

Congenital Hypothyroidism

Congenital Hypothyroidism results from an inadequate production of thyroid hormone, which may be due to a number of causes. Disorders of hormonogenesis may be inherited as autosomal recessive trait and may be X linked. There is a 3:1 sex ratio of females to males. Other causes of hypothyroidism include maternal radioiodine or iodine exposure, TSH or Thyroid Hormone Unresponsiveness, hypopituitarism and developmental defects of the thyroid gland. In newborn infants with congenital hypothyroidism, associated symptoms may vary in range of severity and rate of progression depending on degrees of thyroid hormone deficiency. Symptoms may range from infantile hypotonia, poor feeding and delayed stooling at birth to the classic signs of myxedema, protruding tongue, and poor peripheral circulation and bradycardia. Treatment should begin in the first few weeks of life to prevent permanent retardation of intellectual function and skeletal growth.

Prevalence:	1: 3,600- 1:5,000
Analyte Measured:	Thyrotropin (TSH)
Reporting Ranges:	Decreased T4 Elevated TSH
Feeding Effect:	None
Timing Effect:	< 24 hours of age: Repeat test >24 hours of age: Results are valid
Confirmation:	Repeat filter paper specimen. If results remain abnormal then Serum T4 and TSH levels are indicated.
Treatment:	Referral will be made to a Pediatric Endocrinologist. Initiate oral intake of thyroxin (dose is weight dependent). Additional treatment is symptomatic and supportive.
Comment:	Most newborns are asymptomatic due to the transplacental transfer of moderate amounts of maternal T4. Specimens should be obtained at 48 hours to avoid the TSH surge at birth due to stress producing false positive results. TSH levels return to normal adult levels in about 72 hours. Providers must be alerted to clinical symptoms in older infants despite normal newborn screening results.