
Brief Reports

Late Onset Congenital Syphilis

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Congenital syphilis is transmitted transplacentally from an infected mother to her fetus; it is not spread by sexual transmission. In contrast, acquired syphilis in children is almost always sexually transmitted from abuse by an infected adult(1).

Case Report

An 11-year-old girl presented with progressively increasing swellings of the middle part of sternum, left shoulder and left orbit for the past 20 days associated with minimal local pain. There was restriction of movement of the left shoulder joint. There was no history of preceding trauma, fever, tuberculosis or

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contact with tuberculosis, weight loss, lymphadenopathy, similar swelling anywhere else or skin rash. There was a history of sexual abuse but the patient did not tell the time of insult or the person involved. The mother had not registered in any ante-natal clinic and did not have history of abortions or still births. She had no evidence of any sexually transmitted disease at the time of presentation. The child was of good birth-weight without any significant illness in the past.

On examination she was well nourished with height of 130 cm (10-25 percentile), weight of 29 kg (50 percentile) and head circumference was 51 cm. We noticed four swellings at the above mentioned sites, 3 x 3cm, firm in consistency, minimally tender, fixed to the underlying bone with increased warmth but no redness or discharge from the swellings. There was no involvement of any other bones or joints. Right eye examination revealed nebular opacity 5 x 3 mm in size (*Fig. 7*). Slit-lamp examination by an ophthalmologist showed right-sided interstitial keratitis, iridocyclitis, occlusio pupillae and posterior synechiae. The left eye was normal. There were no other stigmata of late congenital syphilis. Gynecological examination revealed old hymenal rupture without evidence of any genital lesion. There was no deafness.

X-rays of the left shoulder revealed evidence of chronic osteomyelitis (*Fig. 2*). Bilaterally ribs and mid-sternum showed osteolytic areas. Bone-scan confirmed affection of ribs bilaterally, shaft

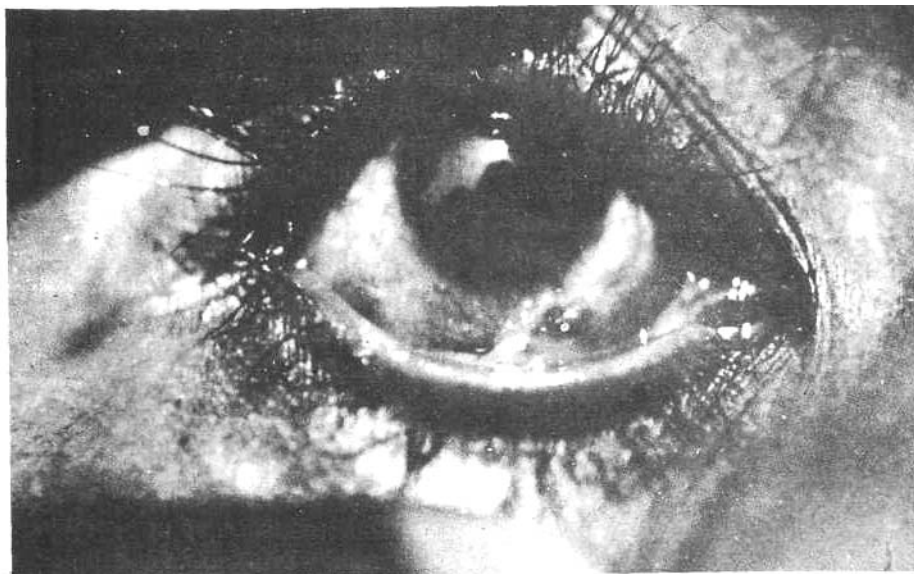


Fig. 1. Nebular opacity in right eye.

