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PATHOLOGICAL FRACTURE AS INITIAL MANIFESTATION OF CONGENITAL SYPHILIS IN A 2-MONTH-OLD INFANT – CASE REPORT

Marcela Tavares Machado
Medical Student
Tiradentes University, Aracaju, Sergipe, Brazil

Enzo Janólio Cardoso Silva
Medical Student
Tiradentes University, Aracaju, Sergipe, Brazil

Gustavo Henrique Cavalcanti Pereira Paixão
Medical Student
Tiradentes University, Aracaju, Sergipe, Brazil

Gabriel Borges Paixão
Medical Student
Tiradentes University, Aracaju, Sergipe, Brazil

Mário Augusto Ferreira Cruz
Pediatric Orthopedic Surgeon
Tiradentes University, Aracaju, Sergipe, Brazil

Izailza Matos Dantas Lopes
Pediatrician, PhD in Health and Environment
Tiradentes University, Aracaju, Sergipe, Brazil

Abstract: Congenital syphilis (CS) is a common pathology in children under 1 year of age in Brazil, which can present with clinical manifestations in various systems, including orthopedic, most often associated with lesions in alternating bands of radiotransparency and radiopacity, areas of osteolysis, and pathological fractures that manifest as exuberant bone calluses. Case report of a patient treated at a public health hospital in Aracaju, Sergipe, diagnosed with CS through radiological manifestations. The case report reaffirms the importance of early diagnosis and treatment, since the infection can trigger complications in infants, such as growth and neurodevelopmental changes, even deafness and blindness.

Keywords: Congenital syphilis, Pathological fracture, Pediatric orthopedics

INTRODUCTION

Congenital syphilis (CS), which currently has a prevalence of 9.6 cases per 1,000 live births in Brazil, is a constant challenge to global public health, impacting thousands of families and revealing the weaknesses of health systems, especially in areas with limited access to quality prenatal care[1]. Despite advances in diagnosis and treatment during pregnancy, vertical transmission remains, causing serious complications in both fetuses and infants. It is worth noting that approximately 40% of pregnant women with untreated syphilis suffer spontaneous abortion[2-4]. In the Brazilian context, current recommendations advise that pregnant women undergo testing at least three times: in the first trimester, as early as possible, in the third trimester, and at the time of delivery[5].

Control of CS is crucial, as this infection can trigger serious complications for babies[6,7]. Although many children are born without apparent signs of the disease, CS can manifest itself in the first years of life—up to the second year—or even later, presenting a wide variety of symptoms[8], a period in which diagnosis may be delayed. These manifestations include dermatological, bone, ophthalmic, auditory, neurological, or dental changes, in addition to other anomalies that can be identified through laboratory tests[9].

Among these manifestations, bone involvement plays a prominent role, since it can present atypically, simulating traumatic conditions or situations of child abuse. With regard to changes, symmetrical lesions are observed in several bones of the extremities, characterized radiologically by the presence of alternating bands of radiotransparency and radiopacity, areas of osteolysis in the metaphysis and periosteal thickening in the diaphysis, as well as pathological fractures in the metaphyses of long bones, which manifest as exuberant bone calluses[10-15].

In this context, recognizing orthopedic lesions as a possible initial presentation of congenital syphilis is essential for the diagnosis and prevention of irreversible sequelae. Thus, the present study aims to report a case of pathological fracture in a 2-month-old infant as an initial manifestation of congenital syphilis, diagnosed based on radiological findings, emphasizing the importance of integrated clinical reasoning. Due to the high incidence of the disease and the systemic inflammatory reaction caused by *Treponema pallidum* in vertical transmission, congenital syphilis is a differential diagnosis for several diseases.

CASE REPORT

Patient A.M.S., female, 2 months old, was taken by her mother to an orthopedic appointment on 01/01/2025, with a report of right arm immobility for 1 week. The patient's guardian did not present a prenatal card, so it was not possible to verify the VDRL history. According to information gathered from the mother, she had a urinary tract infection during pregnancy, which was treated appropriately, and had a vaginal delivery without complications. Physical examination revealed reduced mobility of the right upper limb (RUL) associated with pain when moving the limb, with characteristics of Parrot's pseudoparalysis. Imaging revealed periostitis involving the diaphysis and metaphysis of both humeri, radii, and ulnae (Figures 1 and 2), with evidence of a pathological fracture of the right proximal humerus (Figure 1B). In addition, Wimberger's sign was observed, with bilateral metaphyseal destruction located in the proximal medial tibiae, present in this case mainly on the right (Figure 3A), which is pathognomonic for congenital syphilis. Concluding the radiological observation, there is a sign of metaphyseal serration or Wegner's sign in some of the long bones of both limbs (Figure 3). This raised suspicion of a pathological fracture caused by congenital syphilis.

