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Turner Syndrome

Turner Sendromu

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ABSTRACT

Turner syndrome is a genetic disorder that affects mostly females. Affected females have characteristic features such as short stature, premature ovarian failure, and several other features. Oral manifestations of this condition are not much discussed in the literature. But reported literature includes teeth, palate, periodontal and salivary changes. So the aim of this review is to illustrate the general manifestations, and especially the oral manifestations of Turner syndrome and evaluate their possible management.

Key Words: Turner syndrome, Genetic disorder, X-chromosome, Tooth, Growth

ÖZET

Turner sendromu daha çok kadınları etkileyenbir genetik bozukluktur. Etkilenen kadınların başlıca karakteristik özellikleri kısa boy, premature over yetmezliği, ve diğer bazı özelliklerdir .Literatürde bu durumun oral özellikleri çok fazla tartışılmamıştır. Bununla birlikte literatürde bu hastalarda diş, damak, periodontal ve salyada değişiklikler olabileceği bildirilmiştir. Bu gözden geçirmede, Turner sendromunun genel ve oral (ağızla ilgili) belirtilerinin vurgulanarak, bunların yönetinin değerlendirilmesi amaçlanmıştır.

Anahtar Kelimeler: Turner sendromu, genetic bozukluk, X kromozomu, Diş, Gelişim

Turner syndrome (TS) is a common genetic disorder that has been classically associated with a 45X karyotype,although several other X-chromosomal abnormalities have been identified in these patients, many of which involve mosaicism. It occurs in one out of every 2500 to3000 female births and is associated with a broad array of potential abnormalities, most of them thought to becaused by haploinsufficiency of

genes that are normally expressed by both X chromosomes.[1]

The most common features of TS are short stature and premature ovarian failure. The short stature is mainly due to SHOX haploinsufficiency and is characterized by a shortfall of approximately 20 cm from the predicted adult height. About two thirds of girls and women with TS have a high-arched or ogival palate. Other skeletal features include micrognathia and scoliosis. Evidence of fetal lymphedema is manifest as neck webbing, malrotation of the ears and a low posterior hairline.[2]

Oral abnormalities reported in the literature are few and not clustered, so this review portrays the various oral abnormalities reported in the text under a single roof.

Psychological disorders

Social immaturity is often described, as are difficulties with social relations, particularly during adolescence when individuals with TS tend to be maturationally “out of synch” with their peers. Most individuals with TS have very few friends and often experience significant teasing, including bullying. Due to their particular set of physical stigmata, females with TS generally have a poor body image and low self-esteem. Many individuals with TS demonstrate a high degree of dependence and typically live at home with parents, although a number do marry and have (adopted) families. Incidence of a major depressive disorder and a history of suicidal attempts are relatively low in this population, these are still higher than in the normal population.[4]

Cardiovascular disorders

Hypertension is common and should be treated vigorously, since it contributes to aortic dissection risk. Cardiovascular complications are the major contributor to premature death in Turner syndrome. Echocardiograms or other imaging (MR angiography) should be performed 5 yearly to monitor the aortic root, especially if assisted pregnancy is contemplated, when aortic dissection risk increases. Since Turner women are shorter than average, aortic root diameter norms are inappropriate, and a proposed Turner-specific nomogram has been developed. Oestrogen replacement reduces the risk of atherosclerosis.[5]

Autoimmune disorders

Females with TS have an increased prevalence of autoantibodies and are at increased risk of developing autoimmune diseases such as: Hashimoto thyroiditis,

pernicious anemia, Addison disease, celiac disease, inflammatory bowel disease, diabetes, autoimmune hepatitis, autoimmune colitis, thrombocytopenia, and juvenile rheumatoid arthritis.[6]

Orofacial manifestations

Facial:

Epicanthic folds, ptosis of the eyelids,prominent ears, and micrognathia are common facial features.[7]

Eyes:

Visual abnormalities, particularly strabismus, are found in approximately 20%of patients.[7]

Ears:

Chronic suppurative otitis with resultant hearing loss occurs in some cases.[7]

Neck:

In infants, excess skin on the nape of the neck is common. During embryonic life, neck blebs or cystic hygromas are common. With age, the excess skin on the neck metamorphoses into pterygium colli. The posterior hairline is low.[7]

Palate:

The palate is highly arched in approximately 35% and cleft palate may occur with a somewhat higher than normal frequency.[7]

Tooth:

Several abnormalities in tooth development and morphology have been described in Turner syndrome, including reduced tooth size, thinner enamel, and less dentine, theclinical significance of which is unknown. Girls with TS are at greater risk for root resorption, which can lead to tooth loss, especially during orthodontic treatment.[8] The teeth may erupt prematurely, the first permanent molars appearing between 1.5 and 4 years of age. There is increased molarization of premolars. Cusp height is reduced as is crown size.

