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ORIGINAL ARTICLE

Clinical and Genotypic Insights into Turner Syndrome: Emphasis on Cardiovascular Abnormalities

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Abstract

Objective. Turner syndrome (TS) is a genetic disorder characterized by X chromosome abnormalities in females. It presents with various clinical features, including short stature and cardiovascular anomalies. Limited awareness and diagnostic facilities contribute to the underdiagnosis of TS in India. The current study aims to explore genotype-phenotype associations with clinical characteristics and cardiovascular abnormalities in TS patients in India.

Methodology. A cross-sectional study involving 40 TS patients underwent clinical assessments and karyotyping. Data on demographics, anthropometry, Turner stigmata, cardiovascular evaluation, neurocognitive assessment and biochemical parameters were collected. The statistical analysis was conducted utilizing Statistical Package for Social Sciences (SPSS) version 27.0.

Results. Monosomy 45, X (55%) was the most prevalent genotype, with notable differences in age at diagnosis and height standard deviation score among genotypes. Thirty-five percent (35%) of participants had cardiovascular abnormalities, with a higher prevalence in the monosomy group. Lower IQ scores and increased thyroid autoimmunity were associated with specific genotypes. Additionally, a greater occurrence of skeletal and cutaneous stigmata, including cubitus valgus and multiple nevi, was observed in the monosomy group.

Conclusion. The study underscores the significance of genotype-phenotype associations in TS, emphasizing personalized management strategies. Early detection using sophisticated technologies like MRI, comprehensive assessments and assisted personalized management strategies to individual genetic profiles may improve cardiovascular and overall health outcomes in Turner syndrome patients.

Key words: Turner syndrome, genotype-phenotype associations, cardiovascular abnormalities, karyotyping, clinical characteristics

BACKGROUND

Turner syndrome (TS) is the sole survivable monomer syndrome in humans resulting from complete or partial absence of the X chromosome in females. It is relatively common, occurring in approximately 1 in 2,500 live female births.[1] Despite India's birth rate and the estimated incidence of TS, many cases likely go undetected due to limited awareness and diagnostic facilities.[2]

The primary phenotypic hallmark of TS is short stature, accompanied by characteristic features like a short neck, broad chest, genu valgum and nail dysplasia.[3] Of the complications associated with TS, non-congenital cardiovascular disease emerges as a major threat, responsible for over 40% of deaths, while congenital cardiovascular anomalies contribute around 8% to mortality.[4] Intriguingly, individuals with 45, XO monosomy exhibit a substantially higher prevalence of cardiovascular anomalies (38%) compared to those with 45, X/46, XX mosaicism (11%), consequently leading to elevated mortality rates.[5] Cardiovascular abnormalities such as aortic dissection, represent a significant concern in TS, contributing to a mortality rate three times higher than that of the general population.[6] Additionally, autoimmune diseases, notably thyroiditis,[7] and skeletal issues like osteoporosis elevate health risks, particularly in adult women with TS.[3]

Given the diverse clinical presentations of TS, contingent upon the patient's karyotype, this study aims to investigate the influence of various chromosomal irregularities on clinical and biochemical characteristics in TS patients.

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METHODOLOGY

Study design

This observational, cross-sectional study spanned 18 months from February 2020 to July 2021 in a tertiary care center in India. It focused on patients clinically diagnosed with TS attending the Endocrinology Department of Nil Ratan Sircar (NRS) Medical College Hospital, Kolkata. The diagnosis was confirmed through karyotypic analysis once they had provided informed consent.

Inclusion and exclusion criteria

Participants included patients with TS whose karyotyping was validated by examining 20 to 50 metaphase cells using the Giemsa-Trypsin-Giemsa (GTG) banding technique in a certified genetics laboratory. Patients with Noonan syndrome and those lacking karyotypic confirmation despite exhibiting features suggestive of TS were excluded.

Measurements and assessments

Anthropometric measurements, detailed clinical examination and pubertal status were recorded. Cardiovascular evaluation included electrocardiogram, echocardiography with colour Doppler and Magnetic Resonance Imaging (MRI). A cardiac MRI with aortography was done on a Siemens 3 Tesla MAGNETOM Verio dot machine done by a single operator to look for different cardiac and aortic anomalies. The aortic size index (ASI) is the ratio of the aortic diameter to body surface area. The diameter is usually taken at the level of the ascending aorta or the part of the aorta near the right pulmonary artery. The elongated transverse aortic arch was also noted, if present, which is a kinking of the inferior aortic contour at the aortic isthmus and/or left subclavian artery originating from a position posterior to the trachea in the horizontal plane and/or an increased distance between the left common carotid and left subclavian artery.

