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# Ehlers Danlos Syndrome: A Case Report

**Dr. Kalabarathi S[1], Agilan R[2]**

[1]HOD cum Principal, Department of Obstetrics and Gynecological Nursing, Saveetha College of Nursing,
[2]M.Sc (Nursing) - Nurse Practitioner in Critical Care, Saveetha College of Nursing,
[1,2]Saveetha Institute of Medical and Technical Sciences, Chennai, Tamil Nadu, India

## ABSTRACT

Ehlers-Danlos syndrome (EDS) is an inherited collagen disorder that primarily affects the skin and joints. Clinically, it is characterized by skin hyperelasticity and joint hypermobility. In this article, he described a rare condition observed in her 10-year-old boy who was diagnosed with EDS based on clinical, radiographic, and histological findings.

***KEYWORDS:*** *Hereditary collagen disorders, Connective tissue disorder, Joint hypermobility*

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## INTRODUCTION

Ehlers-Danlos syndrome is a systemic disease of the connective tissue component, manifested clinically by skin fragility and hyperelasticity and joint laxity. It is an inherited disease, and inheritance is usually autosomal dominant with low penetrance. Autosomal recessive and X-linked recessive variants are also known. Already since the 4th century BC. When Hippocrates described loose joints and numerous burn-like scars on several Scythian limbs, the current state of knowledge on different clinical aspects with different genetic patterns was recently described. After the description of hyperelastic skin by Van Meekeren and Koop Ehlers was the first to notice that skin is vulnerable. Focusing on tumorigenesis, we also established four diagnostic criteria: skin hyperelasticity, skin fragility, joint hypermobility, and subcutaneous mollusc pseudotumor formation. The number of cases in the literature is very small. The available information reported only about 500 cases by 1967, mostly in Europeans. Indian literature were chosen as the case of the present publication because the reports are even more sparse.

## Case description

A 10-year-old boy reported to our college with chief complaint of inability in chewing caused by missing teeth since past 2 years. The patient presented with missing deciduous teeth, and he reported premature exfoliation of posterior deciduous teeth within five years, except primary canines. The primary concern was of unerupted permanent teeth, except for the first permanent molars, which displayed delayed eruption. Medical history revealed cerebral fever (at age of 4 years) and hernia 15 days post birth. The family history revealed that the patient's paternal grandfather had highly flexible joints. However, nothing extraordinary was reported / recalled, with respect to any of the existing and deceased family members. Considering the little family information which was available, no conclusions could be drawn to ascertain any genetic pattern.

**Extraoral examination revealed:**

a) A mesomorphic body structure with a dolichocephalic head.

b) A leproprosopic face with orbital hypertelorism.

c) A wide nasal bridge with a depressed bulb of nose and stretched external nares.

d) The skin over the extremities was loose and easily stretchable.

e) The patient appeared to be shy and non-conversant, especially because of his unaesthetic appearance. The patient's score was eight on the Beighton scale (scores which are ≥ than 5 describe hypermobility) as was assessed by the joint mobility test which was suggested by Beighton et al., by using a protractor.

**Intraoral examination and evaluation revealed**

a) Macroglossia.

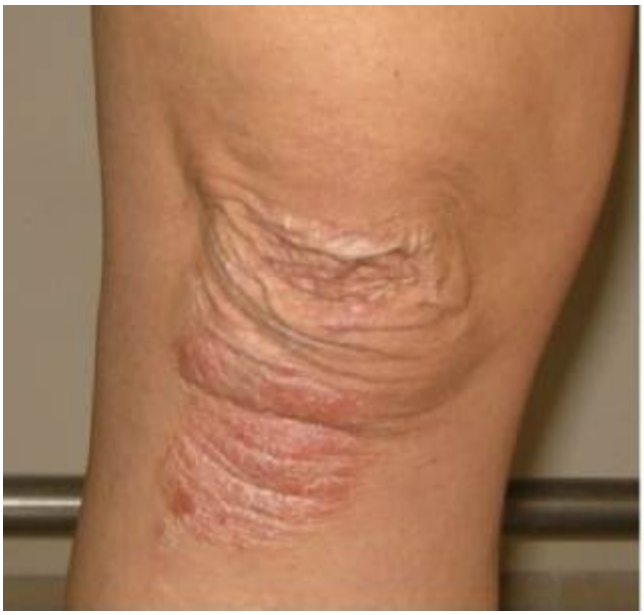
b) Positive Gorlin's sign.

c) The edentulous gingiva appeared to be comparatively thick and fibrous

d) Teeth which were present in the oral cavity were 16, 26, 36, 46, 53, 63, 73 and 83.

