Picture Story

Classic Ehlers-Danlos syndrome

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(Key words: Classic Ehlers-Danlos syndrome; skin hyperelasticity; joint hypermobility)

We report a 12 year old boy with skin hyperextensibility (Figures 1a & 1b), joint hypermobility (Figure 2) easy bruising (Figure 3) and a papyraceous (cigarette paper) scar (Figure 4). He was born to non-consanguineous parents with an uneventful birth. The mother had noted generalized floppiness and skin laxity since birth. Joint hypermobility and easy bruising with poor wound healing was appreciated later. None of the family members have similar abnormalities.

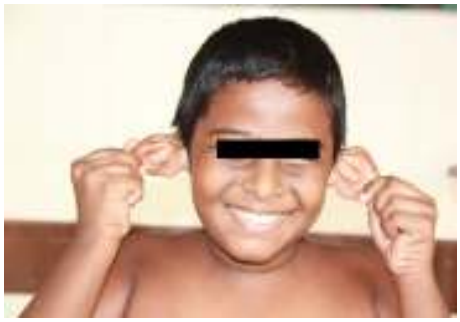

Figure 1a: ***Skin hyperextensibility***
**Permission given by parents to publish photograph*

Figure 1b: ***Skin hyperextensibility***

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Figure 2 Joint hypermobility

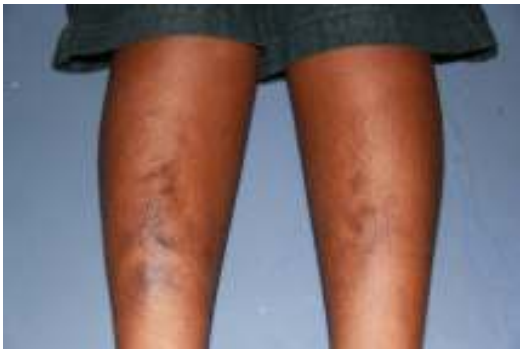

Figure 3: ***Easy bruising***

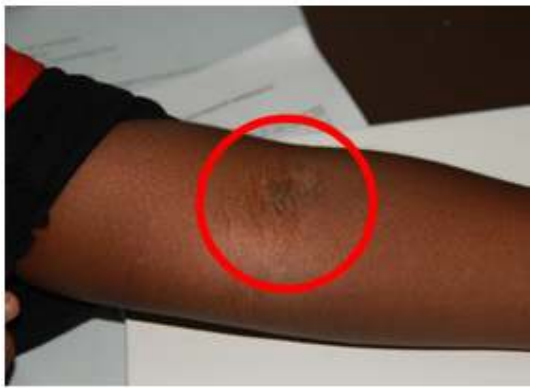

Figure 4: ***Papyraceous (cigarette paper) scar***

Discussion

Classic Ehlers-Danlos syndrome (EDS) is a heritable connective tissue disorder characterized by skin hyperextensibility, fragile and soft skin, delayed wound healing with formation of atrophic scars, easy bruising, and generalized joint hypermobility. It comprises Ehlers-Danlos syndrome types I and II, but it is now apparent that these form a continuum of clinical findings and differ only in phenotypic severity[1]. The prevalence of classic EDS has been estimated to be 1:20,000. It is currently estimated that approximately 50% of patients with a clinical diagnosis of classic Ehlers- Danlos syndrome harbor mutations in the COL5A1and the COL5A2 gene, encoding the 1 and the 2-chain of type V collagen, respectively[1].

Reference

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