Introduction

Septo-optic dysplasia (SOD) is a rare, congenital condition that can be caused by mutations in HESX1, OTX2, SOX2, SOX3 genes. Symptoms of SOD include: optic nerve hypoplasia, hypopituitarism and midline brain abnormalities such as absence of septum pellucidum and/or corpus callosum. Hypopituitarism in SOD usually manifests as growth hormone deficiency followed by central hypothyroidism as well as deficiency of gonadotropins. Precocious puberty is a rare finding among patients with SOD, though it appeared among 2 of our patients.

Materials

2 girls diagnosed with SOD at the age: Patient No 1 - 3rd year of life and Patient No 2 – first year of life. At the moment girls are 10,5 and 13 years old.

Both patients had:
- hypernatremia observed since neonatal period
- Diagnosis: diabetes insipidus

**TREATMENT → DESMOPRESSIN**
- blindness caused by bilateral optic nerve hypoplasia
- developmental delay
- lack of septum pellucidum (MRI)
- obesity, tall stature and since 1st year
- accelerated bone age
- TSH insufficiency **TREATMENT → L-THYROXINE**
- GH deficiency
- ACTH insufficiency excluded

Patients did not receive:
- Growth Hormone treatment
- Hydrocortisonum

**Patient No 1** received **TREATMENT → LONG-ACTING GnRH ANALOGUE** because of rapid progression of puberty. Medication was withdrawn within less than 6 months due to recurrent urticaria.

**Patient No 2** was not treated with GnRH analogue.

Results

**GnRH Test**

<table>
<thead>
<tr>
<th>Hormone</th>
<th>Patient No 1</th>
<th>Patient No 2</th>
<th>Ranges</th>
</tr>
</thead>
<tbody>
<tr>
<td>IGF-1 [ng/mL]</td>
<td>21</td>
<td>37</td>
<td>82-262</td>
</tr>
<tr>
<td>E2 [pg/mL]</td>
<td>31,9</td>
<td>77,6</td>
<td>&lt;8</td>
</tr>
<tr>
<td>PRL [ng/mL]</td>
<td>7,04</td>
<td>9,77</td>
<td>2,7 – 19,7</td>
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<tr>
<td>TSH [ng/dl]</td>
<td>0,24</td>
<td>0,03</td>
<td>0,4-6,0</td>
</tr>
<tr>
<td>ft4 [pmol/L]</td>
<td>0,78</td>
<td>0,72</td>
<td>1,8-4,1</td>
</tr>
<tr>
<td>Cortisol – baseline [ug/dl]</td>
<td>5,2</td>
<td>3,3</td>
<td>5-20</td>
</tr>
<tr>
<td>Cortisol in 60’ ACTH stimulation test [ug/dl]</td>
<td>26,6</td>
<td>24,2</td>
<td></td>
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</tbody>
</table>

**Conclusions**

In most cases SOD is associated with multiple pituitary hormone deficiency, including gonadotropins. This report implicates the necessity for long-term monitoring as pituitary insufficiency may evolve over time and does not rule out precocious puberty.

References