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Neonatal Thyroid Screening

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Introduction

Congenital hypothyroidism may result in mental retardation if treatment is not initiated within the first two weeks of life. This can occur in a range of manners, each with different prognosis and disease progression.

a. Maternal hypothyroidism often leads to significant neuro-intellectual impairment in the infant despite early diagnosis and treatment.
b. Maternally induced hypothyroidism due to medication, iodine deficiency or excess or blocking antibodies may manifest as transient hypothyroidism in the infant.
c. Neonatal hypothyroidism may show very little initial symptoms due to residual maternal hormone, which responds well to early treatment.

Screening Options

Screening should be undertaken between 2 - 4 days after birth, alternatively before discharge, preferably within 7 days.

Samples collected prior to 48 hours are often found to have elevated TSH levels whereas critically ill infants and post-transfusion, infants may have falsely low values.

There are three major approaches to screening for neonatal hypothyroidism each with advantages and disadvantages (table 1).

Results and Diagnosis

The diagnosis of congenital hypothyroidism is established with a TSH >40mU/L (or >40µU/mL) and a low fT4 concentration.

This finding needs to be confirmed, however, confirmation should not delay initiation of treatment.

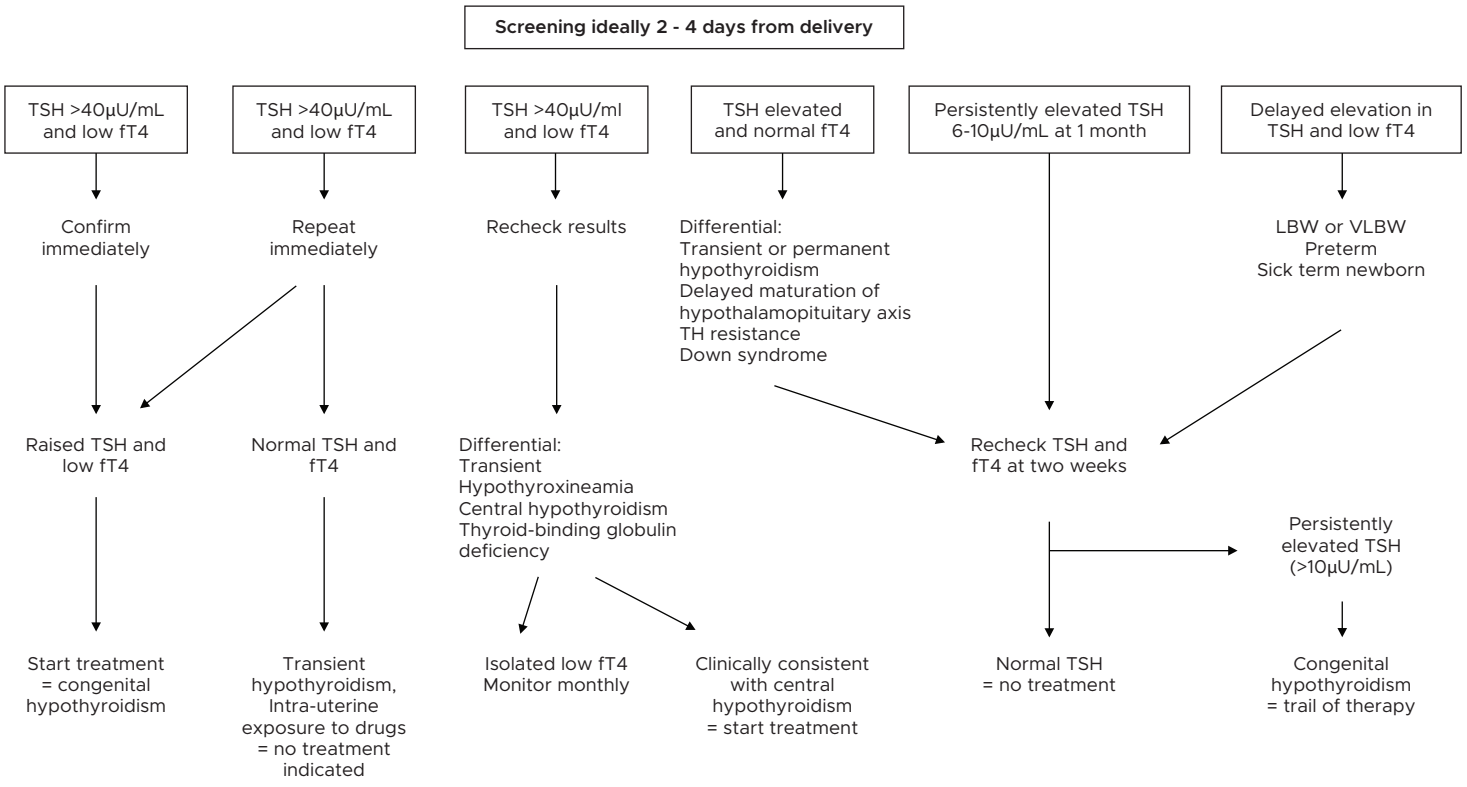
Elevated TSH values, but lower than the diagnostic threshold, represents the majority of clinical cases and requires a second screening using a fresh sample.

