RegioneEmilia-Romagna

SERVIZIO SANITARIO REGIONALE
EMILIA-ROMAGNA

Newborn screening

a simple test to protect the health of babies

Information for parents

What is newborn screening? What is it for? Is it important?

Every baby born in the Emilia-Romagna region is offered newborn blood spot screening. A blood sample is collected by pricking the baby's heel and squeezing out a few drops of blood onto a blood spot card for a simple screening test.
This allows to screen for health conditions that, if not detected early, may negatively impact the development of the baby.
Newborn screening is an important preventive medicine intervention which makes it possible to detect even very rare health conditions that affect one in 2,000 babies. In the event of positive diagnosis, adequate treatment can be provided during the first days of the baby at a clinical centre specialised in the health condition detected, where the necessary assistance will be offered to the baby and family. Genetic counselling is also offered to parents to assess risks related to any future pregnancy.

Newborn screening is important because it looks for health conditions that, if early detected, can be treated with very good results at specialised clinical centres.

Today the blood spot test is offered throughout Italy and it screens for over 40 health conditions, thus making it much more effective.

How is the blood sample analysed?

All blood samples collected from the hospitals where the babies are born in the Emilia-Romagna region and Republic of San Marino, as well as the ones collected from home birth, are sent to the Screening Centre Laboratory. Samples are kept at the Laboratory for at least 5 years to better protect the health of babies, while respecting personal data protection regulation.
Blood samples are analysed using different methods and the ones with altered values are identified. In this case, the test is repeated although some initial alterations are temporary and disappear over time.
Sometimes **a second blood spot sample is needed**. This may be due to technical reasons - insufficient blood sample - or to the presence of special conditions such as some mother's health conditions, diets or drugs, which make it necessary to repeat the test.

If you are called to repeat the test, it doesn't mean there's something wrong with the baby but that further checks are required.

Which conditions is the newborn screening for?

The blood spot test screens for conditions that belong to 3 groups: **endocrine disorders** (congenital hypothyroidism and congenital adrenal hyperplasia), **genetic disorders** (cystic fibrosis) **and metabolic diseases**.
These **conditions are rare** as they affect 1 in 2,000 babies, **congenital** (meaning they exist at or from birth) **and often inherited** (genetically transmitted).
If early detected through newborn screening, it is possible to start an immediate treatment **by administering hormones or enzymes to babies who cannot produce enough of them, or by treating them with an adequate diet or medicines to prevent damage caused by the build-up of harmful substances in the body before the onset of symptoms, resulting in a significant improvement of life quality.**

Metabolic diseases

Phenylketonuria (PKU)
Benign hyperphenylalaninemia (H-PHE)
Biopterin defect in cofactor biosynthesis (BIOPT-BS)
Biopterin defect in cofactor regeneration (BIOPT-REG)
Tyrosinemia, type I (TYR I)
Tyrosinemia, type II (TYR II)
Maple syrup urine disease (MSUD)
Homocystinuria due to CBS deficiency (HCY)
Homocystinuria due to MTHFR deficiency
Citrullinemia, type II (CIT II)
Citrullinemia, type I (CIT I)
Argininosuccinic aciduria (ASA)
Argininemia (ARG)
Carnitine uptake defect (CUD)
Carnitine palmitoyltransferase I (CPT-I) deficiency
Carnitine palmitoyltransferase II (CPT-II) deficiency
Carnitine acylcarnitine translocase (CACT) deficiency
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
Mitochondrial trifunctional protein (TFP) deficiency
Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
Medium/short-chain acyl-CoA dehydrogenase (M/SCHAD) deficiency
Glutaric acidemia, type 2 (GA-2/MADD)
Glutaric acidemia, type 1 (GA-1)
Isovaleric acidemia (IVA)
Beta-ketothiolase (BKT) deficiency
3-hydroxy-3-methylglutaric aciduria (HMG)
Propionic acidemia (PA)
Methylmalonic acidemia (Mut, Cbl A, Cbl B deficiency) (MMA)
Methylmalonic acidemia with homocystinuria (Cbl C, Cbl D deficiency) (MMA-HCYS)
2-methylbutyryl-CoA dehydrogenase (2MCD) deficiency
Multiple carboxylase deficiency (MCD)
Malonic acidemia (MAL)
Galactosemia
Biotinidase deficiency

Metabolic diseases diagnosed through differential diagnosis

Some disorders do not have all the features required to be involved in screening, but they share however some reference values with the conditions detected through screening. If these values are altered further checks are carried out to possibly detect them.

