Turner syndrome (TS) is a genetic condition with different phenotypic expressions depending on karyotype. Due to genetic prenatal testing, its prevalence is getting lower.

**AIM**

The objective was to analyze the presence of different comorbidities associated with TS according to the karyotype and evaluate if there is follow-up in adulthood.

**METHOD**

Descriptive retrospective study including patients genetically diagnosed with Turner syndrome between 1984 to 2019.

Clinical and follow-up variables

Patients classified according to karyotype (monosomy X/mosaicism/isochromosome)

Follow-up to adulthood, gynecologic screening and pregnancy registered

**RESULTS**

- **70 patients**
  - **22 prenatal diagnosis (aborted)**
  - **48 postnatal diagnosis → 38 collected patients**

- **Mean age:** 32.6 years old (SD 17.7)

- **Prescribed treatment**
  - **Growth hormone:** 50% of cases (89% of them were born in the last 25 years)
  - **Hormonal replacement therapy:** 73% of cases

- **Echocardiographic screening:** 81.6% of patients (3 cases with aortic root dilatation, 4 with bicuspid aortic valve and 3 with coarctation of the aorta)

- **Renal screening:** 81.5% of patients (3 cases with renal fusion and 1 case of abnormal renal rotation)

**Conclusions**

Comorbidities are the main issue in follow-up of TS as they affect most part of the patients. It is important to analyze comorbidities depending on the karyotype.

Taking into account prognostic implication of cardiological and renal disease, regular screening is mandatory in all patients.

Since most patients do not have follow-up in adulthood, it is important to take special care on transition from paediatric to adult healthcare.

**REFERENCES**


**ACKNOWLEDGEMENTS**

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**CONTACT INFORMATION**

raquelcorripio@gmail.com

**TABLE**

<table>
<thead>
<tr>
<th>Comorbidities</th>
<th>Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Thyroiditis</td>
<td>19</td>
</tr>
<tr>
<td>High blood pressure</td>
<td>11</td>
</tr>
<tr>
<td>Neuropsychological complications</td>
<td>12</td>
</tr>
<tr>
<td>Osteoporosis</td>
<td>6</td>
</tr>
<tr>
<td>Diabetes mellitus</td>
<td>4</td>
</tr>
<tr>
<td>Celiac disease</td>
<td>3</td>
</tr>
<tr>
<td>Neoplastic disease</td>
<td>3</td>
</tr>
</tbody>
</table>

**Graph**

- **Karyotype**
  - 49% 45X
  - 9% Mosaicism
  - 42% Isochromosome

- **Autoimmune disorders according to karyotype:**
  - **Isochromosome:** 100% of patients
  - **Monosomy X:** 50% of patients
  - **Mosaicism:** 60% of patients