

Cutaneous neonatal lupus with cutis marmorata telangiectatica congenita-like lesions^{*}

Lúpus cutâneo neonatal com lesões semelhantes à cutis marmorata telangiectatica congênita

Flávia Trevisan¹
Clóvis Antonio Lopes Pinto³

Paulo Rowilson Cunha²
Fernanda Gomes Cattete⁴

DOI: <http://dx.doi.org/10.1590/abd1806-4841.20131832>

Abstract: Neonatal lupus is a rare disease caused by the transplacental transfer of maternal autoantibodies to the foetus, characterized by transient clinical manifestations such as cutaneous, haematological, and hepatobiliary events or permanent such as congenital heart block. The typical cutaneous manifestations include erythematous, scaly, annular or arched lesions on the face, with slight central atrophy and photosensitivity, clinically and histologically similar to subacute cutaneous lupus. However, in some cases, the lesions may resemble those in cutis marmorata telangiectatica congenita, although this phenomenon is rare and only eight such cases have been reported to date. We report a case of cutaneous neonatal lupus with atypical lesions on the limbs, which had a reddish-purple marbled appearance, resembling the lesions in cutis marmorata telangiectatica congenita.

Keywords: Antibodies; Autoantibodies; Clinical diagnosis; Congenital, hereditary, and neonatal diseases and abnormalities; Diagnosis, differential; Lupus erythematosus, systemic

Resumo: Lúpus neonatal é uma doença rara causada pela transferência de auto-anticorpos maternos para o feto, caracterizada por manifestações clínicas transitórias como cutâneas, hematológicas e hepatobiliares, ou permanentes como o bloqueio cardíaco congênito. As manifestações cutâneas típicas incluem lesões fotossensíveis descamativas, anulares ou arqueadas na face, com discreta atrofia central, clínica e histopatologicamente similares ao lúpus cutâneo subagudo. Entretanto, em alguns casos, as lesões podem lembrar aquelas da cutis marmorata telangiectatica congênita, mas esse fenômeno é raro e somente oito casos foram reportados até hoje. Nós relatamos aqui um caso de lúpus cutâneo neonatal com lesões atípicas nos membros de cor vermelho-purpúricas e aparência marmórea, semelhantes às lesões da cutis marmorata telangiectatica congênita.

Palavras-chave: Anticorpos; Auto-anticorpos; Diagnóstico clínico; Diagnóstico diferencial; Doenças congênitas, hereditárias e neonatais e anormalidades; Lúpus eritematoso sistêmico

INTRODUCTION

Neonatal lupus, first described in 1954, is a rare disease characterized by the transplacental transfer of the maternal anti-Ro, anti-La, and, less frequently, the anti-U1RNP autoantibodies to the foetus. It has been so named because it was initially described in cases similar to cutaneous subacute lupus in the children of mothers positive for autoantibodies.^{1,2,3} Its clinical presentation includes transient events such as cuta-

neous, haematological, and hepatobiliary manifestations or permanent events such as congenital heart block. Occurs in 1–2% of children whose mothers are positive for anti-Ro, anti-La, or anti-U1RNP autoantibodies, and of these children, heart block occurs in approximately 50%, cutaneous manifestations in 34%, and both in 10% of the cases.¹

Received on 24.04.2012.

Approved by the Advisory Board and accepted for publication on 04.06.2012.

* Study carried out at the Medical School of Jundiaí (Faculdade de Medicina de Jundiaí - FMJ) - Jundiaí (SP), Brazil.

Financial Support: none

Conflict of Interests: none

¹ MD - Resident of Dermatology, Medical School of Jundiaí (FMJ) - Jundiaí (SP), Brazil.

² Full Professor - Head of the Department of Dermatology and Professor of Dermatology, Medical School of Jundiaí (FMJ) - Jundiaí (SP), Brazil.

³ PhD - Associate Professor of the Discipline of Pathology, Medical School of Jundiaí (FMJ) and Attending Physician, Department of Pathology, Cancer Hospital Antônio Cândido de Camargo (Hospital AC Camargo) - Santo André (SP), Brazil.

⁴ MD - Resident of Dermatology, Medical School of Jundiaí (FMJ) - Jundiaí (SP), Brazil.

