Iranian Red Crescent Medical Journal

CASE REPORT

Oral Manifestations of Ehlers-Danlos Syndrome and Presentation of a Case

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Abstract

Ehlers-Danlos syndrome (EDS) is a rare syndrome, primarily diagnosed by clinical findings and family history. The clinical manifestations of EDS in the orofacial region consist of extra-oral and intra-oral manifestations. Its prognosis depends on the (sub) type and the proper, early diagnosis of the syndrome. The purpose of the present paper is to discuss the manifestations of this syndrome, especially its oral manifestations, and to present a case.

Keywords: Ehlers-Danlos syndrome; Oral manifestations

Introduction

Ehlers-Danlos syndrome (EDS) is a rare disorder with an incidence of 1:10,000[1] to 1:200,000 of live births. The syndrome was described for the first time in 1982 by Job Van Meerkeran and then elaborated in 1901 by Ehlers and in 1908 by Danlos.[2] The syndrome is a hereditary collagen disease; its early manifestations are skin and joint involvement.[3] Other terms such as "elastic man" or "Indian rubber man" have also been used. A large number of articles have presented the cutaneous and articular manifestations of the syndrome but there are only a limited number of sources dealing with the oral manifestations of the syndrome.[4-6]

Ehlers-Danlos syndrome is primarily diagnosed by its clinical findings and family history. EDS is genetically transmitted as autosomal dominant and only four types of it (Types IV, VI, VIII and X) are confirmed by biochemical and molecular tests.[7] Differential diagnosis should include Marfan syndrome, generalized familial articular hypermobility syndrome, cutislaxa pseudoxanthoma elasticum, and larsen's syndrome.[8]

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Received: September 10, 2008 Accepted: January 11, 2009

The classic manifestatins of EDS are articular hypermobility, cutaneous hyperelasticity, presence of dystrophic scars, and susceptibility to bleeding, bruising, hematomas and ecchymosis.[8] There are at least 15 subtypes for this syndrome[1,9] eight of which are more prevalent than others.[10] Table 1 summarizes the manifestations of some EDS subtypes.[8] The clinical manifestations of EDS in the orofacial region consist of extra-oral and intra-oral manifestations.

Extra-oral manifestations consist of scars on the chin and forehead, repeated dislocations of TMJ, epicanthus (a vertical fold of the skin on either side of the nose, sometimes covering the inner canthus), strabismus, narrow nasal bridge, shaggy hair, and skin hyperelasticity.[8] Moreover, intra-oral manifestations comprise highly fragile mucosa (similar to the patients' skin), which is easily ruptured when dental instruments touch them and sutures do not remain in place,[11] usually bruising being evident in the oral mucosa.[10]

Periodontal tissue injuries are usually observed subsequent to prophylactic procedures and periodontal surgeries or tooth extraction. Bleeding can be severe and may not be controlled during surgeries.[8] Early-onset generalized periodentitis is a common manifestation in this syndrome[4,12] which can lead to the loss of primary and permanent teeth.[11,13] The tongue is very soft and almost 50% of the individuals with this syndrome can touch the tip of their nose

Table 1: Clinical manifestations of some subtypes of EDS

(sub) type I	Cutaneous hyper elasticity ; bony protuberance of the chin, knees and ankles; delay in skin regeneration and repair; pigmented and atrophic scars on the skin; subcutaneous hematomas; occasional prolapse of mitral valve; articular hyper mobility; sub cutanceous nodules.
(sub) type II	Similar to (sub) type I but less severe; scars are less common and bleeding tendencies are less prevalent; nodules are not common but articular hyper mobility is similar to (sub) type I.
(sub) type III	The afflicted individuals have long and slender extremities similar to what is seen in Marfan syndrome; articular hyper mobility and cutaneous hyperelasticity; rare cases of bruising and dystrophic scars.
(sub) type IV	Characteristic vascular involvement; hypertelorism ; narrow nasal bridge;ear atrophy; shaggy hair,aneurysms in great veins; rupturing of internal organs are common; mitral valve may be involved , only small joints of the hands are involved.
(sub) type V	Only females are carriers of this rare (sub)type. Transmission is dependent on X-chromosome.
(sub) type VI	Similar to (sub) type I except for ocular involvement; severe scoliosis and vascular ruptures are common.
(sub) type VII	Articular hyper mobility is a characteristic feature; short stature and bilateral dislocation of hip joints are common.
(sub) type VIII	Early-onset periodontitis is a characteristic feature; escharatic /scar points on the skin similar to diabetic ulcers; teeth are lost before the individual is 30 years old, cutaneous hyper elasticity; articular hyper mobility; narrow nasal bridge; scars on the arms and chin.

with their tongue (called Gorlin's sign); this can be seen in 8-10% of the normal population.[1] The palate is usually deep and dome-shaped in EDS patients.[11,14] Enamel hypoplasia is a common finding in such individuals.[8,10] The premolars and molars occasionally have deep fissures and high cusps. The teeth seem brittle and microdontia is sometimes manifesting.[8] Radiographs demonstrate pulp stones and root deformities in most cases.[8,10] Prognosis depends on the (sub) type and the proper and early diagnosis of the syndrome. In general, the prognosis is very bad in the vascular (sub) type [(sub) type IV] and is similar to healthy individuals in the mild classic (sub) type [(sub) type II].[10] The purpose of the present paper is to discuss the manifestations of this syndrome, especially its oral manifestations and to present a case, a 33- year- old man who had been referred to Mashhad Dental School to receive dental treatment.

