Case Report

Ehlers-Danlos Syndrome - Clinical presentation

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Abstract

Two cases of Ehlers-Danlos syndrome (EDS) diagnosed on the basis of their clinical presentation are presented. EDS is a rare genetic disorder involving connective tissue of skin and joints, having a variety of presenting signs and symptoms. There is no specific therapy for this disease but it is important to diagnose it as it may end up in fatal complications.

Introduction

Ehlers-Danlos Syndrome (EDS), is described by the Ehlers-Danlos National Foundation as a "heterogeneous group of heritable connective tissue disorders characterized by articular hypermobility, skin extensibility and tissue fragility".[1] The incidence of EDS is approximately 1 in 5000 births. Due to lack of awareness among patients and health professionals, many cases remain unnoticed or misdiagnosed. Two cases are presented here to increase awareness of this disease.

Case Reports

The first case was of a 12 year old boy who presented with recurrent, painless, subcutaneous bruises in his limbs, that occurred either spontaneously or after minimal trauma, for the past 3 years. The bruised areas did not itch, were initially erythematous, gradually becoming dark and resolved without any sequelae. His symptoms neither aggravated during the cold weather nor were they associated with joint pains or fever. Past history was unremarkable with no previous history of jaundice or blood transfusion. He also did not give any history of drug intake or chemical exposure. There was no history of bleeding from any site including the oral mucosa and no history of malaena. His parents and siblings were alive and healthy and there was no history of any illness running in the family.

He was a young boy of thin built, with small body frame for his age. His vitals were normal. The rest of the general examination including the oral cavity was unremarkable. His skin was soft, velvety, and hyperextensible (Figure 1). A thin, 'cigarette paper' scar was present on the dorsum of one hand presumably occurring after a burn (Figure 2). Several erythematous to violaceous, non-blanching lesions, 0.5 to 2.0 cm in diameter were present in both the limbs. These were nonpalpable and

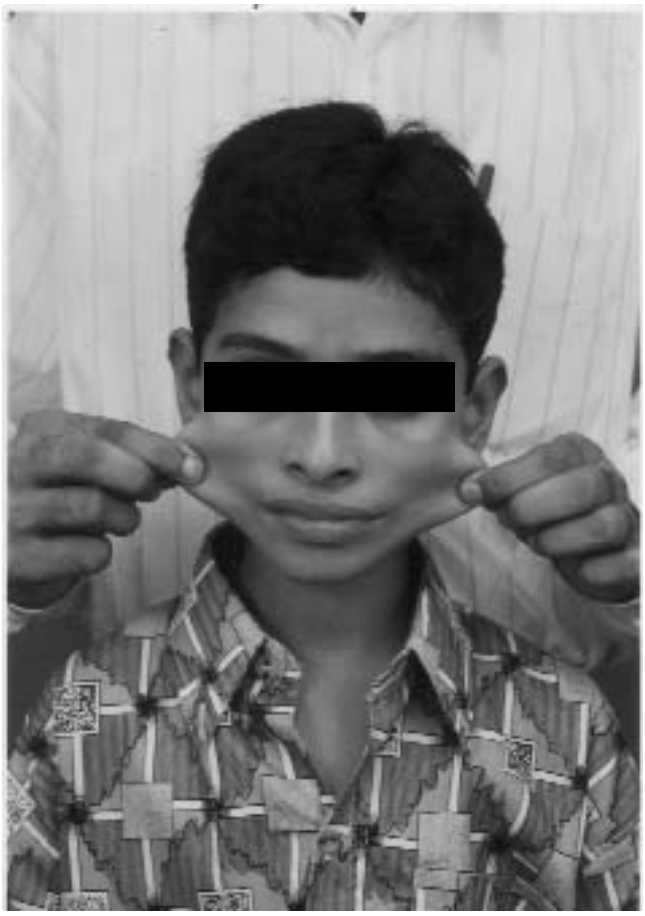

Figure 1. Skin Hyperextensibility.

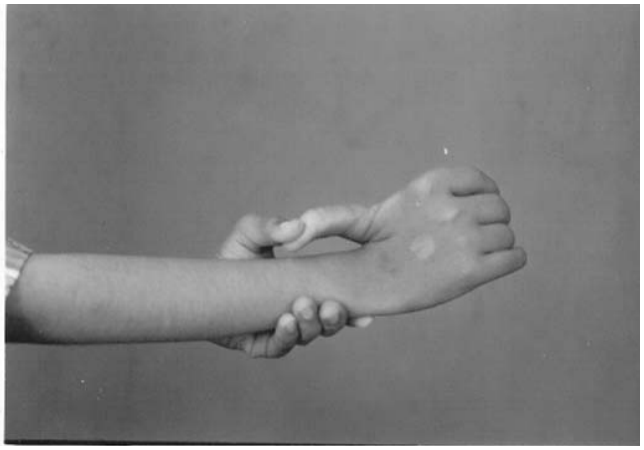

Figure 2. Shows hypermobile joints & a characteristic thin, 'cigarette paper' scar on dorsum of hand. Bruises are also visible near the elbow joint and on dorsum of hand.

nontender. There were no visible veins. Musculo-skeletal examination revealed hypermobile joints without any deformity, dislocation or inflammation (Figure 2). There was no evidence of scoliosis, pes planus, arachnodachtyly or any eye abnormality. Cardiovascular system examination was normal with no evidence of mitral valve prolapse. Examination of other systems was also normal.

