46,XX ovotesticular DSD in the absence of SRY gene associated to SOX3 duplication

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**Background**

Ovotesticular DSD is a rare disorder defined by the presence of both ovarian and testicular tissue in the same individual.

SRY is present in approximately 1/3 of patients with 46,XX ovotesticular DSD. In SRY-negative ovotesticular DSD, the mechanism responsible for the presence of testicular tissue is not yet understood.

**Case presentation**

Male patient referred for hypospadias and bilateral cryptorchidism at 2.5 years of age.

He had a trophic phallus (32 mm x 13 mm) with coronal hypospadias and hypoplastic scrotum. No palpable gonads.

**Results**

<table>
<thead>
<tr>
<th>Hormonal laboratory</th>
<th>Patient</th>
<th>Normal values (range)</th>
</tr>
</thead>
<tbody>
<tr>
<td>LH (IU/litre)</td>
<td>&lt;0.01</td>
<td>0.05-0.3</td>
</tr>
<tr>
<td>FSH (IU/litre)</td>
<td>0.73</td>
<td>0.2-0.9</td>
</tr>
<tr>
<td>Testosterone (ng/dl), Basal</td>
<td>&lt;10</td>
<td>10-32</td>
</tr>
<tr>
<td>Post hCG</td>
<td>30</td>
<td>&gt;150</td>
</tr>
<tr>
<td>AMH (pmol/litre)</td>
<td>216</td>
<td>300-1400</td>
</tr>
</tbody>
</table>

Karyotype: 46,XX (60)

**Discussion**

- Ovotesticular DSD
  - 82% 46,XX
  - 35% SRY+
  - 18% Other karyotypes carrying Y sequences

**Conclusion**

This is the first case of SRY-negative 46,XX Ovotesticular DSD in whom a SOX3 duplication is reported.

These results are in line with evidence in mice indicating that, in the absence of SRY, gain-of-function of SOX3 induces testis differentiation in the XX biotopical gonad.