Ehlers-Danlos type VIII

Review of the literature

Abstract Ehlers-Danlos type VIII is a rare disorder characterized by soft, hyperextensible skin, abnormal scarring, easy bruising, and generalized periodontitis with early loss of teeth. To illustrate the clinical dermatological and dental features, we present the case history of a 20-year-old patient who has suffered from poor healing of wounds at the shins and knees since childhood, which have developed into hyperpigmented atrophic scars. In the course of orthodontic treatment during the last 3 years, severe apical root resorption, gingival recession, and loss of alveolar bone were observed. Family history was noncontributory for any skin or tooth disorders. The typical clinical signs confirmed the diagnosis of Ehlers-Danlos syndrome type VIII. As there is no specific treatment for the disorder, management is limited to the symptomatic treatment of the dental disease. It seems advisable to consider carefully the indications for orthodontic treatment in patients with Ehlers-Danlos type VIII syndrome.

Key words Periodontitis · Gingival recession · Abnormal scarring · Early tooth loss · Ehlers-Danlos syndrome

Introduction

The Ehlers-Danlos syndrome (EDS) is a group of generalized disorders characterized by abnormalities of the connective tissue, leading to fragility of the skin and blood vessels, hyperextensibility of the skin, and joint hypermobility [7]. There are many subtypes and variants of the EDS; at least ten types have been described on the basis of clinical symptoms and inheritance pattern. However, about half of the patients with EDS do not fit into one of the ten types [23]. Therefore, in 1997 a revision of the classification of the Ehlers-Danlos syndromes was proposed which is based primarily on the cause of each type [3].

EDS type VIII is of special interest from a dental viewpoint, due mainly to the resulting fragility of the oral mucosa and blood vessels, and an aggressive type of periodontitis causing severe loss of alveolar bone. EDS type VIII was recognized by McKusick [17] in 1972 in a family with skin fragility, abnormal scarring, early tooth loss, and severe periodontitis. Deformed roots and pulp calcifications in EDS patients have been reported [1]. Up to now the diagnosis of EDS type VIII has been based only on clinical criteria. It is distinguished from the other types of EDS by periodontitis and by the characteristic purplish discoloration of scars on the shins. Unlike EDS types IV, VI, and VII, no underlying biochemical defect has been detected in type VIII yet. We present the case of a young man with EDS type VIII to draw attention to this rare condition and to document its clinical dental and dermatological features.

Case report

A 20-year-old man with dental abnormalities was referred to the Department of Restorative Dentistry and Periodontology for diagnosis and treatment of severe periodontitis. The patient reported that he had suffered from gingival bleeding for many years and that the four first premolars had been extracted about 3 years ago, at the beginning of the orthodontic treatment. The periodontal status had deteriorated after orthodontic treatment had been started with fixed appliances about 3 years ago. Since the age of 5 years, wounds on the shins protractedly healed leaving atrophic, hyperpigmented scars. Mild trauma was followed by easy bruising. The patient also reported suffering from atopic ecema and allergic rhinitis since childhood. The family history was noncontributory for early tooth loss, easy bruising, joint hypermobility, or prolonged bleeding. Growth parameters at birth were normal.
On clinical dermatological examination, there were hyperpigmented scars along the shins with “cigarette-paper-like” thin skin and a clearly visible venous pattern. The patient’s habitus was markedly marfanoid with an asthenic build, pectus excavatus, and long extremities. However, on examination no skeletal, ocular, or cardiovascular abnormalities were found. There was only mild hyperextensibility of the skin and extremities and minimal hypermobility of the joints.

The clinical oral examination showed a dentition with multiple composite resin and amalgam restorations. The first premolars were missing and the wisdom teeth were retained. The gingiva was highly inflamed, with a papillary bleeding index [19] of 92%. Furthermore, severe recession could be observed at the lingual sides of teeth 46, 33, 42, and at the palatine side of tooth 26. The orthodontic appliances were still in situ (Fig. 1). The patient reported pain in all teeth, especially in the 4th quadrant, during tooth brushing and pain was provoked by gentle pocket probing. All teeth reacted positively to CO₂ testing; only tooth 27 was questionable. Due to the presence of the orthodontic devices, tooth mobility could not be tested. The X-ray (OPT) taken during this appointment (1997) could be compared with X-rays (OPT) taken in 1993 and 1994 (Fig. 2). The 1993 X-ray does not show any orthodontic appliance, whereas the one from 1994 does. Comparing the X-rays of 1993 and 1994, no root resorption can be observed. However, between 1994 and 1997, severe apical root resorption of all teeth (grade 3 on a scale of 0–3 [16]) of more than one-third of the total root length took place. Reduction of alveolar bone crest was also graded as severe (grade 2, on a scale of 0–2 [2]) for all teeth with the exception of the upper front, which implies a distance of more than 4 mm from cementoenamel junction to the alveolar bone crest [16]. No therapy was initiated because of lack of patient compliance.

**Discussion**

The distinguishing characteristics in EDS type VIII are the dental symptoms (inflamed gingiva and oral periodontal destruction) [22, 23]. Inheritance has been described as autosomal dominant [6]. However, other clinical manifestations vary, with different degrees of skin hyperextensibility, fragility and scarring, minimal-to-moderate small-joint hypermobility, and a normal-to-slightly increased tendency to bruising on mild trauma. Also abnormalities of hemostasis, e.g., bleeding abnormalities, have been described in some patients with EDS type VIII [8].

The diagnosis of EDS type VIII in our patient was based on a constellation of clinical findings that include rapidly progressive periodontal breakdown in the course of orthodontic treatment and pretibial atrophic and hyperpigmented scarring with prominent veins. There was no hyperextensibility of the joints. The absence of similar features in the family history suggests that EDS arose in this patient as a new mutation or is a recessive gene in his family.

Interestingly, in this case, orthodontic treatment had commenced, resulting in periodontal deterioration accompanied by severe apical root resorption. Root resorption and reduction of the alveolar crest are adverse effects that might be encountered in the course of any orthodontic treatment of adolescents as well as adult patients and which concern not only front but also posterior teeth [11, 12, 16, 18]. Lupi et al. [16] reported that apical root resorption increased from 15% to 73% and alveolar bone loss from 19% to 37% during orthodontic treatment. However, most of the increase is seen in a shift from grade 0 to 1 (blunted roots and less than 2 mm of bone loss, respectively) and is considered to be slight [16], which is in agreement with several other studies [11, 12, 18]. The present case, however, displays severe destructive processes in both apical root resorption and alveolar bone loss. Reasons for apical root resorption as described in the literature include trauma of the teeth, endodontic treatment, harmful habits such as bruxism,

**Fig. 1** Photograph taken in 1997, demonstrating an inflamed gingiva and a severe lingual gingival recession on tooth 36