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"PREVENTABLE TRAGEDY : A CASE OF CONGENITAL SYPHILIS HIGHLIGHTING MISSED ANTENATAL SCREENING OPPORTUNITIES"

Paediatrics

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ABSTRACT

Congenital syphilis presents a significant global burden, contributing to fetal loss, stillbirth, neonatal mortality, and congenital infection. Despite the target established in 2007 by the World Health Organization (WHO) of fewer than 50 cases per 100,000 live births, the global incidence is on the rise, particularly in low- and middle-income regions. T. pallidum can be transmitted from the mother to the fetus transplacentally or during labor when traversing the vaginal canal. Congenital syphilis is associated with significant morbidity and mortality. Despite an increased incidence in recent decades worldwide, including in developed countries, early detection and treatment remain crucial. We present a case of a 3-month-old child with skin rashes, hepatosplenomegaly and diagnosed with early congenital syphilis due to missed opportunities for antenatal screening. This case highlights gaps in prenatal care and emphasizes the importance of adherence to recommended screening protocols during pregnancy.

KEYWORDS

hepatosplenomegaly, maculopapular rash, treponema pallidum

INTRODUCTION :

Sir William Osler aptly quoted" He who knows syphilis, knows medicine."

Congenital syphilis was initially described in 1497 and is, therefore, the oldest recognized congenital infection. It is acquired transplacentally from an infected mother to her fetus (1). The rate of reported congenital syphilis in the United States has increased dramatically since 2012. During 2019, a total of 1,870 cases of congenital syphilis were reported, including 94 stillbirths and 34 infant deaths. The 2019 national rate of 48.5 cases per 100,000 live births represents a 41% increase relative to 2018 (34.3 cases per 100,000 live births) and a 477% increase relative to 2012 (8.4 cases per 100,000 live births). During 2015–2019, the rate of congenital syphilis increased 291.1% (12.4 to 48.5 per 100,000 live births), which mirrors increases in the rate of primary and secondary syphilis among females aged 15–44 years (a 171.9% increase, from 3.2 to 8.7 per 100,000 females) (2). Overall risk of transplacental infection of the fetus is about 60 to 80%, and likelihood is increased during the 2nd half of the pregnancy. Untreated primary or secondary syphilis in the mother usually is transmitted, but latent or tertiary syphilis is transmitted in only about 20% of cases (3). Therefore, physicians should be aware of the diverse clinical features of syphilis to enable early diagnosis of the disease. We report a case of congenital syphilis in a 3-month-old infant who had skin eruptions and hepatosplenomegaly. His mother was tested negative during her antenatal screening; however, she tested positive for syphilis later(4).

Case Report:

A 3-month-old boy was admitted due to a 2-week history of an asymptomatic, skin eruption over upper and lower limbs. His parents had taken him to several pediatric and dermatologic clinics, but could not identify his illness. He had been born to a 27-year-old, G1P1A0 mother who had adequate prenatal care at other clinics. Further history revealed no known high-risk behaviours, but her socio-economic status suggested limited access to healthcare services. She was tested for syphilis once in pregnancy and was told that the tests were negative.

The patient was delivered at 39+6 weeks of gestational age by normal spontaneous vaginal delivery and the birth weight was 2800 g. Throughout the neonatal period, he remained asymptomatic, his skin color remained normal. Physical examinations revealed no abnormalities across all systems.

At admission, upon physical examination, his height was 61.5 cm (25 to 50th percentile), weight was 6.5 kg (25 to 50th percentile), and head circumference was 39.5 cm (10 to 25th percentile). He was only feeding a small amount of milk and his activity was decreased. His abdomen was distended with hepatosplenomegaly. He had characteristic mucocutaneous rash, presenting with erythematous maculopapular or bullous lesions, followed by desquamation involving hands and feet.(Fig. 1A and Fig.1B)

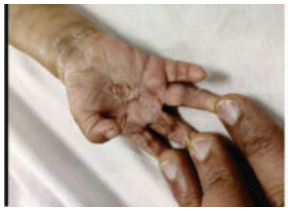

Fig.1a

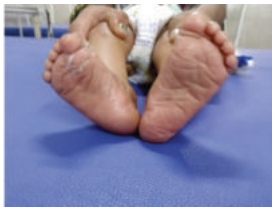

Fig.1b

The infant's laboratory evaluation revealed positive Treponema pallidum particle agglutination (TPPA) and rapid plasma reagin (RPR) tests, with an RPR titer of 1:32. The infant also had elevated liver enzymes and anemia. **This confirmed the diagnosis**.

Long bone x-rays taken, reported to be normal. An ophthalmology reference was taken for ocular syphilis which was reported to be normal. Hearing tests were found to be normal.

