Case Report

Infantile cortical hyperostosis - a report of Saudi family

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ABSTRACT

A 2-weeks-old Saudi neonate was apparently well till the 10th day of life when a swelling of the right groin was noted accompanied by irritability and fever, without history of trauma. On examination: the girl was irritable and febrile, the mass was firm, ill defined, fixed and tender. The state of the underlying skin was normal. There was family history of 3 siblings with similar swellings in the neonatal period and one of them had recurrence of the condition till the age of 7 year. The radiological findings indicated diaphysis hyperostosis, sparring of the epiphysis and the benign course of the disease. With exclusion of syphilis, osteomyelitis and trauma, the likely diagnosis would be infantile cortical hyperostosis. Such diagnosis should not be overlooked when faced by bony swellings in neonates.

Keywords:
Metabolic bone disease; Caffey disease; Infantile cortical hyperostosis; Newborn.

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INTRODUCTION

Infantile cortical hyperostosis (Caffey disease) is a benign multifocal proliferative bone disease with new bone formation with pronounced cortical thickening [1]. It is usually present in the first six months of life with fever, bony swellings due to cortical thickening, high erythrocyte sedimentation rate (ESR), thrombocytosis, and mild elevation of alkaline phosphatase. This condition is characterized by exacerbation and remissions till final recovery at later age in childhood [2]. The etiology of Caffey disease is unknown; however some authors linked it with Ebestian barr virus infection without proof [3]. IgM and C-reactive protein were elevated in some studies [4,5]. Hyperphosphatemia was reported in association with Caffey disease on rare occasions [6,7]. We report on Saudi neonate with the condition who had a groin swelling and a family history of 3 siblings with similar swellings in the neonatal period; one of them had recurrence of the condition till the age of 7 years.

CASE REPORT

A 2-weeks-old Saudi female presented with high-grade fever, painful swelling in the right clavicle and groin and inability to move the right limb. There was no history of trauma, or injection at these sites. She was born at term and admitted to the neonatal intensive care unit (NICU) because of meconium aspiration. She was discharged at the age of 5 days when she was feeding well. Her mother had regular antenatal care and the pregnancy was uneventful. Maternal VDRL, and hepatitis B and C screening were negative in the third trimester. The parents are consanguineous and they had seven children. There were three other siblings with history of recurrent jaw and clavicular swellings during early childhood, which resolved before the age of 7 years. Her examination revealed an active, alert, pink neonate who was febrile (temperature 39.8 ºC). She had right clavicular firm swelling, which was tender with no crepitus. The overlying skin was normal. The right hip was in fixed abduction with hard swelling over the right iliac bone measuring 5x5cm, which was tender and warm to touch. The rest of her examination was unremarkable.

Hematological investigations, including complete blood count (CBC), revealed leukocytosis, thrombocytosis, and elevated ESR (Hb 12.5 gm%, Hct 31.4%, WBC 21.3 x 103/cmm, platelets 1251x103/cmm, ESR 107 mm/hr). The biochemical profile was normal: (Ca 2.45 mmol/l, Phosphate 1.5 mmol/l, alkaline phosphatase 294 IU/L). C-reactive protein was negative; and CSF, urine, and blood cultures were sterile. TORCH screening and VDRL were negative. X-ray of the right clavicle (Figure1) and pelvis (Figure 2) showed thickening of the periostium and no signs of fractures.

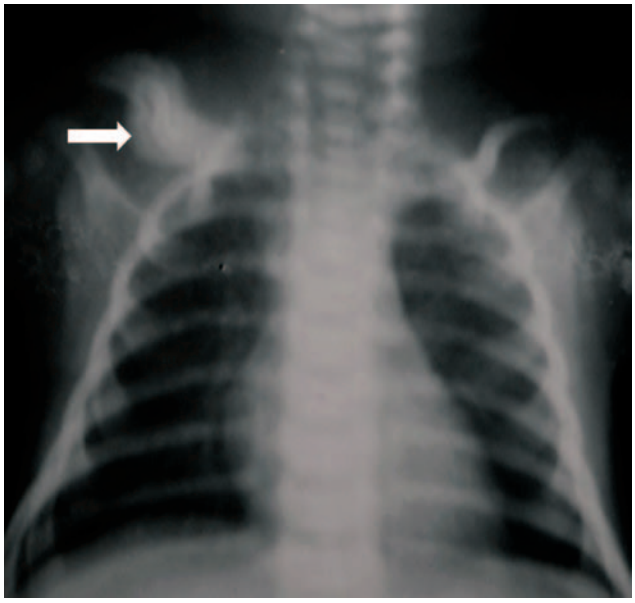

Figure 1 - Periosteal thickening of right clavicle (arrow).