Neurocognitive evaluation included the Weschler Adult Intelligence Score[8] for subjects aged 15 and above, and the Malin's Intelligence Scale for Indian Children for those aged 5-15.

Routine biochemical parameters and hormonal parameters were analysed in an autoanalyzer and a Siemens immunoassay system (IMMULITE 1000), respectively, in our institution laboratory.

Sample size estimation

Previous studies done on genotype-phenotype associations in TS patients had sample sizes ranging from 16 to 52 participants.[9,10]

Considering α=0.05, β = 0.85, the sample size is estimated to be 43.

Similarly, considering an α=0.05, β = 0.80, the sample size is estimated to be 25.

Therefore, we considered a sample size of 40 in our study.

Ethical clearance

Written informed consent was obtained from all participants, and confidentiality was maintained. The study was conducted in accordance with the Declaration of Helsinki (1964) and approved by the Institutional Research Ethics Committee.

Statistical analysis

The study participants were classified into three groups based on their karyotypic profile: Monosomy (Genotype A), Mosaicism (Genotype B) and Structural defects (Genotype C). Continuous data were displayed as mean with standard deviation (SD), and categorical variables were represented as frequency (N) and percentage (%). The data was entered into a Microsoft Excel sheet and analyzed utilizing Statistical Package for Social Sciences (SPSS) (version 27.0; SPSS Inc., Chicago, IL, USA) and GraphPad Prism version 5. Demographics, clinical features and biochemical parameters were compared using Chi-square test (for categorical variables) or one-way ANOVA (for continuous variables). The differences in the prevalence of cardiovascular abnormalities were compared between the groups A, B and C using Chi-square test. The association between the aortic size index and the genotypes identified were compared using One-way ANOVA. A p-value of ≤0.05 was deemed statistically significant.

RESULTS

The study comprised forty (N = 40) subjects diagnosed with TS, confirmed through clinical stigmata assessment and karyotypic analysis. Among the participants, fifty-five percent (n=22) exhibited monosomy, denoted as Genotype A, while twenty-five percent (n=10) presented with structural defects, including seven isochromosomes, two ring chromosomes, and one deletion at Xp22.3, categorized as Genotype C. Additionally, twenty percent (n=8) displayed mosaicism, categorized as Genotype B (Figure 1).

Significant differences were observed in the mean age at diagnosis across genotypes, with individuals of Genotype A diagnosed at an average age of 7.4 years, while those of Genotypes B or C were diagnosed at 11.8 years (P<0.05). Similarly, there was notable variability in mean height SDS, with the monosomy genotype exhibiting the lowest value (-4.4 ± 1.33), followed by the structural defect genotype (-3.80 ± 1.18), and the mosaic genotype showing the least decrease in height (-3.13 ± 0.86) (P<0.05). No significant differences in the IQ scores have been observed between individuals with Genotype A (83.7 ± 10.8), Genotype B (87 ± 10.2) and Genotype C (87.4 ± 14.4). For a comprehensive overview of additional characteristics, refer to Table 1.

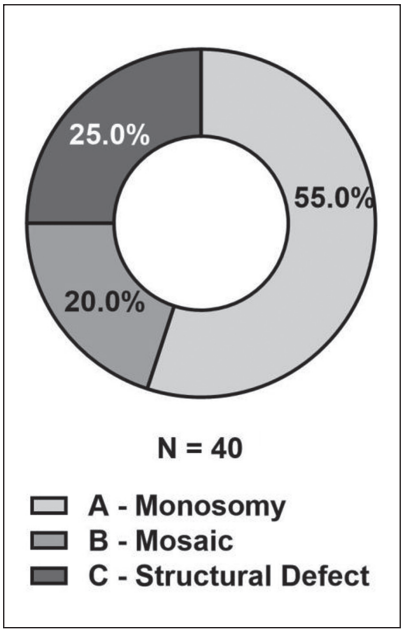

Figure 1. Distribution of karyotypes among subjects with Turner syndrome (n = 40).

