Case report

Oral consideration with Mosaic Turner Syndrome -a case report

Abstract

Turner syndrome (TS) is a chromosomal disorder with a prevalence of approximately 1/2500 live female births. There is complete or partial absence of one of the two sex chromosomes, resulting in a genetic constellation of 45, X monosomy or 45, X/46, XX mosaic, respectively. The chief manifestations of the syndrome are peripheral edema, short stature, extra skin fold, webbing ofneck, renal and cardiovascular anomalies, sexual infantilism, learning disability etc.

We are presenting acase of 13 yr oldfemale whowas diagnosed with MosaicTurner Syndrome.Thekaryotype analysis wasconsistent with the diagnosis.

Key words

Mosaic Turner Syndrome, Chromosome, Mosaicism, karyotyping

Introduction

Turner syndromealso called XO syndrome or Bonnevie-Ulrich syndrome, a disorder of females ischaracterized by the absence of all or part of anormal second sex chromosome.[1]Turner Syndrome was first described by Otto Ulrich in 1930 and Henry Turner in 1938.[2] It occurs in 1 in 2,500 to 1 in 3,000 live-born girls. Approximately 50% of the patients with Turner syndrome have a 45X karyotype (pure form of TS). Several karyotype variations exist including short or long arm deletion, ring X, isochromosome of the long arm, and mosaicism (a combination of cell lines such as 45X/46XX).[1,3] In mosaic Turner syndrome, some cells have the normal number of

46 chromosomes, but other cells are missing one X or there are structural defects in the second X.This syndrome is characterized by sexual infantilism, webbed neck, short stature, peripheral edema, renal and cardiovascular anomalies, gonadal dysplasia, some learning disability etc.[1]

Here we present 13 yr old female with clinical feature of Mosaic Turner Syndrome

Case report

A 13 yr old girl reported our department with a chief complaint of decayed teeth in upper front and back region. Previous medical reports revealed congenital hypothyroidism, congenital renal agenesis of right side. Her family history revealed consanguineous marriage of many of her family members including her parents. None of the family members showed the features of this disease.

On general physical examination patient was of short height (118cm) with short extremities, frontal bossing, hypoplastic mandible,short neckand low posterior hair line [figure1A]. The chest appeared broad and had widely spaced nipples (both the nipples were outside midclavicular line) [figure 1B].There was protuberant abdomen [figure 1C] with slight pitting edema in dorsum of feet, cubitus valgus (forearm is angled away from the body to a greater degree than normal when fully extended) and hypoplastic nails [figure 1D]. She was uncooperative with speech problem. All vital signs were within normal limits.

On intraoral examination there was microstomia [figure2A], high arched palate, dental caries wrt11, 21, 22,23, 24, 25 [figure2B]and generalized microdontia [figure3]. Based on the clinical findings a provisional diagnosis of Turner syndrome was given.

The investigations advised were complete hemogram, complete urine analysis, renal function tests, cardiac tests,Thyroid function tests, ultrasound of abdomen including gonads, karyotyping,

and skeletal age estimation. The karyotype confirmed the 45, XO (mosaic type) [figure4]. Her skeletal age was 12yrs. Thyroid function tests showed normal thyroid hormone levels (as patient was on medication). There were low levels of serum creatinine, positive anti microsomal antibodies. Renal agenesis of right side was confirmed by renogram[figure 5]. Otherblood and urine analysiswere normal. On ultrasound of abdomen right kidney was not visible. Cardiac examinations were normal and ultrasoundof gonadsshowed gonadal agenesis. The provisional diagnosis was confirmed by investigations. Patient was given dental treatment and referred to endocrinologist for further follow up.

