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Clinical profile of patients of turner syndrome (TS) with karyotype-phenotype corelation from a tertiary care hospital in Eastern Uttar Pradesh (UP), India

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ABSTRACT

Objective: The present study was done to study the clinical profile and karyotype-phenotype corelation of turner syndrome patients in eastern UP, India.

Materials and Methods: The present study was a retrospective observational study conducted from January 2018 to December 2020 on newly diagnosed TS patients. All patients were screened for thyroid dysfunction, celiac disease, diabetes mellitus (DM), dyslipidaemia, liver dysfunction, hearing loss, cardiovascular anomalies and renal anomalies. Data was tabulated in Microsoft excel sheet and averages and means were calculated. Fischer exact test was used to assess the corelation of karyotype with clinical phenotypic features.

Results: Total 16 patients were diagnosed with TS and 37.5% were classic 45 XO, 18.5% were mosaic 45X/46XX and rest 43.75% were of rarer TS variants. One patient had mosaicism for X chromosome with reciprocal autosomal translocation- 45X, t(12,20)(q24.1p13), 46X, t(12,20)(q24.1p13) *marker karyotype which is the first case reported so far. The average age of presentations was 16.8years ± 3.4years (range 8 to 23 years). One patient with karyotype 46,X, del(Xq22-28) had DM with negative anti-GAD antibodies and one patient with karyotype 46XX/46,X+marker had systemic lupus erythematosus (SLE). No significant karyotype and phenotype corelation was found in our study.

Conclusion: We report rare association of SLE with TS and a novel karyotype in TS involving mosaicism for X with autosomal translocation t(12,20). No significant karyotype-phenotype corelation was found in our study. More focused studies are needed to study the genes responsible for various manifestations in TS, pathogenic mechanisms of DM and SLE in TS and the effect of autosomal translocations in TS phenotype.

1. Introduction

Turner syndrome (TS) or Ullrich TS was first described independently in 1930 by German paediatrician Otto Ullrich and in 1938 by American endocrinologist Henry Hubert Turner as a syndrome of infantilism, congenital webbed neck and cubitus valutas in phenotypic female.[1] In 1959 missing X chromosome was found to be associated with TS by Ford et al.[2] TS patients are phenotypically females with genotype comprising of one intact X chromosome and

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complete or partial absence of second sex chromosome. The common presentation is with short stature, absence of menses and non-development of pubertal changes with one or other features involving facial features, skeletal system, skin, cardiovascular system, renal system, neurocognitive, endocrinopathies, autoimmunity, hearing. This syndrome is found in all ethnic communities and prevalence is ~ 1 in 2500 live births.[3] The monosomy X (45XO) is the most common genotype observed, others being mosaicism, deletions and structural abnormalities involving X chromosome. Its incidence is not related to maternal age and abnormality mostly is due to paternal meiotic errors (75%)- meiotic nondisjunction, deletion of the short arm or abnormal Y chromosomes.[4] The clinical features result due to haploinsufficiency of genes normally expressed in both sex chromosomes and which escape X inactivation.[5] The present study was done to study the clinical profile of TS patient in our part of India in eastern UP.

2. Materials and Methods

The present study was a retrospective observational study conducted in the department of Endocrinology & Metabolism, IMS, BHU, Varanasi, India, a tertiary care facility. All the patient of TS who were diagnosed from January 2018 to December 2020 were included in the study. Data regarding their age of first presentation, chief complaints, clinical manifestations and investigations were documented. Diagnosis of TS was made on basis of clinical features, raised s.FSH levels along with chromosomal analysis on peripheral blood using standard GTG banding karyotype technique on 20-30 cells obtained from peripheral blood (450-550 resolution). All patients underwent ultrasonography for evaluation of uterus and ovaries and all were screened for thyroid dysfunction, celiac disease, DM, dyslipidaemia, liver dysfunction, hearing loss, cardiovascular anomalies and renal anomalies. Data was tabulated in Microsoft excel sheet and averages and means were calculated. Fischer exact test was used to assess the corelation of karyotype with clinical phenotypic features.

