

Hormones and Me
Congenital Hypothyroidism
A Guide For Parents

This booklet is valuable reading for parents of children with
Congenital Hypothyroidism.

It is also recommended reading for their family and friends.

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About this book

This booklet, *Congenital Hypothyroidism – A Guide for Parents*, aims to provide a basic understanding of how the thyroid gland works and the causes of congenital hypothyroidism and its treatment.

We encourage you to discuss any additional questions or areas of concern with your doctor after reading this booklet.

Merck Serono are proud to bring you this booklet from the *Hormones and Me* educational series. We hope that you find it a valuable and helpful resource.

This booklet was revised in 2014 with the help of Professor Maria Craig (The Children's Hospital at Westmead, NSW, Australia), a Paediatric Endocrinologist specialising in childhood endocrine disorders and a member of the Australasian Paediatric Endocrinology Group (APEG).

Paediatric endocrinologists, Professor Margaret Zacharin (Royal Children's Hospital, VIC, Australia) and Dr Ann Maguire (The Children's Hospital at Westmead, NSW Australia) have reviewed the *Hormones and Me* series on behalf of the Australasian Paediatric Endocrine Group (APEG).

Introduction

The term congenital hypothyroidism (CH) describes a condition that is present at birth (congenital) and in which the thyroid gland is absent or does not produce normal amounts of thyroid hormone.

Congenital hypothyroidism occurs in approximately 1 in 2000 to 3000 babies, so each year in Australia about 100 babies are born with this condition.

There is usually no indication that there is anything medically wrong with these babies at the time of birth. Therefore, it may be a shock to parents to be told that their baby's thyroid gland is not functioning properly, and it may be difficult for parents at first to absorb all the information given about hypothyroidism.

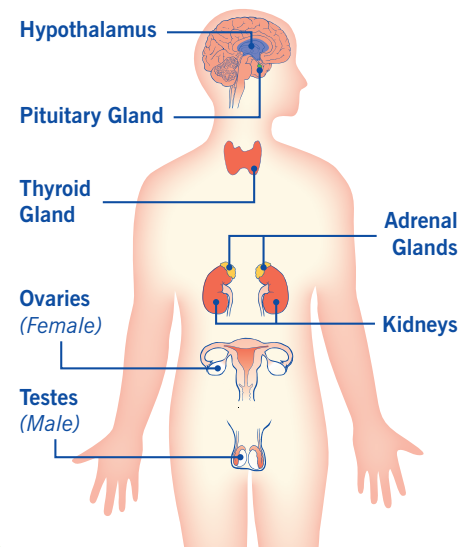
In Australia routine thyroid screening of all babies has been performed since the 1970's. Hypothyroidism is therefore detected by the end of the first week of life and treated. However, in many other countries this type of screening is not yet available or is only available in some centres. Thus, a baby born outside Australia may not have had the opportunity of early treatment and may have problems of development that are not usually seen in babies born in Australia today.

This booklet has been written to help you to learn about congenital hypothyroidism. It explains the function of the thyroid gland and the different types of congenital hypothyroidism; the newborn screening program and how it operates; and the treatment and outlook for babies with this condition. In the final section of the booklet, some of the more common questions that parents ask about congenital hypothyroidism are answered.

Hormones

Hormones are chemicals that carry messages from one part of the body to another via the bloodstream. They are produced by endocrine glands (such as the thyroid gland) and play a vital role in regulating metabolism, growth and development. Low levels or deficiency of a hormone can have dramatic effects on normal bodily functions.

Diagram 1; Location of the thyroid and other glands in the body



action on the body, such as growth hormone, while others stimulate hormone production in other glands in the body, such as the thyroid gland and adrenal glands (see *Diagram 1*).

