Bartter syndrome

Template:Infobox - genetic disease '*Bartter syndrome* ***is an AR hereditary tubulopathy with a combination of impaired water and electrolyte metabolism. The syndrome arises as a consequence of a complex*** **disorder of tubular transport** ***and*** *excretion of ions*.

Etiology

Diseases are caused by **abnormalities'** of three different **transport systems'**: $Na^+/K^+/2Cl^-$- cotransporter, potassium channel and chloride channel.

Pathogenesis

$Na^+/K^+/2Cl^-$-cotransporter defect (NKCC2, an ATP-independent ion channel)[1] in the ascending part of the loop of Henle of the nephron leads to insufficient absorption of sodium, and its reduced level in the *macula densa* will increase the activity of the RAAS, which leads to an increase in the level of aldosteroneu and to **secondary hyperaldosteronism** with all clinical symptoms (blood pressure is normal).

Clinical picture

The main symptoms include:

- hypokalemia (significant muscle weakness)
- alkalosis
- hypercalciuria
- polyuria
- hyperrenin hyperaldosteronism
- growth disorders.

According to the type of defective transport system*, we recognize* *6 types* of Bartter syndrome. [2]
Type I - $Na^+/K^+/2Cl^-$-cotransporter defect (NKCC2, gene "SLC12A"); manifests already in infancy, mostly in children born prematurely to mothers with polyhydramnios[3].
Type II − ATP-dependent apical potassium channel defect (ROMK1, gene *KCNJ1*); phenotypically it is the same as type I.
Type III - basolateral chloride channel defect (ClC-Kb, gene *CLCNKB*); hypomagnesemia is observed in 30% of patients (types I and II do not have it)[3].
Type IVa' − β-subunit defect of the basolateral chloride channel (Barttin, gene *BSND*); characteristic **trias**: Bartter's syndrome, renal insufficiency, hearing impairment[3].
Type IVb - combined dysfunction of two chloride channels ClC-Ka and ClC-Kb (genes "CLCNKA" and "CLCNKB"), prenatal manifestation, polyhydramnios
Type V - transient form (defect in the *MAGED2* gene), polyhydramnios, excessive salt loss with secondary metabolic alkalosis, disappears spontaneously in the first months of life

Therapy

The therapy is only **symptomatic**, its basis is diet adjustment and ion substitution[2].

Prognosis

The prognosis of the disease is uncertain, some patients develop mental retardation or kidney failure.

Links

References

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2. ZIEG, Jacob – DOLEŽEL, Zdeněk. Bartter and Gitelman syndrome. *Čas Lék Czech* [online]. 2022, vol. 161, no. 3-4, p. 131-134, Available from <https://www.prolekare.cz/casopisy/casopis-lekaru-ceskych/2022-3-4-1/bartteruv-a-gitelmanuv-syndrom-131752>. ISSN 0008-7335.
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