3. Results

The total 16 patients were diagnosed with TS during 3 years period and out of which 37.5% were of classic 45,XO, 18.5% were of mosaic 45X/46XX and rest 43.75% collectively were of rarer variants of TS involving structural abnormalities (Table 1). One of the rarer variants was mosaicism for X chromosomes with reciprocal autosomal translocation- 45X, t(12,20)(q24.1p13), 46X,t(12,20)(q24.1p13) *marker which is so far to the best of the literature search have been reported for the first time here.(Table 3). The most common complaint at presentation were primary amenorrhoea (87.5%) and non-development of secondary sexual characteristics (75%) followed by short stature in 68.75%. The average age of presentations was 16.8years ±3.4years (range from 8 to 23 years). One patient had presented in prepubertal years for short stature at age of 8 years. Clinical features and baseline characteristics of total patients and according to karyotypes have been tabulated in Table 2. The average height deficit in these patients was -3.1 SDS with range from -0.9 SDS in 46, X, del(Xq22-28) karyotype to -5 SDS in 45X, t(12,20)(q24.1p13), 46X, t(12,20)(q24.1p13)*marker. Only 2 (12.5%) patients developed spontaneous menarche followed by secondary amenorrhoea (45, XO and 46, X, del(Xq). These patients had partial development of secondary sexual characteristics also. All patient on ultrasonography showed hypoplastic uterus with 50% patients showing streak ovaries and rest 50% hypoplastic ovaries. None of the patient showed signs for virilization. All patients had s.FSH > 40 IU/ml except one patient at age of 8 years had FSH of 12 units. None of the patients had liver dysfunction, dyslipidaemia or cardiac abnormality at diagnosis. One patient had DM with karyotype 46, X, del (Xq22-28). She was detected to have diabetes at presentation through routine screening in TS. She had normal BMI of 21.8 Kg/m2 with no family history and no signs of insulin resistance. Anti-GAD antibodies were negative Table 3. Another patient with karyotype 46XX/46,X+marker had SLE. Few cases so far have been reported associated with TS. She had discoid lupus rashes, arthritis, and thrombocytopenia with ANA and anti-ds DNA antibodies positive Table 3. No significant karyotype and phenotype correlation was found in our study Table 1. We managed patients with sequential hormonal therapy as all but one patient were of age >12 years. One patient who presented at age of 8 years was planned for growth hormone (GH) therapy.

Table 1: Type and frequency of chromosome abnormalities in Turner syndrome

		Karyotype	N (%)
1		45, XO	6 (37.5%)
2		45, XO/46,XX	3 (18.75%)
3	**Total**	Structural variants	7 (43.75%)
	a	46, X, del (Xq)	3 (18.75%)
	b	46, X, del (X q22-28)	1 (6.25%)
	c	46X, +marker	1(6.25%)
	d	46X, i((Xq10)/ 45XO	1(6.25%)
	e	45X, t(12,20)(q24.1p13), 46X, t(12,20)(q24.1p13) *mark	1(6.25%)

4. Discussion

We here present the clinical profile of TS patients from our part of the country. The average of presentation in our study was 16.8 ±3.4 years and 93.75% of these patients were diagnosed after the age of 12 years. There was no significant

Table 2: Clinical features of TS patients and corelation of clinical parameters with karyotype

