

Treponema infection in an infant differential diagnosis: congenital syphilis versus child abuse

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Abstract: Syphilis is a sexually transmitted disease, which was discovered that it might have been dated back to as early as 3000 BC [1]. Among the multiple forms of manifestation, the most redoubtable is the materno-fetal transmission of the disease, which can lead either to the intrauterine death of the fetus and miscarriage or stillbirth, or to the appearance of congenital syphilis [1]. The discovery of clinical signs of the disease in a child, followed by the confirmation of the syphilis diagnosis, often entails the differentiation of the congenital infection from abuse cases. In the current paper we describe the case of a 1-year-old girl who presented with perianogenital lesions suggestive of syphilis infection, with an onset around 10 months of age. The detection of positive serologies among several family members and the precarious living conditions led to the suspicion of abuse. After investigating the case, the diagnosis of early congenital syphilis was established, followed by an adequate treatment.

Key Words: sexually transmitted disease, congenital syphilis, physical abuse.

Congenital syphilis is a disease of an infectious nature, with maternal-fetal transplacental transmission, from a mother affected by a recently evolutionary syphilis. The microorganism responsible for the infection is a spirochaete – *Treponema pallidum*. The disease was first described in a British 17th century pediatric textbook, although Paracelsus (1493-1541) suggested its in utero transmission [2].

In developed countries, the incidence of congenital syphilis has been continuously declining during 1995-2005, parallel to the prevalence of the infection among the female population [3]. In the U.S, there has been recorded a decrease in the incidence of the disease by 14.1% between 1995-2005 (from 30.4 cases/100.000 live births in 1996 to 8.2 cases/100.000 live births in 2005); on the other hand, between 2005-2008, there has been reported a slight increase in incidence (from 8.2 to 10.1 cases/100.000 live births) [4,5,6].

In Romania, although there were no reports of congenital syphilis cases before 1990, since then a massive increase in the incidence of the disease has been enlisted, due to the improvement of screening methods. After the year 2000, a decrease in incidence has been reported, reaching a number of 9703 cases between 2007 - 2009, according to the Laboratory of Epidemiology of the Dermato -Venereological Center from Bucharest and the Center for Sanitary Calculus and Statistics of the Ministry of Health. However, it is mentionable that statistics do not always reflect the real situation, since it is possible for some cases not to be reported.

Clinical Case

We present the case of a 1-year-old girl, of urban provenance, weighing 9 kg, born at term, through a physiological route. Around the age of 10 months, the child's mother observed the appearance of perianogenital lesions, for which reason she was

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directed towards our department. From her personal pathological antecedents, we mention an episode of asthmatic bronchitis at the age of 6 months. In regards of the patient's family history, the detection of a positive serology in both parents, as well as an aunt and her husband are notable.

Clinical examination.

Papulo-erosive lesions with a diameter of ~ 5 mm were found, located in the perianogenital area, along with an erythematous edematous exanthema of the buttocks

and genital region, developed in the diaper-covered area. In terms of neuromotor development, the child was able to hold her head, to sit up and to walk supported – characteristics attributed to a normal developmental age of 10 months (gross motor developmental quotient = 83). Otherwise, general physical examination was normal.

Paraclinical investigations.

Laboratory exams identified a positive serology for syphilis: VDRL ++++ and TPHA ++++ (1/80 dilution).

The penicillin prick test was negative. The remaining examinations were within normal limits. The CSF examination could not be performed, due to the lack of cooperation from the child's family.

Diagnosis. In accordance with the CDC criteria[7], we established the diagnosis of a compatible case of early congenital syphilis.

Treatment. The patient received antibiotic therapy with crystalline penicillin G, in accordance with the treatment regimens recommended by the Ministry of Health, combined with prednisone, in order to avoid the Jarisch-Herxheimer reaction. The evolution was favourable, with a resolution of the lesions. After a two week interval, the patient must return for a new therapeutic course. The first serological check-up will follow within 90 days after the treatment is concluded.

Discussions

Congenital syphilis

The risk of vertical transmission of the infection and disease manifestation in the offspring is dependent on the stage of maternal infection, the most dangerous period being represented by the recent,



Figure 1. Congenital syphilis, condylomata lata – clinical aspect



Figure 2. Perianal papulo-erosive lesions

virulent syphilis of the mother [1,8,9]. According to Kassowitz's law, the longer the duration of the untreated infection before the pregnancy, the less likely it is that intrauterine and neonatal mortality or congenital syphilis will occur [10,11,12]. In primary and secondary forms of the untreated maternal disease, the risk of transplacental transmission of the infection is very high, approaching 100%, while in late stages of syphilis, it varies between 10-30% [13].

Treponemal invasion of the fetus begins after 18 weeks of gestation, when the treponeme may cross the placenta, due to the involution of the cytotrophoblast (layer of Langhans) [1,10]. The spectrum of fetal manifestations is determined by the intensity of the maternal infection, so that it was estimated that in women with disease of few years duration, about half of the pregnancies will be affected, half of which will end in intrauterine fetal death (with miscarriage or stillbirth), and the other half will lead to perinatal death or the clinically manifest form of the neonatal infection – congenital syphilis [8,9]. More than 50% of affected newborns can be asymptomatic, so that many cases remain unreported [8,14].

Congenital syphilis is classified into several clinical forms [1,4,15]. In the case of early congenital syphilis, the clinical manifestations can become apparent at birth or during the first 2 years of life and they are similar to those of acquired secondary syphilis. Late congenital syphilis occurs after the second year of life, anytime within the first 2 decades. The stigmata are characterised by cicatriceal lesions and congenital dystrophies caused by the syphilis infection, that have an important role in the differentiation from acquired forms of the disease. Among the stigmata of congenital syphilis infection, Hutchinson's triad, which is represented by interstitial keratitis, Hutchinson teeth and deafness, is pathognomonic.

