

## Ehlers-Danlos syndrome identified from periodontal findings: case report

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### Introduction

The Ehlers-Danlos syndrome comprises a group of generalized connective tissue disorders characterized by fragile skin, skin hyperextensibility, and joint hypermobility. More than 10 types of Ehlers-Danlos syndrome have been identified based on genetic and biochemical studies.<sup>1</sup> What is now known as Ehlers-Danlos syndrome (EDS) Type VIII was first described in 1972. We report a case of EDS Type VIII in a seven-year-old female.

### Literature review

In 1972, McKusick<sup>2</sup> identified an autosomal dominant form of EDS characterized by periodontal disease in early adulthood, scarring of pretibial skin unaccompanied by joint hypermobility, and hyperextensibility of skin. Stewart et al.<sup>3</sup> reported the case of a family with autosomal dominant transmission, that exhibited the common findings of postpubertal advanced destructive periodontal disease, ecchymotic pretibial lesions, and cigarette-paper scarring. There was moderate joint hypermobility, but no hyperextensibility of the skin.

In describing EDS Type VIII Gorlin<sup>1</sup> states that the onset of skin fragility is noted in childhood. Ecchymoses following slight trauma resolve, except for the pretibial areas. These heal with tender yellow-brown atrophic wrinkled scars that somewhat resemble venous stasis, and scarring becomes worse with age. Periodontal disease appears after puberty, and permanent teeth are usually lost by the third decade.

According to Hartsfield<sup>4</sup> there is a significant amount of overlap between EDS, types IV and VIII. Type IV has demonstrable type III collagen abnormalities, while type VIII has normal collagen as indicated by current gel electrophoresis techniques. Other features that characterize type IV are hyperextensibility of skin, easy bruisability, pes planus, and arterial and intestinal rupture. Type IV can be fatal.

### Case report

HW, a 7-year-old white female, came to the Pediatric Dental Clinic at Arkansas Children's Hospital for initial dental evaluation. Her parents were concerned with the appearance of her teeth. She had been evaluated previously by a dentist who noted a malocclusion and had referred her for orthodontic evaluation.

A thorough review of the medical history indicated that the child had an unremarkable prenatal course and birth. She had no significant illnesses or hospitalizations. Her development was normal, and the family history was unremarkable for genetic anomalies. Her history did reveal occasional, prolonged epistaxis. The patient's dental history was, according to her parents, uneventful, although she had never had routine professional care. Tooth eruption and exfoliation had occurred without difficulty. The child and her parents were not aware of any carious teeth or toothaches. She reported brushing her teeth twice daily.

Physical examination revealed a thin white female with translucent skin. She had extensive bruising covering her anterior shins (Fig 1). There was hyperextensibility of her fingers, elbows, and knees. Her height was in the 95th percentile and her weight was in the 25th percentile. Laboratory examination revealed normal CBC, platelets, PT, and PTT; however, her bleeding time was greater than 15 min.

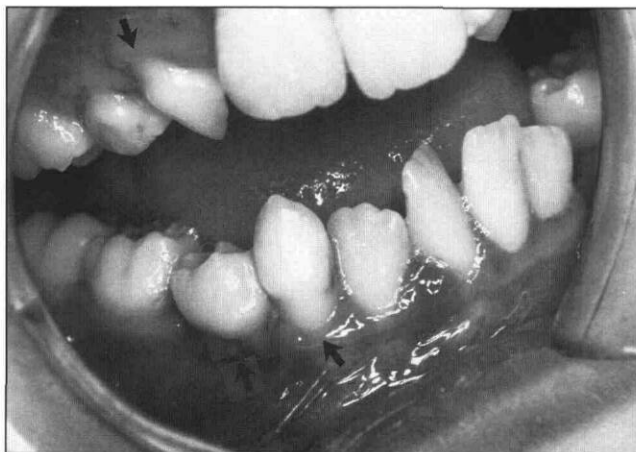
Oral examination revealed early mixed dentition with a Class I relationship of her first permanent molars and primary canines. There was severe crowding of incisors and she had a posterior crossbite on her right side. Severe

generalized gingivitis with gingival recession and exposed root surfaces on the mandibular primary molars were noted (Fig 2). Radiographs showed 4- to 5-mm bony pockets surrounding the primary and permanent molars (Fig 3). There was a heavy accumulation of calculus on the lower incisors and all molars. Her left primary mandibular canine and first molar had exfoliated prematurely.

Due to the unusual findings of prolonged bleeding time and pretibial bruising combined with advanced periodontal disease, systemic disease was suspected. The pa-



**Fig 1.** Ecchymotic pretibial lesions were noted on both legs. They were asymptomatic, and had been present for as long as the child and parent could remember.



**Fig 2.** Severe gingival inflammation with recession was present in all molar and canine regions (arrows).

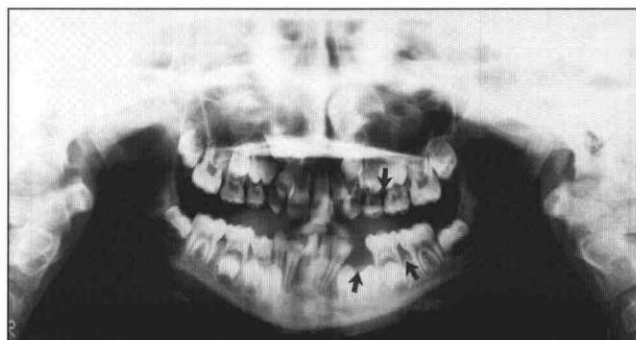
tient was referred to the departments of Pediatric Genetics and Pediatric Hematology for a thorough examination. Biochemical analysis of cultured skin fibroblasts indicated normal type III collagen. Based on clinical findings, a diagnosis of Ehlers-Danlos syndrome Type VIII was made.

### Discussion

Ehlers-Danlos syndrome Type VIII is a rare condition involving advanced periodontal disease. The case presented here demonstrates how a significant medical condition was diagnosed as a result of a thorough dental examination and appropriate medical follow up.

Advanced periodontal disease in children includes a long list of systemic conditions in a differential diagnosis when local factors are ruled out. This differential diagnosis includes: histiocytosis X, cyclic neutropenia, Papillon-Lefèvre syndrome, hypophosphatasia, leukemia, vitamin D-resistant rickets, leukocyte disorders, and acrolynia. EDS Type VIII should be included in this differential diagnosis.

Periodontal disease in combination with connective tissue abnormalities is unusual; one should investigate the diagnosis of Ehlers-Danlos syndrome. As pointed out by other authors, periodontal disease may not become evident until adolescence. Periodontal destruction was a



**Fig 3.** Panoramic radiograph showing bone loss around primary and permanent molars (arrows).

consistent finding when type VIII was diagnosed, but may also be found in individuals with type IV. Due to the potentially fatal nature of EDS type IV, collagen studies are imperative in making a diagnosis.

The long-term prognosis for the periodontal health of individuals with Ehlers-Danlos syndrome Type VIII is guarded, as the underlying connective tissue defect is unknown. Children demonstrate precocious loss of the primary dentition. Beginning early in the second decade of life, aggressive gingival recession occurs, punctuated by episodes of gingival infection and inflammation. Affected individuals generally do not respond well to conventional periodontal therapy.<sup>5</sup>

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