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Case Report

Caffey's Disease: A Case Report

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Abstract

Infantile cortical hyperostosis (Caffey disease) is characterized by spontaneous episodes of subperiosteal new bone formation along 1 or more bones commencing within the first 5 months of life. It is featured by acute inflammation of periosteum and overlying soft tissue which causes fever. Sometimes it can present as fever without focus. Generallyit is treated by sole acetaminophen having a good outcome. An 8 month old boy came with multiple tender non-suppurative swelling over various areas of face and limb with mild fever for prolong duration. The lesions were diagnosed to be infantile cortical hyperostosis by radiological profile showing new bone formation, periosteal reaction, sclerotic changes without osteolytic appearance after ruling out common infective causes like epidemic parotitis. He was treated by oral acetaminophen syrup which proved to be adequately effective on follow up after 4 wks.

Keywords: Caffey's disease, cortical hyperostosis, inflammation, periosteum.

1. Introduction

Caffey's disease or infantile cortical hyperostosis (ICH) is a rare self-limiting condition which usually affects the young infants [1]. It (Caffey disease; OMIM 114000) is a genetic disorder characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. It was first named and recognized as a separate entity in 1945. There is increased bone formation; laboratory findings include an elevated level of alkaline phosphatase. There may be an elevation in white blood cell count (WBC) and erythrocyte sedimentation rate (ESR), indicating an inflammatory response. The bone change begins before 5 months of age and resolves before 2 years of age [1]. Recurrence of ICH is uncommon, incidence of adverse outcome of the hyperostotic lesions in affected individuals are very less [3]. It can be inherited in an autosomal dominant manner where as more severe and often lethal prenatal form of Caffey disease appears to be inherited as an autosomal recessive disorder [1]. Affected individuals have shown link in the α1(I) chain of type I collagen (*COL1A1*)[4]. Trials of antiinflammatory agents like paracetamol, indomethacin and with glucocorticoid have shown remarkable improvement. The diagnosis is done by ruling out other causes like osteomyelitis, hypervitaminosis A, scurvy, bone tumors and child abuse. While there is no particular laboratory investigations to confirm this disease except increased alkaline phosphatase which is highly nonspecific, a high index of suspicion in a typical clinical setting can avoid protracted investigations for this otherwise self-limiting illness.

2. Case Report

An 8 month old boy was admitted to Pediatric Department of MKCG Medical College, Berhampur with one month history of low grade intermittent fever and irritability which was followed by appearance of tender, non-suppurative swelling over right side of the jaw [Figure 1]. The skin over the jaw was normal and there was no discharging sinus or localized lymphadenopathy. It was followed by swelling over opposite jaw and right elbow [Figure 2]. Other systems were absolutely normal. He was fully immunized as per NIS. His birth history was uneventful and motor milestones being normal. The vitals were normal. The swellings were firm in consistency, tender..

Lab investigation revealed normal hematological features except Hb to be 8.8gm/dl and ESR 130mm/hr. Peripheral smear showed microcytic hypochromic anaemia with thrombocytosis. Serum electrolytes and urea creatinine, liver function tests were within normal limit except serum alkaline phosphatase, which was measured to be 1140 IU/dl. Blood culture revealed no growth and maternal serology for

syphilis was nonreactive. The serum amylase was normal which ruled out incidence of epidemic parotitis. X-Ray showed right mandible and right hand forming new bone, periosteal reaction over radius and ulna [Figure 3], inhomogeneous calcification over lower end of radius and ulna, absence of osteolytic lesions. It was diagnosed to be IHC. It was treated by paracetamol only as antinflamatory medication. After 4 wksof treatment child was symptomatically normal.