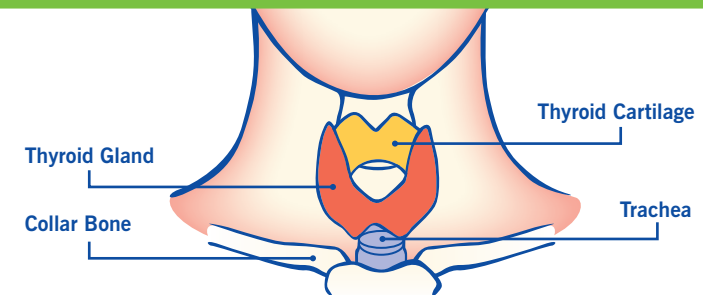
The regulation of hormones in the body starts in a part of the brain called the hypothalamus, which sends chemical messages to the pituitary gland. The pituitary, a pea-sized gland located at the base of the brain, responds to these messages and in turn regulates the release of most of the body's hormones. The pituitary gland secretes many hormones: some of them have a direct

The Thyroid Gland

The thyroid is a butterfly-shaped gland that is located in the front of the lower part of the neck and consists of two connected lobes on each side of the trachea (windpipe) (see *Diagram 2*). It begins to develop very early in the human embryo – about three weeks after conception. It develops first at the base of the developing baby's tongue, and then moves downwards into the neck to take up its final position below the thyroid cartilage (also called the Adam's apple). The thyroid gland is usually situated in its normal position by the end of the first trimester (13 weeks) of pregnancy and starts to produce thyroid hormone (also called thyroxine) by this time. Until the baby's thyroid gland starts to produce enough thyroid hormone later in the pregnancy, he or she will be dependent on the mother's thyroid gland for this hormone.

While the thyroid gland is developing, the hypothalamus and pituitary are also forming. The hypothalamus begins to send signals to the pituitary, which in turn stimulates the thyroid gland to grow and to produce the thyroid hormone. By full term, the thyroid gland is functioning normally and can produce sufficient thyroid hormone for the newborn baby's needs.

Diagram 2 : The shape and position of a normal thyroid gland



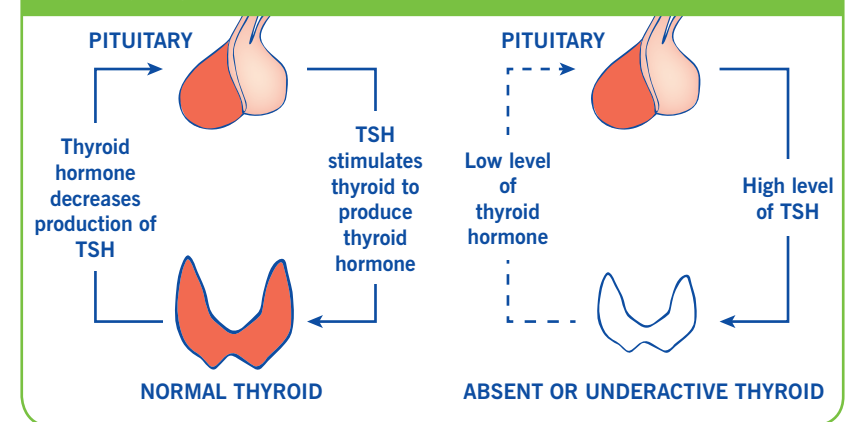
Function and regulation of the thyroid gland

The function of the thyroid gland is to make thyroid hormone, which is released into the bloodstream. Thyroid hormone is involved in regulating the body's metabolic rate, which is the speed at which the body breaks down (metabolises) protein, fat and carbohydrate to provide the energy for many normal daily bodily functions. Thyroid hormone is also needed for the body to make proteins, for brain development and growth throughout childhood. It is an essential hormone for almost every cell in the body. The production of thyroid hormone involves several steps, in which thyroid gland cells use iodine and other raw materials from the bloodstream and converts them into thyroid hormone.

The pituitary gland makes thyroid stimulating hormone (TSH), which is also called thyrotropin. The pituitary gland communicates with the thyroid gland via its production of TSH (see *Diagram 3*). When the thyroid gland is producing too little thyroid hormone, the pituitary is able to sense this. As a result it increases its output of TSH, which circulates in the blood to the thyroid gland and stimulates it to produce more thyroid hormone. When the thyroid is secreting enough thyroid hormone, this causes the pituitary to decrease its production of TSH to a normal level. If the thyroid is producing too much thyroid hormone, this causes the pituitary gland to reduce its production of TSH to a low level.

