European Journal of Clinical Pharmacy
Print ISSN: 2385-409X **Online ISSN**: 3105-0409
Website: https://farmclin.com/

Original Research Article

Clinical Spectrum of Turner Syndrome with Cytogenetic Correlation in a Tertiary Care Hospital of Bagalkote, Karnataka

Article History:

Name of Author:
Sujayendra Kulkarni[1], Naveen Charantimath[2], Anil Satyaraddi[2], Suyamindra S.K[3], Ashwini H[1], Kavitha MM[4] and Sangappa V Kashinakunti[4]

Affiliation: *[1]Department of Genetics and Genomics, S Nijalingappa Medical College and HSK Hospital, Navanagar, Bagalkote-587102*
[2]Department of Endocrinology, Diabetes and Metabolism, S Nijalingappa Medical College and HSK Hospital, Navanagar, Bagalkote-587102
[3]Kalyan Karnataka Institute for Human Genome Research, Department of Microbiology, Adikavi shri Maharshi Valmiki University, Raichur-584133
[4]Department of Biochemistry, S Nijalingappa Medical College and HSK Hospital, Navanagar, Bagalkote-587102

Corresponding Author:

Suyamindra Kulkarni

Received: 28-11-2025
Revised: 05-12-2025
Accepted: 12-01-2026
Published: 28-01-2026

Abstract: ***Background***: Turner syndrome (TS) is a common chromosomal disorder in phenotypic females caused by complete or partial absence of one X chromosome. The condition shows marked clinical heterogeneity, largely influenced by the underlying cytogenetic pattern. Regional data on the clinical spectrum and karyotype–phenotype correlation of TS from India, particularly North Karnataka, remain limited. ***Objectives***: To evaluate the clinical profile of patients with Turner syndrome and correlate phenotypic features with cytogenetic findings in a tertiary care hospital in North Karnataka, India. ***Methods***: This hospital-based observational study was conducted from January 2018 to December 2025. A total of 600 clinically suspected cases of Turner syndrome were referred for cytogenetic evaluation. Conventional G-banded karyotyping was performed in all cases, and fluorescence in situ hybridization (FISH) was used selectively to confirm mosaicism. Clinical, demographic, anthropometric, and phenotypic data were analyzed and correlated with cytogenetic patterns. Statistical analysis was performed using SPSS/R. ***Results***: Turner syndrome was cytogenetically confirmed in 30 patients (5%). Monosomy 45 X was the most common karyotype, observed in 24 cases (80%), while mosaic Turner syndrome was identified in 6 cases (20%). The majority of patients were diagnosed during adolescence, with 66.7% diagnosed between 11 and 15 years of age; none were diagnosed before 5 years. Short stature was the most consistent clinical feature (86.7%), followed by primary amenorrhea (93.3%). Other common features included shield chest (86.7%), cubitus valgus (66.7%), low posterior hairline (70%), and webbed neck (53.3%). Patients with monosomy 45X showed a higher frequency of short stature and primary amenorrhea compared to mosaic cases, although the differences were not statistically significant. ***Conclusion***: Monosomy 45X was the predominant cytogenetic abnormality among clinically diagnosed Turner syndrome patients. The study highlights delayed diagnosis and a high burden of classical phenotypic features, underscoring the need for improved early detection through growth monitoring and timely cytogenetic testing. Comprehensive cytogenetic evaluation, including FISH, is essential for accurate diagnosis and management, particularly in resource-limited settings.

Keywords: Turner syndrome (TS); Monosomy X; Mosaic Turner syndrome; Karyotype–phenotype correlation; Cytogenetics; North Karnataka.

INTRODUCTION

Turner syndrome (TS) is a frequent chromosomal disorder in phenotypic females, resulting from complete or partial loss of one X chromosome. The condition most commonly arises due to monosomy X (45X), mosaic karyotypes (such as 45X/46XX or other cell lines), or structural abnormalities of the X

chromosome, including isochromosomes, deletions, and ring chromosomes. TS exhibits a broad clinical spectrum, encompassing short stature, gonadal dysgenesis with delayed puberty or primary amenorrhea, congenital lymphedema, webbed neck, and distinctive craniofacial features. Affected individuals also have an increased risk of cardiovascular, renal, endocrine, auditory, and metabolic complications, which may lead to significant morbidity in the absence of early diagnosis and appropriate management.

Globally, Turner syndrome occurs in approximately 1 in 2,000–2,500 live-born female infants, making it one of the most frequent sex chromosome aneuploidies [1,2]. Cytogenetic studies of spontaneous abortions have shown that monosomy X accounts for nearly 10–15% of chromosomally abnormal miscarriages, and it is estimated that up to 99% of 45X conceptions result in fetal loss, explaining the discrepancy between conception and live-birth prevalence [3]. Among live-born individuals, approximately 30–50% have classic monosomy 45X, while the remaining cases exhibit mosaicism or structural abnormalities of the X chromosome [4].

In India, population-based data on the incidence of Turner syndrome are limited due to the absence of a national congenital anomaly registry and underdiagnosis, particularly in rural and resource-limited settings. Hospital-based and referral-center studies suggest an incidence broadly comparable to global figures, estimated at 1 in 2,500–3,000 live-born females [5,6]. Indian patients frequently present late, during late childhood or adolescence, most commonly with short stature or primary amenorrhea, reflecting delayed referral, limited awareness, and restricted access to cytogenetic testing. Consequently, milder phenotypes, especially mosaic forms, may remain undetected.

