

CASE REPORT

Interdisciplinarity in oro-maxillofacial dysmorphism rehabilitation of a patient with Turner syndrome. A clinical case report

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Abstract

Statement of the problem: Turner syndrome is a chromosomal disorder that manifests with short stature, gonadal dysfunction, hypothyroidism, congenital heart disease, and distinct craniofacial features including oro-maxillofacial dysmorphism. This paper presents a case of a 30-year-old female patient with Turner syndrome who sought dental care to improve the dental and facial morphology and restore the oral health. Detailed exobuccal examination revealed complex anomalies. Initial periodontal therapy and carious lesions treatment was followed by orthodontic treatment and prosthetic rehabilitation with two porcelain fused to metal fixed partial dental prostheses. Tooth whitening and restoration of the incisal edge of the anterior teeth was performed to complete the smile design improvement. The interdisciplinary approach pursued in this case resulted in successful oral rehabilitation with optimal treatment outcomes and high patient's satisfaction. **Clinical significance:** Patients with Turner syndrome may present with complex morphologic oro-maxillofacial alterations that require individualized dental treatment plans. Appropriate interdisciplinary medical/dental treatment can lead to successful oral rehabilitation with a minimum of invasive procedures.

Keywords: Turner syndrome, orthodontic treatment, prosthetic rehabilitation, dental and facial morphology.

Introduction

Turner syndrome 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 to 3000 female births and is associated with a broad array of potential abnormalities, most of them thought to be caused by haploinsufficiency of genes that are normally expressed by both X chromosomes. Major clinical manifestations include: growth failure, congenital heart disease (e.g., coarctation of aorta, bicuspid aortic valve, atrial and ventricular septal defects), gonadal failure, and learning disabilities [1–3]. Other co-morbidities such as hypothyroidism, diabetes mellitus, deafness, premature osteoporosis, are associated with complex psychosexual factors influenced by hormonal deficiencies and by the disturbed body image due to abnormal physical development [4, 5].

In Turner syndrome (TS) are associated a number of autoimmune manifestations that require regular screening

for an early diagnosis and a timely therapeutic approach [6].

The skeletal characteristics of Turner syndrome are decreased maxillary growth with midface hypoplasia and a wide, micrognathic mandible [7].

Distal molar occlusion (60%), narrow upper arch and wide lower arch with concomitant lateral crossbite (39%) and anterior open bite (17%) are often observed in TS [8–10]. The teeth are smaller than normal [10, 11], because of reduced enamel thickness [11] with the locus responsible for the reduction of the tooth crown size being reported to be situated on the short arm of the X chromosome [12].

The development of the craniofacial structures and the somatic and dental morphology unique to Turner syndrome significantly influence the orthodontic and prosthetic restorative treatment.

Patient, Methods and Results

A 30-year-old female patient with Turner syndrome was referred in April 2008 to the Department of

Prosthodontics, Faculty of Dentistry, “Victor Babeș” University of Medicine and Pharmacy, Timișoara, Romania, for oral rehabilitation.

A detailed medical history revealed that the she was diagnosed with Turner syndrome (45X karyotype) late, at the age of 16 years. Therefore, no growth hormone therapy was initiated and patient was treated with estrogen replacement therapy.

Pertinent general clinical examination findings revealed a short stature (146 cm height), BMI 237 kg/m², and high blood pressure (135/90 mmHg).

At the initial consultation, the patient presented a deviation in craniofacial morphology, a Class II malocclusion, the teeth are smaller than normal in the presence of periodontal disease, diastemas, multiple extractions, carious lesions and a Kennedy Class III. Diagnostic data collection included clinical examination, model casts analysis, cephalometric measurements, panoramic radiograph (Figure 1), and photographic documentation (Figure 2).



Figure 1 – Preoperative panoramic radiograph.



Figure 2 – Lateral view of the patient's dentition in maximum intercuspal position.