Cardiac manifestations usually occur in the foetus in the second trimester of pregnancy. Third degree blockage, the most severe development, once established is irreversible with mortality rates of up to 20%.² Hepatic manifestations are seen in 10% of all such cases and are commonly associated with cardiac or cutaneous manifestations and the prognosis for patients is generally good, with spontaneous resolution.^{1,2} The haematological manifestations, which occur in approximately 10% of cases, include thrombocytopenia, neutropenia, anaemia, and pancytopenia, which are usually transient manifestations.²

The cutaneous manifestations include macules and scaly, erythematous plaques, mainly in the peri-orbital region. The lesions may be present at birth but occur more commonly after sun exposure until the infant is three months old, remaining until the age of seven months, rarely leaving residual lesions or atrophic telangiectasias.¹⁴ The polycyclic lesions must be distinguished from the urticaria, erythema marginatum, tinea corporis, seborrheic dermatitis, and ichthyosis lesions. The annular lesions can be confused with those of erythema annulare centrifugum, familial annular erythema, erythema multiforme, *Pityrosporum* infections, annular erythema of infancy, and erythema gyratum atrophicans transiens.

In addition, eight cases of cutaneous neonatal lupus (CNL) have been previously described in patients with lesions similar to those in *cutis marmorata telangiectatica congenita* (CMTC), a sporadic and unusual vascular anomaly.^{5,6} CMTC most usually presents as marbled-like, reticular, and violaceous segmental lesions with onset at birth or soon after birth and gradually regress during childhood or remain until adulthood.^{5,6,7} The diagnosis is mainly based on clinical findings and involves criteria that have not yet been completely established.⁸ The differential diagnosis includes physiological persistence of *cutis marmorata*, reticulated capillary malformation, Bockenheimer syndrome, Adams-Oliver syndrome, phakomatosis pigmentovascularis, and Klippel-Trenaunay syndrome.⁸

CNL is diagnosed by examining the characteristic skin lesions and detecting the presence of autoantibodies in the mother or in the patient. Diagnostic doubts may be dispelled if vacuolar alteration of the epidermis and adnexal structures, hyperkeratosis, acanthosis, and superficial or deep perivascular lymphocytic infiltrations, which are characteristic of sub-acute cutaneous lupus lesions, are observed during histopathological examination.⁹ Direct immunofluorescence analysis for the presence of IgG may aid in diagnosis; however, the results are negative in more than 50% of cases.⁶

CASE REPORT

A 3-month-old female infant born at term showed erythematous and scaly lesions on the face after photoexposure at 4 weeks of life. At 2 months, she showed marbled-like, slightly infiltrative erythematous, purpuric lesions, which resembled CMTC lesions, on the limbs (Figures 1 and 2). Histopathological analysis of the lesions on the limbs showed chronic lichenoid interface dermatitis, acantholysis, spongiosis, and vacuolization of the basal layer, which was highly suggestive of cutaneous lupus (Figure 3). The results of anti-Ro autoantibody analysis were positive for both the patient and her mother, confirming the diagnosis of neonatal lupus. The results of haematologic screening involving blood cell count and erythrocyte sedimentation rate (ESR), hepatic screening involving liver function and liver enzyme analysis, and cardiological testing involving electrocardiography and echocardiography were normal.



FIGURE 1: (*Cutis marmorata telangiectatica congenita*-like lesions: erythematous-purpuric marbled macules on the upper limbs.)



FIGURE 2: *Cutis marmorata telangiectatica congenita*-like lesions: erythematous-purpuric marbled macules on the limbs

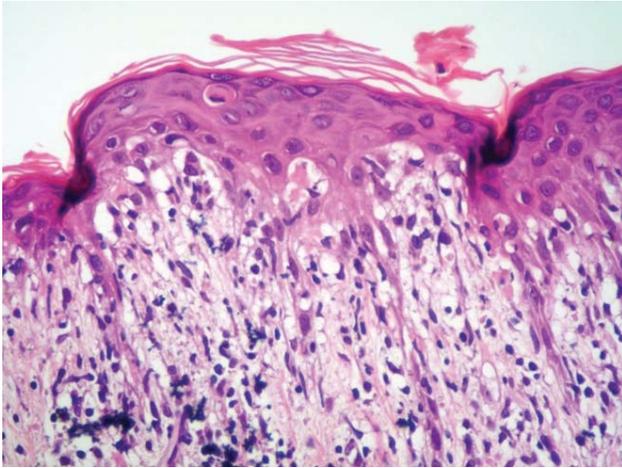


FIGURE 3: Epidermis with hyperkeratosis, spongiosis, irregular acanthosis and basal layer vacuolization. In the dermis, moderate lymphohistiocytic infiltrate with perivascular distribution. (Hematoxylin-eosin, 200X)

The treatment of lesions was only preventive, avoiding sun exposure, which caused regression of lesions at 7 months without scarring. The patient's mother was asymptomatic at the time of pregnancy, with an ESR of 116 mm/h, a fine speckled anti-nuclear antibody pattern, and an ANA titre of 1:640. Therefore, she was instructed to ensure outpatient follow-up because of the possibility of developing connective tissue diseases.

