Bartter's syndrome

Bartter's syndrome is AR inherited tubulopathy with a combination of disorders of water and electrolyte metabolism.

The syndrome arises as a result of a complex disorder of tubular transport and ion excretion .

Etiology: The disease is caused by abnormalities of three different transport systems : Na + / K + / 2Cl - cotransporter, potassium channel and chloride channel.

Pathogenesis: A defect in the Na + / K + / 2Cl - transporter (NKCC2, ATP-independent ion channel) [2] in the ascending part of the Henle loop of the nephron leads to insufficient sodium absorption and its reduced level in the macula densa increases RAAS activity , which leads to increased levels aldosterone and secondary hyperaldosteronism with all clinical signs (blood pressure is normal).

The clinical picture

The main **symptoms** include:

Hypokalaemia (severe muscle weakness)

Alkalosis

Hypercalciuria

Growth disorders.

According to the type of defective transport system, we distinguish 5 types of Bartter's syndrome.

- Type I - Na + / K + / 2Cl - cotransporter defect (NKCC2); manifest already in infancy, mostly premature babies to mothers with polyhydramnios [3]
- Type II - ATP-dependent apical potassium channel defect (ROMK1); phenotypically the same as type I.
- Type III - basolateral chloride channel defect; Hypomagnesaemia is observed in 30% of patients (types I and II do not have it) [3]
- Type IV - β-subunit defect of the basolateral chloride channel; Characteristics: Bartter's syndrome, renal insufficiency, hearing impairment [3] .
- Type V - calcium receptor defect [1] ; Hypocalcaemia occurs with decreased PTH [3] .

Therapy: The therapy is only symptomatic.

Prognosis: The prognosis of the disease is uncertain, with some patients experiencing mental retardation or kidney failure .

References

References ORPHANET ,. Bartter syndrome [online]. [feeling. 2015-12-08]. < https://www.orpha.net/consor/cgi-bin/Disease_Search.php?lng=EN&data_id=259&Disease_Disease_Search_diseaseGroup=Bartter-syndrome&Disease_Disease_Search_diseaseType=Pat&Disease (s)/group% ndBartTter = ter = ter = tertitle Disease_Search_Simple ,>. CASTROP, Hayo and Ina Maria SCHIEßL. Physiology and pathophysiology of the renal Na-K-2Cl cotransporter (NKCC2). Am J Physiol Renal Physiol [online] . 2014, vol 307, no. 9, pp. F991-F1002, also available from < https://www.ncbi.nlm.nih.gov/pubmed/25186299 >. ISSN 1522-1466. HEROLD, Gerd, et al. Internal medicine. 1st edition. 2016. 1000 pp. ISBN 9783981466058 . References CHILD, P., et al. Internal Medicine. 2nd edition. Prague: Galén, 2007. ISBN 978-80-7262-496-6 . KLENER, P, et al. Internal Medicine. 3rd edition. Prague: Galén, 2006. ISBN 80-7262-430-X .