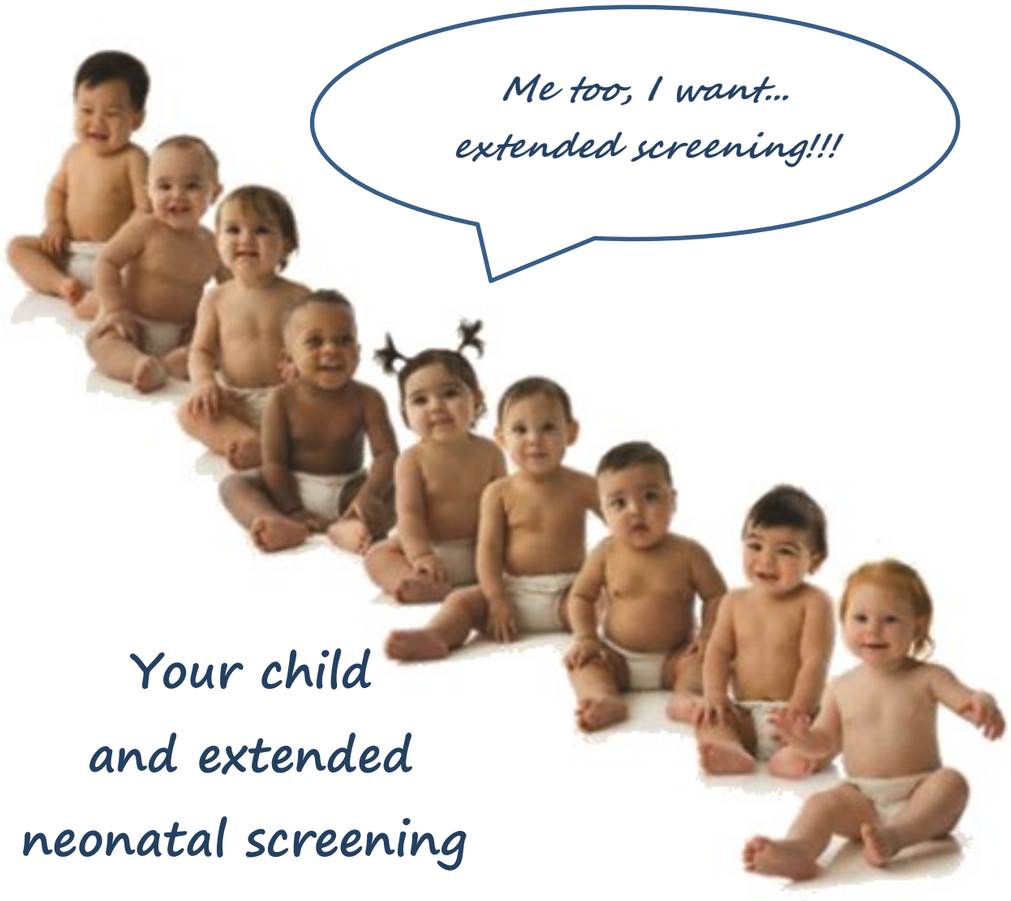

Your child and extended neonatal screening

SISTEMA SANITARIO REGIONALE
REGIONE MARCHE

AZIENDA OSPEDALIERA
OSPEDALI RIUNITI MARCHE NORD

CENTRO SCREENING NEONATALE
REGIONE MARCHE

Dear parents,
This leaflet seeks to explain why it is important for your son or daughter, when between 48 and 72 hours old, to undergo screening for early detection of diseases that could have serious consequences for his or her development.

WHAT IS NEONATAL SCREENING?

As part of preventive medicine programmes, all newborns can undergo testing for free to allow early detection of congenital diseases. Under National Law No. 104 of 05/02/92, compulsory neonatal screening for Phenylketonuria, Congenital Hypothyroidism and Cystic Fibrosis has been in place for some time.
With Decree of the Regional Government (DGR) No. 918/2013, the Marche Regional Government added extended metabolic screening as of 2014 by screening for another 40 metabolic disorders with the use of an innovative technology, based on Mass Spectrometry, which identifies genetically transmitted metabolic diseases, many of which are among the rare diseases. This test panel, together with screening for **Galactosaemia and biotinidase deficiency**, was made compulsory throughout the country with Law No. 167 of 19/08/16.

WHY UNDERGO NEONATAL SCREENING?

The aim of the Neonatal Screening is to identify some congenital diseases early, before the onset of symptoms. Although the likelihood of your child suffering from one of the diseases researched through screening are very low, early diagnosis allows your child to be treated sooner, ensuring his or her proper development and optimum health. **A simple test can prevent the effects of a disease that might not be recognised otherwise.**

HOW IS THE SCREENING CARRIED OUT?

