

## SELF ASSESSMENT ANSWERS

### Unusual scars in a young female patient

**Q1: What other signs and symptoms would she present with?**

- Skin—skin feels velvety to touch and is soft. Stretches and springs back easily. Atrophic “cigarette paper” scars.
- Musculoskeletal—pronounced joint hypermobility.
- Others—easy bruising and poor muscle tone.

**Q2: What could be the cause of scars across her back?**

Chickenpox scars when she was 10 years of age.

**Q3: What is the diagnosis?**

Ehlers-Danlos syndrome type 1.

**Discussion**

Ehlers-Danlos syndrome is a group of genodermatoses affecting connective tissues predominantly collagen fibres that are structurally abnormal.<sup>1</sup> Defects in elastin<sup>2</sup> and fibronectin<sup>3</sup> metabolisms have been found in Ehlers-Danlos syndrome type IV and V respectively. Typically the dermal collagen is disorderly, sparse, and vortex-like. The mode of inheritance is variable and can be autosomal dominant, recessive, and X-linked recessive. Table 1 summarises the various types of Ehlers-Danlos syndrome.

This patient has Ehlers-Danlos syndrome type 1. The clinical features are predominantly cutaneous and musculoskeletal.

Hyperextensible skin with normal springiness is a characteristic feature. Laxity of skin occurs later in life. The skin is fragile such that minor trauma or even cutaneous infections lead to gaping “fish mouth” wounds, which heal slowly leaving behind atrophic “cigarette paper” scars. Bruising is also an important feature. Molluscoid pseudotumours, which are spongy and blue-grey in appearance, may form in scars and over pressure points. These facts should be taken into consideration when surgical procedures are planned.

The musculoskeletal manifestations include joint laxity with recurrent dislocation and postural abnormalities such as kyphoscoliosis.<sup>4</sup> There is weakness of supporting structures with increased risk of inguinal hernias, eventration of the diaphragm, and torsion of the stomach.<sup>5</sup> Prematurely ruptured fetal membranes with premature births and uterine prolapse in pregnancy have also been described. Life expectancy, physical, and mental developments are normal.

**Final diagnosis**

Ehlers-Danlos syndrome type I with chickenpox scars.

**References**

- 1 **Pope FM**, Narcisi P, Nicholls AC, *et al*. COL3A1 mutations cause variable clinical phenotypes including acrogeria and vascular rupture. *Br J Dermatol* 1996;**135**:163–81.
- 2 **Christiano AM**, Uitto J. Molecular pathology of the elastic fibres. *J Invest Dermatol* 1994;**103**:53–75.
- 3 **Arneson MA**, Hammerschmidt DE, Furcht LT, *et al*. A new form of Ehlers-Danlos syndrome. Fibronectin corrects defective platelet function. *JAMA* 1980;**244**:144–7.
- 4 **Beighton P**. *Hypermobility of joints*. Berlin: Springer-Verlag, 1983:178.
- 5 **Champion RH**, Burton JL, Burns DA, *et al*. *Rook, Wilkinson, Ebling textbook of dermatology*. Vol 3. 6th ed. Oxford: Blackwell Science, 1998:2034.

**Table 1** The various types of Ehlers-Danlos syndrome

Type	Inheritance	Clinical features
I	AD	Soft, velvety, hyperextensible skin with easy bruising, cigarette paper scars, and pseudotumours
II	AD (Rarely AR)	Milder but similar to type I
III	AD	Joint involvement with early osteoarthritis and minimal skin abnormality
IV	AD	Increased susceptibility to great vessel and bowel rupture. Cutaneous features as in type I with normal joints.
V	XLR	Similar to type II with more ecchymoses
VI	AR	Cutaneous features as in type I but with ocular involvement such as keratoconus and ocular fragility.
VII A + B	AD	Prominent musculoskeletal features with floppy infant, congenital hip dislocation, and short stature. Normal scarring.
VII C	AR	Pronounced hypermobile joints with very soft, fragile skin and easy bruising.
VIII	AD	Similar to type II with severe periodontitis
IX	XLR	Hyperextensible skin with short arms, occipital horns, broad clavicles, and bladder diverticula.
X	AR	Similar to type II

AD, autosomal dominant; AR, autosomal recessive; XLR, X-linked recessive (adapted from Champion *et al* (page 2034).<sup>5</sup>