Laboratory tests were requested (Table 1), conservative treatment was performed for the fracture, and the patient was referred to pediatrics for follow-up of congenital syphilis. On the same day at the pediatric care center ( ), despite the physical examination showing no abnormalities other than MSD mobility, she was admitted, prescribed crystalline penicillin 50,000 IU/kg/dose every 6 hours, and new maternal serological tests and the prenatal card were requested. The mother's test showed VDRL: 1:128, ANTI-HIV 1 and 2 negative, and ANTI--HC: non-reactive.

When retrieving the prenatal card, VDRL was recorded as "non-reactive" at two points during the second trimester. Normal delivery without complications at 39 weeks. Neonatal screening (heart, eyes, tongue, and ears) performed ( ) with no abnormalities, no results from the heel prick test. Birth weight 3.565 kg and APGAR 7/9, adequate for gestational age. According to the daily report, the patient is hemodynamically stable, with no signs of poor perfusion, eupneic, with no neurological changes or other complaints. Urination and bowel movements present and without changes, afebrile.

As a course of action, a red blood cell concentrate transfusion was performed, antibiotic therapy was maintained, and cerebrospinal fluid was collected to rule out neurosyphilis. During hospitalization, the VDRL on 01/08/2025 showed a result of 1:32, and on 01/11/2025, 1:512.

After treatment was completed on 01/13/2025, the patient, aged 2 months and 12 days, continued to be followed up at the pediatric outpatient clinic. At the first consultation on January 21, 2025, at 2 months and 20 days of age, the mother reported complaints of secretory cough, regurgitation after breastfeeding, and vomiting during coughing fits, with no complaints related to mobility. Regarding her pregnancy history, she had 11 prenatal consultations, with the mother testing "non-reactive" for serology tests (VDRL, HIV, HBsAg, and HCV), in addition to testing "non-immune" for toxoplasmosis and "immune" for rubella. In addition, the child had adequate lifestyle

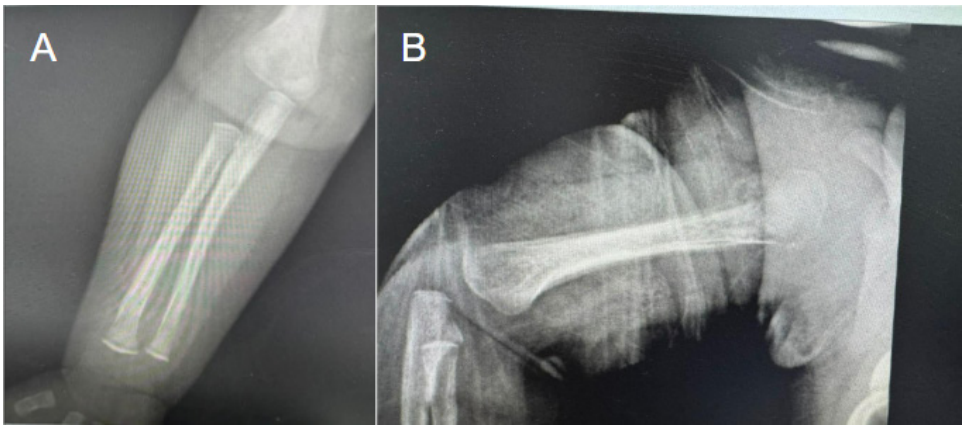

Figure 1 - Periostitis involving the diaphysis and metaphysis of the right humerus, radius, and ulna (A and B); Presence of pathological fracture in the right humerus (B).