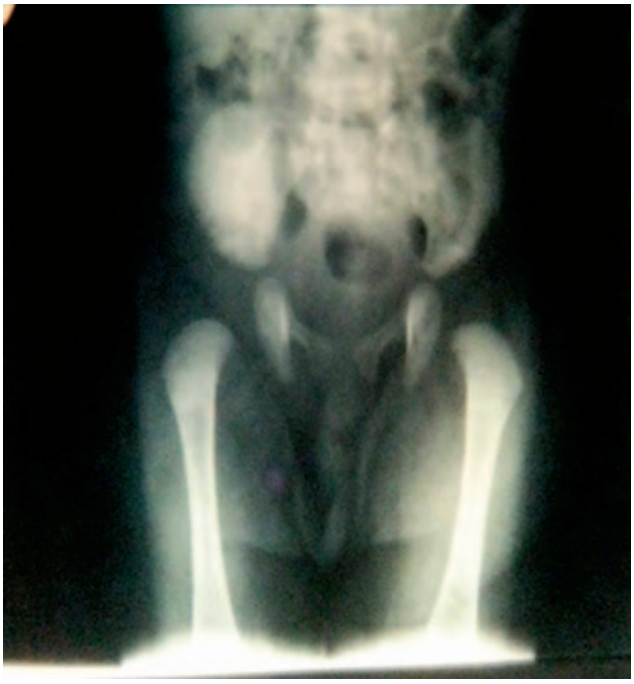

Figure 2 - Periosteal thickening of the whole right iliac bone with no signs of fracture.

An initial working diagnosis was osteomyelitis based on history of fever and bone swelling, but the general wellbeing of the patient as well as absence of early bone radiological signs made it unlikely. The possibility of trauma was unlikely because of negative history, presence of two sites of swellings and normal skin, and absent fractures. The diagnosis of congenital syphilis was excluded by the negative tests for the mother and the patient. Therefore, the diagnosis of infantile cortical hyperostosis was entertained based on the clinical presentation, hematological profile, classical radiological appearance and positive recurrent bone swellings in the family history.

The patient was started on prednisolone 1.5mg/kg/day for two weeks and the fever subsided after three days of the treatment. The pseudoparalysis resolved and the patient could move all his limbs freely. The swelling subsided gradually with radiological resolution by the age of 6 weeks. The platelates count returned to normal (223 x103/cmm) and ESR to 5 mm/hr. She presented again at the age of 14 weeks with similar swelling over the left iliac bone, which recovered spontaneously after two months.

DISCUSSION

Caffey disease is a metabolic bone disease characterized by periosteal reactive bone formation with resorption of the immediate underlying cortex, which tend to fleet from one bone to another [8]. The disease is either sporadic or familial. Inheritance appears to be autosomal dominant with variable penetrance. Some affected patients were found to be heterozygous for a missense mutation encoding the alpha-1 collagen type I gene (COL1A1) which manifests in children with collagen diseases.

Since Caffey and Silverman [3] described their first case, many authors reported clusters of cases, which affect 3:1000 children. There was unexplained sharp decline in reporting this disease since 1970s. The disease was reported in only few occasions from the Middle East including two Arabs from Palestine [8]. The recognition of Caffey disease is important as the differential diagnosis includes bone tumors, osteomyelitis, child physical abuse, vitamin C deficiency, prolonged prostaglandin E1 infusion and chronic vitaminosis A. Careful history and radiological appearance can differentiate these entities.

CONCLUSION

The natural history of Caffey disease is being self-limiting and of benign course, which usually settles after exacerbations and remissions without sequel. More reports on cases in Arab countries are needed to show if it has a different phenotype/genotype background.

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