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MOSAIC FORM OF TURNER SYNDROME WITH NORMAL STATURE: A RARE CLINICAL PRESENTATION

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ABSTRACT

Mosaic TS may present with subtle or absent somatic features and normal height, complicating diagnosis. We report a 16-year-old female with primary amenorrhea and delayed puberty, but normal height (164 cm) and absent dysmorphic features. Cytogenetic analysis revealed mosaicism: 45, X [10]/47, XXX [20].

Introduction

Turner syndrome (TS) is a chromosomal disorder resulting from complete or partial monosomy of the X chromosome, with clinical manifestations influenced by the degree and pattern of mosaicism. Classical 45, X TS typically presents with short stature, primary ovarian insufficiency, and characteristic somatic stigmata such as webbed neck, shield chest, cubitus valgus, and high-arched palate. However, the presence of an additional X chromosome, as in 47, XXX mosaicism, can attenuate these classical features, contributing to preserved stature and subtle phenotypic expression through compensatory effects on both somatic growth and gonadal development.

We report a 16-year-old female presenting with absent secondary sexual characteristics, including Tanner stage I breast development and lack of pubic and axillary hair. Menarche had not occurred by age 16. Parental histories indicated normal pubertal timing. Mid-parental height was 160 cm; the patient's height was 164 cm (56.8th percentile) and weight 47 kg (34.9th percentile). Bone age was delayed at 13 years, with a predicted adult height of 175.1 ± 5.9 cm, suggesting growth potential exceeding the familial target.

Physical examination revealed no classical somatic features of TS, including webbed neck, high-arched palate, shield chest, or cubitus valgus. Pelvic ultrasound and MRI demonstrated a hypoplastic uterus and underdeveloped ovaries, without Wolffian structures. Cardiac and renal evaluations were unremarkable. Thyroid function and glucose metabolism were within normal limits. Hormonal evaluation was consistent with severe ovarian insufficiency: anti-Müllerian hormone (AMH) <0.1 ng/mL, follicle-

stimulating hormone (FSH) -112.1 mIU/mL (reference 3.0–12.0), luteinizing hormone (LH) -63.3 mIU/mL (1.9–11.6), and estradiol 133.9 pmol/L (68–1269).

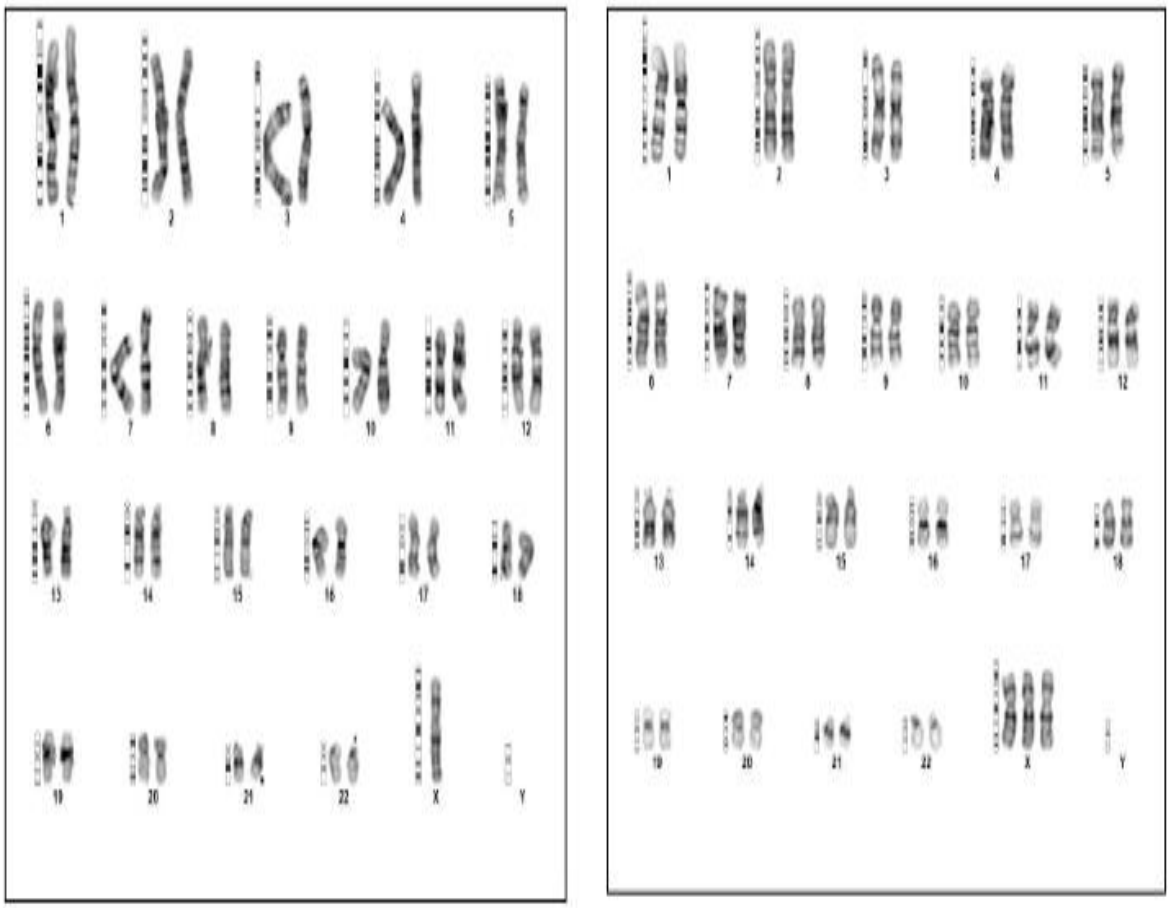

Figure 1. Cytogenetic analysis revealed mosaicism: 45, X [10]/47, XXX [20].

This case underscores the broad phenotypic variability of mosaic TS. Individuals with a 47, XXX cell line may retain near-normal ovarian function, spontaneous pubertal development, and normal or increased stature. Population studies indicate that 47, XXX individuals are, on average ~5.3 cm taller than 46, XX controls, while 45, X/47, XXX mosaics exhibit short stature in only ~64.3% of cases, markedly lower than classic 45, X TS.

The relative growth advantage in mosaic individuals may arise from gene dosage effects within the pseudoautosomal region (PAR1), which escapes X-inactivation, and from the presence of a functional second or third X chromosome in specific tissues. Tissue-specific mosaicism further complicates genotype-phenotype correlations, as peripheral blood karyotypes may not accurately reflect the cellular composition of skeletal or gonadal tissue.

Delayed diagnosis is frequent in individuals lacking classical TS stigmata, yet early recognition is crucial for monitoring cardiovascular, renal, endocrine, skeletal, and psychosocial health. Despite normal stature and minimal physical abnormalities, our patient exhibited markedly elevated gonadotropins and undetectable AMH, confirming ovarian insufficiency. This emphasizes the importance of cytogenetic evaluation in adolescents presenting with unexplained primary amenorrhea.

In summary, this case highlights the wide phenotypic spectrum of mosaic Turner syndrome and demonstrates that absence of classical somatic features should not preclude genetic testing. Early cytogenetic diagnosis enables timely endocrine management, appropriate reproductive counseling, and surveillance for comorbidities, optimizing long-term health outcomes.

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