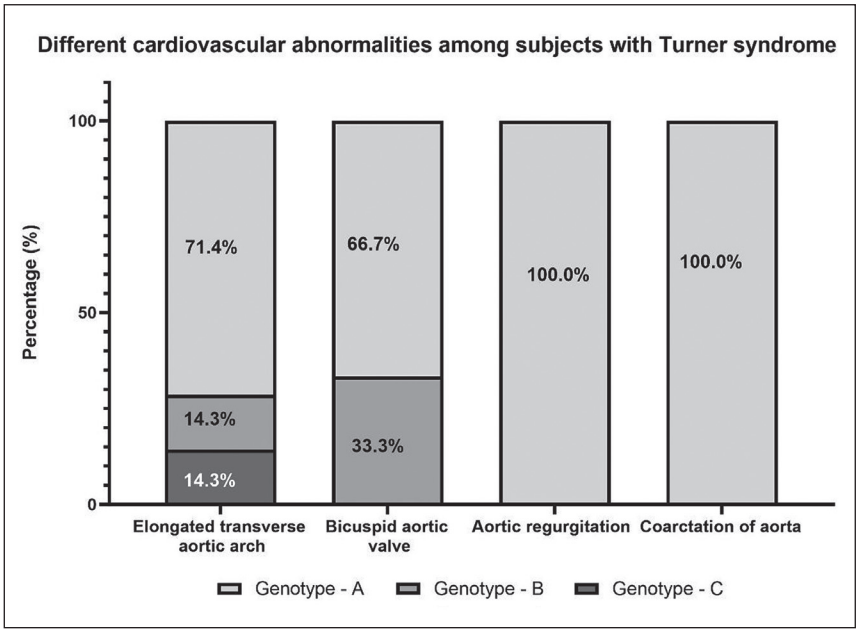

Figure 2. Different cardiovascular abnormalities among subjects with Turner syndrome (n = 40).

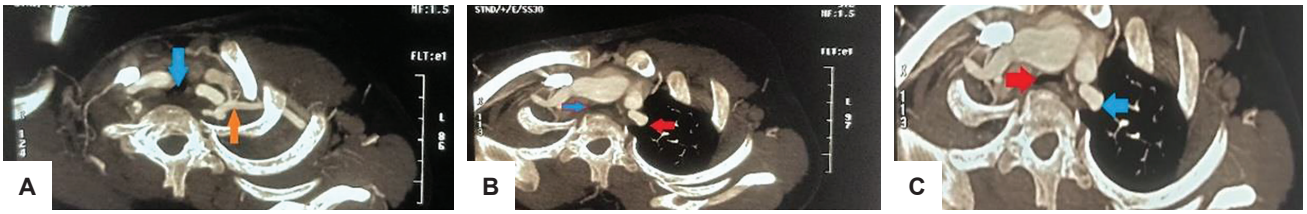

Figure 3. Cardiac MRI showing cardiovascular abnormalities in subjects with Turner syndrome. **(A)** Dilated subclavian artery *(blue arrow)* and elongated transverse aortic arch *(orange arrow)*; **(B)** Coarctation of aorta *(blue arrow)* and dilated subclavian artery *(red arrow)*; **(C)** Coarctation of aorta *(red arrow)* and dilated subclavian artery *(blue arrow)*.

Among the study participants, 14 cases exhibited cardiac abnormalities, accounting for 35% of the total cohort (Figure 1 and Table 2). Additional analysis indicated that the monosomy group exhibited a higher percentage of cardiovascular abnormalities than the other genotype groups (Figure 2). Figure 3 shows a representative image suggestive of an elongated transverse aortic arch.

Additionally, the mean aortic size index in the monosomy group was 2.7 cm/m^2 in the monosomy group, while it measured 2.3 cm/m^2 in the non-monosomy group (Table 3).

DISCUSSION

This study examined the correlation between genotypes and the diverse clinical and biochemical characteristics in individuals of Indian descent diagnosed with TS, with a specific focus on cardiovascular abnormalities. Our study revealed significant associations between genotypes and certain clinical and biochemical features.

The distribution of genotypes among the 40 patients with TS in our study closely resembles that reported in existing literature. Monosomy 45, X (45, XO) was the most prevalent genotype, comprising 55% of the cases, followed by structural defects and mosaic genotypes. A previous study in Indian TS patients observed monosomy in 69% of cases, followed by mosaic genotypes (12%) and structural defects (19%).[11] Similarly, studies across the globe have reported monosomy to be the most common (~50%) genetic defect, followed by mosaic genotypes (10-30%) and structural defects (10 to 38%).[12,13]

Monosomy X in Turner syndrome increases the risk of heart defects due to the loss of genes on the second X chromosome, which are critical for normal cardiovascular development.[14] For example, a haploinsufficiency of *TIMP1* has been associated with increased risk for bicuspid aortic valve, aortic aneurysm formation and dissection.[15,16] This highlights the need for early cardiac screening and lifelong monitoring to prevent severe complications.

