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Case Report

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Triple Mosaic Turner Syndrome: An Exceptionally Rare Cytogenetic Variant

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Abstract

Background: Turner syndrome (TS) demonstrates substantial clinical, phenotypic, and cytogenetic heterogeneity, particularly in mosaic forms that often create diagnostic and prognostic challenges. Triple mosaic variants involving the X chromosome are exceedingly rare and remain poorly characterized, limiting the understanding of their clinical significance and genotype-phenotype correlations. Reporting such a rare mosaic pattern contributes to expanding the cytogenetic spectrum of TS and aids in the clinical interpretation of complex mosaic outcomes associated with primary amenorrhea (PA).

Case Presentation: A comprehensive clinical, endocrinological, cytogenetic, and molecular cytogenetic evaluation was carried out on a 17-year-old female presenting with PA. Peripheral blood lymphocyte chromosomes were analyzed using conventional G-banded karyotyping and fluorescence in situ hybridization (FISH) with the DXZ1 centromeric probe, following the AGT Cytogenetics Laboratory Manual (4th edition). A conventional karyotype revealed a rare triple mosaic karyotype pattern of mos 45,X[30]/46,X,i(X)(q10)[7]/46,XX[3], consistent with TS. FISH confirmed the presence of X chromosome mosaicism by demonstrating nuclei with one and two X-centromeric signals; however, it did not differentiate between the 46,XX and 46,X,i(X)(q10) cell populations.

Conclusion: The current case highlights the diagnostic significance of the rare triple mosaic TS karyotype in the evaluation of PA. Although the predominance of the 45,X cell population may have influenced the phenotype, genotype-phenotype correlations should be interpreted cautiously because mosaicism was assessed only in peripheral blood lymphocytes. This rare cytogenetic finding presents diagnostic challenges that require integrated conventional G-banded karyotyping and complementary FISH for accurate cytogenetic characterization.

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Introduction

Medical conditions pertaining to pubertal development and menstrual dysfunction constitute a significant concern in the field of reproductive medicine and often reflect underlying genetic or endocrine abnormalities. Among these conditions, primary amenorrhea (PA) is one of the most common clinical presentations encountered in adolescent gynecology. PA is described as the absence of menstruation by 15 years of age in females demonstrating typical secondary sexual characteristics or by 13 years of age in those without pubertal development (1). According to the American College of Obstetricians and Gynecologists (ACOG), the average age of menarche is approximately 12.43 years; however, this mean age significantly varies among popula-

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tions, with an earlier onset noted in developed countries relative to developing nations. The frequency of PA is estimated to be between 2% and 5% among adolescent girls, with chromosomal abnormalities accounting for roughly 50% of the cases (2). Clinically, PA signifies a multifaceted clinical outcome arising from disruptions in the hypothalamic-pituitary-ovarian axis, anatomical irregularities of the reproductive system, or genetic defects influencing gonadal maturation. Prevalent etiologies encompass Müllerian agenesis, hypogonadotropic hypogonadism, and other chromosomal abnormalities (3).

Turner syndrome (TS) is the most prevalent chromosomal disorder, occurring in approximately 1 in 2,000-2,500 live-born females. Classic TS results from complete or partial monosomy of one X chromosome (45,X) and displays distinctive phenotypic traits, including short stature, delayed puberty, webbed neck, shield chest, heart abnormalities, and primary ovarian insufficiency (4).

