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 IgG-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).

Moreover, the first day of admission we noted polyuria and polydipsia not reported by mother:
- ingestion 3750 ml/24 h,
- excretion 3950 ml/24 h.

Dehydration test:

<table>
<thead>
<tr>
<th>T0</th>
<th>End of test</th>
</tr>
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<tbody>
<tr>
<td>urine density:</td>
<td>1000</td>
</tr>
<tr>
<td>urine osmolality:</td>
<td>51 mOsm/kg</td>
</tr>
<tr>
<td>plasma osmolality:</td>
<td>285 mOsm/kg</td>
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The suspicion of diabetes insipidus was confirmed.

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.