CASE REPORT

Neonatal lupus with atypical cardiac and cutaneous manifestation

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SUMMARY

Neonatal lupus erythematosus is a rare, passively acquired autoimmune disease, caused by maternal autoantibodies. The most common manifestations are skin rash and congenital heart block. Cutaneous manifestations may be present at birth, but often develop within a few weeks after delivery. Congenital heart block may present as bradycardia in utero or during physical examination at birth. Approximately 40–60% of mothers are asymptomatic when the infants are diagnosed. We present a case of a child, born with erythematosus lesions in the face, scalp, trunk, limbs and nodules/papules on the palmar and plantar surfaces. He also had hepatosplenomegaly and thrombocytopenia. Echocardiography showed hyperechoic lesions on the anterior papilar muscle of the left ventricle and on the lateral cusp of the tricuspid valve. The mother had unexplained fever and vasculitic lesions in her hands and feet. Antinuclear antibodies, anti-SSa/Ro and anti-SSb/La were positive in the mother and child, making the diagnosis of neonatal lupus.

BACKGROUND

Neonatal lupus erythematosus syndrome (NLE) is a rare immune disease that results from the passage through the placenta of anti-SSA/Ro and/or anti-SSB/La antibodies. It is characterised by cardiac, cutaneous, hepatic and haematological abnormalities. In 40–60% of cases, maternal autoimmune disease is unknown. The most common types of skin rashes consist of annular, erythematosus and scaly lesions in the face, scalp and periocular regions. Apart from complete and incomplete congenital heart block, other cardiac manifestations have been reported in children born to affected mothers. We report a case with atypical cardiac and cutaneous manifestations.

CASE PRESENTATION

A male infant was born at 40 weeks gestational age with a birth weight of 3.6 kg, by caesarian delivery due to fetal–pelvic incompatibility. The pregnancy was uneventful with innocent serologies and normal echographies. On physical examination, he had a skin rash with annular erythematosus lesions in the face, scalp, trunk and limbs, petechiae and purpuric lesions and nodules/papules on the palmar and plantar surfaces (figures 1–4). He also had hepatosplenomegaly. Because of an episode of chocking and desaturation with the presence of a murmur in the cardiovascular exam, he was admitted to neonatal intensive care unit. He had a respiratory distress syndrome with oxygen needs

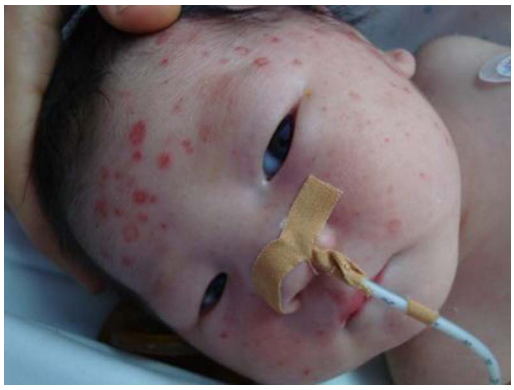

Figure 1 Skin rash with annular erythematosus lesions in the face.

(maximum of 34%) until seventh day of life. Meanwhile, his mother had unexplained fever and vasculitic lesions in her hands and feets and erythema in the nose (figure 5).

INVESTIGATIONS

Laboratory investigations showed thrombocytopenia with normal haemoglobin and leukogram. The septic workup, including haemoculture, was negative and liver function was normal. Serologies for virus, syphilis and toxoplasmose were negative.

The echocardiography showed pulmonary hypertension with hyperechogenic lesions on the anterior papilar muscle of the left ventricle and on the lateral cusp of the tricuspid valve. The ECG was normal.

Cerebral ultrasound was normal. Antinuclear antibodies, anti-SSa/Ro and anti-SSb/La were positive in the mother and child.

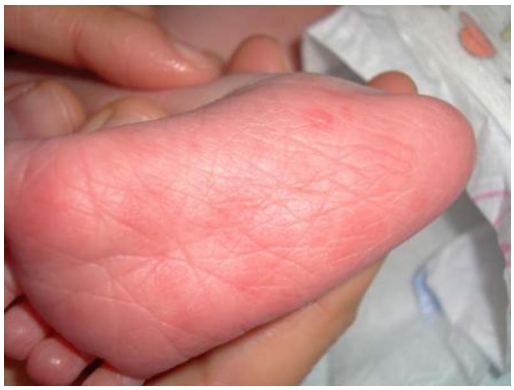

Figure 2 Nodules/papules on the plantar surface.