Discussion

Turner Syndrome is a disorder affecting girls with abnormalities of the X chromosome and associated phenotypic features such as short stature, congenital lymphedema and ovarian failure.[4] The frequency among live-born females is 1 in 2500 to 1 in 3000. [1, 3] approximately98-99% of TS fetuses are spontaneously aborted before 28th week ofgestation. About 20% of all spontaneously aborted fetuses have TS. [5, 14]

Approximately half have monosomy X (45,X), and 5 to 10 percent have a duplication (isochromosome) of the long arm of one X (46,X,i(Xq).[3]Most of the rest have mosaicism for 45X, with one or more additional cell lineages. More than one half of patients with the condition will have a missing X chromosome (45,X) in all cells studied or a combination of monosomy X and normal cells (45,X/46,XX; mosaic Turner syndrome).A mosaic result does not necessarily predict severity because karyotyping only investigates lymphocytes, not the relevant tissues like brain, heart and ovaries.[6]

The presentation of Turner's syndrome varies at different ages.Clinical features of Turner syndrome are lack of breast development and amenorrhea, with elevated follicle stimulating hormone levels by 14 years of age, infertility,[7]congenital lymphedema,short stature, short webbed neck, low posterior hairline, epicanthal fold, ptosis, strabismus, prominent ears, congenital hearing loss, prominent thorax, hypoplastic nails[1],broad chest with widely placed nipples, cubitus valgus, scoliosis, kyphosis[4], multiple pigmented nevi[9,10], cardiac abnormalities, renal abnormities, GIT disorders and gonadal dysgenesis.[1,3]Females with TS also have significantlyhigher risk for certain diseases includinghypothyroidism, diabetes mellitus, heartdisease, osteoporosis, congenitalmalformation of some organs, neurovasculardisease, liver cirrhosis aswell as colon and rectal cancer.[11,3]

Oral manifestations include high arched and narrow palate, hypoplastic mandible, thinnerenamel, decreased amount of dentin,reduced tooth size,[8, 10]tooth mobility and periodontal pocket,[1] malocclusion[10].Girls with Turner syndrome typically have normal intelligence (i.e. mean full scale IQ of 90); however, they may have difficulty with nonverbal, social, and psychomotor skills. [1]

There is no cure for Turner syndrome. However, much can be done to minimize theSymptoms by growth hormonereplacement, either alone or with a low dose of androgen, to increase growth and estrogen replacement therapy such as the birth control pill, to promote development of secondary sexual characteristics. [14]

Conclusion

Turner syndrome is a chromosomal disorder affecting females with a frequency of 1 in 2,500 live births. While most of the physical findings are harmless, there can be significant medical

problems associated with the syndrome. Early detection, specialized counseling methods, management of symptoms and complications, and regular follow up may help reduce the mortality and improve the quality of life for these patients. Dentists should become familiar with this condition to provide optimal oral health care for the affected individuals.