DISCUSSION

Since 1980, eight cases of CNL patients with CMTC-like lesions have been reported.^{5,6,7,10} Of these, four cases are from a series of 18 cases in some of which CNL and CMCT were assumed to be coexisting diseases, whereas in some other cases the authors had concluded that the livedoid CMTC-like pattern was a

residual phase of CNL that occurs at the intrauterine stage.^{7,10} These hypotheses have been questioned ever since because in some cases, as the one here described, lesions are absent at birth.

We report a case of a 3-month-old infant who had CNL that was difficult to diagnose clinically because the characteristics of the lesions on the limbs were not very specific; the diagnosis was confirmed by the history of sun exposure, histopathologic examinations and by the patient and the patient's mother positive anti-Ro autoantibodies. The CNL lesions were erythematous, with scaly and patch-like appearance on the face and reddish-purple marbled-like appearance on the limbs, which resembled CMTC lesions; the lesions completely regressed by the time the patient was seven months old.

On the basis of this information and that obtained from previously published cases, we hypothesize that the CMTC-like lesions are part of the spectrum of possible clinical presentations of CNL, but not specific for it.^{5,7} In agreement with some authors, we consider that clinical and immunological examination is required only if the patient has typical CNL lesions or if the patient's mother is suspected of having connective tissue disease.⁵ Because of the rare but possible association of cardiac, haematologic, and hepatic clinical manifestations with CNL, patients should be screened for these manifestations. Due attention should be given to subsequent pregnancies because the risk of CNL occurrence accompanied with heart block among siblings of symptomatic mothers increases from 2% to 25% in subsequent pregnancies.¹ Asymptomatic mothers, who constitute almost half of all CNL cases, should be followed-up every six months or annually because they are likely to develop Sjogren syndrome (20%), systemic lupus erythematosus (18%), and other connective tissue diseases (18%).¹ □

REFERENCES

1. Carvalho JF, Viana VST, Cruz RBP, Bonfá E. Neonatal lupus syndrome. Rev Bras Reumatol. 2005;45:153-60.
2. Monari P, Gualdi G, Fantini F, Giannetti A. Cutaneous neonatal lupus erythematosus in four siblings. Br J Dermatol. 2008;158:626-8.
3. del Boz J, Serrano MM, Gómez E, Vera A. Neonatal lupus erythematosus and cutis marmorata telangiectatica congenita-like lesions. Int J Dermatol. 2009;48:1206-8.
4. Heughan CE, Kanigsberg N. Cutis marmorata telangiectatica congenita and neonatal lupus. Pediatr Dermatol. 2007;27:320-1.
5. Lee LA. The clinical spectrum of neonatal lupus. Arch Dermatol Res. 2009;301:107-110.
6. Buyon JP, Clancy RM. Neonatal Lupus: Review of Proposed Pathogenesis and Clinical Data from the US-based Research Registry for Neonatal Lupus. Autoimmunity. 2003;36:41-50.
7. Weston WL, Morelli JG, Lee LA. The clinical spectrum of anti-Ro-positive cutaneous neonatal lupus erythematosus. J Am Acad Dermatol. 1999;40:675-81.
8. Kienast AK, Hoeger PH. Cutis marmorata telangiectatica congenita: a prospective study of 27 cases and review of the literature with proposal of diagnostic criteria. Clin Exp Dermatol. 2009;34:319-33.
9. Peñate Y, Guillermo N, Rodríguez J, Hernández-Machín B, Montenegro T, Afonso JL, et al. Histopathologic characteristics of neonatal cutaneous lupus erythematosus: description of five cases and literature review. J Cutan Pathol. 2009;36:660-7.
10. Carrascosa JM, Ribera M, Bielsa I, Coroleu W, Ferrándiz C. Cutis marmorata telangiectatica congenita or neonatal lupus? Pediatr Dermatol. 1996;13:230-2.

*MAILING ADDRESS:**Flávia Trevisan**Rua Marechal Mallet, 181 - São Pedro
83005-530 - São José dos Pinhais - PR
Brazil**E-mail: flaviatrevisan1@yahoo.com.br*

How to cite this article: Trevisan F, Cunha PR, Pinto CAL, Cattete FG. Cutaneous neonatal lupus with cutis marmorata telangiectatica congenita-like lesions. An Bras Dermatol. 2013;88(3):428-31.