disorders in the patients with AS (20 patients with AS on a background of diabetes and 10 stroke patients without diabetes). **Results.** The diabetic history were present in 110 of 416 patients (26.4 %), another 48 patients (11.5 %) diagnosed with diabetes was first installed. Thus, the incidence of diabetes in patients with AS in our study was approximately 38.0%, much higher than in the corresponding age population. In general, the different types of electrolyte disorders were observed in 258 of 349 (73.9 %) patients with AS, while in patients with underlying disorders of carbohydrate metabolism, these violations occurred significantly more often than patients without such. In particular, carbohydrate metabolism occurred in 82 (81.2 %) and 36 (83.7 %) patients 1 and 2 groups versus 134 (65.4 %) in the control group ($p < 0.05$). Hypomagnesemia (less than 0.8 mmol/L) was observed in 2 of 10 patients (20 %) in AS without carbohydrate disturbances and in 6 of 20 patients (30 %) in AS with concomitant diabetes. Hypophosphatemia (phosphate levels less than 0.8 mmol/L) was found in patients without diabetes and AS in 2 patients with concomitant DM or newly diagnosed diabetes. Following the correction of oral medication containing phosphates and magnesium blood electrolyte levels was stabilized and that coincided with the improvement of the patients and the degree of disability. **Conclusions.** Electrolyte disorders are fairly common problem in patients with concomitant diabetes and AS. In patients with impaired carbohydrate metabolism observed significantly higher frequency (more than 81.2 % of patients) occurrence of electrolyte disorders than patients without them. In the case of AS on a background of diabetes electrolyte disturbances occur significantly more frequently than in patients without such comorbid disorders. Further research is needed to elucidate the role of individual electrolyte disorders (eg, magnesium and phosphate) in the course and consequences of AS.

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X-linked myotubular myopathy and dilated cardiomyopathy as the cause of respiratory failure in a ventilated child in the ICU (case report)

Background. Centronuclear myopathies (CNMs) are a group of clinically and genetically heterogeneous muscle disorders [1]. Myotubular myopathy, an X-linked form of CNM is characterized by neonatal hypotonia and inability to maintain unassisted respiration. The MTM1 gene, responsible for this disease, encodes myotubularin — a lipidic phosphatase involved in vesicle trafficking regulation and maturation [2]. **Case report and discussion.** We report a 7-month old male infant who has required mechanical ventilation since birth due to suspected neuromyopathy. Congenital adrenal hyperplasia, Pompe disease (type II glycogenosis), Prader — Willi syndrome, and SMA were excluded. Further genetic testing revealed a hemizygous variant (c. 64–2A > G) in MTM1. This variant is predicted to abolish the intron 2 splice donor site of MTM1 and has been reported in a male infant with severe X-linked myotubular myopathy [3]. Our patient's mother and maternal grandmother were found to be heterozygous carriers of the c. 64–2A > G variant. Two variants of uncertain significance were detected in this patient's MYH7 gene. MYH7 encodes the cardiac-specific beta heavy chain myosin protein and is a cause of autosomal dominant dilated cardiomyopathy and distal myopathy. One variant (p.Leu881Met) was inherited from the father and the other (p.Arg1749Gly) was inherited from the mother. The paternally inherited variant is found in a region of the MYH7 protein where a significant number of previously reported MYH7 missense mutations are found [4]. Both MYH7 variants are absent from the ExAC public database. Thus, the genetic testing allowed us to diagnose the combined genetic pathology: myotubular myopathy and possibly dilated cardiomyopathy. This pathology causes the respiratory failure and the need of permanent respiratory support in a patient. **Conclusions.** The case report demonstrates importance of: 1) genetic screening in a population, especially in genetics-compromised parents, for family planning and timely detection of hereditary diseases during pregnancy; 2) early genetic testing to confirm the diagnosis of a sick child; 3) development of palliative and hospice medicine with the possibility of providing ventilation support at home by parents (guardians).

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