

Screening tests for newborns



- ☆ Genesis would like to inform new parents about the innovative program of screening tests for newborns.
- ☆ Screening tests started with the clinical examination of hearing, vision and the ultrasound scan of hips, to follow latter on with the screening test for 45 metabolic diseases, such as the fibrocystic disease
- ☆ The benefit of these screening tests is to diagnose in early stages congenital abnormalities, which usually do not present any symptoms in infant age but will eventually give symptoms in the future. Early diagnosis can lead either to an immediate treatment or to the adoption of the appropriate therapeutic regime, limiting in this any possible risky complications.
- ☆ Congenital abnormalities which are detected through the screening tests programs are present in very low percentages, however not negligible for the level of medical services Genesis seeks to provide.
- ☆ The results of clinical tests are recorded in the child´ s health book and parents are informed only in case of pathological findings. With regard to the results of the metabolic diseases tests, these are posted to the contact address you stated during your admission in the Clinic.
- ☆ Please take some time to read the rest of this brochure, where you will find more information about the Genesis screening tests for newborns. These procedures are performed by specialized medical personnel and laboratories, under the scientific supervision of the Clinic's Neonatal Department and according to the international scientific developments. The Head Midwife in your floor will pass you a concession form to fill on the previous mentioned tests.
- ☆ In case of questions, please feel free to consult a Pediatrician • Neonatologist of our department.



Hearing Test

■ What is a screening hearing test?

During the last few years painless and safe methods have been developed, such as otoacoustic emissions to check the hearing system of a newborn. The method of otoacoustic emissions allows hearing examination from the very first few days following birth. In many advanced countries, such as the USA or Germany, newborns are submitted to this hearing test before their discharge from the maternity hospital, since the early 90s.

■ How this hearing test is performed?

The method applied is called "recording of otoacoustic emissions". It is a painless and quick method, performed with special equipment by specialized Otolaryngologist (ENTs). During the examination, a very small tube with an integrated sounder is placed inside the infant's ear, sending specially arranged sounds to the inner ear (cochlea). The sounds produce an impulse in the cochlea cells, thus leading them to produce a special signal, which is recorded and analyzed. Results come out immediately.

■ How significant are the results?

The test results are characterized either as "normal recording of otoacoustic emissions" or as "non normal recording of otoacoustic emissions". In the latter case there is an indication for further examination concerning the hearing ability and our ENT will inform you on the following necessary steps.



■ Should screening hearing tests be performed in all newborns?

Natural hearing from the very first months of life is a basic prerequisite to understand speech and language. If a baby presents any difficulty in hearing, then the normal course of his / her lingual and speaking development is significantly disturbed, with consequences not only on the child's socialization but even his / her school performance.

Ophthalmologic Test

■ What does the eye examination assess in newborns?

Any possible anatomic abnormalities of the eyeball, opacities in cornea and lens and the fundus oculi. In addition, this examination assesses also any possible conditions which could restrain newborn's good eyesight. Indicatively, damages as congenital cataract, persistent hyperplastic primary vitreous, congenital glaucoma could be diagnosed early and can be treated.

■ What is necessary for the examination?

A prerequisite for the ophthalmologic screening is the mydriasis. This takes place with infusion of collyrium 30 minutes before the test. The test is performed in the infant's cradle, by a specialized oculist. This procedure is painless and lasts a few minutes.



■ How can I tell if my baby can see?

Vision is a sense which develops after birth and a prerequisite for its correct development is to make sure that the light can reach the back part of the eyeball, the retina. During the ophthalmologic screening we check whether there's any factor preventing light from reaching the retina. This test cannot fully diagnose the operating condition of the eye, as this depends also on other factors as well which can be altered after the birth and during the child's life, until the optical system matures.

Ultrasound Hip Test

■ What does the ultrasound hip test check for in infants?

One of the most common orthopedic conditions we see in newborns is the development of hip dysplasia. This can range from a mild limpness of the joint to a severe deformation, as when the head of the thigh bone has no contact with the acetabulum and is in dislocation position.



■ **Why is the early diagnosis of hip dysplasia important?**

Early assessment of a pathological condition in the hips is very significant in order to select the right kind of treatment and to achieve a good therapeutic outcome. Late hip dislocation diagnosis could lead towards an invasive treatment, when an early diagnosis can be dealt conservatively, such as with the placement of special splints.

