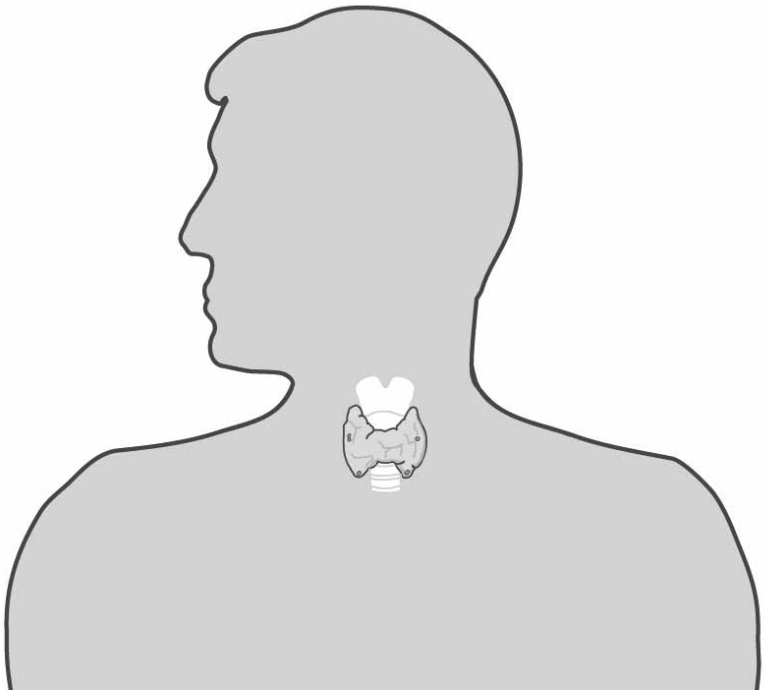
Congenital hypothyroidism

This leaflet explains about congenital hypothyroidism and how it is treated at Great Ormond Street Children's 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. 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. 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.

Other investigations

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.

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 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 this type of congenital hypothyroidism, the chance of having another baby who is affected 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 ask to talk to a genetics nurse about this.

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 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 and make sure that your child takes their medicine regularly each day and therefore keeps a steady level of thyroxine in their blood.

For the first couple of years, your child will need regular blood tests to check these levels. Doctors use the information from these tests to work out the right dose of thyroxine for your child, which changes as they gain weight and develop.

Once your child is two or three years old, they will need fewer blood tests as the dose of thyroxine will be calculated according to how he or she is growing. They will need to take thyroxine for the rest of their life, but this quickly becomes routine. The medicine is easily available and can be ordered on repeat prescription from your child's family doctor (GP).

Does the treatment have any side effects?

Because thyroxine 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 given too little thyroxine, your child 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 thyroxine, he or she may develop mild diarrhoea, not put on weight, may be more restless than usual and over a long period may grow more quickly than usual. But, as described above, the correct dose for your child will be calculated on a regular basis, so these effects are unlikely to 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.

Is there a support group for congenital hypothyroidism?

There is no support group specifically for people with congenital hypothyroidism. An informal network exists within the British Thyroid Foundation.

The British Thyroid Foundation
PO Box 97, Clifford, Wetherby,
West Yorks LS23 6XD

Tel: 01423 709707
Website: www.btf-thyroid.org

The following organisation is also helpful:

Child Growth Foundation
2 Mayfield Avenue, Chiswick London W4 1PW

Tel: 020 8994 7625
Website:
www.childgrowthfoundation.org

Notes

Compiled by the Endocrinology Departments
in collaboration with the Child and Family Information Group

Great Ormond Street Hospital for Children NHS Trust
Great Ormond Street
London WC1N 3JH

www.goshfamilies.nhs.uk www.childrenfirst.nhs.uk

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