However, the phenotypic expression is highly variable, especially in mosaic forms, which account for approximately 30-40% of TS cases and arises from postzygotic nondisjunction or mitotic errors occurring during early embryogenesis. Among these, isochromosome Xq is one of the most prevalent structural Xchromosome anomalies in TS. It results from atypical centromeric division, leading to the duplication of the long arm (Xq) and deletion of the short arm (Xp). Therefore, haploinsufficiency of genes located on Xp, particularly those associated with growth and ovarian development, leads to short stature, gonadal dysgenesis, and primary ovarian insufficiency, whereas duplication of Xq may also affect the clinical phenotype. The coexistence of an isochromosome Xq with additional mosaic cell lines further contributes to cytogenetic complexity and leads to variable genotype-phenotype associations, making accurate diagnosis particularly challenging. Rare triple mosaic karyotypes, such as mos 45,X[30]/46,X,i(X)(q10)[7]/46,XX[3], are exceedingly rare and sparsely documented in the literature (5-7). The relative proportion of each cell lines may profoundly influence both phenotypic severity and reproductive outcomes, highlighting the necessity for cytogenetic characterization by complementary diagnostic methods. In addition, the identification of the occult Y-chromosomal material is clinically important in TS because its presence has been associated with an increased risk of gonadoblastoma and may influence patients management and genetic counseling. Therefore, accurate cytogenetic characterization of complex mosaicism is important for establishing the diagnosis and understanding genotype-phenotype association.

The present case highlights a rare triple mosaic TS variant 45,X[30]/46,X,i(X)(q10)[7]/46,XX[3], identified in a 17-year-old female presenting with PA. This case highlights the diagnostic value of integrating conventional G-banding with FISH for accurate characterization of complex X chromosome and emphasizes the importance of comprehensive cytogenetic evaluation in a patient with atypical TS.

Case Presentation

A 17-year-old female presented with PA and failure to reach menarche. There was no history of dysmenorrhea or spontaneous pubertal development. The patient was referred for further evaluation at inDNA Center for Research and Innovation in Molecular Diagnostics, inDNA Life Sciences Private Limited, located in Bhubaneswar, Odisha, India, on 9 October 2025, to rule out any underlying endocrine or genetic causes.

Clinical findings: The patient presented with a thin body habitus, with a height of 136 *cm* and a body weight of 30.8 *kg*. She exhibited delayed pubertal development characterized by Tanner stage II breast development and sparse pubic hair. Dysmorphic features included a heart-shaped face, low-set ear, and cubitus valgus which are consistent with the phenotypic spectrum of TS. Additionally, no other obvious phenotypic anomalies or classical dysmorphic features suggestive of TS were observed (Figure 1).

Laboratory investigation: Endocrine evaluation performed through the chemiluminescence assay revealed both the luteinizing hormone (LH) and follicle-stimulating hormone (FSH) levels were significantly elevated, consistent with hypergonadotropic hypogonadism. Additionally, thyroid function testing revealed increased levels of thyroid-stimulating hormone (TSH), confirming hypothyroidism (Table 1).

Radiological findings: Pelvic transabdominal ultrasonographic evaluation revealed non-visualization of the uterus, likely secondary to marked uterine hypoplasia associated with profound estrogen deficiency, and the presence of bilateral streak gonads with a hypoechogenic appearance, each measuring around 0.2 cm^3, suggestive of

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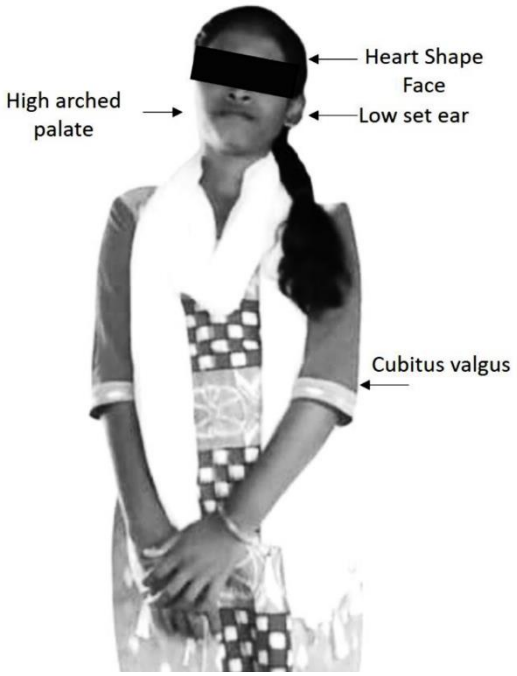


Figure 1. Phenotypic presentation of the mosaic Turner variant

gonadal dysgenesis. No identifiable ovarian follicles or normal ovarian tissue were visualized. Based on these clinical, radiological, and biochemical findings (Table 1), a strong suspicion of an underlying sex chromosome anomaly was raised. Consequently, a peripheral blood sample was collected for cytogenetic investigation.

Diagnostic assessment

Cytogenetic analysis: Written informed consent for publication of the clinical information, cytogenetic findings, and accompanying clinical images was obtained from the patient's parent prior to sample collection. The study was conducted in accordance with the ethical principles of the Declaration of Helsinki (2013 revision), and all patient-identifiable information was anonymized prior to publication. Subsequently, 2-3 *ml* of whole blood was collected in a heparin sodium vial for cytogenetic investigation. As per the AGT Cytogenetics Laboratory Manual (4th edition), routine 72-*hr in vitro* culture was initiated using whole blood with RPMI-1640 complete media (Cat No: 61870-036; Gibco, USA) containing 10% fetal bovine serum (Cat No:10270-106; Gibco, USA), 1% antibiotics (A007-100ML; HiMedia, India), and phytohemagglutinin-M (Cat No: 10576-015; Gibco, USA). At the 67th *hr*, 0.1 *µg/ml* of Colcemid (Cat No:15210-040; Gibco, USA) was added to arrest the cells in the metaphase stage. Metaphase cells were harvested by hypotonic treatment with 0.075 *M* potassium chloride solution (Merck, Germany) followed by a fixation procedure using pre-chilled Carnoy's fixative (methanol: acetic acid, 3:1) (Merck, Germany) until a clear, white lymphocyte cell pellet was obtained, and slides were prepared. Trypsin-Giemsa banding (GTG-banding) was then conducted, followed by chromosomal analysis using an Olympus BX-43 microscope (Olympus Corporation, Japan). An average banding resolution of approximately 450-550 bands per haploid set was

Table 1. Hormonal profile of mosaic Turner variant

Test particulars	Case indications	Reference value	Methodology	Clinical interference
FSH	>110.0 *mIU/ml*	Males: 2.1–18.6 *mIU/ml* Females (Follicular): 1.9–10.0 *mIU/ml* Females (Mid-cycle): 3.6–20.6 *mIU/ml* Females (Luteal): 1.5–10.8 *mIU/ml*	Chemiluminescence	FSH and LH levels were elevated which indicated hypergonadotropic hypogonadism condition
LH	22.265 *mIU/ml*	Follicular: 2.20–13.50 Ovulatory: 13.0–96.0 Luteal: 0.9–11.65 Postmenopausal: 7.2–58.50	Chemiluminescence	
Prolactin (S)	6.936 *ng/ml*	>17 years: 4.75–23.1 Pregnancy: 15.83–183.24 Postmenopausal: 3.17–17.51	Chemiluminescence	No evidence of hyperprolactinemia was observed
Testosterone (by ECLIA)	0.138 *ng/ml*	> 17 years: <0.71	Chemiluminescence	Androgen deficiency was not apparent
FT3 (S)	6.022 *pmol/ml*	≥7 years: 3.9–8.4	Chemiluminescence	Elevated TSH with near-normal FT4 suggested subclinical or early overt hypothyroidism
FT4 (S)	13.874 *pmol/dL*	≥7 years: 12.49–23.0		
TSH	19.573 *µIU/ml*	≥7 years: 0.5–5.0		

