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FAILURE TO THRIVE WITH ELEVATED SWEAT CHLORIDE CONCENTRATION: SUSPECTED CYSTIC FIBROSIS – FINAL DIAGNOSIS NEPHROGENIC DIABETES INSIPIDUS

V. Skalicka[1], K. Blahova[1], V. Vavrova[2], J. Janda[1]
[1]1st Paediatric Department [2]2nd Paediatric Department, Charles University, 2nd Medical Faculty, University Hospital Motol, Prague, Czech Republic
veronika.skalicka@lfmotol.cuni.cz

Introduction: Cystic fibrosis (CF) is an autosomal-recessive disorder caused by cystic fibrosis transmembrane conductance regulator (CFTR) gene mutation leading to inspissated secretion in respiratory, gastrointestinal and reproductive tracts and elevated sweat salt content. Nephrogenic diabetes insipidus (NDI) is caused by renal resistance to antidiuretic hormone and/or impairment of the countercurrent mechanism in the loop of Henle leading to water diuresis resulting in high plasma sodium concentration. The primary form is an X-linked recessive.
Case report: Boy 7-months, growth retardation below 10th centile, negative family history. At 4 months failure-to-thrive. Laboratory: serum Na+ 159, Cl- 116 mmol/l, osmolality 325 mmol/kg, sweat chloride 56.9 mmol/l. Suspected CF. High fluid intake and diuresis>200 ml/kg/day reported. Urine: Na+ 6 mmol/24 hrs., osmolality 64 mmol/kg, DDAVP-test only 66 mmol/kg. Dg.: NDI. Treatment: iv. correction of hypovolemic hypernatremia, then frequent fluid serving. Hydrochlorothiazide 2 mg/kg/day resulted in reduction of fluid requirement to 800 ml/day. Serum ionogram and sweat Cl- normalized. Further development normal. Investigation of mother's native urine osmolality: normal.
Conclusion: growth failure is pathognomonic for both CF and NDI. In typical CF, sweat Cl- values (pilocarpine iontophoresis) exceed 60 mmol/l. In children values between 40 and 60 mmol/l are "indeterminate" and suggestive of CF. In CF serum Na+ is in the low normal range or decreased. In our NDI patient elevated sweat Cl- concentration was due to its high plasma concentration. Query on fluid intake/urine output was an essential point in the failing-to-thrive patient's history that led us to the diagnosis of NDI.