Mosaicism ratios of 45,X to 46,XY idicY can explain a phenotype in a case with mixed gonadal dysgenesis.

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Take-home message
The sclerotic lesions (streak gonad and coarctation of the aorta, characteristics of Turner’s syndrome) in a mixed gonadal dysplasia patient with 45,X/46,XY, idicY are attributable to higher proportions of 45,X cells.

Methods
Fluorescence in situ hybridization (FISH) and/or chromogenic in situ hybridization (CISH) was performed for the gonadectomy and aorta specimens to determine the proportion of cells with 45,X to those with 46,XY.

A normal testis with a 46,XY karyotype (1 year old) and an ovary Tuner’s syndrome were also similarly examined as controls. Each subject gave their informed consent and patient anonymity was preserved.

Results
The distribution and proportion of the 45,X cells in the gonads, aortic tissue and control testicular tissues are summarised in Table 2.

Table 2: The proportion and distribution of 45,X cells in the aortic, gonadal and Müllerian duct tissues

<table>
<thead>
<tr>
<th>%45,X cells</th>
<th>Patient</th>
<th>Control</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>45,X</td>
<td>46,XY</td>
</tr>
<tr>
<td>Peripheral lymphocytes</td>
<td>23.3% (7/30)</td>
<td>100%</td>
</tr>
<tr>
<td>Gonadal tissue</td>
<td>Streak gonad</td>
<td>98.7% (224/227)</td>
</tr>
<tr>
<td>Müllerian duct</td>
<td>100% (100/100)</td>
<td>100% (100/100)</td>
</tr>
<tr>
<td>Aortic tissue</td>
<td>87.6% (149/170)</td>
<td>NA</td>
</tr>
</tbody>
</table>

Discussion
1) Higher proportions of 45,X cells might cause a regressive change (apoptosis and stromagenesis), leading to sclerotic lesions such as streak gonad and coarctation of the aorta in MGD.

2) Since MGD is sometimes associated with cardiovascular anomalies, all patients with 45,X cell lines should be proposed for both endocrine and cardiologic follow-up as is performed for Turner syndrome.

References:
6. J.W.Keepling, Fetal Pathology, monosomy X, p28-29

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