Table S3: Compilation of all reported SOX9 mutations associated to CD and ACD.

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**SOX9 – C-terminal extensions**

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<th>Survival</th>
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### SOX9 – C-terminal extensions [continued]

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### SOX9 – missense mutations

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<td>HMG</td>
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### SOX9 – missense mutations [continued]

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#### SOX9 – splice-site mutations

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<th>Phenotype</th>
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<th>Survival</th>
<th>Reference</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>IVS1-2A&gt;C</td>
<td>-</td>
<td>-</td>
<td>46,XY</td>
<td>CD</td>
<td>F*</td>
<td>NS</td>
<td>[Kwok et al., 1995]</td>
</tr>
<tr>
<td></td>
<td>IVS1-2A&gt;G</td>
<td>-</td>
<td>-</td>
<td>46,XX</td>
<td>CD</td>
<td>F</td>
<td>NS [neonatal]</td>
<td>[This study]</td>
</tr>
<tr>
<td></td>
<td>IVS2+1G&gt;A</td>
<td>-</td>
<td>-</td>
<td>46,XY</td>
<td>CD</td>
<td>F*</td>
<td>NS [neonatal]</td>
<td>[Wagner et al., 1994]</td>
</tr>
</tbody>
</table>

#### SOX9 – in-phase deletions

<table>
<thead>
<tr>
<th>Exon</th>
<th>cDNA</th>
<th>Protein</th>
<th>Domain</th>
<th>Karyotype</th>
<th>Phenotype</th>
<th>Sex</th>
<th>Survival</th>
<th>Reference</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>c.196_225del30</td>
<td>p.E66_E75del10</td>
<td>Dimer</td>
<td>46,XX</td>
<td>CD</td>
<td>F</td>
<td>&gt;1 day</td>
<td>[Sock et al., 2003]</td>
</tr>
<tr>
<td>3</td>
<td>c.1047_1130del84</td>
<td>p.P301_P328del28  d</td>
<td>PQA</td>
<td>46,XX</td>
<td>ACD</td>
<td>F*</td>
<td>Alive at 1.8 yrs</td>
<td>[Chen et al., 2012]</td>
</tr>
</tbody>
</table>

*With dysgerminoma. \(^b\)With true hermaphroditism. \(^c\)Patient characterized as ‘intersex’. \(^d\)Mutation in homozygosis. \(^e\)Mild CD/ small patella syndrome. M: male; [M]: male with hypospadias; {M}: male with small penis; M*: 46,XX sex reversal; F: female; F*: 46,XY, sex reversal, as proposed by Fonseca et al. [32]. ACD: acampomelic campomelic dysplasia; CD: campomelic dysplasia; Dimer: dimerization domain; HMG: high-mobility group DNA binding domain; NS: not specified; PQA: proline-glutamine-alanine-rich domain; TA: transactivation domain.


