A girl with progressive fatigue and hyponatremia: question

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Case summary

A 9-year-old girl was admitted to hospital due to emesis, fatigue and lethargy. She is the only child of unrelated Caucasian parents. Her mother has recently been diagnosed with Crohn's disease and diabetes mellitus type I. During the 3 months preceding admission, the child had three episodes of gastroenteritis-like illness with vomiting and lethargy; for the two of these episodes, she was admitted to hospital where hyponatraemia was determined. The patient was treated with intravenous fluids, and she quickly recovered. The current episode was the fourth one of emesis and had started 24 h prior to admission, with recurrent abdominal pain and no diarrhoea. On the day of admission she deteriorated and became lethargic. She was afebrile. The mother had noticed progressive fatigue and weakness during the last few months as well as a weight loss of 5 kg.

On examination, the patient was sleepy with a slow response to external stimuli. She was mildly dehydrated with dry mucous membranes, an increased heart rate (101/min), normal blood pressure (100/50 mmHg), normal capillary refill and no cool extremities. Orthostatic measurements were not possible due to her clinical condition. There was no clitoridomegaly, and her pubertal development was at Tanner stages B1, P2, A1. She was at the 75th centile for height and 10th centile for weight.

The answer to this question can be found at <http://dx.doi.org/10.1007/s00467-009-1404-6>.

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Laboratory investigations revealed hyponatremia (Na 126 mEq/l), normokalaemia (K 4.0 mEq/l), glucose at 73 mg/dl, urea at 40 mg/dl and creatinine at 0.6 mg/dl. The acid–base status, full blood count, C-reactive protein, erythrocyte sedimentation rate and liver function tests were all normal (Table 1). The thyroid function tests were in normal range [thyroid stimulating hormone 2.8 mIU/l, free thyroxine (FT₄) 1.4 ng/dl]. Urine electrolytes showed increased renal Na⁺ and K⁺ secretion (Table 1). The serum and urine osmolalities were not measured upon admission. The findings from a lumbar puncture were normal. Her bone age was accelerated for 2 years. The patient was initially treated with intravenous fluids to maintain circulating volume, and sodium chloride was administered to raise plasma Na⁺.

Questions

1. What important history and which diagnostics signs are absent from the case summary?
2. What is the possible diagnosis?
3. What diagnostic test would you perform to confirm it?

Table 1 Laboratory findings

Clinical parameters	On admission		Day 10	
	Plasma	Urine	Plasma	Urine
Na ⁺ (mEq/l)	126	148	139	153
K ⁺ (mEq/l)	4.0	32	4.8	22.5
Urea (mg/dl)	40	-	25	-
Creatinine (mg/dl)	0.6	38	0.5	34
Glucose (mg/dl)	73	-	85	-
Haemoglobin (g/dl)	13.0	-	11.6	-
FE _K (normal 10–30%)	-	12%	-	13.2%
Renal K ⁺ excretion (normal 1–2 mEq/kg/24 h)		1.3		0.9
K ⁺ /creatinine (mmol/g)		84		66

FE_K, Fractional excretion of K⁺