

Case Report

Elastosis Perforans Serpiginosa in a Case of Down's Syndrome: A Case Report

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Abstract: Elastosis perforans serpiginosa (EPS), is a rare primary perforating dermatosis characterized by trans epidermal elimination of fragmented elastic fibres, clinically presenting as hyperkeratotic papules. EPS is classified into three types: (1) Idiopathic (2) reactive, which is associated with systemic conditions including Down's syndrome, osteogenesis imperfecta, Ehlers Danlos syndrome, Marfan's syndrome, diabetes mellitus, chronic renal failure and connective tissue disorders like pseudoxanthoma elasticum (3) the one that is induced by D-penicillamine. We report a case of Down's syndrome with associated lesions that were histopathologically compatible with EPS.

Keywords: Elastosis perforans serpiginosa, Tran's epidermal elimination, Down's syndrome.

INTRODUCTION

Elastosis perforans serpiginosa (EPS) is the most distinctive of primary perforating disorders of dermal origin because it demonstrates the best example of trans epidermal elimination of elastic fibres. It is considered a benign condition, since there are no extra cutaneous manifestations. Clinically they present as hyperkeratotic papules usually affecting the upper extremities, face, neck and trunk in groups, arranged in circinate or serpiginous pattern, with predilection in males of 2nd decade of life. Histopathology is the gold standard for diagnosis. There are increased number of thickened elastic fibres in upper dermis and altered elastic fibres are extruded through the epidermis. We report a case of Down's syndrome with associated lesions that were histopathologically compatible with EPS.

CASE REPORT

A 31 year old male with Down's syndrome, who presented with asymptomatic hyper pigmented lesions in both forearms of 3 months duration.

There was no history of any material extruding from the lesion, trauma or any other precipitating factors.

A past history of congenital heart disease, Atrial Septal Defect (ASD) uncorrected with moderate

Pulmonary Arterial Hypertension and low moderate Tricuspid Regurgitation was present.

There was a past history of gouty arthritis of 1 year duration and he was on Allopurinol 100 mg intake.

He is youngest among two siblings, born to a non-consanguineous marriage. Other sibling was normal.

Dermatological examination revealed hyper pigmented annular papules about 4-5 mm in size arranged in an annular fashion in extensor aspect of both forearms, some of them showing keratotic plugs on the surface.



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FIG 1 &2: Hyper pigmented lesions forearm

General examination showed microcephaly ,slanting palpebral fissures ,almond shaped eyes, flat facies and sandal gap.

Histopathology of the biopsy specimen from a hyperkeratotic papule revealed hyperplastic epidermis. Follicular infundibulum was dilated with an overlying keratin plug. A trans epidermal channel was identified through which the basophilic nuclear debris and eosinophilic elastic fibres were eliminated mixed with necrotic keratinocytes and inflammatory cell infiltrate. No granuloma/no fungal elements/ no calcification was identified. Special staining with Elastic Van Gieson highlighted elastic fibres in the focus of trans epidermal elimination. With the above clinical and histopathological evidence, a diagnosis of EPS associated with Downs syndrome was made.