It is a small prick in the heel of the newborn, made with a sterile needle, performed on the maternity ward, between 48 and 72 hours after birth, before discharge. The child will barely realise it is happening but the few drops of blood collected onto a special absorbent paper are enough to search for metabolic diseases. The blood is sent to the Regional Screening Centre of Santa Croce Hospital, Fano, where the testing is performed. In the event that the tests are abnormal, the specialists from the Birthing Centre and family members are contacted.

WHEN DO PARENTS FIND OUT THE RESULTS?

Negative results are not disclosed; therefore, if you are not called, this means that all tests are normal. If your child tests positive for one of the diseases investigated, family members will be called from the Birthing Centre or the Screening Centre for further testing.

- **Remember: if screening is negative, you will not receive any notification.**
- **Being called to give a new sample does not necessarily mean that your child is ill.**

WHAT ARE THE DISEASES INVESTIGATED BY NEONATAL SCREENING?

PHENYLKETONURIA (PKU) is due to a hereditary defect that causes accumulation of a protein component – phenylalanine – in the body. The increase of phenylalanine in the blood of the child, if left untreated, causes a developmental delay. Treatment consists of a diet low in protein (meat, fish, milk, eggs, etc.) and allows for normal growth and development.

INCIDENCE In Italy, phenylketonuria affects one child in every 3,700 births. In the Marche Region, screening for this disease was started in 1973.

CONGENITAL HYPOTHYROIDISM (CH) is due to insufficient production of thyroid hormones by the thyroid gland. These hormones are essential for the growth and mental development of the child. Treatment consists of taking the deficient hormone, thyroxine, by mouth daily. This ensures normal intellectual and physical development.

INCIDENCE In Italy, Congenital Hypothyroidism affects one child in every 1,000 births. In the Marche Region, screening for this disease was started in 1982 .

CYSTIC FIBROSIS (CF)is due to an inherited defect that causes the anomaly of a protein that regulates the movement of salt in and out of cells. This causes the accumulation of viscous secretions and high levels of salt in the sweat. The classic form affects the respiratory system and gastrointestinal tract, with repeated infections and poor growth. In a very small number of children – about 2% of all born – screening may also involve DNA analysis. If you do not want any diagnostic study with genetic testing (DNA), please notify the staff in writing. In this case, the testing for cystic fibrosis will be done in ways that do not involve genetic investigation. Diagnosis by neonatal screening, and before the onset of symptoms, can prevent the occurrence of some complications with a major improvement in the clinical outcome. ***INCIDENCE In Italy, Cystic Fibrosis affects one child in every 3,400 births. In the Marche Region, screening was started in 1995*** .

GALACTOSAEMIA is due to an inherited defect that causes the accumulation in the body of a sugar, galactose, which is derived from the metabolism of carbohydrates. This accumulation has toxic effects on various organs and systems (in particular, the nervous system and eye). Treatment consists of a galactose-free diet. If instituted early, this allows for a significant improvement in mental and physical development. ***INCIDENCE In Italy, it is estimated in about one baby in every 25,000. In the Marche Region, screening was started in 2019.***

BIOTINIDASE DEFICIENCY is due to an inherited defect that causes deficiency of biotin (vitamin B8), a nutritional disorder that if left unchecked or untreated can become a very serious condition. Biotin participates in metabolic reactions including the synthesis and metabolism of energy substances. Symptoms range from skin reactions, rash and alopecia to neurological symptoms (seizures, ataxia), developmental delay, loss of eyesight and hearing. Treatment consists of a biotin supplemented diet. If initiated early, these complications can be prevented.

INCIDENCE The frequency in Italy is estimated at about 1 infant in every 60,000. In the Marche Region, screening was started in 2019.

EXTENDED METABOLIC SCREENING

This involves testing for about 40 rare metabolic diseases that can be classified into the following three groups:

- Amino acid metabolism disorders;
- Organic acid metabolism disorders;
- Fatty acids oxidation disorders.

INCIDENCE In Italy, these three classes affect about one child in every 25,000 births. In the Marche Region, screening was started in 2014.

It is important to remember that for general concerns,
you must contact your family paediatrician.
In case of a positive result in a screening test,
the Regional Centre for Neonatal Screening will coordinate all the diagnostic and treatment pathway in collaboration with you and your child's paediatrician.

YOUR FAMILY PAEDIATRICIAN

Contact information for the Regional Centre for Neonatal Screening
Child Neuropsychiatry
North Marche Ospedali Riuniti Hospital Trust
“Santa Croce” Hospital
Via Veneto, 2-61032 Fano (PU)
Laboratory tel: 0721 882401
Department tel: 0721 882419
Fax 0721 882414 – 0721 882401
Email address
screeningneonatale@ospedalimarchenord.it

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