

CASE REPORT

Elastosis perforans serpiginosa in a patient with Down's syndrome *

Elastose perfurante serpiginosa em portadora da síndrome de Down

Ana Carolina Figueiredo Pereira ¹
Sérgio Rodrigues da Costa Júnior ³
Everton Carlos Siviero do Vale ⁵

Isabela Guimarães Ribeiro Baeta ²
Oswaldo Macedo Gontijo Júnior ⁴

Abstract: Elastosis perforans serpiginosa is a rare, primary perforating dermatosis, frequently associated with certain genetic diseases and characterized by the transepidermal extrusion of elastic fibers. The present case report describes this dermatosis in a 19-year old female patient with Down's syndrome, who presented with asymptomatic erythematous, keratotic papules in an arciform pattern, located on her right forearm and knee, which had been present for five years. Following histopathological confirmation, treatment with cryotherapy was initiated, resulting in partial remission of the lesions.

Keywords: Conjunctive tissue; Down's syndrome; Elastic tissue; Elastic tissue abnormalities

Resumo: A elastose perfurante serpiginosa é dermatose perfurante primária incomum, frequentemente associada a determinadas doenças genéticas e caracterizada por eliminação transepidérmica de fibras elásticas. Relata-se um caso raro dessa dermatose em paciente feminina de 19 anos, portadora da síndrome de Down, que apresentava pápulas eritematocerotóticas em arranjo arciforme, localizadas no antebraço e joelho direitos, assintomáticas, com cinco anos de evolução. Após confirmação histopatológica, foi iniciado tratamento com crioterapia, ocorrendo remissão parcial das lesões.

Palavras-chave: Síndrome de Down; Tecido conjuntivo; Tecido elástico; Tecido elástico/anormalidades

INTRODUCTION

Elastosis perforans serpiginosa (EPS) is a rare skin condition included among the primary perforating dermatoses of dermal origin. It is considered a benign condition, since there are no extracutaneous manifestations. Its etiopathogenesis remains to be clarified. Focal dermal irritation is believed to induce the formation of a transepidermal channel to eliminate the irritant agent. It is normally associated with hereditary diseases of the connective tissue and with the use of D-penicillamine for Wilson disease, cystinuria and rheumatoid arthritis. ^{1,2}

It is characterized clinically by papules and ker-

atotic, desquamative, normochromic or erythematous plaques, usually affecting the face, neck, trunk and upper limbs. ^{1,2} Histopathology is the gold-standard test for diagnosis, typical findings consisting of qualitative and quantitative changes in dermal elastic fibers and their transepidermal elimination. ³

There is no standard therapy; however, successful treatment has been reported with the use of cryotherapy with liquid nitrogen, isotretinoin, tazarotene, imiquimod and CO₂, Er:YAG and pulsed dye lasers. ⁴⁻⁶ In a minority of cases, spontaneous resolution of lesions has been reported. ^{1,2} The present

Received on 10.08.2009.

Approved by the Advisory Board and accepted for publication on 19.03.2010.

* Study conducted at the Department of Dermatology, Teaching Hospital of the Federal University of Minas Gerais, Minas Gerais, Brazil.

Conflict of interest: None / *Conflito de interesse: Nenhum*

Financial funding: None / *Suporte financeiro: Nenhum*

¹ Medical Resident, Dermatology Department, Teaching Hospital of the Federal University of Minas Gerais, Minas Gerais, Brazil.

² Medical Resident, Dermatology Department, Teaching Hospital of the Federal University of Minas Gerais, Minas Gerais, Brazil.

³ Medical Resident, Dermatology Department, Teaching Hospital of the Federal University of Minas Gerais, Minas Gerais, Brazil.

⁴ Adjunct Professor, Department of Internal Medicine, Federal University of Minas Gerais. Preceptor of the Medical Residency Program, Department of Dermatology, Teaching Hospital of the Federal University of Minas Gerais, Minas Gerais, Brazil.

⁵ Assistant Professor, Department of Internal Medicine, Federal University of Minas Gerais. Preceptor of the Medical Residency Program, Department of Dermatology, Teaching Hospital of the Federal University of Minas Gerais, Minas Gerais, Brazil.

report describes the case of a patient with Down's syndrome who has had EPS for the past five years.