Extraoral examination indicated relative facial symmetry, convex facial profile, asymptomatic mastication muscles and no sign of temporomandibular joint disorder. Endo-oral examination indicated a Class II malocclusion with narrow upper arch and wide lower arch, bilateral crossbite and anterior open bite. The patient's maxillary dental midline was inclined and deviated 1.5 mm to the right, tremas and diastemas caused by a low and anterior position of the tongue, high and narrow palatal vaults are also present.

Cephalometric measurement was evaluate and demonstrated a hyperdivergent facial skeleton (<ML-SNL, <PL-SNL), cranial base angle was increased (<N-S-Ba), shortened clivus (S-Ba) and the mandible was more retroghathic (<SNB) than maxilla (<SNA). A reduced posterior upper face height (S-Art) has been observed. The length of the body of the mandible (Go-Pog) and the total length of mandible (Art-Pog) were shortened.

A detailed cardiology evaluation including echocardiography and electrocardiogram was performed that revealed high blood pressure and the presence of small aortic root diameter. Following the cardiologist's instructions, medication for hypertension was initiated with (ADD agents) and a minimally invasive dental treatment plan was established.

Prior to beginning the treatment, approval from the Ethical Committee of the “Victor Babeș” University of Medicine and Pharmacy was obtained. The patient gave written consent for full-face photography (Figure 3).



Figure 3 – Full-face view of the patient at presentation.

The shade of the maxillary central incisor was determined as 3M2 using the shade guide Vitapan 3D Master (Vident, Brea, CA, USA) (Figure 4).

After initial periodontal therapy and extraction of root 16 with antibiotic prophylaxis, treatment of carious lesions and endodontic treatment for teeth 21, 35 were performed. The orthodontic treatment consisted of maxillary and mandibular Roth .022 fixed appliances (Figure 5) and straightwire technique.

The orthodontic treatment followed normal phases: alignment and leveling (accomplished with nickel titanium round and rectangular wires), space closure (with stainless steel SS rectangular wires) and finishing (SS wires) (Figure 6).

The duration of the active orthodontic treatment was 18 months, followed by retention with vacuum formed retainers, and after that, the patient was referred for prosthodontic rehabilitation. Implants were considered as a first option, but due to general health status and financial reasons, the chosen treatment plan called for two porcelain-fused-to-metal fixed partial dental prostheses, one at the maxilla (13–17), and one at the mandibula (44–46) (Figure 7).

After the cementation of the fixed partial dental prostheses, it was decided to bleach the anterior teeth 11, 12, 21, 22 with carbamide peroxide bleaching gel (Opalescence Boost, Ultradent, USA). Two weeks after bleaching, the incisal edges of the maxillary central incisors were restored with composite resin (Gradiac Direct, GC America), with the goal of improving the smile design of the patient (Figure 8).

Three years after beginning the treatment, the enhancements of dentofacial aesthetics accomplished through the oral rehabilitation resulted in a marked increase in patient's satisfaction and self-esteem (Figure 9).



Figure 4 – The shade of the maxillary central incisor determined with Vitapan 3D Master Shade Guide.



Figure 5 – The orthodontic treatment consisted of maxillary and mandibular fixed appliances with Roth .022 bands on the molars: frontal view.



Figure 6 – Lateral view during the orthodontic treatment.



Figure 7 – Occlusal view of the mandibular treatment after cementation of the two fixed partial dental prostheses.

Figure 8 – The smile of the patient after bleaching and restoring the incisal edge of the superior central incisors.



Figure 9 – Postoperative view of the patient's dental and facial harmony three years after beginning the treatment.

Discussion

Turner syndrome is a rare genetic disorder, affecting approximately one in 2500 live-born female, due to total or partial absence of the X chromosome. The phenotype is highly variable with slight or even normal phenotype [8]. In monosomy X (45,X), the X chromosome is of maternal origin in 68–80% of women, and it is of paternal origin in 20–32%, indicating that paternal sex chromosome loss is the most common cause of this condition [13].

Women with monosomy X generally have the most severe phenotype, the isochromosome X anomaly is more frequently associated with auto-immunity and the ring X karyotype is also associated with psychological and learning difficulties [14].

