The Neonatal Screening Program - Useful Information and FAQs

CONGENITAL HYPOTHYROIDISM

What is congenital hypothyroidism?

This is a disorder in the neonate whereby the thyroid gland either doesn't function well enough or not all, resulting in the under production or total absence of the all-important hormone, thyroxine. In Cyprus we have estimated that 1 baby in every 2000 born have congenital hypothyroidism.

How exactly is congenital hypothyroidism screened for?

The most efficient way to determine whether a child has this disorder is to check for the gland's functionality within the first days of life. Following the baby's first 48 hours after delivery, before being discharged from the birth clinic or hospital, or during the baby's first outpatient visit to the pediatrician, several drops of blood from the baby's heel are taken and applied onto a special absorbent blood collecting card. It is important that the blood sample is taken by the 5th or 6th day of life and sent to the Center for Preventive Paediatrics without delay. Upon receipt, the Center's clinical lab tests for the levels of the Thyroid Stimulating Hormone, (TSH) also known as thyrotropin in the baby's blood collected on the card. In the event that congenital hypothyroidism in the child is a possibility/indicated/suspected following screening, the Center informs the referring paediatrician for any further action that needs to be taken so as to lead to a diagnosis.

I have been told my child is hypothyroid. What exactly does this mean?

Untreated hypothyroid babies tend to be lethargic with decreased activity. They often have difficulty in passing stools and their skin feels cold and dry to the touch. Learning is delayed and their development is slow often leading to stunted growth. Evidently, hypothyroid children are deficient in growth and development in comparison to their non-hypothyroid counterparts. Most importantly, when hypothyroidism is present before the brain has fully developed, the child's cognition and mental development is irreversibly affected. As the years go by, their untreated condition may lead to more severe symptoms whereby their skin gathers a substance causing generalized swelling of their face and body. This condition is known as Myxedema and can be life threatening. It is of paramount importance that a child who has been diagnosed early on in life with hypothyroidism is placed on exogenous thyroxine therapy so as to develop normally and avoid all the serious complications of the disorder.

If my child is hypothyroid, will all the serious symptoms of the disorder manifest?

No. From the moment hypothyroidism is diagnosed early on in life via the *Neonatal Screening Program* and the administration of intervention therapy has begun, then the symptoms will not arise and the child's mental and physical development will be normal.

What causes a child to be born with congenital hypothyroidism?

The causes of hypothyroidism diagnosed in the neonate is still unknown in the majority of cases. Only a small proportion of incidents are hereditary whereby the mutant gene is inherited from both parents.

Medication administered to the pregnant mother for her thyroid conditions or overuse of antiseptics containign iodine may also bring on a transient type of hypothyroidism in the neonate. With the correct caution in the use of these products during pregnancy, such incidents in the neonate can be easily avoided.

The chances of parents of a hypothyroid child having another child with the same condition is actually negligible. In fact, their chances are almost comparable to a couple without a child diagnosed with congenital hypothyroidism.

Is therapy difficult to administer?

No. However, it is absolutely necessary that your child is given his/her assigned dose of thyroxine without fail. Thyroxine tablets can be ground to a powder and given to your child in his/her milk or creamy solids. Periodically, your paediatrician will take a blood sample from your child to check whether the administered dose of thyroxine needs adjusting. The Center for Preventive Paediatrics provides a follow-up sampling schedule to all parents to indicate when blood needs to be sent to the Center to check for this. The frequency of blood samples to be taken is higher during the child's first year of life. In general, the follow up period after the child's initial diagnosis of hypothyroidism is coordinated by the Center in association with its medical collaborators/coworkers.

How long does the therapy last for?

Usually throughout their whole lives. However, some children suffer from a transient form of the condition and will be able to come off their therapy once this has been proven. This will be assessed from the age of 2 onwards. Specialized tests will be carried out to assess whether your child will need to take exogenous thyroxine for life. Whatever the case, the therapy is straightforward, cheap and easy to administer, without side effects or other drawbacks for your child.

How will I know my child is developing normally?

The Center for Preventive Paediatrics offers standardized psychometric testing to all children diagnosed with congenital hypothyroidism up until the age of 7. These tests are given so as to assess the child's physical and cognitive development.

Always remember:

The etiology of the majority of congenital hypothyroid cases are unknown. There is therefore, nothing that could have been done to have avoided your child's condition.

However, if the diagnosis of this condition and intervention therapy all occur in a timely manner soon after the baby's birth, then his/her physical and cognitive development should be no different to any non-hypothyroid child.

Never interrupt your child's therapy, even if they seem to be developing normally. This is owed to the exogenous thyroxine your child has been administered. If you stop then you risk the symptoms of hypothyroidism manifesting.