Acute dehydration as a primary manifestation of Cystic fibrosis

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Objectives: To present a case of acute hyponatriemic dehydration as a primary manifestation of cystic fibrosis.

Methods: male infant 6 months old from normal pregnancy and with normal development. Four days with reduced appetite, weaker food intake and fatigue. On the day of examination subfebrile (37,4C) with nausea, refusing food and liquids. At admittion subfebrile (37,4C), pale, with fatigue and perioral cyanosis. Signs of dehydration were present: dry tongue, reduced turgor and elasticity of the skin. Big fontanela under the calvaria and with mild hypotonia. Heart rate tahycardic. Other physical examination was normal. Laboratory analysis with decreased levels of Na=126 mmol/l, K=2.8 mmol/l, Cl=68mmol/l and urea= 9.1mmol/l. Urine positive for ketones. ABS–Ph= 7.61; BE=14,6mmol/l(metabolic alkalosis). Chest X ray and abdominal ultrasound were normal. Treatment started with parenteral rehydration (0,9%NaCl) and per oral administration of 7,4% KCl with resolving of acute hyponatriemic dehydration, metabolic alkalosis and normalization of levels of electrolytes.

Results: Due to the occurrence of acute hyponatriemic dehydration without visible signs of additional fluid loss during the summer period and with positive family history of cystic fibrosis in a close relative we also thought of cystic fibrosis. An outpatient sweat test was made with increased levels of chlorides in sweat 124 mmol/l. Genetic analysis for CFTR mutations showed the presence of a mutation F508/del, second was not confirmed. The results of sequencing of the CFTR gene are still pending.

Conclusion: The occurrence of acute hyponatraemic/hypochloremic dehydration and metabolic alkalosis in the summer period without visible additional fluid loss in childhood should lead us to think of cystic fibrosis.