

## **P29. Evaluation of Differences in Clinical Characteristics Between Patients with the Classical Karyotype of Turner Syndrome and Mosaics**

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**Context:** Turner Syndrome (TS) is a chromosome disorder characterized by a total or partial loss of a chromosome X. It has been described as affecting 3% of all conceptions, and having an incidence rate of 1:2500 newborn live. The classical karyotype, 45X is frequently associated with higher comorbidities rates. **Objective:** The aim of this study is to compare clinical characteristics between patients with the classical karyotype of TS (45X) with mosaic patients.

**Methods:** Retrospective data extracted from charts of 33 TS patients and 19 stigmas were analyzed. Karyotype was characterized by G band with a mean of 26±13 cells analyzed. **Population:** TS Patients **Interventions:** None. **Outcome Measures:** clinical characteristics **Results:** 45X karyotype was found in 39% patients. Other found were: 45X/46XX (6%), 45X/46Xi (15.6%), 45X/46XY (3%), 45X/46X+mar (9%), 45X/46Xr (3%), 46Xi (12%), 45X/47XXX (3%), 45X/44XX/45Xi(y) (3%), 45X/46XX+mar+mar/47X-c+mar+mar (3%) and 45X/46Xpsuidic(x) q24 (3%). All patients were adults with Tanner 5. When dividing patients in two distinct groups (45X vs Other), we found no differences in age at diagnosis (14.0±8.7 vs 13.3±5.1 years old, p=0.77), final height (1.43±0.5 vs 1.43±0.5 m, p=0.91), wingspan (1.43±0.7 vs 1.45±0.1 m, p=0.46) and proportion of spontaneous telarche (23% vs 42%, p=0.27). There was a trend towards a higher rate of spontaneous menarche in mosaics (0 vs 21%, p=0.08). The only patient with signs of virilization was the one with the 45X/46XY karyotype. Regarding differences in stigmas frequency, lower lip corner orientation 54%vs16%, (p=0.02) and cubitus valgus (92% vs 53%, p=0.02) were higher in 45X patients than mosaics. Only 45X patients presented with square, shield-like chest (31% vs 0, p=0.01). Crowded teeth were more frequently found in mosaics than in 45X patients (42% vs 7%, p=0.03, respectively). There were no differences in the proportion of renal and cardiac malformation, mental retardation, hearing deficiency, hypotiroidism, diabetes, dyslipidemia, and liver enzymes abnormalities between the two groups. Celiac disease was not found.

**Conclusions:** In our study, mosaics patients had more chance of spontaneous menarche. Although some stigmas were more commonly found in a group over the other, there were no differences in the proportion of associated diseases and malformations between the two groups. Our data suggests that patients in the adult range with the 45X karyotype had the same comorbidities as mosaics.

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