Case Report

Journal Homepage: http://crcp.tums.ac.ir

Beyond Monosomy: A Mosaic Turner Syndrome Presenting with Coarctation of the Aorta with Left Persistent Superior Vena Cava and Primary Ovarian Failure

Shivani Shukla*, Nancy Garg, Apoorva Suran, Jaideep Khare

Department of Endocrinology, People's College of Medical Sciences, Bhopal, India.

Use your device to scan and read the article online

Citation Shukla Sh, Garg N, Suran A, Khare J. Beyond Monosomy: A Mosaic Turner Syndrome Presenting with Coarctation of the Aorta with Left Persistent Superior Vena Cava and Primary Ovarian Failure. Case Reports in Clinical Practice. 2025; 10(6): 280-284. DOI:10.18502/crcp.v10i6.21615

Running Title Mosaic Turner Syndrome presenting with Coarctation of the Aorta

Article info:
Received: November 13, 2025
Revised: November 25, 2025
Accepted: December 17, 2025

Keywords:
Mosaic turner syndrome; Coarctation of the aorta; Primary amenorrhea; Growth hormone; Puberty induction

ABSTRACT

Turner syndrome is a rare but not uncommon genetic syndrome due to partial or complete loss of X-chromosome in females. Various chromosome anomalies identified are 45X0, isochromosome Xq, ring X, deletion Xp, or an abnormal Y chromosome, most common being 45X0.Clinical features depends on the type of chromosome anomaly present while generally include short stature, primary amenorrhea, hypogonadism.
Hereby, we report a case of 18 year old female with Mosaic turner syndrome who presented with clinical finding of short stature with primary amenorrhea with coarctation of aorta.
Patient was managed with multi-disciplinary approach that include growth hormone replacement, puberty induction and planning for ductal stenting.

Introduction

Turner syndrome (TS) is a genetic syndrome with a female phenotype consisting of sex-chromosome abnormalities, with monosomy 45,X being the most common, and other variants such as mosaic 45,X/46,XX; 45,X/46,XY; 45,X/47,XXX, etc., that may lead to significant multisystem morbidity in the future [1]. Classically, it presents as short stature with delayed puberty or amenorrhea, and left-sided outflow congenital heart defects. Turner›s stigmata include down-slanted palpebral fissures, epicanthal folds, low-set ears, short broad neck, micrognathia, multiple nevi, and Madelung deformity. Other clinical features include renal anomalies such as horseshoe kidney, renal aplasia, or renal hypoplasia, and neuropsychologic problems.

Turner syndrome is also associated with various congenital heart diseases such as coarctation of the aorta, aortic stenosis, mitral valve anomalies, and left heart syndrome, which further is a major determinant of mortality and warrants screening

*** Corresponding Author:**
Shivani Shukla
Address: *Department of Endocrinology, People's College of Medical Sciences, Bhopal, India.*
E-mail: *drshivani27894@gmail.com*

with 2D echocardiography and CT angiography [1,2]. While individuals with mosaic Turner (45,X/46,XX) have a milder phenotype, with left-sided congenital heart defects, obesity, and hypertension being less common, and age at menarche being near normal with spontaneous pregnancy, unlike our patient who has primary amenorrhoea [3,4]. Thus, timely diagnosis and early intervention are important for better prognosis.

Here we report an interesting case of Turner syndrome with mosaic genotype (45,X/46,XX) (Figure 1a) with normal IQ, with a hypoplastic uterus with streak gonads, and with coarctation of the aorta.

Case Presentation

An 18-year-old female presented with the chief complaint of failure to gain height for the past 4 years. She was apparently well until 4 years ago, when she noticed that other students had overtaken her in height and she was the shortest in her class. On enquiry, she also gave a history of primary amenorrhea, scanty axillary and pubic hair, and absent breast budding. The patient has a normal IQ and attention span.

There is no history of weight gain, neck swelling, diminished sweating, paresthesia, or constipation. There is no history of bone pain or bone deformity. No history of headache, vomiting, visual complaints, polyuria, polydipsia, or renal stones. No history of perioral numbness or CNS infection. No history of consanguinity in parental marriage.

Birth history: Normal delivery at home; no significant h/o of perinatal complications. However, patient gives history of recurrent lower respiratory tract infection. No significant family and personal history of short stature.

On general physical examination, there were no signs of malnutrition or micronutrient deficiency. IQ and attention span were normal. Dental examination was normal, and there was no evidence of a palatal anomaly. She had a short neck with cubitus valgus and a short right-hand metacarpal. There was no pallor, icterus, cyanosis, clubbing, goitre, pedal oedema, or kyphoscoliosis. Pulse was 70/min in the right hand and feeble in the left; all peripheral pulses were feeble. Blood pressure (supine) was 160/90 mmHg in the right arm and 112/60 mmHg in the left and lower limb was not recordable. Patient's height was 126.1cm(below 3rd centile), with height age of 8 years 2 months while bone age was 11 years (Figure 1b). Sexual maturity rating (Tanner) was B1P2. Detail anthropometric findings are described in Table 1.

