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CASE REPORT

Neonatal lupus: case report with exuberant findings

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Abstract

Neonatal lupus erythematosus (NLE) is an immunological disease characterized by the presence of maternal specific antibodies IgG such as anti-Ro, anti-La and anti-U1RNP with capacity of crossing the placental barrier and causing lesions in neonates. It is manifested by cardiac, cutaneous, hepatic or hematological alterations. Atrioventricular block is the most frequent, severe and permanent manifestation. Non-cardiac manifestations are transient and disappear until six or eight months of life. The diagnosis is made mainly through the clinical and presence of specific antibodies in the maternal and fetal circulation. The prognosis and treatment will depend on the manifestations presented and the degree of systemic involvement. Most manifestations require expectant behavior since they are self-resolving, and the degree of cardiac involvement is the most important factor in NLE mortality.

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INTRODUCTION

Neonatal lupus erythematosus (NLE) is a rare disease and was first described in 1954 by McCuiston. This autoimmune condition is caused by the passive transfer of autoantibodies from the mother to the fetus.^{1,2} Incidence varies from 1% to 2% in infants born to mothers with systemic lupus erythematosus (SLE), Sjögren syndrome, or who are asymptomatic but have anti-Ro/SSA, anti-La/SSB, or anti-RNP autoantibodies.³

The most common clinical manifestations are congenital atrioventricular block (AVB, 50% of cases) and cutaneous involvement (34% of cases); both are seen in 10% of cases.¹ Other less common systemic alterations, such as neonatal hepatitis, cholestasis, and cytopenias, may occur.³

The main clinical characteristics of the cutaneous form typically include multiple annular erythematous lesions or arcuate macules. The face is the most commonly affected site, but the disease can also affect the palms of hands, soles of feet, or diaper area. Periorbicular involvement is frequent and described as raccoon eyes,² which is considered a characteristic finding of this syndrome.

Cutaneous NLE is diagnosed by observation of characteristic cutaneous lesions and exclusion of systemic involvement combined with the detection of autoantibodies in the mother and/or the patient.¹ Differential diagnosis and appropriate treatment should be initiated as early as possible to avoid complications.²

Here we report the case of a neonate with NLE with cutaneous involvement followed in our hospital.

CASE REPORT

A male infant born at term by uncomplicated vaginal birth presented with crusted and exulcerated erythematous lesions at birth, mainly involving the face, trunk, and limbs, in addition to desquamation of the hands and feet. Mother denied previous comorbidities and reported adequate prenatal care. No other alterations were identified upon clinical examination. Maternal serological screening during gestation (collected in the 1st and 3rd trimesters) showed negative results for HIV, syphilis, toxoplasmosis, rubella, cytomegalovirus, and herpes simplex virus. After birth, red reflex, cardiac, fundoscopic, audiometric, and Guthrie tests were conducted, and no alterations were found. The newborn was tested for Epstein-Barr virus, cytomegalovirus, and herpes simplex, which were all negative. Laboratory examinations showed low platelet count (41,000 cells/mm³). The patient was referred from the neonatology to outpatient dermatology and hematology clinics for clinical investigation.

Clinical dermatological examination at 21 days revealed annular polycyclic erythematous macules, disseminated but sparing the extremities (Figure 1), leading to the suspicion of neonatal lupus. Tests were conducted and showed a reactive FAN of 1:320 with mixed pattern (fine nuclear speckled

and homogeneous nucleolar) and positive anti-Ro/SSA and anti-SSB/La autoantibodies of >320. These tests were also performed in the mother and showed a FAN of 1:320 with nuclear speckled pattern and positive anti-Ro/SSA and anti-SSB/La autoantibodies of >320. Electrocardiographic and echocardiographic results showed no alterations. Hemogram was repeated at 1 month of age and showed normalized platelet count (196,000 cells/mm³).

Diagnosis of cutaneous NLE was made based on the clinical presentation and positive autoantibodies in the mother and the newborn.

We decided to closely observe the infant without any other intervention. We advised the mother to avoid exposure of the newborn to the sun and to follow up through regular consultations at the pediatrics and dermatology clinics. When the patient returned after a month of outpatient consultation, the cutaneous lesions had receded and were lighter and grayed out, with evident improvement in relation to the initial consultation (Figure 2).

