Severe High Blood Pressure with Renal Failure in a Neglected Case of 11β-Hydroxylase Deficient Congenital Adrenal Hyperplasia

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Introduction

- Congenital adrenal hyperplasia (CAH)
  - a group of autosomal recessive disorders
  - characterized by impaired cortisol synthesis
- 11β-Hydroxylase Deficient Congenital Adrenal Hyperplasia
  - an enzymatic defect in 11-beta-hydroxylase
    - the second most common variant of CAH (1)
    - accounts for approximately 5-8% of cases (1)
    - patients present with features of androgen excess (2)
    - approximately two thirds of patients also have high blood pressure (HBP), which is initially responsive to glucocorticoid replacement, but may become a chronic condition subsequently requiring standard antihypertensive therapy. (2)

Case Report

- Patient - A.P., female, 17 years
  - Medical history:
    - the first child of a consanguineous couple
    - Family history of CAH (2 third-degree relatives)
    - diagnosed with CAH in the neonatal period
    - ambiguous genitalia: clitoral and vaginal reconstruction at the age of 2 years.
    - female genetic sex
    - Barr chromatin 17% positive

Daily treatment with glucocorticoids was initiated, but the medical follow-up and self-administered therapy were extremely irregular.

Several hospital admissions due to acute adrenal insufficiency reported episode in April 2014, precipitated by an infectious disease, she presented with severe hypertension, hyperkalemia, renal failure

Endocrinology department – further investigations
- FE=142.5 cm (3.7 SD), G= 40 kg, BMI=20 kg/m²
- Breast development: Tanner stage III (fig.1)
- Male pattern baldness was present (fig.2)
- Hirsutism (fig.3) was evaluated based on Ferriman-Gallwey score (result = 15)
- Deepening of the voice
- Amenorrhea
- Severe hypertension (maximum value 220/140 mmHg)
- Complete baseline endocrine evaluation (before beginning steroid replacement) revealed absolute cortisol deficiency, with elevated aCTH

Laboratory findings and evolution:

<table>
<thead>
<tr>
<th>Date</th>
<th>ACTH (KU/ml)</th>
<th>Cortisol (μg/dl)</th>
<th>17-OH-Progesterone (ng/dl)</th>
<th>DHEAS (mg/dl)</th>
<th>Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>14/06/2014</td>
<td>42 ± 29</td>
<td>5.36 ± 0.6</td>
<td>27.60 ± 0.8</td>
<td>16.20 ± 0.2</td>
<td>Prednisol 15 mg/day Astron 0.1 mg /day</td>
</tr>
<tr>
<td>03/07/2014</td>
<td>15.6 ± 0.6</td>
<td>31.3 ± 0.03</td>
<td>-</td>
<td>2.28 ± 0.04</td>
<td>Prednisol 15 mg/day Astron 0.1 mg /week</td>
</tr>
<tr>
<td>20/02/2015</td>
<td>16.7 ± 0.6</td>
<td>5.22 ± 0.01</td>
<td>-</td>
<td>0.4 ± 0.02</td>
<td>Prednisol 15 mg/day Astron 0.1 mg /week</td>
</tr>
</tbody>
</table>

Irreversible consequences:
- severe hypertension
- left ventricular hypertrophy
- stage IV renal failure

Conclusions

- Care of adolescents with congenital adrenal hyperplasia has unique challenges. (4)
- Children with rare congenital diseases are now living full, productive lives and the issue of effectively transitioning these children to adulthood is a major public health problem.
- This case illustrates that CAH due to 11 beta hydroxylase deficiency can progress to severe acute and chronic complications.
- While early treatment to prevent hypertension is mandatory in patients with CAH, once renal failure occurs, renal transplantation may be the best choice of treatment.
- Early recognition and compliance to treatment can prevent morbidity and mortality.

References: