Case Report

Primary Amenorrhea and Delayed Puberty Secondary to Isochromosome Mosaic Turner Syndrome: A Case Report

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Abstract

Turner Syndrome is a chromosome disorder that affects women and results from the loss or partial loss of one of the X chromosomes. Isochromatidosis mosaic Turner syndrome is a rare variant that tends to manifest with milder or unusual features, so that problems may go undetected. Our case is that of a 17-year-old girl whose short stature, delayed puberty, and primary amenorrhea do not have the classical features of Turner. Assessment found a severe short stature, prepubertal sexual development, hypergonadotropic hypogonadism, delayed bone age, an infantile uterus with absent ovaries, and a mosaic karyotype 45, X/46, X, i(X)q10), which confirmed isochromosome mosaic Turner syndrome. The patient has been put on oestrogen therapy and is being put on follow-up. This case demonstrates the importance of cytogenetic testing in short, statistically immature children with short stature to identify and properly treat them.

Keywords: Turner syndrome; Isochromosome; Mosaics; Primary amenorrhea; Hypergonadotropic hypogonadism; Delayed puberty.

Received: 17 January 2026 **Revised: 02 February 2026** **Accepted: 19 February 2026** **Published: 27 February 2026**

Introduction

Turner syndrome (TS) is a common chromosomal disorder that affects phenotypic females having a total or partial absence of one X chromosome, with the prevalence estimated at 1 per 2,0002,500 of the births of females that survive.[1,2] The traditional karyotype is monosomy X (45, X), which accounts for almost 50 percent of cases. The rest are mosaic forms and structural aberrations of the X chromosome, including isochromosomes, diephrism chromosomes, or deletions.[3] Isochromosome mosaic Turner Syndrome (IMTS), the most frequent cases of which are duplication of the long arm of the X chromosome [i(Xq)], is an uncommon variant, with an incidence of approximately 818 out of 100,000 cases of TS.[4-6] IMTS can have milder or non-typical clinical Features and can have no classic stigmata of Turner syndrome, due to mosaicism and partial preservation of genetic material, and this results in delayed diagnosis.[7]

A common executive complaint in TS is short stature, delayed puberty, and primary amenorrhea, which is due to gonadal dysgenesis and oestrogen deficiency and manifests as hypergonadotropic hypogonadism.[1,8] We present a case of a 17-year-old girl who has been complaining of short stature, delayed puberty, and primary amenorrhea, and later was diagnosed with isochromosome mosaic Turner syndrome.

Case Report

A 17-year-old girl was seen at the outpatient department complaining of short stature, retarded puberty, and amenorrhea. The client did not have any history of chronic conditions, developmental delay, or consanguinity. The family history was not remarkable, nor was the short stature of parents or siblings.

Vital signs were normal on general examination. Anthropometric measurements showed the height to be below 1.5 standard deviations for the age group, consistent with severe short stature. General physical examination revealed polydactyly of the left foot. Neck, shield chest, cubitus valgus, no classical Turner stigmata were present. The normal examination was normal systemically.

The Sexual maturity rating (SMR) indicated the development of breast and pubic hair (Tanner stage I). Neurological examination and intelligence quotient (IQ) analysis were normal.

Investigations

The patient was checked for short stature and primary amenorrhea. Evaluation of bone age showed significant bone underdevelopment relative to age. Hormonal analysis was done and showed that there was hypergonadotropic hypogonadism with an elevated level of Follicle-stimulating hormone (FSH),

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DOI:
10.21276/amit.2026.v13.i1.389

How to cite this article: Gowadia N, Bhattacharjee A, Bhattacharjee P, Parashar AK. Primary Amenorrhea and Delayed Puberty Secondary to Isochromosome Mosaic Turner Syndrome: A Case Report. Acta Med Int. 2026;13(1):551-552.

and an extremely low level of serum oestrogen. Tests of thyroid functioning were stereotypical.

An abdominal and pelvic magnetic resonance imaging showed that the infant had an infantile uterus, non-visualised ovaries, and a normal vaginal canal. MRI of the brain and echocardiography showed normal findings, and there was no sign of related congenital abnormalities.

PBL cytoplasmic analysis showed the presence of two cell lines, namely 45, X [20] and 46, X, i(X) [30], which confirmed the diagnosis of isochromosome mosaic Turner syndrome.

Management and Outcome

As a result of economic issues, it was not possible to start the growth hormone therapy. The patient has received oestrogen replacement therapy to induce puberty and treat the effects of hypogonadism. She has already been placed on routine follow-up for pubertal development and growth and has also been screened for related comorbidities.

Discussion

Isochromosome mosaic TS is another type of TS that has a rare manifestation, where half of the X chromosome (usually the long arm) is duplicated (Xq) with the short arm (Xp) deleted, which lacks genes essential to normal development and ovarian function.[3,6]

MTS is usually milder and phenotypically abnormal than classic 45, X Turner syndrome, leading to late diagnosis.[4,7] As was previously reported on this and similar cases in earlier literature, our patient debuted short stature, late puberty, and primary amenorrhea devoid of characteristically typical phenotypic features of TS.[5,9,11]

Hypergonadotropic hypogonadotrophic with delayed bone age and non-existent ovaries has been noted as a regular occurrence in IMTS as reported in several case series.[4,9,10] It is important to begin growth hormone and oestrogen therapy early, and an early start leads to better height gains, better pubertal progression, healthier bones, and a better quality of life.[1,8]

The presented case demonstrates the significance of karyotype tests in cases of short stature and primary amenorrhea in adolescent girls, even when no typical signs of Turner syndrome are present, to prevent cases of either missed diagnosis or delayed diagnosis.

Conclusion

The clinical presentation of Isochromosome mosaic Turner syndrome is mild and does not exhibit the typical phenotypic features of Turner syndrome. Turner syndrome should thus be considered as the cause of short stature, late puberty, and primary amenorrhea in any female teenager, to diagnose and initiate proper treatment as soon as possible.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Sharma L, Kikkeri NS. Turner syndrome. StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Nov 10.
2. Martin-Giacalone BA, Lin AE, Rasmussen SA, et al. Prevalence and descriptive epidemiology of Turner syndrome in the United States, 2000–2017. Am J Med Genet A. 2023;191(6):1545-1556. doi:10.1002/ajmg.a.63181.
3. Ferdousi T, Banu H, Sultana N, Mahrukh H, Hasanat MA. Turner syndrome with isochromosome structural abnormalities: a case report. Cureus. 2023;15(7):e40516. doi:10.7759/cureus.40516.
4. Llanes MRV, Uyking-Naranjo M. Isochromosome mosaic Turner syndrome: a case report. J ASEAN Fed Endocr Soc. 2019;34(2):203-207. doi:10.15605/jafes.034.02.15.
5. Tamrakar R, Rai A, Maskey R. Isochromosome mosaic Turner syndrome with delayed growth and puberty: a case report. J Diabetes EndocrinolAssoc Nepal. 2023;7(1):45-49.
6. Gravholt CH, Andersen NH, Conway GS, et al. Clinical practice guidelines for the care of girls and women with Turner syndrome. Eur J Endocrinol. 2017;177(3):G1-G70.
7. Sybert VP, McCauley E. Turner's syndrome. N Engl J Med. 2004;351(12):1227-1238.
8. Bondy CA. Turner syndrome study group. Care of girls and women with Turner syndrome: a guideline of the Turner Syndrome Study Group. J ClinEndocrinolMetab. 2007;92(1):10-25.
9. Akasaka M, Kamei A, Ito J, Oyama K. Turner syndrome associated with refractory seizures and intellectual disability: a case study. Cureus. 2020;12(12):e12364.
10. Tamrakar R, Rai A, Maskey R. Isochromosome mosaic Turner syndrome presenting with hypergonadotropic hypogonadism. J Diabetes EndocrinolAssoc Nepal. 2023;7(1):45-49.
11. Llanes MRV, Uyking-Naranjo M. Atypical presentations of mosaic Turner syndrome. J ASEAN Fed Endocr Soc. 2019;34(2):203-207.