References

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Figures with legends

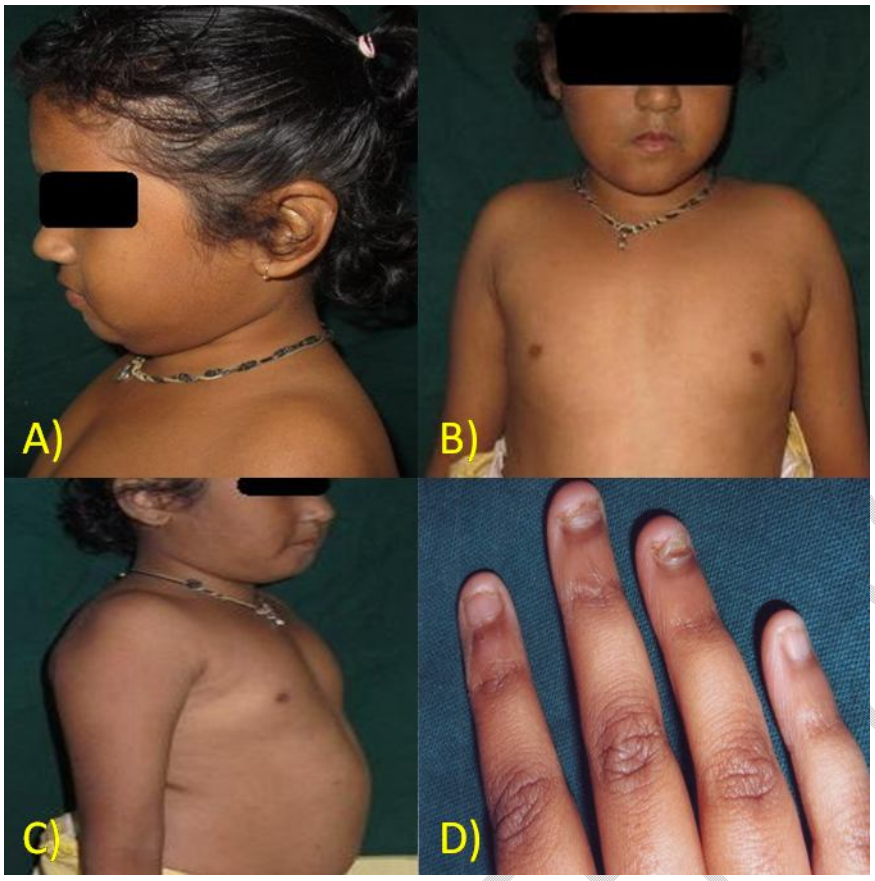

Figure1- A)Frontal bossing, Hypoplastic mandible, Short neck B) The chest appeared broad and had widely spaced nipplesC) Protuberant abdomenD) Hypoplastic nails