The case presented in this paper was diagnosed at the age of 16 years with Turner syndrome (karyotype

45,X) and was treated only with estrogen replacement therapy.

Typical clinical features are short stature and premature ovarian failure and less constantly phenotypic particularities such as congenital malformations, acquired cardiovascular, otological (hearing impairment), autoimmune and metabolic pathologies [15]. Epidemiological studies have revealed a 3-fold higher mortality in the Turner syndrome population than the general population, with circulatory disease (ischemic heart disease, from cerebro-vascular disease or aortic aneurysms) accounting for 41% of the excess mortality [5].

Gonadal failure and short stature occur in most individuals with Turner syndrome and are associated with complex psychosexual factors influenced by hormonal deficiencies and by the disturbed body image due to abnormal physical development [16].

Other major contributing risk factor for cardiovascular events is hypertension, which affects up to 25% of adolescents and 40–60% of adults with Turner syndrome. In some cases, the elevated blood pressure is due to narrowing of the aorta or a kidney abnormality [3]. This should be monitored during routine health maintenance visits and treated if necessary. Early diagnosis and regular screening for potentials associated complications are essential in the medical follow-up of the patients with Turner syndrome. Our patient was treated with medication for hypertension (ADD agents) and following the cardiologist instructions minimally invasive dental treatment plan was elaborated.

Females with Turner's syndrome with numerical and/or structural aberration of the X chromosome, present also with deficits in the oro-maxillofacial somatic development.

The deviations in craniofacial morphology of Turner syndrome females are largely influenced by changes in the cranial base structure. These changes have been suggested to arise from retardation in cartilage growth during early development [17].

The abnormal form of the cranial base can be observed in TS fetuses already at the time of ossification [18].

Distinct craniofacial features include micrognathia, high arched palate and malocclusion. Some studies

reported that a shorter posterior cranial base length (sella-basion) and a reduced mandibular prognathism angle (sella-nasion-supramentale) are two variables that strongly predict Turner syndrome [7].

Dentofacial analysis of the patient presented in this paper reveal all this data and the low rest position of the tongue increases the relative pressure of the cheeks on the upper dental arch, which leads to its narrowing. On the other hand, it is known that the sex chromosomes influence the palatal width.

Other studies [19] confirm that numeric aberration of the X chromosome most likely affects the quantitative and qualitative excretion of amelogenin so that teeth often present enamel defects (reduced crown size and enamel hypoplasia).

High caries index values highlight the demand of early preventive measures mostly focused on special care patients [19].

One investigation performed by Midtbø M and Halse HA demonstrates that X-chromosome deficiency influences root formation [20].

In our case, no root morphology modifications were present, but we can reveal high carious index and the presence of periodontal disease.

Developmental anomalies requiring orthodontic intervention are more frequent and severe in patients with Turner syndrome. The rate and the timing of growth, the development of the craniofacial structures, and the somatic and dental morphology unique to Turner syndrome significantly influence orthodontic treatment. Some authors have presented case reports of patients with Turner syndrome who were treated with orthodontic therapy and orthognathic surgery [21, 22].

In most cases, the complexity of the condition requires an interdisciplinary approach for optimal treatment outcome [23].

This paper presents a case of a 30-year-old female patient with Turner syndrome who is in need of oral rehabilitation.

The complexity of the case requires interdisciplinary approach: orthodontic, periodontic and prosthodontic, treatment following the instructions of the cardiologist for minimally invasive treatment.

The sequences of the treatment are described and illustrated and the patient was recall every two months during the three-year period for the follow up care.

Three years after beginning the treatment, the results of oral rehabilitation and dental and facial morphology improvement of the patient contribute in raising her self-esteem.

✉ Conclusions

In spite of physical and psychological factors and other problems that can occur in Turner syndrome, with appropriate medical care, interdisciplinary collaboration and ongoing support, a person with Turner syndrome can lead a normal and healthy life. This case reveals a consistent collaboration between the prosthodontist,

orthodontist and cardiologist that leads to a minimally invasive oral rehabilitation for a 30-year-old female patient with Turner syndrome.

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