The phenotypic variability observed in Turner syndrome is strongly influenced by the underlying karyotype. Individuals with monosomy 45X typically exhibit more severe clinical features, including pronounced short stature, congenital lymphoedema, and a higher prevalence of congenital heart disease, particularly bicuspid aortic valve and coarctation of the aorta. In contrast, mosaic and structurally abnormal karyotypes often present with milder phenotypes and may retain partial ovarian function [7,8]. Establishing genotype–phenotype correlations is therefore crucial for prognosis, targeted screening, and individualized management.

Given the limited regional data from India, especially from Karnataka, studies documenting the clinical spectrum and cytogenetic profile of Turner syndrome are essential. Such data contribute to improved early diagnosis, better understanding of local presentation patterns, and optimized multidisciplinary care. The present study aims to analyze the clinical features of patients with Turner syndrome and correlate them with cytogenetic findings in a tertiary care hospital in North Karnataka.

MATERIALS AND METHODS

Study design and setting

This was a hospital-based observational study conducted in the Department of Genetics at S Nijalingappa Medical College and HSK Hospital Bagalkote, Karnataka, India a tertiary care teaching hospital in North Karnataka and probe deign and molecular work help obtained from Kalyan Karnataka Institute for Human Genome Research, Raichur, Karnataka, India. The study period extended from January 2018 to December 2025. Ethical clearance was obtained from the Institutional Ethics Committee, and informed consent was obtained from the patients or their legal guardians prior to enrollment.

Study population

A total of 600 clinically suspected cases of Turner syndrome were referred to the cytogenetics laboratory from Endocrinology department and Obstetrics and gynecology department during the study period for chromosomal analysis. Clinical suspicion was based on features such as short stature, delayed puberty, primary amenorrhea, webbed neck, shield chest, congenital lymphoedema, and other characteristic phenotypic features.

Out of these, 30 patients were confirmed to have Turner syndrome on cytogenetic analysis, yielding a diagnostic positivity rate of 5%. Among the confirmed cases, 6 patients (20%) demonstrated mosaic karyotypes, while the remaining cases showed non-mosaic chromosomal abnormalities.

Clinical and demographic evaluation

Detailed clinical and demographic data were collected using a structured proforma at the time of evaluation. Patients were classified according to:

- Place of residence: urban or rural
- Educational status: literate or illiterate (for adult patients or parents/guardians in pediatric cases)
- Age at diagnosis of Turner syndrome
- Growth parameters: height, weight, and body mass index (BMI), plotted against age-appropriate standard growth charts; height standard deviation score (SDS) was calculated where applicable
- Pubertal status: assessed using Tanner staging
- Phenotypic features: including short stature, webbed neck, low posterior hairline, broad shield chest, cubitus valgus, congenital lymphoedema, scoliosis, cardiac activity and

other dysmorphic features
- Relevant clinical investigations such as echocardiography, renal ultrasonography, and hormonal profiles were reviewed where available.
- Cytogenetic analysis (Karyotyping)
- Peripheral venous blood (2–3 mL) was collected in sodium heparinized vacutainers from all patients. Lymphocyte cultures were established using standard protocols. Cells were harvested after 72 hours of incubation, followed by hypotonic treatment and fixation.
- Chromosomal analysis was performed using Giemsa-Trypsin-G banding (GTG-banding) at a resolution of approximately 450–550 bands per haploid set. A minimum of 20 metaphases were analyzed per case, and in cases suspected of mosaicism, up to 50 metaphases were examined. Karyotypes were reported according to the International System for Human Cytogenomic Nomenclature (ISCN) guidelines.
- Fluorescence in situ hybridization (FISH)
- Fluorescence in situ hybridization (FISH) was performed in selected cases to:
- Confirm mosaicism
- Detect low-level mosaicism not evident on conventional karyotyping
- Identify the presence or absence of X-chromosome centromeric material or Y-chromosome sequences
- Commercially available probes specific for X-chromosome centromere (DXZ1)(Wuhan health care PVT Ltd) and, where indicated, Y-chromosome centromere (DYZ3))(Wuhan health care PVT Ltd) were used according to the manufacturer's protocol. A minimum of 100 interphase nuclei were analyzed per case. FISH analysis done by ASI software- GenASIs v8.4.1, Japan (Olympus BX53 fluorescent microscope) and results were interpreted independently and correlated with karyotype findings.
- Data analysis and karyotype-phenotype correlation
- Confirmed Turner syndrome cases were categorized based on cytogenetic findings into:
- Monosomy 45X
- Mosaic Turner syndrome
- Structural abnormalities of the X chromosome
- Clinical features, growth parameters, and age at diagnosis were correlated with karyotype patterns. Demographic factors such as rural/urban residence and literacy status were also analyzed in relation to age at presentation and disease severity.

Statistical analysis

Data were entered into Microsoft Excel and analyzed using statistical software (SPSS/R). Categorical variables were summarized as frequencies and percentages, while continuous variables were presented as mean ± standard deviation or median with range, as appropriate. Comparisons of categorical variables were performed using the chi-square test or Fisher's exact test, and continuous variables were analyzed using the independent t-test or Mann–Whitney U test. A p-value < 0.05 was considered statistically significant.

