# Postnatal screening test to identify early-onset conditions

Babynext®

CARING FOR LIFE

**Waiting for a child is a period characterized by excitement but also a lot of concern. Fortunately, many checkups during pregnancy, tests, instrumental investigations, and even genetic screenings in the prenatal phase allow to monitor and protect children's health.**

## Prenatal genetic screenings:

- Do not identify diseases so-called "de novo", which arise for the first time in the unborn child and are not transmitted by parents.
- Are not always extended to investigation of monogenic diseases
- Are not always preceded by parental survey (carrier screening test)

The healthcare system now applies nationwide expanded newborn screening.

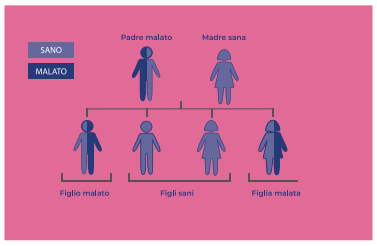
- 49 metabolic diseases
- Biochemical investigation only A blood sample taken
- From the baby's heel at the time of birth nascita.

# BABYNEXT® Caring for life: The added value of genetics

For several years, the scientific community has been showing interest in the use of sequencing techniques for newborn screening.

Babynext® CARING FOR LIFE

**New tool for prevention and health protection of newborn babies.**

| | | |
|---|---|---|
|  | NGS ANALYSIS | Technique |
|  | MORE THAN 280 | Detected conditions |
|  | 66 | Dominant Transmission Conditions |
|  | 12 | X-Linked conditions |
|  | MORE THAN 10 | Clinical areas of interest |

# Conditions detected by Babynext®

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| CLINICAL AREA | SOME OF THE CONDITIONS/DISEASES DETECTED BY THE TEST |
|---|---|
| Hematology | > Favism<br>> Beta Thalassemia<br>> Sickle Cell Anemia |
| Endocrinology | > Familial thyroid dyshormonogenesis<br>> Congenital hypothyroidism type 1<br>> Congenital adrenal hyperplasia from 3-beta-hydroxysteroid dehydrogenase deficiency |
| Gastroenterology | > Congenital defect of bile acid synthesis, types 1, 2 and 3 |
| Immunology | > Severe combined immunodeficiency from ADA deficiency<br>> Reticular dysgenesis<br>> X-linked agammaglobulinemia |
| Metabolic disorders | > Galactosemia<br>> Phenylketonuria<br>> Tyrosinemia |
| Nephrology | > Alport Syndrome<br>> Cystinuria A<br>> Distal Renal Tubular Acidosis |
| Oncology | > Retinoblastoma<br>> Predisposition to rhabdoid tumor type1 and 2<br>> Nephroblastoma or Wilms' tumor |
| Otolaryngology | > Pendred's syndrome<br>> Deafness with vestibular aqueduct enlargement<br>> Cystic Fibrosis |
| Syndromic | > Neurofibromatosis type 1 |

# Why Babynext®

Conditions analyzed by Babynext Caring for life are characterized by:

- **early onset**
- **high penetrance**
- **ability to take charge**

- Specifically, the survey in NGS conducted with Babynext Caring for life allows: to investigate preventable/treatable genetic diseases that cannot be identified by other methodology,
- To identify cases with mild variants and define ambiguous cases,
- To provide a fast and accurate diagnosis on infants not affected by parameters of prematurity, birth weight etc,
- Possible reduction of false positives and time (Tandem Mass Spectrometry - TMS on which SNE is based requires confirmatory testing).

# Benefits of Babynext®

**For the CHILD:**

- Schedule clinical audits
- Define controlled diets or dietary supplements
- Apply targeted treatment protocols

**For the FAMILY:**

- Early and effective taking charge
- Reproductive risk information for future pregnancies or on children already born
- Useful information for collateral risk assessment

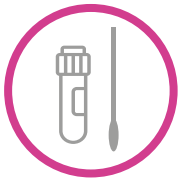
**For the SOCIETY:**

- Optimize health care costs

# WHOM IS THE TEST ADRESSED TO

It is recommended to take the test as **soon as possible for timely diagnosis and possible management of the child**. It is recommended for all children, from birth to 3 years old who do not show obvious symptoms

# How to perform the test



**How the test is performed**:
Simple buccal swab executable **even by yourself**

- Buccal swab
- Information form and informed consent
- Instructions for **collecting** storing and sending the sample
- Acceptance form Courier
- Pickup envelope

**Reporting times:**
45 working days.

**PRE- AND POST-TEST GENETIC COUNSELING INCLUDED**