

CONGENITAL PRIMARY HYPOTHYROIDISM

Definition

Congenital primary hypothyroidism, or CH, is the most common disorder detected by newborn screening. It is caused by inadequate production of thyroid hormone. Thyroid hormone is important for the normal functioning of all the body's organs and is essential for normal brain development. The most common causes of primary hypothyroidism are total or partial failure of the thyroid gland to develop (aplasia or hypoplasia), or its development in an abnormal location (an ectopic gland). The screening program is not designed to detect less common causes of hypothyroidism such as those caused by pituitary insufficiency.

Incidence

The incidence of congenital primary hypothyroidism is estimated at 1 in 4,000 live births.

Characteristics

Deficiency of thyroid hormone in an infant may result in mental and growth retardation if it is not diagnosed and treated early in life. Many infants with CH may appear clinically normal before three months of age, by which time some brain damage has usually occurred.

When symptoms or signs are present, they may include prolonged neonatal jaundice, constipation, lethargy and poor muscle tone, feeding problems, a large tongue, puffy face, large fontanel, distended abdomen, and umbilical hernia. However, these signs and symptoms are nonspecific for CH, are found in fewer than 30% of neonates with CH, and may be present in infants without the condition. **Therefore, in the newborn, clinical signs and symptoms are not reliable indicators of CH.**

Newborn Screening Methodology

The laboratory method used is a time-resolved fluoroimmunoassay for thyroid stimulating hormone (TSH, Thyrotropin).

Results	Actions
Within Normal Limits ($< 25 \mu\text{IU/mL}$ serum TSH)	Normal report is sent to submitter.
Borderline ($25 - < 60 \mu\text{IU/mL}$ serum TSH)	Borderline report is sent to submitter. 1. Repeat newborn screen. 2. If repeat screen remains elevated, obtain endocrine consult.
Presumptive Positive ($> 60 \mu\text{IU/mL}$ serum TSH)	1. Attending physician will be notified by the program follow-up staff. 2. Repeat newborn screen immediately.

Confirmation

Consultation with a Pediatric Endocrinology Consultant should be made for confirmation and diagnosis, (319) 356-2838.

Treatment and Outcome

Treatment should be initiated immediately upon the confirmation of congenital hypothyroidism. Proper dose adjustment of thyroid hormone replacement therapy is important during infancy, the most critical period of brain development. Therefore, management of thyroid hormone therapy is usually performed in consultation with a Pediatric Endocrinology Consultant. Periodic cognitive testing is also recommended.

Screening Practice Considerations

- Congenital primary hypothyroidism is the most common disorder detected by the program.
- Detection of CH does **not** depend on nutritional factors.
- The normal newborn demonstrates a TSH surge in the first hours of life as an adaptation to the extrauterine environment. To be valid, a specimen must be collected when the infant is at least 24 hours of age. If an infant is tested at less than 24 hours of age, a repeat specimen must be collected within seven (7) days of age, regardless of prior test results.
- Thyroid medications administered to the mother during pregnancy, affect congenital hypothyroidism screening results. Contact the Pediatric Endocrinology Consultant regarding management for these situations, (319) 356-2838.
- Blood collection using preservatives (EDTA) can result in false negative results.
- Prompt confirmatory testing is required even if there is evidence to suggest that one of the situations associated with false positive screens is present. These situations can include early specimen collection and prematurity. The presence of any of these does **not** exclude the possibility of disease.
- This screening test is not designed to detect thyroid binding globulin (TBG) deficiency or causes of hypothyroidism other than congenital primary hypothyroidism. In addition, some infants develop late onset congenital primary hypothyroidism. If TBG deficiency or other forms of hypothyroidism are suspected, or in the presence of clinical symptoms, appropriate exams and studies are indicated. Therefore, in the presence of clinical symptoms, contact the Pediatric Endocrinology Consultant.
- TSH ranges apply to the newborn period. Interpretation of results from specimens collected after the newborn period should be performed in consultation with the Pediatric Endocrinology Consultant.

Other Sites of Reference

The MAGIC Foundation

Website: www.magicfoundation.org/

National Organization For Rare Disorders - Hypothyroidism

Website: www.rarediseases.org/search/rdbdetail_abstract.html?disname=Hypothyroidism

American Academy of Pediatrics' Policy Statement - Newborn Screening for Congenital Hypothyroidism: Recommended Guidelines (RE9316)

Website: www.aap.org/policy/04407.html

The Thyroid Society

Website: www.the-thyroid-society.org/

Thyroid Foundation of Canada

Website: www.thyroid.ca/wehavemoved.html

What should I know about Congenital Hypothyroidism?

Website: www.doh.wa.gov/EHSPHL/PHL/Newborn/chgo.htm

MUMS - National Parent-to-Parent Network

Website: www.netnet.net/mums/

Support Groups

The MAGIC Foundation for Children's Growth (MAGIC)

1327 North Harlem Avenue

Oak Park, IL 60302

Website: www.magicfoundation.org/

(708) 383-0808 or 1-800-362-4423