Congenital hypothyroidism

- The most common congenital endocrine diseases (prevalence 1:3000–4000);[1]
- Thyroid hormones have a key role in brain development, especially up to 8 months of age (a little less until 3 years of age);
- Without substitution therapy, irreversible brain damage occurs – with clinical diagnosis, the brain is already irreversibly damaged;
- Since 1985, a nationwide neonatal screening – determination of the level TSH;
- etiopathogenesis: in 75–80 % **thyroid dysgenesis** (agenesis, aplasia, hypoplasia, hemithyreoidea, cystic malformation, ectopia) 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 icterus (due to transplacental transmission of thyroid hormones from the mother), later (in the first 2-3 months of life) failure to thrive, delayed growth rate and bone maturation – late closure of fontanelles, delayed eruption of milk dentition, macroglossia, muscle hypotonia, omphalocele, constipation, hoarse crying, disorders of thermoregulation, anemia; even later growth disorder, psychomotor retardation, sensorineural hearing impairment;
- neonatal goiter or thyroid gland of normal size;
- 2–5 times increased risk of associated congenital malformations compared to the general population → ultrasound examination of the heart, kidneys and CNS is recommended;
- laboratory findings: ↑TSH, ↓fT_4; (for the central form ↓TSH i fT_4);
- therapy: lifelong replacement therapy with levothyroxine (started as soon as possible); intestinal absorption of L-thyroxine is aggravated by simultaneous ingestion of fiber, soy milk, calcium or iron preparations and malabsorption as such.[2][1]

Links

Related Articles

- Thyroid disease • Children's goiter
- Examination of thyroid function

Ref

1. AL TAJI, E – HNÍKOVÁ, O. Tyreopatie v dětství a adolescenci. *Pediatr. praxi* [online]. 2014, y. 15, vol. 3, p. 134-137, Available from <https://www.pediatriepropraxi.cz/pdfs/ped/2014/03/04.pdf>.
2. LEBL, J – JANDA, J – POHUNEK, P. *Klinická pediatrie.* 1. edition. Galén, 2012. 698 pp. pp. 185-188. ISBN 978-80-7262-772-1.