Our study revealed a lower mean age at diagnosis, particularly in the monosomy group, compared to the non-monosomy groups. This finding aligns with recent Indian studies reporting a mean age at presentation of approxi-

Table 1. Prevalence of different clinical and biochemical features among subjects with Turner syndrome (n = 40)

Abnormalities	Genotype-A (N = 22) n (%) or mean ± SD	Genotype-B (N = 8) n (%) or mean ± SD	Genotype-C (N = 10) n (%) or mean ± SD	*p*-value
Age at diagnosis (yr)	7.4 ± 2.1	12 ± 5	11.8 ± 2.7	**0.0002*** **
Height SDS	-4.4 ± 1.3	-3.12 ± 0.86	-3.8 ± 1.2	**0.03***
Weight SDS	-1.67 ± 0.7	-1.09 ± 0.83	-1.12 ± 0.6	0.05
TPO positive	13 (59.1%)	0 (0%)	7 (70%)	**0.01***
FSH (mIU/mL)	86.4 ± 18.7	60.8 ± 42.6	62.5 ± 28.3	**0.02***
Estradiol (pg/mL)	7.2 ± 1	19.5 ± 6.8	16.4 ± 3.5	**0.0001*** **
AMH (ng/mL)	1.6 ± 0.7	4 ± 1.8	2.9 ± 2	**0.0007*** **
IQ Scores	83.7 ± 10.8	87 ± 10.2	87.4 ± 14.4	0.64
Verbal-Performance IQ	11.4 ± 1.9	12.5 ± 0.5	15.2 ± 1.4	**<0.0001**** **
Multiple pigmented nevi	18 (81.8%)	2 (25%)	6 (40%)	**0.01***
Cubitus valgus	19 (86.4%)	2 (25%)	6 (60%)	**0.01***
Short 4th Metacarpal	19 (86.4%)	2 (25%)	6 (60%)	**0.01***
Low posterior hairline	19 (86.4%)	2 (25%)	6 (60%)	**0.01***
Shield chest	4 (18.2%)	4 (50%)	0 (0%)	**0.03***
Low set ears	17 (77.3%)	6 (75%)	4 (40%)	0.1
Short/webbed neck	11 (50%)	2 (25%)	4 (40%)	0.46
Scoliosis	3 (13.6%)	0 (0%)	2 (20%)	0.43
High arched palate	16 (72.7%)	2 (25%)	6 (60%)	0.06

OR: Odds Ratio; SDS: Standard Deviation Score; TPO: Thyroid Peroxidase; FSH: Follicle-Stimulating Hormone; AMH: Anti-Müllerian Hormone. *, ***, **** represents *P* <0.05, *P* <0.001, *P* <0.0001 respectively.

Table 2. Association between different cardiovascular abnormalities among subjects with Turner syndrome and the genotype

Abnormalities	Genotype-A (N = 22) n (%)	Genotype-B (N = 8) n (%)	Genotype-C (N = 10) n (%)	*p*-value	Picked up by Echocardiography	Picked up by Cardiac MRI
Bicuspid aortic valve	2 (9.1%)	1 (12.5%)	0 (0%)	0.5545	Yes	Yes
Elongated transverse aortic arch	5 (22.7%)	1 (12.5%)	1 (10%)	0.6236	No	Yes
Aortic regurgitation	3 (100%)	0 (0%)	0 (0%)	-	Yes	Yes
Coarctation of aorta	1 (100%)	0 (0%)	0 (0%)	-	Yes	Yes

Table 3. Association between ASI: Genotype

ASI	Number	Mean	SD	Minimum	Maximum	Median	*p*-value
Genotype-A	11	2.7009	0.1053	2.4500	2.8000	2.7100	**0.0013**
Genotype-B	2	2.3500	0.0707	2.3000	2.4000	2.3500	
Genotype-C	1	2.3800	0.0000	2.3800	2.3800	2.3800	

ASI – Aortic Size Index

mately 14 years, with many patients primarily presenting with gonadal failure.[17] This discrepancy in age at diagnosis may reflect varying clinical presentations influenced by karyotype. Therefore, healthcare providers must be vigilant to facilitate early detection of TS in adolescents, particularly among those presenting with gonadal issues.[17]