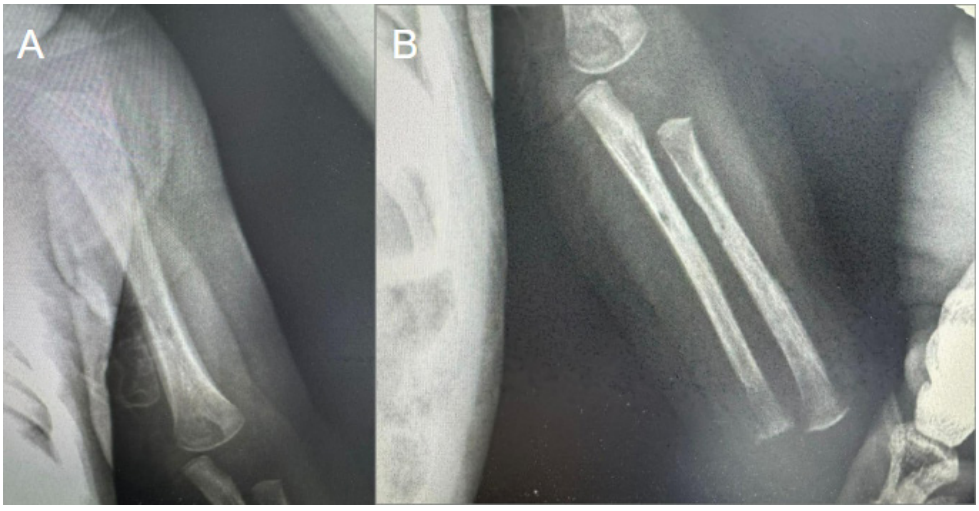

Figure 2 - Periostitis involving the diaphysis and metaphysis of the left humerus, radius, and ulna (A and B).

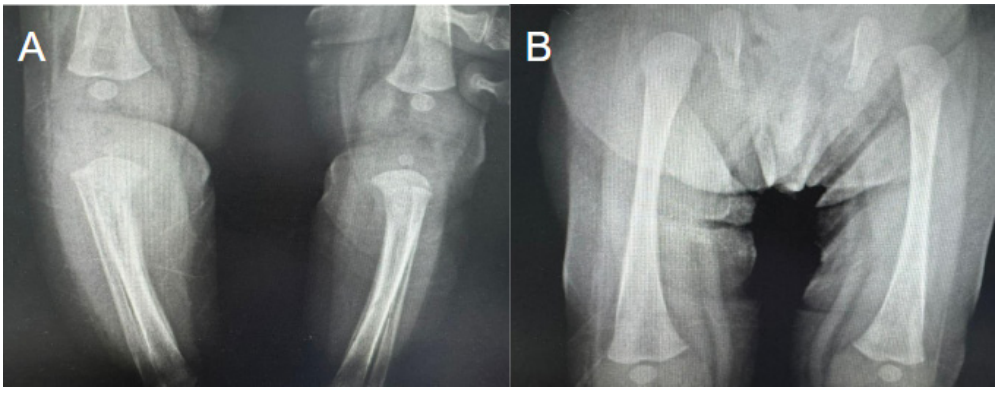

Figure 3 - Presence of Wimberger's sign (A); Metaphyseal serration, or Wegner's sign in long bones (A and B).

Parameter Evaluated	Result	Reference Value for Age Group
Hemoglobin	7.8 g/dL	10.0 - 20.0 g/dL
Hematocrit	24.8	31.0 - 55.0
White blood cells	8,200/mm3	5,000 - 15,000/mm3
Platelets	681,000/mm3	140,000 - 380,000/mm3
C-reactive protein	48 mg/dL	< 1.0 mg/dL
Urea	16 mg/dL	9 - 40 mg/dL
Creatinine	0.3 mg/dL	0.3 - 0.7 mg/dL
TGO	300 U/L	< 45 U/L
TGP	270 U/L	< 40 U/L
ESR	60 mm/h	< 10 mm/h
VDRL	1:768	Non-reactive
HBSAg	Non-reactive	Non-reactive
Anti-HIV 1 and 2	Non-reactive	Non-reactive

Table 1: Tests collected on admission (01/01/25)

Parameter Evaluated	Result	Reference Value for Age Group
Alkaline phosphatase	558 U/L	29-474 U/L
TGO	68 U/L	< 45 U/L
TGP	76 U/L	< 40 U/L
Gamma-GT	183 U/L	< 81 U/L
Albumin	2.4 g/dL	2.8 - 5.4 g/dL
VDRL (child)	1:32	Non-reactive
VDRL (maternal)	1:16	Non-reactive
Serum Iron	45.7 ng/mL	40 to 120 ng/mL

Table 2: Tests requested during outpatient follow-up (01/28/25)

habits, social and dietary history, and an up--to-date vaccination schedule.

On physical examination, the patient presented with 5 cm hepatomegaly, and laboratory tests were requested due to suspicion of congenital syphilitic hepatomegaly, in addition to vitamin D and ferrous sulfate supplementation. She weighed 5.360 kg (Z-score 0), had a head circumference of 40 cm (Z-score between 0 and +1), a height of 57 cm (Z-score between -2 and 0), and a BMI of 16.4 (Z-score between 0 and +1), which were appropriate for her age.