CASE REPORT

A young, 19-year old, white female patient with Down's syndrome complained of asymptomatic skin lesions on her right arm and knee that had appeared three years previously. She reported having had a similar condition on the medial surface of her left arm five years previously when she received a diagnosis of granuloma annulare, which was treated unsuccessfully with clobetasol propionate cream 0.05%; however, the lesions disappeared spontaneously. There were no other signs or symptoms. The patient also had hypercholesterolemia, which was being treated with simvastatin 20 mg/day.

Dermatological examination revealed erythematous papules with a keratotic surface, arranged in an arciform or annular pattern, with atrophic, scarred centers. The lesions were located on the medial surface of the patient's right forearm (Figure 1) and on the extensor surface of her right knee (Figure 2).

Histopathology revealed a transepidermal channel, eliminating amorphous material composed of eosinophilic elastic fibers, basophilic mononuclear cells and cell debris (Figure 3). Orcein staining showed a large quantity of thick, irregular and elongated elastic fibers in the upper dermis (Figure 4).

A diagnosis of EPS was confirmed and cryotherapy with liquid nitrogen was performed over multiple sessions. Up to the present time, partial remission has been achieved.

DISCUSSION

EPS is characterized clinically by papules and



FIGURE 1: Erythematous papules with keratotic surface arranged in an arciform or annular pattern with an atrophic center, located on the patient's right forearm



FIGURE 2: Erythematous, keratotic, crusted papules, located on the front of the right knee

keratotic plaques and histologically by focal elastosis of the dermis and transepidermal elimination of abnormal elastic fibers. It is a rare skin disease classified as a primary perforating dermatosis similar to reactive perforating collagenosis, perforating folliculitis and Kyrle disease. It was initially described by Lutz in 1953 and denominated *keratosis follicularis serpiginosa*.³ In 1955, Miescher found elastin in the material eliminated transepidermally and suggested the name of *elastoma intrapapillare perforans verruciforme*.¹ It was given the current denomination of *elastosis perforans serpiginosa* in 1958 by Dammert and Putkonen.³

The incidence of EPS has yet to be established. 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 at onset may range

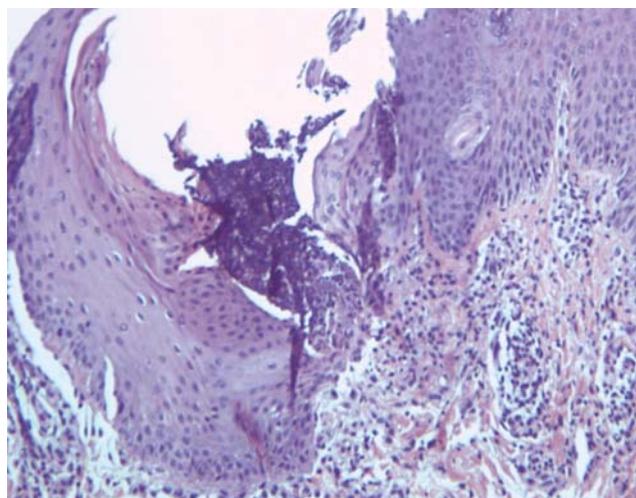


FIGURE 3: Acanthosis and transepidermal elimination channel containing amorphous material and nuclear fragments. Upper dermis with perivascular lymphohistiocytic inflammatory infiltrate. (Hematoxylin and eosin staining; magnification 200x)

