NEWBORN SCREENING TESTS AND YOUR CHILD
A Parents' Guide

Dear Parents, the Region of Tuscany is giving the opportunity for your baby to avail of a newborn screening programme free of charge.

What is Newborn Screening?

In the field of preventative medicine programmes all newborn babies undergo simple tests, free of charge, that allow us to detect several congenital illnesses early-on. National legislation 104 05/02/1992 called for the screening of Phenylketonuria, congenital hyperthyroidism and cystic fibrosis. In 2004, the Region of Tuscany introduced screening for over forty different metabolic disorders. National legislation 167/2016 made such tests obligatory for all newborns. The regional resolution 909/2018 extended screening to include three other illnesses, from Lysosomal Storage Disease to Severe Combined Immune Deficiency disorder.

Why do Newborn Screening?

The objective of Newborn Screening is the early detection, prior to the appearance of symptoms, of several congenital illnesses, and thus to prevent or limit the serious typical damage that such illnesses can cause and guarantee a good quality of life through the timely introduction of specific therapies.

A simple test can save a life

How is it done?

Drops of blood that are taken through a heel prick test on the newborn baby between 48 and 72 hours after birth, are analysed. The drops of blood are placed on a special piece of absorbent paper that is attached to a card that contains all the baby's details. For babies who weigh less than 1.8kg at birth, this test needs to be carried out at 48 hours from birth and also at 14 days and 30 days from birth. The sample is sent to Meyer University Hospital where analysis is carried out for all infants born in Tuscany. The hospital conserves the blood drop sample for a period of ten years.

When do you get the results?

If the child results to be positive for one of the illnesses that being investigated, he/she will be called to go to the hospital where he/she was born or to the screening centre for further tests. Normal tests results are not communicated to the parents so if you are not contacted this means that the test results were negative.

Important: if you are contacted, this does not mean that your baby is ill, but only that further tests need to be carried out.

What illnesses are identified during the screening process?

Phenylketonuria and other metabolic illnesses

Phenylketonuria was the first metabolic illness to be tested during the newborn screening process. It is caused by a congenital defect in an enzyme that controls the build up of Phenylalanine, a protein component, in the body. A build up of Phenylalanine is toxic for the brain. Treatment consists of a diet that is low in this particular substance and that allows for cognitive development and normal growth. In some cases pharmaceutical treatment is used. Apart from Phenylketonuria, the screening process can identify more than 40 metabolic disorders by means of a complex technique called Mass Spectrometry (MS). Metabolic disorders are a large group of hereditary illnesses caused by genetic metabolic defects. Symptoms can appear even during the first days of life, but more often during the first year of life or later on, even as an adult.

These illnesses, if not treated properly, can effect various organs and apparatus, such as the central nervous system, the heart, the liver, the kidneys, the skin etc. In some cases, they can be the cause of Sudden Infant Death Syndrome (SIDS).
The ability to identify these illnesses in affected babies at an early stage, prior to the onset of symptoms, can allow for a therapeutic diet and/or medication to improve the prognosis and quality of life on a long term basis.
The metabolic disorders that are tested for during the newborn screening process are: defects in amino acid metabolism, organic acid disorders, urea cycle defects, fatty acid beta-oxidation disorders (Frequency: about 1 infant in every 2,000)

Biotin Deficiency
This is a congenital defect of the metabolism of a specific vitamin, biotin, that leads to multiple carboxylase deficiency. The symptoms can vary and can include psychomotor developmental delays, convulsions, immune disorders and skin problems. Screening tests the activity of the biotin vitamin. Treatment involves the oral administration of biotin.

Congenital Hyperthyroidism
This is caused by the absence of, or insufficient production of, thyroid hormones that are indispensable for brain development and maturation and for normal growth in a child. Treatment involves the oral administration of thyroxine. Early diagnosis and treatment results in normal child development. (Frequency: about 1 infant in every 1,500).

