Case Report

EHLERS-DANLOS SYNDROME-TYPE II

Surg Cdr PLK DESYLVA ⁺, Surg Capt KM SHAH ⁺,
Lt Col S BHATTACHARYA [#]

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Introduction

The Ehlers Danlos syndromes (EDS) are a group of disorders of collagen metabolism characterised by skin hyperextensibility, joint hypermobility & sometimes systemic involvement. The severity ranges from mild to lethal. There are eleven types of EDS identified based on the clinical features; however sometimes these clinical features do not fit into a distinct subtype [1,2].

EDS is an inherited disorder transmitted by autosomal dominant, autosomal recessive or linked recessive genes. The basic defect is in deficient biosynthesis of type I or type III collagen fibres [3]. These disorders are rare, some cases have been reported from India [4-6]. We are reporting one such case of EDS type II (mitis).

Case Report

A nineteen-year-old male reported for a trivial skin ailment. During examination he was incidentally noticed to have hyperextensible skin. History revealed skin hyperextensibility since early childhood, presence of easy bruisability & delayed wound healing but there was no dislocation of joints or relevant family history. His development milestones were apparently normal.

Examination revealed a tall individual of height 178 cms & distance between fingertips 187 cms. There was no dolichostenomelia, ratio of upper to lower segment being 1.19 (upper segment 97 cms, lower segment 81 cms).

His eyes were widely spaced with a broad nasal bridge (Fig 1). The skin was hyperelastic & could be pulled away easily (Fig 2) but quickly returned to normal position when released. There were no loose folds of skin or striae. One atrophic scar was present over the left knee. The fingers were long with increased extension, however the thumb sign and wrist signs were negative. The gait, eyes, palate and foot arches were normal and there were no bony defects, varicosities or hernias. General & systemic examination revealed no abnormality.

Haemogram, urinalysis, biochemical parameters, ECG, echo cardiography & chest X-ray, were within normal limits. Skin biopsy revealed no significant abnormality.

He was advised long term treatment with oral ascorbic acid one

gram per day.

Discussion

EDS type II or mitis type is a rare disease that has an autosomal dominant inheritance [1-3], however in this case no family history was available. The exact mechanism of biochemical defects is obscure however abnormalities occur in dermal collagen fibres which may become small, disorganised, having irregular contour & cross section shows a flower like arrangement [3]. However, in most cases of EDS types I & II,

Fig 1 : Widely spaced eyes with broad nasal bridge

^{*} Classified Specialist (Derm, STD & Leprology), INHS Sanjivani, Naval Base, Kochi 682004, ⁺ Senior Adviser & Head of the Department of Dermatology, STD & Venereology, [#] Classified Specialist (Pathology), INHS Asvini, Colaba, Mumbai 400 005.

Fig. 2 : Hyperelastic skin that is easily pulled away and returns to normal when released

there is no obvious alteration in collagen or elastic fibres as observed in this case [7].

The disease is characterised by hyperextensible skin which when released returns quickly to its normal position as observed in this case. Easy bruisability & delayed wound healing leaving atrophic scars was observed in this case. Characteristic facial appearance with wide spread eyes & wide nasal bridge [1,3] was also observed, however epicanthic folds were not present.

EDS type II is a milder form than EDS type I (gravis) where hypermobility & subluxation of joints, hypotonia and other musculoskeletal defects like scoliosis and pes planus, hernias and varicosities may occur [3]. This case had increased extension of fingers, but no other abnormality.

Cutis laxa, pseudo xanthoma elasticum, osteogenesis imperfecta, renal tubular acidosis, medullary sponge kidney, osteolysis of terminal pharynx and Marfan's syndrome have been associated with EDS type I [1]. Although this individual was tall and appeared Marfanoid there was no dolichostenomelia and other skeletal deformities eg pectus excavatum or carinatum, pes planus, scoliosis, dislocations etc, Thumb and wrist signs were negative. There were no other eye, cardiovascular or respiratory signs to suggest Marfan's syndrome.

Physical and mental development is normal in EDS Types I and II as observed in this case. Life expectancy is also normal as opposed to some of the other types of EDS [1-3].

Ascorbic acid in the dose of 1 gram per day is reported to be beneficial in cases of EDS type I [8] and hence has been exhibited. Plastic surgery may help remove redundant skin. Appropriate treatment for systemic involvement is required.

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