

**Disease Name:**

**CONGENITAL HYPOTHYROIDISM, THYROID DYSGENESIS, ECTOPIC THYROID, HYPOPLASTIC THYROID**

**Classification:**

**Genetic Information:**

**Inheritance:** 85% are sporadic; 13% represent various autosomal recessive forms of defects in thyroid metabolism; 2% are the result of transplacental passage of maternal thyrotrophin blocking antibody

**Population Incidence:** 1:3,000

**Ethnic Incidence:** Caucasian 1:4,000; Hispanic 1:2,000; Native American 1:2,000; African American 1:32,000; 50% more females than males

**Gene & Location:**

**Common Mutation:** NA

**OMIM #** 218700

**Disease Information:**

**Symptom Onset:** Generally not before 3 months of age

**Symptoms:** Unfortunately, long before clinical symptoms become apparent, the brain is damaged and mental retardation is the primary symptom. Other symptoms, if present, may include prolonged neonatal jaundice, constipation, lethargy, poor muscle tone, feeding problems, a large tongue, puffy face, large fontanelle, distended abdomen and/or umbilical hernia.

**Physical Findings:** In addition to the symptoms listed above, there may be delayed bone age and growth problems.

**Treatment:** Thyroxine, in pill form, is crushed, mixed with breast milk or non-soy formulas and given once a day. It is critically important to normalize T4 levels and suppress TSH as soon as possible and to monitor these levels every few weeks in the first year of life.

**Natural History without treatment:** Severe mental retardation (cretinism), growth failure

**Natural History with treatment:**

While the mental outcome of early treated hypothyroidism is expected to be normal, outcome depends on the severity of prenatal hypothyroidism, early, rapid normalization of thyroid levels, and compliance with therapy.

**Newborn Screening Information:**

**T4  
TSH**

T4 levels are low, TSH increased. 10% of all affected infants will be identified only on the routine second screening specimen. T4 levels may be low in very low birth weight or ill infants.

**Prenatal testing:**

For maternal antibody-mediated congenital hypothyroidism; suspected with previous affected sibling and mother with autoimmune hypothyroidism

**Miscellaneous Information:**

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4. Fisher DA, Schoen EF, La Franchi S, Mandel SH, Nelson JC, Carlton EI, et al. The hypothalamic-pituitary-thyroid negative feedback control axis in children with treated congenital hypothyroidism. *J Clin Endocrinol Metab* 2000;85:2722-7.
5. Leger J, Larroque B, Norton J. Influence of severity of congenital hypothyroidism and adequacy of treatment on school achievement in young adolescents: a population-based cohort study. *Acta Paediatr* 2001;90:1249-56.
6. Simoneau-Roy J, Marti S, Deal C, Huot C, Robaey P, Van Vliet Guy. Cognition and behavior at school entry in children with congenital hypothyroidism treated early with high-dose levothyroxine. *J of Pediatr* 2004;144:747-52.
7. Larson C, Hermos R, Delaney A, Daley D, Mitchell M. Risk factors associated with delayed thyrotropin elevations in congenital hypothyroidism. *J Pediatr* 2003;143:587-91.
8. LaFranchi SH, Hanna CE, Krainz PL, Skeels MR, Miyahira RS, Sesser DE. Screening for congenital hypothyroidism with specimen collections at two time periods: results of the Northwest Regional Screening Program. *Pediatrics*. 1985;76:734-740.