Cardiovascular examination: on inspection, suprasternal pulsations were present; apex beat at the 4th intercostal space; S1 and S2 normal; an interscapular murmur was present. All other systems were normal. Cardiovascular findings were confirmed in radiological investigations which was suggestive of coarctation of aorta (Figure 1e & 1f). MRI pelvis

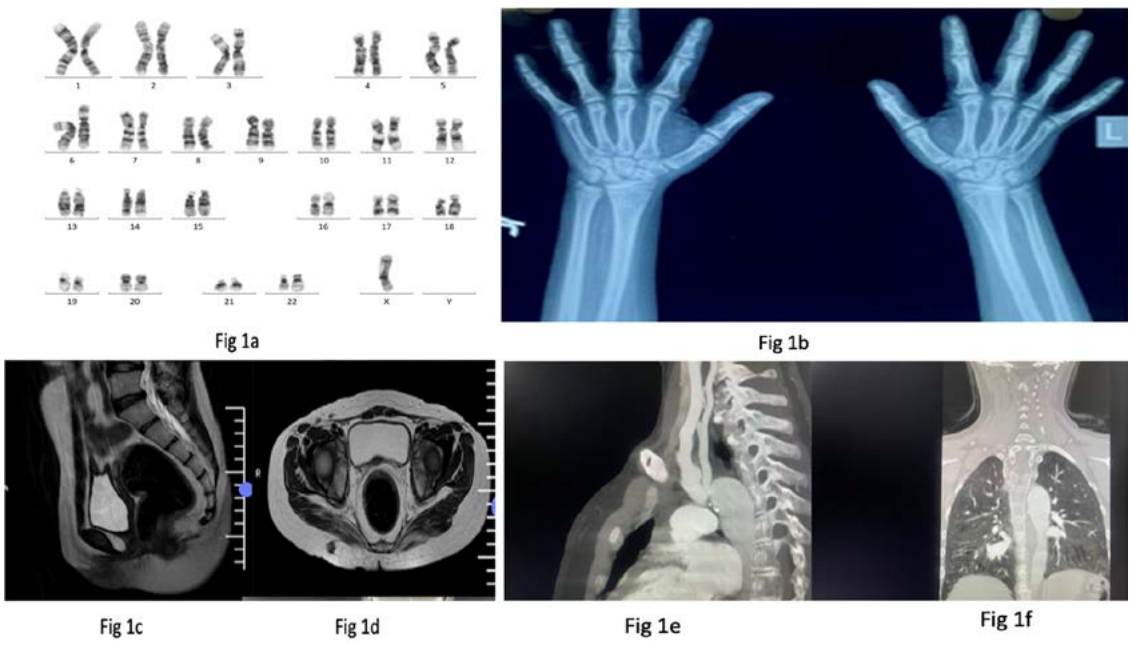

Fig. 1. 1a: It shows the karyotyping of the patient suggesting mosaic pattern 46XX:X0
1b: X ray left hand of the patient showing 12 years of bone age and non-closure of epiphysis of the wrist of left hand. 1c and 1d: showing MRI pelvis with contrast suggestive of hypoplastic uterus. 1e and 1f: showing CT angiogram with contrast showing pre ductal coarctation of aorta.

Table 1. Anthropometric Data of patient

Parameter	Value
Age / Sex	18 years / Female
Height	126.1 cm (Below 3rd centile)
Upper Segment (US)	66.1 cm
Lower Segment (LS)	60 cm
US : LS Ratio	1.1 (High ratio for age)
Arm Span	125 cm
Weight	26.9 kg (Below 3rd centile)
BMI	16.9 kg/m²
Height-for-Age	8 years 2 months
Weight-for-Age	9 years 2 months
Mid-Parental Height (range)	140.5–157.5 cm
Expected Target Height	149 cm
Target Weight	32 kg
Neck length	8 cm
Birds index	15.7