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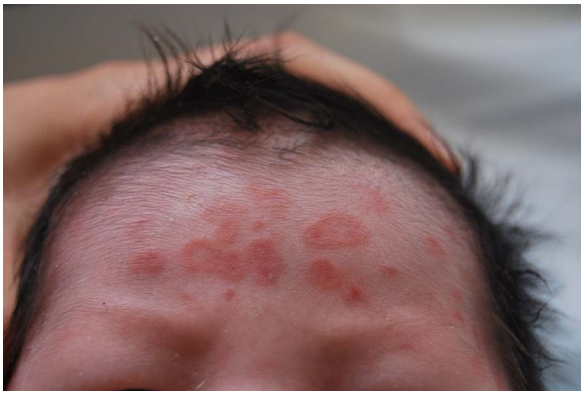

Figure 3 Skin rashes in the face.

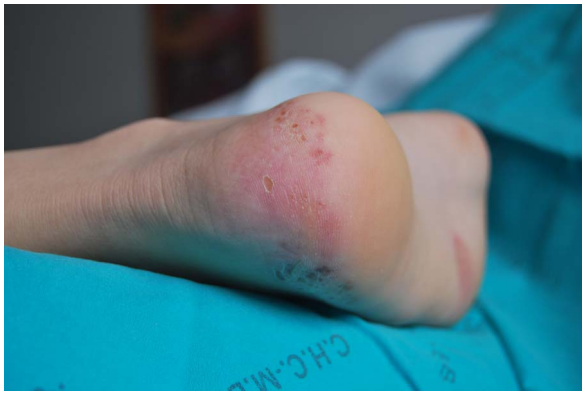

Figure 5 Vasculitic lesions in the mother's foot.

DIFFERENTIAL DIAGNOSIS

Perinatal infections (cytomegalovirus, rubella, group B *Streptococcus* and syphilis) and other non-infectious neonatal diseases such as erythema multiforme.

OUTCOME AND FOLLOW-UP

Oxygen needs were progressively reducing, and at day 7 he was with 21% of FiO_2. Platelet count was normal at the day 15. The echocardiography at discharge did not reveal pulmonary hypertension, but all the hyperechogenic lesions already described were still present. He was discharged home at the 29th day of life, with skin rash less erythematosus, evolving to scaly lesions (figure 6).

At follow-up, the skin lesions resolved within 1 year, leaving hyperpigmented macules on the trunk (figure 7). Neurodevelopment was normal. At the cardiology consultation (2 months), the hyperechogenic lesions disappeared, but he was diagnosed with moderate mitral insufficiency with diuretics needs until nowadays.

The mother was referred to an autoimmune consultation, where she was diagnosed with Sjogren syndrome.

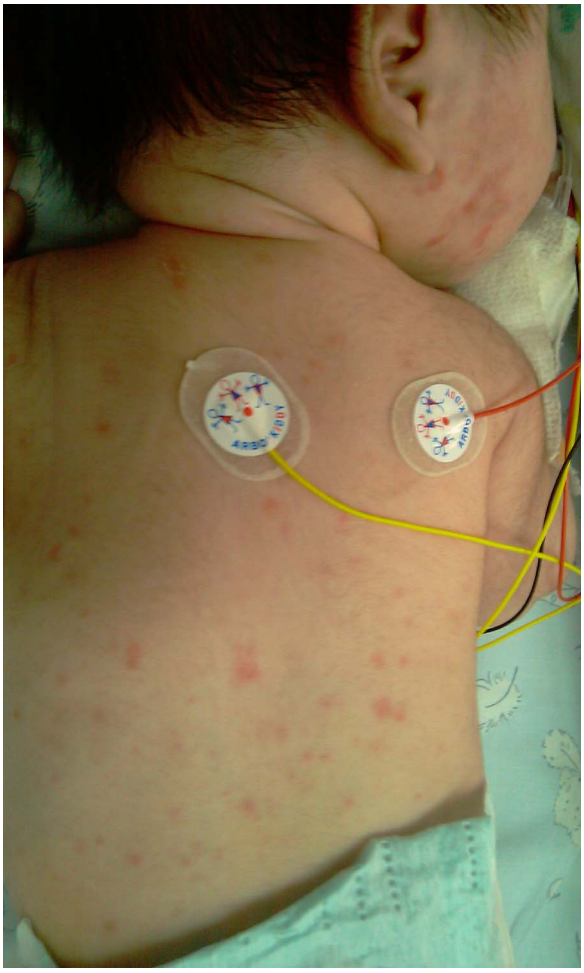

Figure 4 Annular lesions in the trunk.

