

# Hormones and Me Congenital Hypothyroidism



**ANZSPED**

AUSTRALIA AND NEW ZEALAND  
SOCIETY FOR PAEDIATRIC  
ENDOCRINOLOGY AND DIABETES

# Congenital Hypothyroidism



## Table of Contents

About this book	2
Introduction	3
Hormones	4
The Thyroid Gland	5
Types of Congenital Hypothyroidism	10
Newborn Screening	14
Investigations to Determine the Type of Hypothyroidism	16
Treatment	18
Outlook for Babies with Permanent Congenital Hypothyroidism	21
Questions and Answers	22
Glossary	25
Support Organisations & Further Reading	29
Record of Clinical Visits	30
References	32
The Hormones and Me Booklet series	34

## About this book

This booklet, *Congenital Hypothyroidism*, aims to provide a basic understanding of how the thyroid gland works, 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 is 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 2023 by members of the Thyroid working group of the Australia and New Zealand Society for Paediatric Endocrinology and Diabetes (ANZSPED): Dr Natasha Heather, A/Prof Michelle Jack, Dr Yoon Hi Cho, Dr Joel Vanderniet, Dr Sarah McMahon and Dr Sathyakala Vijayanand.

Prof Margaret Zacharin (Royal Children's Hospital, VIC, Australia) and Dr Ann Maguire (The Children's Hospital at Westmead, NSW, Australia) previously reviewed the Hormones and Me series on behalf of the Australasian Paediatric Endocrine Group (APEG), now known as ANZSPED.

This booklet was first updated and reproduced in 2006 with the help of A/Prof Maria Craig (The Children's Hospital at Westmead), who also revised it in 2011. Originally published in 1987 by the Royal Children's Hospital, VIC, as 'Congenital Hypothyroidism. An Information Guide for Parents'. Special thanks to the original authors and editors, Dr Joan Coakley (The Children's Hospital at Westmead, NSW, Australia), Dr John Connelly (Royal Children's Hospital, VIC, Australia), Dr Philip German (Monash Medical Centre, VIC, Australia) and Jane Drury.



## Introduction

The term congenital hypothyroidism (CH) describes a condition that is present at birth (congenital) and in which the thyroid gland is underactive or absent.

Congenital hypothyroidism occurs in approximately 1 in 3,500 babies, so each year in Australia about 70 babies are born with this condition.

At the time of birth, there are usually no visible signs of any medical issues in these babies. Therefore it may be a shock to parents to be told that their baby's thyroid gland is not functioning properly. It may also be difficult, at first, to absorb all the information given about hypothyroidism.

In the past, children born with congenital hypothyroidism often had serious problems because of late diagnosis. With the introduction of newborn screening in the 1970's in New Zealand and Australia, the outlook for those children changed dramatically. Early treatment allows them to lead normal healthy lives.

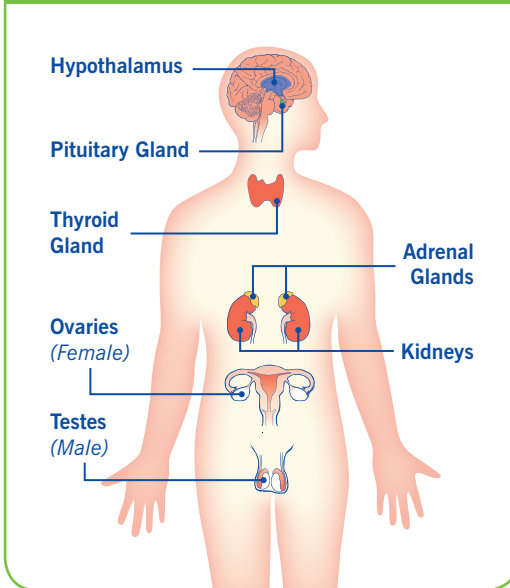
This booklet has been written to help you learn about congenital hypothyroidism. It explains the function of the thyroid gland; the different types of hypothyroidism; the newborn screening program and how it operates; and the treatment and outlook for babies with this condition. The final section of the booklet addresses some of the most commonly asked questions by parents concerning congenital hypothyroidism.

