

Karyotype is associated with timing of ovarian failure in women with Turner syndrome.

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Journal of Pediatric Endocrinology and Metabolism 2021; 34(3): 319–323

Background : Turner Syndrome (TS) or Monosomy X is the most common sex chromosome abnormality in females and is associated with premature ovarian failure in almost 100% of the cases.

Objective: To characterize the age of ovarian failure in Turner Syndrome (TS) patients by karyotype.

Methods: Retrospective cohort study of individuals with TS at an academic university hospital in USA. Subjects were seen in TS Clinic at UNC Hospital between 2014 and 2018. Individuals were analyzed by karyotype category (45X, 45X/46XX mosaicism, miscellaneous) and percentage of 45X cells. Age at follicle-stimulating hormone (FSH) > 30 IU/ml was defined as the age at loss of ovarian function. A total of 79 patients were included in the study. They were divided into 3 karyotype categories: Group 1 (45X, n=30); group 2 (45X/46 XX, n=13) mosaic Turner ; group 3 (miscellaneous with no Y chromosome material, n=36).

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RESULTS: Mean age of the children was 13 years (SD: 5.4) and the median age at diagnosis of TS in the population was 1.0 years

- Fifty-five of 79 (70%) patients had evidence of ovarian failure, median age of failure 11 years
- Ovarian failure was more prevalent among individuals with 45X karyotype (100%). (Group 1)
- Of patients with 45X/46XX karyotype or Mosaic Turner (group 2), only one individual (7.7%) had experienced ovarian failure and in the miscellaneous karyotype category (group 3), 24 of 36 (66.6%) had ovarian failure.
- The median age of ovarian failure for 45X patients (n=30) was 10 years old, which is significantly younger than other karyotypes (n=49), which had a median age of 15 years for ovarian failure, $p < 0.01$.

DISCUSSION : Understanding of the timing of ovarian failure in children with Turner Syndrome is important for appropriate counseling and planning of interventions. In this study, 45X individuals had complete and early loss of ovarian function compared to the other karyotypes. The mosaic and miscellaneous karyotypes of TS had later age of onset of ovarian failure and were significantly older than with 45X patients. Mosaic Turner individuals have the highest chance of preserved ovarian function and fertility depending on the percentage of 45X cells in the peripheral blood karyotype. The late age of onset of ovarian failure in non-classic Turner phenotypes may provide a window of opportunity for oocyte cryopreservation and hence fertility options in later life.

Key messages: This study from the US has shown that timing of ovarian failure in Turner syndrome is related to the karyotype. Measurement of FSH (>30 IU/ml) and AntiMullerian Hormone levels (AMH <0.01ng/ml) may be used to assess ovarian failure in Turner syndrome. **Mosaic Turner syndrome has the highest chance of spontaneous puberty and possible fertility and this is inversely associated with the percentage of 45X cells in the peripheral blood.**

EXPERT COMMENT

“With advances in fertility preservation options, an understanding of the timing of ovarian failure in Turner Syndrome is important for counseling and planning of treatment options like sex steroid replacement and oocyte cryopreservation for future fertility. Karyotype has a definite role in predicting ovarian failure timing according to this study.”

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With warm regards,

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Editor – Academic Pearls
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Reference

Fitz VW, Law JR, Peavey M. Karyotype is associated with timing of ovarian failure in women with Turner syndrome. J Pediatr Endocrinol Metab. 2021 Feb 26;34(3):319-323. doi: 10.1515/jpem-2020-0304. PMID: 33650387.