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Diagnosis at first sight

Massive hepatomegaly and skin rash as manifestations of congenital infection☆

Hepatomegalia masiva y exantema como manifestación de infección connatal

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Clinical description of the case

A three-month-old male infant brought to the emergency department due to respiratory distress and marked whining in the last few hours. Normal controlled pregnancy with negative serologies in the first trimester. Serology for HIV was repeated in the third trimester and was negative. Despite belonging to the high-risk group for sexually transmitted diseases due to risky sexual relations, other serologies were not extended in the third trimester.

On physical examination, significant abdominal distension was observed, with marked hepatosplenomegaly with normal cardiopulmonary auscultation. He has also presented a hyperchromic

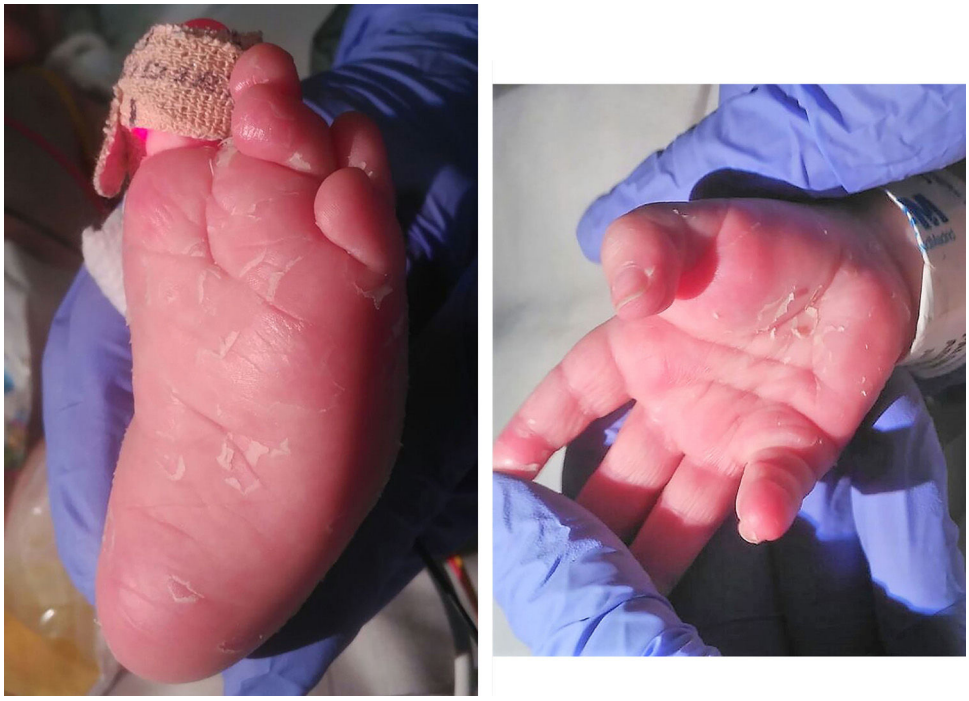

Fig. 1. Palmoplantar desquamation.

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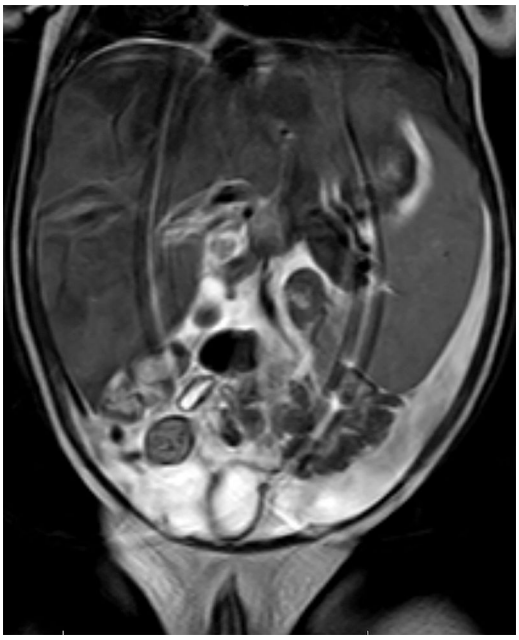