At the follow-up appointment on January 28, 2025, at 2 months and 27 days of age, she brought laboratory test results (Table 2) showing liver transaminases and canalicular enzymes above the upper limits of normal, in addition to VDRL 1:32, maternal VDRL 1:16, and serum iron 45.7. At this consultation, he had no complaints or changes in physical examination and neuropsychomotor development. Suspicion of congenital syphilitic hepatitis was confirmed due to elevated liver enzymes.

At follow-up on March 11, 2025, the infant was 4 months and 10 days old, with the mother reporting no new complaints. Physical examination and neuropsychomotor development analysis revealed no significant changes, including abdominal examination. He weighed 6.820 kg (Z-score 0), was 63 cm tall (Z-score between 0 and +2), had a head circumference of 42 cm (Z-score +1), a BMI of 17.1 (Z-score between 0 and +1), and a weight gain of 29 grams/day.

On April 22, 2025, at 5 months and 21 days, she weighed 7.260 kg (Z-score between 0 and +2), measured 63.5 cm (between Z-score 0 and - 2), head circumference 43 cm (between Z-score between 0 and + 1), and BMI 18.0 (between Z-score between 0 and + 1). The mother had no complaints, and physical examination of the abdomen revealed diastasis of the rectus abdominis muscles and a 1 cm umbilical hernia, with no visceromegaly. In addition, she brought the results of the laboratory tests requested at the last consultation (Table 3).

Finally, on May 21, 2025, at 6 months and 20 days, she weighed 7.905 kg (between Z-score 0 and +2), measured 66 cm (between Z-score 0), head circumference 44 cm (between Z-score +1 and +2), and BMI 18.4 (Z-score 0 and +1). As a course of action, laboratory tests were requested and a return visit was scheduled. However, the parents and patient stopped attending appointments, thus losing outpatient follow-up.

DISCUSSION

The incidence rate of congenital syphilis in Brazil in 2024 showed a slight reduction of 5.0% compared to 2021, reaching 9.6 cases per 1,000 live births[16] . However, according to the Pan American Health Organization (PAHO), it still accounted for the majority of cases, despite the implementation of projects such as "Sífilis Não!" (No to Syphilis), launched in 2016[17] . Among affected infants, it is estimated that only half of the cases of congenital syphilis in Brazil had undergone long bone radiology in 2024. Among children with known results, 720 had bone changes[16], however, the occurrence of pathological fractures as an initial manifestation is considered rare.

Between 1998 and 2024, 3,739 deaths from congenital syphilis in children under 1 year of age were recorded in the Mortality Information System (SIM)[16]. Factors such

Parameter Evaluated	Result	Reference Value for Age Group
Hemoglobin	11.2 g/dL	10.0 - 20.0 g/dL
Hematocrit	32	31.0 - 55.0%
White blood cells	8,820/mm3	5,000 - 15,000/mm3
Platelets	687,000/mm3	140,000 - 380,000/mm3
Total bilirubin	0.1 mg/dL	< 1.2 mg/dL
Direct Bilirubin	0.1 mg/dL	< 0.3 mg/dL
Indirect Bilirubin	0.1 mg/dL	< 1.0 mg/dL
Alkaline phosphatase	347 U/L	29-474 U/L
Gamma-GT	16 U/L	< 81 U/L
TGO	42 U/L	< 45 U/L
TGP	17 U/L	< 40 U/L
Serum iron	67 ng/mL	40 to 120 ng/mL
C-reactive protein	5.4 mg/mL	< 1.0 mg/dL
Reticulocytes	2.2	1 - 2
Ferritin	117 ng/mL	50 - 200 ng/mL
VDRL (child)	1:8	Non-reactive
VDRL (maternal)	1:4	Non-reactive

Table 3: Tests requested during outpatient follow-up (04/22/25)

Date	VDRL Child	VDRL Maternal
Prenatal	-	Non-Reactive
01/01/2025	1:768	1:128
01/08/2025	1:8	-
01/11/2025	1:512	-
01/28/2025	1:32	1:16
04/22/2025	1:8	1:4

Table 4: Comparison of VDRL results throughout outpatient follow-up

as inadequate prenatal care and delayed diagnosis and treatment favor the onset of complications that can range from asymptomatic manifestations to fetal death[18].