The testing and follow-up algorithm as proposed by the American Academy of Pediatrics (AAP) may facilitate management of often confusing results obtained within the diagnostic testing window (figure 1).

Table 1

Approach	Advantages	Disadvantages
Primary TSH measurement with back up fT4 if TSH is high.	Cost effective screening effective in large portion of population.	Misses delayed rise in TSH with thyroxine-binding globulin deficiency, central hypothyroidism and hypothyroxinaemia. Normal postnatal increased TSH becomes an issue upon discharge.
Primary fT4 measurement with backup TSH if fT4 is low.	Will diagnose thyroxine-binding globulin deficiency, central hypothyroidism and hypothyroixinaemia.	Misses hyperthyroitoxinaemia where TSH increase is delayed.
Simultaneous measurement of TSH and fT4.	Most comprehensive testing.	More costly.

Figure 1. Testing and management algorithm as adapted from the American Academy of Pediatrics.



Follow-up

Follow-up testing is essential to ensure appropriate patient response and compliance (figure 2).

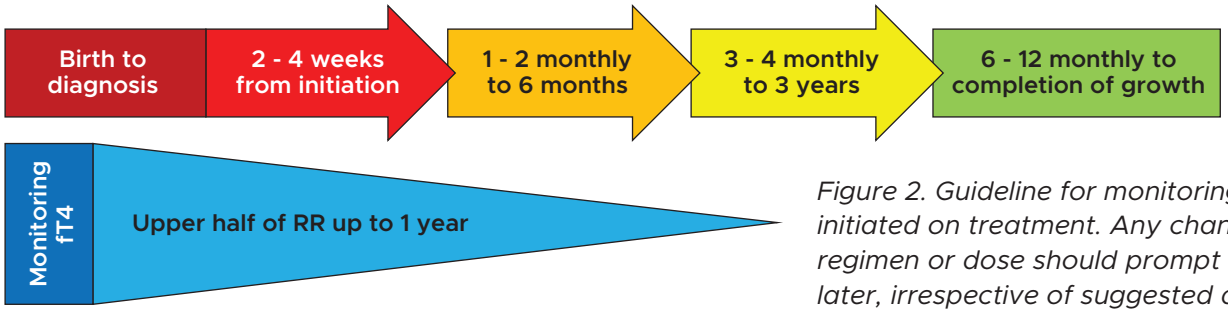


Figure 2. Guideline for monitoring tests in patients initiated on treatment. Any change in treatment regimen or dose should prompt testing 4 weeks later, irrespective of suggested algorithm.

References

1. Smith L. 2007. Updated AAP Guidelines on Newborn Screening and Therapy for Congenital Hypothyroidism. Am Fam Physician. 76(3):439-444

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