# Congenital Hypothyroidism

## 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: Thyroid and adrenal glands in the body



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

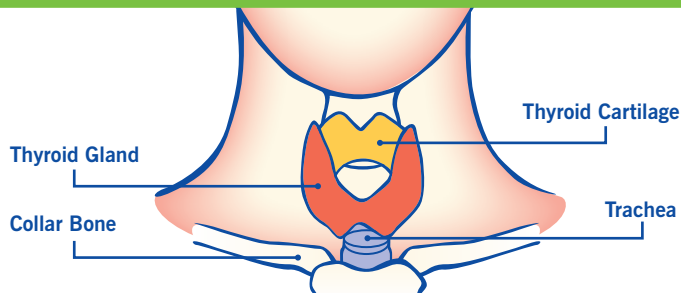
them have a direct action on the body, such as growth hormone, while others stimulate hormone production in other glands in the body, such as the thyroid gland and adrenal glands (see *Diagram 1*).

## The Thyroid Gland

The thyroid is a butterfly-shaped gland located in the front of the lower part of the neck. It consists of two connected lobes on each side of the windpipe (trachea) (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 migrates 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 is able to produce thyroid hormone (also called thyroxine) by this time. Early in the pregnancy, the baby receives thyroid hormone from the mother.

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 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



# Congenital Hypothyroidism

## Function and regulation of the thyroid gland

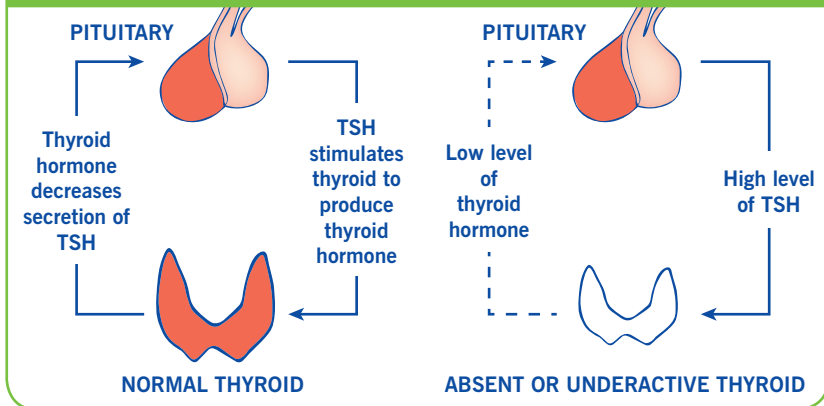
The function of the thyroid gland is to make thyroid hormone and secrete it into the bloodstream. Thyroid hormone is involved in regulating the body's metabolic rate, which is the speed at which the body breaks down (metabolises) food to provide the energy for many normal daily bodily functions. The 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 secretion of TSH to a normal level. If the thyroid is secreting too much thyroid hormone, this causes the pituitary gland to reduce its secretion 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' on page 10.



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



The control of thyroid hormone secretion is illustrated in *Diagram 3*. In the underactive thyroid diagram the thyroid is small and misshapen. This is one cause of an underactive 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 growth in the first two years of life as well as for brain function throughout life. Thyroid hormone is important in regulating the body's metabolic rate, so that when there is a lower level (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

# Congenital Hypothyroidism

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:

- 1 Sluggish physical and mental activity**
- 2 Low body temperature**
- 3 Slow pulse rate**
- 4 Intolerance of cold**
- 5 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 vital for normal growth and development before and after birth. Because the unborn baby also received thyroid hormone from the mother, most babies with congenital hypothyroidism will have normal growth measurement at birth, including their weight and length. However, if a baby's hypothyroidism is not treated, they will remain small through infancy and childhood and end up being very short. This slowness of growth not only affects the skeleton but all parts of the body.