The clinical manifestations of early congenital syphilis occur in the first 2 years of life, most commonly in the first 3 months after birth[1]. The disease can be asymptomatic in up to 90% of newborns[11], but cutaneous, mucosal, hematological, and hepatological manifestations may occur, these being the most frequently reported signs and amenable to prenatal diagnosis[19].

In the orthopedic field, fractures are less frequent findings, but they are associated with greater severity[14] . A differential diagnosis with child abuse should also be made, which, in the reported case, would be disregarded due to the findings of hepatomegaly associated with changes in liver enzymes and anemia, as these are manifestations related to the diagnosis of congenital syphilis[20] . Bone changes are associated with local and systemic inflammatory processes, which facilitate the occurrence of trauma and infections, with periosteal reaction being the most common, representing 34% of cases[10] . In the case presented, diaphysis in long bones, signs of Wimberger and Wegner, and pathological fracture of the right proximal humerus were the changes observed.

Among the bone manifestations reported by the mother, one that deserves mention is Parrot's pseudoparalysis, characterized by reduced or stopped limb movement, which can be accompanied by sudden pain, a condition that is easily confused with trauma or other neuromuscular diseases[6]. Bone changes are often underdiagnosed due to the lack of radiological examinations and studies in the literature on the prevalence and prognosis of children with this comorbidity, which compromises medical evaluation. Pseudoparalysis, when its timely and early treatment is delayed, corroborates with the emergence of bone deformities, such as syphilitic facies, characterized by a saddle nose, Olympic forehead, shortening of the maxilla, and saber tibias, in addition to delayed neurodevelopment and growth in children. The pathophysiology, described by Joseph Marie Jules Parrot in the 19th century[21], is related to syphilitic osteochondritis and periostitis, mainly affecting the long bones. The radiograph in this case report reveals bone rarefaction, periosteal thickening, and changes in the bone growth cartilage[4].

Studies show that radiography of long bones can be useful for early identification of fractures in about 20% of asymptomatic patients with reactive treponemal tests and 60-80% of patients with clinical signs and children of mothers who were inadequately treated or untreated. Follow-up of radiological changes is performed after 1 year of age, and after 2 to 4 years, the lesions disappear. The patient in the report did not undergo further radiographic examinations because the Pediatrics team was waiting for her to reach one year of age. In addition to the manifestations noted in the reported case, there are other radiographic changes that are more frequent in early CS, such as metaphyseitis (lesions in alternating bands of radiotransparency and radiopacity), periostitis (irregular periosteal thickening), epiphysitis (enlargement and irregularity of the epiphyseal plate), and Wimberger's sign (localized symmetrical demineralization and bone destruction of the medial portion of the proximal metaphysis of the tibia), which meet the criteria for presumed congenital syphilis[4].

Although considered by many authors to be an additional expense, long bone radiography is beneficial, as approximately 60 to 90% of infants are asymptomatic in the clinical setting, but 20% of them have skeletal manifestations of congenital syphilis, comprising a significant portion[22] .

According to the Ministry of Health, testing of pregnant women is recommended at least three times: in the first and third trimesters of pregnancy; at the time of delivery; or in cases of miscarriage/ d fetus. The diagnosis can be made with immunological tests, such as the non-treponemal test (VDRL), which should be used for confirmation and evidence of dilution decline, and the treponemal test (rapid test)[4]. After treatment, the infant should have the VDRL test performed quarterly until it becomes negative[6]. If the VDRL does not show a significant reduction after 6 months, or if symptoms persist, retreatment should be considered[8].

The approach adopted in this case followed current guidelines, with administration of intravenous crystalline penicillin at a dose of 200,000 IU/kg/day, due to its high efficacy in eradicating *Treponema pallidum* and its ability to prevent late complications, being considered the only effective antibiotic for preventing vertical transmission of the disease[6] . A conservative approach to the pathological fracture was indicated because, after treatment of the underlying cause, spontaneous consolidation occurs satisfactorily in most cases, without the need for surgical intervention. However, orthopedic and physiotherapeutic follow-up is necessary for the child's full recovery.

CONCLUSION

Congenital syphilis (CS) is a common pathology in children under 1 year of age, which can present with clinical manifestations in various systems, making early diagnosis difficult and remaining an important public health problem. This report highlights the importance of the differential diagnosis of pathological fractures with congenital syphilis, showing the importance of radiography, which can be useful even in asymptomatic patients, allowing for timely treatment, since the infection can trigger complications with neurodevelopmental and growth changes, as well as permanent sequelae in infants, such as bone deformities, deafness, and blindness.

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