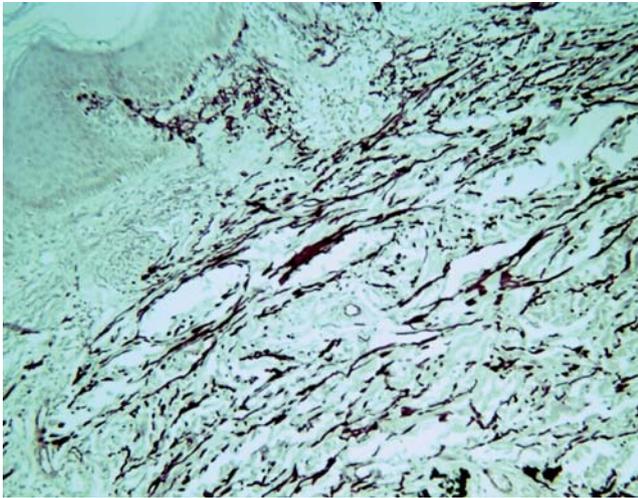


FIGURE 4: Upper dermis with a large quantity of thick, irregular elastic fibers (Orcein staining; magnification 100x)

from 5 to 89 years, as reported in the literature. Approximately 75% of those affected are male and there appears to be no preference with respect to race.² Familial groupings have been reported with varying hereditary patterns.⁷

Although the etiopathogenesis is as yet unclear, it is believed that the focal inflammation in the dermis, which has a biochemical or mechanical origin, may induce the formation of epidermal and follicular channels to expulse abnormal elastic fibers considered irritants. Fujimoto et al. reported interactions between elastin peptides and their 67kDa receptors expressed in the epidermis surrounding the transepidermal

elimination channel, which may participate in the pathogenesis of the disease.⁸

EPS is classified into three 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; and 3) induced by D-penicillamine.¹ The association between EPS and Down's syndrome is unknown; however, factors present in this syndrome such as premature skin aging, joint hyperelasticity and acrocyanosis may suggest an underlying connective tissue disorder.^{1,2} table 1 illustrates some clinical and epidemiological features of the association between EPS and Down's syndrome.^{2,3,9-12}

The disorder may present with papules and erythematous or normochromic, keratotic, asymptomatic or pruriginous plaques grouped in an annular, arciform or serpiginous pattern, surrounded by satellite lesions. They have umbilicated centers from which dermal material is eliminated. The lesions are characteristically symmetrical except in cases associated with Down's syndrome or D-penicillamine treatment.⁷ Disseminated lesions, albeit rare, have been described.¹³⁻¹⁵ The condition most commonly affects the face, the neck and the upper limbs. Lesions develop slowly, disappearing spontaneously in some cases after six months to five years, leaving superficial scarring.^{1,2,4}

The gold-standard for diagnosis is histopathological examination of the skin lesions.

TABLE 1: Clinical and epidemiological features of the association between elastosis perforans serpiginosa and Down's syndrome

Authors	Number of cases	Sex	Age (years)	Time since onset	Site of the lesions	Progression
Guimarães et al. (1981) ³	1	Male	12	3 years	Upper limbs, thighs and knees	No information
Schepis et al. (1997) ⁹	1	Male	16	2 months	Jaw line	Spontaneous remission in 12 months
Andrade et al. (1999) ¹⁰	1	Male	10	1 year	Upper and lower limbs	Not submitted to treatment. Lesions stable.
Mehta et al. (2001) ²	1	Female	21	20 years	Trunk and limbs	Partial remission with curettage. No response to topical corticoids.
De Pasquale et al. (2002) ¹¹	1	Female	20	2 years	Thighs	No information
Espinosa et al. (2008) ¹²	1	Female	16	NI	Forearms and legs	No information

Histopathology is characterized by 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 epidermis may be acantholytic and hyperkeratotic.^{1,2}

Clinically, elastosis perforans serpiginosa must be differentiated from granuloma annulare, tinea corporis, annular sarcoidosis, cutaneous calcinosis and porokeratosis of Mibelli.

Various forms of treatment have been indicated; however, management of the disease is difficult and there is no standard treatment. There have been reports of therapeutic success using cryotherapy with liquid nitrogen and oral isotretinoin.¹ Outland et al. described remission of the condition following treat-

ment with 0.1% tazarotene gel.⁶ Kelly et al reported success after 10 weeks of therapy with imiquimod.⁴ Treatment using CO₂, Er:YAG and pulsed dye lasers has also been reported in the literature, although results are controversial. Kaufman reported benefits with one or two sessions of pulsed dye laser, while Abdullah et al. reported resolution of the lesions following a single session of CO₂ laser. Saxena et al., on the other hand, failed to obtain a good response with this form of treatment.⁵ □

ACKNOWLEDGEMENTS

The authors gratefully acknowledge the important contribution made by Professor Antônio Carlos Martins Guedes in the histopathological analysis of this case, enabling diagnosis to be reached.

