Newborn Critical Care Center (NCCC) Guidelines

Thyroid Screening and Therapy for Congenital Hypothyroidism

BACKGROUND

Congenital Hypothyroidism (CH) is the most common preventable cause of MR. It occurs in 1:2000 to 1:4000 of all newborns, and is increased in VLBW (1:250) and LBW (1:1500) infants.[1] Untreated infants will progress to severe neurodevelopmental impairment as well as delayed bone age with impaired linear growth.

Unfortunately, regardless of the type of approach used, 0.1-1% of newborns with congenital hypothyroidism will have normal screening hormone concentrations due to errors in sample collection and processing, delayed TSH rise and mild forms of disease. This could be higher in individuals with Down Syndrome because of pituitary thyroid axis dissociation, amongst other factors.[2]

Reports of abnormal thyroid function tests in infants that were not identified by newborn screening suggest the need for longitudinal studies to determine if current recommendations should be modified for timely detection of thyroid disease in individuals with Down Syndrome.

INITIAL SCREENING

- Newborn Metabolic Screen: ideally sent at 2-4 days of life; must be at least 24 hours old (fluro-immuno assay)
- T4 and TSH are performed on all specimens by the NC State Laboratory

REPEAT SCREENING

- For VLBW infants, repeat screen at 4-6 weeks of age.
- For infants with congenital heart disease or Trisomy 21, repeat screen at 30 days of age.
- Consider repeat newborn screen at 30 days of life for all infants who remain in NCCC. [3]

NOTE: *VLBW infants as well as infants with CHD or Trisomy 21 are at increased risk for a delayed rise in TSH in response to low serum T4 concentrations.*

TEST RESULTS FOR REPEAT NBS		
TSH (mU/L)	**T4 (µg/dL)**	**Action**
>40	Any value	• Obtain serum Free T4, TSH • Consult Pediatric Endocrinology
>20	Any value	• Obtain serum FT4, TSH • Consult Pediatric Endocrinology
Any value	<5*	• Repeat NBS – see table below

LOW T4 WITH NORMAL TSH:

- Diagnosis may be transient hypothyroxinemia of prematurity (THOP), TBG deficiency or sequelae of a non-thyroidal illness. Consider checking FreeT4 and TSH while awaiting repeat NBS results.

TEST RESULTS FOR REPEAT NBS		
TSH (mU/L)	**T4 (µg/dL)**	**Action**
>15	<5	• Obtain serum FT4, TSH • Consult Pediatric Endocrinology
≤15	<5	• Obtain serum FT4, TSH • Consult Pediatric Endocrinology
≤15	>5	• Normal • Repeat at 4 weeks if birthweight < 1500 grams • C*onsider* repeat at 4-6 weeks if diagnosis of CHD or Trisomy 21 and still inpatient

TREATMENT AND MONITORING

- Begin treatment with levothyroxine (Synthroid)* with guidance from Pediatric Endocrinology on initial dosing, as treatment will depend on the underlying cause of hypothyroidism.
- The timing of additional thyroid function testing should be discussed with Pediatric Endocrinology.
- Arrange out-patient follow up with Pediatric Endocrinology for long term management.

✴ *Synthroid should never be compounded. It needs to be in tablet form then crushed into a small amount of formula/breast milk (do not use soy-based formula). Synthroid is preferred - generic levothyroxine may have variable absorption in the newborn period.*

References:

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Reviewed May 2022 – Mackay MD / Jackson MD / MacIver MD (Endo)