CASE REPORTS

TWO CASES OF EHLERS-DANLOS SYNDROME

BY

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The Ehlers-Danlos syndrome is a congenital abnormality of the skin, of which the following may be considered the chief characteristics: (1) Abnormal friability of the skin and subcutaneous tissue; (2) loose and hyperelastic skin; (3) hyperextensibility of joints; (4) the presence of nodules of various types in the skin; (5) well-marked epicanthic folds.

The clinical picture is not invariably complete and, as in other congenital conditions, the incidence of associated congenital abnormalities is higher than in normal subjects. The friability of the skin is nearly always evident from the history of these patients. They are described as being subject to cuts or even large gaping wounds from the most trivial injuries, and a glance at the skin of their knees or foreheads will show scarring of an unusual degree. The laxity of the skin is easily demonstrable as a rule by the ease with which a fold may be drawn away from the deeper tissues, and its lack of attachment is suggested by the absence of skin creases which, according to Le Gros Clark (1945) are due to the attachment of the skin to deep fascia by strands of collagen fibres.

Observation of the wounds in these patients shows an unusual freedom of the fat lobules, which are easily detached from the wound surface, and it is noticed that when sutures are being placed in such wounds it is necessary to space them close together, owing to the marked degree of retraction of the skin between the sutures. There is an abnormal tendency in these patients to bruise easily, and this has been described as due to abnormal friability of the blood vessels. In fact, no abnormal histological appearance of the vessel walls has been described and it may well be that the cause of the vascular damage is due to lack of supporting tissue. There is no abnormality of the platelet count.

Hyperelasticity of the skin is demonstrated by the rapid return to its original unwrinkled state when tension is released. It may be mentioned that abnormal looseness of the skin can occur as an isolated abnormality unrelated to the Ehlers-Danlos

syndrome, when it should be referred to as cutis hyperelastica.

Hyperextensibility of the joints is presumably due to an abnormality of the mesodermal tissue in the joint capsule analogous to that of the skin. It is not associated with any radiological alteration in the joints.

Nodules in the skin, which are described in the majority of these patients, are of two main types, though there appears to be some confusion in the terminology. The subcutaneous, freely movable nodules described by Parkes Weber and Aitken (1938) and Tobias (1934) appear histologically to be small lipomata.

The soft, raised thickenings of the skin which follow bruising are most probably due to organization taking place in the haematomata. Ronchese (1936) states that these nodules contain numerous foreign-body giant cells.

Marked epicanthic folds, which occurred in both our cases described below, have also been noted by Benjamin and Weiner (1943) and others, and could be included as one of the chief characteristics of the syndrome.

Pathology

Ronchese (1936) has described sections of the skin which reveal thinning and torsion of the collagen fibres, and Burrows and Turnbull (1938) found that the collagen bundles were less regularly arranged and less closely packed than in the normal subject. There appears to be an increase of elastic tissue, which is arranged in irregular and coarse bundles. The individual elastic fibres are rough, and tend to be curled up at the edges.

Case Histories

Case 1. A boy, aged three and a half years, weighed 27 lb. His mother stated that 'when he falls he doesn't cut himself, he just tears open,' and that he bruised very easily.

Case 2. A girl, aged nine years, weighed 51.5 lb. She has always bruised easily and on six occasions has suffered large cuts in her left knee.

Both patients had been frequent visitors to a hospital casualty department for suturing of superficial injuries. Their mental and physical development had otherwise been normal, and no abnormal physical signs were elicited other than those described. In neither case was there consanguinity of the parents, nor was there any family history of the condition. All the characteristic features mentioned above were present in both patients. Hyperextensibility of the joints was marked, and the girl was able to touch her heels with the back of her head, when supported, without effort.

The boy had numerous scars on both knees, forehead, and chin. The scars had the typical appearance described in this condition, being thin, papyraceous, and atrophic, and the epicanthic folds mentioned above were well marked. The bleeding time and clotting times, platelet counts, and tourniquet tests were all within normal limits in both patients.

The histological report on a biopsy specimen of the girl is as follows.

SECTION 1 (JUNCTION OF SCARRED AND NORMAL SKIN). The epidermis is atrophic and the corium condensed, resulting in narrow bundles of collagen disposed almost entirely in a horizontal plane. In

one area there is a complete absence of elastin in the pars papillaris and subjacent superficial part of the pars reticularis.

SECTION 2 (NODULE). There is no appreciable pathological change.

SECTION 3 (SMALL ARTERY). The vessel wall is cut almost longitudinally, and is surrounded by loose areolar tissue into which there has been haemorrhage attributable to the biopsy.

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(For illustrations to this article see Plate VII.)

(A) Case 2, showing exaggeration of the inner canthus; (B to E) Case 1, showing (B) exaggeration of the inner canthus and scarring of the nose and forehead, (C) loose and hyperelastic skin, and scarring of the knees, (D) hyperextensibility of joints, (E) typical thin, papyraceous, and atrophic scarring of knees, and multiple bruises on legs.