



## Congenital Hypothyroidism

### What is Congenital Hypothyroidism?

Hypothyroidism means that the *thyroid gland* at the front of the neck is underactive or not fully functioning. Congenital hypothyroidism means that a newborn baby is unable to make normal amounts of *thyroid hormone*. This can be a permanent condition and need lifelong treatment. Thyroid hormone is important for your baby's growth and brain development. Therefore, untreated congenital hypothyroidism can lead to poor growth and developmental delay. However, with early detection and appropriate treatment, your baby will likely be able to lead a normal healthy life.

### Causes of Congenital Hypothyroidism

A common cause of congenital hypothyroidism is when the thyroid gland has not developed properly because the gland is missing, too small or located at a different location at the neck. In some cases, the thyroid gland is present at the front of the neck but does not manufacture the hormone correctly. In rare cases, the thyroid gland misses a signal from the pituitary gland to produce thyroid hormones. Occasionally, in a very small number of cases, anti-thyroid medications taken by the mother to treat an overactive gland can result in a temporary hypothyroidism which should resolve 3 months later.

### Symptoms and Signs of Congenital Hypothyroidism

A majority of babies with congenital hypothyroidism look no different from the unaffected baby, as the symptoms are not usually obvious at birth. Some babies with severe hypothyroidism may have signs of poor feeding, weak cry, excessive drowsiness, constipation and a prolonged period of yellowish discoloration of the skin (jaundice). The doctors will examine your baby to evaluate if your baby has an enlarged fontanelle (a diamond-shaped soft spot at the top of your baby's head), a large tongue or poor muscle strength.

### Confirming the Diagnosis of Congenital Hypothyroidism

Given the difficulty in detecting congenital hypothyroidism at birth, almost all babies born in Singapore undergo the *newborn screening program* where a small sample of blood is collected from the baby's umbilical cord immediately after birth and sent for laboratory testing.

Where the result of thyroid screening is *slightly abnormal* (meaning there is a slightly low level of thyroid hormone and a slightly high or normal level of thyroid stimulating hormone), the doctor will explain to you that a second sample of blood will be taken at day 6 - 8 to confirm the diagnosis. The blood test will be taken from a vein on the baby's hand or foot by the doctor.

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Where the result of thyroid screening is *very abnormal* (meaning there is a very low level of thyroid hormone and/or normal to high level of thyroid stimulating hormone), the doctor will speak with you and recommend a second blood sample to be taken early and arrange for a thyroid scan to determine the location and function of the gland before treatment.

Where the result of thyroid screening is *negative* (meaning the thyroid hormone and thyroid stimulating hormone are both within the normal range), you will not need to bring for repeat testing.

## **Treatment of Congenital Hypothyroidism**

Congenital hypothyroidism is treated by giving thyroid hormone medication as a pill called *levothyroxine*. Many babies will require treatment for life. The levothyroxine pill should be crushed and given once daily, mixed with a small amount of water, breast milk or formula milk using a dropper or syringe. It is important to serve the thyroid hormones every day and have regular check-up with your doctor to ensure that your baby will have normal growth and brain development. As your baby grows in weight, the doctor will recommend slightly higher doses to keep up with your baby's weight. You should avoid giving your baby soy milk, vitamins with iron and calcium supplements when he/she is taking thyroid hormones, because these foods can interfere with the absorption of thyroid hormone from your baby's gut.

The levothyroxine pill is the same thyroid hormone as what is made by the body so you are just replacing what is missing. In general, side effects only occur if the dose of thyroid hormone is too high which the doctor can avoid by checking the level of hormone in the baby's blood test. Ask your doctor if you have any questions.

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