Case Report: A Review of Neonatal Lupus Erythematosus with a Case Illustration

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Introduction:

Neonatal Lupus Erythematosus (NLE) is a rare autoimmune disease of the newborn caused by the passive transplacental transfer of maternal autoantibodies, particularly anti-SSA/Ro and anti-SSB/La (1). The global incidence of NLE remains uncertain due to numerous undiagnosed cases, but in the USA, it affects one in every 20,000 live births (1). NLE was first reported in 1954 in a baby born to an ANA-positive mother, and it is characterized by a constellation of manifestations affecting various systems, including the cardiovascular, cutaneous, hepatobiliary, and hematological systems (6).

Case Illustration:

A six-month-old female presented with a five-month history of an erythematous, itchy facial rash exacerbated by sun exposure. Born full-term following an uncomplicated pregnancy and birth, she had achieved age-appropriate developmental milestones. Notably, her family history revealed a grandmother with lupus.

Clinical Examination:

Upon examination, pronounced erythematous patches were noted on her face, particularly around the periorbital region, and a more reticulated eruption on the left arm. However, the rest of the systemic examination was normal, with no evidence of hepatosplenomegaly.

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Assessment and Management:

NLE was the primary consideration given the striking periorbital distribution (so-called “raccoon eyes”), exacerbated by sun exposure. Atopic dermatitis was also considered, and the baby was managed with hydrocortisone 2.5% topical ointment since the parents reported associated pruritus. To confirm the clinical suspicion of NLE, maternal serologies were ordered. Due to the slow waning of maternal antibodies, it was deemed that serological testing on the infant, who was already five months old at the time of presentation, might yield false-negative results.

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Maternal serology confirmed positive anti-Ro/SSA at >8.0 and positive anti-La/SSB antibodies at 2.3, far exceeding the range values of <1.0 Neg AI. Following the confirmation of NLE, a re-

ferral to pediatric cardiology was made, and subsequent ECG and cardiac ultrasound showed a structurally normal heart with minor T-wave abnormalities. A follow-up EKG was planned for six months later. Notably, the mother was referred by us to rheumatology and was subsequently diagnosed with Sjögren's syndrome.

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Clinical Presentation in NLE:

Cutaneous Manifestations: The disease typically presents with cutaneous lesions, which can emerge at birth or in the first few weeks of life. These rashes are commonly found on the scalp, face, and neck, often forming a periorbital "raccoon-eye" pattern. The rash varies in appearance, showing characteristics like redness, circular shape with or without central scaling, and other forms such as polycystic plaques, urticarial, ulcerative, or bullous (4,5). While sunlight exposure is not a requirement for rash development, it can worsen existing lesions or trigger new ones. Infants with neonatal lupus erythematosus (NLE) may also exhibit symptoms like petechiae, persistent cutis marmorata, and discoid lesions (4)

Cardiac Manifestations: The most severe cardiac manifestation is congenital heart block (CHB), which carries a 2% recurrence risk in subsequent pregnancies. It is often irreversible and necessitates pacemaker placement (5). Conduction pathway damage usually occurs in utero and is already established at birth. Mortality associated with cardiac NLE, particularly CHB, remains significant, with a rate of around 10% in the neonatal period. The damage the conduction system usually occurs in utero (5). Other cardiac issues can include endocardial fibroelastosis or dilated cardiomyopathy (5). The risk factors for cardiac complications in NLE include maternal antibody status, antibody titers, timing of exposure, and previous affected siblings.

Long-term Concerns: While skin lesions usually resolve within the first six months of life, complications like CHB can have lifelong implications (4). Additionally, a history of NLE does not predispose the child to develop systemic lupus erythematosus (SLE) or other autoimmune diseases later in life.

Other Manifestations: NLE can also manifest with hematologic (thrombocytopenia, hemolytic anemia), hepatic (transient liver enzyme elevation), and neurologic (chorioretinitis, hydrocephalus, etc.) abnormalities.

Diagnostic Assessment:

Maternal Serology: Detection of anti-SSA/Ro and anti-SSB/La antibodies in the mother is diagnostic. The risk of NLE is higher if both antibodies are present, but not all infants born to mothers with these antibodies will develop NLE. This suggests a multifactorial etiology. Higher titers of these antibodies and earlier detection during gestation are associated with an elevated risk (7).

Infant Evaluation: Infants suspected of having NLE should undergo a complete blood count, liver function tests, and urgent referral to a pediatric cardiologist or at least the local emergency department for an electrocardiogram (ECG) (6).

Management and Follow-Up:

Cutaneous NLE: Topical corticosteroids and strict sun protection are the mainstays of treatment (6).

Cardiac NLE: Infants with cardiac NLE require close monitoring. Those with third-degree CHB may require a pacemaker (7).

Pregnancy Monitoring: Pregnant women with anti-SSA/Ro and/or anti-SSB/La antibodies should have fetal echocardiography to monitor for CHB.

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Conclusion:

This case illustrates the complex and varied presentation of NLE in infants arising from the passive transfer of maternal autoantibodies. While the presented child primarily exhibited cutaneous manifestations and did not show major cardiac complications, this case emphasizes the need for a high index of suspicion, as early recognition of the diverse manifestations of NLE may be lifesaving. A collaborative, multidisciplinary approach with dermatology and cardiology specialties working together to ensure early detection, vigilant monitoring, and management is crucial for improving outcomes for affected infants.

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