Therefore, when the thyroid gland is underactive (producing too little thyroid hormone), the blood will contain a high level of TSH and a low level of thyroid hormone. There are different causes of an underactive thyroid gland and some of these are discussed in the section 'Types of Congenital Hypothyroidism'.

Diagram 3: The pituitary gland's regulation of thyroid hormone production by the thyroid gland



Actions of thyroid hormone

Normal levels of thyroid hormone are essential for normal physical growth and development throughout childhood. It is also necessary for normal brain development during pregnancy, brain growth in the first two years of life and for brain function throughout life. Thyroid hormone is important in regulating the body's metabolic rate throughout life, so that when there is not enough (deficiency) of thyroid hormone, the metabolism slows down, and when there is too much



(excess), the metabolism speeds up. Metabolic rate varies from person to person and can be influenced by various factors including age, body size, diet and exercise, as well as hormone levels. A slow metabolic rate may bring about the following effects:

- **Tiredness, low energy levels**
- **Poor memory and concentration**
- **Low body temperature**
- **Feeling cold**
- **Slow pulse rate**
- **Constipation**

These effects may be seen at all ages. Early in life, there are additional effects of thyroid hormone deficiency and babies with congenital hypothyroidism may also have the following features:

Slow growth

Thyroid hormone is essential for normal growth. Even if a baby is born without a thyroid gland, he or she will have had the benefit of the mother's thyroid hormones during the pregnancy. Therefore, the baby will be a normal size at birth. After birth the infant must have normal levels of thyroid hormone to grow and develop. If these are low or absent a child will not grow normally and if untreated would eventually be a very short adult.

Slow mental development

Thyroid hormone is essential for many parts of the body, including the brain. The brain develops normally before birth if the mother's thyroid function is normal. However, the development of the infant brain, up to the age of about two and a half years, is dependent upon on normal levels of thyroid hormone.

Intellectual impairment can occur if thyroid hormone levels are low. If the thyroid hormone level is only slightly lowered, there may be little or no effect on intellectual function. However, when thyroid hormone is completely absent and if a baby does not receive any treatment, brain growth is damaged, with consequent intellectual disability. This can be prevented with early diagnoses and treatment (see section: 'Outlook for Babies with Permanent Congenital Hypothyroidism').

Persistent jaundice

The term jaundice means that the skin has a yellow colour due to the presence of bilirubin, which is a product of the breakdown of old red blood cells. It is common for healthy newborn babies to have mild jaundice, which may last for a week or two and is usually not harmful. In untreated congenital hypothyroidism jaundice may persist for much longer, but once thyroid hormone treatment is started the jaundice will usually disappear quickly.

Other features may be seen including excessive sleepiness, poor feeding and weight gain, dry skin, low body temperature, low muscle tone, puffy face and a hoarse cry. Some babies can have an umbilical hernia, a large fontanelle or a large tongue.



Types of Congenital Hypothyroidism

Congenital hypothyroidism can be divided into three categories:

- 1 Thyroid dysgenesis (due to abnormal formation, development and growth of the thyroid gland)**
- 2 Thyroid dyshormonogenesis (due to abnormal production or release of thyroid hormone)**
- 3 Central hypothyroidism (due to abnormal formation or function of the hypothalamus and/or pituitary gland)**

The first two categories are sometimes called primary hypothyroidism because they are due to disease of the thyroid gland itself.

1. Thyroid Dysgenesis

In most babies with congenital hypothyroidism, the thyroid gland is either absent, has not formed properly or is in an abnormal position.

Absent thyroid gland (thyroid athyreosis)

In babies with this condition, the thyroid gland fails to develop before birth: it is absent and will never grow. Consequently no thyroid hormone is produced. This condition is called athyreosis or thyroid agenesis.

