Localised Scleroderma: An Atypical Case

Ana Ventura[1], Paula Maio[3], Mafalda Casinhas Santos[2], Helena Sousa[2]

Port J Pediatr2018;49:373-4
DOI: https://doi.org/10.25754/pjp.2018.12235

A 16 year-old adolescent, female, with multidisciplinary follow-up for intellectual developmental disorder and minor dysmorphias (microcephaly, ogival palate, hypertelorism and pseudo-strabismus), with normal previous genetic investigation (karyotype, array comparative genomic hybridization and molecular analysis of X-fragile). She came to the consultation with oval cutaneous plaques, which had evolved over seven months (Fig. 1). She did not have systemic repercussion, dysphagia, dyspnoea or Raynaud's phenomenon. Physical examination revealed cutaneous thickening of anterior trunk and proximal region of the lower limbs, limited range of motion of the elbows, hips and lumbar spine, with no other inflammatory signs, and thoracic kyphosis; without uveitis. Generalised morphoea was diagnosed and medicated with topical corticosteroids by dermatology. Analytically she had slight iron-deficiency anaemia, normal leukocyte formula and serum proteins electrophoresis, inflammatory parameters, autoantibodies (antinuclear, anti-Scl-70, anticentromere, anti-SSA, anti-RNP and anti-histones), rheumatoid factor and serology for *Borrelia burgdorferi* negative. She had thickening of the dermis and subdermis, with involvement of the superficial adipose tissue in the dermal ultrasound scan. A dorsal lumbar and pelvic magnetic resonance showed no inflammatory signs or degenerative alterations. Echocardiogram, pulmonary function tests with DLCO, capillaroscopy and limbs x-rays were normal. She started methotrexate (subcutaneous 15 mg/m^2/week) and oral prednisolone 0.5 mg/kg/day, besides phototherapy (UV-B ultraviolet radiation) and physiotherapy. Three months later, due to progression of the lesions, she suspended oral corticotherapy and started monthly intravenous methylprednisolone pulses (1 g/dose, three days). She completed six months of pulse therapy with stabilization and partial regression of the cutaneous

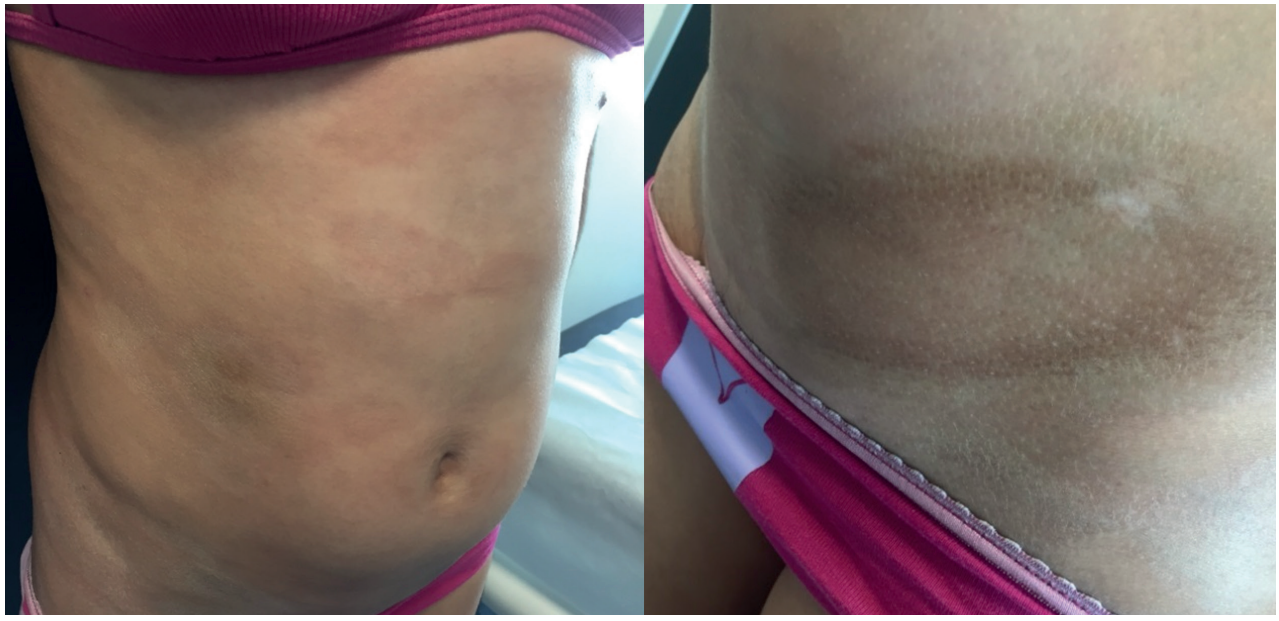

Figure 1. View of the lesions in the diagnosis. We observe multiple infiltrated oval plaques, hypo and hyperpigmented, with ill-defined margins, surrounded by violet erythema, with dimensions between 4 to 15 cm on the longer axis, bilateral, localised at the anterior and posterior region of the trunk and proximal region of the lower limbs.