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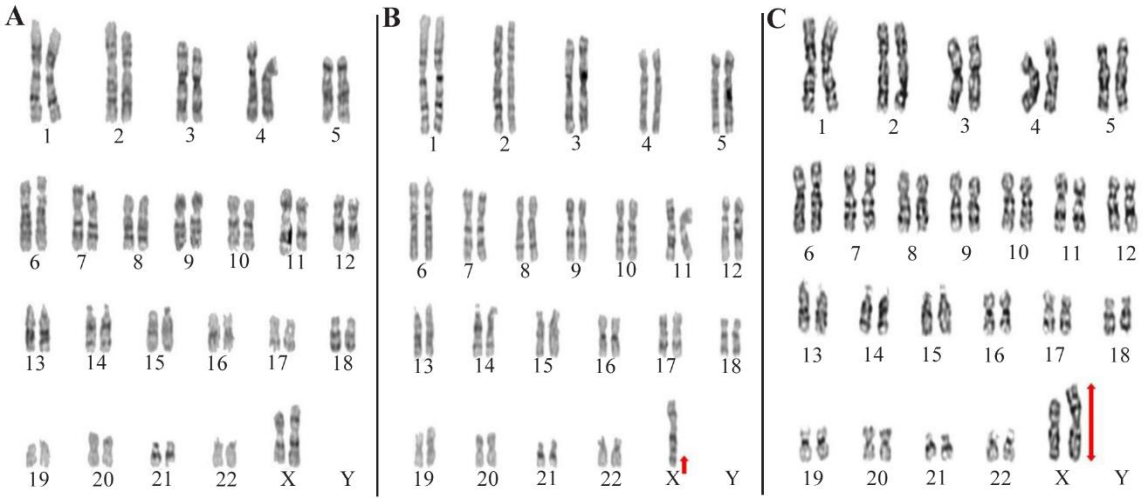


Figure 2. Triple cell-line mosaicism identified by cytogenetic karyotyping, showing (A) normal chromosome complement, (B) monosomy X, and (C) isochromosome X

achieved, ensuring accurate identification of numerical and structural alterations, including isochromosome X. Initially, 40 metaphase spreads were captured and analyzed to evaluate mosaicism. Examination of 40 metaphase spreads showed three different cell lines. Monosomy X (45,X) (Figure 2B) was observed in 30 metaphases, and iso-X (Figure 2C) was observed in 7 metaphases, while the remaining cells demonstrated a normal chromosome complement (46,XX) (Figure 2A). An additional 10 metaphases were examined to improve the precision of clone frequency estimation and to rule out the suspicion of potential mosaicism. According to ISCN 2024 guidelines, the karyotype was defined as mos 45,X[30]/46,X,i(X)(q10)[7]/46,XX[3] (Figure 2).

Molecular cytogenetic analysis (Fluorescence in situ hybridization): To further characterize and reconfirm the percentage of X chromosome mosaicism detected by conventional cytogenetic investigation, fluorescence in situ hybridization (FISH) was performed using the XA X/Y Aneusomy Probe (Cat No: D-5608-100-OG; MetaSystems Probes GmbH, Germany).The commercial probe set contains both the DXZ1 alpha-satellite X chromosome probe targeting the X chromosome centromeric region (Xp11.1-q11.1) which was directly labelled with Spectrum Green, and DYZ3 alpha-satellite Y chromosome probe targeting the Y chromosome centromeric region (Yp11.1-q11.1) which was directly labelled with Spectrum Orange. Hybridization was carried out based on the manufacturer's instruction. Briefly, slides were pre-treated, co-denatured with the probe at 72-75°*C* for 1 *min*, followed by overnight hybridization at 37°C in a humidified chamber. After post-hybridization stringency washes, nuclei were counterstained with 4',6-diamidino-2-phenylindole (DAPI) and analysis was performed on Olympus BX-43 fluorescence microscope (Olympus Corporation, Japan) equipped with appropriate filter sets and Image-Pro Express version 6.3 image analysis software.

A total of 500 non-overlapping interphase nuclei containing intact nuclear morphology and well-defined hybridization signals with 95% hybridization efficiency were scored. Nuclei demonstrating overlap, truncation, excessive background, weak or diffused signals, or ambiguous hybridization patterns were excluded.