Twice as many girls as boys are affected. It occurs in about 1 in 10,000 births and accounts for about one-third of the cases detected by newborn screening. We do not understand the reason why the thyroid gland fails to develop in most of these babies. However, a number of genes have been identified that can cause this problem.

Abnormally positioned thyroid gland (ectopic thyroid)

The term 'ectopic' means something is in an abnormal position. An ectopic thyroid gland is not in its normal position in the neck and is also small and malformed. It is often found at the base of the tongue, near the place where the gland first began to form in the developing baby. An ectopic thyroid gland may have varying degrees of function. Some ectopic thyroids are very small and very underactive, while others are initially able to produce a nearly normal amount of thyroid hormone. Thus, there are degrees of severity in this condition.

We know that after birth an ectopic thyroid gland will not improve its function or descend to its normal position. An abnormally placed thyroid may occasionally work well enough in an infant to provide sufficient thyroid hormone for normal growth and development, but the gland is unlikely to continue to function throughout life and usually stops working within a few months, or occasionally a few years.

An ectopic thyroid gland is about twice as common in girls as in boys. This type of problem accounts for about half of the cases of thyroid dysgenesis and is slightly more common than athyreosis. Again, it is not known for sure why in some babies the thyroid gland is located in an ectopic position, but some of the same factors that cause athyreosis may well give rise to this problem.

Malformed thyroid gland in the normal position (thyroid hypoplasia)

Hypoplasia means that an organ has failed to develop fully and/or reach its normal size, and in thyroid hypoplasia the gland is small, malformed and occasionally consists of only one lobe. The condition only accounts for about 5% of all cases of congenital hypothyroidism. Some cases are due to an abnormal gene inherited from one or both parents.



2. Thyroid Dyshormonogenesis

Babies with thyroid dyshormonogenesis have a normally positioned thyroid gland in the lower neck, but it cannot produce a normal amount of thyroid hormone. This condition accounts for about 15% of cases detected by neonatal screening. It can be either transient, in which case it gets better with time, or it can be permanent and last throughout life. In babies with this condition, the thyroid gland is often enlarged and may be seen or felt in the front of the neck (this is called a goitre).

Permanent dyshormonogenesis

When thyroid hormone is made, each step is controlled by a particular protein called an enzyme. Occasionally one of these enzymes is missing and causes permanent dyshormonogenesis. This abnormality is usually an inherited condition and therefore other babies born into the family are also at risk of having it. Usually both parents need to carry a gene for this problem, with a one in four chance of a child being affected (boys and girls have an equal chance of being affected).

Transient dyshormonogenesis

In this condition, the function of the thyroid returns to normal after a variable period of time. There may be several causes of this problem. Sometimes, antibodies from the mother's blood cross the placenta during the pregnancy and decrease the function of the baby's thyroid gland. Women who have had abnormal thyroid function themselves are at particular risk of this happening. It is, therefore, particularly important for a pregnant woman to tell her doctor if she has an underactive thyroid and any family history of thyroid disease.

Another cause of transient dyshormonogenesis is the presence of too much iodine in the body. Whilst iodine is essential for the manufacture of thyroid hormone and iodine deficiency can lead to hypothyroidism, the opposite can happen too. Too much iodine can overload the thyroid gland and cause it to stop working for a while. This can happen if a mother consumes a lot of iodine containing medication during pregnancy, while breast feeding or if a baby needs surgery (e.g. for a heart condition) and the body is washed with an iodine preparation. However, once the excessive iodine intake stops, the thyroid gland will return to normal and the baby will not suffer any long-term ill effects.

3. Central Hypothyroidism

In this condition, the thyroid gland develops normally and is in the correct position, but TSH is either not produced or not released properly from the pituitary gland. As a result, the thyroid gland is not stimulated by TSH to produce and release enough thyroid hormone to meet the body's needs. This is a very rare condition. It only accounts for less than 5% of all cases of congenital hypothyroidism and occurs in about 1 in 30,000 newborns.

