CONGENITAL HYPOTHYROIDISM
A GUIDE FOR PARENTS

This booklet is essential reading for the families of children diagnosed with congenital hypothyroidism. It is also recommended reading for children with this condition.
DISCLAIMER - speak to an appropriate healthcare professional

The information contained in this booklet is a general guide only and should not be relied upon, or otherwise used, in place of medical advice. Any medical information contained in this booklet is not intended as a substitute for informed medical advice. You should consult with an appropriate healthcare professional on (1) any specific problem or matter which is covered by any information in this booklet before taking any action; or (2) for further information or to discuss any questions or concerns.

Whilst we have taken reasonable steps to ensure the accuracy of the contents of this booklet, it is provided on the terms and understanding that Serono Symposia International, Serono Australia Pty Ltd and Serono S.A. (and their respective officers and employees) and all other persons involved in the writing, development, publication, distribution, sponsorship or endorsement of this booklet, to the fullest extent permitted by applicable law, are not responsible for (1) any error or any omission from this booklet; (2) make no warranties, representations or give any undertakings either express or implied about any of the content of this booklet (including, without limitation, the timeliness, currency, accuracy, correctness, completeness or fitness for any particular purpose of the booklet or its content); (3) are not responsible for the results of any action or inaction taken on the basis of any information in this booklet; (4) are not engaged in rendering any medical, professional or other advice or services; (5) expressly disclaim any and all liability and responsibility to any person in respect of anything done by any such person in reliance, whether wholly or partially, upon the whole or any part of the contents of this booklet.
About this book

Serono Symposia International is pleased to bring you *Congenital Hypothyroidism – A Guide for Parents* as a service to the Australasian community. It forms part of the ‘Hormones and Me’ educational booklet series.

This booklet 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 the booklet.
This booklet is an updated version of ‘Congenital Hypothyroidism. An Information Guide for Parents’, published by the Royal Children’s Hospital Victoria in March 1987 and was reviewed and edited by Dr Maria Craig (The Children’s Hospital Westmead, NSW, Australia), a Paediatric Endocrinologist specialising in childhood endocrine disorders and a member of the Australasian Paediatric Endocrine Group. It has been retitled Congenital Hypothyroidism – A Guide for Parents.

Special thanks to the original authors, Dr John Coakley (Children’s Hospital Westmead) and Dr John Connelly (Royal Children’s Hospital, Victoria) and also to Dr Philip Bergman (Monash Medical Centre, Victoria) and Jane Drury for their invaluable assistance with editing.

4 Introduction
5 Hormones
6 The Thyroid Gland
10 Types of Congenital Hypothyroidism
14 Newborn Screening
16 Investigations to Determine the Type of Hypothyroidism
18 Treatment
20 Outlook for Babies with Permanent Congenital Hypothyroidism
21 Questions and Answers
24 Further Information
25 Glossary
29 Support Organisations
30 Congenital Hypothyroidism – Record of Clinical Visits
34 References for Text
36 The ‘Hormones and Me’ Booklet series
37 Copyright
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.

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 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 1970s, the outlook for these children changed dramatically and with early treatment they can now lead normal healthy lives.

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 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.

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 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 (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 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.

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.

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

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

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 low 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 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:

- Sluggish physical and mental activity
- Low body temperature
- Slow pulse rate
- Intolerance of cold
- 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 receives thyroid hormone from the mother, most babies with congenital hypothyroidism will have normal growth measurements 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.**
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 agenesis)**

In babies with this condition, the thyroid gland has failed to develop before birth: it is absent and will never grow. Consequently no thyroid hormone is produced. This condition is called thyroid agenesis or athyreosis. 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 fails to develop in this small number of babies is at present unknown, but some cases may 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 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, but others are 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 become bigger or 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. This accounts for about half of the cases of thyroid dysgenesis and is slightly more common than athyrosis. Again, it is not known for sure why in some babies the thyroid gland remains in an ectopic position, but some of the same factors that cause athyrosis 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.
Types of Congenital Hypothyroidism

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. 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 for 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

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, and this is what 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 the chance of this is 1 in 4 and 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. Women who have had abnormal thyroid function themselves, either an underactive thyroid gland or an overactive gland (the condition known as Graves’ disease), are at particular risk of this happening, so it is important to tell the doctor about any family history of thyroid disease.

Another cause of transient dyshormonogenesis 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.

On the other hand, 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 on the baby’s skin can block the thyroid gland. 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.

In a few cases of thyroid dyshormonogenesis, both permanent and transient, the precise cause of the condition is unknown.

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 50,000 newborns.

Central hypothyroidism may be associated with a deficiency of other pituitary gland hormones (for example, congenital hypopituitarism), brain abnormalities or other uncommon medical conditions. Some of these conditions are linked to a gene defect, which can be hereditary.
Newborn Screening

All babies born in Australia are now screened for congenital hypothyroidism. There are five centralised screening laboratories located in Western Australia, South Australia (which also covers Tasmania and part of the Northern Territory), Victoria, New South Wales (also covers the Australian Capital Territory) and Queensland (also covers the rest of the Northern Territory). There is a single laboratory for New Zealand.

Between two to five days after birth, a small amount of blood is 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. A high level of TSH in the blood spot identifies babies who have congenital primary 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.

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. 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 3,500 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. A blood test is done to confirm the results of the newborn screening. Sometimes the blood sample is analysed to check for jaundice (elevated bilirubin). The mother may also be given a blood test to check for antibodies, if there is a risk that the baby’s hypothyroidism is a result of thyroid disease in the mother, but it is not routine to test the mother’s blood. Usually the baby’s bones are x-rayed because the growth of the skeleton can be delayed in hypothyroidism.

