Objective: to analyze karyotype of Turner's syndrome (TS) patients in two tissues of different lineage, and to assess which correlates better with phenotype.

Methods: an observational study was designed in the Gynaecological Endocrinology Unit of a teaching hospital. Patients between 20-50 years old, diagnosed of TS by blood karyotype, were included. A new 50-cell count blood karyotype and a urethral cell karyotype from urine samples were performed. Some TS-related comorbidities were collected.

Results: Twenty-seven TS patients were included. Urine cultures of 12 patients were contaminated by microorganisms. With 50-cell count blood karyotype, 3 cryptic mosaicisms were found. Six patients with mosaicism in blood karyotype showed pure monosomy in urine karyotype. Correlations exist between blood karyotype and phenotype considering spontaneous menarche, height, dysmorphology, congenital malformations and hypothyroidism, whereas it did not appear in urine analysis.

Conclusions: T-lymphocytes karyotype from blood samples is the gold standard technique. 50-cell count may be considered if TS or ovarian failure is suspected, to detect cryptic mosaicisms. Urethral cell culture presents technical difficulties and limitations, due to the easier lost of abnormal X-chromosome that worsens its correlation with phenotype. A partial correlation between blood karyotype and phenotype exists.