Central hypothyroidism may be associated with deficiencies of other pituitary gland hormones (for example, due to congenital hypopituitarism), brain abnormalities or other uncommon medical conditions.



Newborn Screening

All babies born in Australia, as well as many other countries, are screened for congenital hypothyroidism.

To do a newborn screening test, it is necessary for the baby to have a very small sample of blood removed, usually taken from the baby's heel, and placed on a piece of absorbent paper. This blood spot is forwarded to a central laboratory for measurement of TSH. Screening is usually carried out between two and three days after birth.

A high level of TSH in the blood spot identifies babies who have primary congenital hypothyroidism (that is, congenital hypothyroidism due to dysgenesis or dyshormonogenesis). The test does not identify babies with central hypothyroidism. If the TSH level is high the test is repeated, on a sample of blood taken from a vein, to confirm the original result. Thyroid hormone levels are tested at the same time.

The reasons why newborn screening is carried out are as follows:

- It is very difficult to diagnose congenital hypothyroidism at birth just by examining the baby. Features described previously, such as slow growth and impaired mental development, are not present at birth, and are only able to be detected after weeks or months of untreated hypothyroidism. Therefore, if newborn screening is not carried out, many cases of hypothyroidism will be missed at birth and treatment delayed, leading to a high risk of impaired brain and physical development.
- Congenital hypothyroidism is not a rare condition. About 100 new cases are diagnosed in Australia each year (1 in 2000 to 3000 births).

In some babies, the newborn screening test needs to be repeated. This includes twins, babies born prematurely, those with a very low birth weight (less than 1000 g), or if they have been unwell and admitted to a neonatal intensive care unit. All these conditions can alter thyroid hormone levels, but in most cases the baby will not need treatment.

Early diagnosis and treatment of all forms and severity of congenital hypothyroidism has an excellent outcome - with normal physical and intellectual development of the almost all affected children.



Investigations to Determine the Type of Hypothyroidism

If the results of a baby's newborn screening are abnormal, this information is immediately given to a doctor who will arrange for the baby to be seen on the same day. At this appointment a medical history is taken from the parents and the baby is carefully examined to gain clues as to what type of hypothyroidism is present.

At the first visit, some special investigations are also carried out. A blood test is done to confirm the results of the newborn screening and to test the level of thyroid hormone in the blood. Sometimes, if the baby has jaundice the blood sample is also analysed to check for jaundice (elevated bilirubin). If the mother has a history of thyroid disease she will also be asked to have a blood test as it may help to diagnose the cause of the baby's problem. Usually, the baby's knee is x-rayed because bone growth can be delayed in hypothyroidism.

Another investigation that is usually performed at this appointment is a thyroid scan. For this test, the baby is given an injection of a radioactive substance called technetium, which is taken up by the thyroid gland. This makes it possible to see the position and shape of the thyroid and to determine if the gland is absent, in the wrong place (ectopic) or present in the normal position.

The administered radioactivity disappears within hours and technetium is excreted from the body very quickly. A thyroid scan should only be performed when the baby is not already being treated for hypothyroidism. This is why it is usually done at the first visit. There is virtually no risk associated with the use of technetium because only a low dose is used.

Many babies will also have a thyroid ultrasound scan. This shows whether the thyroid is present or not, and whether the size is normal. Sometimes, it may not be necessary to do both a thyroid scan and a thyroid ultrasound if one of these investigations shows a definite abnormality.

If the scan or ultrasound cannot be done straight away, treatment should not be delayed as these tests can be done after treatment has commenced. A technetium scan can, however, only be done for a few days after treatment is commenced. An ultrasound can be done at any time.

For some babies with thyroid dysmorphogenesis, more detailed investigations are carried out after two or three years of treatment to determine if the child has an enzyme deficiency. All babies should have their hearing screened at or by 4 months of age. Occasionally an additional hearing test may be needed, especially for those with dysmorphogenesis, because progressive loss of hearing, although rare, can sometimes occur with this type of thyroid problem.



