Sex development is a process that directs both the bi-potential gonads to become either a testis or an ovary, and the consequent differentiation of internal ducts and external genitalia. Disorders of sex development (DSD) are those congenital conditions in which development of chromosomal, gonadal, or anatomical sex is atypical. These individuals can be classified according to their karyotype in: sex chromosome DSD, 46,XY DSD and 46,XX DSD.

### Results

Twenty one gonads (corresponding to 12 patients) showed ovotesticular characteristics and 4 (2 patients) showed only testicular parenchyma. OT: ovotestis, T: testis, M: male, F: female, CA: chronological age, GB: gonadoblastoma, UGT: undifferentiated gonadal tissue

**Figure 1. Histological analysis of the patients’ gonads**

- **Table 1. Clinical material and characteristics of the gonads**

<table>
<thead>
<tr>
<th>Cases</th>
<th>Total 14</th>
<th>Sex of rearing (M/F)</th>
<th>CA (months)</th>
<th>OCT ¾+</th>
<th>GB/UGT</th>
<th>OCT ¾+ &amp; GB/UGT</th>
</tr>
</thead>
<tbody>
<tr>
<td>OT</td>
<td>12</td>
<td>M:8 F:4</td>
<td>1.16 - 50 (M=14)</td>
<td>3</td>
<td>6 (3/3)</td>
<td>3 (2/1)</td>
</tr>
<tr>
<td>T</td>
<td>2</td>
<td>M:2 F:0</td>
<td>5-26 (M=15.5)</td>
<td>1</td>
<td>0</td>
<td>0</td>
</tr>
</tbody>
</table>

OT: ovotestis, T: testis, 1 (2m) dysembryotic testis with seminiferous cords and ovarian follicles, 25-3 (14m) Meandering and dysgenetic seminiferous cords, one of them with an oogonia cell inside (50m), 3 (1.5m) Leydig cell with prominent and primary follicles, and seminiferous cords, 40x.

T: testis, 4 (2m) Branched seminiferous cords in tunica albuginea, and sciatric germ cells, 20x, 5 and 6 (3.7m) Righ gonad, seminiferous tubules, with a significantly thick basal membrane, vacuolated cells, Sertoli cells with sparse cytoplasmic and eosinophilic cytoplasm, oil droplets in the cytoplasm, 40x, and GB, gonadoblastoma, 7 (3m) Left gonad, oval shape structure with germ cells, sex cord cells, and lumen, 40x, 8 (1.5m) Testicular area of an ovotesticular gonad, needs reassembling GB, 40x, 9 (20m) Ovarial structure with two types of cells and a thick membrane, 40x.

**Table 2. IHC staining pattern**

<table>
<thead>
<tr>
<th>Cases</th>
<th>IHC</th>
<th>SOX9</th>
<th>FOXL2</th>
<th>OCT ¾+</th>
</tr>
</thead>
<tbody>
<tr>
<td>3 GB</td>
<td>2</td>
<td>1</td>
<td>1</td>
<td></td>
</tr>
<tr>
<td>3 UGT</td>
<td>2’</td>
<td>2’</td>
<td>2’</td>
<td></td>
</tr>
</tbody>
</table>

**Figure 2. SOX9, FOXL2 and OCT3/4 immunoexpression**

**Conclusions**

- A careful histological analysis is crucial for the diagnosis. Nevertheless, the addition of several IHC markers is important to achieve a thorough characterization of the gonads.
- In all testicular parenchyma signs of dysgenesis were found.
- A second biopsy in 2 former testicular cases revealed the presence of ovarian parenchyma.
- Considering the histopathological findings in early childhood, a close clinical follow up of patients with a specialized DSD team is suggested.

**Discussion**

- How representative is a biopsy?  
- How important are SOX9 and FOXL2 in histopathological assessment of DSD gonads?

**References**

- Suárez J et al. Gonadoblastomas and selected other aspects of gonadal pathology in young patients with disorders of sex development 2014.