## Slow mental development

Development of the brain is dependent on normal levels of thyroid hormone and intellectual impairment can occur if the level is low. The degree of impairment depends on how severe the deficiency is. When the thyroid hormone level is only slightly lowered, there may be little or no effect on mental function. When thyroid hormone is completely absent



and the baby receives no treatment, mental retardation may be severe. However, this will not occur if treatment is begun early. (See the 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 newborn babies to have mild jaundice which may last for a week or two and is usually not harmful. In untreated hypothyroidism, jaundice may persist for much longer but once thyroid hormone treatment is started, the jaundice will ordinarily disappear quickly.

Other features may be seen including excessive sleepiness, poor feeding and weight gain, dry skin, puffy face and a hoarse cry.

## 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 absent or has not formed properly or is in an abnormal position.

#### **Absent thyroid gland (thyroid agenesis or athyreosis)**

In babies with thyroid agenesis, the thyroid gland has failed to develop before birth. It is absent and will never grow. As there is no thyroid gland, no thyroid hormone can be produced. About 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. The reason why the thyroid gland failed to develop in these babies is at present unknown, but some cases have a genetic basis.



### **Abnormally positioned thyroid gland (ectopic thyroid)**

The term ectopic means in an abnormal position. The thyroid does not occupy its normal position in the neck and is also small and abnormally formed. 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. Most ectopic thyroids are very small and underactive, but occasionally some can 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 descend to its normal position. In fact, its function will often deteriorate further over time. Sometimes its function is enough to be normal at birth so that the newborn screening tests are normal, then it may gradually decrease in childhood.

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

### **Abnormally formed 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, abnormally formed 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

The prefix dys means 'abnormal' and genesis means 'production of'.

Babies with thyroid dyshormonogenesis have a normally positioned gland that cannot produce a normal amount of thyroid hormone. 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). This condition accounts for about 30% 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 for life. In some children, thyroid dyshormonogenesis arises due to genetic changes that can be associated with other clinical features.

### Permanent Dyshormonogenesis

As mentioned previously, thyroid hormone is made in a number of steps. Each step is controlled by a particular protein called an enzyme. Occasionally one of these enzymes is missing or not working properly, and this is what causes permanent dyshormonogenesis. This is usually an inherited condition and therefore other babies born into the family are also at risk of having it. Usually the chance of this is 1 in 4. Boys and girls are equally 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, so it is important to tell the doctor about any family history of thyroid disease.



Another cause of transient dysmorphogenesis is the presence of too much iodine in the body. Iodine is essential for the manufacture of thyroid hormone and iodine deficiency can lead to hypothyroidism or goitre in older children and adults.

However, too much iodine can also decrease the function of the thyroid gland. A baby may be at risk of becoming hypothyroid if the mother takes a lot of medication containing iodine during pregnancy and/or while breast feeding. Occasionally the use of large amounts of iodine containing antiseptics used on the baby's skin can block the thyroid gland. This is more common in sick, premature babies. However, once the excessive iodine intake stops, the thyroid gland will usually return to normal and the baby will not suffer any long-term ill effects.

### 3. Central Hypothyroidism

In this condition, the thyroid gland has developed 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. The condition only accounts for less than 5% of all cases of congenital hypothyroidism and occurs in about 1 in 16,000 to 1 in 50,000 newborns.

Central hypothyroidism usually occurs together with a deficiency of other pituitary glands hormones and occasionally with brain abnormalities or other uncommon medical conditions. Some of these conditions are linked to a gene defect, which can be hereditary.

## Newborn Screening

Newborn blood spot screening is offered free of charge to all babies born in Australia and New Zealand. Congenital hypothyroidism is the most common disorder detected by newborn blood spot screening.

Between one and three days after birth, a small amount of blood is collected from the baby's heel and placed on a piece of absorbent paper. This blood spot is forwarded to a central screening laboratory for measurement of TSH.

