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Oral Consideration with Mosaic Turner Syndrome: A Case Report

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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Case Report

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ABSTRACT

Turner Syndrome is a disorder of the X chromosome affecting girls with a prevalence of approximately 1/2500 to 1 in 3000 live female births. About 50 % of girls with Turner syndrome have monosomy X (45XO, pure form of TS). About 30% of girls with this disorder have Turner mosaic with mixed chromosome pattern (46XX/45X). The chief manifestations of the syndrome are short stature, peripheral edema, webbing of the neck, extra skin fold renal and cardiovascular anomalies, sexual infantilism, learning disability etc. Turner syndrome is a major concern for the dentist due to its various oral manifestations which include Class II facial profile, hypoplastic mandible, high arched

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and narrow palate, thinner enamel, decreased amount of dentin, reduced tooth size, tooth mobility and periodontal pocket, and malocclusion. We are presenting a case of a 13 yr old female with Mosaic Turner Syndrome, reported to our department with oral complaints.

Keywords: Mosaic turner syndrome; chromosome; mosaicism; karyotyping.

1. INTRODUCTION

Turner Syndrome also called Bonnevie-Ulrich syndrome or XO syndrome was first described by Otto Ulrich in 1930 and Henry Turner in 1938, characterized by the absence of all or part of a normal second sex chromosome in girls [1,2]. It occurs in 1 in 2,500 to 1 in 3,000 live-born girls. Most often (Approximately 50% of the patients) Turner syndrome presents with 45X karyotype (pure form of TS) whereas several karyotype variations is also exist including ring X, isochromosome of the long arm, short or long arm deletion, and mosaicism [1,3]. In mosaic Turner syndrome, cells inside the same person have different chromosome packages i.e. some cells have missing or structurally altered one X chromosome while other cells have normal number of 46 chromosomes. This syndrome is characterized by difference clinical presentations at different ages, as several physical features may be apparent at an early age or at a later age. Most common features are short stature, sexual infantilism, webbed neck, peripheral edema, cardiovascular anomalies, renal anomalies, gonadal dysplasia and some learning disability etc. [1].

Oral manifestations described in various literatures includes Class II facial profile, hypoplastic mandible, high arched and narrow palate, thinner enamel, decreased amount of dentin, reduced tooth size, tooth mobility and periodontal pocket, and malocclusion [4,5,6,7]. Here we present 13 yr old female with clinical feature of Mosaic Turner Syndrome with typical oral manifestations.

2. CASE REPORT

A 13-year-old girl reported to our department with a chief complaint of decayed teeth in the front tooth region of the upper jaw. Previous medical reports revealed congenital hypothyroidism, congenital renal agenesis of right side. Her family history revealed consanguineous marriage of her parents.

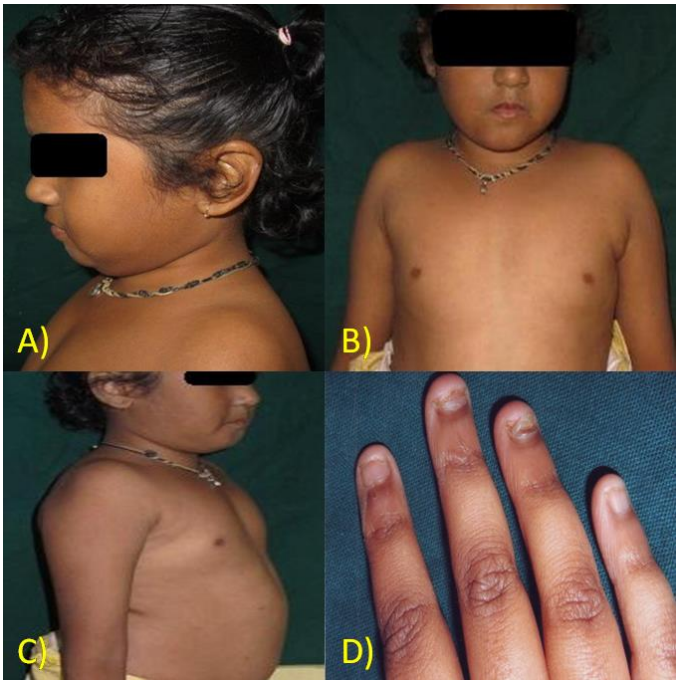

Fig. 1. A) Frontal bossing, Hypoplastic mandible, Short neck B) The chest appeared broad and had widely spaced nipples C) Protuberant abdomen D) Hypoplastic nails