RESULTS:

The demographic characteristics of patients clinically suspected and cytogenetically confirmed to have Turner syndrome. Among the 600 suspected cases, the age at referral ranged from 6 to 22 years. The 30 confirmed Turner syndrome patients were also referred within the same age range, while the age at diagnosis ranged from 12 to 20 years.

With respect to residence, 450 (75.5%) of the suspected cases were from urban areas and 150 (25%) were from rural areas. Among confirmed cases, 18 (60%) belonged to urban areas and 12 (40%) to rural areas.

Based on educational status, 550 (91.6%) of the suspected cases were literate and 50 (8.3%) were illiterate. Among confirmed Turner syndrome patients, 30 (100%) were literate and 00 (00%) were illiterate. (Table-1)

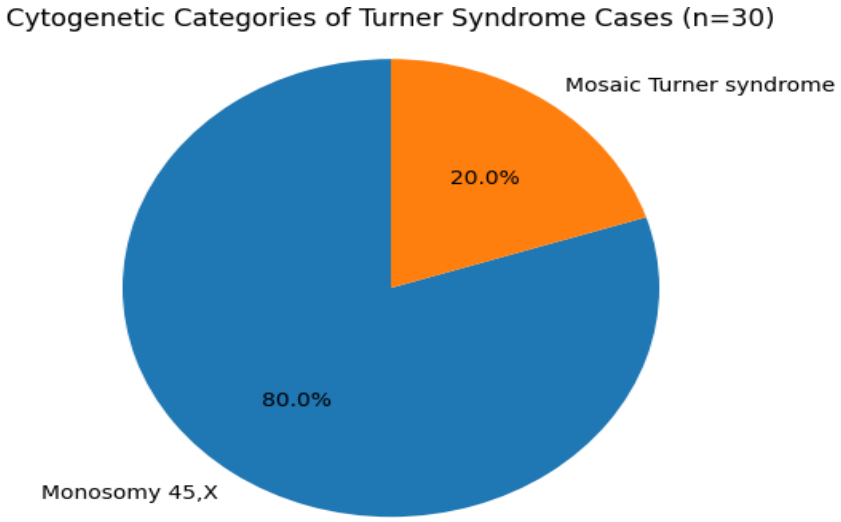
Table 1. Demographic profile of patients with suspected and confirmed Turner syndrome (2018–2025)

Variable	Suspected TS (n=600)	Confirmed TS (n = 30)
|Mean age at referral (years)	6-22	6-22
Age at diagnosis (years), mean ± SD	--	12-20
Residence		

• Urban	450 (75.5%)	18 (60%)
• Rural	150 (25%)	12 (40%)
Educational status*		
• Literate	550 (91.6%)	30 (100%)
• Illiterate	50 (08.3%)	00 (00%)

*According to the Census of India, a literate person is defined as someone aged 7 or above who can both read and write with understanding in any language. An illiterate person is someone who cannot read and write with understanding, or any child aged 0–6, regardless of their ability to read or write.

The cytogenetic distribution among the 30 confirmed cases of Turner syndrome. Monosomy 45X was the most common karyotype, identified in 24 patients (80.0%). Mosaic Turner syndrome was observed in 6 patients (20.0%). No other cytogenetic variants were identified in this cohort.(Picture-1)



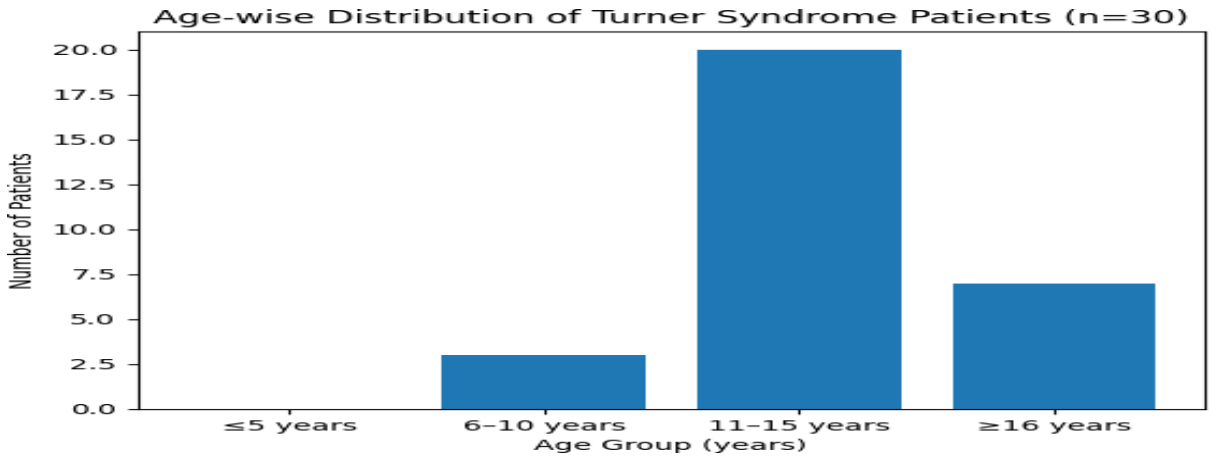
Picture 1: Cytogenetic Categories of Turner Syndrome Cases (n=30)

The mosaic karyotype patterns identified by conventional karyotyping and fluorescence in situ hybridization among the 6 patients with mosaic Turner syndrome. Mosaicism was confirmed in all six cases; however, the specific mosaic cell line patterns could not be further sub-classified in the present analysis. The total number of mosaic cases identified was six. (Table-2)