A high level of TSH in the blood spot identifies babies who are likely to have congenital primary hypothyroidism (that is, congenital hypothyroidism due to dysgenesis or dyshormonogenesis). It is not possible to detect all affected babies through a screening test. The test does not identify babies with central hypothyroidism and will occasionally miss babies with primary hypothyroidism if the abnormal thyroid gland is still functioning briefly in the early days of life (see ectopic thyroid).

If the TSH level is high, babies are referred to a specialist doctor for a blood test to confirm that they have congenital hypothyroidism. Babies with congenital hypothyroidism should start treatment as soon as possible.





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

- It is very difficult to diagnose congenital hypothyroidism at birth just by examining a baby. Some of the features described previously, such as slow growth and impaired mental development, are not present yet. In fact, it may be months before they can be seen. Therefore if newborn screening is not carried out, many cases of hypothyroidism will be missed at birth and treatment delayed, leading to an unacceptable risk of impaired brain and physical development.
- Congenital hypothyroidism is not a rare condition. About 70 new cases are diagnosed in Australia each year (1 in 2,000 births).

With early diagnosis and treatment almost all infants with congenital hypothyroidism, including those affected severely, will develop normally, both physically and mentally.

## Investigations to Determine the Type of Hypothyroidism

When 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 as soon as possible. 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 they have.

At the first visit, some special investigations are also carried out including a blood test to confirm the results of the newborn screening. Sometimes the blood sample is analysed to check for jaundice (elevated bilirubin).

Another investigation that is usually performed at this time 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. There is virtually no risk associated with the use of technetium because only a low dose is used. In addition, its radioactivity disappears within hours and technetium is excreted from the body very quickly.

A thyroid scan should be performed when the baby is not already being treated for hypothyroidism. This is why it is usually done at the first visit. Treatment should not be delayed while waiting for imaging.



Many babies will also have a thyroid ultrasound. This shows whether the thyroid is present or not and if the size is normal. The combined results of the thyroid scan and thyroid ultrasound may assist in establishing the underlying diagnosis.

For some babies with thyroid dysmorphogenesis (in which the thyroid gland is present but not functioning normally), more detailed investigations may be carried out after two or three years of treatment. Occasionally a hearing test is needed, as hearing problems are more common in babies with hypothyroidism, especially those with dysmorphogenesis. Increasingly, genetic causes have been identified.

## Treatment

In all forms of hypothyroidism, the thyroid gland is unable to make enough thyroid hormone for the body's needs, so this hormone must be replaced. It is given as a tablet by mouth once a day. The tablet can be crushed and dissolved in a small amount of breast milk, formula or water and given by syringe placed at the back of the mouth before a feed. It should NOT be put into a bottle, in case the infant doesn't finish the bottle. The thyroid hormone in the tablets is exactly the same as the thyroid hormone that is naturally made 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, which is used for the treatment of diabetes.

The dose of thyroid hormone that is required varies with body size and, as the baby grows, the dose usually needs to be increased. The dose may also need to be changed if the baby is changed to a soy-based formula. There are other factors that may affect absorption of thyroid hormone that your doctor will discuss with you.

Once treatment has been started, blood tests are done at regular intervals to measure the amounts of thyroid hormone and TSH in the blood. These tests are done more frequently (usually every one to three months) while the baby is young and less frequently (usually every three to six months) when they are a toddler and child. The frequency of testing depends on various factors, such as a child's age and whether there has been a recent change in dose.



The aim is to keep the thyroid hormone level in the blood in the normal range. With appropriate treatment, the pituitary gland no longer needs to make large amounts of TSH. Therefore the amount of TSH in the blood will drop to a normal level, though sometimes this may take several weeks.

If treatment is inadequate (for example, if the dose is too low or the tablets are not being given or absorbed properly) 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.

For most babies with transient hypothyroidism, treatment with thyroid hormone is given for a variable amount of time. It may be difficult initially to tell whether the under-activity of the thyroid gland is transient or permanent. The treatment may be needed for several years, or sometimes may not be required at all.

