The beast behind the dwarf

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Introduction:

Abnormalities of growth are one of the most common reasons for the pediatric-endocrinology consults. It's an obvious manifestation with countless possible causes behind, and sometimes we can have unexpected diagnosis.

Case report:

We investigated the case of a **4 years old girl**, born at term, naturally, SGA: birth weight= 1950 g, who presented in the Endocrinology Department for **short stature**.

Clinical evaluation:

- 89,5 cm (- 4 SD),
- 11 kg (-2,5 SD),
- discreet ocular asymmetry.

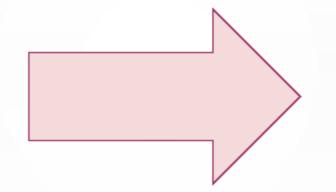
Blood test:

- anemia,
- decreased IGF-1: 40,8 ng/ml (49-289),
- normal adrenal function:
 - ACTH= 36 pg/ml (0-46)
 - cortisol= 14 ug/dl (5-25)
- normal thyroid function:
 - TSH= 0, 849 uIU/ml (0,3-6,3)
 - FT4=1,25 ng/dl (0,89-1,76).

- normal karyotype,
- normal thoracic X-ray and abdominal ultrasound.

Moreover, the first day of admission we noted **polyuria** and **polydipsia** not reported by mother:

- \triangleright ingestion 3750 ml/24 h,
- > excretion 3950 ml/24 h.



Dehydration test:

T0 End of test:

urine density: 1000 1010

urine osmolality: 51 mOsm/kg
141 mOsm/kg

plasma osmolality: 285 mOsm/kg 304 mOsm/kg

The suspicion of diabetes insipidus was confirmed.

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Ophthalmologic exam - important papillary edema.

MRI revealed an expansive mass with contrast enhancement located in posterior fossa. An **angio MRI** and a **biopsy** were performed and established the final diagnosis: **histiocytosis**

The patient received treatment for anemic syndrome and for diabetes insipidus (Desmopressin 30 ug/day). She was directed to the Pediatric Clinic where chemotherapy was initiated.

Conclusions:

Even if in our patient's case, the short stature and diabetes insipidus, were considered initially easy to manage and benign, the histiocytosis was found to be an unexpected and unpleasant diagnosis which involves more aggressive treatment and complications.







