



# Congenital hypothyroidism

**This leaflet explains about congenital hypothyroidism and how it is treated at Great Ormond Street Hospital.**

## What is congenital hypothyroidism?

This is a disorder affecting the thyroid gland, which is in the neck. The thyroid gland produces a hormone (chemical substance) called thyroxine, which is needed for normal growth and development as well as a normal metabolic rate. If the thyroid gland does not produce enough thyroxine, it causes hypothyroidism. If the disorder is present at birth, it is called congenital hypothyroidism.

## How is congenital hypothyroidism diagnosed?

In the UK, all babies are tested for congenital hypothyroidism soon after birth, using a tiny amount of blood taken from pricking their heel (Newborn Bloodspot test). If this test shows that your baby possibly has hypothyroidism, it will be recommended that he or she have further blood tests to confirm the diagnosis.

He or she will also have a special scan of the neck that allows doctors to see if your child's thyroid gland is present and in the right place (explained below). The scan is painless and uses a special intravenous marker that is only taken up by the thyroid gland. This is extremely useful information as it allows us to tell you whether there is a chance of the condition happening in another child that you might have.

## What are the symptoms of hypothyroidism?

Most babies with congenital hypothyroidism are diagnosed very early, before they have any symptoms. If these signs and symptoms are present, they may include feeding difficulties, sleepiness, constipation and jaundice (the skin may look yellow).

It is very important that the above blood tests are carried out soon after the heel prick blood tests are known. This is because if congenital hypothyroidism is not diagnosed and treated soon after birth, it can cause problems with mental development, learning and clumsiness.

## What causes hypothyroidism and is it inherited?

During the early months of pregnancy, when your baby's organs are developing, the thyroid gland moves from the back of the tongue to its normal position in the neck. In some babies this does not happen, which means that the gland cannot work properly. On other occasions the thyroid gland does not develop at all. If you have one child with either of these types of congenital hypothyroidism, the chance of having another baby who is also affected with congenital hypothyroidism is very low.

There is another very rare type of hypothyroidism in which a child's thyroid gland is in the right place, but it cannot



produce thyroxine. This type is inherited and so there is a risk that if you have another child in the future they may have the same condition. If you are worried, please talk to your Hospital doctor about this.

Finding out the location of the thyroid gland is important as this will help confirm if the problem with the thyroid gland is lifelong or not, but also whether or not there is a chance of the condition happening in another child that you might have.

## **How common is congenital hypothyroidism?**

In the UK, around one in every 3500 newborn babies have congenital hypothyroidism. It is more common in girls than boys, but at the moment we do not understand why.

## **How is congenital hypothyroidism treated?**

Congenital hypothyroidism is treated by replacing the thyroxine that the body cannot produce. This is usually in liquid form (as levothyroxine oral solution) but tablets can also be used. The medicine only needs to be given once a day. Although missing an occasional dose will not cause any immediate problems, it is best to try to make sure that your child takes their medicine regularly each day and therefore keeps a steady level of thyroxine in their blood.

## **Does the treatment have any side effects?**

As levothyroxine medicine is simply replacing a normal chemical produced by the body, giving the correct dose every day should not have any side effects. However, if your child is not given enough levothyroxine, they will develop the symptoms of hypothyroidism outlined earlier, and over a long period, may grow more slowly than usual. If your child has too much levothyroxine, he or she may develop mild diarrhoea, may be more

restless than usual and will not put on weight. However, as described above, the correct dose for your child will be calculated on a regular basis, so these effects are unlikely to occur.

## **Other investigations**

Your baby may also have a special scan (thyroid technetium scan) of the neck that allows us to see if your child's thyroid gland is present and in the right place. In order for the special x-ray camera to pick up the image of the thyroid gland, a weakly radioactive marker is injected through a cannula and is taken up by the thyroid gland. The cannula is inserted into a vein in the back of your baby's hand or inner elbow by skilled nursing staff on Kingfisher Ward. The scan is done in the Nuclear Medicine department and takes about 45 to 60 minutes to complete. The scan is done only once.

For a very small number of children born with hypothyroidism, hearing problems can occur as part of a rare syndrome (collection of symptoms often seen together) or if there is severe hypothyroidism at birth. For this reason, all young children coming to GOSH for diagnosis and treatment of congenital hypothyroidism will have a detailed hearing assessment at about six weeks of age. This will still need to be done even if your baby has passed the neonatal hearing assessment. Please remember that it is extremely rare for serious hearing problems to occur as a result of congenital hypothyroidism. However, it may still be necessary for your child to be monitored by the audiology department for some months.

## **Congenital hypothyroidism and future pregnancies**

If your baby has no thyroid gland or the gland is at the back of the tongue, the chance of having another baby who is also affected with congenital hypothyroidism is very low, but your baby will usually need life-long medication.



When hypothyroidism is present, but the child's thyroid gland is in the right place, there is a risk that if you have another child in the future they may have the same thyroid problem. At present, it is difficult to identify the potential genes that cause the various types of congenital hypothyroidism. However, the team at GOSH is beginning to look at very specific groups of children with congenital hypothyroidism, so parents may be asked about having a blood sample taken and stored for future genetic testing. If you are worried, please talk to your specialist nurse or doctor, and if appropriate, you will be referred to a geneticist.

## Monitoring

For the first couple of years, your child will need regular blood tests to check thyroxine concentrations and regular outpatient appointments at GOSH to check growth, development and levothyroxine doses. The blood tests can be done easily in Outpatients at GOSH or sometimes at your local hospital. You should discuss with your Clinical Nurse Specialist or Consultant which is most appropriate for your child. The Endocrine team will use the information from these tests to work out the right dose of levothyroxine for your child, which changes as they gain weight and develop.

By the age of three, but sometimes much earlier, the Endocrine team will have been able to confirm whether or not your child has permanent hypothyroidism, requiring lifelong treatment with levothyroxine. Once this has been confirmed, your child will continue to need regular check-ups. These can be done closer to home by a local paediatrician, as, once your child is at nursery or school, having hospital appointments closer to home is far less disruptive to family life. If your child has additional problems, they may continue to have appointments at GOSH although

routine monitoring may continue at your local hospital. As children get older, blood tests are needed less frequently so that at school age, four- to six-monthly is frequent enough. A local paediatrician can make a new referral back to GOSH if any problems occur.

## What is the outlook for children with congenital hypothyroidism?

It is difficult to predict whether any young child will grow up normally. However, screening for congenital hypothyroidism has been happening in the UK for long enough for us to know that almost all children who are diagnosed and treated from an early age will grow up normally.

However, a small proportion of children who have had severe hypothyroidism in the womb may have some difficulties later in life, like poor hearing, clumsiness or trouble with learning. These problems can be reduced if hypothyroidism is picked up early and treated as described above.