NBS Newborn Screening

Igenomix® WITH SCIENCE ON YOUR SIDE

In newborn screening, early detection is key

Around

3%-4%

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

Igenomix NBS identifies genetic disorders in newborn babies

Igenomix Newborn Screening is a genetic test that **analyzes 237 genes** linked to **more than 200 conditions.**

In addition, this test identifies a child **is a healthy carrier** of any of these genetic alterations.

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

https://www.igenomix.net/genomics-precision-diagnostic/newborn-screening/

1.

These genes are responsible for developmental, genetic and metabolic disorders that **cause serious health problems starting in early childhood.**

2.

The ultimate benefit is an early intervention to prevent intellectual and physical disabilities as well as life-threatening illnesses.

3.

This test allows the **detection of many more disorders** than with a conventional heel prick test.

Indication

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

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

www.igenomix.net

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

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What genes are included and why?

+ Infancy

Diseases with presentation in infancy.

+ Severity

Conditions with serious clinical repercussions, affecting the development of the newborn.

+ Actionable

Potentially treatable and actionable alterations.

+ Evidence

Genetic disorders with sufficient medical knowledge and scientific evidence.

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	✓	✓

Clinical utility of Igenomix Newborn Screening

Igenomix uses Next Generation Sequencing (NGS) technologies to perform NBS.

High clinical utility, by detecting +200 diseases.

Detect potentially treatable genetic conditions.

Allows an early clinical intervention of the baby.

Can avoid the development of symptoms, some of which may be irreversible.

Option for additional analysis and diagnosis at any stage of life.

Supports reproductive decisions and family planning.

Scientific advice for doctors and genetic counselling for patients, before and after the test.

Results: 25 calendar days

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Referral Workflow

1. Igenomix Genetic Counselling
2. Doctor/Patient receive the genetic kit
3. Sample collection: buccal swab
4. Samples shipped to Igenomix
5. Igenomix perform testing and analysis
6. Generation and review of the report
7. Results in 25 calendar days
8. Igenomix Genetic Counselling*

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

Sample type	Container	Transportation temperature	Volume
Buccal swab	Kit provided by Igenomix	20-25°C	According to the kit instructions

Contact your local Igenomix representative for more information about the test or email at **diagnostic@igenomix.com** and **+34 96 390 5310**

More information can also be found on our website **www.igenomix.eu**

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