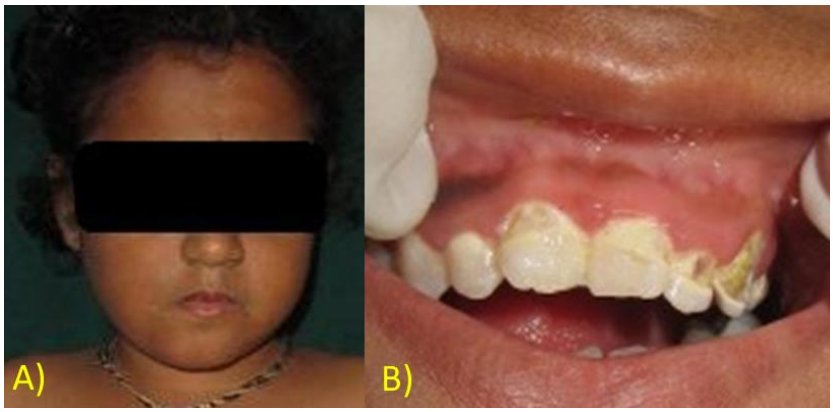

Fig. 2. A) Microstomia B) Dental caries wrt11, 21, 22, 23, 24, 25

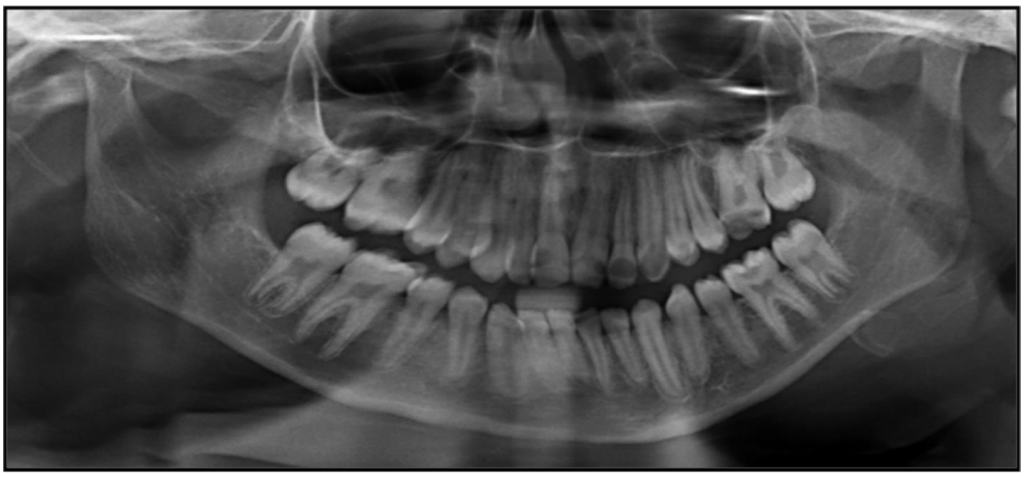

Fig. 3. OPG showed generalized microdontia

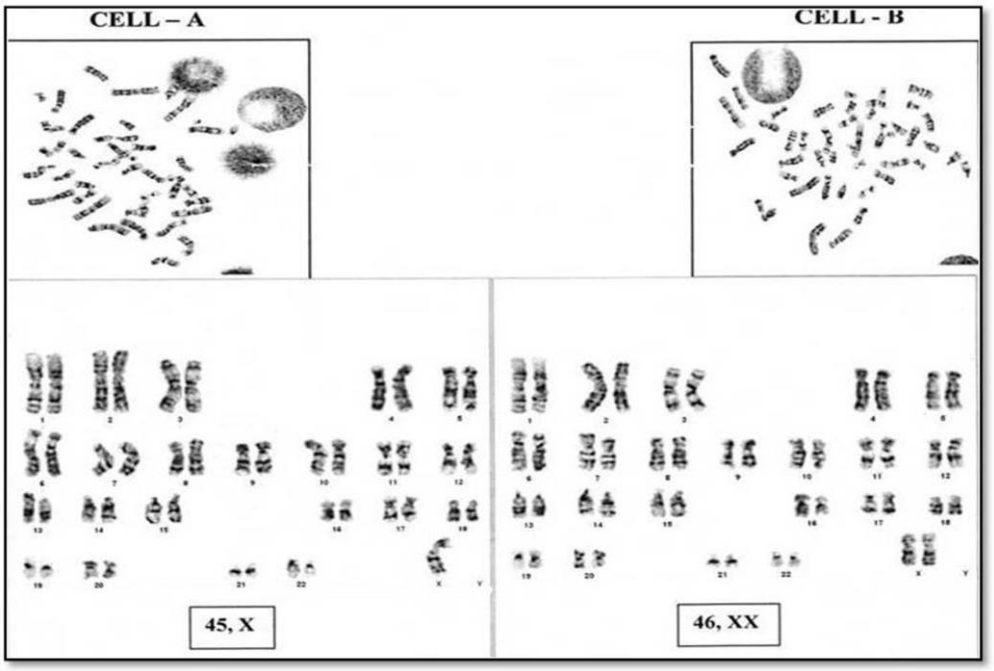

Fig. 4. 45, XO Karyotype (mosaic type)

