ADDISON’S DISEASE: DELAY IN DIAGNOSIS IN A GIRL WITH LONGSTANDING SYMPTOMS

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BACKGROUND
Autoimmune destruction of the adrenal cortex is the cause of primary adrenal insufficiency (AI) in 45% to 55% of cases in children.

CASE PRESENTATION
10·1012 year old girl referred because of suspicion of AI. Longstanding complaints of extreme fatigue, loss of appetite, recurrent gastric symptoms and salt craving.

MEDICAL HISTORY
2 years: Idiopathic thrombocytopenic purpura (ITP) 4·1·12 years: Hospitalization with vomiting, dehydration and electrolyte abnormalities.

She was found underweight (BMI SDS: -5.71), with a biochemical profile (Table 1) indicative of AI (hypoglycemia, hyperkalemia, hyponatremia and inappropriately high sodium excretion). She was managed with iv hydration. The family failed to attend a scheduled appointment at the Pediatric Endocrinology Clinic.

Table 1: Biochemical profile at the age of 4·1·12 years

<table>
<thead>
<tr>
<th>Test</th>
<th>Result</th>
<th>Reference range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Glucose (mg/dl)</td>
<td>56</td>
<td>70-100</td>
</tr>
<tr>
<td>Urea (mg/dl)</td>
<td>39</td>
<td>5-45</td>
</tr>
<tr>
<td>Creatinine (mg/dl)</td>
<td>0.4</td>
<td>0.5-1.0</td>
</tr>
<tr>
<td>Potassium (mEq/L)</td>
<td>5.8</td>
<td>3.5-5.5</td>
</tr>
<tr>
<td>Sodium (mEq/L)</td>
<td>131</td>
<td>134-148</td>
</tr>
<tr>
<td>Calcium (mg/dl)</td>
<td>8.8</td>
<td>8.6-10.6</td>
</tr>
<tr>
<td>Urine sodium (mmol/l)</td>
<td>50</td>
<td>&lt;25</td>
</tr>
</tbody>
</table>

At the age of 10·1012 years
Extreme exhaustion and daily consumption of large amounts of salt prompted a second referral by the pediatrician.

Somatometry
Height: 146 cm (67th percentile)
Weight: 25 kg (11th percentile)
BMI SDS: -4.26 (underweight)

Tanner stages
Breast: Tanner II–III
Pubic hair: Tanner I
Axillary hair: Tanner I

Physical examination
normal female external genitalia
generalized hyperpigmentation
no mucosal candidiasis
blood pressure 93/66 mm Hg

Fig 1-4: Hyperpigmentation

Laboratory tests
<table>
<thead>
<tr>
<th>Test</th>
<th>Result</th>
<th>Reference range</th>
</tr>
</thead>
<tbody>
<tr>
<td>cortisol (μg/dl)</td>
<td>0.7</td>
<td>6.2 – 19.4</td>
</tr>
<tr>
<td>ACTH (pg/ml)</td>
<td>5690</td>
<td>7 – 64</td>
</tr>
<tr>
<td>PRA (ng/ml/h)</td>
<td>24.48</td>
<td>0.5-4.7</td>
</tr>
<tr>
<td>aldosterone (ng/dl)</td>
<td>11.8</td>
<td>3.0-28</td>
</tr>
<tr>
<td>DHEA-S (μg/ml)</td>
<td>0.067</td>
<td>0.4-1.4</td>
</tr>
<tr>
<td>Δ4-A (ng/ml)</td>
<td>0.16</td>
<td>0.25-0.8</td>
</tr>
<tr>
<td>TSH (μIU/ml)</td>
<td>6.2</td>
<td>0.4-5.0</td>
</tr>
<tr>
<td>fT4 (ng/ml)</td>
<td>1.06</td>
<td>0.9-1.9</td>
</tr>
</tbody>
</table>

250 μg cosynotropin stimulation test [Synacthen*] (17-OHP, Cortisol)
0 min 30 min 60 min
0.32 ng/ml 0.31 ng/ml 0.26 ng/ml
0.7 μg/dl 0.75 μg/dl 0.73 μg/dl

INVESTIGATION

She was started on stress doses of hydrocortisone (HC) with dramatic improvement in the patient’s well-being. A biochemical profile at the age of 4·1·11 years showed appropriate HC levels and normalization of electrolytes and glucose.

DIAGNOSIS AND TREATMENT

Major manifestations
autoimmune Addison’s disease + autoimmune thyroid disease

Minor manifestations
cesi disease + history of ITP

REFERENCES


CONCLUSIONS

Clinical and biochemical profile of the patient at the age of 4 years and 11 months elucidates that the diagnosis was present for over 6 years.

This case report highlights that symptoms of adrenal insufficiency are often underestimated by patients and possibly physicians, leading to delayed diagnosis.