CONGENITAL HYPOTHYROIDISM

In Lithuania, newborn screening for congenital hypothyroidism (CH) has been implemented since 1993. All neonates are tested for thyroid stimulating hormone (TSH) levels in dry blood samples.

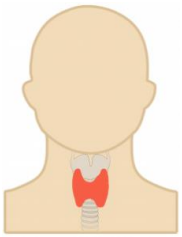
What is congenital hypothyroidism?

Congenital hypothyroidism (CH) is characterized by underdevelopment of thyroid gland. Sometimes thyroid gland grows normally but does not produce hormone thyroxine.

What is the incidence rate of congenital hypothyroidism?

In Lithuania incidence of CH is 1 in 4,600 newborns.

What causes congenital hypothyroidism?



Thyroid gland is a small gland located in neck below larynx (Adam's apple).

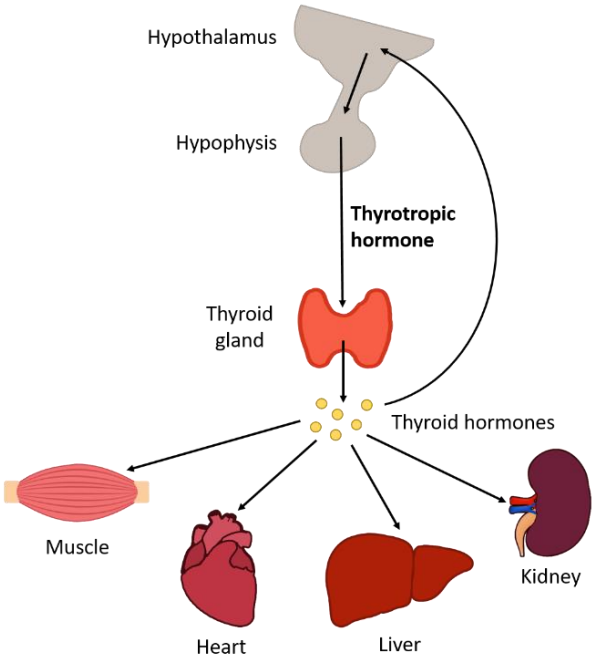
It produces and secretes thyroid hormones that regulate growth and metabolism. Main hormone produced by thyroid gland is thyroxine, which controls amounts of energy used to support vital processes such as breathing, metabolism and digestion. Too much thyroxine speeds up metabolism abnormally and too little of thyroxine slows it down. Thyroid hormones are also essential for brain development from fetal period to two/three years of age.

Normally, thyroid gland should be fully formed by 22nd week of gestation. Sometimes thyroid gland is underdeveloped or not developed at all. In these cases thyroxine is not produced.

80–85 % CH cases are caused by thyroid dysgenesis (underdevelopment).

10–15 % CH cases are caused by thyroid dishormonogenesis (disorder of thyroid hormone production, when gland itself is intact).

Up to 5 % CH cases are secondary due to abnormalities in pituitary or hypothalamic areas. This form is not detected by NS since blood levels of thyrotropic hormone are not elevated.



How is congenital hypothyroidism inherited?

About 90 % CH cases are spontaneous, with remaining 10 % related to inheritance (rare cases of dysmorphogenesis).

How does congenital hypothyroidism manifest?

CH causes growth, sexual maturation, and psychomotor development disturbances. Common symptoms for infants are feeding difficulties, drowsiness, constipation, prolonged neonatal jaundice (yellowing of the skin). However, all of the above mentioned symptoms are quite common in neonates, who do not have hypothyroidism. Therefore, all neonates should be tested for CH.

Extremely preterm newborns (23–27 weeks of gestation) with CH may not be discovered during the initial NS program. Re-sampling of blood is required 2 weeks after birth.

How is congenital hypothyroidism treated?

When hypothyroidism is diagnosed, treatment is immediately initiated and often continues throughout life. Oral thyroxine tablets are prescribed. Doctor determines exact dose of medicine needed for the infant, and makes necessary adjustments as child grows. Blood levels of thyroxine should be monitored regularly.

Compliance to treatment is important since in absence of thyroxine child will grow and develop poorly, irreversible brain damage will occur. On the other hand, when dose is too high, symptoms of hyperthyroidism such as restlessness, weight loss and severe diarrhea may manifest.