Ehlers-Danlos Syndrome Type VIII with Severe Periodontitis and Apical Root Resorption After Orthodontic Treatment

Sir,

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 (1). There are many subtypes and variants of EDS; at least 10 types have been described on the basis of clinical symptoms and inheritance pattern. However, about half of patients with EDS do not fit into 1 of the 10 types (2). Therefore, in 1997 a revision of the classification of the Ehlers-Danlos syndromes was proposed, based primarily on the cause of each type (3).

EDS type VIII was recognized by McKusick (4) 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 (5). Up to now the diagnosis of EDS type VIII has been based only on clinical criteria. Unlike for EDS types IV, VI and VII, no underlying biochemical defect has yet been detected in type VIII. We present the case of a young man with EDS type VIII, in order to draw attention to this rare condition and document its clinical dermatological and dental 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 4 first premolars had been extracted about 3 years ago at the beginning of the orthodontic treatment. His periodontal status had deteriorated after orthodontic treatment had been started with fixed appliances about 3 years previously. Since the age of 5 years, wounds at the shins protractedly healed leaving atrophic, hyperpigmented scars. Mild trauma was followed by easy bruising. The patient also reported suffering from atopic eczema and allergic rhinitis since childhood. The family history was non-contributory for early tooth loss, easy bruising, joint hypermobility or prolonged bleeding. Growth parameters at birth were normal.

On clinical dermatological examination hyperpigmented scars were visible along the shins, with "cigarette-paper-like" thin skin and clearly visible venous pattern (Fig. 1). 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 (6) of 92%. Furthermore, severe recession could be observed at the lingual sides of teeth 46, 33, 42 and at the palatal side of tooth 26. The orthodontic appliances were still in situ. The X-ray (OPT) taken during this appointment (1997) could be compared with X-rays (OPT) taken in 1993 (Fig. 2).

Between 1993 and 1997 severe apical root resorption of all teeth (grade 3 on a scale of 0 – 3 (7)) beyond one-third of the total root length took place. Reduction of alveolar bone crest was also graded severe (grade 2, on a scale of 0 – 2 (8)) for all teeth, with the exception of the upper front teeth.

DISCUSSION

The distinguishing findings in EDS type VIII are the dental symptoms (inflamed gingiva and oral periodontal destruction) (2, 9). Inheritance has been described to be autosomal dominant (10). However, other clinical manifestations vary, with different degrees of skin hyperextensibility, fragility and scarring, minimal to moderate small-joint hypermobility and normal to slightly increased tendency to bruising on mild trauma. In addition, abnormalities of haemostasis, e.g. bleeding abnormalities, have been described in some patients with EDS type VIII (11).

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 an orthodontic treatment and pretibial atrophic and hyperpigmented scars 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 been commenced, resulting in periodontal deterioration accompa-
abnormalities of type I collagen. Cellular autoimmunity to collagen analysis identified morphological and physical condyles and peripheral skeleton and cardiac valvular disease. cutaneous vasculitis, resorptive osteolysis of the mandibular developed not only destructive periodontal disease, but also reported on a patient with Ehlers Danlos type VIII who have been reported in the literature. Hoffman et al. (15) itself. reason for the enhanced tissue destruction, as well as the EDS existing periodontal disease before the orthodontic treatment period (14). In the present case the potentially enhanced alveolar bone loss during the active orthodontic treatment are the age of the patient, time and type of treatment.

Factors for enhanced alveolar bone loss during orthodontic treatment are the age of the patient, time of orthodontic treatment, but also retention of plaque (7). Reasons for apical root resorption as described in the literature include trauma to the teeth, endodontic treatment, adverse habits, such as bruxism, and the time of orthodontic treatment including the applied forces (7, 13). However, endocrine disorders, hormonal imbalances and genetic predisposition are also documented (7, 13). Family-based osteolysis has been described as a cause for extensive apical root resorption (8). The present case may be indicative that EDS patients belong to a high-risk group for root resorption in the course of an orthodontic treatment.

Factors for enhanced alveolar bone loss during orthodontic treatment are the age of the patient, time and type of orthodontic treatment, but also retention of plaque (7). Existing periodontitis as such is also a causative factor for enhanced alveolar bone loss during the active orthodontic treatment period (14). In the present case the potentially existing periodontal disease before the orthodontic treatment or poor plaque control during the treatment may be a major reason for the enhanced tissue destruction, as well as the EDS itself.

Since EDS type VIII is a very rare disease, only a few cases have been reported in the literature. Hoffman et al. (15) reported on a patient with Ehlers Danlos type VIII who developed not only destructive periodontal disease, but also cutaneous vasculitis, resorptive osteolysis of the mandibular condyles and peripheral skeleton and cardiac valvular disease. Collagen analysis identified morphological and physical abnormalities of type I collagen. Cellular autoimmunity to type I collagen was found in vitro, which may be responsible for this patient’s intractable clinical condition. Sloomtew & Beemer (2) found gingival fibrinoid deposits on histological examination in a 7-year-old girl with EDS type VIII and speculate that these deposits are due to the disordered collagen metabolism that underlies the EDS symptomatology.

In children or young adults presenting with gingival inflammation and/or periodontal disease associate with a history of skin or joint symptoms should be considered as possible candidates for EDS type VIII. These patients may be considered to be a high-risk group for orthodontic treatment in respect to both enhanced apical root resorption and alveolar bone loss, which are practically irreversible processes.

REFERENCES

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Fig. 2. Orthopantomograms taken in (a) 1993 and (b) 1997 showing severe root resorption since 1993.