



CH **(Congenital Hypothyroidism)**

What is it?

Congenital hypothyroidism is an inherited disease caused by the thyroid gland not making enough thyroid hormone. Thyroid hormones play a vital role in body growth and brain development. It acts as a chemical messenger to help control the body's metabolism and calcium balance. In the first hour after birth, there is a surge in the thyroid stimulating hormone (TSH) release, probably caused by a drop in the infant's temperature. This leads to a rise in both T3 and T4. These elevated levels may last four to six weeks. CH may also be referred to as CHT, cretinism, endemic cretinism (iodine deficiency), or congenital myxedema.

What causes it?

In most cases, there is no specific reason why the thyroid gland did not develop normally; however, CH can be the result of a number of underlying causes, such as:

- 1) Missing or misplaced thyroid gland.
- 2) Hereditary causes.
- 3) Maternal iodine deficiency.
- 4) Maternal thyroid condition and medications.

What are the symptoms of CH?

Most neonates are asymptomatic, though a few can manifest some clinical features, such as:

- 1) Prolonged jaundice.
- 2) Puffy face.
- 3) Delayed passage of stools.
- 4) Large anterior/posterior fontanel.
- 5) Macroglossia.
- 6) Poor growth.
- 7) Poor weight gain.

- 8) Cold hands or feet.
- 9) Hypothermia.
- 10) Hypotonia.
- 11) Lethargy.
- 12) Poor feeding.
- 13) Respiratory distress in infant weighing less than 5.5 pounds.
- 14) Umbilical hernia.

How is CH detected?

Newborn screening is done on tiny samples of blood taken from the infant's heel 24 to 48 hours after birth. If the test is positive, other tests may be done to confirm that a baby has CH.

What immediate actions may be taken if an infant screens positive for CH?

- 1) Contact family to inform them of newborn screening test result.
- 2) Consult pediatric endocrinologist; referral to endocrinologist if considered appropriate.
- 3) Evaluate infant.
- 4) Initiate timely repeat of newborn screening specimen and/or confirmatory/diagnostic testing as recommended by the specialist.
- 5) Initiate treatment as recommended by the consultant as soon as possible.
- 6) Educate parents/caregivers that hormone replacement therapy prevents mental retardation.
- 7) Follow up as recommended by the specialist.
- 8) Report findings to state newborn screening program.

Diagnostic evaluation

Diagnostic tests may include serum free T4 and thyroid stimulating hormone (TSH); consultant may also recommend total T4 and T3 resin uptake. Sometimes an imaging test of the thyroid, either an ultrasound examination or a thyroid uptake scan is used to help determine the cause for the CH.

What problems can it cause?

Untreated CH causes feeding problems, sleepiness, constipation, jaundice, poor growth, mental retardation and developmental delay. There is a chance that children with CH will have hearing problems. It is very important to follow the doctor's instructions about caring for a child with CH.

How is it treated?

Hypothyroidism is treated by giving the child the missing thyroid hormone (L-thyroxine) and by regular monitoring of growth and thyroid hormone levels. This hormone is usually in the form of a pill and can be crushed into milk for the child to drink. The amount of hormone needed is determined by additional tests. The child may develop normally if treatment begins in the first few weeks of life. If damage to the brain and nerves happen because treatment is delayed, it is usually permanent and cannot be reversed.

CH in children

The child should have a primary care doctor and a pediatric endocrinologist. Treatment for CH is lifelong, and a child with CH should see a doctor regularly.

Contacts

Consultation with Dr. Alan Kenien (pediatric endocrinology) is strongly encouraged.
701.234.2431

Consultation with Dr. John Martsolf (medical genetics) is available. 701.777.4277

North Dakota Department of Health –
Newborn Screening Program
www.ndhealth.gov/newbornscreening
701.328.2493

Where can I find more information?

American Academy of Pediatrics
<http://pediatrics.aappublications.org/cgi/content/abstract/91/6/1203>

American College of Medical Genetics
www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

Genetic Alliance
www.geneticalliance.org

Gene Tests
www.genetests.org

March of Dimes
www.marchofdimes.com

MUMS National Parent-to-Parent Network
www.netnet.net/mums

National Library of Medicine Genetics Home Reference (Congenital Hypothyroidism)
<http://ghr.nlm.nih.gov/condition=congenitalhypothyroidism>

The Thyroid Foundation of America
www.allthyroid.org

University of Iowa – Division of Medical Genetics
www.uihealthcare.com/dept/med/pediatrics/divisions/genetics.html

Children's Special Health Services (CSHS)

State Capitol Judicial Wing
600 E. Boulevard Ave., Department 301
Bismarck, ND 58505-0269
Toll Free: 800.755.2714
701.328.2436
Relay TDD: 701.328.3975
CSHS website: www.ndhealth.gov/CSHS
North Dakota Department of Health website:
www.ndhealth.gov

Family support resources available from CSHS:

- Guidelines of Care Info
- Family Support Packet
- Financial Help Packet
- Insurance Fact Sheet

Family Resources

Family to Family Network
Center for Rural Health
University of North Dakota
School of Medicine and Health Sciences
P.O. Box 9037
Grand Forks, ND 58202-9037
Toll Free: 888.434.7436
701.777.2359
Fax: 701.777.2353
E-mail: NDF2F@medicine.nodak.edu
www.medicine.nodak.edu/crh

Pathfinder Services of ND
Pathfinder Family Center
1600 2nd Ave. SW, Ste. 19
Minot, ND 58701
Toll Free: 800.245.5840
701.837.7500
Relay TDD: 701.837.7501
E-mail: ndpath01@ndak.net
www.pathfinder.minot.com

Family Voices of North Dakota, Inc.
P.O. Box 163
Edgeley, ND 58433
Toll Free: 888.522.9654
701.493.2634
Fax: 701.493.2635
www.geocities.com/ndfv

**This fact sheet has general information. Every child is different and some of these facts may not apply to your child specifically. Certain treatments may be recommended for some children but not others. All children should be followed by a metabolic doctor in addition to their primary-care provider.

Acknowledgement

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Disclaimer

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