Table 2. Mosaic patterns identified by karyotype and FISH analysis (n = 6)

Mosaic karyotype pattern	Number of cases
45X / 46XX	06
45X / 46,X,i(Xq)	No
45X / 46,X,r(X)	No
45X / other cell line	No
Total mosaic cases	06

The age distribution at diagnosis among the 30 confirmed cases of Turner syndrome. No patient was diagnosed at or before 5 years of age. Three patients (10.0%) were diagnosed between 6 and 10 years, while the majority, 20 patients (66.7%), was diagnosed between 11 and 15 years. Seven patients (23.3%) were diagnosed at 16 years or older. (Table-2)(Picture-2)

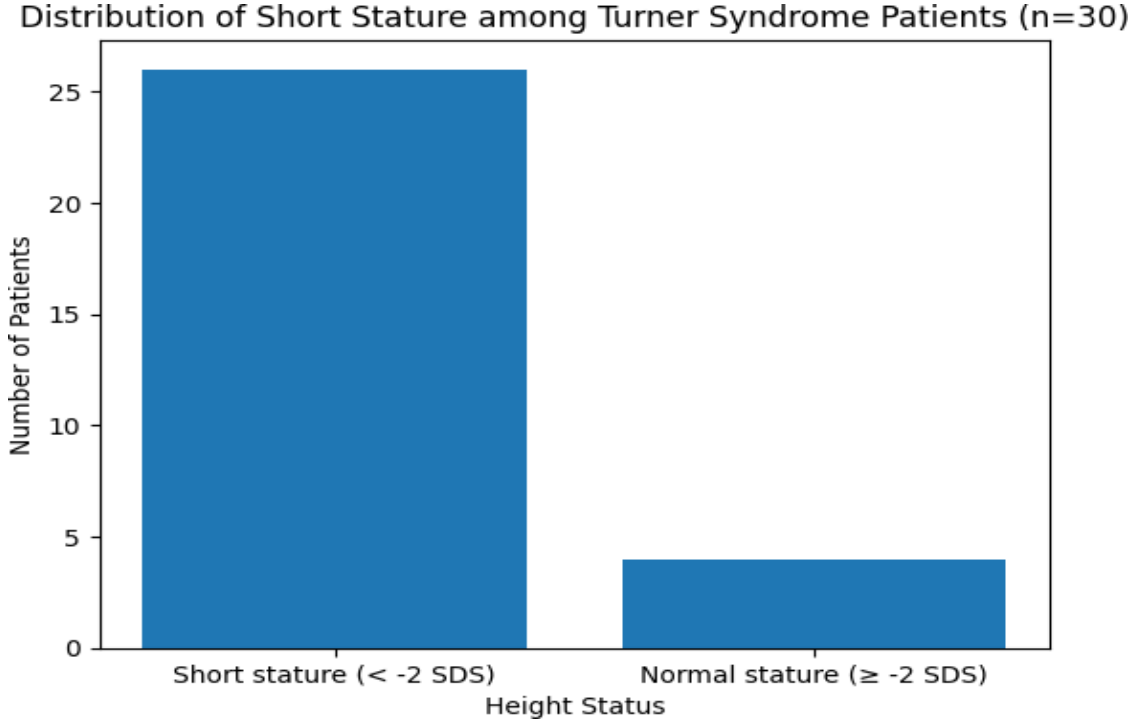


Picture 2: Age-wise Distribution of Turner Syndrome Patients (n=30)

The growth parameters of the 30 patients with confirmed Turner syndrome. Height ranged from 130 to 150 cm, and the mean height standard deviation score (SDS) was −3.1 ± 0.7. Body weight ranged from 25 to 40 kg, and body mass index (BMI) ranged from 18.4 to 22.7 kg/m². Short stature, defined as height less than −2 SDS, was observed in 26 patients (86.7%). (Table-3)(Picture-3)

Table 3. Growth parameters in confirmed Turner syndrome patients (n = 30)

Parameter	Mean ± SD / Median (Range)
Height (CM)	130-150
Height (SDS)	−3.1 ± 0.7. Short stature (height < −2 SDS)
Weight (kg)	25-40
BMI (KG/M2)	18.4-22.7
Short stature (< −2 SD)	26 (86.7%)



Picture 3: Bar chart showing height status of Turner syndrome patients based on standard deviation score

(SDS). Short stature (height < −2 SDS) was observed in 26 patients (86.7%), while only 4 patients (13.3%) had height ≥ −2 SDS.

The phenotypic features observed among the 30 patients with Turner syndrome. Short stature was present in 26 patients (86.7%). Primary amenorrhea was observed in 28 patients (93.3%). Other commonly observed features included shield chest in 26 (86.7%), cubitus valgus in 20 (66.7%), low posterior hairline in 21 (70.0%), and webbed neck in 16 (53.3%).

Congenital lymphoedema was present in 15 patients (50.0%), scoliosis in 8 (26.7%), cardiac abnormalities found in 08 patients (26.7%)(bicuspid aortic valve (BAV) and coarctation of the aorta (CoA)) and other dysmorphic features in 6 (20.0%). (Table-4)

Table 4. Phenotypic features observed in Turner syndrome patients (n = 30)

Clinical feature	Number of patients	Percentage (%) |
Short stature	26	86.7
Primary amenorrhea	28	93.3
Webbed neck	16	53.3
Shield chest	26	86.7
Cubitus valgus	20	66.7
Low posterior hairline	21	70
Congenital lymphoedema	15	50
Scoliosis	08	26.7
Cardiac Abnormalities	08	26.7
Other dysmorphic features	06	20.00

The correlation between cytogenetic pattern and selected clinical features among the 30 patients with Turner syndrome. Short stature was observed in 22 (91.7%) patients with monosomy 45X and 4 (66.7%) patients with mosaic

Turner syndrome (p = 0.17).

