NHS Newborn Blood Spot Screening Programme

Congenital hypothyroidism is suspected
When your baby was about a week old, your midwife took some blood from your baby’s heel, the ‘heel prick test’. The blood was used to test for some rare conditions including congenital hypothyroidism (CHT). The screening test result suggests your baby may have CHT. This result will need to be confirmed by further tests.
Congenital hypothyroidism

About 1 in 3,000 babies born in the UK has congenital hypothyroidism (CHT). Congenital means the baby is born with the condition.

Babies with CHT do not make enough of the hormone thyroxine, an important chemical made in the body. Thyroxine is produced by a gland in the neck called the thyroid. Without thyroxine, babies do not grow properly and can develop permanent physical and mental disabilities. CHT cannot be cured but can be treated simply and successfully.

There is currently no reliable way to detect CHT before birth and no way to prevent babies being born with CHT. There is nothing the parents of a baby with CHT could have done to prevent it.

What happens next?

You will be given an appointment to see a member of your healthcare team who will be able to discuss the screening test result with you and examine your child. They will arrange further blood tests to check if your baby has CHT.
Causes and types of CHT

Babies may develop CHT for different reasons.

1. Abnormal development of the thyroid gland

The thyroid gland may not reach its proper place in the neck during development in the womb, or it may be too small or even missing completely. There is usually no family history of CHT in these babies. The chance of a parent having another baby with CHT is very low.

2. Thyroid gland does not make thyroxine

In 10 to 20% of babies with CHT the thyroid gland is in the normal place and might even be enlarged, but it still does not produce enough thyroxine. In these families, there may be other relatives with thyroid conditions and there is a chance of having another baby with CHT. If you plan to have more children you may wish to discuss this with your healthcare team.
Living with CHT

Children with CHT are able to live full and active lives, like other children, as long as treatment is taken every day. A few children may develop problems with learning and clumsiness, and may need extra help. There may be a slightly increased risk of hearing problems.
Diagnosis of CHT

Newborn blood spot (‘heel prick test’)

The newborn blood spot screening test measures the level of thyroid stimulating hormone (TSH) in the baby’s blood. TSH triggers the thyroid gland to make more thyroxine. A high level of TSH suggests the thyroid is not working properly to make enough thyroxine.

Other blood tests

A repeat blood test is done to check the results of the newborn blood spot screening test. This measures the level of TSH again and also the level of free thyroxine (free T4) in the blood. A high level of TSH together with a low level of free T4 helps confirm the diagnosis of CHT.

Thyroid scans

A thyroid scan may be recommended. Thyroid scans are very safe and can give information about the type of CHT and whether this is likely to be permanent. They can help find out whether there is a chance of CHT occurring in future children.
There are 2 different types of thyroid scan:

**Thyroid uptake scan**

This provides a picture of the thyroid gland and can help see how it is working. A small dose of a chemical (radioiodine or technetium) is injected into the blood before a scan of the neck is performed.

**Ultrasound scan of the neck**

This looks at the thyroid position, shape and size. No injection is required and this is completely pain-free.

**Tests for the mother**

The baby’s mother may also have some blood tests to aid diagnosis.

**Treatment**

The treatment for CHT is to take levothyroxine by mouth, once a day. This replaces the thyroxine the body cannot make. Babies should start their treatment promptly, during the first few weeks after birth. Most children will need to continue it throughout life. Studies of children taking levothyroxine show this treatment is safe and effective.
More information

NHS.UK:
- www.nhs.uk/bloodspot

British Thyroid Foundation:
- www.btf-thyroid.org

The Child Growth Foundation:
- www.childgrowthfoundation.org

British Society for Paediatric Endocrinology and Diabetes:
- www.bsped.org.uk

Find out how Public Health England and the NHS use and protect your screening information at www.gov.uk/phe/screening-data.

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More information about screening: www.nhs.uk

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