Investigations:

1.CBC	Hb/Hct:9.3/30, TLC:14,000 (P:73, L:19, E:1, M:5) Plt:4.5lakh
2.Peripheral smear	Anisocytosis, Poikilocytosis, Microcytic hypochromic anemia
3.Reticulocyte count	0.5% (0.5%-1.5%)
4.RBC Indices	MCV: 76, MCH: 21, MCHC:28
5.CRP	22mg/dL(0-6mg/dL)
6.RBS	92mg/Dl
7.Total bilirubin	3.8mg/dL
8.Direct bilirubin	1.2mg/dL
9.AST	168 U/L
10.ALT	68 U/L
11.ALP	140
12.Total Protein	6.8
13.Albumin	4.5
14.Sodium	136mEq/L
15.Potassium	4.4mEq/L

16.Chloride	97mEq/L
17.CSF	Within normal limits
18.Blood Cultures	No growth
19.Treponemal serological test (TPPA)	Positive
20.RPR titre	1:32

The infant was started on IV acqueous crystalline penicillin G 50,000units/kg/dose 12th hourly for 7 days and every 8hrs thereafter for a total of 10days, and there was significant clinical improvement by the one-month follow-up, with complete resolution of skin rashes and hepatosplenomegaly. (Fig.2A and 2B- healed lesions)

Fig. 2a

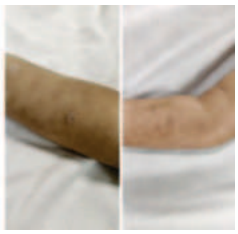

Fig. 2b

DISCUSSION :

Syphilis is often referred to as "the great masquerader". Due to its versatile clinical presentations, recognition of syphilis infection can be a challenge even for the most experienced clinicians. Furthermore, the natural history of syphilis, whether treated or untreated, can be quite unpredictable(5). T. pallidum can enter the fetal blood circulation through the placenta and cause congenital syphilis. The risk of syphilis transmission to the fetus through the placenta is 60–80%, and the risk increases in the second half of pregnancy. Neonatal exposure to secretions, blood or genital lesions of women with syphilis during delivery could also result in T. pallidum infection. Congenital syphilis most commonly occurs in infants of infected women who have not received adequate treatment or any treatment(6). Congenital syphilis is arbitrarily divided into early and late congenital syphilis. The former has onset of clinical manifestations before 2 years of age while the latter has onset of clinical manifestations after 2 years of age (usually manifesting around puberty). Although the majority of infants with congenital syphilis are asymptomatic at birth, among symptomatic ones, clinical manifestations of early congenital syphilis include skin rash, snuffles, jaundice, hepatomegaly with or without splenomegaly, fever, generalized lymphadenopathy, and failure to thrive(7). Rhinitis ("snuffles") has been reported in as many as 73% of infants. It usually develops in the 2nd–3rd week of life and may be the earliest clinical sign(8). Our case had involvement of skin and viscera. This case highlights a preventable instance of congenital syphilis due to missed opportunities for maternal screening during pregnancy. According to CDC guidelines, all pregnant women should be screened for syphilis during their first prenatal visit, with repeat screening in the third trimester and at delivery for high-risk individuals.

The WHO emphasizes that congenital syphilis stands as the most preventable cause of stillbirth and advocates for antenatal screening programs for all pregnant women, alongside treatment of all positive cases and their partners. In our patient's case, her mother underwent syphilis screening during a routine visit, and TORCH results returned negative. Clinical symptoms in our patient manifested at 3 months. This study shows the majority of infants with congenital syphilis are asymptomatic at birth and in most untreated children clinical features manifest within the first 3 months of life(9). Previous studies have shown that maternal serological titers are positively correlated with the risk of congenital syphilis. In our study, baby was diagnosed with early congenital syphilis even with mother being tested negative during pregnancy. If pregnant women are not screened for syphilis before childbirth, and those with syphilis are not treated or do not undergo syphilis interruption treatment, the incidence of adverse pregnancy outcomes is as high as 69 %, and the proportion of children diagnosed with congenital syphilis is even higher. Therefore, screening for syphilis during pregnancy and early standardized treatment of infected pregnant women are of great importance in reducing the incidence of congenital syphilis(10). The standard treatment for early congenital syphilis was 50,000IU/Kg of IV Acqueous Crystalline Penicillin G 12th hourly for first 7 days followed by every 8th hour for next 3 days as per CDC 2021 guidelines for a confirmed case of congenital syphilis. Cho et al. administered Penicillin G at 50,000 IU/kg/dose every 8 hours for 14 days to an extremely preterm newborn baby in their study(11). An alternative treatment is aqueous procaine penicillin G, 50,000 units/kg/day intramuscularly for 10 to14 days. For infants who are at least four weeks of age or older children, treatment is aqueous penicillin G, 50,000 units/kg/dose every 6 h intravenously for 10 to 14 days because of the difficulty in excluding neurosyphilis in this age group(12). It is recommended that NTT(non-treponemal test) titres should be monitored with 3-month-intervals after the treatment of seroreactive newborns. Fourfold reduction is expected by the sixth month in the titres of the patients receiving adequate treatment. If there is no decrease, or an increase is detected in the titres, the cases should be re-evaluated in terms of treatment resistance, efficacy and neurosyphilis. The main drawback to preventing congenital syphilis is the inadequate diagnosis and treatment processes of antenatal regular follow-ups of the pregnant women(13). This case reinforces the importance of early and repeated screening during pregnancy, as the risk of congenital syphilis is significantly reduced with timely maternal treatment.

CONCLUSION :

Congenital syphilis is entirely a preventable tragedy. With the regular follow-up of syphilis during pregnancy, the effective treatment of the pregnant women, and with the early treatment of infants after a rapid evaluation, the complications of congenital syphilis can be prevented. This case underscores the need for universal screening during pregnancy and highlights the adverse outcomes that can occur when maternal screening is missed or misinterpreted. Addressing the barriers to healthcare access and ensuring adherence to screening protocols are critical steps in reducing the incidence of congenital syphilis. Public health efforts should focus on improving awareness and providing comprehensive care to all pregnant women.

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Conflicts Of Interest : Nil

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