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Congenital hypothyroidism screening guidelines: A review

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Abstract
Congenital hypothyroidism (CHT) is one of the most common preventable causes of mental retardation. In most case, it is due to defect in thyroid gland and usually asymptomatic at birth. To maximize the early detection of CHT so that pre-symptomatic treatment can be initiated to reduce the long-term morbidity associated with the condition if untreated and minimize **s**econd heel pricks, diagnostic delay and start treatment within 14 days of age of diagnosis and normalize TSH level within 1 month of treatment. The blood spot sample should be taken on day 5 (birth day count as zero) and in exceptional circumstances between day 5—8, for all babies regardless of medical condition, milk feeding and prematurity.

Keywords: Congenital hypothyroidism, screening guidelines, TSH

Introduction
Congenital hypothyroidism (CHT) is one of the most common preventable causes of mental retardation [1]. Incidence rate of about 1 in 3000, and more common in females [2]. Hypothyroidism is due to inadequate thyroid hormone production. In most case it is due to defect in thyroid gland (primary congenital hypothyroidism) as shown in figure # 1.
In transient congenital hypothyroidism, initially there is hypothyroid presentation but later become normal or Euthyroid. It is mainly common in preterm babies, also babies with acute illnesses, on medications, Idiopathic, exposure to iodine-containing compounds in imaging and/or surgery, maternal Ab induced or excess iodine intake.

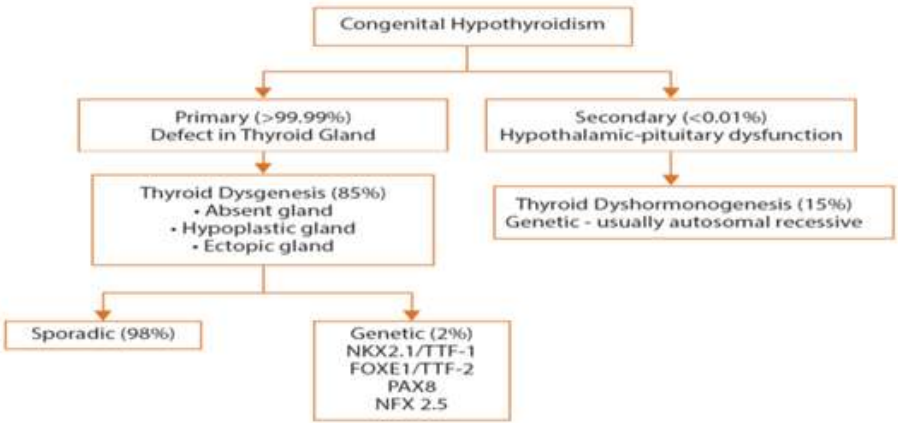

Fig 1: Congenital hypothyroidism- Aetiology

Presentation
It is usually asymptomatic at birth in most neonates, some may present as puffy face, enlarged protruding tongue, cold mottled skin, hoarse cry, large posterior fontanels, umbilical hernia, jaundice, constipation, feeding difficulties and lethargic behavior, hypotonic.

Screening protocols
To maximize the early detection of CHT so that pre-symptomatic treatment can be initiated to reduce the long-term morbidity associated with the condition if untreated and minimize **s**econd heel pricks, diagnostic delay and start treatment within

14 days of age of diagnosis and normalize TSH level within 1 month of treatment.

The blood spot sample should be taken on day 5 (birth day count as zero) and in exceptional circumstances between day 5—8, for all babies regardless of medical condition, milk feeding and prematurity. In Preterm infants (born <32 weeks), current screening will not detect CHT at 5-8 days due to immaturity of the hypothalamic-pituitary axis. In these cases repeat TSH at 28 days of Age or just before discharge whichever comes first.

Unscreened babies

If presenting within 12 month of age, do routine screening. After 12 months of age if the family or GP have any clinical concerns a referral for pediatric assessment and test should be done.

TSH interpretation

1. <10 mU/L: Whole Blood (WB) label as negative, CHT not suspected.
2. ≥20 mU/L: label as positive, CHT suspected and refer to pediatric endocrine team.
3. Between 10—19 mU/L: label as borderline, CHT borderline and repeat test in 7-10 days after the initial sample.

