



CONGENITAL HYPOTHYROIDISM SCREENING INFORMATION FOR PARENTS/CARERS

What is the Newborn Screening Programme?

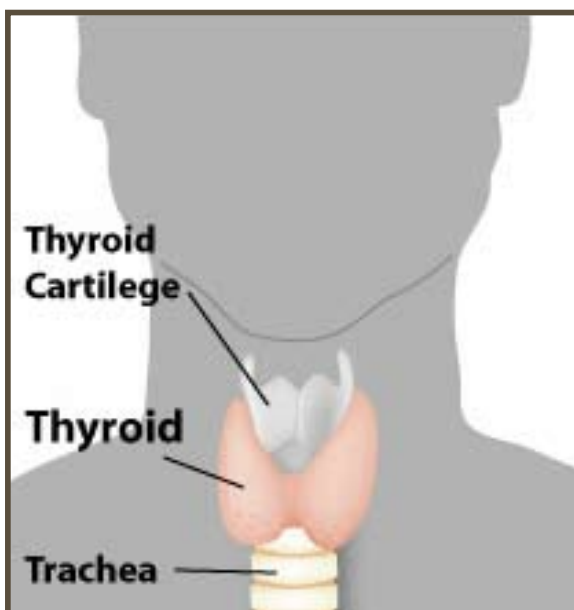
Your baby had a blood sample taken from the heel at about three days of age. The blood was absorbed into a special card and sent to the NWU Newborn Screening Laboratory in Potchefstroom where it was tested for several disorders. Congenital Hypothyroidism (CH) was one of the disorders.

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?

Hypothyroidism (or underactive thyroid) is a common condition in which the thyroid gland produces too little thyroid hormone. About 1 in 5,000 babies is born with congenital hypothyroidism, in which the thyroid fails to grow normally and cannot produce enough of its hormone. There is no known cause for most cases of congenital hypothyroidism. But about 10 to 20 percent of the time, the condition is caused by an inherited defect that alters the production of thyroid hormone.

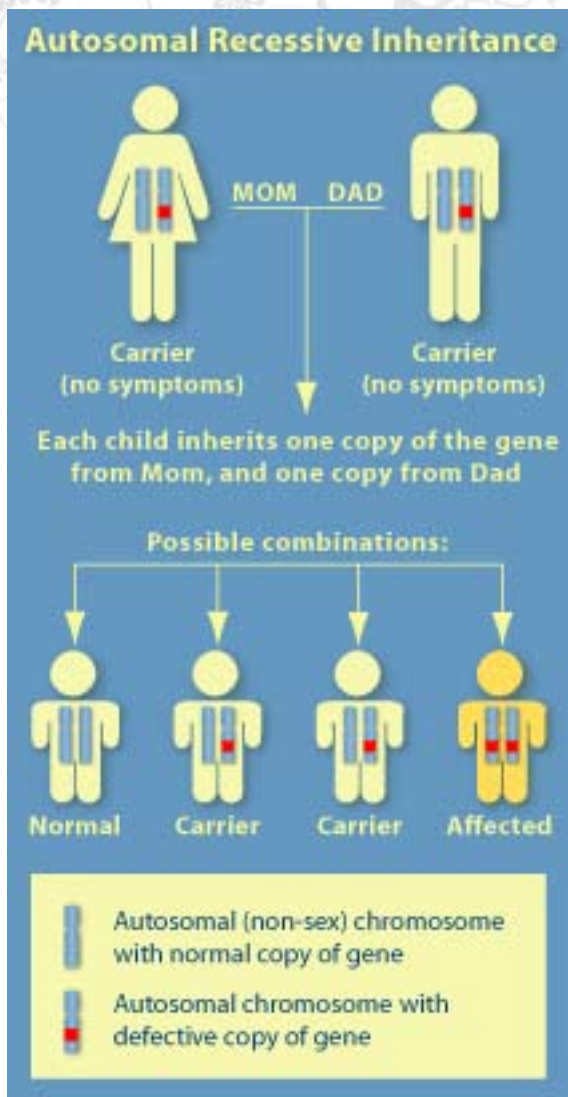


Congenital Hypothyroidism is a condition where a baby is born with a thyroid gland that does not work properly. A normally working thyroid gland is critical for normal growth and brain development.

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).





How do people get hypothyroidism?

Hypothyroidism is a recessive disorder, which means that both parents must pass on the defective gene for any of their children to get the disease. If a child inherits only one copy of the faulty gene, he or she will be a carrier. Carriers don't actually have the disease, but they can pass it on to their children.

The most common inherited form of hypothyroidism is a defect of the TPO (thyroid peroxidase) gene on chromosome 2. This gene plays an important role in thyroid hormone production.

What are the symptoms of Congenital Hypothyroidism?

In babies with the inherited form of hypothyroidism, the condition affects growth and cognitive development. It may cause mental retardation, delayed puberty, stunted growth, and ataxia (inability to coordinate muscle movements).

In adults, hypothyroidism slows the body's metabolism, making the patient feel mentally and physically sluggish. Symptoms may include weakness, fatigue, muscle aches, mood swings, hair loss, memory loss, or slow speech. A person's symptoms will depend upon how little thyroid hormone they produce, and for how long they have had the disorder.

When the body is deprived of thyroid hormone, the pituitary gland works overtime, producing extra thyroid-stimulating hormone (TSH). This glut of TSH may enlarge the thyroid into a condition called a goiter.





How do doctors diagnose hypothyroidism?

Babies are normally screened for hypothyroidism 24 hours after birth. A tiny sample of blood taken from the baby's heel is tested for low thyroid hormone levels or high thyroid-stimulating hormone (TSH) levels.

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 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.
3. A thyroid ultrasound, if no thyroid is seen on scanning, or if a scan cannot be performed.
4. 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 simple. Thyroid hormone is given by crushing a tablet, adding a small amount of milk and giving it to the baby by spoon to make sure the baby gets the full dose. 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 paediatric endocrinologist or 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.



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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?

At present the answer is “no”. The reasons for the under-development of the thyroid gland are not known, so we do not know any way of preventing it.