Based on laboratory validation, the cutoff values for female control specimens were established at 5.7% for nuclei exhibiting a single DXZ1 signal and 4.5% for nuclei exhibiting three DXZ1 signals. In the current case, 357 of 500 interphase nuclei (71.4%) showed a single DXZ1 centromeric signal, consistent with monosomy X, whereas 143 nuclei (28.6%) showed two DXZ1 centromeric signals, substantially exceeding the cutoff, thereby confirming the presence of monosomy X mosaicism. The DYZ3 Y-centromeric probe was simultaneously evaluated in all 500 nuclei. No DYZ3 signals were detected in any nucleus, indicating the absence of cytogenetically detectable Y-chromosomal material in the peripheral blood sample. Because the DXZ1 probe detects X chro-

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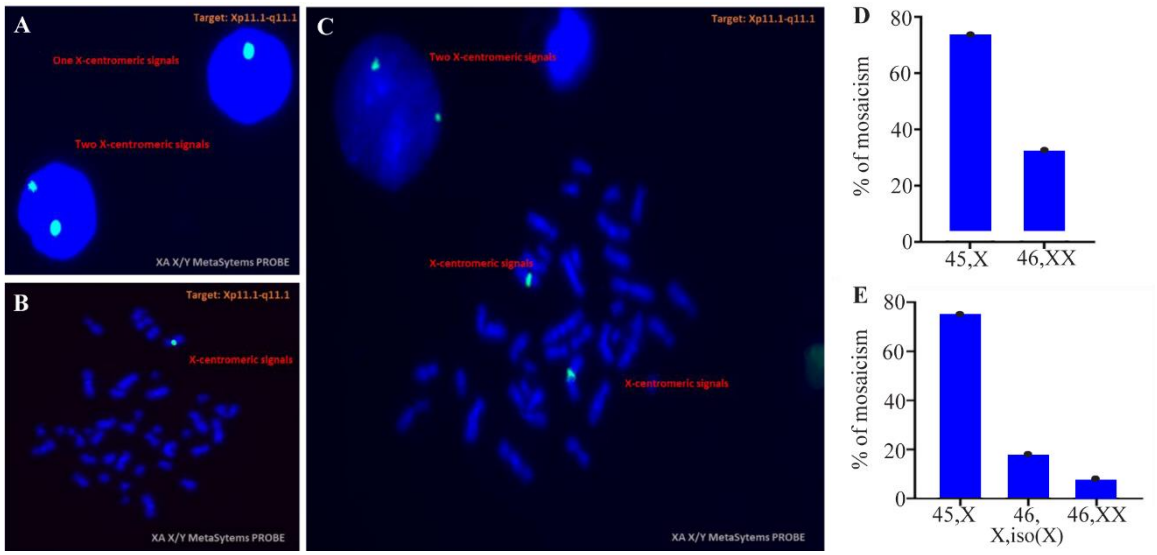


Figure 3. FISH and conventional cytogenetic analysis demonstrating mosaic X chromosome complement. A) Interphase FISH showing a single X-centromeric signal and two X-centromeric signals, B) metaphase spread demonstrating a single X-centromeric signal, C) representative interphase and metaphase cells with two X-centromeric signals, D) graphical representation of mosaic distribution percentage determined by FISH analysis, and E) graphical representation of mosaic distribution percentage determined by conventional karyotyping

mosome centromeres rather than assessing the structural integrity of the X chromosome, nuclei displaying two DXZ1 signals may indicate either normal 46,XX cells or 46,X,i(X)(q10) cell populations, both of which include two X centromeres. Consequently, the DXZ1 assay was used to confirm the presence and estimate the overall frequency of X chromosome mosaicism but could not independently distinguish or quantify the relative frequencies of the normal 46,XX cells or 46,X,i(X)(q10) cell populations. Identification and quantification of the three cytogenetically distinct clones therefore relied on conventional G-banded karyotype analysis. As per the ISCN 2024 guidelines, the FISH results were designated as nuc ish (DXZ1×1,DXZ1×2)[357/143] (Figure 3). A comprehensive summary of the clinical, endocrine, radiological, conventional cytogenetic, and molecular cytogenetic findings are presented in table 2.