Treatment

In all forms of permanent hypothyroidism, the thyroid gland is unable to make enough thyroid hormone for the body's needs, and therefore, this hormone must be replaced. Treatment should be commenced as soon as possible after the results of the repeated TSH level and thyroid function tests results are available. Thyroid hormone is given in tablet form by mouth as a daily dose. The thyroid hormone that is present in the tablets is exactly the same as the thyroid hormone that is naturally present in the body.

Thyroid hormone is well absorbed from the digestive tract and readily enters the bloodstream. Thus, it does not have to be given by injection like some other hormones such as insulin used for the treatment of diabetes.

Your doctor will tell you the dose of thyroid hormone your baby needs, which is based on your baby's weight. The tablets should be crushed and administered in a few millilitres of water or breast milk. This can be done by putting the dissolved tablet mixture in a 1 ml syringe, provided by the hospital, and placed in the back of the baby's mouth. The dose is usually given before a feed to minimise the chance of losing the medicine if the baby regurgitates a bit of milk. Naturally, as the baby grows, a higher dose may be needed. It has also been shown that the dose of thyroid hormone may need to be altered if the baby is changed to a soy-based formula.

Once treatment has been started, blood tests are done at regular intervals in order to measure the amounts of thyroid hormone and TSH in the blood. These tests are usually done 1–2 weeks after starting thyroid hormone treatment, then every 2 weeks until the TSH level is normal, and then every 2-3 months. The doctor will tell the family how often testing is needed for each infant. Between the ages of one and three years, children should have the tests done every 2 to 4 months, and after the age of 3 years, every 3 to 12 months until growth is completed. The frequency of testing also depends on various factors, such as a child's age and whether there has been a recent change in dose. A repeat test usually needs to be performed 4–6 weeks after a dose change.

The aim is to keep the thyroid hormone level in the blood in the upper half of the normal range for the child's age. With appropriate thyroid hormone replacement treatment, the pituitary gland no longer needs to secrete large amounts of TSH. Therefore, the amount of TSH in the blood will drop to a normal level, though this may take several weeks.

If treatment is inadequate (for example, if the dosage is too low or the tablets are not being given or absorbed properly, or they are out of date) the thyroid hormone level in the blood will be low and there will be an elevated TSH level.

Treatment for permanent hypothyroidism is life-long. If the tablets are not taken for a prolonged time, the symptoms of hypothyroidism will return.



Occasionally, an underactive thyroid detected in a baby may be transient and may get better with time. In this case treatment may only be required for a relatively short period. A baby's brain is particularly susceptible to damage from low thyroid levels, so extra care is needed for the first 2 years of life.

Thyroid tablets are only available in blister packs to avoid inadvertent intake by other children. It is usually advised that they be kept in the refrigerator. If there is a problem with keeping the tablets in a refrigerator it is reasonable to leave them at room temperature, for example, when travelling.

If your baby has been prescribed iron supplements these should not be taken at the same time as their thyroid tablets, as the iron can prevent the normal absorption of thyroxine.

Follow-up

Babies with congenital hypothyroidism are seen by a doctor at regular intervals to assess progress.

An appointment will be made for your baby to be seen by an endocrine specialist every 4-12 weeks. The specialist will make sure that the child is developing normally and that the thyroid hormone levels are satisfactory. A blood test will be performed at every visit to check thyroid hormone levels and dose adjustments will be made, according to the results. Usually there are very few problems once treatment has begun.

Outlook for Babies with Permanent Congenital Hypothyroidism

Before the newborn screening program for hypothyroidism was instituted, diagnosis was frequently delayed and, as a result, some infants suffered permanent mental retardation. With early diagnosis and treatment this can be prevented. Children with permanent congenital hypothyroidism can be expected to have normal intelligence and growth, and be as healthy as children without hypothyroidism.

However, it should be appreciated that, just as there is a range of levels of intelligence in the general community, there is a range of intellectual abilities among hypothyroid children. While the majority of them have average intelligence, some are a bit below average and some above. In addition, a small proportion of children with hypothyroidism have other medical problems, such as hearing impairment or visual processing problems, which may also have an effect on their intellectual development.