1. Paediatrics Department, Hospital de Santa Maria, Centro Hospitalar Lisboa Norte, Lisbon, Portugal
2. Paediatric Department, Hospital Vila Franca de Xira, Vila Franca de Xira, Portugal
3. Dermato-Venereology Service, Hospital Vila Franca de Xira, Vila Franca de Xira, Portugal
Corresponding Author
Ana Ventura
patriciaventura26@gmail.com
Paediatrics Department, Hospital de Santa Maria,
Avenida Professor Egas Moniz, 1649-035 Lisbon, Portugal
Received: 03/01/2018 |Accepted: 28/04/2018

plaques (Fig. 2), although without improvement of joint amplitudes. In this particular case, contractures may not be exclusively related with the morphoea, given that a limited range of motion is also present in joints without underlying cutaneous involvement, admitting that this can be a pre-existing alteration, related to an underlying disorder. Diagnostic investigation continues with more specific genetic study, in order to relate all the phenotypic changes found.

The authors aim to demonstrate the characteristic lesions of juvenile localised scleroderma, generalised subtype or generalised morphoea, a rare disease with incidence that varies between 0.34-2.7 cases per 100 000 individuals / year, warning about the importance of excluding systemic involvement. Although the diagnosis is frequently established based on the characteristic cutaneous commitment, some cases are doubtful and a biopsy of the lesions can be indicated.

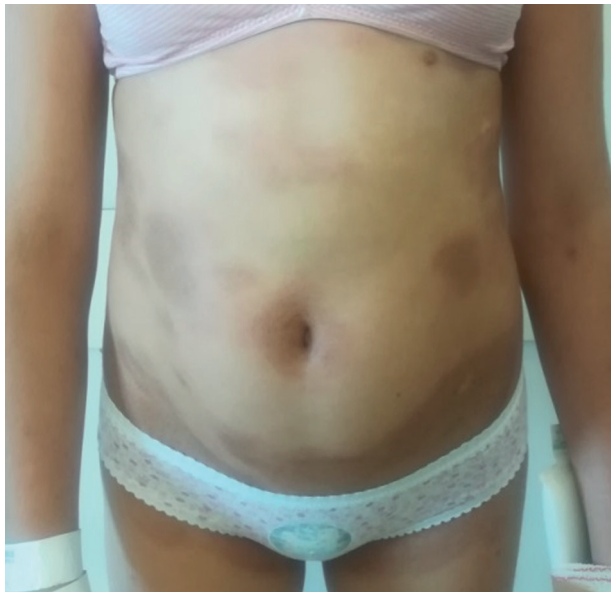

Figure 2. View of the lesions after six months of methylprednisolone. We observe the anterior trunk with multiple hyperpigmented infiltrated plaques, fewer in number and of smaller dimensions in relation to their initial situation. Bright and infiltrated skin, with absence of hair distribution.

Keywords: Adolescent; Scleroderma, Localised/diagnosis

WHAT THIS REPORT ADDS

- Juvenile localised scleroderma is a rare entity, and diagnosis is mainly clinical.
- It can be an important cause of morbidity and up to a quarter of cases have extra-cutaneous manifestations, namely articular.
- Although its treatment is not consensual, the immunosuppressive therapy seems to be effective in delaying the progression of the disease.
- Early diagnosis and orientation are fundamental for the prognosis.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

Funding Sources

There were no external funding sources for the realization of this paper.

Protection of human and animal subjects

The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).

Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

References

1. Torok K. Pediatric scleroderma: Systemic or localized forms. Pediatr Clin North Am 2012;59:381-405. doi: 10.1016/j.pcl.2012.03.011.
2. Sawhney S, Aggarwal A. Pediatric rheumatology. A clinical viewpoint. Singapore: Springer Singapore; 2017.
3. Bielsa Marsol I. Update on the classification and treatment of localized scleroderma. Actas Dermosifiliogr 2013;104:654-66. doi: 10.1016/j.adengl.2012.10.012.
4. Zulian F. Localized scleroderma in childhood [Accessed 29 June 2017]. Available at: http://www.uptodate.com
5. Denton CP. Overview and classification of scleroderma disorders [Accessed 29 June 2017]. Available at: http://www.uptodate.com