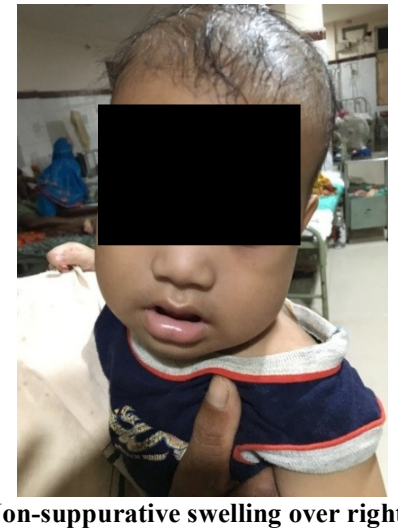

Figure 1: Non-suppurative swelling over right side of the jaw

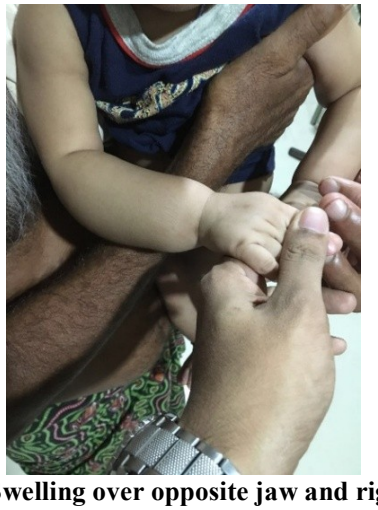

Figure 2: Swelling over opposite jaw and right elbow

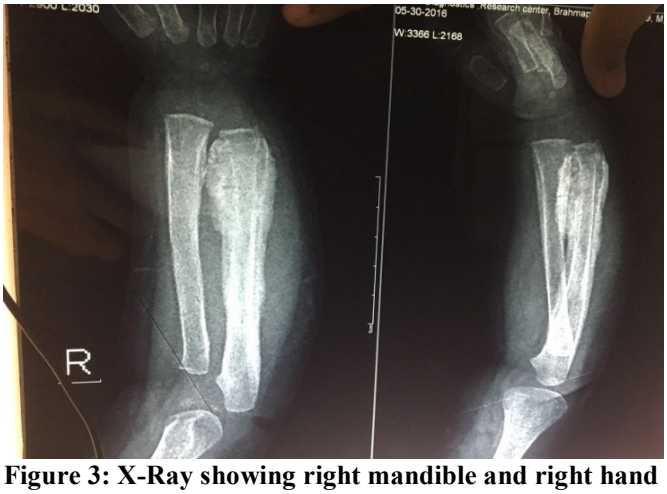

Figure 3: X-Ray showing right mandible and right hand forming new bone, periosteal reaction over radius and ulna

3. Discussion

Infantile Cortical Hyperostosis is a self-limiting disorder pictured by a triad of symptoms (irritability and fever), soft tissue swelling and underlying cortical bone thickening. The exact etiology of this condition is still unknown [5]. Most cases are sporadic, but a few familial cases with autosomal dominant and recessive patterns have been described. Among the proposed causes are infections, immunological defects and genetic abnormalities [5]. Two forms of Caffey disease has been described in literature, a classical mild infantile form (ICH) delineated by Caffey and Silverman and a severe form with prenatal onset. The incidence of the disease is unknown. A total of 44 cases have been reported with the severe prenatal onset of Cortical Hyperostosis [6]. The classic form has an onset within the first 6 months of life with irritability, painful swelling of the overlying soft tissue that precedes the cortical thickening of the underlying bones, fever and anorexia (like our case). The swelling is painful with a wood like in duration but with no redness or warmth, thus suppuration is absent. Mandible is the most commonly involved site followed by scapula, clavicle, ribs and long bones. Otherwise children are asymptomatic [5,6]. Isolated cases of facial nerve palsy and Erb's palsy have been reported by some authors [7]. The pain ranges from mild to severe along with other features like dysphagia and proptosis.[8] These features are not found in our patient. Laboratory findings include elevated inflammatory markers, thrombocytosis and anemia (all are seen in our patient). Sometimes alkaline phosphatase is widely elevated, like our case. The severe prenatal onset form is generally autosomal recessive characterized by extensive hyperostotic bone involvement, angulations and shortness of long bones, as well as polyhdramnios and fetal hydrops. In these conditions possibility of osteogenesis imperfecta should be ruled out [8].

Radiography is the most valuable diagnostic study in ICH. Cortical new bone formation (Cortical Hyperostosis) beneath the regions of soft tissue swelling is the characteristic feature. X-ray shows periosteal hyperostosis confined to diaphysis of long bones. Bone scan shows increased uptake of radioisotope in many areas. This is a very useful test which may reveal increased uptake of radioisotope in involved areas before radiographic changes are present. Highly characteristic scintigraphic image, when the mandible is involved, can play the most important role in diagnosis and its recognition can also spare many unnecessary investigations.MRI is also very useful in the diagnosis. No laboratory tests are specific for diagnosis of ICH. Awareness of the existence of this rare condition and its typical clinicoradiological profile will avoid the patient being subjected to many unnecessary investigations.

Caffey disease is mostly self-limiting and resolves within six months to one year and may not need any treatment. Paracetamol, Indomethacin or Naproxen could be

used in really symptomatic cases. Steroids can be administered if there is poor response to Indomethacin. In this case, paracetamol was used and the outcome appeared to be satisfactory. In some cases, the bone lesions can recur suddenly at their original sites or at newer sites and can have an unpredictable clinical course with remissions and relapses. Hence, relapse can happen several years later [10].

4. Conclusion

It is a self-limiting disease of children. This report can help the pediatricians and orthopedicians a better view on the condition which will reduce unnecessary head banging in several investigations and will simultaneously decrease heavy stress in parents. Simple treatment by paracetamol will save the child from unnecessary toxicity of many drugs.

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