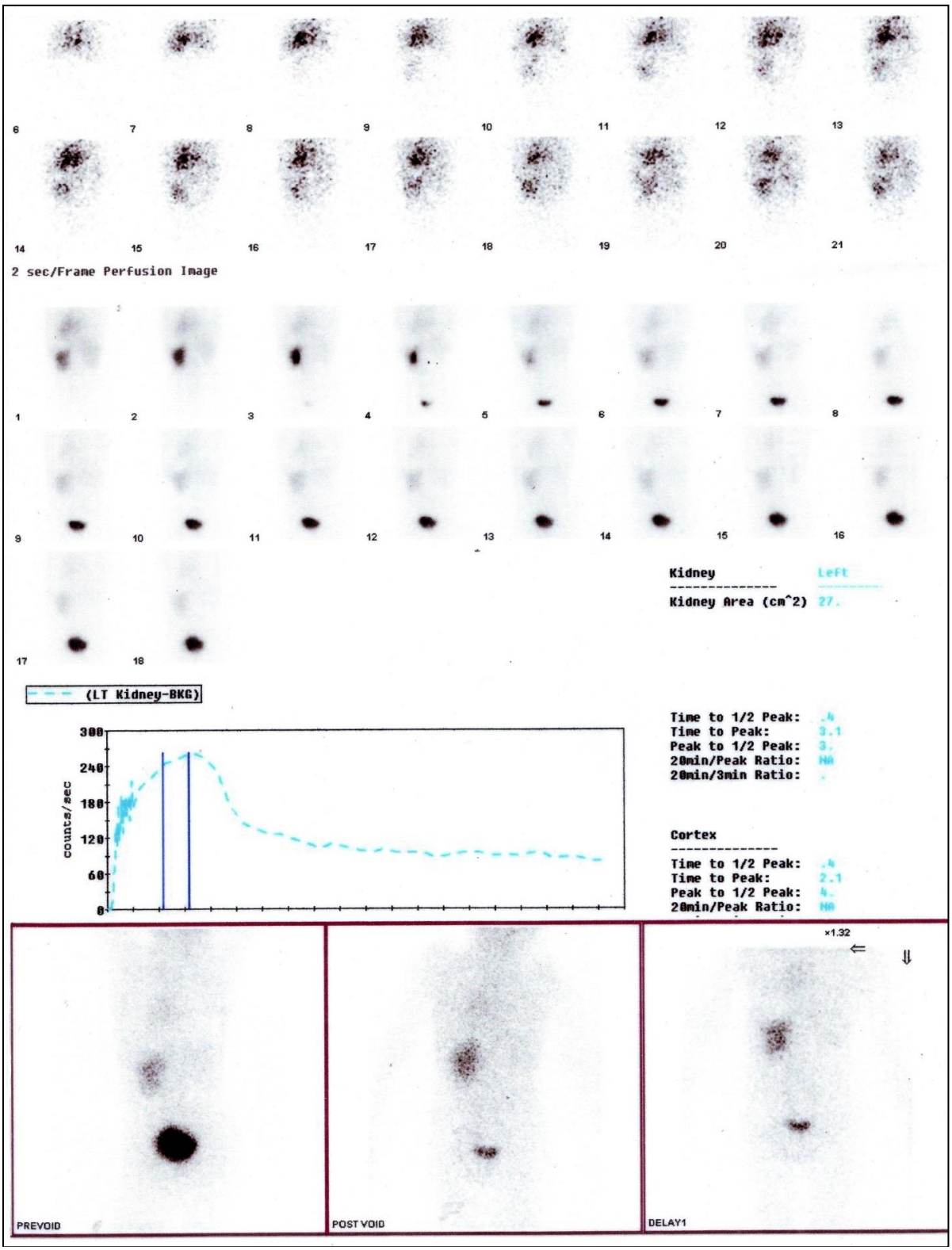

Fig. 5. Renogram

On general physical examination patient was of short height (118cm) with short extremities, frontal bossing, hypoplastic mandible, short neck and low posterior hair line [Fig. 1A]. The chest appeared broad and had widely spaced nipples (both the nipples were outside midclavicular line)

[Fig. 1B]. There was protuberant abdomen [Fig. 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 [Fig. 1D]. She was uncooperative with speech problem. All vital signs were within normal limits.

