

Inborn hypothyroidism

- the most common congenital endocrine disease (prevalence 1: 3000–4000);^[1]
- thyroid hormones play a key role in brain development, especially by 8 months of age (slightly less so by 3 years of age);
- without substitution treatment, irreversible brain damage occurs - at the clinical diagnosis, the brain is already irreversibly damaged;
- since 1985, nationwide neonatal screening has been introduced - determination of TSH levels;
- etiopathogenesis: in 75–80% **thyroid dysgenesis** (agenesis, aplasia, hypoplasia, hemithyroidism, cystic malformation, ectopy) or **dyshormonogenesis** (disorder of any stage of hormone synthesis or secretion; neonatal goiter), or rare **isolated congenital central hypothyroidism** (congenital TSH defect - cannot be detected by neonatal screening)
- clinical picture without treatment: initially only prolonged neonatal jaundice (due to transplacental transmission of thyroid hormones from the mother), later (in the first 2 - 3 months of life) failure, slower growth rate and bone maturation - late closure of the fontanelle, delayed eruption of deciduous teeth, muscle hypotension, omphalocele, constipation, hoarseness, thermoregulation disorders, anemia; even later growth failure, psychomotor retardation, sensorineural hearing loss
- neonatal goiter or thyroid gland of normal size
- 2-5 times increased risk of associated congenital malformations compared to other populations → ultrasound examination of the heart, kidneys and CNS is recommended
- laboratory findings: ↑ TSH, ↓ fT4; (for the central form ↓ TSH and fT4);
- therapy: lifelong levothyroxine replacement therapy (started as soon as possible); intestinal absorption of L-thyroxine is impaired by concomitant ingestion of fiber, soy milk, calcium or iron preparations and malabsorption.^{[2][1]}

Links

Related articles

- Diseases of the thyroid gland • Goitre
- Examination of thyroid gland

Reference

1. AL TAJI, E a O HNÍKOVÁ. Tyreopatie v dětství a adolescenci. *Pediatr. praxi* [online]. 2014, roč. 15, vol. 3, s. 134-137, also available as <<https://www.pediatricpropraxi.cz/pdfs/ped/2014/03/04.pdf>>.
2. LEBL, J, J JANDA a P POHUNEK, et al. *Klinická pediatrie*. 1. vydání. Galén, 2012. 698 s. s. 185-188. ISBN 978-80-7262-772-1.