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Case Letter

Elastic oddity: A case report of a rare occurrence of elastosis perforans serpiginosa in Down's syndrome

Gowrappala Shanmukhappa Asha, Ujwal Manjunatha Shetty

Department of Dermatology, Venerology, Leprosy, Bangalore Medical College and Research Institute, Bengaluru, India

Corresponding author: Ujwal Manjunatha Shetty, MD, MBBS, E-mail: ujwal.hm@gmail.com

Sir,

Elastosis perforans serpiginosa (EPS) is a rare disorder classified as a primary perforating dermatosis. It is a benign condition, where altered elastic fibers are extruded through the papillary layer and epidermis by transepithelial elimination. Focal dermal irritation is believed to induce the formation of a transepidermal channel. It is associated with connective tissue disorders like Ehlers-Danlos syndrome, Marfan syndrome, cutis laxa and osteogenesis imperfecta. Drug induced EPS is associated with D-penicillamine [1,2].

It is characterized clinically by papules and keratotic, desquamative, normochromic or erythematous plaques, usually affecting the face, neck, trunk and upper limbs. Histopathology is the gold standard for diagnosis where thickened, vertically arranged elastic fibres are seen penetrating the skin forming narrow canals [1,2].

There is no standard therapy; however, successful treatment has been reported with the use of cryotherapy with liquid nitrogen, isotretinoin, tazarotene, imiquimod and CO_2, Er: YAG and pulsed dye lasers. Spontaneous resolution is reported in a minority of cases [1].

Here we present a 17-year-old, known case of Down's syndrome, male patient with bizarrely arranged papules, in whom we diagnosed EPS.

A young, 17 year old, male patient with Down's syndrome complained of asymptomatic skin lesions on both his forearms that had appeared 2 years back. He did not report any current or past similar complaints. He did not have any other signs.

Dermatological examination revealed multiple hyperpigmented umbilicated papules, with trailing linear scars, symmetrically arranged in serpiginous pattern on posterior aspect of bilateral forearms (Figs. 1a and 1b).

Histopathology revealed transepidermal channels and clumps of thick, coarse elastic fibres in the dermis with lymphocytic infiltrate (Fig. 2). Van Gieson staining showed vertically oriented coarse elastic fibres.

A diagnosis of EPS was confirmed. But the patient refused all treatment and was lost to follow-up.

EPS is A perforating dermatosis in which the material extruded through the epidermis is derived from elastic fibres in the upper dermis. It is a rare skin disease classified as a primary perforating dermatosis similar to reactive perforating collagenosis, perforating folliculitis and Kyrle disease. The first description of EPS was published in 1927 by Fisher), as a disease with hyperkeratosis. In 1953, Lutz described the morphology of EPS and named it serpiginosa follicular keratosis. In 1955, Miescher showed that the extruded material contains elastin and suggested the term elastomer intrapapillare perforans veruciforme. Dammert and Putkonem changed into EPS in 1958 [1,3].

EPS usually presents between 5 and 20 years of age, but the age can range from 5 to 89 years [2]. More than 75% of the affected cases are male, and there appears to be no race predilection [1-3].

Exact etiopathogenesis is still elusive, but focal inflammation in the dermis of mechanical origin,

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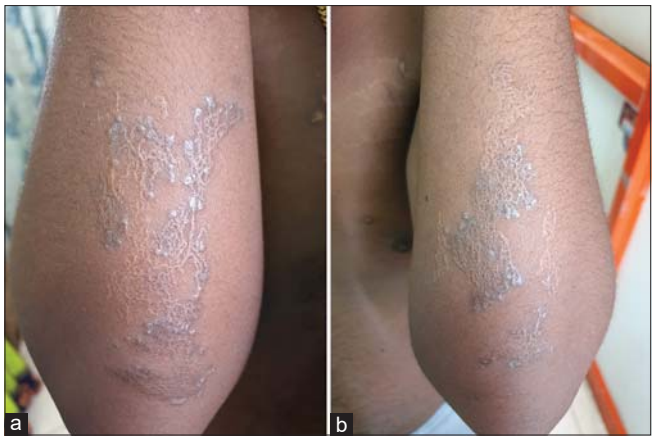

Figure 1: (a) Umbilicated papules arranged in a bizzare serpiginous pattern on the right forearm, (b) Umbilicated papules arranged in a bizzare serpiginous pattern on the left forearm.

