

Congenital Hypothyroidism (CH)

(endocrine condition)

Also known as:

- congenital myxedema
- cretinism

What is CH?

CH is a condition in which thyroid hormone deficiency is present at birth. Thyroid hormone is essential for normal growth and brain development. Without treatment infants may have developmental delay and growth retardation.

What causes CH?

Most cases result from a failure of the thyroid gland to develop normally. A minority of cases result from defects in the synthesis of thyroid hormone by the thyroid gland.

How common is CH?

The incidence of CH is about 1 in every 3,000 to 4,000 infants born in Canada.

What are the clinical features of CH?

Most newborns with CH are asymptomatic. A minority may have prolonged neonatal jaundice, hoarse cry, constipation, umbilical hernia, and large tongue.

What is the screening test for CH?

Increased thyroid stimulating hormone (TSH) concentration is detected on the newborn blood spot screen. The screening will not identify newborns who have TSH deficiency. Transient forms of increased TSH as the result of transfer of maternal antibodies may be detected. Newborn blood spot screening is not 100% perfect. Infants with clinical symptoms need timely assessment and diagnostic testing even if their screen result is normal.

How is the diagnosis confirmed?

The diagnosis of CH is confirmed by measurement of serum TSH and free T4. An ultrasound or thyroid scan may also be carried out to assess the size, shape or location of the thyroid gland in determining the underlying cause for the deficiency.

How is CH treated?

Thyroid hormone replacement with L-thyroxine provides an effective treatment for CH. The treatment must be started as soon as possible after birth. Parent education is an important component of treatment, including advising parents on actions to be taken when their child develops a minor illness. The treatment is lifelong.

What is the outcome of treatment of CH?

With early treatment and regular monitoring of TSH and free T4, the outcome is excellent for normal growth and development.

Is CH inherited?

Most cases of CH are transient with low risk of recurrence. A minority of patients with CH are missing enzymes involved in the synthesis of thyroid hormone. These conditions are inherited as autosomal recessive traits.

For additional resources, please call:

Pediatric Endocrinology

Stollery Children's Hospital
1C4, 8440 – 112 St. NW
Edmonton, AB T6G 2B7
Phone: 780-407-8249
Fax: 780-407-1509

Emergency consultations:

Phone 780-407-8822 and ask for the specialist on call for pediatric endocrinology.

Pediatric Endocrine Clinic

Alberta Children's Hospital
28 Oki Drive NW
Calgary, AB T3B 6A8
Phone: 403-955-7003
Fax: 403-955-7639

Emergency consultations:

Phone 403-955-7211 and ask for the specialist on call for endocrine diseases.

Early screening, detection and treatment – every infant, every time

For more information about the NMS Program, visit www.ahs.ca/newbornscreening
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