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## Case report

# Generalized morphea in a child with harlequin ichthyosis: a rare association



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## ARTICLE INFO

### Article history:

Received 24 October 2013

Accepted 21 May 2014

Available online 26 November 2014

### Keywords:

Harlequin ichthyosis

Children

Morphea

Scleroderma

## ABSTRACT

**Introduction:** Harlequin ichthyosis (HI) is a severe and rare hereditary congenital skin disorder characterized by excessive dryness, ectropion and eclabion. The association of ichthyosis with systemic sclerosis has been described in only three children. No patient with generalized morphea (GM) associated with harlequin ichthyosis was described.

**Case report:** A 4-years and 6-months girl, diagnosed with harlequin ichthyosis based on diffuse cutaneous thickening, scaling, erythema, ectropion and eclabium since the first hours of birth was described. She was treated with acitretin (1.0 mg/kg/day) and emollient cream. At 3 years and 9 months, she developed muscle contractures with pain on motion and limitation in elbows and knees, and diffuse sclerodermic plaques on the abdomen, back, suprapubic area and lower limbs. Skin biopsy showed rectified epidermis and mild hyperorthokeratosis, reticular dermis with perivascular and periadnexal infiltrates of lymphocytes and mononuclear cells, and reticular dermis and sweat gland sclerosis surrounded by a dense collagen tissue, compatible with scleroderma. The patient fulfilled the GM subtype criteria. Methotrexate and prednisone were introduced. At 4 years and 3 months, new scleroderma lesions occurred and azathioprine was associated with previous therapy, with no apparent changes after two months.

**Discussion:** A case of harlequin ichthyosis associated with a GM was reported. The treatment of these two conditions is a challenge and requires a multidisciplinary team.

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## Morfeia generalizada em uma criança com ictiose arlequim, uma associação rara

### R E S U M O

**Introdução:** Ictiose arlequim é uma doença cutânea congênita grave, autossômica e rara, caracterizada por ressecamento excessivo da pele e hiperqueratose. A associação de ictiose

### Palavras-chave:

Ictiose arlequim

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<http://dx.doi.org/10.1016/j.rbre.2014.05.004>

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Crianças  
Morfeia  
Esclerodermia

com esclerose sistêmica foi descrita em apenas três crianças. Ainda não foi descrito nenhum paciente com morfeia generalizada (MG) associada à ictiose arlequim.

*Relato de caso:* Menina de quatro anos e seis meses de idade com diagnóstico de ictiose arlequim baseado em espessamento cutâneo difuso, com fissuras, descamação, eritema e sangramento da lesão desde as primeiras horas de vida. A paciente foi tratada com acitretina (1,0 mg/kg/dia) e creme emoliente. Aos três anos e nove meses, desenvolveu contraturas musculares com dor à movimentação e limitação nos cotovelos e joelhos e placas esclerodérmicas difusas no abdômen, nas costas, na região suprapúbica e nas extremidades inferiores. A biópsia de pele mostrou epiderme retificada e hiperqueratose leve, derme reticular com linfócitos, infiltrado mononuclear perivascular e perianaxial e esclerose da derme reticular e glândula sudorípara rodeada por um tecido colágeno denso, compatível com esclerodermia. A paciente preencheu os critérios para o subtipo MG. Metotrexato e prednisona foram introduzidos. Aos quatro anos e três meses, apresentou novas lesões esclerodérmicas, associando-se azatioprina à terapêutica anterior, sem resposta após dois meses.

*Discussão:* Um caso de ictiose arlequim associada à MG foi descrito. O tratamento dessas duas condições é um desafio e requer uma equipe multidisciplinar.

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## Introduction

Juvenile cutaneous scleroderma is a rare condition in pediatric patients characterized by the involvement of the skin and/or subcutaneous tissue.<sup>1,2</sup> Generalized morphea (GM) is an uncommon subtype of localized scleroderma and occurs in 7% of the patients.<sup>2</sup> The association of GM with other cutaneous diseases was rarely reported in the literature, especially with ichthyosiform diseases.

