EHLERS-DANLOS SYNDROME

Prasanta Basak, A J Kanwar, Surrinder Kaur and Arti Nanda

A female patient had Ehlers-Danlos syndrome type II since infancy, manifesting with hyperextensible skin and cigarette paper scars at the sites of trauma. Treatment with vitamin C 1 gm a day seemed to be useful.

Key words : Ehlers Danlos syndrome, Hyperextensible skin.

Ehlers-Danlos syndrome (EDS) was first described by Van Meckeren in 1682.[1] It is an inherited generalized disorder sharing phenotypic features including hyperextensible skin and joints, poor wound healing, easy bruisability and occasional fragility of large blood vessels and viscera.[2] At least 11 varieties of EDS have been recognized on the basis of clinical and biochemical criteria.[3] The syndrome may be inherited as an autosomal dominant, autosomal recessive or as an X-linked recessive trait. In the present communication we report a case of EDS recently seen by us.

Case Report

A 8½-year-old female student presented with a history of easy bruisability of the skin and poor wound healing since infancy. Minor trauma during play resulted in gaping wounds, many of which had to be sutured. The resultant scars were thin and cigarette paper like. The child had been born normally at term without any complications. Mental and physical development was normal. She was the youngest of 3 sibs; there was no family history of similar disorder and the parents were non-consanguineous.

Examination revealed cigarette paper scarring on the forehead, elbows, fingers, knees and shins, at the sites of previous trauma (Fig. 1). The skin was soft, thin and hyperextensible (Fig. 2). There was no evidence of purpura or ecchymosis and the teeth were normal. Hypermobility of the joints was not marked and was

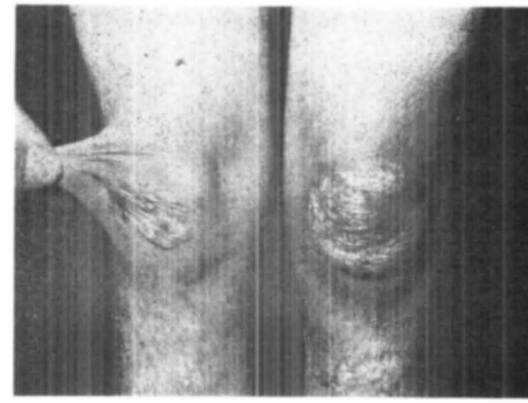

Fig. 1. Cigarette paper scars on the knees.

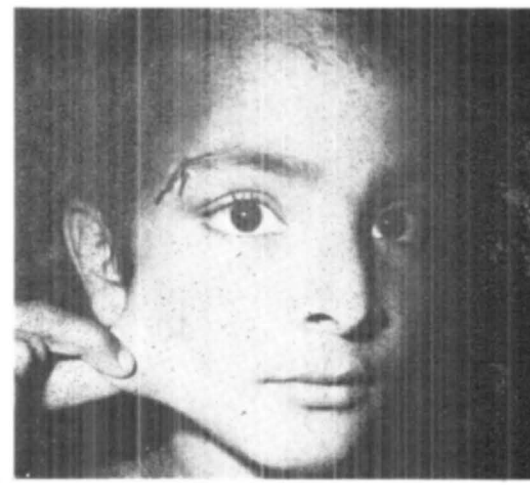

Fig. 2. Hyperextensible skin of the cheeks.

From the Department of Dermatology, Post-Graduate Institute of Medical Education and Research, Chandigarh-160 012, India.

Address correspondence to : Dr A J Kanwar

restricted only to the small joints of the hands. Systemic examination was non-contributory and the fundus was normal.

Routine blood and urine investigations were normal. Coagulation studies and platelet adhesion and aggregation tests did not reveal any abnormality. Skiagram of the skull was normal. A skin biopsy from the forearm revealed normal epidermis. The thickness of the dermis was about one half of that of normal skin. The collagen was fibrillary and appeared normal. Special stains revealed a relative increase in elastic fibres.

Comments

EDS has earlier been reported from India.[4-6] Singh et al[6] reported the occurrence of this syndrome in 4 generations. All had type II (mitis) variety with autosomal dominant transmission.

Eleven types of EDS have been formulated according to the clinical and biochemical criteria, though many patients do not meet the criteria for any of the 11 types.[7] Marked hyperextensibility of the skin, easy bruisability and joint hypermobility, in the absence of other vascular or skeletal abnormalities characterize types I, II and X. The clinical features of type II (mitis) are milder than that of type I (gravis). Type X (fibronectin deficient) which may clinically resemble types I and II is diagnosed biochemically by the presence of abnormal fibronectin in association with platelet function defects. Clinically our patient was categorized as type I EDS, the negative family history being accounted for by the variable penetrance of the autosomal dominant gene. Histopathologically, the increase in dermal elastic tissue with the disorderly arrangement of collagen was consistent with EDS.[8] In addition the dermal thickness was one-half that of the normal skin which is believed to be diagnostic of type I EDS.[9] Skin thickness is normal in type II EDS.[9] Though fibronectin studies could not be done, platelet function studies revealed no abnormalities which ruled out the possibility of our case being type X.

Type III (benign hypermobile type) was not considered as the skin is minimally affected in this condition and there was no marked hypermobility of the large joints. Type IV (ecchymotic type) is clinically distinctive with peaked nose, thin lips and with hands and feet having a premature ageing appearance. The skin is thin with prominent underlying veins. Bruising is a constant feature and the friability of the tissues make the patient prone to rupture of the aorta, bowel or uterus. Our patient did not have the above characteristic features. Type V is transmitted as an X-linked trait. As our patient was female, this was not considered. Type VI (ocular type) and type VII (arthochalasis multiplex congenita) were easily ruled out clinically as our patient did not have any eye signs such as keratoconus or intraocular haemorrhage, nor was there any extreme joint laxity resulting in congenital bilateral hip dislocation. Type VIII was easy to exclude as it is associated with periodontal disease resulting in resorption of the gum and loss of teeth. Type IX is characteristically associated with bony occipital horns diagnosed radiologically, which were absent in our patient. Type XI (familial joint instability) is characterized by normal skin with marked joint laxity and hence was not considered.

EDS type VI may respond to oral ascorbic acid,[7] but it can also be empirically administered in other types of EDS in doses of 2 to 4 gm per day.[10] Children are to be given appropriately lower doses. This medication should be taken with a full glass of water or other fluid. Ascorbic acid is ordinarily safe, although it is metabolized by the body to oxalic acid. Since this compound may be found as a part of some kidney stones, it is important to ensure that the patient has not had previous kidney stones and does not have a history of kidney stones in the family. The

improved wound healing in response to vitamin C is due to the enhanced secretion of collagen.

Because the skin is already thin in EDS, premature ageing of the skin is an added risk. Since the sun markedly accelerates this process, it is prudent to protect exposed skin from the sun. Sunscreen should be used daily but especially when sun exposure is contemplated. Wearing a wide brimmed hat is a simple and effective measure.

Protection of the skin from trauma is important owing to the cosmetic disfigurement that may result. This ordinarily occurs in the first 3 or 4 years of life. Injury is especially risky as the child attempts to learn how to walk on loose, unstable joints. During this period, extra care is necessary to prevent falls. Furniture should be appropriately padded and any obstacles such as loose carpets should be removed. Lacerated wounds or any surgery performed need meticulous haemostasis and pressure dressing. Angiographic procedures are hazardous owing to blood vessel fragility. Life expectancy is normal.

Our patient was put on oral vitamin C 500 mg twice daily. When seen 3 months later, she did not have any fresh wounds and there was no clinical evidence of bruising.

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