Webbed neck was present in 12 (50.0%) monosomy 45X cases and 4 (66.7%) mosaic cases (p = 0.65). Congenital lymphoedema was observed in 11 (45.8%) patients with monosomy 45X and 4 (66.7%) patients with mosaic karyotype (p = 0.39).

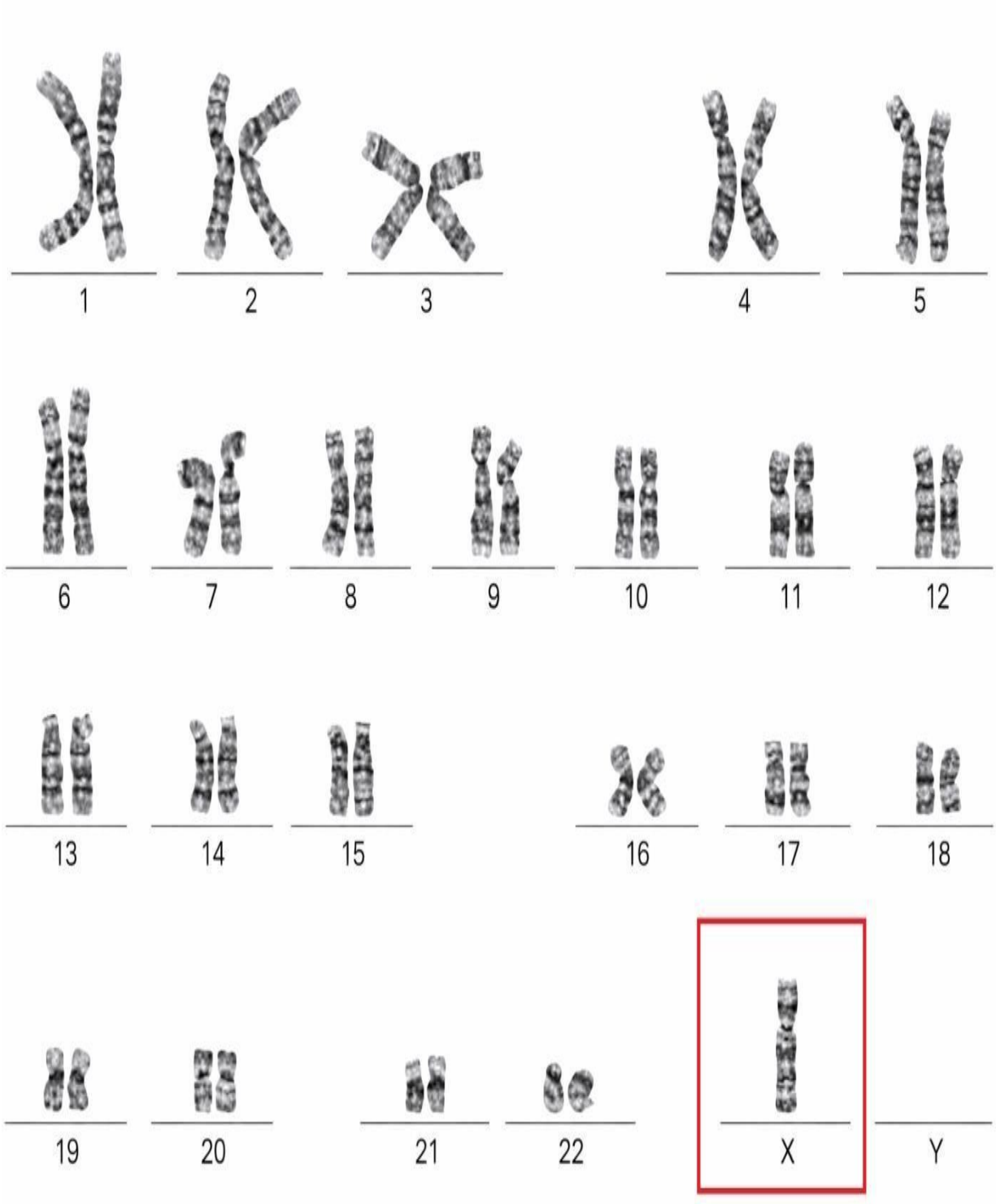
Primary amenorrhea was noted in 21 (87.5%) patients with monosomy 45X and 3 (50.0%) patients with mosaic Turner syndrome (p = 0.06).(Table-5)

Table 5. Correlation of cytogenetic pattern with selected clinical features in Turner syndrome (n = 30)

Clinical feature	Monosomy 45X (n = 24)	Mosaic TS (n = 6)	p-value
Short stature	22 (91.7%)	4 (66.7%)	0.17
Webbed neck	12 (50.0%)	4 (66.7%)	0.65
Congenital lymphoedema	11 (45.8%)	4 (66.7%)	0.39
Primary amenorrhea	21 (87.5%)	3 (50.00%)	0.06