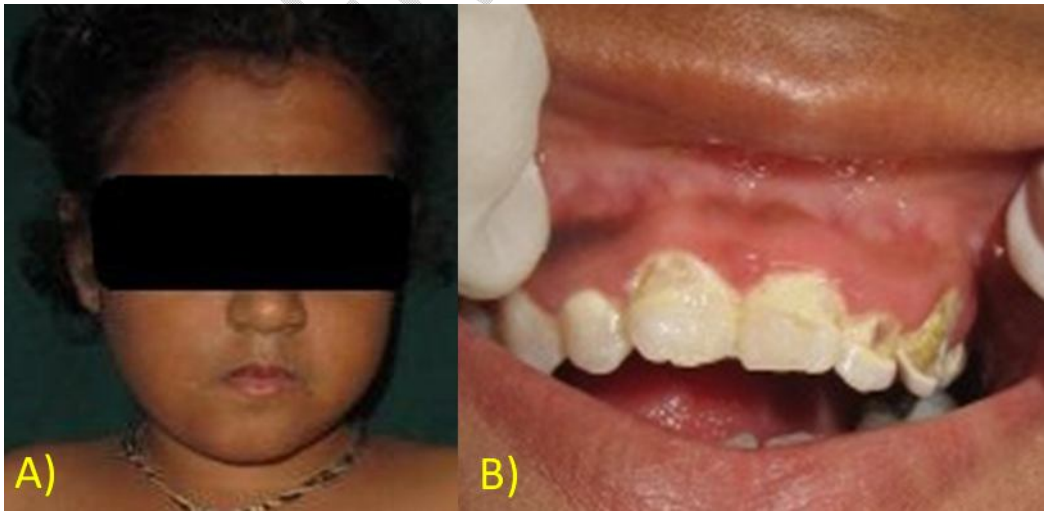

Figure 2- A) Microstomia B)Dental caries wrt11, 21, 22, 23, 24, 25

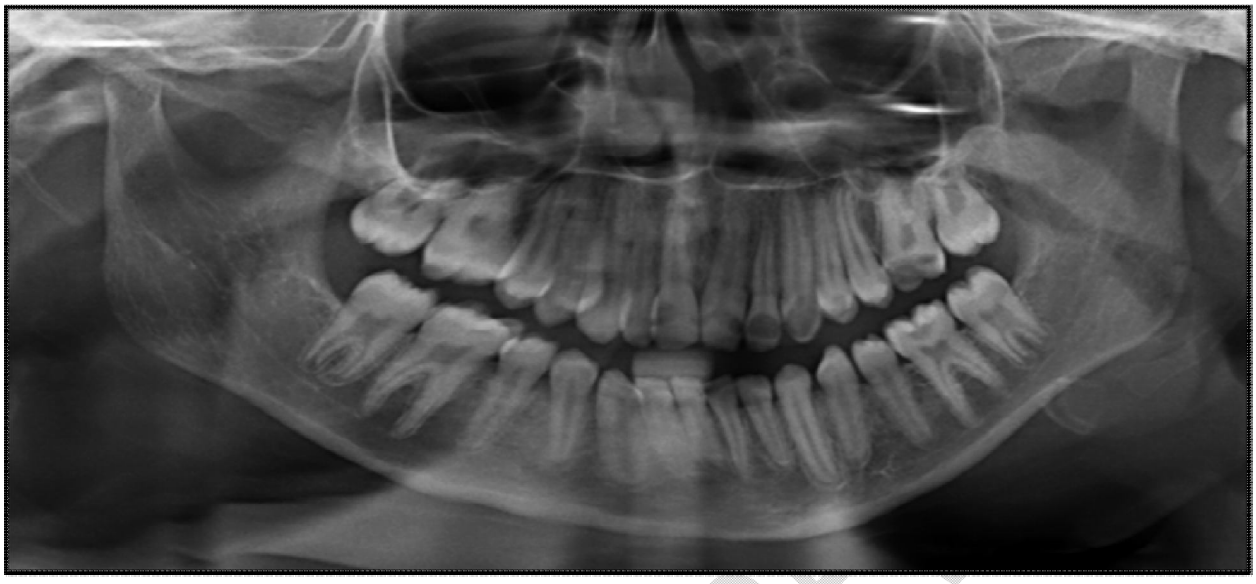

Figure 3-OPG showed generalized microdontia

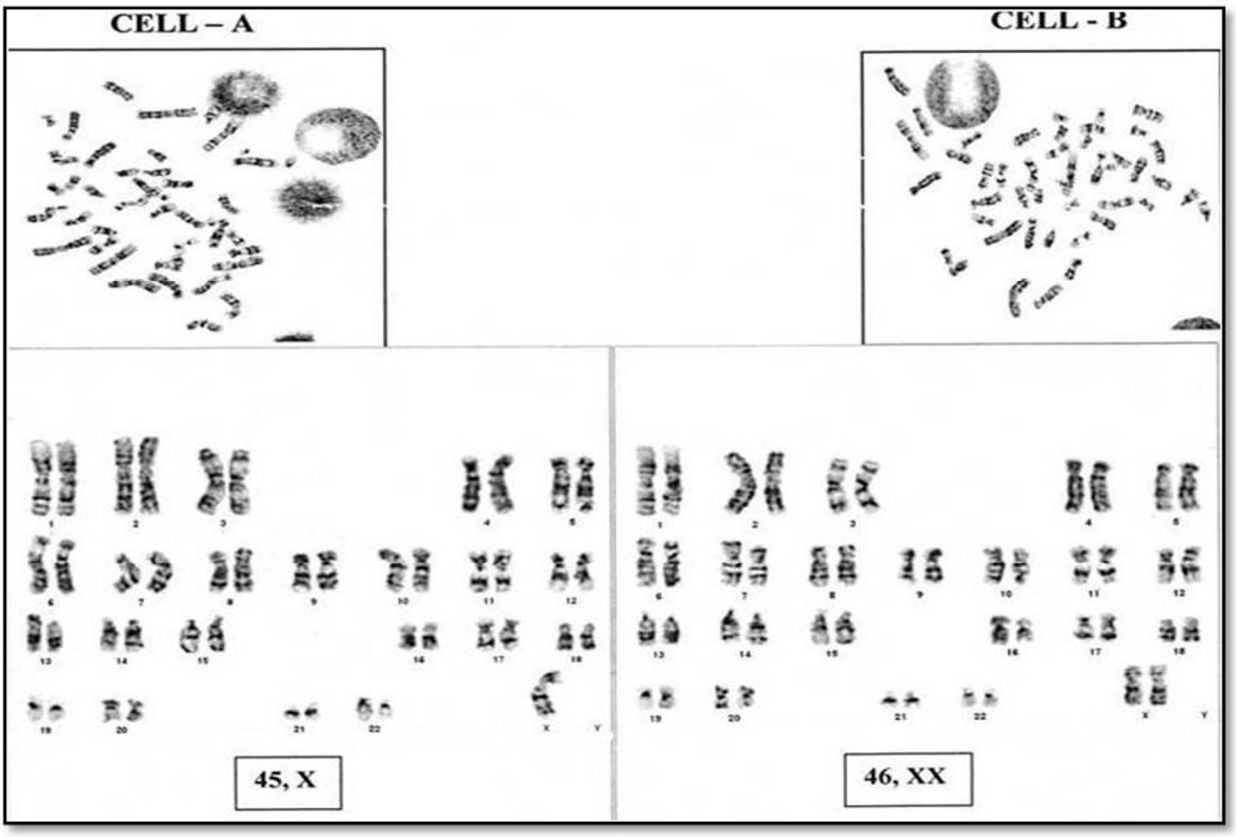

Figure 4- 45, XO Karyotype (mosaic type)