Because of newborn screening, babies who are born with a thyroid problem today can be expected to grow and develop just like other healthy children who have a normal thyroid gland.

A small proportion of children with congenital hypothyroidism may have another medical condition, so it is recommended that they are seen by a paediatrician in the newborn period, and for follow-up checks during childhood.



Questions and Answers

What is the best way to give thyroid hormone tablets to a baby?

It is best to crush the tablet on a spoon, mix it with a few millilitres of breast milk or sterile water. Safe and effective dosing can be improved by using a 1 ml syringe (without a needle!) to administer the mixture. The syringe should be placed towards the back of the baby's mouth, to avoid it being spat out. It can be taken either in the morning or evening, either before feeding (to avoid losing any medicine if the baby regurgitates) or with milk/food, but it should be administered in the same way every day.

Are there any foods that may interfere with the absorption of thyroid hormone tablets?

Soy-based formulas, and iron and calcium supplements can reduce the absorption of thyroid hormone in the intestine, necessitating larger doses. Therefore, it is best to avoid giving thyroid hormone tablets at the same time as these types of foods. If a baby with congenital hypothyroidism is changed to a soy-based formula, it would be wise to let the doctor know.

What will happen if a baby misses a dose of thyroid hormone or vomits up one of the tablets?

No harm is done if only one or two doses are missed. It is, of course, important that the tablets are administered regularly, so obtaining a repeat prescription from the doctor when supplies are getting low is recommended. If the baby vomits within half an hour of receiving the dose of thyroid hormone, the same dose should be repeated later in the day when the vomiting has stopped.

Can treatment be stopped at any stage – for example, on reaching adulthood?

It is not possible to stop treatment for permanent congenital hypothyroidism. Thyroid hormone must be taken regularly throughout life, even when physical growth is completed.

Will any problems occur when a child has immunisations or has to take other medication?

No. Children with hypothyroidism can have the usual immunisations and take other medication without any problem.

What are the side effects of thyroid hormone treatment?

Because the treatment of hypothyroidism involves replacement therapy with a natural hormone, there are no side effects if the dosage is appropriate. However, if too much thyroid hormone is given, the effects will be the same as those that occur with an overactive thyroid gland, such as rapid pulse, loss of weight, restlessness and overactivity.

What are the risks of a subsequent child in the family having hypothyroidism?

The risks depend on the cause. Most cases of congenital hypothyroidism are due to abnormal development of the thyroid gland; absent (athyreosis), small or malformed (hypoplasia) or developed in an abnormal position (ectopic). Because these problems are not usually inherited the risk of a subsequent child having a similar condition is very small. In the general population, the likelihood of a baby being born with an absent or malformed thyroid gland is about 1 in 2000 to 3000. The risk may be higher when there is already one affected child in the family. The risk of familial recurrence should be discussed with the doctor.

If the baby is one of the few babies with permanent dysmorphonogenesis due to an enzyme deficiency, the risk of having a second infant with the same problem is usually one in four (see the section 'Types of Congenital Hypothyroidism'). With transient dysmorphonogenesis, there is usually no risk of recurrence, unless the cause of the problem (for example, excess iodine) is still present.

When a person with congenital hypothyroidism has children, what risk do these children face of having the same condition?

There is only a low risk that the children of a mother or father with congenital hypothyroidism will have the same problem. In general, this applies to all types of congenital hypothyroidism. Even with permanent dysmorphonogenesis due to an enzyme deficiency, the risk remains low, unless the affected person has a partner who either has the same condition or is a carrier of it.

It is true that some families do have a higher frequency of thyroid problems than others, so it is possible that both parent and child may have congenital hypothyroidism.

Is a child with hypothyroidism more likely to get other diseases later in life?

No, not in the majority of cases.

[illegible]

Not in the normal position.



