Congenital Hypothyroidism: When To Intervene ? (NNF-TALK)

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Epidemiology

- Incidence 1 in approximately 1000 Indian newborns
- most common preventable causes of mental retardation
- Etiology:
 - Dysgenesis-85%
 - Dyshormogenesis-15%
- Transient or permanent

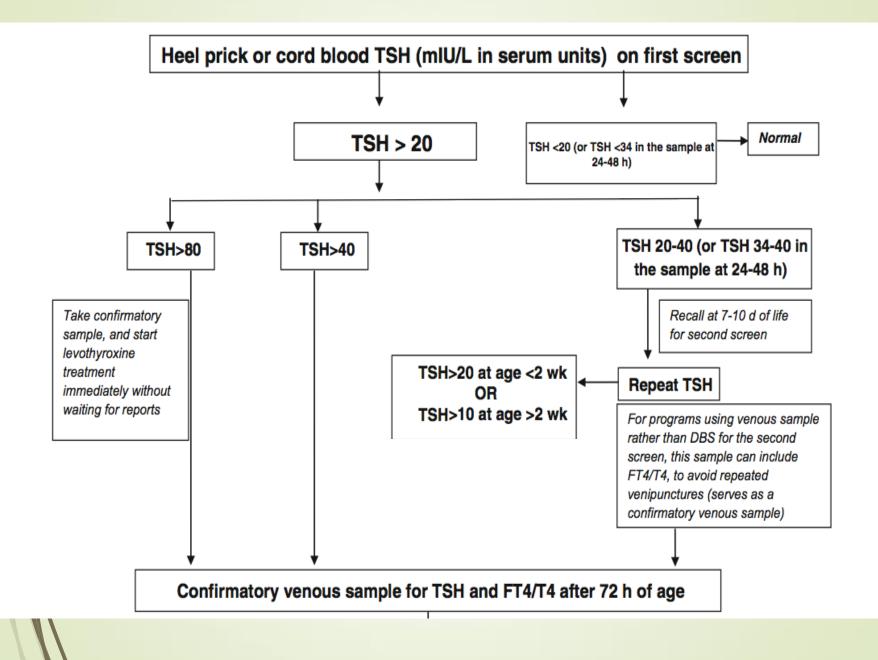
Mathai S. Newborn screening for congenital hypothyroidism- experience from India. Abstract presented at 8th Asia Pacific Regional Meeting of the International Society for Neonatal Screening. New Delhi, Sep 2013.

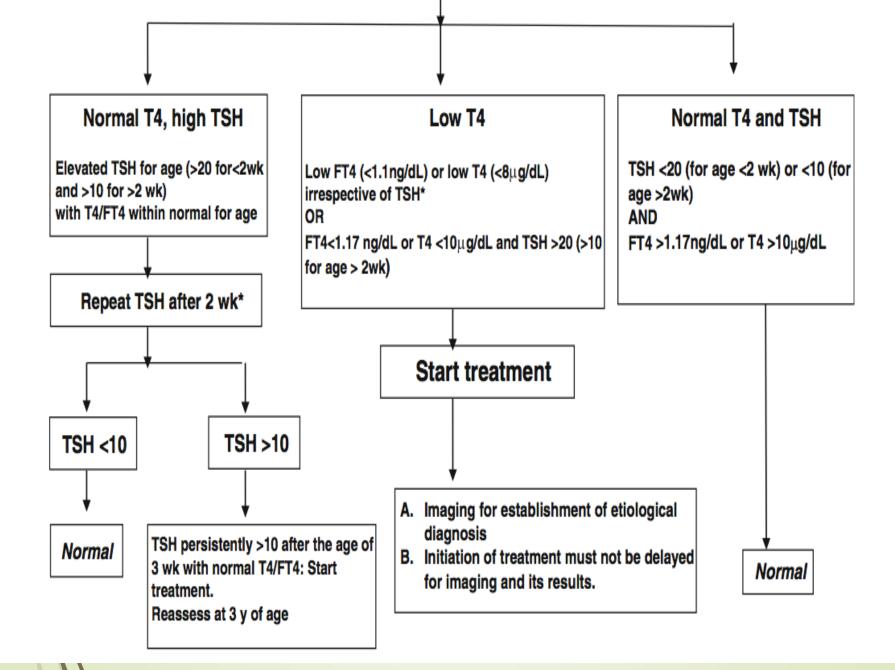
CHANGES AT THE TIME OF BIRTH

- TSH rises to 60-80mU/ml within 30 mins of birth (cooling & stress)
- Followed by a 2-6 fold rise in T₄ to 15-19 µg/dl within 24 hours
 - T₃/rises to about 300 ng/dl
- Stage of physiological hyperthyroidism
- SH surge is short-lived
- Levels decline by 72 hours

Biochemical criteria to initiate treatment (ESPE)

- If capillary TSH neonatal screening: ≥ 40 mU/L whole blood,
- Start treatment (obtain sample), without waiting result,
- Unless venous thyroid function test (TFT) results are available on the same day
- Capillary TSH < 40 mU/l of whole blood, may wait for TFT (provided results available the following day)





Biochemical interventions (ISPAE)

- TSH >20 mIU/L (serum units) is recommended cut-off for recall for cord blood and postnatal screen samples after 48 h of age.
- Screen TSH >40 mIU/L: screen positive cases, immediate recall
 - Mildly elevated TSH from 20 to 40 mIU/L should have a second TSH screen at 7 to 10 d of age.

Criteria on initiation of levothyroxine therapy in term newborns

Low T4 (<100 nmol/L or 8 µg/dL) or low FT4 (<12 pmol/L or <1.1 ng/dL) irrespective of TSH.

Mild low T4 (<128 nmol/L or 10 μ g/dL) or low FT4 (<15 pmol/L or 1.17 ng/dL) in the presence of elevated venous TSH >20 mIU/L if age is <2 wk and >10 mIU/L if age is >2 wk.

Normal T4/FT4 with persistently elevated TSH >10 mIU/L at age>3 wk.

When to screen

- All newborns, preterm and LBW/VLBW infants should undergo routine screening for CH only at 48–72 h postnatal age, not earlier
- Sick neonates should be screened at least by 7 day of age.
- Second screen at 4 wk of age (or at 2 wk of age if discharged early).

Slaughter JL, Meinzen-Derr J, Rose SR, et al. The effects of gestational age and birth weight on false-positive newbornscreening rates. Pediatrics. 2010;126:910–6.

Radioimaging

Ultrasonography
Scintigraphy

 I¹²³ / I¹³¹
 Tc- pertechnetate

Required for etiological diagnosis
Improved diagnostic accuracy
Better to use both together (ISPAE)
Both scintigraphy and ultrasound in neonates with high TSH concentrations (ESPE)

Chang Y, Lee DH, Hong YH, Hong HS, Choi DS, Seo DY. Congenital hypothyroidism: analysis of discordant US and scinti- graphic findings. Radiology. 2011;258:872–9.

Dosing

Standard dose of 10-15 µg/kg/d should be initiated depending upon the severity of CH

- Very low T4, the initial dose should be on the higher side
- Administration at a consistent time of the day is more important than administration on an empty stomach.



Serum T4/FT4 is measured at 2 wk and TSH with T4/FT4 at 1 mth

Then T4/FT4 and TSH are measured every 2 mo till 6mth of age

Every 3mths during 6mth- 3y

Every 3–6 mo thereafter, till completion of growth and puberty.

Thank you