Extraorally, the patient was presented with microstomia [Fig. 2A] and hypoplastic mandible. On intraoral examination, there was high arched palate, dental caries in relation to 11, 21, 22, 23, 24, 25 [Fig. 2B] and generalized microdontia [Fig. 3]. 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, and Thyroid function tests, ultrasound of the abdomen including gonads, karyotyping, and skeletal age estimation. The karyotype confirmed “Mosaic Turner syndrome” (45X in cell A and 46XX in cell B) [Fig. 4]. Her skeletal age was 12 yrs. Thyroid function tests showed normal thyroid hormone levels (as the patient was on medication). There were low levels of serum creatinine and positive anti microsomal antibodies [8,9]. On ultrasound of the abdomen, the right kidney was not visible. Normal renal function of the left kidney was appreciated on Renogram [Fig. 5]. Other blood and urine analysis were normal. Cardiac examinations were normal and ultrasound of gonads showed gonadal agenesis. The provisional diagnosis was confirmed by investigations. The Patient was given dental treatment and referred to an endocrinologist for further follow-up.

3. DISCUSSION

Turner Syndrome is a disorder of the X chromosome affecting girls with associated phenotypic features [10]. TS fetuses are spontaneously aborted before the 28th week of gestation in approximately 98-99% of cases while the frequency among live-born females is 1 in 2500 to 1 in 3000 [1,11].

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 45 X, 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) [12].

The presentation of Turner’s syndrome varies at different ages, as several physical features may be apparent at an early age or at a later age. Signs of Turner syndrome at birth or during infancy may include congenital lymphedema, short stature, short webbed neck, low posterior hairline, epicanthal fold, Swelling of the hands and feet, especially at birth, ptosis, strabismus, prominent ears, congenital hearing loss, prominent thorax, hypoplastic nails [1], broad chest with widely placed nipples, cubitus valgus, scoliosis, kyphosis [10], multiple pigmented nevi [13], cardiac abnormalities, renal abnormalities, GIT disorders and gonadal dysgenesis [1,3].

Signs of Turner syndrome in childhood, teens, and adulthood may include short stature, ovarian insufficiency due to ovarian failure, slowed growth, No growth spurts at expected times in childhood, lack of breast development and amenorrhea with elevated follicle-stimulating hormone levels, Failure to begin sexual changes expected during puberty, Sexual development that stops during teenage years, an early end to menstrual cycles and infertility [14]. Females with TS also have significantly higher risk for certain diseases including diabetes mellitus, hypothyroidism, osteoporosis, heart disease, neurovascular disease, liver cirrhosis as well as colon and rectal cancer [3,15].

Girls with Turner syndrome may have difficulty with nonverbal, social, and psychomotor skills although TS girls have normal intelligence with a mean full scale IQ of 90 [1].

A study was conducted to analyze the oral health and craniofacial characteristics in a cohort of 15 females with TS. The subjects presented various oral manifestations like Class II facial profile evidencing insufficient mandible development. (85.71%), Class II division 1 malocclusion, with an increased overjet (42.85%), Class II division 2 malocclusion (42.15%), deep bite(35.71%), cross bite (21.42%) and open bite(14.28%), Dental crowding (57.14%), Tooth anomalies of number, size, structure, shape, position and eruption, Tooth number anomalies, including maxillary incisors agenesis and supernumerary molars (42.8%), tooth size anomalies(35.71%), Enamel dysplasia (21.42%), eruption alterations (35.7%) and premature eruption (21.42%). 42.85% presented more than four out of the seven reported TS characteristic dental signs and

symptoms (supernumerary molars, agenesis, microdontia, enamel defects, alterations in eruption patterns, including advanced and delayed for chronological age, crowding, rotations and transpositions) [4].

Other oral manifestations described in various literatures are high arched and narrow palate, thinner enamel, decreased amount of dentin, reduced tooth size, tooth mobility and periodontal pocket, and malocclusion [5,6,7]. Our case is in accordance with previous literature and presented with hypoplastic mandible high arched palate, thinner enamel, decreased amount of dentin, and reduced tooth size. Our case also presented with dental caries in a few teeth possibly due to poor oral hygiene and thinner enamel (enamel hypoplasia). This finding is also as per the study performed by Szilagyi A et al 2000 [7] where decayed teeth scores were higher than the control group and Faggella et al. [16] where caries index (DMFT) was higher.

There is no cure for Turner syndrome. However, much can be done to minimize the Symptoms by growth hormone replacement, either alone or with a low dose of androgen, to increase growth and estrogen replacement therapy such as the birth control pill, to promote the development of secondary sexual characteristics [17,18].

4. CONCLUSION

Oral manifestations are not uncommon with turner syndrome, thus oral physicians should become familiar with this condition and greater emphasis should be placed on early diagnosis and treatment. Also 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.

CONSENT

As per international standards, parental written consent has been collected and preserved by the author(s).

ETHICAL APPROVAL

It is not applicable.

DISCLAIMER (ARTIFICIAL INTELLIGENCE)

Author(s) hereby declare that NO generative AI technologies such as Large Language Models (ChatGPT, COPILOT, etc.) and text-to-image generators have been used during the writing or editing of this manuscript.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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