Fig. 2. MRI image of the patient in which moderate–severe hepatomegaly and splenomegaly can be seen.

maculopapular rash for 3 weeks, which on admission was accompanied by palmoplantar desquamation (Fig. 1). Blood tests were performed, revealing anaemia, thrombocytopenia and leukocytosis with neutrophilia. A slight GOT elevation at 89 U/l was observed with other transaminases normal, hyperbilirubinaemia of 2.9 mg/dl (direct 1.6 mg/dl), elevated acute phase reactants and prolonged clotting times. Reactive leukocytosis was seen in the blood smear. The chest X-ray was normal.

Abdominal ultrasound was performed, revealing a possible hepatic mass, and a study with abdominal magnetic resonance imaging was performed, ruling out the presence of space-occupying mass, but showing severe moderate hepatomegaly as a result of diffuse infiltration and splenomegaly of 10 cm (Fig. 2).

The patient's mother presented a hyperchromic macular rash of similar characteristics (Fig. 3) with elevated liver enzymes. Maternal serologies were collected with positive treponemal and non-treponemal tests (1/64 titres), with a diagnosis of secondary syphilis.

The test to detect anti-*Treponema pallidum* IgM was not available, therefore the RPR test was performed with a result of 1/2,048, or four times the maternal values, resulting in a diagnosis of congenital syphilis.

Evolution

Given the patient's respiratory difficulty, he required admission to the PICU and support with non-invasive mechanical ventilation. Antibiotic treatment for congenital syphilis with aqueous intravenous benzylpenicillin was initiated for 15 days. Correction of the analytical alterations and a reduction in hepatosplenomegaly were observed, along with cessation of the respiratory distress and the gradual stabilization of the patient.

In the extension study, a bone series with radiolucent metaphyseal bands in the femur, tibia and distal radius, and periosteal reaction in the pubis, femur and tibia were performed (Fig. 4). The fundus was normal. BAEPs showed bilateral alterations in the wave V appearance threshold, and is to be repeated. Lumbar puncture was performed with no alterations in the cytochemistry, but with a positive VDRL with titres of 1/2.

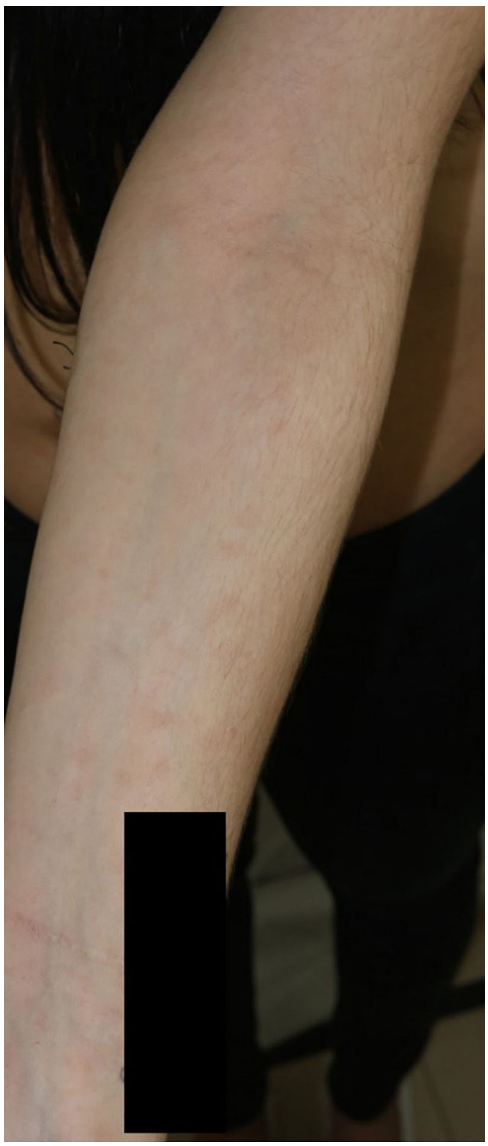

Fig. 3. Hyperchromic macular rash on the forearm of the patient's mother.