The complete blood picture, ESR and bleeding profiles were normal. Rheumatoid factor and antinuclear antibody were negative. X-ray showed no evidence of osteoporosis. Ultrasound abdomen for involvement of viscera and abdominal aorta did not reveal any abnormality. A diagnosis of classical EDS was made on clinical grounds.

The patient did not require any surgical repair and was advised to limit physical activity and avoid trauma. He was given Folic acid and Vitamin C to reduce the likelihood of bruising.

The second case was that of a 27 year old man presenting with diarrhoea of recent onset with a frequency of 4-5 stools / day. It was not associated with fever. On systemic review he was found to have the symptoms of reflux oesophagitis for several years, such as heartburn, regurgitation and temporary relief with antacids. His past medical/ surgical as well as drug history was insignificant. There was also no evidence of any familial illness.

The patient was of average height and had normal vitals. His facial appearance was quite remarkable with prominent upper lip, a broad base of nose and epicanthic folds displaced laterally though the interpupillary distance was normal. Oral examination revealed a high arched palate and a haemangioma on the undersurface of his upper lip. His skin was very soft, velvety and hyperextensible though there were no scars or bruises present. The joints of his body were hypermobile, non-tender, and without crepitus or dislocation. Examination of other systems revealed no abnormality.

On investigation his complete blood picture, ESR and bleeding profile were all normal. Imaging studies including X-ray chest and ultrasound abdomen were normal. No abnormality was detected on ECG or echocardiography. Upper GI endoscopy revealed reflux esophagitis without any other pathology. He was diagnosed as hypermobility type of EDS.

His diarrhoea subsided spontaneously and he was treated for his reflux esophagitis. A biopsy of upper lip was planned to exclude malignancy. Otherwise he was advised to avoid strenuous physical activity.

Discussion

Ehlers-Danlos Syndrome comprises a group of heterogeneous disorders affecting skin, ligaments, joints, and blood vessels. The current classification system for EDS proposes six major types, classical, hypermobility, vascular, kyphoscoliosis, arthrochalasis, and dermatosparaxis.[1] Presentation may be mild to severe, depending upon the type of syndrome affecting the patient. It is important to diagnose and classify EDS as affected individuals with certain types are at risk of dramatic and often fatal complications such as failure of wound closure, degenerative joint disease, rupture of major vessels as the aorta and hollow organs as bowel or uterus, rupture of eyeball, and premature rupture of membranes during pregnancy.[2]

Classical-type EDS[3] includes type I and II of previous classification, having major and minor clinical manifestations respectively. The first case was included under this category (type II of previous classification), as his skin was soft, velvety and hyperextensible. He also had the characteristic 'cigarette paper' scar and his joints were hypermobile (Figure 2). A 9-point scoring system, mentioned by Beighton[4], was used to measure joint hypermobility. Tolat and Gokhale[5] have reported a similar case in all respects except for the easy bruising found in this case unaccounted for by a normal bleeding and coagulation profile. Our case also did not have any subcutaneous nodules (fibrous transformation of subcutaneous haematomas, called molluscoid pseudotumours), or blue sclerae, as have been described by other workers.[6,7] Classical EDS is an autosomal dominant disorder due to mutation in the gene that affects collagen type V. Half of the cases are due to new mutation with no prior family history, as in this case, although we cannot exclude the possibility of a milder variety in the family that may have gone unnoticed. It is important to recognize this syndrome as it plays a role in the prognosis of diseases involving collagen. These include skin (acne, cutaneous surgery, and bruising disease) and joint diseases (dislocations).

The second case was diagnosed as hypermobility-type EDS[8] on the basis of his soft, velvety, hyperextensible skin and hypermobile joints. He did not have any evidence of bleeding under the skin. His facial appearance revealed epicanthic folds which are the commonest ophthalmic feature of EDS, producing apparent broadening of the nose as was evident in this case.[9] He had a haemangioma in his upper lip. A case of vascular-type EDS with mediastinal epithelioid haemangioendothelioma has been reported with no etiologic association between the two.[10]

No specific therapy exists for EDS; however, patients should be advised regarding the type and extent of physical activity so as to prevent self-trauma. Furthermore, non-weight-bearing muscular exercises done under

physician guidance can increase muscular strength and improve joint stability. Surgical wounds heal less rapidly therefore stitches need to be in place for a longer period of time. Finally, patients with EDS need to take care of their skin and eyes to prevent sunburn and nearsightedness. The role of vitamin C is not proven, but it is given nevertheless.[11]

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