

EHLER DANLOS SYNDROME - A CASE REPORT

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ABSTRACT

Ehlers-Danlos syndrome (EDS) is a generalized disorder of one element of connective tissue manifesting clinically by fragility and hyperelasticity of the skin and joint laxity. It is a hereditary disorder, the inheritance being usually autosomal dominant with low penetrance. Autosomal recessive and X-linked recessive varieties are also known. First described by Hippocrates in 4th century B.C., the various clinical types with variable penetrance have been described lately. The number of cases EDS reported in the literature is very meagre.^[1]

KEYWORDS: Adolescent, Ehlers-Danlos Syndrome, skin hyper extensibility, joint hyper mobility.

INDRODUCTION

The Ehlers-Danlos syndromes (EDS) are a heterogeneous group of disorders characterized by joint hypermobility, hyperextensible skin, and fragile tissues that are extremely susceptible to trauma. As many as 1 in 5000 individuals may be affected by some form of the disease. Categorization is based on clinical and corresponding molecular pathogenetic findings in collagen types I, III and V, or processing enzymes. The classic form of EDS is one of the most common, occurring in 1 *per* 10,000 to 20,000 infants. It includes two previously designated subtypes (EDS type I, or *gravis* and EDS type II, or *mitis*) that are now recognized to form a continuum of clinical findings and differ only in phenotypic severity.^[2]

CASE REPORT-1

A 13-year-old boy born of a consanguineous marriage reported to our Department of Oral Medicine and

Radiology with the chief complaint of inability in chewing caused by irregular arrangement of upper front teeth for past 2 years. The past medical, surgical, dental history were not contributory. Patient having positive family history revealed that his younger sister was also having similar problems. On general examination patient was calm, cooperative, conscious and oriented with time, place and person. Stature and build were normal. On examination of musculoskeletal system the patient was having hyperextensibility of joints and hyperelasticity of skin.

Extraoral examination revealed a mesomorphic body structure with a mesencephalic head. He had a mesoprosopic face with orbital hypertelorism. A wide nasal bridge with a depressed bulb of the nose and stretched external nares were present. The skin over the extremities was loose and easily stretchable especially in the ears.



Figure 1: Facial profile.



Figure 2: Anterior crossbite.



Figure 3: Hyperextensibility of joints.



Figure 4: Hyperextensibility of ear lobe.



Figure 5: Orthopantomogram reveals presence of all permanent teeth and horizontally impacted 35.

Intraoral examination and evaluation revealed normal mouth opening. The teeth which were present in the oral cavity were 16,15, 14, 12, 11, 21, 22, 24, 25, 26, 36,34,32,31,41,42,44,45,46. Anterior crossbite with retrognathic maxilla was present. A noticeable loss of vertical dimension was present. An orthopantomogram (OPG) revealed presence of all the teeth.

CASE REPORT-2

A 10 year old girl born of a consanguineous marriage reported to our Department of Oral Medicine and Radiology with a chief complaint of irregular teeth arrangement of upper front tooth for the past 2 years. The past medical, surgical, dental history were not contributory. Patient was having positive family history with her elder brother also having similar problems. On general examination the patient was calm, cooperative, conscious and oriented with time, place and person.

Stature and build were normal. On examination of the musculoskeletal system, the patient was having hyperextensibility of joints and hyper elasticity of skin.

Extraoral examination revealed a mesomorphic body structure with a mesencephalic head. A mesoprosopic face with orbital hypertelorism was present. Patient was having deficit in malar prominence with mild concave profile. A wide nasal bridge with a depressed bulb of nose and stretched external nares was present. The skin over the extremities was loose and easily stretchable especially in ears.

On Intra-oral examination, mouth opening was normal. The teeth which were present in the oral cavity were 55,54,53,52,51,61,62,63,64,65,75,74,73,72,31,41,82,83,84,85.



Figure 5: Facial profile.



Figure 6: Deficit malar prominence.



Figure 7: Hyperextensibility of joint.



Figure 8: Intraoral photograph.

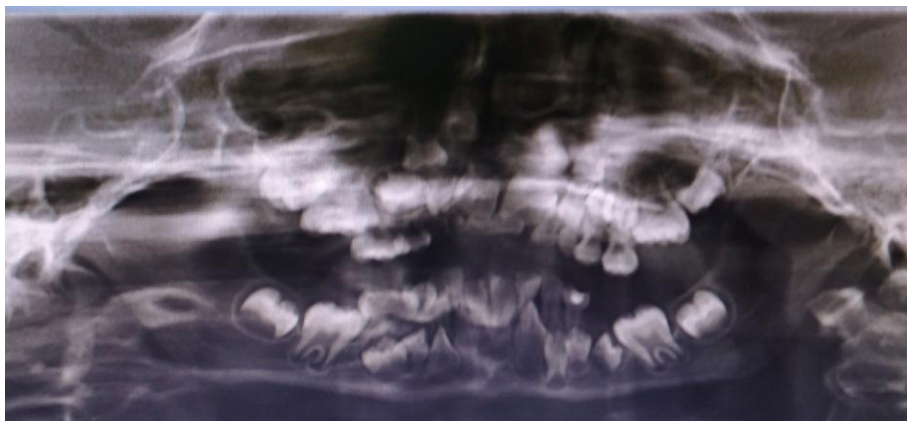


Figure 9: Orthopantomogram reveals presence of mixed dentition period.