Endocrine System

A system of glands in the body that secrete substances called hormones into the bloodstream. The endocrine glands include the thyroid, the pituitary, the ovaries and the testicles.

Endocrinologist

A doctor specialising in the treatment of hormone disorders, including thyroid disorders.

Enzyme

A special protein, formed in a living cell, which helps chemical reactions to occur.

Genetic

Pertaining to genes, which are the units on the chromosomes that transmit inheritance of one or more characteristics.

Goitre

A visible swelling of the thyroid gland in the front of the neck.

Hormone

A chemical substance that is made by an endocrine gland and then secreted into the bloodstream. There are a large number of hormones that have widespread effects on the body, such as thyroid hormone, growth hormone, insulin and cortisol.

Hypoplasia

An organ that has failed to develop fully or reach its normal size.

Hypothalamus

Part of the base of the brain that controls the release of hormones from the pituitary gland.

Hypothyroidism

A condition in which the thyroid gland is underactive or absent. The term congenital hypothyroidism means that the condition is present at birth.

Jaundice

A yellow colouring of the skin due to an excessive amount of the pigment bilirubin, which is a product of the breakdown of old red blood cells.

Metabolism

All of the processes that occur in the body that turn the food you eat into energy your body can use. It is the chemical activity that occurs in cells, releasing energy from nutrients or using energy to create other substances, such as proteins.

Paediatric Endocrinologist

A doctor who specialises in the disorders of endocrine glands in children.

Paediatrician

A doctor specialising in the treatment of children, including those with congenital hypothyroidism.

Pituitary Gland

An endocrine gland about the size of a pea and situated at the base of the brain.

This gland is very important because it controls the other endocrine glands, including the thyroid.

Primary Hypothyroidism

Hypothyroidism that is caused by disease of the thyroid gland itself.

Thyroid Gland

An endocrine gland that produces thyroid hormone. It is a butterfly-shaped gland that is located in the front of the lower part of the neck and consists of two connected lobes on each side of the windpipe.

Thyroid Hormone

The hormone produced by the thyroid gland (also called thyroxine). The tablets given to treat hypothyroidism contain this hormone. Thyroid hormone is sometimes called T4 or Free T4, particularly on blood test request forms and results.

Thyroid Scan

This is a test to see the position and shape of the thyroid gland and to determine if the gland is absent, ectopic or present in the normal position.

Thyroid Stimulating Hormone (TSH)

A hormone that is produced by the pituitary gland and which stimulates the thyroid gland to secrete thyroid hormone (TSH is also called thyrotropin).

Support Organisations & Further Reading

Further Information

Endocrine Society (USA) Hormone Health Network

<http://www.hormone.org/questions-and-answers/2012/congenital-hypothyroidism>

Consensus guidelines on screening, diagnosis, and management of congenital hypothyroidism.

<http://press.endocrine.org/doi/abs/10.1210/jc.2013-1891>

Support Organisations

CH Kids - Congenital Hypothyroidism Australia

<http://chkids.ning.com/>

Thyroid Australia Ltd

<http://www.thyroid.org.au/>

Newborn Screening

<http://www.genetics.edu.au/Publications-and-Resources/Genetics-Fact-Sheets/FactSheet20>

The MAGIC Foundation

<https://www.magicfoundation.org/www/docs/1185.2433/congenital-hypothyroidism.html>

British Thyroid Foundation

<http://www.btf-thyroid.org/>



Record of Clinical Visits

The following table may be used to keep a record of your child's visits to the doctor, test results and thyroid hormone doses.

Date	Age	Clinical Review		Blood Tests	
		Weight (kg)	Height (cm)	TSH	T4

Treatment		Comments
Current Dose of Thyroxine	New Dose of Thyroxine	



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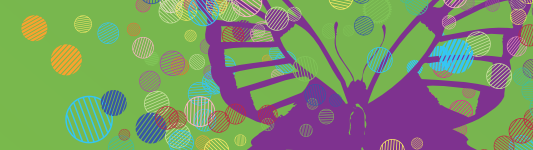
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