REFERENCES

- Lewis KG, Bercovitch L, Dill SW, Robinson-Bostom L. Acquired disorders of elastic tissue: Part I. Increased elastic tissue and solar elastotic syndromes. *J Am Acad Dermatol.* 2004;51:1-21.
- Mehta RK, Burrows NP, Payne CM, Mendelsohn SS, Pope FM, Rytina E. Elastosis perforans serpiginosa and associated disorders. *Clin Exp Dermatol.* 2001;26:521-4.
- Guimarães NS, Pinto JM, Guedes ACM, Armond S, Corgozinho Filho AA, Furtado T. Elastose perfurante serpiginosa - Relato de quatro casos. *An Bras Dermatol.* 1981;56:189-94.
- Kelly SC, Purcell SM. Imiquimod therapy for elastosis perforans serpiginosa. *Arch Dermatol.* 2006;142:829-30.
- Saxena M, Tope WD. Response of elastosis perforans serpiginosa to pulsed CO₂, Er:YAG and dye lasers. *Dermatol Surg.* 2003;29:677-9.
- Outland JD, Brown TS, Callen JP. Tazarotene is an effective therapy for elastosis perforans serpiginosa. *Arch Dermatol.* 2002;138:169-71.
- Vearrier D, Buka RL, Roberts B, Cunningham BB, Eichenfield LF, Friedlander SF. What is standard of care in the evaluation of elastosis perforans serpiginosa? A survey of pediatric dermatologists. *Pediatr Dermatol.* 2006;23:219-24.
- Fujimoto N, Tajima S, Ishibashi A. Elastin peptides induce migration and terminal differentiation of cultured keratinocytes via 67 kDa elastin receptor in vitro: 67 kDa elastin receptor is expressed in the keratinocytes eliminating elastic materials in elastosis perforans serpiginosa. *J Invest Dermatol.* 2000;115:633-9.
- Schepis C, Barone C, Siragusa M, Pettinato R, Romano C. An Updated Survey on Skin Conditions in Down Syndrome. *Dermatology.* 2002;205:234-8.
- Andrade ZMV, Pacheco THD, França ER, Alencar E. Elastosis perforans serpiginosa associada à Síndrome de Down: relato de caso. *An Bras Dermatol.* 1999;74:585-87.
- De Pasquale R, Nasca MR, Musumeci ML, Micali G. Elastosis perforans serpiginosa in an adult with Down's syndrome: report of a case with symmetrical localized involvement. *J Eur Acad Dermatol Venereol.* 2002;16:387-89.
- Espinosa PS, Baumann RJ, Vaishnav AG. Elastosis Perforans Serpiginosa, Down Syndrome, and Moyamoya Disease: a case report. *Pediatr Neurol.* 2008;38:287-8.
- Pedro SD, Garcia RL. Disseminated elastosis perforans serpiginosa. *Arch Dermatol.* 1974;109:84-5.
- Macaulay WL. Symmetry in elastosis perforans serpiginosa: its significance. *Arch Dermatol.* 1963;88:215-7.
- Holbrook KA, Byers PH. Structural abnormalities in the dermal collagen and elastic matrix of patients with inherited connective tissue disorders. *J Invest Dermatol.* 1982;79 (Suppl 1):7s-16s.

MAILING ADDRESS / ENDEREÇO PARA CORRESPONDÊNCIA:
 Ana Carolina Figueiredo Pereira
 Alameda Álvaro Celso, 55, Santa Efigênia
 30150-260 Belo Horizonte - MG, Brazil
 Telephone: +55 31 3409 9560
 E-mail: anacarol@bc.ufmg.br

How to cite this article/Como citar este artigo: Pereira ACF, Baeta IGR, Costa SR Jr, Gontijo OM Jr, Vale ECS. Elastosis perforans serpiginosa in a patient with Down's syndrome. *An Bras Dermatol.* 2010;85(5):691-4.