteria:

- Diseases with **presentation in infancy**
- Diseases that have early
- Potentially **treatable and actionable**
- Diseases with **sufficient medical knowledge** and scientific evidence.



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## Why Igenomix Newborn Screening Test?

- Igenomix Newborn Screening Test is a comprehensive genetic test that **analyzes 237 genes** to reach a rapid, accurate diagnosis using Next Generation Sequencing (NGS) technologies.
- In addition, this test identifies if a child is a **healthy carrier** of any of these **genetic disorders**.

\* This study can provide, if desired, information on the carrier status of recessive diseases of the newborn. This status does not usually have clinical implications for the patient, but this information may be of interest to their parents in the face of family planning for a new pregnancy.

## Our new universal approach to expanded newborn screening using whole exome sequencing

Disease Group	Igenomix NBS (+200 conditions)	Conventional Newborn (Heel-Prick Test)
Congenital errors of metabolism	✓	✓
Immunodeficiencies	✓	✗
Endocrine diseases	✓	✓
Hemoglobinopathies	✓	✓
Neuromuscular diseases	✓	✗
Deafness of genetic origin	✓	✗
Lung diseases	✓	✓

## Who is the Igenomix NBS for?

**Indicated for all newborns.  
Performed as early as the first days of life.**

Early treatment is crucial to prevent complications and improve the prognosis for newborns.





# How does it work?



\*In positive cases or if there are questions regarding test results.



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