Additionally, the mean height Standard Deviation Score (SDS) across all ages was notably shorter (between -3.12 and -4.4) than that found in international studies (~ -2.0 to -3.0),[18] suggesting a characteristic of the Indian TS population.[19]

The shorter stature observed in the current study may be attributed to delayed diagnosis and a subsequent delay in initiating growth-promoting therapies, certain ethnic and genetic factors specific to the Indian population, and associated comorbidities such as renal anomalies and cardiac defects. Additionally, factors such as access to healthcare resources and nutritional status could influence growth outcomes in this group.[11] In our study, individuals with monosomy (45, X) had a lower mean height SDS than other genotypes. This could be attributed to the additional effect of haploinsufficiency of genes such as SHOX on the X chromosome.[11]

The current study also observed an increased frequency of skeletal and cutaneous markers, with cubitus valgus and multiple nevi being the most prevalent (Table 1). Previous studies have also observed that the clinical manifestations of TS are most pronounced in individuals with monosomy 45, X, least severe in those with mosaicism 45, X/46, XX, and intermediate in individuals with iso-chromosome and Y-material genotypes.[20,21]

The observed variation in clinical severity among TS karyotypes suggests differential gene expression and epigenetic influences. Unique genetic makeup and interactions between X chromosome genes and other factors likely contribute to phenotypic variability.[20,21] In the current study, stigmata were more common in the monosomy group, particularly cubitus valgus, short fourth metacarpal, low posterior hairline, multiple pigmented nevi, and shield chest.

Early detection of cardiovascular abnormalities in TS is crucial due to their potential impact on morbidity and mortality. In the current study, a slightly higher prevalence (35%) was observed compared to previous reports of 25 – 30%, consistent with findings from a Taiwanese cohort that noted these abnormalities predominantly in the classical 45, XO TS group.[22] This highlights the importance of cardiac MRI over echocardiography, particularly for detecting abnormalities like elongated transverse aortic arch (ETAA) and cardiac lesions.[23,24] While cardiac MRI offers detailed imaging, accessibility and cost can limit its widespread use, necessitating a balanced approach in integrating both techniques for comprehensive cardiovascular assessment in TS patients.[23]

Examining IQ scores in TS patients reveals potential cognitive impairments, impacting academic, social and daily functioning, reflecting the importance of tailored interventions for improved quality of life.[25] Regarding cognitive function, we found lower IQ scores in our TS patients (Avg. IQ – 83.7) compared to previous studies.[25,26] This observation is consistent with research indicating an increased incidence of intellectual disability, particularly in patients with ring chromosome r(X).[27] Additionally, hypothyroidism was found to be most common in isochromosomes, possibly due to enhanced autoimmunity involving the thyroid gland.[17,28]

Limitations

The study's small sample size limits the generalizability of findings to the broader TS population. Limited awareness and diagnostic facilities in India may contribute to a biased sample that does not fully represent the spectrum of TS, particularly milder cases. Lack of information on confounding variables and significant delays in diagnosing TS in patients could impact the observed phenotype and the severity of cardiovascular abnormalities.

CONCLUSION

Our cohort revealed that classic monosomy is the most common form of Turner Syndrome. This group had the lowest SDS height and had more cardiac defects, the most common being an elongated transverse aortic arch. Advanced imaging techniques like cardiac MRI are crucial for accurately diagnosing cardiovascular abnormalities. Additionally, our findings highlight lower IQ scores and increased thyroid autoimmunity in certain genotypes, emphasizing the need for comprehensive assessments and targeted interventions. Early detection and appropriate management are essential for improving outcomes in TS patients.

Statement of Authorship
All authors certified fulfillment of ICMJE authorship criteria.

CRediT Author Statement
MC: Conceptualization, Methodology, Software, Validation, Formal Analysis, Investigation, Resources, Data Curation, Writing – original draft preparation, Writing – review and editing, Visualization, Supervision, Project administration, Funding acquisition; **AM:** Validation, Formal analysis, Data Curation, Writing – review and editing; **PKS:** Conceptualization, Methodology, Validation, Resources, Data Curation, Writing – review and editing, Visualization, Supervision, Project administration, Funding acquisition.

Data Availability Statement
Datasets generated and analyzed are included in the published article.

Author Disclosure
The authors declared no conflict of interest.

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