

CAGAR syndrome

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Key words: Gitelman syndrome, CCTGA, arterial hypertension, MS

We present the patient with the combination of different genetic and autoimmune diseases: Congenitally corrected transposition of great arteries (CCTGA), Gitelman syndrome, resistant arterial hypertension and urinary retention that was the first sign of multiple sclerosis. A 54-year old female patient presented to the department of internal medicine due to hypokalaemia and hypomagnesemia when Gitelman syndrome was diagnosed. Concentrations of potassium in blood measured from 2014 to 2019 were: 2.9-3.1-3.2-3.3-3.7 mmol/L. She also had resistant arterial hypertension which is uncommon in Gitelman sy., but now is successfully controlled with antihypertensive therapy. The patient also suffers from urinary retention (lower than 3 L of urine) which is an atypical clinical presentation, but it was also the first sign of multiple sclerosis. Because of urinary retention she undergoes micturition cystography after which low residual volume (80 mL) was detected and potential diverticula were suspected. She often develops fevers up to 42°C (due to sinusitis, otitis, UTI) during which oedema of soft tissue is observed (she gains on her weight up to 15 kg), that condition is considered and treated as hyperpyrexia syndrome. A positive history of hypersensitivity to many different drugs such as penicillin, ciprofloxacin, tramadol, metoclopramide, trospium chloride, ezetimib, ivabradine, clarithromycin, pregabalin was identified. Statin and fibrates-induced myopathy and urinary retention while taking amitriptyline and carbamazepine were also observed. Gitelman syndrome has been considered as a benign variant of salt-losing nephropathies presenting asymptomatic or with mild symptoms. This view has since been challenged by recent reports emphasizing the phenotype variability and the potential severity of the disease. This is the first case where Gitelman sy. is associated with CCTGA, resistant arterial hypertension and urinary retention which all together can be described as a CAGAR syndrome.