Of note, ichthyosis comprehends a heterogeneous group of skin diseases characterized by cutaneous hyperkeratinization and may be congenital or acquired.<sup>3</sup> Harlequin ichthyosis is a congenital form, transmitted in an autosomal recessive mode. It is the most severe and often fatal form of ichthyosis.<sup>4</sup>

The association of ichthyosis with scleroderma has been described in only four patients. Three of them had juvenile systemic sclerosis, and one of them had localized scleroderma. Two of the juvenile systemic sclerosis patients were associated with ichthyotic-appearing lesions,<sup>5</sup> and one of them with acquired ichthyosis.<sup>6</sup> The localized scleroderma patient was associated with ichthyotic-appearing lesions.<sup>5</sup> However, to our knowledge, no patient with GM scleroderma associated with the harlequin subtype of ichthyosis was reported. This case is described herein.

## Case report

A 4-years and 6-months girl was diagnosed with harlequin ichthyosis at neonatal period, based on diffuse cutaneous thickening, diffuse plate-like scales, erythema, fissures, bleeding lesions, bilateral ectropion and eclabium since the first hours of life. No history of parental consanguinity and harlequin ichthyosis were observed. She was a term newborn after cesarean delivery. Her birth weight and height were 2.650 gm and 43 cm, respectively. Microcephaly, auricular pinna and nasal abnormalities, alopecia and flexion contracture were not evidenced. She was treated with acitretin

(1.0 mg/kg/day) and emollient cream. During the first 3 months of life, she had been hospitalized three times due to skin infections, with improvement after antibiotic treatment. At 3 years, she presented growth retardation with height of 84 cm, with delayed motor development. Skin biopsy of abdominal epidermis showing hyperkeratosis parakeratotic, agranulose, regular epidermal acanthosis and thickening of the dermis with proliferation of vessels with mild lymphocytic inflammatory infiltrate and psoriasiform dermatitis (Fig. 1) confirmed diagnosis of ichthyosis.<sup>7</sup> After 9 months, she developed painful flexion muscle contractures, with pain on motion and limitation affecting elbows and knees, associated with generalized ichthyosis lesions, with diffuse pain upon palpation, bilateral ectropion and diffuse scleroderm plaques without distribution pattern on: abdomen (11 lesions ranged from 0.5 × 0.5 cm to 3.3 × 3.0 cm), back (13 lesions ranged from 1.0 × 1.0 cm to 2.8 × 3.0 cm), supra-pubic area (3 lesions varied from 0.8 × 1.4 cm to 3.5 × 2.0 cm) and proximal aspects of thighs (2 lesions ranged from 1.0 × 1.0 cm to 3.0 × 2.5 cm). Skin biopsy showed rectified epidermis and mild hyperorthokeratosis, reticular dermis with lymphocyte and mononuclear cells that infiltrate perivascular and periadnexal, and sclerosis of reticular dermis and sweat gland surrounded by a dense collagen tissue, compatible with scleroderma. Hemoglobin, leukocyte and platelet counts, renal function and urinalysis were normal. The C-reactive protein was 14.4 mg/dL (reference value <5). The rheumatoid factor, antinuclear antibody (ANA), anti-double-stranded DNA (anti-dsDNA), anti-Sm, anti-RNP, anti-Ro and anti-La antibodies were negative. Thoracic computer tomography, echocardiography, barium meal examination and slit-lamp examination were normal. The nailfold capillaroscopy was performed without being able to visualize capillaries due to crusts periungual. Therefore, the patient fulfilled the juvenile localized scleroderma criteria (GM subtype)<sup>8</sup> and methotrexate (0.5 mg/kg/week) and prednisone (1.0 mg/kg/day) were introduced at the age of 3 years 11 months. At 4 years and 1 month, methotrexate dosage was increased to 1.0 mg/kg/week. At 4 years and 3 months, she presented generalized ichthyosis lesions with new scleroderma

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