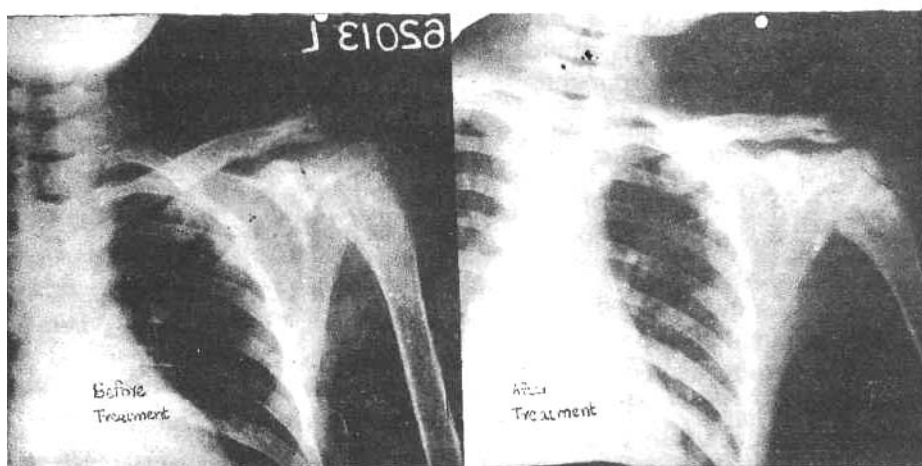


Fig. 2. Left clavicle and humerus showing evidence of osteomyelitis (before treatment) and improvement (after treatment).

of right humerus, and left shoulder joint indicating an active osseous pathology. X-ray chest did not reveal any evidence of tuberculosis. Mantoux test was negative.

The child's VDRL titer was significantly positive 1:64 (>1:8 significantly positive). TPHA (*Treponema pallidum* heme-agglutination) was also positive 1:160 (>1:80 significantly positive). CSF

examination was normal. CSF-VDRL was negative. Mother's VDRL titer was also significantly positive 1:64. The child's and mother's HIV were negative. The father had abandoned the mother and children. Consequently, the other sib did not report for investigations.

The child was treated with benzathine penicillin 2.4 mega units intramuscularly weekly for 3 weeks. Iridocyclitis was treated with local atropine and steroid drops. Pain and restriction of left shoulder joint gradually disappeared along with complete disappearance of the other mentioned swellings within 10 days of penicillin therapy. A repeat X-ray of the left shoulder joint done 2 weeks later showed healing (*Fig. 2*).

Discussion

Late congenital syphilis occurs from 2 to 30 years of age. The characteristics of late congenital syphilis include Hutchison's triad (Hutchison's teeth, interstitial keratitis, and eighth nerve deafness); bone changes such as frontal bossing, saddle nose, sabre tibia, perforation of the hard palate, Clutton's joint and rhagades(2). Most of the stigmata described in the literature may occur in other conditions and are etiologically indistinguishable. Some are associated with vitamin deficiencies and some have no pathological significance. Only clinically characteristic interstitial keratitis, mulberry molars, and upper central incisors of the second dentition as described by Hutchison may be classified as pathognomonic(3).

Interstitial keratitis is seen only in congenital syphilis and is not seen in acquired syphilis(4). Eye changes in

acquired syphilis include only uveitis and chorioretinitis. Interstitial keratitis usually starts in one eye but, irrespective of timely diagnosis and early antisyphilitic treatment, the other eye is likely to be involved after an interval which may vary from a few weeks to many years(2).

Mother also had VDRL titer significantly positive. Gummatous periosteitis which occurs in both congenital and acquired syphilis results in sclerosis and considerable new bone formation. Radiological changes in long bones are seldom pathognomonic(5). Late acquired syphilis occurs 3-10 years after primary stage and bony changes are seen 5 to 20 years after original infection(6). Our patient had history of sexual abuse with hymenal rupture as supportive evidence. In humans, treponemes inoculated at new sites during the incubation period of experimental infection, will multiply and produce lesions(6). After the primary stage has appeared, no further inoculation will produce tissue response (Colles' law)(2,6). Reinfection is possible if early and successful treatment of the first attack of the disease has been achieved(6). Since our patient was never treated for her congenitally acquired syphilis, the possibility of later reinfection due to sexual abuse causing bone lesions does not arise.

We conclude that this is a case of late onset congenital syphilis with interstitial keratitis and chronic osteomyelitis and history of sexual abuse as an incidental finding.

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48 XXXY Variant of Klinefelter Syndrome

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In 80% of the cases, the karyotype in Klinefelter syndrome is 47 XXY. In the

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remaining 20% chromosomal variants including mosaicism is seen. Among the variants, 48 XXXY is the commonest observed¹. The most significant effect of the additional X chromosomes on the phenotype is an increase in somatic malformations and mental retardation⁽²⁾. Dysmorphic features are very mild in 47 XXY, but are multiple and widespread in 49 XXXXY while 48 XXXY represents the intermediate level of the spectrum^(2,3). The abnormality is rare and features overlap with many chromosomal syndromes, notably Down's syndrome. This communication documents 48 XXY variant of Klinefelter syndrome.

Case Report

An 11 month old boy born of a non-consanguineous marriage was referred for delayed milestones and blindness. Paternal and maternal ages were 25 and 21 years, respectively. A detailed history including family history of infertility, mental handicap, past abortions and