The diagnosis of congenital syphilis is primarily a clinical one, which is supported by the paraclinical examinations, among which we mention the following: detection of *T. Pallidum* from a florid lesion or lymph-node puncture by dark-field microscopy and serological tests - non-treponemal tests (VDRL, USR, RPR, TRUST) or treponemal tests (TPHA, FTA, FTA-abs, ELISA, TPI) [1,4]. Due to the central nervous system involvement in ~ 40% of the cases, CSF examination is recommended. Among the usually performed set of tests are also included: a complete blood count, liver functions tests, tests for other sexually transmitted disease (HIV, chlamydia, gonorrhea, viral hepatitis B and C) [16].

According to the CDC (Atlanta, 1998), when the diagnosis of congenital syphilis is being established, several possibilities must be taken into consideration. Thus, the diagnosis of a confirmed case of congenital syphilis is established after detecting the etiologic agent by means of dark-field microscopy, immunofluorescence or other specific staining methods of specimens obtained from lesions, placenta, the umbilical cord or other tissues.

The diagnosis of a compatible case of congenital syphilis is established in several situations such as when a newborn whose mother presented at birth with an untreated or inadequately treated syphilis infection, including recurrences, reinfection, antibiotherapy with other drugs than penicillin, penicillin-therapy that started within less than 4 weeks before birth or the lack of any supporting documents concerning the treatment or post-therapeutic serological surveillance; the clinical manifestations of the child are not taken into consideration in these cases. Another situation in which the diagnosis of a compatible case of congenital syphilis is considered includes the case of a newborn or a child with a positive serology for *T. Pallidum* and any of the following criteria: clinical signs of congenital syphilis, radiological changes found in congenital syphilis, CSF changes in the absence of other causes (positive VDRL, an increased proteinorahia - 0.46-2g/l, moderate mononuclear pleocytosis - 10-400 cells/ml), positive treponemal tests (ELISA for IgM class anti-treponemal antibodies, FTA-Abs for 19S-IgM) [4,7].

Differential diagnosis includes multiple disease. Syphilitic pemphigus requires differentiation from endemic pemphigus, that does not involve the palmo-plantar region. Snuffles must be distinguished from a common viral rhinitis or diphtheric coryza, which is characterized by the frequent association with angina. Bone lesions must be differentiated from: severe malnutrition, rickets, poliomyelitis (with an onset after 4 months of age), obstetrical paralysis (becomes apparent immediately after birth), physical abuse. Hepatosplenomegaly may be confused with other types of hepatosplenomegaly. Dental abnormalities must be distinguished from those caused by other infections or maternal disease that occurred during the second half of the pregnancy [1,4].

The treatment of choice in syphilis is represented by antibiotherapy with penicillin, which can be combined with low-dose corticosteroids, in order to avoid the Jarisch-Herxheimer reaction [1]. According to the CDC (Atlanta, 1998), the therapeutic decision must be based upon the following aspects: identification of the maternal treponemal infection and implementation of an adequate treatment, the presence of clinical and paraclinical signs of syphilis in children, as well as the comparison between the child's non-treponemal antibody titres and the ones found in the mother[7]. Considering the fact that an adequate treatment of the mother before the 18th week of gestation prevents the transplacental transmission of the disease, public education promoting a rigorous sexual hygiene and implementing effective screening methods could successfully prevent the appearance of congenital syphilis [8,10,17]. The American College of Obstetricians and Gynecologists along with the American Academy of Pediatrics recommend screening for syphilis at the first prenatal examination for all women and again at 32-36 weeks of pregnancy for those included in the high risk groups [16,18]. In 2003, the American Academy of

Pediatrics recommended that serological tests should be repeated at delivery [16,19,20]. A study from Haiti has shown that decentralizing screening for syphilis could decrease the incidence of congenital infection, even in peripheral areas, with a limited infrastructure [8,21].

Child maltreatment

Regarding the issue of maltreatment, it is defined as a behavior toward a child that is outside the norms of conduct and entails substantial risk of causing physical or emotional harm and can be classified into four categories: physical, sexual or psycho-emotional abuse and neglect [22].

Congenital syphilis is a disease that causes skeletal changes in one fourth of the cases, that may mimic the lesions found in physical abuse cases, such as metaphyseal fractures and the vicious callus that results from the fracture consolidation [23,24]. The family anamnesis, a thorough physical examination and detection of a positive serology establish the differential diagnosis.

Another important aspect in assessment of children diagnosed with syphilis is represented by the distinction between the congenital form of the disease and an acquired infection, in which case the issue of sexual abuse can be raised. The diagnosis is made by correlating the anamnestic data with clinical findings and the results of serological tests.

Conclusions

Treponemal infection continues to be a topical epidemiological issue, therefore it is necessary to draw attention to this entity, particularly concerning the serious implications of the disease among children, including vital organ involvement, lethal intercurrent infections and the possibility to progress towards forms of late congenital syphilis in lack of an adequate treatment. Thus, the authors decided to report the present case, which has the distinctiveness of an abuse suspicion being raised. Upon hospitalization, the child's mother admitted to the detection of a positive serology during pregnancy and, by correlating the lesions found on physical examination with the infant's reactiv serological test, a presumptive diagnosis of congenital syphilis was established. The abuse issue came into question after the discovery of a positive serology in the child's uncle, given the fact that the mother confessed to a long absence from her daughter's life, leaving the infant in the care of her relatives. After a thorough anamnesis, associated with the clinical lesions that were incompatible with primary syphilis and in the lack of a forensic report or of any physical or paraclinical signs of abuse, the diagnosis was established – a compatible case of early congenital syphilis.

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