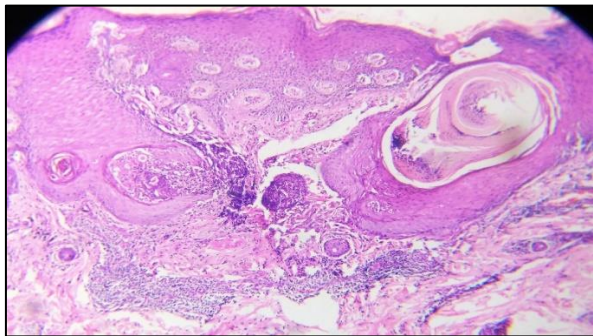


FIG 3: 40 X H&E

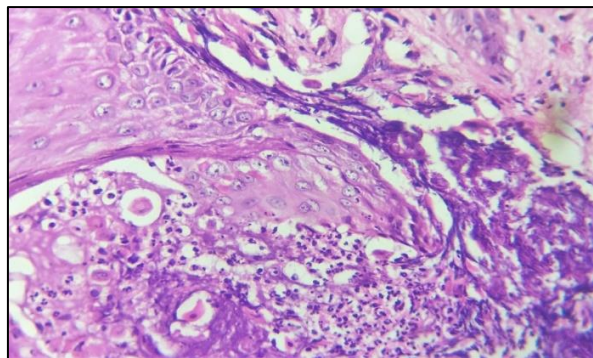


FIG 4: 40X H& E

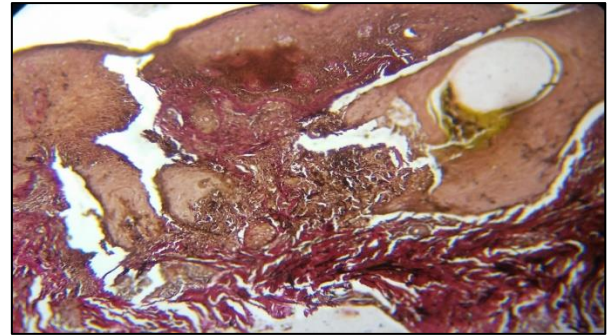


FIG 5: 40X ELASTIC VAN GIESON

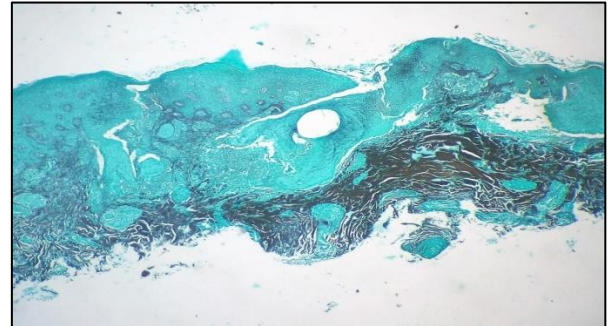


FIG 6: 10 X MASSON TRICHROME

DISCUSSION

Elastosis perforans serpiginosa (EPS) is a rare disorder classified as a primary perforating dermatosis. This group of diseases also includes reactive perforating collagenosis, perforating folliculitis and Kyrles disease.

EPS is characterised clinically by papules and keratotic plaques and histologically by focal elastosis of the dermis and Tran's epidermal elimination of abnormal elastic fibres.

The first case of EPS was most likely described by Fisher in 1927 and at that time, this condition was classified into a group of diseases with hyperkeratosis. In 1953, Lutz described the morphology of EPS and it was denominated as keratosis follicularis serpiginosa. In 1955, Miescher found elastin in the material eliminated Trans epidermally and the name elastoma intrapapillare perforans verruciforme was given. It was given the current term elastosis perforans serpiginosa in 1958 by Dammert and Putkonen.

The disease prevalence has not been established yet. Around 90% of patients develop symptoms of the disease prior to 30 years of age, the majority between 6 and 20 years of age, however age of onset may range from 5 to 89 years as reported in the literature.

EPS is classified into three types:

- Idiopathic
- Reactive,

which is associated with systemic conditions including Down syndrome, osteogenesis imperfecta, Ehlers Danlos syndrome, Marfans syndrome, diabetes mellitus, chronic renal failure and connective tissue disorders like pseudoxanthoma elasticum (3) induced by D-penicillamine.

The association between EPS and Down syndrome is not unknown; however factors present in this syndrome such as premature skin aging, joint hyperelasticity and acrocyanosis may suggest an underlying connective tissue disorder.

The etiopathogenesis is not fully understood. It is believed that focal inflammation in the dermis, which has a mechanical or biochemical origin, may induce the formation of epidermal and follicular channels to expulse abnormal elastic fibres considered irritants. Fujimoto *et al* reported interactions between elastin peptides and their 67kDa receptors expressed in the epidermis surrounding the trans epidermal elimination channel, which may participate in the pathogenesis of the disease.

The disorder may present with papules and erythematous, keratotic asymptomatic plaques grouped in an annular, serpiginous pattern, surrounded by satellite lesions. They have umbilicated centres from which dermal material is eliminated. The condition most commonly affects the upper limbs, face and neck. Lesions develop slowly, disappearing spontaneously in some cases after 6 months to 5 years, leaving superficial scarring.

The gold standard for diagnosis is histopathological examination of skin lesions, which is characterised by trans epidermal or perifollicular channels that extend from dermis in a linear or spiral pattern, containing a mixture of eosinophilic elastic fibres, basophilic material consisting of keratinocytes and inflammatory cells. The elastic fibres are thick and numerous causing foci of chronic inflammation in the upper dermis. The fibres are compacted, twisted and fragmented, which can be clearly seen in the staining for elastic fibres such as Van Gieson staining.

Clinically EPS must be differentiated from granuloma annulare, tinea corporis, sarcoidosis, cutaneous calcinosis and parakeratosis of Mibelli.

Various forms of treatment have been indicated. However management of the disease is difficult and there is no standard treatment.

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