DISCUSSION

Ehlers-Danlos syndrome, classic type is a disorder of connective tissue inherited in an autosomal dominant manner. It is estimated that approximately half of the affected individuals have inherited the disease causing mutation from an affected parent, and the other half have a *de novo* mutation. Mutations in the *COL5A1* and *COL5A2* genes, encoding the $\alpha 1$ and $\alpha 2$ -chain of type V collagen, respectively, are identified in about 50% of patients with a clinical diagnosis of classic Ehler danlos syndrome. . It is estimated that 46% of cases of classic EDS can be attributed to mutations in *COL5A1* and 4% to mutations in *COL5A2*. Most mutations identified result in a reduced amount of type V collagen in the connective tissues available for collagen fibrillogenesis. Considerable inter- and intra familial differences in the phenotypic expression are observed in classic EDS, but no genotype-phenotype.^[2]

Classic EDS is characterized by hyperextensible and fragile skin, which is smooth and velvety to the touch,

delayed wound healing with formation of atrophic scars, easy bruising, and generalized joint hypermobility. The skin manifestations can vary in severity from mild to severe; milder forms were previously termed the *mitis* type, or EDS type II. Traumatic or surgical scars are thin, papyraceous, and may stretch considerably after healing. In more severe cases, scars may have a characteristic “fish mouth”- or “cigarette paper”-like appearance. Thin, atrophic, darkly pigmented scars are formed as a consequence of intradermal or subdermal hematomas, and occur mainly at pressure points.^[2] In our cases joint hypermobility was present elicited by finger hyperextensibility. Skin was hyperelastic in both cases. No scars and hematomas were found.

The oral manifestations of EDS include the following: the mucous membrane is fragile, which may bleed on instrumentation and which sutures cannot hold ; Dental aberrations like pulp stones, short and deformed roots; A high incidence of caries in the deciduous teeth. Spontaneous fractures of teeth have been reported. Early

onset of generalized periodontitis is one of the most significant oral manifestations of the syndrome. Tongue is supple. 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 hyper extensibility of the tongue, with increased incidence of TMJ dysfunction may be seen in some cases.^[3] In both cases reported here, Gorlin's sign was negative.

The following preventive, therapeutic and surveillance measures are followed in classic Ehlers-Danlos syndrome: 1. Wearing pads or bandages over the forehead, knees, and shins to avoid skin tears – young children with skin fragility. 2. Wearing soccer pads or ski stockings with shin padding during activities – older children. 3. Ascorbic acid (vitamin C) to reduce bruising; in general, a dose of 2 g/day is recommended for adults, with proportionally reduced doses for children. 4. Avoid acetylsalicylate (aspirin) and sports that strain joints (contact sports, fighting sports, football, running). 5. Physiotherapy for children with hypotonia and delayed motor development. 6. Promotion of muscle strength and coordination with non-weight-bearing exercise, such as swimming. 7. Anti-inflammatory drugs to alleviate arthralgias. Adapt lifestyles in patients with hypotonia, joint instability and chronic pain. 8. Close dermal wounds without tension, preferably in two layers; apply deep stitches and left skin stitches in place twice as long as usual; carefully tape the borders of adjacent skin to prevent stretching of the scar. 9. Treat cardiovascular problems. 10. Yearly echocardiogram when aortic dilatation and/or mitral valve prolapse are present. 11. Refer pregnant women with EDS to high-risk obstetric practices when possible because of increased pregnancy risks. 12. It is important to include blood test results like prothrombin time (PT), partial thromboplastin time (PTT), and international normalized ratio blood test (INR) as a routine part of evaluation in patients with EDS.^[2,4]

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 dental and oral clinicians must be aware of, for making correct diagnosis of such patients and for proper treatment planning.

REFERENCE

1. Ehlers-Danlos syndrome Farhana Tahseen Taj, Vijaya V. Sajjan, Dolly Singh Indian Dermatology Online Journal, 2014; 5(1).
2. Classic Ehlers-Danlos syndrome: Case Report and Brief Review of Literature Paulo Morais¹, Olga Ferreira¹, Sofia Magina¹, Cármen Silva², Miguel Leão³, Ana Maia², Filomena Azevedo¹ Acta Dermatovenerol Croat, 2013; 21(2): 118-122.

3. Ehler danlos syndrome: A case report Pragati Kaurani¹, Nikhil Marwah², Mayank Kaurani³, Narendra Padiyar⁴ Journal of Clinical and Diagnostic Research, 2014 Mar, 8(3): 256-258.
4. Dental Considerations in Ehlers-Danlos Syndrome: A Case Report Ghassem Ansari ¹; Masoud Fallahinejad Ghajari ^{1,*}; Bahareh Nazemi ²; Behnaz Erfanian J Compr Ped, 2015 February; 6(1): e22463.47.