Tyrosinemia, type III (TYR III)
Glycine N-methyltransferase (GNMT) deficiency
Methionine adenosyltransferase (MAT) deficiency
S-adenosylhomocysteine hydrolase (SAHH) deficiency
3-methylglutaconic aciduria (3-MGA)
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency
2-methyl-3-hydroxybutyryl-CoA dehydrogenase (MHBD) deficiency
Isobutyryl-CoA dehydrogenase (IBD) deficiency
Short-chain acyl-CoA dehydrogenase (SCAD) deficiency

Key Information to remember:

- Newborn screening is an important preventive medicine intervention that can significantly improve the quality of life of babies with a positive diagnosis.
- Sometimes a second blood spot sample is needed but it doesn't necessarily mean there's something wrong with the baby.
- Often, despite a second blood spot sample, the screening result is negative. This creates serious emotional distress for the family, but it is however motivated by some rare cases in which the result is positive and the baby can be immediately supported.
- **Your paediatrician** will be your primary contact person for your child's care. In the event of diagnosis of a condition through newborn screening, the diagnosis and treatment plan will be coordinated by the Clinical centres for endocrine disorders, inherited metabolic diseases and cystic fibrosis in close cooperation with the paediatrician and the family.
- The conditions involved in newborn screening are associated with codes that entitle patients to co-pay fee (ticket) exemptions.

From newborn screening test to assistance

- Within 48-72 hours from birth (at the hospital where the birthing centre is located) a blood sample is collected by pricking the baby's heel and squeezing out a few drops of blood onto a blood spot card.
- The personnel of the birthing centre send off the blood spot card to the Newborn Screening Laboratory via dedicated transport.
- The card reaches the Lab within 24/48 hours from sample collection.
- If the test result is positive, meaning the values tested are altered, the Regional Newborn Screening Centre will contact the birthing centre to recall the baby and forward the family to the reference Clinical Centre and agree on further checks.
- If the diagnosis of congenital disorder is confirmed, the family is contacted immediately and the process of taking charge of the baby by the reference Clinical Centre is organised.
- The reference Clinical Centre welcomes the baby, defines the clinical-care path and plans the checkups; it works in close cooperation with the paediatrician and supports the family throughout the process.
- The reference Clinical Centre certifies and records the condition in the regional registry of rare diseases and summary data is sent to the national registry of rare diseases in compliance with the data protection regulation.

Regional centres

Regional Newborn Screening Centre

• Newborn screening laboratory

S. Orsola-Malpighi Polyclinic,
Padiglione 20, Via Massarenti 9,
Bologna University Hospital Authority

• Clinical centre

S. Orsola-Malpighi Polyclinic's Paediatrics Care Unit,
Padiglione 16, Via Massarenti 9,
Bologna University Hospital Authority

Regional Hub Centre for inherited metabolic diseases

S. Orsola-Malpighi Polyclinic's Paediatrics Care Unit,
Padiglione 16, Via Massarenti 9,
Bologna University Hospital Authority

Neonatal and paediatrics care unit,
Guglielmo da Saliceto Hospital, Via Taverna 49,
Piacenza Local Health Authority

Regional Hub Centre for endocrine disorders

S. Orsola-Malpighi Polyclinic's Paediatrics Care Unit,
Padiglione 16, Via Massarenti 9,
Bologna University Hospital Authority

Regional Hub Centres for cystic fibrosis

CF centre of Parma University Hospital Authority,
Via Gramsci 14

CF centre of Romagna Local Health Authority,
Bufalini Hospital, Cesena, Viale Ghirotti 286

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