Table 2. Biochemical and Radiological Investigation of patient

Test	Patient value with normal reference range
Cortisol	15.50 µg/dL (5–23 µg/dL)
Free thyroxine (FT4)	21.84 pmol/L (12.0–21.9 pmol/L)
Prolactin	25.31 ng/mL (<25 ng/mL)
FSH	108 mIU/mL (4–30 mIU/mL premenopause; 40–250 mIU/mL postmenopausal/ovarian failure)
Estradiol	5.3 pg/mL (30–100 pg/mL follicular; <15 pg/mL postmenopausal)
IGF-1	143 ng/mL (114–492 ng/mL adult female 25–39)
Serum calcium	9.6 mg/dL (8.4–10.2 mg/dL)
Phosphorus	13.17 mg/dL (3.0–4.5 mg/dL)
iPTH	11 pg/mL (10–60 pg/mL)
MRI pelvis	Hypoplastic uterus and small ovaries. Uterus 6 × 2 × 3 cm; left ovary 14 × 8 mm; right ovary 10 × 7 mm
2D Echocardiography	Concentric LVH; normal valves and wall motion; preserved EF. No MR or TR; no RWMA at rest; LVEF 60%
CT neck & thoracic angiography	Preductal coarctation with post-stenotic dilatation. Left subclavian from stenosed arch; mild narrowing vs contralateral; left ECA→subclavian collaterals
DEXA scan	Low bone density (Z-scores). Left femur −3.9; right femur −3.7; spine −4.5.
Karyotyping	mos 45,X/46,XX. Diagnosis: abnormal female karyotype consistent with mosaic variant pattern of turner syndrome

suggestive of hypoplastic uterus (Figure 1c & 1d). Details of radiological and cardiovascular finding are described in Table 2.

A multidisciplinary approach was taken for the patient. She was advised therapeutic lifestyle modification. As advised by the cardiologist for management of the hypertension, the patient was started on an angiotensin-converting enzyme inhibitor (telmisartan 20 mg). For endocrine issues, the patient was started on recombinant growth hormone subcutaneously daily in the evening for short stature and on estradiol valerate 0.5 mcg for pubertal induction.

For bone health, nutritional supplementation such as calcium and vitamin D was started.

After multidisciplinary planning, definitive repair of pre-ductal coarctation (endovascular stenting) with the cardiologist is planned.

The prognosis of the natural history of TS was explained to the patient and attendant, which included the risk of metabolic syndrome, bone health, cardiovascular disease, and pregnancy outcomes.

Discussion

Our patient with mosaic Turner syndrome (TS), 45,X/46,XX, presented with the classic Turner's triad consisting of gonadal dysgenesis presenting as primary amenorrhea, short stature with classic somatic stigmata, and a major cardiovascular lesion: pre-ductal coarctation of the aorta (CoA).

Mosaic 45,X/46,XX show greater likelihood of spontaneous pubertal progression and potential for menses/fertility than 45,X; nevertheless, some still require puberty induction owing to primary ovarian insufficiency, as in our patient (FSH 108, very low estradiol) [5–7]. Turner Guidelines were followed

and physiologic 17β-estradiol at very low dose was started with plan for gradual escalation and followed by cyclic progestogen after adequate estrogenization, to establish secondary sex characteristics, support uterine growth and optimize metabolic and skeletal health [1]. Patient was also explained that future pregnancy must be approached cautiously as pregnancy confers elevated aortic risk [2,5-7,11].

In our patient, since her height was below the 3rd centile on the growth chart, guidelines recommend offering growth hormone treatment early because growth failure in TS starts before birth and progresses rapidly early in life. GH treatment can be offered as long as the epiphyses remain open. It is recommended to start GH at a dose of 45–50 µg/kg. [1]

Skeletal fragility is well recognized in TS. While mosaics may, on average, exhibit higher BMD than 45,X, clinically meaningful bone deficits remain common due to hypogonadism, smaller bone size, and microarchitectural alterations [8-10]. Our patient also has low Z-scores (spine −4.5; femoral sites −3.8), which warranted initiating management with sex-steroid replacement, calcium/vitamin D, and follow-up with DXA scanning after 1 year of treatment [1,8-10].

Although the prevalence of CHD is higher in monosomy 45,X than in mosaic karyotypes, CoA has been reported in mosaics [1-4]. TS guidelines suggest that cardiovascular disease is the principal cause of morbidity and mortality across the TS spectrum, and recommend aortic imaging using echocardiography and MRI/CT angiography for arch disease and aortopathy surveillance [1,2]. For hypertension, guidelines recommend that it is essential to rule out secondary causes of hypertension such as renal anomalies, obstructive uropathy, or coarctation, and to use a beta-blocker or angiotensin receptor blocker in patients who have a dilated aorta with or without hypertension. If the patient has hypertension without any secondary cause or dilated aorta, they are treated according to the hypertensive guidelines for normal adults [1,12].

Normally, coarctation of aorta is of two type, i.e preductal and post ductal type. Preductal coarctation of aorta is generally severe and presents early in life [13]. In our patient, she was of adolescence group and still was asymptomatic. Also, patient had left persistent left superior vena cava which is more common in classic type of turner's syndrome. Also 80% drain into right atrium and only 20 % into left atrium. Our patient stands unique as she is mosaic turner with left persistent superior vena cava draining into left atrium and patient is still asymptomatic [14].