Case Report

A 33-year-old man diagnosed with EDS had been referred to the Dental Faculty at Mashhad University of Medical Sciences to receive dental treatment. The patient had a familial history of EDS (his brother was afflicted with EDS). There were no indications of any systemic diseases in the patient's medical history and he was not taking any medications. His vital signs were normal. The patient's dental history indicated a scaling procedure in the past and a cosmetic skin surgery.

In the extra-oral examination of the patient, severe hyperelasticity of the skin was manifest but no articular hypermobility was observed (Figure 1 and 2). The patient had shaggy hair and cutaneous scars in the neck region. He also had epicanthuses in his eyes. Intra-oral manifestations included some retained primary teeth and the absence of some permanent teeth. The premolars demonstrated enamel hypoplasia. The patient's palate was very deep and dome-shaped (Figure 3). His tongue was soft and fissured but Gorlin's sign was not observed and his lip was bumpy (Figure 4 and 5).

A panoramic radiographic view revealed that the teeth $\frac{5\ 4\ 3\ |\ 3\ 5\ E}{5\ 3|\ 3\ 4\ 5}$ were impacted. The panoramic view also revealed calcification of stylomandibular ligament (Figure 6) and the periapical view revealed pulp stone (Figure 7).

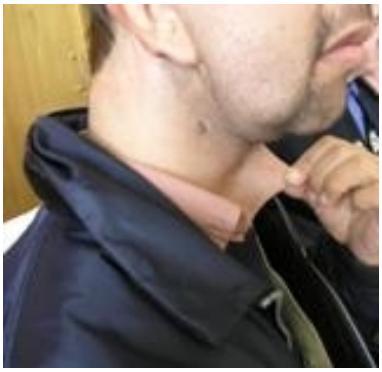

Fig 1: Patient with severe hyper elasticity of the skin.

Fig 2: No articular hypermobility was observed in the patient.

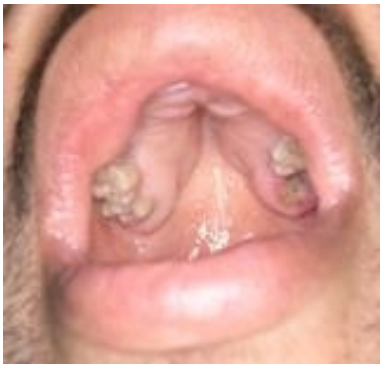

Fig 3: The patient's palate was very deep and dome-shaped.

Discussion

Mitral valve is usually involved in these individuals and there is a need for prophylactic antibiotics.[14] Appointments with such patients should be as short as possible so that iatrogenic traumas to TMJ can be avoided. The dental surgeon should be aware that inferior alveolar block injection may lead to hematoma.[14,15]

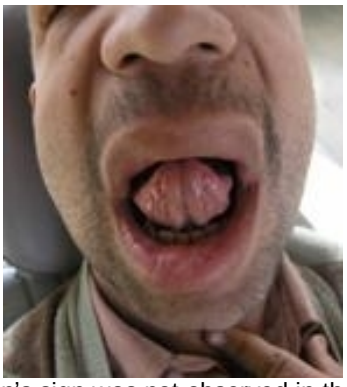

Fig 4: Gorlin's sign was not observed in the patient.

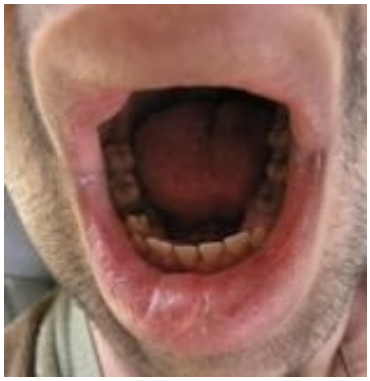

Fig 5: The patient had bumpy lip.

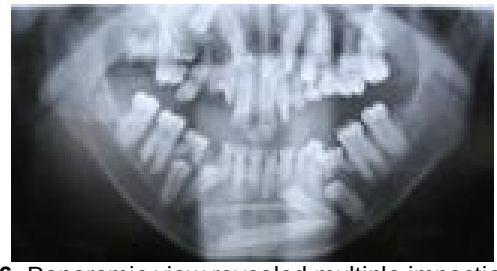

Fig 6: Panoramic view revealed multiple impaction and calcification of stylomandibular ligament

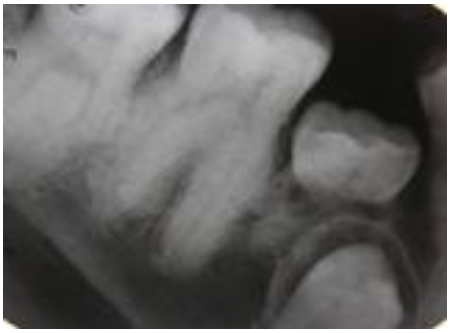

Fig 7: Periapical view revealed pulp stone.

Orthodontic forces should be lighter than those in healthy individuals because such forces damage periodontal ligaments. The teeth are moved more easily and with milder forces, and root resorption is not considered a problem in such patients.[9] A relapse is common, a longer retention period should be considered[9] and the dental surgeon should pay special attention to the fact that the oral mucosa is susceptible to injuries in such patients.[16] It is advisable not to perform maxillofacial surgeries for EDS patients. In case such surgeries are absolutely necessary, the blood factors should be carefully checked preoperatively. Sutures may not be able to keep the mucosa in place and acrylic covers should be used.[14]

Oral examinations can be of great help in the diagnosis of EDS and if the classic manifestations of the syndrome are present, the patient should be immediately referred to a specialist. The dental surgeon should be aware of the complications of rendering dental treatments to such patients so that they will not be exposed to any danger.

Acknowledgement

We thank Shiraz School of Dentistry for their cooperation.

Conflict of interest: None declared.

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