It is important to store thyroid hormone tablets as instructed on the packaging to ensure they don't lose their potency. Some brands require refrigeration but can be kept out of the fridge for up to 2 weeks if needed (ie. when travelling). Other brands don't require refrigeration, but should be stored in a cool, dry place (below 25°C). For tablets that come in a bottle, keep the lid closed tightly when not in use and ensure that no moisture builds up in the bottle. It is important to speak to your doctor before changing brand of thyroid hormone tablets, as extra monitoring may be required after changing.

# Congenital Hypothyroidism

## Follow-up

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

The doctor will monitor your child's growth and development and check for any signs of under-treatment or over-treatment. They will arrange regular blood tests and make sure the appropriate dose of thyroid hormone is being given. 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, the diagnosis was frequently delayed and, as a result, some infants suffered permanent brain damage, resulting in intellectual disability. With early diagnosis, 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, so there is a range of intellectual abilities among hypothyroid children. In addition, a small proportion of children with hypothyroidism have other medical problems such as hearing impairment, 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.

## Questions and Answers

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

It is best to crush the thyroid hormone tablet on a spoon, mix it with a small amount of liquid (water, breast or formula milk). The liquid can be given via spoon or with a syringe.

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

In one study it was shown that soy-based formulas seemed to reduce the absorption of thyroid hormone in the intestine, necessitating larger doses. This is the only food that may cause a problem with absorption, so if a baby is changed to a soy-based formula it would be wise to let the doctor know.

### **Are there any medicines that may interfere with the absorption of thyroid hormone tablets?**

If a baby or child has to take iron supplements, iron causes the thyroid hormone to be poorly absorbed. Iron supplements must be given at the opposite end of the day to thyroxine.

### **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 a dose is missed occasionally. However, if doses are repeatedly missed, this will result in inadequate thyroid hormone levels. It is important to ensure you don't run out of medication by obtaining a repeat prescription from the doctor before supplies get low. If the baby vomits within half an hour of receiving the dose of thyroid hormone, the same dose should be repeated 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 must take other medication?**

No. Children with hypothyroidism can have the usual immunisations. It is important to check with your doctor if your child has been started on any other medication.

### **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; agenesis (absent), hypoplasia (small or malformed) or ectopic thyroid (developed in an abnormal position). Because these problems are not usually inherited the risk of a subsequent child having a similar condition is small.

# Congenital Hypothyroidism

If the baby has dysmorphogenesis 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').

**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 dysmorphogenesis 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.

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

No, in most cases.



## Glossary

### **Agnesesis**

Absence or failure of an organ or part of the body to develop normally (similar to aplasia).

### **Aplasia**

Lack of development of an organ or tissue.

### **Athyrosis**

Absence of the thyroid gland (sometimes spelt athyreosis).

### **Bilirubin**

A product of the breakdown of red blood cells.

### **Central Hypothyroidism**

Hypothyroidism that is due to abnormal formation or function of the hypothalamus and/or pituitary gland.

### **Congenital**

Present at birth

### **Dyshormonogenesis**

Abnormal production of a hormone (dys means 'abnormal' and genesis means 'production of')

### **Ectopic**

Not in the normal position.

# Congenital Hypothyroidism

## 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.

# 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

**Australia and New Zealand Society for Paediatric Endocrinology  
and Diabetes (ANZSPED)**

[www.anzsped.org](http://www.anzsped.org)

**Australian Thyroid Foundation**

[www.thyroidfoundation.org.au](http://www.thyroidfoundation.org.au)

**The Magic Foundation**

[www.magicfoundation.org](http://www.magicfoundation.org)

**Thyroid Australia**

[www.thyroid.org.au](http://www.thyroid.org.au)

## Record of Clinical Visits

The following table may be used to keep a record of your child’s visit 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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Hormones and Me

# Congenital Hypothyroidism

This booklet is valuable reading for anyone with  
Congenital Hypothyroidism.

It is also recommended reading for their family and friends.