For treatment of coarctation of the aorta, if it presents in infancy (more than 4 months and weight less than 25 kg), balloon angioplasty and surgical repair are equally good; after 24 months or if the patient weighs more than 25 kg, then stenting is preferred [15].

Conclusion

Turner syndrome is a rare but not uncommon genetic syndrome that requires a multidisciplinary approach. Our patient was initiated on recombinant growth hormone and estradiol for puberty induction with future progestogen, bone health restoration with vitamin D and calcium supplementation, antihypertensive therapy, and was planned for CoA repair and stenting once blood pressure is controlled.

Ethical Considerations

Compliance with ethical guidelines

There were no ethical considerations to be considered in this article.

Funding

No funding was received to assist with the preparation of this manuscript.

Conflict of Interests

The authors have no conflict of interest to declare.

References

[1] Gravholt CH, Andersen NH, Christin-Maitre S, Davis SM, Duijnhouwer A, Gawlik A, et al. Clinical practice guidelines for the care of girls and women with Turner syndrome. Eur J Endocrinol. 2024 Jun 5;190(6):G53-G151. https://doi.org/10.1093/ejendo/lvae050

[2] Silberbach M, Roos-Hesselink JW, Andersen NH, Braverman AC, Brown N, Collins RT, et al. Cardiovascular Health in Turner Syndrome: A Scientific Statement From the American Heart Association. Circ Genom Precis Med. 2018 Oct;11(10):e000048. https://doi.org/10.1161/HCG.0000000000000048

[3] Álvarez-Nava F, Lanes R, Quintero MS, Soto M, Maciel-Guerra AT, Skakkebæk A. Impact of karyotype on congenital heart disease in Turner syndrome. Am J Med Genet C Semin Med Genet. 2020;184(1):41-53. https://doi.org/10.1002/ajmg.c.32146

[4] Gorito V, Baptista C, Santos P, Costa AM, Carvalho J. Coarctation of Aorta in Turner Mosaicism. Int J Pediatr Adolesc

Med. 2021 Dec;8(4):268-70. https://doi.org/10.1016/j.ijpam.2020.03.005

[5] Doğer E, Çakıroğlu Y, Ceylan Y, Ulak E, Özdamar Ö, Çalışkan E. Reproductive and obstetric outcomes in mosaic Turner's Syndrome: a cross-sectional study and review of the literature. Reprod Biol Endocrinol. 2015 Jun 10;13:59. https://doi.org/10.1186/s12958-015-0055-7

[6] Folsom LJ, Fuqua JS. Reproductive Issues in Women with Turner Syndrome. Endocrinol Metab Clin North Am. 2015 Dec;44(4):723-37. https://doi.org/10.1016/j.ecl.2015.07.004

[7] TTuke MA, Ruth KS, Wood AR, Beaumont RN, Tyrrell J, Jones SE, et al. Mosaic Turner syndrome shows reduced penetrance in an adult population study. Genet Med. 2019 Apr;21(4):877-886. https://doi.org/10.1038/s41436-018-0271-6

[8] Sylvén L, Hagenfeldt K, Bröms J, et al. Bone mineral density in 45,X versus mosaic Turner syndrome: effects of HRT duration. Acta Paediatr. 1995;84(10):1179-83.

[9] Augoulea A, Zachou G, Lambrinoudaki I. Turner syndrome and osteoporosis. Maturitas. 2019 Dec;130:41-9. https://doi.org/10.1016/j.maturitas.2019.09.010

[10] Faienza MF, Ventura A, Colucci S, et al. Bone fragility in Turner syndrome and DXA size-artifact considerations. Int J Endocrinol. 2016;2016:6562891.

[11] American Society for Reproductive Medicine (ASRM) Practice Committee. Maternal cardiovascular morbidity and mortality in pregnancy among individuals with Turner syndrome. Fertil Steril. 2024;122(3):401-8.

[12] Turner HE, Johannsen EB, Smyth A, Orchard E, Gravholt CH. Approach to the patient with Turner syndrome. J Clin Endocrinol Metab. 2025;110(5):dga f517.

[13] Law MA, Collier SA, Sharma S. Coarctation of the aorta. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025.

[14] Azizova A, Onder O, Arslan S, Ardali S, Hazirolan T. Persistent left superior vena cava: clinical importance and differential diagnoses. Insights Imaging. 2020 Oct 15;11(1):110. https://doi.org/10.1186/s13244-020-00906-2

[15] Saxena A, Relan J, Agarwal R, Awasthy N, Azad S, Chakrabarty M, et al. Indian Guidelines for Indications and Timing of Intervention for Common Congenital Heart Diseases: Revised and Updated Consensus Statement of the Working Group on Management of Congenital Heart Diseases. Abridged Secondary Publication. Indian Pediatr. 2020 Feb 15;57(2):143-57. https://doi.org/10.1007/s13312-020-1733-x