Alberta Health Services
Alberta Newborn Screening

Talking with Parents > When talking to a parent whose baby has received an abnormal screen result, let them know that an abnormal screen result doesn't mean that the infant has the condition. It means that they have a higher chance of having the condition, and they need more tests to find out for sure. A printable sheet for parents with more condition specific information can be used to support your conversation, visit ahs.ca/nms and under Quick Reference click on *What conditions are screened for?*

Congenital Hypothyroidism (CH)

(endocrine condition)

Information for Health Professionals

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 and growth delays.

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 infants 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 infants who have TSH deficiency. Transient forms of increased TSH as the result of transfer of maternal antibodies may be detected. Newborn blood spot screening will not detect all infants with CH. 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. The treatment is lifelong in most cases. 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 sporadic with low risk of recurrence. A minority of individuals with CH are missing enzymes involved in the synthesis of thyroid hormone. These conditions are inherited as autosomal recessive disorders.

Additional resources are available through:

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 pediatric endocrinology.

Early screening and follow-up care – every baby, every time

For more information about the Alberta Newborn Screening Program, visit www.ahs.ca/newbornscreening