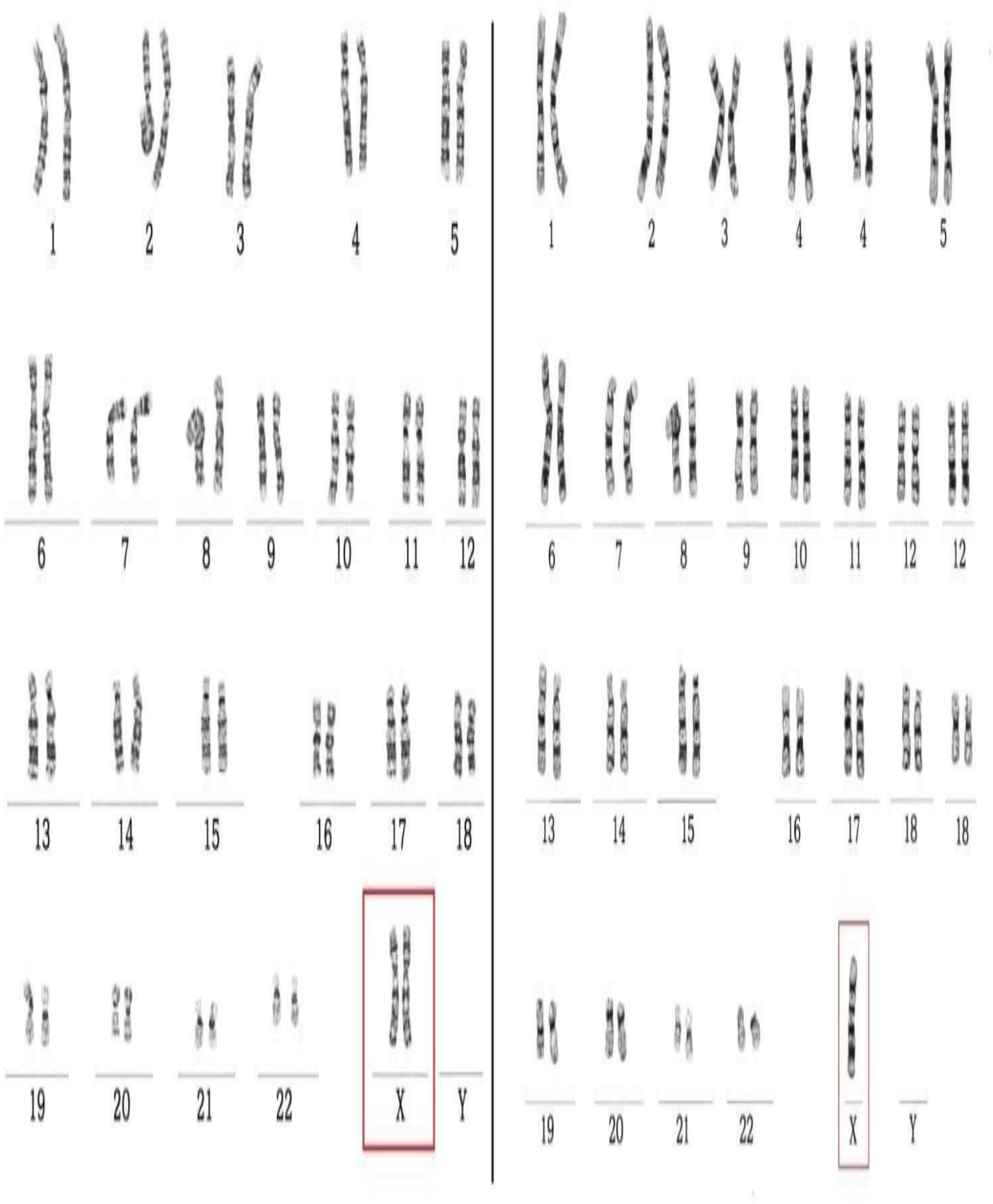
Conventional karyotype analysis (GTG banding) showing monosomy X (45X).

The karyotype demonstrates 22 pairs of autosomes with normal morphology and banding pattern. Analysis of the sex chromosomes reveals the presence of a single X chromosome with absence of the second sex chromosome, consistent with monosomy X (45X). This cytogenetic finding is diagnostic of Turner syndrome. The chromosomes are arranged in standard karyotypic order according to size and centromere position. (Picture-4)



Picture 4. Turner syndrome (45X)

GTG-banded karyotype demonstrating mosaic Turner syndrome.
The composite karyotype illustrates mosaic Turner syndrome, with two distinct cell lines observed on conventional cytogenetic analysis. One cell line shows a normal female karyotype (46XX), while the second cell line demonstrates monosomy X (45X) characterized by the presence of a single X chromosome and absence of the second sex chromosome. All autosomes (chromosomes 1–22) appear normal in number and morphology in both cell lines. The X chromosome is highlighted to emphasize the mosaic pattern. These findings are consistent with Turner syndrome mosaicism (45X/46XX). (Picture-5)

