Case Report

Ehlers Danlos syndrome in two siblings

M. N. Das, A. Ghorpade, P. Mercy, T. K. Pandey*, R. Sharma*
Department of Dermatology, Venereology and Leprology, *Department of Medicine, JLN Hospital and Research Centre, Bhilai Steel Plant, Bhilai, India

Address for correspondence: Dr. M.N. Das, 4-A/41/7, Bhilai, Chattisgarh, India. E-mail: sysantosh@yahoo.com

ABSTRACT

Two cases of Ehlers-Danlos syndrome affecting two real brothers are being reported. Both of them presented with features of atrophy and hyperextensibility of skin, hypermobility of joints and scarring at the sites of trauma. The elder brother also had kyphoscoliosis and hypogonadism with testicular failure.

Key Words: Ehlers Danlos syndrome, Kyphoscoliosis, Hypogonadism

INTRODUCTION

Ehlers-Danlos syndrome is a group of generalized disorders of connective tissue characterized by hyperextensibility of skin, hypermobility of joints and fragility of skin and blood vessels.[1] More than ten clinical types have been described on the basis of clinical, genetic and biochemical information.[2] The basic defect in this disease is abnormality in the biosynthesis of collagen. It may be inherited as an autosomal dominant, autosomal recessive or X-linked recessive trait.

CASE REPORTS

A 15-year-old boy (Case 1) born out of a consanguineous marriage presented with repeated scarring over both the knee and elbow joints since infancy. He was referred to us to rule out leprosy. His elder brother (Case 2), aged 17, also had similar skin lesions.

Cutaneous examination of both the brothers revealed cigarette paper scarring over the extensor aspects of both the knee [Figure 1] and elbow joints. The skin was soft and hyperextensible, particularly over the elbow joints [Figure 2]. The interphalangeal joints were hypermobile [Figure 3]. In addition, the younger brother had a high arched palate and the elder brother had kyphoscoliosis [Figure 4] along with bilateral soft, small-sized testes with decreased testicular sensation. He also had pseudotumor formation around the elbow joints. There was no evidence of purpura, ecchymosis, cyanosis, jaundice, digital clubbing or lymphadenopathy in both the brothers.

The fundus, routine blood tests, urine, coagulogram, blood urea, sugar and serum electrolytes were normal in both the brothers. X-ray skull, barium meal and enema did not show any abnormality in Case 1. Echocardiography of the elder brother did not reveal any abnormality. Left ventricular ejection fraction was 62 percent and there were no regional wall motion abnormality. All the valves were normal and pliable.
Testicular FNAC revealed decreased spermatogenesis, increased hyalinization and maturation arrest. Hormonal assay revealed serum cortisol at 8 am 12 ug/dl, testosterone 3.2 ng/ml, FSH 26 uI V/ml, LH 18 u IV/ml and serum prolactin 18 ng/ml. Skin biopsy taken from the atrophic skin lesion over the elbow revealed normal epidermis and scanty, whorled, disorderly dermal collagen. Special staining revealed relatively increased and irregularly oriented elastic fibers.

DISCUSSION

A few cases of Ehlers-Danlos syndrome were reported earlier in the Indian literature.\[3,4\]

In one case report, Singh et al\[5\] reported this syndrome in four generations. This genetic disorder has variable prognosis varying from mild to lethal. Its exact etiology is still unknown. Deficiency of some enzymes has been detected in certain types. Several congenital defects like flat feet, myopia, lop ears, prognathism may be associated with this condition.\[6\] Abnormal bleeding tendency and mitral valve prolapse have also been reported.

The treatment is mainly preventive with avoidance of trauma to prevent cosmetic disfigurement. Vit C has been tried in certain types with some benefit. Plastic surgery can offer some help for the cosmetic improvement. The purpose of reporting these two cases

Figure 1: Cigarette paper scarring over the knees (case 1)

Figure 2: Hyperextensible skin over the elbow joints (case 1)

Figure 3: Hypermobile interphalangeal joints (case 1)

Figure 4: Kyphoscoliosis (case 2)
is to create awareness about this condition so that it can be diagnosed early and parents can be counseled at the earliest. Although association of kyphoscoliosis has been reported earlier\textsuperscript{[7,8]} hyper-gonadotrophic hypogonadism along with testicular failure has not been mentioned, though here, it could be coincidental.

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