

## Elastosis Perforans Serpiginosa in a patient with Down syndrome treated with imiquimod 5% cream

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### Abstract

*Elastosis Perforans Serpiginosa* (EPS) is a rare skin disease characterised by hyperkeratotic papules, transepidermal elimination of abnormal elastic fibres, and focal dermal elastosis. The aetiology is unknown, but an association with underlying systemic disorders, including Down syndrome has been described. Treatment is often difficult. A 45-year old man with Down syndrome presented with symmetrical annular elements on forearms and femora. The elements were erythematous with atrophic hypopigmented central healing and peripherally, infiltrated keratotic papules with desquamation. A punch biopsy showed the classical histopathologic features of EPS. We found no clinical signs of cerebrovascular or cardiovascular disease. We initiated topical therapy with imiquimod 5% cream once a day for 6 weeks followed by 3 times weekly for 4 weeks to a single element. As regression of EPS was observed and the patient tolerated the therapy well, treatment of other lesions was commenced, and further regression was seen.



Figure 1. *Elastosis Perforans Serpiginosa* on right forearm before start of treatment. Annular erythematous lesions with hyperkeratotic papules peripherally and central atrophy.

### Introduction

*Elastosis Perforans Serpiginosa* (EPS) is a rare skin disease with unknown etiology belonging to the group of perforating dermatoses.<sup>1</sup> The disease is associated with systemic disorders like Down syndrome, Ehlers-Danlos syndrome, Marfan syndrome<sup>2,3</sup> and it also exists in a drug-induced form by pencillamine.<sup>4</sup> EPS is characterized by increased elastic tissue in papillary dermis, inflammation and transepidermal elimination of abnormal elastic fibers.<sup>5</sup> This response is more generalised and prolonged in cases associated with Down syndrome. Clinically EPS is characterized by grouped hyperkeratotic papules and plaques with central heeling, often symmetrically located on upper and lower extremities, trunk and face. The treatment modalities are numerous and include cryotherapy, curettage, lasers, isotretinoin.<sup>6,7</sup> Long-term results are variable and treatment may cause discomfort, atrophy and scarring. Recently imiquimod therapy has been introduced to treat EPS.<sup>8</sup>

### Case Report

A healthy 45-year old man with Down syndrome presented with symmetrical annular elements on forearms and thighs varying in size from 1 to 8 cm (Figure 1). Small erythema-

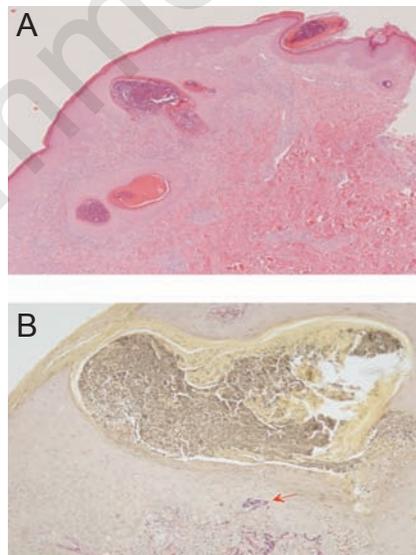


Figure 2. *Elastosis Perforans Serpiginosa* – skin from thigh. (A) HE staining. Epidermis is thickened and presents with transepidermal perforation channels containing inflammatory debris and degenerated elastic fibers. (B) Elastic van-Gieson staining; coarse elastic fibers enter the epidermis (arrow). Fibers loose stain, as they undergo transepidermal elimination.

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tous and hyperkeratotic papules appeared five years prior and progressed centrifugal to form a pattern of linear, annular and serpiginous lesions with central heeling and atrophy. Peripherally infiltrated reddish keratotic papules with central depression and overlying desquamation, was seen. The patient had been treated unsuccessfully with topical corticos-



Figure 3. The treatment result after topical therapy with imiquimod 5% cream, once a day for 6 weeks followed by 3 times weekly in a period of approximately 5 months. The elements became less inflamed and infiltrated.

teroids and systemic antifungal drugs before clinical examination at the Department of Dermatology. A punch biopsy from the thigh (Figure 2a,b) showed a histopathological picture compatible with *Elastosis Perforans Serpiginosa*.<sup>9</sup> There were transepidermal and transfollicular perforation channels containing inflammatory debris and degenerated elastic fibers. In the surrounding dermis the elastic fibers were thicker than normal and partly degenerated, however the content of elastic tissue was not significantly increased. An inflammatory infiltrate consisting of mainly mononuclear cells, histiocytes and some neutrophils was present in the dermis. The patient was further examined with full blood screen, blood pressure, echocardiogram and ultrasound of: heart, aa. carotis, aorta abdominalis and aa. ilicae and there was no signs of vascular disease.

In reference to Kelly and Purcell 2006, topical therapy with imiquimod 5% cream was initiated once a day for 6 weeks followed by 3 times weekly for 4 weeks. The clinical response after three months of Imiquimod treatment was promising and the treatment was continued 3 times weekly for further 2 months (Figure 3). Twenty milligrams of Imiquimod cream was used per treatment week. Skin lesions became less inflamed and infiltrated and further dissemination was stopped. The patient had no adverse effects.

## Discussion

This case report describes a patient with Down syndrome with EPS successfully treated with imiquimod 5% cream. EPS is a rare disease associated among others with Down syndrome and often runs a progressive course in this group. Differential diagnostic considerations include: dermatophytosis, granuloma annulare, reactive perforating collagenosis and porokeratosis. There can be considerable diagnostic delay, as in this case, where the patient had skin lesions for five years, before repeated skin punch biopsies with staining with both hematoxylin and eosin stain and elastic van-Gieson stain revealed EPS. During this time the patient was treated with antifungal and steroids and skin lesions progressed to large plaques.

Our case report is in accordance with the prior observations,<sup>8</sup> that the immune-stimulating treatment with imiquimod 5% cream is well tolerated and can be used for treatment of EPS. The mechanism is unknown, but imiquimod might accelerate the immune response to the abnormal elastic fibers resulting in a quicker resolution. Treatment with imiquimod 5% cream offers a well tolerated and painless alternative to destructive therapies like cryotherapy, laser therapy, and curettage. However the long term effect of imiquimod cream in treatment of EPS needs to be established.

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