Parameter	Total	45, XO	45, X/46, XX	Structural variant	Fisher's Exact Test (p value)
Number (n)	16 (100%)	6 (100%)	3 (100%)	7(100%)	-
Av age at presentation	16.8 ±3.4	14.6 ±3.8	18.3± 4.04	18±2.3	0.457
Av height (cm)	135.5±12.36	127.7±10.3	144±9.5	138.5±12.4	0.383
Ht SDS	-3.1	-3.66	-2.3	-3.1	
Primary amenorrhea	14 (87.5%)	5 (83.3%)	3 (100%)	6(85.7%)	0.562
Secondary amenorrhoea	2(12.5%)	1 (16.6%)	0	1 (14.3%)	-
Absent Secondary sexual characteristics	12 (75%)	5 83.3%)	3 (100%)	4(57.1%)	0.430
Autoimmune thyroiditis	11 (68.75%)	3 (50%)	2 (66.6%)	6 (85.7%)	0.519
On treatment for hypothyroidism	6 (37.5%)	2 (33.3%)	0	4 (57.1%)	
Coeliac ds	2 (12.5%)	1 (16.6%)	0	1(14.3%)	1.0
Eye	1(6.5%)	1 (16.6%) strabismus	0	0	0.562
Ear	33 (18.75%)	2(33.3%) Middle ear infections, hearing defect, low set ears	1 (33.3%) Low set ears	0	0.250
Mouth	8(50%)	5(83.3%) High arched palate, abnormal teeth, micrognathia	1(33.3%) Micrognathia	1(14.3%)	0.441
Neck	7 (43.75%)	4 (66.6%) Webbing, low post hair line, SN	1(33.3%)	2(28.6%)	0.431
Thorax	7(43.75%)	1 (16.6%) SC	2(66.6%) SC, inverted nipples	4(57.1%)	0.287
Nevi	8(50%)	4 (66.6%)	1 (33.3%)	3(42.9%)	0.674
Skeletal system	14(87.5%)	5(83.3%) CV, short MT 4 and 5 B/L, scoliosis, PE	3 (100%) CV, short MC, scoliosis, clinodactyly	6(85.7%)	0.562
Heart	0	0	0	0	-
Kidney	1(6.25%)	0	0	1(14.3%) Horseshoe kidney	-
Neurocognitive	4(25%)	3(50%) Cognitive, nonverbal, calculation	0	1(14.3%)	0.519
Other associations	2(12.5%)	0		2(28.6%) SLE, DM	-
Audiometry	1(6.25%)	1(16.6%) SNHL	0	0	-

SN: Short neck, SC: Shield chest, CB: Cubitusvalgus, MT: Metatarsal, MC: Metacarpal, PE: Pectus excavatum, DM: Diabetesmellitus, SLE: Systemic lupus erythematosus, SNHL: Sensorineural hearing loss

Table 3: Clinical features of TS patients with karyotypes involving structural abnormalities in chromosomes

Parameter	46, X, del(Xq)	46, X, del (Xq22-28)	46xx/46, X+marker	46, X, i(Xq10)/ 45XO	45, XO, t(12,20)(q24.1p13)/ 46X, t(12, 20)(q24.1p13) *mark
Number (n)	3 (100%)	1 (100%)	1 (100%)	1 (100%)	1 (100%)
Av age at presentation	18.3±1.5	17	19	14	21
Av height (cm)	138.6± 11.1	152	150	120	132
Ht SDS	-3.4	-0.9	-1.2	-4.3	-5
Primary amenorrhea	2	1	1	1	1
Secondary amenorrhoea	1	0	0	0	0
Absent Secondary sexual characteristics	1	1	1	1	0
Autoimmune thyroiditis	2	1	1	1	1
Coeliac ds	1	0	0	0	0
Eye	0	0	0	0	0
Ear	0	0	0	0	0
Mouth	1 Abnormal teeth	0	0	1 High arched palate	0
Neck	2 Webbing, SN, low posterior hair line	0	0	0	0
Thorax	2 SC, inverted nipples	1 SC	0	1 SC	0
Nevi	2	0	0	0	1
Skeletal system	2 CV, short MC, scoliosis, brachydactyly	1 CV, splayed foot	1 CV, short MT	1 CV	1 Genu varum
Heart	0	0	0	0	0
Kidney	0	0	0	1 horse shoe kidney	0
Neurocognitive	0	0	0	0	1
Other associations	0	DM, Anti GAD negative	SLE	0	0
Audiometry	0	0	0	0	0