DISCUSSION

NLE is a rare condition characterized by transplacental transfer of maternal IgG autoantibodies (anti-Ro, anti-La, and less often anti-U1RNP) to the fetus. Its incidence is estimated at 1:12,500 live births in the population with anti-Ro/SSA/Ro4 antibodies. This incidence increases to 1 in every 86 (1.2%) live births in the population of mothers with SLE.⁴

Skin changes begin in the first weeks of life or may already be present at birth and are characterized as annular plaques that are scaly and photosensitive affecting the whole body, especially the face and scalp, as seen in our patient. The literature also describes a peculiar and characteristic change, which consists of confluent lesions in the periorbital region reminiscent of an owl or raccoon, but this was not seen in our case.³ The lesions are polymorphic and transitory, lasting for



Figure 1. Crusted and exulcerated erythematous polycyclic annular plaques on the face, trunk, and limbs. Note the desquamation on the hands.



Figure 2. Annular erythematous eruptions, more evident on the arms, sparing the extremities, and receding. Complete improvement of hand desquamation.

approximately 6 months, which coincides with the disappearance of maternal IgG autoantibodies, which were present in the infant's circulation.^{1,6} After recovery, there are generally no residual lesions.³

The most frequent and severe clinical manifestation is complete congenital AVB, which is generally permanent⁴; this was not seen in our patient after clinical screening. Damage to the cardiac conduction system originally occurs in the normal heart, usually between 18 and 24 weeks. Coincidentally, it is precisely at this time that there is an abrupt elevation in the transplacental transfer of maternal IgG autoantibodies to the fetus, which reinforces the pathophysiological mechanism of anti-Ro/SSA and anti-SSB/La antibodies in this syndrome.⁴

Other systemic manifestations, such as the presence of hemocytopenia in the newborn including hemolytic anemia, leukopenia (neutropenia), and thrombocytopenia, are observed; the latter is the most common alteration, occurring in 10%–20% of cases in a transitory manner.⁴ Our patient had a low platelet count of 41,000 cells/mm³, with transient manifestation and normalization of hemogram values after 1 month of life.

The frequency of hepatobiliary manifestations in patients with NLE is 9%, and in approximately 80% of these

cases, hepatic involvement is associated with cardiac and/or skin abnormalities.⁴ When the finding is isolated, the newborn may present with elevated hepatic enzymes as the only laboratory finding a few weeks after birth. Other organs and systems involved are the lungs (pneumonia and capillaritis), osteoarticular system (chondrodysplasia punctata), and central nervous system (convulsions, aseptic meningitis, macrocephaly, and hydrocephalus).³ Our patient did not exhibit any of these manifestations.

Diagnosis is confirmed based on the presence of characteristic cutaneous lesions and detection of autoantibodies in the mother and the patient. In case of diagnostic uncertainty, histopathological examination should be performed, which presents the same characteristics of subacute cutaneous lupus. Direct immunofluorescence for IgG testing can aid in diagnosis, but the results are negative in >50% of cases.¹ Early diagnosis is essential in NLE to avoid permanent sequelae and even death from cardiac complications.³ The diagnosis in the case presented herein was confirmed by clinical examination of the newborn, associated with the presence of anti-Ro/SSA and anti-SSB/La antibodies exceeding 320 in the patient and the mother.

Differential diagnosis is important. Annular lesions may be mistaken for those of centrifugal annular erythema, erythema multiforme, pityrosporum infection, annular erythema of childhood, and erythema gyratum atrophicans transiens.¹

Treatment requires a multidisciplinary approach for detection and early intervention in situations of risk.⁵ Watchful observation is required for the majority of NLE symptoms because they tend to resolve spontaneously, and this was the intervention chosen for our patient. In cases of heart block, corticosteroids can be considered during pregnancy, along with the use of diuretics and early pacemaker implant. Serial cardiovascular assessments during pregnancy is indispensable in future pregnancies, as the risk of NLE occurrence accompanied by heart block among siblings born to symptomatic mothers increases from 2% to 25% in subsequent pregnancies.¹

The prognosis of the disease varies according to the presence and severity of AVB because the other manifestations recede between 6 and 8 months of age. It should be noted that the majority of patients with NLE without congenital heart block have a good prognosis, similar to our patient.⁷ In cases of NLE, approximately half of the mothers are asymptomatic and do not have any diagnosis of autoimmune disease, but should be followed every 6 months or once a year because of their susceptibility to developing Sjögren syndrome (20%), SLE (18%), and other connective tissue diseases (18%).¹ The mother of our patient was asymptomatic and has been referred to rheumatology for examinations and monitoring.

Even though the prognosis of cutaneous NLE is excellent because it is transitory, coinciding with the end of the movement of maternal antibodies, appropriate measures should be taken to rule out coexisting systemic diseases or similar cutaneous manifestations that may be more severe. It is also important to guide the mother due to the risk of emerging

autoimmune disease in the future when there is no previous diagnosis and to the increased risk of manifestation of NLE with heart block in future pregnancies, with serial cardiovascular assessments being essential.

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