e) Grade II mobile 53, 63, 73, 83.

f) A noticeable loss of vertical dimension. A maxillary occlusal radiograph was taken to rule out the presence of any supernumerary teeth. An orthopantomograph (OPG) revealed the presence of all the remaining unerupted permanent teeth. Biopsies of the gingival tissues which overlay the unerupted 13, 12, 11, 21, 22, 23 were obtained and they were sent for a histopathologic examination. The histological sections of the tissues were stained with Haematoxylin and Eosin, which showed hyperplastic keratinized epithelium with dense, fibrotic connective tissue, with numerous plump fibroblasts and blood vessels. On the basis of the classic clinical and histopathologic findings, the patient was diagnosed with Ehlers Danlos Syndrome Type III, [1]. A treatment plan was formed and it consisted of a) Extraction of the deciduous teeth b) Use of a removable partial denture prosthesis (RPD) for restoring form, function and aesthetics subsequently



**Discussion:**

Ehlers Danlos syndrome is a rare connective tissue syndrome that mainly affects the skin and the joints. EDS is a rare type of disorder of the connective tissue that affects the collagen metabolism in which deficiency and /or disordered deposition of collagen takes place One of the first descriptions given was of a young Spaniard who could stretch the skin which overlay his right pectoral muscle, over to the left angle of his mandible The skin and joint manifestations of this syndrome have been studied; however, only little emphasis has been laid on the oral manifestations of this syndrome

Pommeau-Delille and Soussie christened this condition as Ehlers-Danlossyndrome Ehlers, first described this syndrome as one with hyperplastic skin and a strong tendency to get bruised. Danlos, found a pseudo-tumour of a molluscoid or fibrous type. This syndrome affects 1 in 5,000 people, it is inherited as an autosomal dominant trait and it may also be associated with X chromosome Depending on which type of collagen is affected, the disorder has a wide range of expressivity and there are many phenotypes. A new classification which was based on cause was developed, which described 6 types of EDS Out of the known six types, only 4 types of EDS, namely types IV, VI, VII and X can be confirmed by doing biochemical and molecular tests As 19 types of collagen are found in the human body, it is especially difficult to establish a precise diagnosis. Despite bleeding disorders being associated with EDS, blood investigations are not of much diagnostic value, as no correlation has been found between the findings and the various types of EDS A few of the EDS types can be confirmed by genetic testing, after the clinical criteria are met. However, many cases may still remain ambiguous and they do not fit in any of the well-described subtypes

**The characteristic features of this syndrome are**

1. Hypermobility of the joints.
2. Hyperelasticity, fragility and softness of the skin.
3. Deficient healing of wounds.
4. Ecchymosis caused by minor traumas.

Besides the cutaneous and articular anomalies, the patients may show cardiovascular complications (such as aneurysms and mitral valve prolapse), gastrointestinal complications (hernias and gastrointestinal diverticulosis), and ocular defects

**The oral manifestations of EDS include the following:**

1. The mucous membrane is fragile, which may bleed on instrumentation and which sutures cannot hold
2. Dentinal aberrations like pulp stones, short and deformed roots.
3. A high incidence of caries in the deciduous teeth
4. Spontaneous fractures of teeth have been reported.
5. Early onset of generalized periodontitis is one of the most significant oral manifestations of the syndrome
6. Hyperelasticity, fragility and softness of the skin.
7. A supple tongue. Approximately 50% of those with this syndrome can touch the end of their noses with their tongue (Gorlin's sign), as compared to 8-10% in the normal population who can do this
8. Hyper mobility of the TMJ, with increased incidence of dysfunction may be seen in some cases.
9. In this patient, the connective tissue hyperplasia which was manifested in the gingiva, resulted in its thickening, which must have prevented the eruption of the permanent teeth and resulted in the chief complaint of the patient. The classic clinical findings, along with histopathologic findings, confirmed the diagnosis. The removal of the deciduous teeth prematurely, was primarily guided by the unusual mobility which presented and the prosthodontic rehabilitation of the patient was aimed at rapidly addressing the chief complaint. The patient was periodically recalled for two years for the post insertion adjustments of the prosthesis. The permanent teeth did not show any signs of erruption during this period of follow up.

## Conclusion

Ehlers-Danlos Syndrome is an autosomal dominant hereditary connective tissue disorder with skin and joint manifestations. This condition is also known to have oral manifestations that clinicians must be aware of, for making correct diagnoses of such patients and for proper treatment planning..

**Conflict of Interest:**
None

**Funding:**
None

**Consent for publication:**
Informed consent was obtained from the parents of the patients to publish this case in medical journal.

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