Sgrinio Smotyn Gwaed Newydd-anedig Cymru
Newborn Bloodspot Screening Wales

GIG CYMRU NHS WALES
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Public Health Wales

Newborn bloodspot screening programme

Your baby's screening result

Congenital hypothyroidism (CHT) is suspected

Your baby's screening result

The result of your baby's 'heel prick' screening blood test suggests they might have congenital hypothyroidism (CHT). Your baby now needs further tests to check whether he or she has CHT or not.

This leaflet gives some information about CHT and explains what happens next.

What is CHT?

Each year in Wales, approximately 18 babies are born with congenital hypothyroidism (CHT). Congenital means the baby is born with the condition.

Babies with CHT do not make enough of the hormone thyroxine, which is an important natural substance 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. Newborn bloodspot screening is vital because it helps to identify babies with CHT before the low thyroid hormone levels cause permanent harm.

A few babies born with CHT may have symptoms such as jaundice (which causes yellowing of the skin and the whites of the eyes), dry skin, puffy eyelids, a large tongue, a hoarse cry, feeding problems, constipation and sleepiness.

Children with CHT can live full and active lives, like other children, as long as they take their medication every day. A few children may develop mild problems with learning and clumsiness, and may need extra help. There may be a slightly increased risk of hearing problems.

Treatment

The treatment for CHT is to take levothyroxine (thyroxine) by mouth once a day. This replaces the thyroxine the body cannot make. Babies should start treatment promptly during the first few weeks after birth. Studies of children taking levothyroxine show this treatment is safe and effective because it puts back what the body isn't making properly.

What happens next?

You will be given an appointment to see a specialist doctor as soon as possible. The doctor will be able to discuss the screening test result with you and examine your baby. They will arrange further blood tests to find out if your baby does have CHT

Causes and types of CHT

CHT may happen by chance but in some cases it may be inherited. There is nothing the parents of a baby with CHT could have done to prevent it. Babies may develop CHT for different reasons, as follows.

1. The thyroid gland has not developed normally

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. The thyroid gland does not make thyroxine

For some babies with CHT, the thyroid gland is in the normal place and might even be enlarged, but it still does not produce enough thyroxine because of an underlying 'production line' problem. 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 want to discuss this with your healthcare team.

Diagnosing CHT

Newborn bloodspot ('heel prick' test)

The newborn bloodspot screening test measures the level of thyroid-stimulating hormone (TSH) in the baby's blood. TSH triggers the thyroid gland to make thyroxine. A high level of TSH suggests the baby's thyroid is not working properly to make enough thyroxine. More blood tests will be needed to find out whether your baby does have CHT.

Further blood tests

A repeat blood test is done to check the results of the newborn bloodspot screening test. This measures the level of TSH again and also the level of thyroxine in the blood. A high level of TSH together with a low level of thyroxine (measured as 'free T4') is the pattern that is usually seen with 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 to find out whether there is a chance of future children having CHT.

There are two different types of thyroid scan, as follows.

Ultrasound scan of the neck

The ultrasound scan looks at the position, shape and size of the thyroid.

Thyroid uptake scan

This test can provide a picture of the thyroid gland and can help to see how it is working. A small dose of a chemical is injected into the blood before a scan of the neck is done. An active (normal) thyroid gland 'takes up' the chemical and its position can be seen on the scan. An underactive thyroid may not take up much chemical.

Tests for the mother

The baby's mother may also be asked to have blood tests to check for any abnormalities that may have affected the baby's results.

More information and support

The healthcare team responsible for your baby's care will be happy to discuss any questions you might have.

You can find more information at the following websites.

- British Thyroid Foundation
  www.btf-thyroid.org/congenital-hypothyroidism

- Medicines for Children
  www.medicinesforchildren.org.uk/levothyroxine-hypothyroidism

- The Child Growth Foundation
  www.childgrowthfoundation.org

- British Society for Paediatric Endocrinology and Diabetes (BSPED)
  www.bsped.org.uk

- Newborn Bloodspot Screening Wales
  phw.nhs.wales/newborn-bloodspot-screening

Notes

You can write down any notes here from discussions with your health professionals.

Using your information

For us to contact you as part of the programme, we will need to handle some of your and your baby's personal information. If you need more information about this, you can:

- visit the website: phw.nhs.wales/use-of-site/privacy-notice
- email PHW.InformationGovernance@wales.nhs.uk, or
- phone 029 2010 4307.

We also keep personal details to make sure that the standard of our service is as high as possible. This includes checking your baby's records if your baby is found to have a condition after having a screening test which showed a 'not suspected' result.

We only ever publish information as statistics and we never publish personal details. We pass on your personal information to health professionals or organisations that need it, including your GP, health visitor and consultant paediatrician. These professionals must protect the personal information in the same way as we do.

All our paper and computer records are stored and processed securely, and the public cannot access them.

Information courtesy of Public Health England.

Version 1
December 2020