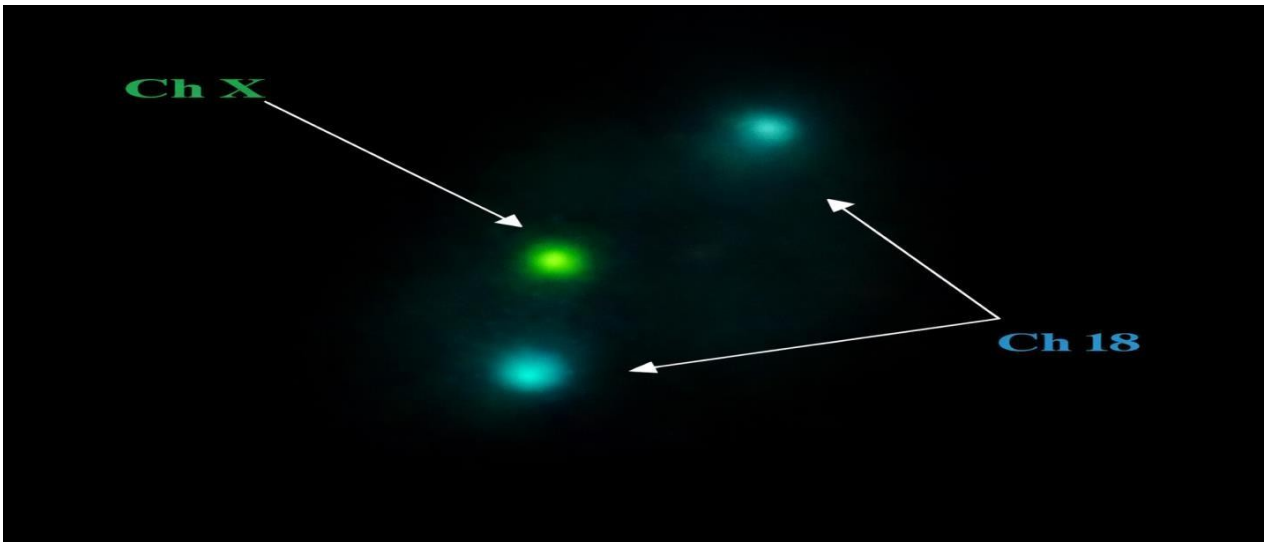


Picture 5. GTG-banded karyotype demonstrating mosaic Turner syndrome.

The clinical photographs depict a girl with Turner syndrome, showing characteristic phenotypic features including webbed neck (pterygium colli), short stature, and shield chest. The posterior view demonstrates a broad, webbed neck with low posterior hairline, while the frontal and lateral views highlight the typical body habitus associated with Turner syndrome. Facial features are partially masked to maintain patient confidentiality. These phenotypic findings correlate with the underlying chromosomal abnormality observed on cytogenetic analysis. (Picture-6)

Picture 6. Clinical features of a patient with Turner syndrome.

The interphase nucleus displays one green signal representing the X chromosome and two blue signals representing chromosome 18. This signal distribution is consistent with monosomy X and normal disomy 18, supporting the karyotype findings. (Picture-7)



Picture 7. Interphase FISH using X-specific (green) and chromosome 18-specific (blue) probes.

DISCUSSION:

Turner syndrome (TS) represents one of the most common chromosomal disorders affecting females, characterized by partial or complete monosomy of one X chromosome and a wide spectrum of phenotypic manifestations. The present study highlights the clinical profile and cytogenetic correlation of Turner syndrome patients from a tertiary care hospital in North Karnataka, India, and provides valuable regional data from a resource-limited setting.

In this study, monosomy 45X was the most frequent cytogenetic abnormality, accounting for 80% of confirmed cases, while mosaic Turner syndrome constituted 20%. This distribution is comparable with previous studies reporting monosomy X in approximately 30–50% of live-born TS patients, with mosaic forms comprising the remaining cases [11, 12]. However, the relatively higher proportion of monosomy X in our cohort may reflect referral bias and underdiagnosis of milder mosaic phenotypes, which often present later or remain clinically unrecognized [10,16].

A notable finding of the present study was the delayed age at diagnosis, with the majority of patients (66.7%) being diagnosed between 11 and 15 years of age, and none diagnosed before 5 years. Similar trends have been reported in Indian studies, where TS is often diagnosed during adolescence due to short stature or primary amenorrhea rather than during early childhood [13, 14]. This delay contrasts with developed countries, where early recognition through growth monitoring and prenatal screening is more common [9, 15]. Late diagnosis may limit the benefits

of early growth hormone therapy and timely pubertal induction.

Short stature was the most consistent clinical feature, observed in 86.7% of patients, which aligns with global data identifying growth failure as the hallmark of Turner syndrome [9,12]. The mean height SDS of −3.1 ± 0.7 in our cohort indicates significant growth impairment, emphasizing the need for early referral and endocrine evaluation. Primary amenorrhea was present in 93.3% of cases, reflecting gonadal dysgenesis, a classic feature of TS, particularly in monosomy 45X individuals. Turner syndrome is frequently associated with congenital cardiovascular anomalies, most notably bicuspid aortic valve (BAV) and coarctation of the aorta (CoA), contributing to increased cardiovascular morbidity in our study 08 (26.7%) patients shows similar cardiac abnormalities [15].

Other characteristic phenotypic features such as shield chest, cubitus valgus, low posterior hairline, and webbed neck were observed at frequencies comparable to previously reported series [12, 14]. Congenital lymphoedema was present in 50% of patients; a feature commonly associated with monosomy X and thought to result from abnormal lymphatic development during fetal life [11].

Genotype–phenotype correlation analysis revealed that patients with monosomy 45X had a higher prevalence of short stature and primary amenorrhea compared to mosaic TS patients, although the differences did not reach statistical significance. Similar observations have been reported by Hook and Warburton, who demonstrated that mosaic individuals often have milder phenotypes due to the presence of a normal cell line [11]. The trend toward a lower frequency of primary amenorrhea in mosaic cases in the present study (50% vs. 87.5%) suggests partial preservation of ovarian function, as documented in earlier studies [12, 16].

