**Introduction**

Reeves (1941) described absence of the septum pellucidum and an optic nerve anomaly occurring together in the same patient, but De Morsier (1956) was the first convincingly to prove an association. At the beginning, growth retardation was not recognized as part of the syndrome, but in 1970 Hoyt et al described the association between SOD and hormonal insufficiency.

Septo-optic dysplasia (SOD) is a congenital affection characterized by classic triad: optic nerve hypoplasia, hypothalamic-pituitary endocrine deficits and midline abnormalities of the brain including absence of septum pellucidum and corpus callosum dysgenesis. It is diagnosed usually in infancy and has a variable presentation that includes visual abnormalities, neurologic disorders, and/or hypothalamic-pituitary endocrine deficits.

The majority of SOD are sporadic, but familial cases have been described.

**Case presentation**

The patient is a 5 year and 10 months old Algerian girl. She was born after 39 weeks gestation by normal vaginal delivery and was the first child of consanguineous parents. She had no asphyxia and her birth weight was 2800 g. After birth, she developed noncholicastic jaundice and was treated with ultraviolet light. She had no developmental delay or mental retardation. Here parents were healthy and there was no family history of growth disorders.

At birth, bilateral congenital nystagmus and strabismus was noted. But right blindness was suspected by parents only at age of 2 years but confirmed at 4 years old when ophthalmological evaluation revealed pallor and hypoprophic right optic nerve mild pallor in the left eye. The intraocular pressure was normal in both eyes. Hearing was normal.

Brain magnetic resonance imaging demonstrate hypoplasia of the optic nerves, chiasm, and optic tracts mainly on the right with small pituitary gland. Besides above malformations, other cerebral malformations were not detected.

At 5 years 10 months she was referred to our hospital because of short stature.

Physical examination of the patient was weight 23 kg = 0,5SD, height 117 cm = 0,5SD but at -2,5SD from her target size (= 184,5cm), blood pressure 107/60 mmHg. We noted a retardation of Bone age: three old.

The endocrine examination at this time showed symptoms suggestive of hypothyroidism such as fatigue, chronic constipation, dry skin, and hair loss. The neurological examination was normal.

**Endocrine investigations demonstrated somatotroph, corticotropic, and thyrotroph deficiencies (Table 1).**

<table>
<thead>
<tr>
<th>Patient</th>
<th>Results</th>
<th>Normal range</th>
</tr>
</thead>
<tbody>
<tr>
<td>ACTH</td>
<td>&lt; 1 pg/ml</td>
<td>5 – 46</td>
</tr>
<tr>
<td>CORTISOL 8h</td>
<td>35 nmol/l</td>
<td>138-551</td>
</tr>
<tr>
<td>TSH</td>
<td>8.68 mU/L</td>
<td>0.1-4</td>
</tr>
<tr>
<td>FT4</td>
<td>6 pmol/l</td>
<td>12 – 22</td>
</tr>
<tr>
<td>IGF 1</td>
<td>10 ng/ml</td>
<td>8 - 293</td>
</tr>
</tbody>
</table>

She was treated with hydrocortisone, L-thyroxine and GH.

Unfortunately, the parents did not reconsult and the genetic study could not be realized at our small patient.

**Discussion**

Septo-optic dysplasia is a rare congenital disorder. It has an estimated prevalence of 1 in 50,000 births. Both sexes are equally affected [1].

The diagnosis of septo-optic dysplasia (SOD) is a clinical one and can be made when two or more features of the classical triad of optic nerve hypoplasia, pituitary hormone abnormalities and midline brain defects, including agenesis of the septum pellucidum and/or corpus callosum, are present [2].

The clinical presentation may be mild or extremely severe and depending upon the type and extent of associated anomalies. Most cases are detected in infancy [3].

Examination of the optic nerves is best performed by means of clinical evaluation since the imaging of the optic nerves and chiasm are normal in about half the patients with SOD [4].

Nearly two thirds of SOD patients have pituitary hormones deficiency. It is not always clear if the hypothyroidism is due to a primary pituitary dysfunction or is secondary to a hypothalamic dysfunction [5,6].

Morishima and Aranoff cite that 30% of SOD cases have complete manifestations, 62% have the complication of hypothyroidism and 60% have an absent septum pellucidum [7].

The majority of cases of SOD are sporadic. In consanguineous families, the likelihood of recessive inheritance is significantly higher, the likely recurrence risk is one in four [8].

The possibility of diagnosing prenatal cerebral changes by three-dimensional ultrasound and fetal MRI has been reported in the literature. MRI is usually most helpful to visualize midline and other brain abnormalities in children with optic nerve hypoplasia [9].

**Conclusion:**

A multi-disciplinary team approach is necessary in the management of these patients. This team included a pediatric endocrinologist, pediatric neurologist and pediatric ophthalmologist.

The prognosis varies according to the severity of the disease. An early diagnosis, allowing the correction of hormonal deficits in due time, is associated with a more favorable evolution.

**REFERENCES**