INTRODUCTION

- Steroid hormone biosynthesis is initiated by the cholesterol side-chain cleavage enzyme (P450scc), which converts cholesterol to pregnenolone, the precursor of all steroid hormones.
- This enzyme is encoded by the CYP11A1 gene and is responsible for glucocorticoid, mineralocorticoid and sex steroid synthesis in the adrenal glands, gonads and placenta.
- Early or late onset adrenal insufficiency, 46, XY sex development disorder, and hypergonadotropic hypogonadism may develop in patients with CYP11A1 mutation.
- Herein, a case presenting with adrenal insufficiency and found to have a CYP11A1 mutation will be presented.

CASE REPORT

- 3.5 year old male patient
- He was admitted to an external center with vomiting and change in consciousness.
- Upon detection of sodium 126 mEq / L and potassium 6.4 mEq / L in his examinations, he was referred to us with a pre-diagnosis of adrenal insufficiency.
- In the physical examination; height: 101.8 cm (0.34 SDS), weight: 16 kg (0.06 SDS), BMI: 15.4 kg/m2 (SDS: 0.24), BP: 90/60 mmHg, testicular volume 2 ml/2 ml stretched penile length 4 cm, hyperpigmentation was present on the skin, gums, axilla, and genital area.
- He was born at term, weighing 3400 g, with C/S.
- Parents were first degree cousins.
- The laboratory findings of the patient are presented in Table 1.
- In abdominal ultrasonography; bilateral adrenal glands were normal.
- Hydrocortisone and fludrocortisone treatments were initiated.
- Genetic analysis revealed a mutation in the CYP11A1 gene.
- At the last control of the patient at the age of 14; testicular volumes were determined as 25 ml, FSH: 8.4 μIU/ml, LH: 11.1 μIU/ml, Total testosterone: 62.5 ng/dl.
- The patient is being followed up for hypergonadotropic hypogonadism.

Table 1. The laboratory findings of the patient

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<table>
<thead>
<tr>
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<tbody>
<tr>
<td>Glucose (mg/dl)</td>
<td>89</td>
</tr>
<tr>
<td>Cortisol (μg/dl)</td>
<td>2.9</td>
</tr>
<tr>
<td>ACTH (pg/ml)</td>
<td>&gt;1250</td>
</tr>
<tr>
<td>Renin (ng/ml/h)</td>
<td>2.78</td>
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<tr>
<td>Aldosterone (pg/ml)</td>
<td>20</td>
</tr>
<tr>
<td>17-Hydroxyprogesterone (ng/ml)</td>
<td>0.08</td>
</tr>
<tr>
<td>DHEA-SO4 (μg/dl)</td>
<td>17.1</td>
</tr>
<tr>
<td>Peak cortisol response to ACTH stimulation test (μg/dl)</td>
<td>2.2</td>
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</table>

CONCLUSIONS

P450scc deficiency is a rare, new disease that can present with acute adrenal insufficiency at any time from infancy to early childhood. Since gonadal steroid synthesis may also be affected, patients should be carefully monitored in terms of gender development disorder and gonadal insufficiency.

REFERENCES


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