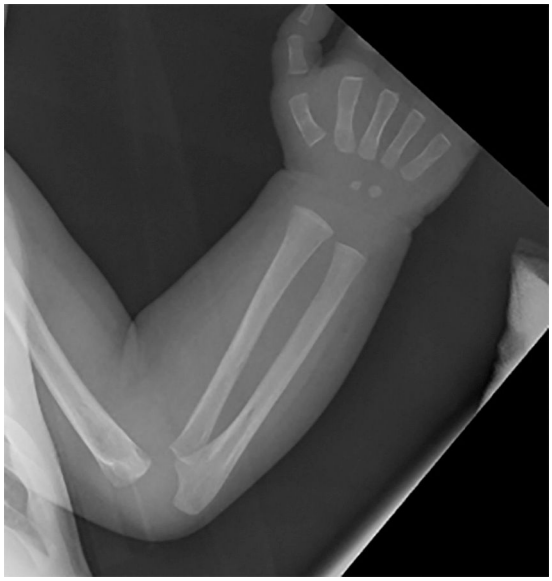

Fig. 4. X-ray with radiolucent bands in distal radial metaphysis.

In the patient's follow-up, he presented gradual improvement of the hepatosplenomegaly with a gradual decrease in non-treponemal antibody titres, with VDRL negativization in cerebrospinal fluid at 6 months and serum RPR negativization at 12 months.

Final comments

Thanks to universal screening for syphilis in pregnancy, which allows the infection to be detected and transmission to the newborn prevented, cases of congenital infection are rare.[1,2] However, there are circumstances where the infection can go unnoticed in pregnancy, such as cases in which the infection occurs during gestation and the diagnostic test is not repeated. The guidelines of the Spanish Society of Gynaecology and Obstetrics (SEGO) recommend repeating the test during pregnancy if it was negative in the first trimester in those patients belonging to high-risk groups.[3] There is currently an increase in the number of syphilis cases in Spain. Although so far these cases have been mostly in men, a rebound of cases in women is beginning to be observed, and it is significant in the reproductive age range.[2]

The rate of transmission to the foetus depends on the stage of the maternal disease, being higher in primary and secondary syphilis, where it reaches over 80%.[4] The presence of compatible symptoms together with direct observation by dark field microscopy, positive PCR for *T. pallidum* or non-treponemal antibody titres four times those of the mother give a diagnosis of congenital syphilis.[5,6] A mortality rate of around 6.5% has been reported, while 33.6% of congenital syphilis cases present with symptoms.[7,8]

Other characteristic manifestations of early congenital syphilis are haemorrhagic rhinitis, cytopenias or alterations in long bones that produce Parrot's pseudoparalysis. Hepatosplenomegaly is caused by infiltration of immune system cells, as well as extramedullary haematopoiesis. Therefore, it is important to conduct an extension study that includes blood count, biochemistry, fundus, bone series, auditory screening and lumbar puncture.[4,9,10] Cases of confirmed syphilis should be treated with aqueous intravenous benzylpenicillin for 10 days. If there is cerebrospinal fluid involvement, a longer treatment should be considered.[4,6,9] It is essential to monitor the patient both clinically and serologically since the negativization of non-treponemal antibodies in serum and cerebrospinal fluid is what confirms the response to antibiotic treatment.[5,6,9] The diagnosis of cases of congenital syphilis makes it necessary to reinforce the performance of serological screening on pregnant women with risk factors in the third trimester of pregnancy.

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