SN: Short neck, SC: Shield chest, CB: Cubitus valgus, MT: Metatarsal, MC: Metacarpal, DM: Diabetes mellitus, SLE: Systemic lupus erythematosus

difference in average age of presentation of classic 45XO and non-classic karyotypes. Similar age of presentation of 15.8±3.6 years was observed by Purvar N et al. from Jaipur, India in 2021.[6] This age of presentation is quite late as compared to western population. In a follow-up study from Belgium in 2005, median age of presentation decreased from 11.2 years in 1991 to 6.6 years in 2003 with patients diagnosed at age >12 years decreased from 45% to 22%. More number of patients diagnose prenatally and infancy (30%).[7] Similar trend was seen in study from UK, with average age of diagnosis at 5.89 ± 5.3 years and 20% patients diagnosed at age >12 years.[8] This persistent trend of delayed diagnosis in our population is unfavourable as we miss the critical window of opportunity to use GH and hormonal replacement therapy at an appropriate age to have normal final adult height. This also reflects the neglect of girls' health in Indian households and also less awareness among paediatrician and primary care physicians for the syndrome.[8–10]

Our study had 37.5% of patients with classical 45, XO genotype, 18.75% with mosaic 45XO/46XX and 43.75% with structural variants with or without mosaicism. The overall worldwide reported prevalence of 45, XO karyotype is 40-50%, 45XO/46XX mosaicism of 15-25% and isochromosome of 10% and rest being rare karyotypes.[5,11] Mondal S et al. from India reported 44.1% of classical 45, XO karyotype followed by 26.5% of isochromosomes.[12] We did not find any significant corelation of karyotype with phenotypic features in our study likely due to small study group. Literature has variable findings on genotype-phenotype corelation. There is corelation of classic turner's stigmata in 45 XO, severe short stature with loss of Xp in 45 XO, isochromosome, and Xp deletion, 2-fold increased autoimmune diseases (AD) prevalence with isochromosomes, mental retardation, learning disability and autism spectrum disorder in ring chromosomes, sensorineural hearing loss with 45 XO and isochromosomes, virilization and gondaoblastomas with Y chromosome or part of Y-chromosome. These typical

phenotypic features are mostly due to haploinsufficiency of the genes normally expressed in both sex chromosome that escape X inactivation.[5] But specific genotype does not always predict karyotype. In a study from India on 103 patients, higher skeletal and cutaneous stigmata (cubitus valgus-68.3%, multiple nevi -68.2%) were found in classic 45,XO karyotype, bicuspid aortic valve in 45,X/46,XX mosaics (33.3%), aortic coarctation exclusively in classic 45, XO 7.2%). Overt hypothyroidism, conductive deafness and recurrent otitis media were commonest in isochromosomes (P<0.03) and 45,X/46,XY mosaics had highest IQ (P<0.005).[12] In another study from Saudi Arabia on 52 patients did not find reliable evidence of karyotype predict the clinical presentation.[13]

