POLYURIA AND POLYDIPSIA: THE DEEP WATERS OF DIAGNOSIS

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Introduction

Central diabetes insipidus (CDI) is a rare condition. The management of diabetes insipidus is quite misleading and an ethilogical diagnosis is not always possible at first assessment. Water deprivation test (WDT) is not always diagnostic, therefore in patients with only mild or moderately impaired urine concentration capacity, the differentiation between primary polidypsia (PP), partial CDI, and partial nephrogenic DI should currently rely on a comprehensive assessment of patient and family history, clinical data and cranial MRI imaging of the hypothalamic-pituitary region.

Objectives

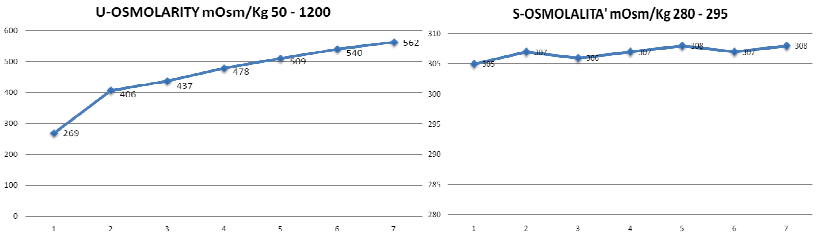
A 7 year old girl presented with polyuria, polydipsia and nocturia of mild severity and of one year duration, during which she underwent outpatient follow up with her general practitioner and urologist. No weight loss or other endocrine signs. A urinary tract problem was suggested, but urinalysis, urine colture and urinary tract ultrasonography were negative.

Methods

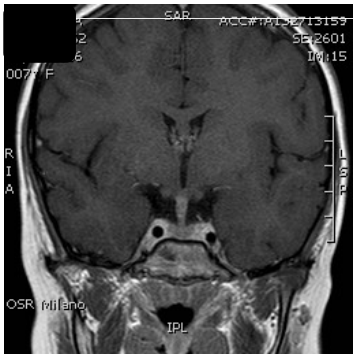
One year after the onset of symptoms polyuria was worsened and confirmed by water balance (120 ml/kg/24h); urinary osmolality, plasma osmolality and electrolytes were variable, no other hormone deficits were diagnosed:

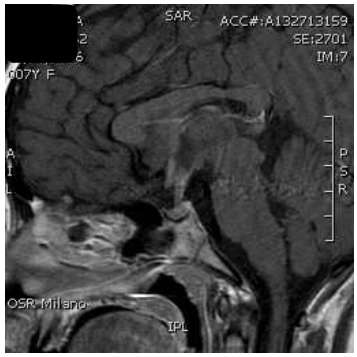
S-SODIUM (mmol/L)	133	135 - 148
U-OSMOLARITY (mOsm/Kg)	222	50 - 1200
S-OSMOLARITY (mOsm/Kg)	278…296	280 - 295
U-Osm/S-Osm	0,75	>1
IGF1 (ng/ml)	74	80-244
PRL (ng/ml)	14.8	2.8-29.9
TSH (uU/mL)	2.82	0.25-5
LH (mU/mL)	<0.1	0.1-6
FSH (mU/mL)	<0,3	
Cortisol h8-16-24 (ng/mL)	187…50…11	
ACTH h8-16-24 (pg/mL)	41…15…10	

During WDT (Fig 1) urine osmolarity partially increased whereas desmopressin, plasma osmolarity and sodium were stable. WDT revealed a partial DI, MRI was not performed.



For a second opinion, she was admitted at our center.
T1-weighted MRI showed:
- thickening of the pituitary stalk
- the lack of posterior pituitary hyperintensity.
-diffuse pathological enhancement of the optic chiasm and optic nerves
Hypothesis: chronic inflammatory granulomatosis disease suspect for Langherans-cell histiocytosis (LCH). No other organs were involved (spine MRI, abdomen ultrasonography, chest X-rays, hand-wrist x-ray, total body scintigraphy, ophthalmic visit were normal). Cerebrospinal fluid was negative for ß-HCG and α-fetoprotein.
We believed it was mandatory to proceed with biopsy of the brain tissue for histological diagnosis but her parents refused.
The patient was discharged with sublingual desmopressin acetate treatment and the water balance progressively improved.





Results

After one month of follow up, the patient presented neurological deterioration (decline in school performance, difficulty in concentration and memory).

Brain MRI showed worsening of the neuroradiological features with a neurodegenerative pattern of the white matter.

There was no apparent involvement of the cerebellum, which instead is a typical complication of LCH. These neuroradiological aspects were suspect of dysimmune encephalitis.

After many consultations, 3 months later, parents agreed to biopsy: evaluation of the pituitary stalk was compatible with a **germinoma**.

Conclusions

WDT is not always easy to understand especially in case of partial urine concentration. Furthermore ethiology of partial diabetes insipidus is very difficult to enstablish and this case shows the importance of performing brain MRI and to successively evaluate the need for bioptic examination. If the cause should remain unclear, tumors should always be excluded. Moreover, the thickening of the stalk and the inflammatory aspect initially led to hypothesize LCH, but may also have been suggestive of germinoma. Therefore, this neuroradiological aspect should be monitored in time. Finally, the neurodegenerative component (paraneoplastic or dysimmune) is an atypical presentation of germinoma and may be misleading.