

Congenital Hypothyroidism (TSH, NBS)

Use

Quantitative determination of human thyroid stimulating hormone (hTSH) in blood specimens dried on filter paper as an aid in screening newborns for congenital (neonatal) hypothyroidism.

Clinical Significance

Congenital hypothyroidism occurs in about 1 in 3,000 to 4,000 newborns and is usually caused by abnormal development or absence of the thyroid gland. Untreated, low levels or absence of circulating thyroid hormone can lead to intellectual disability and growth failure; however, excellent treatments are available, which can provide normal healthy development and normal life expectancy. Clinical diagnosis of neonatal hypothyroidism in infants is difficult to establish, and the condition needs early medical intervention. An elevated hTSH concentration in infant blood is the earliest available laboratory manifestation of primary hypothyroidism. Due to its high specificity and sensitivity, hTSH testing is the screening method of choice for the detection of neonatal hypothyroidism.

Further information and ACT Sheets can be found at the OSDH Newborn Screening Program [website](#).

Methodology

GSP Neonatal hTSH solid phase fluoroimmunoassay

Specimen Type

See [Guidance for Collection of NBS Dried Blood Spots](#)

Minimum Volume/Size

See [Guidance for Collection of NBS Dried Blood Spots](#)

Collection Instructions

See [Guidance for Collection of NBS Dried Blood Spots](#)

Common Causes for Rejection

See [Guidance for Collection of NBS Dried Blood Spots](#)

Shipping

See [Guidance for Collection of NBS Dried Blood Spots](#)

Turn-around Time

Within 5 working days from receipt

Reference Range

TSH < 27.0 μ IU/mL serum

Reportable Results

- Within Normal Limits
- Outside Normal Limits

Interpretation

- Within Normal Limits: Not consistent with congenital hypothyroidism
- Outside Normal Limits
 - Elevated: Submit repeat specimen within 48 hours or perform serum TSH and free-T4 tests at 10 to 14 days of life
 - Repeat Elevated: Possible congenital hypothyroidism; recommend immediate serum TSH and free-T4 confirmatory testing
 - High: Consistent with congenital hypothyroidism; recommend immediate serum TSH and free-T4 confirmatory testing

Limitations/Interferences

- This is a screening test only. A diagnostic procedure performed on a serum sample should be used to confirm diagnosis of congenital hypothyroidism.
- Heterophilic antibodies in the sample may interfere with the assay.
- Specimens improperly collected, processed or transported may result in erroneous results.

CPT Code

84443

Notes

This test is approved for *in vitro* diagnostic use by the U.S. Food and Drug Administration.