



INFORMATION FOR PARENTS/CARERS FURTHER INVESTIGATIONS REQUESTED FOR CONGENITAL HYPOTHYROIDISM

What is the Newborn Screening Programme?

Your baby had a blood sample taken from the heel at about three days of age. The blood was absorbed into a special card and sent to the NSW Newborn Screening Laboratory in Sydney where it was tested for several disorders. Congenital Hypothyroidism (CH) was one of the disorders.

Why are babies screened for CH?

Most babies with CH are not obviously different from unaffected babies. Without a screening test the child might be months or years old before the diagnosis is made. This delay in diagnosis and treatment can lead to poor growth and intellectual disability.

What is Congenital Hypothyroidism?

Congenital Hypothyroidism is a condition where a baby is born with a thyroid gland that does not work properly. A normally working thyroid gland is critical for normal growth and brain development.

The thyroid gland could be completely absent, or small and underdeveloped or not in the normal position in the neck.

More rarely CH is caused by the absence of an enzyme in the thyroid gland, preventing it from making thyroid hormone (Thyroxine).

The incidence of CH is one in about every 3,500 babies born in New South Wales and the Australian Capital Territory.

What are the symptoms of Congenital Hypothyroidism?

A baby with CH may have no obvious symptoms. However, some babies born with CH may be very sleepy and feed slowly. They may have a tendency to be constipated and suffer from yellowing of the skin (jaundice) that lasts a long time.



My doctor says that my baby has an elevated result for CH from Newborn Screening, and may have CH. What other tests will my baby need to have?

Your baby may need three other tests to confirm the result obtained by screening:

1. A blood test for further thyroid function tests.
2. A thyroid scan to see the position and shape of the thyroid gland. For this test the baby is given an injection of a substance called technetium. There is no known risk associated with this test. To be successful the thyroid scan needs to be performed before any treatment for CH starts.
3. A bone age x-ray usually of the knee to detect if there is any delay in your baby's growth.

IT IS VERY IMPORTANT TO HAVE THESE TESTS DONE AS QUICKLY AS POSSIBLE, SO THAT TREATMENT CAN BEGIN.

What is the treatment for Congenital Hypothyroidism?

The treatment for CH is very simple, just a replacement of the thyroid hormone by thyroxine syrup or tablets crushed and added to milk. Enough thyroxine is given to your baby to increase thyroxine levels to that of an unaffected baby. Thyroxine needs to be given every day for the rest of your child's life.

Your baby will need to see a paediatrician at intervals and have regular blood tests to make sure that thyroxine levels are normal.

The paediatrician will also check your baby's growth and development. Blood tests and check ups will become less frequent as your child gets older.

What does the thyroid gland do?

The thyroid gland is responsible for making thyroid hormone which has 3 main functions:

1. Thyroid hormone helps develop your baby's brain in the first 2 years of life. A lack of thyroid hormone during this time leads to intellectual disability.
2. Thyroid hormone is needed for normal growth, so not having enough can lead to poor growth and short height as an adult.
3. Thyroid hormone keeps the body running at normal speed - a child without it may feel cold, tired and be constipated (not able to move their bowels).

Will my child grow up normal?

The outlook is usually excellent. Early diagnosis and adequate treatment with thyroxine has resulted in normal growth and development for the hundreds of CH children diagnosed by newborn screening.

However, normal growth and development for your child requires constant monitoring and effective treatment every day, and regular check ups with your doctor.

Could CH have been prevented during pregnancy?

Almost certainly "no". The reasons for the under-development or the thyroid gland are not known, so we do not know whether there is any way of preventing it.

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For further information please contact your paediatrician or the NSW Newborn Screening Programme
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