DISCUSSION

The most common clinical manifestations of NLE are in decreasing order of frequency— dermatological, cardiac and hepatic abnormalities. Some infants may also have haematological, neurological or splenic abnormalities.[1]

Cutaneous lesions most often appear within the first few weeks of life and less frequently at birth. Typical rashes are erythematosus or polycystic plaques with or without fine scales—predominately on the scalp, neck or face (with a periorbital distribution), but similar plaques may appear on the trunk and extremities. The lesions can be urticarial, desquamative, ulcerative or crusted.[1]

There are some uncommon skin lesions in children with NLE: multiple morphea, papulo-erythematosus rash, congenital cutaneous lupus with atrophic lesions.[2] Although rare, cutaneous manifestations of lupus with nodules/papules on the palmar and plantar surfaces has already been described in the literature, in two reports.[3 4] In our case, cutaneous manifestations were

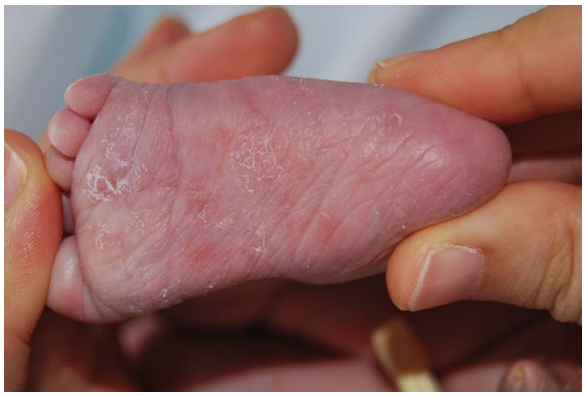

Figure 6 Scaly lesions in the feet.

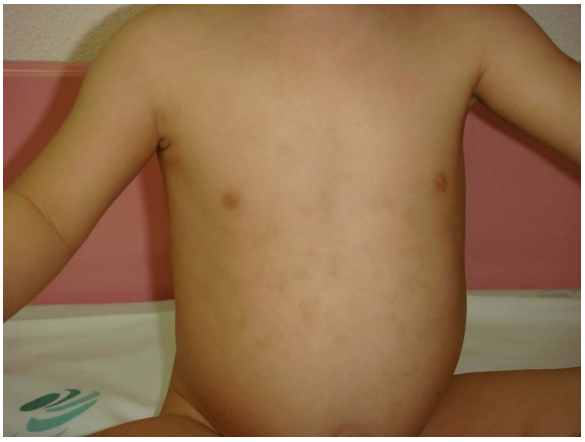

Figure 7 Hyperpigmented macules in the trunk.

atypical: the presence of nodules/papules on hands and feet is unusual, similarly to the presence of skin rash at birth.

In some cases, solar exposure seems to precipitate the eruption. The lesions last for weeks or months and then resolve spontaneously coincident with the disappearance of maternal antibodies. Hypopigmentation or hyperpigmentation is frequent, and occasionally atrophic lesions and scars may develop.[1 2]

The most commonly reported manifestation of neonatal lupus is cardiac disease, with third-degree heart block being the most common finding. Cardiac manifestations usually dictate the prognosis of NLE. Other cardiovascular manifestations of NLE have been more recently recognised: atrial and ventricular arrhythmias and other conduction abnormalities, myocarditis, cardiomyopathy often with endocardial fibroelastosis and structural heart disease, particularly valvular lesions.[5]

These last lesions evolved stenosis, regurgitation and dysplasia. Some authors speculate that these lesions can be caused by inflammation and fibrosis. The hyperechogenic lesions on the anterior papilar muscle of the left ventricle, as seen in our case, could be a fibrotic scar resulting from some type of inflammation during the fetal period, leading posteriorly to mitral insufficiency. Two previous case reports have already documented chordal rupture due to a fibrotic scar formation at the papilary muscle, probably due to anti-SSA antibody.[6 7]

It is common to see that autoimmunity in the mother is not detected until the birth of a child with neonatal lupus. The mothers, who were asymptomatic, eventually develop signs and symptoms of autoimmunity, in particular Sjogren syndrome, systemic lupus erythematosus and undifferentiated connective tissue disease.[8]

In our case, the mother, after a full evaluation, was diagnosed with Sjogren syndrome 3 months after her first child was born.

Children with cutaneous disease may be more prone to develop systemic lupus erythematosus or autoimmunity later in life, mainly due to their genetic predisposition. Mothers of infants with NLE, particularly infants with congenital heart block, have a twofold to threefold increased risk of having an affected infant in a subsequent pregnancy.[1]

Learning points

- Neonatal lupus should be suspected in babies with atypical skin lesions, even if present at birth.
- Besides heart block, other cardiovascular manifestations of neonatal lupus erythematosus syndrome (NLE) have been more recently recognised, including structural heart disease.
- Some of these lesions are the result of ongoing inflammation due to maternal autoantibodies.
- Many mothers are asymptomatic when NLE is diagnosed; symptoms and signs in the child are the first indications of the disease in the mother.

Contributors SM was involved in the conception and design, acquisition of data or analysis and interpretation of data. DFP and ICS were involved in the drafting of the article or revising it critically for important intellectual content. GM gave the final approval of the version published.

Competing interests None.

Patient consent Obtained.

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