A CASE OF EHLERS-DANLOS DISEASE

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Although during the last few years a number of cases showing the syndrome described by Ehlers in 1901 and Danlos in 1908 have been recorded, a further case which has come to our notice seems of sufficient interest to merit publication.

The classical features of this disease are: (1) abnormal elasticity of the skin; (2) friability of the skin and blood vessels, resulting in large subcutaneous haematomata and gaping wounds from trivial injuries; (3) hyperextensibility of the joints; (4) papyraceous scars, sometimes associated with pseudo-tumours; (5) tiny movable subcutaneous nodules, probably of a lipomatous nature; (6) a familial incidence of one or more of the above features.

Case Report

A., a man aged 28, was admitted to an Army hospital following a trivial fall over a doorstep on December 31, 1939. He felt no immediate pain or disability, but on returning to his billet found difficulty in removing his trousers owing to an enormous swelling around the left knee. There was also a gaping wound over the right tibia, with a haematoma the size of an orange below it, together with swelling and discoloration around the right ankle. The swelling over the ankle did not pit on pressure. The wound over the right shin was sutured and the haematoma of the left knee incised, "about a pint of blood clot" being evacuated from it.

On transfer to us twenty-one days later he still had a fluctuating swelling, measuring about three by four inches, below the left patella, and an indefinite mass on the anterior aspect of the right leg, above which was a wide discoloured scar, in shape a wide V, from a recent wound. The discoloration of the ankle had disappeared. His general appearance was that of a healthy well-built young man. His skin had a peculiar velvety look and feel, being finely "rippled" on close inspection; and over the thorax, arms, and face it showed a marked elasticity (Figs. 1 and 2). Elsewhere some

FIG. 1.

FIG. 2.

undue laxity was present, but not to the same degree. He was able to pull the skin over his sternum up to his mouth.

In the subcutaneous tissues beneath the chin was one movable fatty nodule the size of a small pea, such as described by Parkes Weber and Aitken (1938). None of the pseudo-tumours (described by Ronchese (1936) as "molluscoid, empty, and raisin-like") was present in any of the numerous old

scars, which, however, had the characteristic tissue-paper feel and appearance. Where linear these scars were all unusually wide. Flexibility of the large joints was marked, though hyperextensibility of the thumbs or fingers, or both, to which this sign is not infrequently confined, was only slight. No other physical abnormalities were discovered on examination. Investigation of the blood showed no chemical or cytological abnormalities, though platelets were more numerous than usual; bleeding and clotting times were within normal limits. Radiographs of skull and long bones were also normal. No biopsy was performed.

The patient's history was that from early boyhood he had been unable to play games, as severe bruising or wounds were apt to result from trivial injury; otherwise his health had been good and the bleeding time was not prolonged. There was no tuberculosis, consanguinity, or gross malnutrition as a possible aetiological factor. The family history was interesting, inasmuch as various members suffered from the same disease, the females being affected as well as the males, as the accompanying family tree shows. In this the

FIG. 3.

affected members of the family are shown in black. The ages of the children of the next generation range up to 12 years; all those members of the family showing the disease have done so from the age of 3 to 4 years or even earlier.

The points of interest about this case appear to us to be as follows:

1. That what has hitherto been regarded as a curiosity of medicine, and either a source of profit to the "sufferers" (it seems probable that many of the elastic men and contortionists at side-shows may be affected with this disease) or a matter of slight inconvenience, will prove a grave disability if the patient is allowed on full active service with the fighting Forces. Owing to the easily overlooked signs of the disease and the generally good health of the sufferer it may easily be missed, and so lead to dire results from what should be relatively small injuries.

2. So far as we have been able to ascertain in the English and American literature on the subject, no investigations have been made into the familial incidence of the disease. It is possible that the collection of "family trees" may throw light on its mode of transmission and probable appearance in particular members of the family group.

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Dr. Charles Slater, consulting bacteriologist to St. George's Hospital, who died on March 15, aged 83, bequeathed £10,000 to St. George's Hospital for the purpose of teaching bacteriology or for research work in that science, £5,000 to Manchester University for the equipment and maintenance of the laboratories, and £4,000 to Cambridge University for teaching or research work in medical science.