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HYPOKALEMIA – EARLY MARKER OF AUTOSOMAL RECESSIVE TUBULOPATHY (GYTELMAN'S SYNDROME) - CASE REPORT

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ABSTRACT

Hypokalemia is the most common feature of Gitelman syndrome, which is a rare, inherited, autosomal recessive kidney disease associated with tubule disease. In addition to hypokalemia, it is also characterized by hypomagnesemia, metabolic alkalosis, hyperrenemic hyperaldosteronism, normal or lower blood pressure, while the presence of arterial hypertension does not exclude the diagnosis. It affects men and women equally, with a prevalence of 1 to 10 cases per 40,000 inhabitants. The most common cause are mutations in the SLC12A3 gene, which encodes the thiazide-sensitive sodium chloride cotransporter (NCCT) in the renal distal tubules, and the TRPM6 (cation channel subfamily 6 protein claudin 16) gene, which controls distal tubular magnesium transport. The aim of the paper is to present an adult patient with pronounced hypokalemia as part of Gitelman's syndrome. Case report: We present a 21-year-old man with severe hypokalemia as part of Gitelman's syndrome. The disease manifested itself in non-specific complaints, and laboratory findings showed hypokalemia of 2.0 mmol/L, which was the reason for urgent hospitalization. Further examinations of the patient verified the following: hypomagnesemia, hypocalciuria, metabolic alkalosis, preserved kidney function and arterial hypotension. Other potential causes of hypokalemia were excluded by differential diagnosis. He was treated with potassium and magnesium replacement therapy, after which the symptoms of hypokalemia disappeared, and the electrolyte values were closer to the reference values. The diagnosis of Gitelman's syndrome was made based on clinical and laboratory findings. A geneticist was also consulted. Hypokalemia as part of Gitelman's syndrome is rarely encountered in clinical practice, and it is rarely thought of. Severe forms of hypokalemia should arouse suspicion of its existence and lead to a final diagnosis, for which rich clinical experience and teamwork are necessary. The patients with symptoms should be treated symptomatically, and those without symptoms should be monitored 1-2 times a year.

Key words: *kidney diseases; Gitelman syndrome, genetic diseases, inborn; hypokalemia, alkalosis.*

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