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 and to determine if the gland is absent, ectopic or present in the normal position. 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 only be performed when the baby is not already being treated for hypothyroidism. This is why it is usually done at the first visit.

Many babies will also have a thyroid ultrasound. 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.

For some babies with thyroid dyshormonogenesis, more detailed investigations are carried out after two or three years of treatment to determine if they have an enzyme deficiency. Occasionally a hearing test is needed, as hearing problems are more common in babies with hypothyroidism, especially those with dyshormonogenesis.
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. It 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.

The dose of thyroid hormone that is required varies with body size and 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 three to six weeks after starting thyroid hormone treatment, then every two to three months for the first two to three years of life, and at about four to six month intervals thereafter. The frequency of testing depends on various factors, such as a child’s age and whether there has been a recent change in dose.

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 aim is to keep the thyroid hormone level in the blood at about the upper limit of normal. With appropriate treatment, the pituitary gland no longer needs to secrete large amounts of TSH. Therefore the amount of TSH in the blood will drop to the normal level, though sometimes 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) 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 period because it may be difficult initially to tell whether the under activity of the gland is transient or permanent. The treatment may be needed for several years, or sometimes may not be required at all.

Tablets should be kept in the fridge, with the lid of the bottle closed tightly. Repeated opening and closing of the lid can lead to condensation in the bottle and may alter the effect of the tablets. For this reason, new packaging is being introduced consisting of five bottles each containing 40 tablets. Tablets can be removed from the fridge and stored at room temperature (below 25°C) for up to four weeks if needed (for example when travelling).

Follow-up

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

The doctor will want to make sure that the appropriate dosage of thyroid hormone is being prescribed and that there are no signs of under-treatment or over-treatment. 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 mental retardation. With early diagnosis, this can be prevented for the majority of children. 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. 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, 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 tablet on a spoon, mix it with a little milk or sterile water, then place the mixture in the baby’s mouth. Alternatively, an eye dropper can be used.

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.

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.
Questions and Answers

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; 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 very small. In the general population, the likelihood of a baby being born with an absent or malformed thyroid gland is about 1 in 3,500. 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.

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.

If a baby is one of the few with permanent dyshormonogenesis 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 dyshormonogenesis, 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 dyshormonogenesis 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 the possibility of both parent and child having congenital hypothyroidism cannot be completely discounted. Nevertheless it is reassuring to note that of all the children so far diagnosed in the Victorian screening program, only one has a parent who was also born with congenital hypothyroidism over a 20 year period.

Is a child with hypothyroidism more likely to get other diseases later in life?
No, in the majority of cases.
Further Information

Australasian Paediatric Endocrine Group Position Paper: Guidelines for Diagnosis, Management and Follow-up of Children with Congenital Primary Hypothyroidism
www.racp.edu.au/apeg/cph.htm

Australian Prescriber – Taking care of thyroxine
www.australianprescriber.com/cc/95

The Hormone Foundation: Thyroid Information
www.hormone.org/public/thyroid.cfm

Other
www.ghr.nlm.nih.gov/condition=congenitalhypothyroidism
www.magicfoundation.org/www
www.thyroid.org

Glossary

Agenesis
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.
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

Thyroid Australia Ltd
333 Waverley Road
Mount Waverley VIC 3149
Australia
Tel: (03) 9888 2588
Email: support@thyroid.org.au
www.thyroid.org.au/

The Australian Thyroid Foundation Limited
PO Box 186
Westmead NSW 2145
Australia
Tel: (02) 9890 6962
Email: thyroid@icpmr.wsahs.nsw.gov.au

Thyroid Federation International
797 Princess Street
Suite 304
Kingston, ON K7L 1G1
Canada
Tel: +1 613 544 8364
Email: tfi@on.aibn.com
www.thyroid-fed.org/
The following table may be used to keep a record of your child's visit to the doctor, test results and thyroid hormone doses.

<table>
<thead>
<tr>
<th>Date</th>
<th>Age</th>
<th>Clinical Review</th>
<th>Blood tests</th>
<th>Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>Weight (kg)</td>
<td>TSH</td>
<td>Current Dose of Thyroxine</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Height (cm)</td>
<td>T4</td>
<td>New Dose of Thyroxine</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Comments</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

The table includes columns for Date, Age, Clinical Review (Weight, Height, Comments), Blood tests (TSH, T4), and Treatment (Current Dose of Thyroxine, New Dose of Thyroxine).
## Congenital Hypothyroidism – Record of Clinical Visits

<table>
<thead>
<tr>
<th>Date</th>
<th>Age</th>
<th>Clinical Review</th>
<th>Blood tests</th>
<th>Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>Weight (kg)</td>
<td>TSH</td>
<td>Current Dose of Thyroxine</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Height (cm)</td>
<td>T4</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Comments</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>


References for Text


The ‘Hormones and Me’
Booklet Series

Congenital Hypothyroidism — A Guide for Parents is one in a series of Patient Education Library booklets on Metabolic Endocrinology, which aims to provide people with a better understanding of the issues relating to paediatric growth.

If you would like more information about other titles available or have any suggestions for other topics in the ‘Hormones and Me’ booklet series, please contact Serono Symposia International on (+ 61 02) 8977 4163.

The ‘Hormones and Me’ series includes:-

- Intrauterine Growth Retardation (IUGR)
- Puberty and its Problems
- Delayed Puberty
- Craniopharyngioma
- Multiple Pituitary Hormone Deficiency (MPHD)
- Turner Syndrome
- Congenital Adrenal Hyperplasia (CAH)
- Growth Problems in Children
- Diabetes Insipidus
- Management of Emergency or ‘Stress’ Situations where Hypoglycaemia or Cortisol Deficiency Occur
- Growth Hormone Deficiency in Adults
- Klinefelter Syndrome