The use of fluorescence in situ hybridization (FISH) in selected cases strengthened the detection of mosaicism, particularly low-level mosaic cell lines that may be missed by conventional karyotyping alone. Identify the presence or absence of X-chromosome centromeric material or Y-chromosome sequences. This highlights the importance of adjunct molecular cytogenetic techniques in accurately characterizing TS, especially in patients with subtle phenotypes [15].

The predominance of urban referrals observed in this study likely reflects better access to healthcare facilities and diagnostic services, while rural patients may remain underdiagnosed. This urban–rural disparity has been consistently reported in Indian healthcare settings and underscores the need for improved awareness and referral mechanisms at the primary care level. [13].

Overall, the findings of this study reinforce the heterogeneity of Turner syndrome and the strong influence of cytogenetic pattern on clinical presentation. Early diagnosis through vigilant growth monitoring, timely cytogenetic testing, and multidisciplinary management is essential to reduce long-term morbidity and improve quality of life in affected individuals.

CONCLUSION:

This hospital-based study highlights the clinical spectrum and cytogenetic profile of Turner syndrome in patients evaluated at a tertiary care center in North Karnataka. Monosomy 45 X emerged as the most common cytogenetic abnormality followed by mosaic Turner syndrome, reinforcing the predominance of classic karyotypes in clinically recognized cases. Short stature and primary amenorrhea were the most consistent clinical features, with a high burden of characteristic dysmorphic findings such as shield chest, cubitus valgus, webbed neck, and congenital lymphoedema.

A key observation of this study was the delayed age at diagnosis, with the majority of patients being identified during early to mid-adolescence and none diagnosed in early childhood. This delay underscores gaps in early growth monitoring, awareness, and timely referral, particularly in resource-limited settings. Although monosomy 45X patients showed a trend toward more severe phenotypic manifestations compared to mosaic cases, the genotype–phenotype correlations did not reach statistical significance, likely due to the small sample size. Nevertheless, mosaic cases demonstrated relatively milder features, supporting the role of residual normal cell lines in phenotypic modulation.

The combined use of conventional karyotyping and FISH proved valuable in confirming diagnoses and detecting mosaicism, emphasizing the importance of comprehensive cytogenetic evaluation in suspected Turner syndrome cases. Overall, the findings reiterate the heterogeneity of Turner syndrome and the critical influence of cytogenetic patterns on clinical presentation.

Early diagnosis through vigilant growth surveillance, increased awareness among primary care physicians, and timely cytogenetic testing is essential to enable early initiation of growth hormone therapy, appropriate pubertal induction, and systematic screening for associated comorbidities. Strengthening multidisciplinary care and improving access to genetic services, especially for rural populations, can significantly improve long-term health outcomes and quality of life for girls and women with Turner

syndrome.

REFERENCES

1. 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.
2. Nielsen J, Wohlert M. Chromosome abnormalities found among 34,910 newborn children: results from a 13-year incidence study in Århus, Denmark. *Hum Genet*. 1991;87:81–83.
3. Hook EB, Warburton D. The distribution of chromosomal genotypes associated with Turner's syndrome: livebirth prevalence rates and evidence for diminished fetal mortality and severity in mosaics. *Hum Genet*. 1983;64:24–27.
4. Sybert VP, McCauley E. Turner's syndrome. *N Engl J Med*. 2004;351:1227–1238.
5. Verma RS, Lubs HA. Cytogenetic observations on Turner syndrome in Indian patients. *Indian J Med Res*. 1975;63:1125–1132.
6. Lippe B. Turner syndrome. *Endocrinol Metab Clin North Am*. 1991;20:121–152.
7. Bondy CA; Turner Syndrome Study Group. Care of girls and women with Turner syndrome: a guideline of the Turner Syndrome Study Group. *J Clin Endocrinol Metab*. 2007;92:10–25.
8. Gravholt CH. Epidemiological, endocrine and metabolic features in Turner syndrome. *Eur J Endocrinol*. 2004;151:657–687.
9. 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.
10. Nielsen J, Wohlert M. Chromosome abnormalities found among 34,910 newborn children: results from a 13-year incidence study in Århus, Denmark. *Hum Genet*. 1991;87:81–83.
11. Hook EB, Warburton D. The distribution of chromosomal genotypes associated with Turner's syndrome: live-birth prevalence rates and evidence for diminished fetal mortality and severity in mosaics. *Hum Genet*. 1983;64:24–27.
12. Sybert VP, McCauley E. Turner's syndrome. *N Engl J Med*. 2004;351:1227–1238.
13. Verma RS, Lubs HA. Cytogenetic observations on Turner syndrome in Indian patients. *Indian J Med Res*. 1975;63:1125–1132.
14. Lippe B. Turner syndrome. *Endocrinol Metab Clin North Am*. 1991;20:121–152.
15. Bondy CA; Turner Syndrome Study Group. Care of girls and women with Turner syndrome: a guideline of the Turner Syndrome Study Group. *J Clin Endocrinol Metab*. 2007;92:10–25.
16. Gravholt CH. Epidemiological, endocrine and metabolic features in Turner syndrome. *Eur J Endocrinol*. 2004;151:657–687.