Jaw:

The cranial base is short, so the face is retrognathic.[4] In addition, micrognathia can cause crowding of the lower teeth.[8]

Periodontal status:

Increased Plaque and gingival indices[9]

Tori in the mandible which manifest as lump in the gingiva.[10]

Table 1. General Features of Turner Syndrome [3]

Common physical features[3]	**Associated conditions**[3]
Short stature (reduced final height averaging 143 cm [56 inches])	Tendency toward obesity
Gonadal failure	Recurrent otitis media
Cubitus valgus (>10(carrying angle between the extended supinated forearm to the upper arm)	Hearing loss
Low posterior hairline	Renal abnormalities
Multiple pigmented nevi	Left-sided cardiac anomalies; aortic coarctation
Webbed neck	Strabismus
Lymphedema of hands and feet	Hypertension
Nail dysplasia	Hypothyroidism
Deep-set hyperconvex nails	Glucose intolerance
Short fourth metacarpal	Hyperlipidemia
Unusual shape and rotation of ears	Kyphoscoliosis
Broad shieldlike chest with inverted or hypoplastic nipples	Lordosis
Tibialexostosis	Osteoporosis
	Gonadoblastoma Inflammatory bowel disease
	Colon cancer
	Neuroblastoma
	Juvenile rheumatoid arthritis
	Liver disease

Alveolar arches:

The alveolar arch of the maxilla was narrower and of normal length, but the mandibular arch was shorter and broader.[9]

Saliva and Turner Syndrome:

Several salivary changes reported in Turner syndrome that includes[11]

1. Decreased salivary flow rate
2. Higher concentration of ionic calcium
3. Higher concentration of total protein and inorganic phosphates

4. Higher concentration of IgA, lactoferrin and lysozyme in the saliva

Management

Turner syndrome is a lifelong condition. Most people live long and healthy lives, yet some are susceptible to numerous chronic conditions. Health supervision involves careful medical follow-up care, which includes screening for commonly associated chronic diseases. Early preventive care and treatment are also essential.[12]

Recombinant human growth hormone replacement therapy with estrogen and psychosocial support are the treatment modalities.[13]

Estrogen replacement therapy is usually required, but starting too early or using doses that are too high can compromise adult height. Estrogen is usually started at age 12-15 years. Treatment can be started with continuous low-dose estrogens at 12 years, or as early as 5 years. These can be cycled in 3-weeks on, 1-week off regimen after 6-18 months; progestin can be added later. Some authors believe that conjugated estrogens are contraindicated in pediatric patients.Transdermal estrogens are associated with physiologic estrogen levels, and may be preferred treatment, if tolerated.[12]

Cardiac and hearing assessments need to be regularly made for girls with Turner syndrome, approximately every two or three years. The webbing of neck can be surgically corrected by a plastic surgeon. Scarring can occur due to surgery. The decision to operate needs to becarefully considered.[14]

At around the time of puberty, the growth hormone treatmentis stopped and hence height development also stops. Female sex hormones are introduced over a period of time and breast and sexual hair development occur. Menstruation will occur but this is not an indication of fertility.[14]

Most women with Turner syndrome who have had children have a mosaic Turner pattern in their chromosomes ie. they have two types of cells including some normal 46,XX cells. Other women with Turner syndrome have achieved apregnancy using assisted reproductive technologies (ART) such as*in vitro* fertilisation (IVF).[14]

There are special intervention programs for children with Turner syndrome, with a focus on odontology. Tooth age determination is recommended as part of the diagnostic work-up. A specialist in pediatric dentistry or orthodontia may be a useful resource, and should also be consulted if treatment with growth hormone is being considered, and between the ages of 7 and 9 in order to identify dental aberrations or

malocclusions and to plan any necessary orthodontic treatment to correct bite problems.[15]

- Children with eating disorders often required extra dental care, including assistance with oral hygiene and fluoride treatments.[15]
- Individuals with cardiac defects may require prophylactic antibiotics when oral interventions associated with bleeding are undertaken.[15]
- Training in oral motor skills and extra stimulation may be relevant.[15]
- Eating and swallowing problems will need to be investigated and treated by hospital specialist teams.[15]

A patient with Turner's syndrome receiving systemic androgen therapy who experienced marked gingival enlargement, bleeding, and discomfort. The patient was treated with full mouth gingivectomy and 2-week recall visits for a period of 2 months consisting of oral physiotherapy instructions, scaling, and topical fluoride application. She was then placed on 1-month recall for the next 4 months. This mode of therapy was effective in treating severe gingival hyperplasia in a patient receiving systemic androgen therapy.[16]

Life expectancy

Patients with Turner’s syndrome appear to have a decreased life expectancy, primarily as a result of complications of heart disease and diabetes.[17]

Turner syndrome is a disorder that can be diagnosed with its precise features. Oral hygiene maintenance is essential to have a good systemic health. It’s the role of dentist to counsel the patients and their parents for proper oral hygiene. Cardiac and diabetic patients have further compromised oral health so they require proper prophylactic antibiotic measures. In conclusion, a multispecialty approach is crucial for the management of such patients.

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