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

Pernille Axél Gregersen, Birgitte Stausbøl-Gron, Mette Ramsing, Mette Sommerlund

Department of Dermato-Venereology and Department of Pathology, Aarhus University Hospital, Denmark

Abstract

Elastosis Perforans Serpiginosa (EPS) is a rare skin disease characterized 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.

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

**References**

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