Discussion

Turner syndrome is defined by significant cytogenetic and phenotypic variability due to complete or partial monosomy X, structural anomalies of the X chromosome, or numerous forms of mosaicism. Mosaic TS results from postzygotic mitotic nondisjunction in early embryogenesis, leading to the presence of two or more genetically different cell populations. Among the structural X chromosomal abnormalities linked to TS, isochromosome Xq [i(X)(q10)] is one of the most prevalent rearrangements and is thought to arise from aberrant centromeric misdivision or U-type exchange. This rearrangement causes duplication of the long arm (Xq) and deletion of the short arm (Xp), resulting in modified X chromosome gene dosage. Haploinsufficiency of genes on Xp, especially those related to growth and ovarian development, leads to short stature, gonadal dysgenesis, and primary ovarian insufficiency, while duplication of Xq may additionally affect endocrine function and phenotypic variability due to altered dosage of Xq-linked genes (9-13).

The current report describes an exceptionally rare case of triple mosaic TS with the karyotype mos 45,X[30]/46,X,i(X)(q10)[7]/46,XX[3], ascertained using standard G-banded karyotyping. Triple mosaic constitutions including these three cytogenetically different cell populations have been infrequently documented, thereby constraining the existing comprehension of their biological function, genotype–phenotype correlations, and long-term clinical outcomes.

Gollop et al. documented a prenatally diagnosed baby with the identical triple mosaic composition, characterized by a majority of the 46,XX cell line, which correlated with an apparently normal postnatal phenotype (10). Conversely, the current patient exhibited a predominance of the 45,X cell

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Table 2. Summary of the clinical, laboratory, radiological, cytogenetic, and molecular cytogenetic findings in the patient

Category	Parameter	Finding	Clinical interpretation
Clinical	Age	17 years	Adolescent female
	Presenting complaint	Primary amenorrhea	Delayed sexual maturation
	Height	136 *cm*	Short stature
	Weight	30.8 *kg*	Low body weight
	Secondary sexual characteristics	Tanner stage II breast, scanty pubic hair	Delayed pubertal development
	Dysmorphic features	Heart-shaped face, low-set ears, high-arched palate, cubitus valgus	Turner-associated phenotype
Pelvic imaging	Uterus	Non-visualized	Mullerian anomaly/ gonadal dysgenesis
	Ovaries	Bilateral streak (0.2 cm^3)	Gonadal dysgenesis
Hormonal profile	FSH	>1100 *mIU/ml*	Hypergonadotropic hypogonadism
	LH	22.265 *mIU/ml*	Elevated
	THS	19.573 *µIU/ml*	Hypothyroidism
	FT4	13.874 *pmol/dL*	Within reference range
	FT3	6.022 *pmol/dL*	Within reference range
	Prolactin	6.936 *ng/ml*	Normal
	Testosterone	0.138 *ng/ml*	Normal female range
Conventional cytogenetics	Chromosome analysis	mos 45,X[30]/46,X,i(X)(q10)[7]/46,XX[3]	Rare triple mosaic Turner syndrome
Molecular cytogenetics	FISH analysis	nuc ish (DXZ1×1,DXZ1×2)[357/143]	Supports X chromosome mosaicism
Final findings	Integrated diagnosis	Triple mosaic Turner syndrome with isochromosome Xq	Established by combined karyotyping and FISH

population in peripheral blood lymphocytes and displayed classic manifestations of TS, including short stature, primary amenorrhea, delayed pubertal development, hypergonadotropic hypogonadism, bilateral streak gonads, and primary hypothyroidism. These divergent data indicate that phenotypic severity may be affected by the relative proportions of individual mosaic cell types. Nonetheless, due to the variability of mosaicism among tissues, genotype–phenotype associations must be approached with caution.

The presence of mos 45,X[30]/46,X,i(X)(q10)[7]/46,XX[3] cell types provides additional insight into genotype–phenotype correlations in mosaic TS. The preponderance of the 45,X clone correlated with the typical Turner phenotype, while the minor presence of normal 46,XX cells likely contributed modestly to phenotypic variability. Similarly, the 46,X,i(X)(q10) clone may have affected the endocrine phenotype due to modified X chromosome gene dosage caused by the deletion of Xp and the duplication of Xq. Nonetheless, because of the infrequency of this cytogenetic configuration and the lack of functional genomic investigations, these findings should be considered biologically plausible interpretations rather than conclusive determinations (11-14).