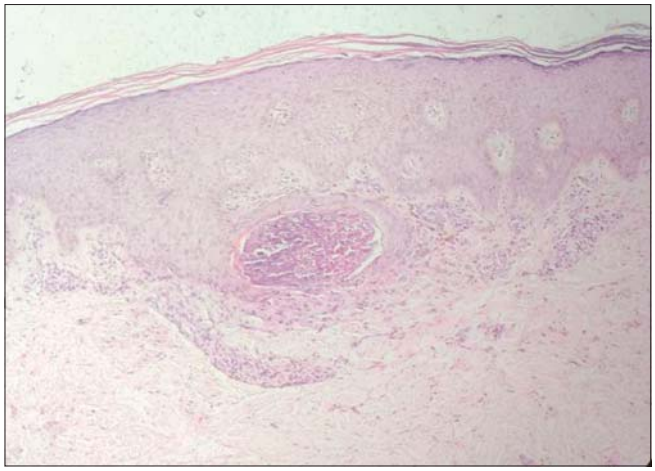

Figure 2: Histopathology showing clumps of thick, coarse elastic fibres in the dermis with lymphocytic infiltrate.

is believed to induce the formation of epidermal channels to expel abnormal elastic fibers. Fujimoto et al. found that interactions between elastin peptides and their 67kDa receptors expressed in the epidermis surrounding the transepidermal elimination channel may participate EPS pathogenesis [1].

EPS has three distinguished types:

1. Idiopathic
2. Reactive, associated in 25% of cases with connective tissue diseases such as Ehlers-Danlos syndrome, cutis laxa, Marfan syndrome, pseudoxanthoma elasticum and osteogenesis imperfecta among others, as well as Down's syndrome
3. Drug-induced by D-penicillamine

EPS usually presents with erythematous as well as skin-colored papules or plaques with keratotic surface which are grouped together and form serpiginous or annular lesions is usually observed. The middle of each lesion is umbilicated from which the material containing damaged elastic fibers are extruded. The lesions are mostly located on the arms, face and neck. This a disease with a slow course and in some cases a spontaneous regression of lesions in the period from 6 months to 5 years. Scarring can be seen [1,2,4].

Histopathology is the gold standard for diagnosis. It shows transepidermal or perifollicular channels that extend from the dermis in a linear or spiral pattern, containing a mixture of eosinophilic elastic fibers, basophilic debris and inflammatory cells. The elastic fibers are thick and numerous, causing foci of chronic inflammation in the upper dermis, thus justifying their extrusion. The surrounding epidermis can be acanthotic [1-3].

Differential diagnoses include granuloma annulare, tinea corporis, annular sarcoidosis, cutaneous calcinosis and porokeratosis of Mibelli.

Even though various modalities of treatment have been advocated, but the management is difficult with limited success and there is no standard treatment. Outland et al. described remission of the condition following treatment with 0.1% tazarotene gel [5] success after 10 weeks of therapy with imiquimod by Kelly et al. Pulsed dye, CO_2 and Er: YAG lasers have been tried successfully in anecdotal cases, but Saxena et al. didn't yield same results [1].

EPS is a rare and interesting disorder with characteristic clinical and histopathological picture. Although it is asymptomatic but as the lesions are chronic it is very distressful to the patient. Inspite of various trials effective modality of the treatment is still lacking.

Consent

The examination of the patient was conducted according to the principles of the Declaration of Helsinki.

The authors certify that they have obtained all appropriate patient consent forms, in which the patients gave their consent for images and other clinical information to be included in the journal. The patients understand that their names and initials will not be published and due effort will be made to conceal their identity, but that anonymity cannot be guaranteed.

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