Galactosemia
This is caused by a hereditary defect that provokes the accumulation of galactose in the body. Galactose is a substance that derives from the metabolism of carbohydrates and has a toxic affect on various organs like the liver and the eyes. Early treatment through a galactose-free diet results in a significant improvement in the child's psycho-physical development.

Cystic Fibrosis
This is caused by a genetic defect that can cause, in very different ways for each person affected, growth-related problems and breathing difficulties. The screening test is based on trypsin dosage. An altered result is not uncommon so a full interpretation requires further tests. Diagnosis of this illness in a newborn baby, prior to the onset of symptoms results in the prevention of various complications and improved clinical progress; it also allows for the genetic consultation of the family in question. Today, apart from the symptomatic treatments available, there are effective medications for a growing number of children. (Frequency: about 1 infant in every 4,000).
In the screening process for cystic fibrosis it is possible that, based on the results and in a very limited number of children, that further genetic testing is required. You will be asked for your consent in this regard at the time of the heel prick test.

Lysosomal Storage Diseases (LSD)
LSDs are caused by a genetic defect of lysosomal enzymes with an accumulation of substances in the lysosomes in organs and body tissue. They are progressive diseases and can cause serious disability or death at an early age. More than 50 LSDs have been identified and they are are all extremely different in character in terms of the age of onset, symptomatology, clinical progress and seriousness, even within the same illness or enzymatic/genetic defect. There are three diseases included in the screening process, by means of an enzyme dose blood test, and they are as follows: Pompe Disease, Fabry's Disease and Mucopolysaccharidosis type 1. Enzyme Replacement Therapy has changed the natural progression of these diseases and has modified the quality of life and life expectancy of patients. In the case of diagnosis through newborn screening, the starting phase for therapy can vary based on the enzymatic/genetic defect and the clinical phenotype.

Pompe Disease
It is caused by a deficiency of lysosomal acid α-glucosidase which leads to an accumulation of glycogen in the cardiac muscle, skeletal muscle and smooth muscle. The symptoms of Pompe Disease vary based on the age of onset (infancy, childhood or adulthood), type of progression and the seriousness of muscular involvement.

Fabry's disease

It is caused by a deficiency of the galactosidase enzyme which leads to the accumulation of glycosphingolipids, especially in the kidneys, heart and nervous system causing kidney failure, cardiomyopathy or a stroke. Newborn screening in general does not allow for diagnosis in baby girls.

Mucopolysaccharidosis type 1
It is caused by a deficiency of the - iduronidase enzyme which leads to the accumulation of mucopolysaccharides, especially in the liver, the bones, the eyes and the nervous system. In some cases, an early hematopoietic stem cell transplant could be recommended.

Severe Combined Immune Deficiency (SCID)
Severe immune deficiency diseases are a vast group of rare illnesses (today, we are aware of over two-hundred of these) that are all characterised by a defect in the immune system. Babies with Sever Combined Immune Deficiency (SCID) are born apparently healthy. However, precisely due to the grave defect in the immune system which does not protect them from infectious diseases, newborn babies can suffer serious, irreversible damage from an early age or even die due to infections which are no problem to babies who possess a normal immune system. Newborn screening allows for the diagnosis of SCID in the first days of the baby's life and so, in cases where a defective immune system is suspected, treatment can be started to protect against all possible infections. Screening is carried out by looking for molecules called TREC in the baby's drop of blood. TREC (T cell receptor excision circles) are small molecules that are produced during T-cell development and maturation and they are fundamental for the proper functioning of the immune system.

If a diagnosis of a congenital illness is confirmed, the baby and his/her parents will follow a treatment programme in coordination with Meyer University Hospital, in collaboration with regional Maternity wards, the Family Pediatrician and Clinics that are specialised in the treatment of specific diagnosed pathologies.

The data owner of the newborn screening programme is Meyer University Hospital – Florence. For more information on how information is handled, it is possible to consult the relevant section on the hospital website: www.meyer.it

For more information

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