In repeat sample result

1. <10 mU/L: Whole Blood (WB) label as negative, CHT not suspected.
2. ≥20 mU/L: label as positive, CHT suspected and refer to pediatric endocrine team.

Inaccurate results

False negative results may be due to; insufficient sample, compressed sample, preterm babies, Hypothalamic-pituitary hypothyroidism (delay Increase of TSH), blood transfusion. While false positive results due to; early sampling, defect in sampling (Multi-larying) or transient hypothyroidism.

Dose

10-15mcg/kg/day. Target: TSH < 20 mU/L and FT4 increase 2 ng/dl, lower initial dose in at risk of cardiac failure in neonates.

Monitoring

First time recheck Thyroid function test within 1—2 weeks of treatment (at least 4 hours after last dose), then every 2 weeks until normal. Every 1—3 months until 1year age, 2—4 months until 3yrs of age and every 3—12 months once complete growth age [3].

Prognosis

It can be permanent CHT if TSH > 20 mU/L on treatment or confirmed on genetic testing or suspected cases with thyroid dysgenesis on imaging. In suspected cases with normal TSH on treatment or imaging not done, in these cases stop treatment after 2—3 years and recheck TSH after 4—6 week off treatment. If repeat test came normal then label as transient CHT as shown in figure # 2.

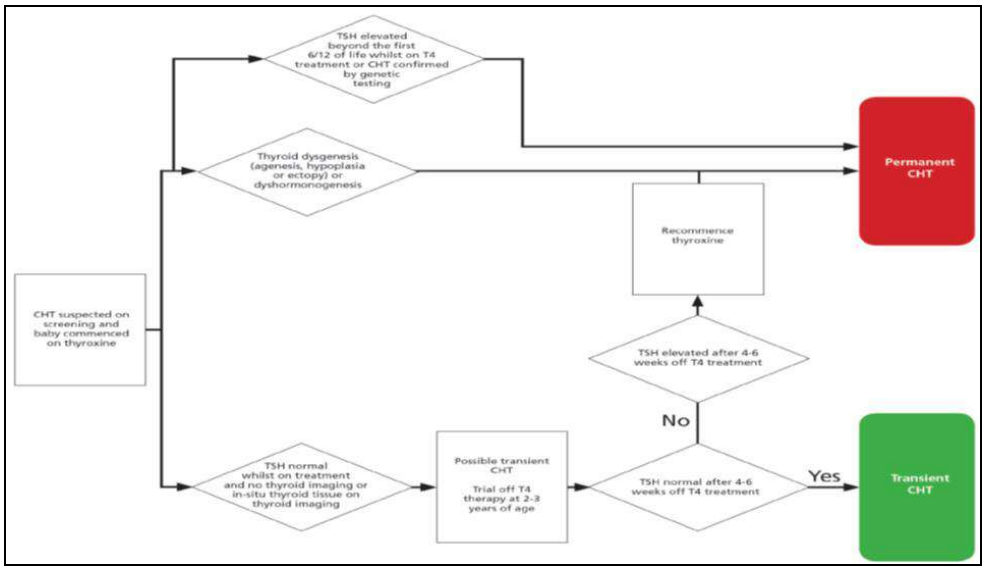

Fig 2: Repeat test came normal then label as transient CHT

References

1. Update of Newborn Screening and Therapy for Congenital Hypothyroidism. American Academy of Pediatric, 2006.
2. NHS Newborn Blood Spot Screening Program guideline updated 2017. https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/753471/Congenital_hypothyroidism_lab_guide.pdf
3. Congenital hypothyroidism. Pediatric Endocrine Society, 2014. https://www.endocrine.org/~/media/endosociety/files/performance-measures/ch_measures-091814_final.pdf?la=en
4. Juliane Léger *et al*. European Society for Pediatric Endocrinology Consensus Guidelines on Screening, Diagnosis, and Management of Congenital Hypothyroidism 2014. The Journal of Clinical Endocrinology and Metabolism (DOI: 10.1210/jc.2013-1891).
5. American Thyroid Association Guidelines. https://www.thyroid.org/congenital-hypothyroidism/
6. Updated AAP Guidelines on Newborn Screening and Therapy for Congenital Hypothyroidism, 2007.