■ **What is the diagnostic value of ultrasound scans?**

The ultrasound test of hips completes the clinical examination of the Neonatologist, in ages less than 6 months, where X-ray tests present serious disadvantages. The ultrasound hip screening has high diagnostic credibility, does not burden the newborn with radiation, can be performed from the first few hours after birth and is a significant aid in the doctor's diagnostic work.

■ **Which infants are submitted to ultrasound hip screening?**

It is recommended to perform the ultrasound hip scan to all newborns with high risk to develop any kind of hip dysplasia. Newborns born with breech presentation or those with positive medical history of congenital hip dislocation are candidates for this screening assessment.

Metabolic Diseases Examination

■ **What is the screening for Metabolic Diseases and why is it useful?**

This blood screening test uses mostly the Tandern MS (mass spectrometry) technology and allows the rapid detection of many metabolic diseases. These diseases may be initially non symptomatic, however in the future will lead to the clinical expression of the underlying disease. Furthermore, it is possible to detect also the carriers of a metabolic disease. These newborns do not present any symptoms, nevertheless they can transfer a metabolic disease to their offspring. This screening test can reveal these carries, even when there is a negative family history for hereditary diseases.

■ **When and how is the blood sample collected?**

When the baby is 2 to 3 days old, we collect a few drops of blood from the infant's heel on a special card, the Guthrie card. This procedure is performed at the same time with the collection of blood for the standard test of 4 metabolic diseases (phenylketonuria, congenital hypothyroidism, galactosemia and G6PD deficiency). The latter one screening test is obligatory and is performed as part of the corresponding national detection program. In premature newborns, the time of examination is the same, as the aim is to check the infant as soon as possible.

■ **Is there a case where the examination may need to be repeated?**

Re-examination is rarely required. In case the sample is unsuitable or the result is non-conclusive, you will be informed as soon as possible in order to collect a new sample.

■ **What diseases or groups of diseases are detected?**

The program may detect more than 40 diseases such as:

- ☆ Aminoacids disorders, organic acidurias, disorders of fatty acids metabolism (b-oxidation)
- ☆ Fibrocystic disease

In case of suspicion, definite diagnosis is made further to complementary special tests.

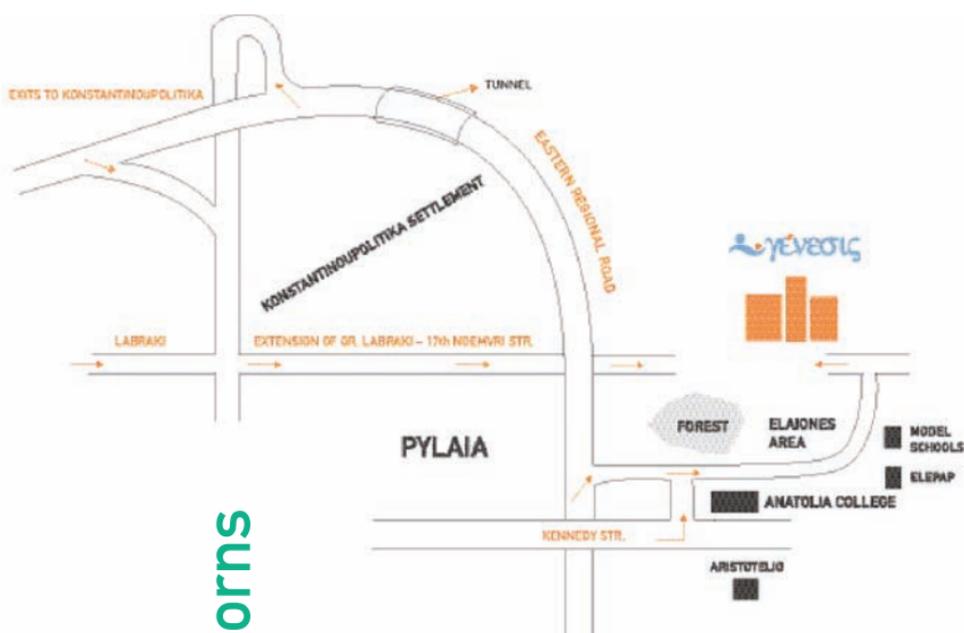
■ **Is there a cure for these diseases?**

Congenital diseases which are assessed can be treated symptomatically and early diagnosis will allow the best possible course of the diseases, in relation with the existing abilities offered by the medical science.

■ **How will I know about the results?**

One week after the sample collection, results are posted via mail by the scientific team performing the tests in collaboration with the Clinic. In case of a delay or any other problem, please let us know.





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