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INFANTILE CORTICAL HYPEROSTOSIS OR CAFFEY'S DISEASE. A CASE REPORT AND REVIEW OF THE LITERATURE

Radiology

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ABSTRACT

Infantile Cortical Hyperostosis (ICH) or Caffey's disease is a self limiting benign bone disease affecting young infants under six months of age. It is characterized by acute inflammation of the periosteum along with swelling of the overlying soft tissue in a child who presents with irritability and fever. There are a wide range of diseases disease such as osteomyelitis, scurvy, bone tumors, hypervitaminosis A and child abuse which mimick ICH ,causing a delay in reaching the correct diagnosis. The emphasis here is to remind clinicians of the very existence of the disease in this era.

KEYWORDS:

Introduction

Infantile cortical hyperostosis (ICH) or Caffey's disease is a self-limiting benign bone disease which occurs in early infancy and is sometimes characterised by an unusual relapsing course. The disease frequently affects infants under six months of age, but it may occur in the prenatal period also.[1] In a child presenting with episodic painful swelling of flat bones and adjacent soft tissue ,it could often be thought of as the recurrence of ICH at an older age when the disease might not have been severe during infancy.[2,3] ICH has a multifaceted clinico-radiological presentation, with clinical symptoms including irritability and fever, acute inflammatory reaction of the periosteum, especially of flat bones with an associated involvement of adjacent soft tissue, and cortical thickening of the underlying bone.[4] Clinical diagnosis of this condition is not so modest because of a wide range of possible differential diagnoses mimicking ICH, such as osteomyelitis, hypervitaminosis A and C, scurvy, bone tumors, tuberculosis, congenital syphilis, prostaglandins E1 and E2 overdose and child abuse. Although, a highly precise clinical and radiological scenario may concur to reach the diagnosis of Caffey's disease without a need for any further investigation, its rarity precludes the establishment of the diagnosis. Here, we emphasize the clinical and radiological features of ICH to help clinicians in making an early diagnosis of the disease.

Case report

A 6 month male baby presented with irregular, low to moderate, on and off fever, excessive crying and swelling of face since one month, and irritability since one week. He had no evidence of bladder or bowel irregularities, skin rash, or respiratory discomfort. He was born by normal vaginal delivery at full term with a normal birth weight. He had no history of recent travel, consumption of a supplementary milk product, or any kind of social or medical problems in his family.Only medication that the child had received was some multivitamin syrup twice (**a day**). On general examination, he was anxious and irritable, with normal body morphology and nourishment. He was able to roll over, transfer objects and recognize strangers, with a Development Quotient of 100. The psychological assessment was within normal limits, and vaccine schedule was up to date. He weighed 7 kg, and his head circumference was 44cm. He was febrile with a temperature of 100.2°F, with normal other vital signs. There was no evidence of pallor, icterus, generalized lymphadenopathy, hepatosplenomegaly or any skin changes. He had swelling and redness over the face, more in its lower aspect, with woody induration and raised local temperature on palpation. He was started on empirical intravenous antibiotics and primary supportive treatment in the form of intravenous fluids with antipyretics. Rest of the systemic examinations revealed no abnormality.

His clinical condition improved with the treatment given, and he remained afebrile with an improvement in oral intake. Biochemical investigations revealed normocytic normochromic anemia (Hb 9.6g/dL), granulocytic leucocytosis (WBC 21,100/µL), normal platelet count (598000/µL), raised CRP (42.7mg/L, normal limits 0-6mg/L), and ESR 80mm/Hr (normal 0-20). Peripheral smear showed normal cellular components. Liver and renal function tests, serum amylase, and serum sodium, potassium, calcium, and phosphate were within normal limits, except a raised serum alkaline phosphate (240 IU/mL). Urine microscopy did not reveal any abnormalities, and bacterial culture of urine and blood was sterile. Radiograph of mandible PA-view showed sclerotic changes in both sides of the mandible with periosteal reaction and adjacent soft tissue swelling. There was no evidence of osteolytic lesions. Chest and pelvic radiographs showed normal bones. On ultrasonography of abdomen and pelvis, no abnormality was seen. After an initial symptomatic improvement, an MRI of face was done, which showed marked cortical thickening with a lamellated periosteal reaction involving the whole length of the mandible. [Image I]Also a STIR hyperintense signal was seen within the expanded mandible with a significant post-contrast enhancement of the cortex.[Image II] In the absence of any other systemic or local disease, a diagnosis of infantile cortical hyperostosis or Caffey's disease was made based on the clinical and radiological features. He was managed conservatively and improved symptomatically and biochemically within a period of 3 weeks. He was discharged in a good general condition, with an adequate tolerance for oral intake. At two weeks post-discharge, he had progressive symptomatic improvement, with a partial reduction in mandibular swelling and no local tenderness.

