Diagnostic Evaluation

Because CH is one of the most common preventable causes of cognitive impairment, early diagnosis and treatment of this disease are essential interventions. Neonatal screening consists of an initial filter paper blood spot T_4 measurement followed by measurement of thyroid-stimulating hormone (TSH) in specimens with low T_4 values.

Tests are mandatory in all US states and territories. Although a blood sample obtained by heel stick for the spot test is best obtained between 2 and 6 days of age, specimens are usually taken within the first 24 to 48 hours or before discharge as part of a concurrent screen for other metabolic defects. Early screening can result in overdiagnosis (false-positives) but is preferable to missing the diagnosis.

For screening results that show a low level of T_4 (<10%), obtain TSH levels, and if these are elevated (>40 mU/L), further tests to determine the cause of the disease should be carried out (Stokowski, 2014). Additional tests include serum measurement of T_4, triiodothyronine (T_3), resin uptake, free T_4, and thyroid-bound globulin. Tests of thyroid gland function (thyroid scan and uptake) usually involve oral administration of a radioactive isotope of iodine (^{131}I) and measurement of iodine uptake by the thyroid, usually within 24 hours. In CH, protein-bound iodine, T_4, T_3, and free T_4 levels are low, and thyroid uptake of ^{131}I is decreased. Skeletal radiography is used to assess age.

In newborns, thyroid function studies are elevated in comparison with values in older children; therefore, it is important to document the timing of the tests. In preterm and sick full-term infants, thyroid function tests are usually lower than in healthy full-term infants; a repeat T_4 and TSH may be evaluated after 30 weeks (corrected age) in newborns born before that time and after resolution of the acute illness in sick full-term infants.

Therapeutic Management

Treatment involves lifelong thyroid hormone replacement therapy as soon as possible after diagnosis to abolish all signs of hypothyroidism and reestablish normal physical and mental