From a diagnostic perspective, the present case highlights the synergistic functions between conventional cytogenetics and FISH from a diagnostic standpoint. Conventional G-banded karyotyping precisely detected and quantified the three cytogenetically different cell types, thereby confirming the definite diagnosis. The DXZ1 centromeric FISH technique confirmed the presence of X chromosomal mosaicism by revealing the ratio of nuclei displaying one versus two X-centromeric signals. The DXZ1 probe exclusively targets the X chromosome centromere, rendering it incapable of differentiating between 46,XX and 46,X,i(X)(q10) cell populations, both of which possess two X centromeres. Consequently, FISH cannot independently determine the relative frequencies of all three mosaic cell lines and should

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be regarded as a supplementary approach rather than a replacement for conventional karyotype analysis.

An important limitation of the current study is that mosaicism was evaluated just in peripheral blood cells, which is another significant aspect. Tissue-specific mosaicism is well-documented in TS, with the distribution of cytogenetic clones in the gonads, skin, buccal mucosa, or other tissues potentially differing significantly from that found in peripheral blood. Thus, the proportions found in this investigation may not accurately represent mosaic distribution in clinically significant tissues, and genotype–phenotype relationships should be evaluated with due caution (15).

The evaluation of Y-chromosomal material is clinically significant in TS due to the association of occult Y chromosome sequences with a higher incidence of gonadoblastoma (16). The current investigation utilized the DYZ3 Y-centromeric probe, revealing no discernible Y-centromeric signals in any of the 500 interphase nuclei analyzed, hence showing the lack of cytogenetically identifiable Y chromosomal material in peripheral blood. While these findings significantly diminish the probability of detecting Y-chromosomal mosaicism by centromeric FISH, it is not possible to entirely rule out the presence of cryptic Y-derived sequences that fall below the assay's analytical resolution.

A significant observation was the non-visualization of the uterus on transabdominal ultrasonography. Given that full uterine agenesis is rare in TS, this finding is more likely indicative of significant uterine hypoplasia resulting from prolonged estrogen deprivation linked to ovarian insufficiency, rather than genuine Müllerian agenesis. Correlation with pelvic magnetic resonance imaging would have yielded a more definitive anatomical characterization.

The current case underscores the necessity of combining comprehensive clinical examination with endocrine evaluation, pelvic imaging, conventional cytogenetics, and molecular cytogenetic methods for diagnosing atypical TS. This comprehensive approach enables individualized endocrine management, ongoing monitoring, reproductive guidance, and suitable genetic counseling for impacted individuals and their families.

Conclusion

The current case expands the literature describing the exceptionally rare triple mosaic TS karyotype, 45,X[30]/46,X,i(X)(q10)[7]/46,XX[3], and highlights its diagnostic significance in the evaluation of PA. The findings suggest that the predominance of the 45,X cell population may have contributed to the classical TS phenotype; however, because mosaicism was evaluated only in peripheral blood lymphocytes, genotype-phenotype correlation should be interpreted with appropriate caution. This report highlights the pivotal role of conventional G-banded karyotyping in accurately identifying and characterizing complex mosaic TS, while FISH analysis serves as a complementary technique by demonstrating the proportion of nuclei exhibiting one versus two X-centromeric signals. Because the DXZ1 centromeric probe cannot distinguish between 46,XX and 46,X,i(X)(q10) cell populations, FISH cannot independently determine the relative frequencies of all three mosaic clones and should therefore be interpreted in conjugation with conventional cytogenetic analysis. In addition, the current case underscores the importance of comprehensive clinical, endocrine, radiological, conventional cytogenetic, and molecular cytogenetic evaluation for precise diagnosis, optimized clinical management, informed genetic counseling, and appropriate long-term follow-up in patients with atypical TS. Documentation of such uncommon cytogenetic variants further expands the understanding of TS heterogeneity and contributes to improved diagnostic and prognostic frameworks.

Conflict of Interest

The authors declare no conflicts of interest.

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