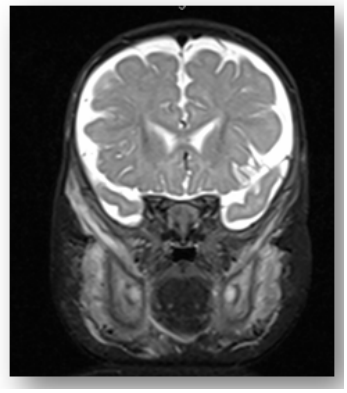

Image 1

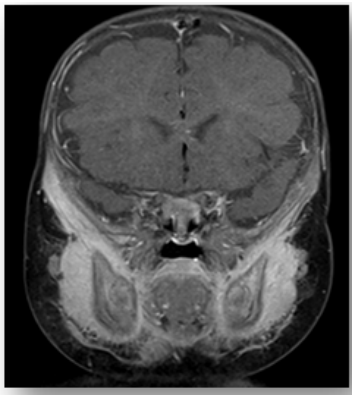

Image II

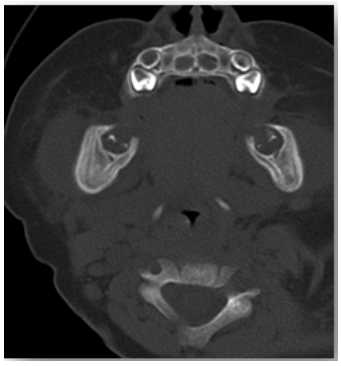

Image III

Discussion

Caffey and Silverman described this idiopathic benign bone disease of infants in 1945.[5] It was defined as a triad of symptoms, clinical signs, and radiological features, such as excessive irritability and fever, unexplained soft tissue swelling adjacent to flat bones, and underlying cortical bone thickening.[5-7] Till now, less than 500 cases of Caffey's disease have been reported in the English literature. The incidence of sporadic cases of disease is also decreasing over time.[1,3,6,7]. Prenatal onset of this disease had been reported to be in less than 50 cases in the literature.[8]

The ultimate aetiology of this disease is not well known.[10] Among the proposed aetiological factors; infection, autoimmune disease and genetic disorders have variously been described.[7,9,10] ICH is described as a type of collagenopathy with commonly an autosomal dominant pattern of inheritance involving alterations in the COL1A1 (17q21) gene, which encodes Alfa-1 chain of type-1 collagen.[11] The majority of the cases are sporadic, with only a few familial cases reported with an autosomal dominant and recessive inheritance. [1,7,10] The prolonged exposure to prostaglandin E1 in infants with cyanotic heart disease, to maintain ductal patency, has also been reported as an aetiology of ICH.[12,13]

The occcurence of this disease with respect to race and gender, and the variability of its incidence due to an impact of environmental factors are not well studied.[1,2,7] The ultimate diagnosis is based on the exclusion of common differential clinical conditions. Normal serum amylase and ultrasonography help to rule out parotitis. In a thriving baby with low-grade fever and the absence of systemic toxicity, it is dubious to go with a diagnosis of septic or aseptic osteomyelitis. Any primary or secondary bone tumour is not known to cause diffuse involvement with sclerosis of mandible. Although, rarity of Caffey's disease precludes its diagnosis, the clinical presentation of an infant with raised ESR, and systemic features as irritability and low-grade fever with diffuse sclerosis of mandible made it possible to diagnose the disease in our case.

Infantile cortical sclerosis,the Caffey's disease occurs usually in flat bones, and most commonly includes mandible followed by scapula, clavicle, ribs, and long bones.[1,5,9,14-16] Several associated complications have been observed in reported cases such as facial nerve palsy, dysphagia, nasal obstruction, and proptosis.[16-19]

The usual biochemical abnormalities in an infant with ICH are elevated ESR, anemia, thrombocytosis, raised alkaline phosphate and serum immunoglobulin levels.[20] Our case had similar findings, except for the serum immunoglobulin level which was not performed. The peculiar feature on radiograph such as cortical new bone formation underneath the soft tissue swelling is the most valuable clue to the diagnosis of ICH in the absence of disease-specific laboratory tests.[6,12,21]

Infantile form of the disease is less severe than the prenatal-onset form, and usually resolves within months with conservative treatment, without leaving any residual deformity. For symptom relief, Naproxen or Indomethacin can be used, with steroids reserved for non-responders. The baby recovered symptomatically well, with general conservative treatment. However it is said that the bone lesions may recur after a variable period of time in same or different bone, with an unpredictable severity of disease.[11]

Conclusion

Caffey's disease is a rare infantile disease with a self-limiting but relapsing course; recovery occurs well with an empirical treatment without a need for any intervention. Clinicians need to be aware of the clinical and radiological features of the disease in reaching a proper diagnosis and also avoid unnecessary investigation. Thus, to confirm the diagnosis, a well-performed clinical examination with basic biochemical tests & radiological evaluation suffice.

Authors contribution

NS, KS, KS, AA participated in acquisition of data, conception and design of manuscript. All authors participated for literature search and approved the final manuscript for the publication.

Conflict of interest

All authors declare that they don't have any conflict of interest.

Acknowledgment

We acknowledged to patient for consent to publish images.

Figure legends

Image I Coronal T2W MR image shows marked marrow odema involving the body of the mandible with marked periosteal reaction and associated soft tissue swelling.

Image II Post contrast coronal T1W fatsat MR image depicts thickened cortex of the mandible shows marked enhancement

Image III Axial plain CT at the level of alveolar arch show diffuse lamellated periosteal reaction with thickened overlying cortex involving body of mandible

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