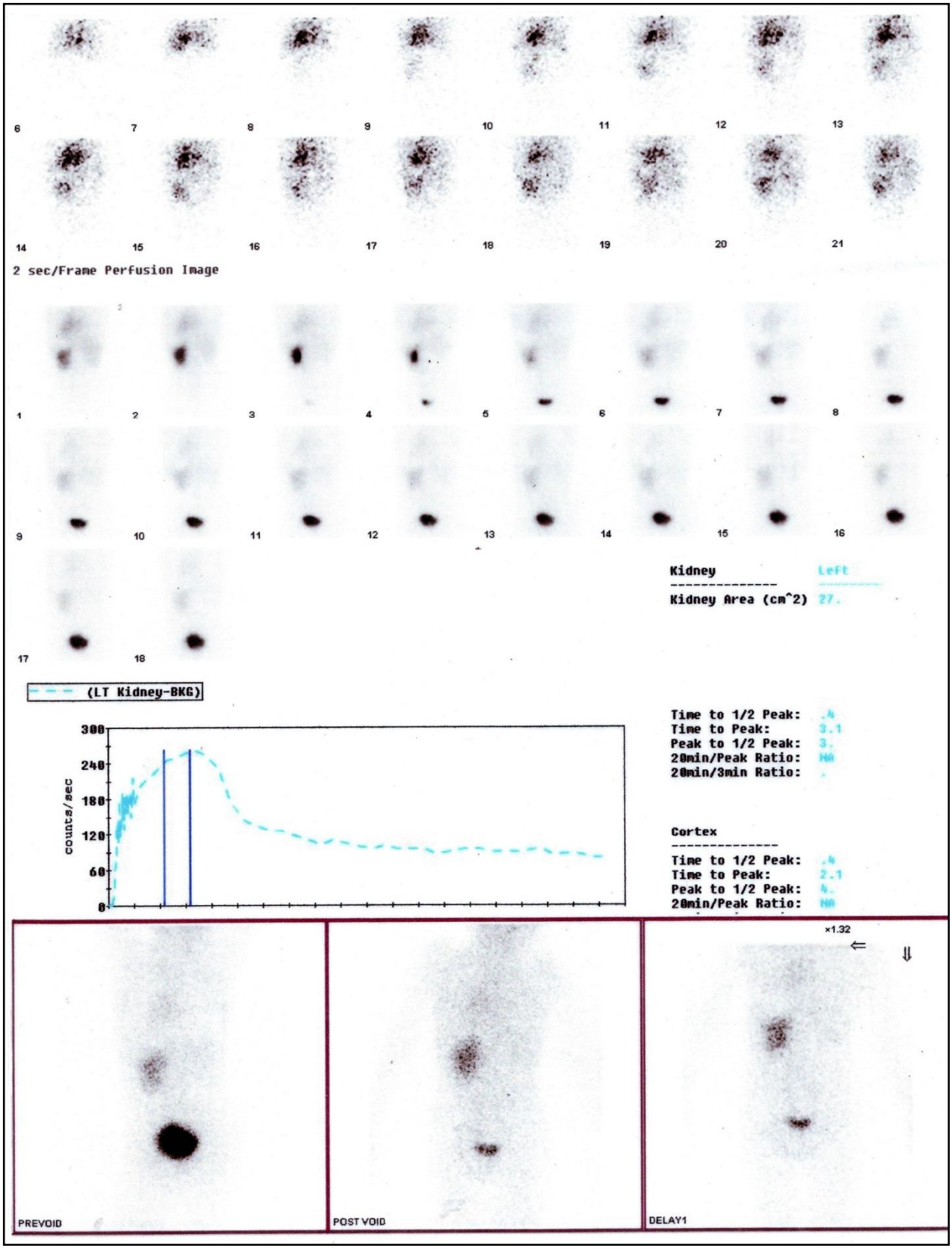

Figure 5- Renogram