Our one patient with karyotype 46X, del(Xq22-28) had DM. She was managed with sulfonylureas and was well controlled on follow up. Abnormal glucose metabolism is found in >70% of adults with TS and about 30% patients developing diabetes.[14] Specific DM phenotype in TS is unclear but adult phenotype resembles Type 2 DM. A study from Denmark in 1998, showed that the incidence of type 1 DM and Type 2 DM in TS patients is 11 times and 3–4 times greater than that in healthy people, respectively.[15] In a study evaluating X chromosome gene dosage and risk of DM in TS, found overall prevalence of DM 25% which was 3 -4-fold more and type 1 DM 0.45% which was similar to adult U.S. population prevalence. DM prevalence was significantly more in 45, XO (17%) and delXp (23%) group, but not in the delXq group (9%). They also found incidence of DM was significantly higher in the iXq (3 copies of Xq) than the 45, XO group (40 vs. 17.3%; P=0.003). The prevalence of anti-GAD antibodies was 21.4% in iXq group as compared to 5% in 45Xgroup (p=0.03).)[16] These findings suggests that haploinsufficiency for Xp genes confers a significantly increased risk for DM that is amplified in the iXq group by the presence of additional doses of Xq genes. Haploinsufficiency for Xp genes causes the basic deficit in beta -cell functions and excess dosage of Xq genes in isochromosomes exacerbates the deficit by altering other genes involved in beta -cell development and function or by stimulating low-grade chronic autoimmunity in the beta cells. Additional Xq copies leads to overexpression of escape-inactivated gene that includes diabetes-related genes such as islet cell antigen (ICA), C-reactive protein (CRP), insulin-like growth factor-II (IGFI-II), and other genes for normal physiological functions of islet cells (GLIS3, KLF11).[14,16]

Women with TS have 2–3-fold higher incidence of AD than normal female population.[17] In a study from Denmark, a standardized incidence ratio (SIR) for male-predominant AD and female predominant AD observed was 3.9 and 1.7 respectively. Among the rheumatological AD, juvenile idiopathic arthritis (JIA) is the most frequently observed (6-fold).[18] SLE in TS patients is extremely rare. There are only few isolated case reports of association of SLE with TS - one each with karyotype 46, X, del(X)(q13), 45X/46,XXq+ and 45, XO.[19] Our patient had 46, X+ marker karyotype which is novel karyotype association with SLE. We did not do FISH for assessment of origin of marker chromosome in our patient due to financial constraints. If the origin of the marker chromosome is from the Y chromosome, it increases the risk of gonadoblastoma and removal of the undeveloped gonads becomes necessary. The exact pathogenetic mechanisms of SLE in TS have not been clearly elucidated. Though autoimmune diseases are more associated with iXq karyotype, neither ours and nor previously reported cases were of iXq karyotype.

Here we also reported one patient of TS mosaicism with t(12;20) autosomal reciprocal translocation and to our knowledge it is the first case reported so far. Reciprocal translocations are most common structural abnormalities with incidence of 1 in 500 to 650 livebirths. These patients can have autism, epilepsy, intellectual defects and congenital abnormalities in ~ 6% cases.[20] Autosomal translocation with structural or numerical abnormalities of X chromosome are rare. The first case of TS with autosomal translocation reported was karyotype 45, X, t(1;2) (q32;q21) in 1979.[21] So far 13 cases have been reported- robertsonian translocation between chromosome 13 and 14 (5 cases-38.5%), followed by t(1;2) (2 cases- 15.4%) and one case each of autosomal translocation t(1;9), t(2;12), t(2;22), t(4;16), t(8;19) and t(15;22).[22] Our case had severe height deficit -5SDS as compared to classical 45, XO of -3.66SDS and had primary amenorrhoea, absent secondary sexual characters with no turner stigmata but neurocognitive defects were present. Balanced reciprocal translocation t (12;20) (p12.1; p12.3) has been reported to cause mild intellectual disability, speech delay, characteristic facial appearance and autistic features in a child. Therefore, short stature, primary ovarian can be attributed to TS and neurocognitive defects can be due to both TS and autosomal t(12:20) translocation in this patient.

5. Conclusion

TS is still underdiagnosed entity in our part of country and there should be low threshold for evaluation for TS in early childhood among females with one or more turner‘s stigmata and/or short stature. No significant karyotype-phenotype corelation was found in the study. More focused studies are needed to study the exact genes responsible for various manifestations in TS, pathogenic mechanisms of DM and SLE in TS and the effect of autosomal translocations in TS phenotype. We report rare association of SLE with TS and a novel karyotype involving mosaicism for X with autosomal translocation t(12,20) in TS.

6. Source of Funding

None.

7. Conflict of Interest

The authors have no conflicts of interest to declare.

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