

Caffey Disease or Infantile Cortical Hyperostosis: A Case Report

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Abstract: Infantile cortical hyperosteosis, also known as Caffey's disease, is a rare, idiopathic mostly self limiting condition affecting infants under 6 months of age. It is characterized by acute inflammation of the periostium and the overlying soft tissue and is accompanied by systemic changes of irritability and fever. Diagnosis may be delayed as this disorder mimics a wide range of diseases including osteomyelitis, hypervitaminosis A, scurvy, bone tumors and child abuse.

I. Introduction

Caffey disease or Infantile Cortical Hyperostosis (ICH) is a rare and mostly self limiting condition affecting young infants. It is characterized by acute inflammation of the periostium and the overlying soft tissue and is accompanied by systemic changes of irritability and fever. Diagnosis may be delayed as this disorder mimics a wide range of diseases including osteomyelitis, hypervitaminosis A, scurvy, bone tumors and child abuse. While there are no laboratory tests to confirm the diagnosis of Caffey disease, a high index of suspicion in a typical clinical setting can avoid protracted investigations for this otherwise self-limiting illness.

II. Case Report

A 3 day old girl child was admitted to MKCG Medical College and Hospital, Berhampur, with intermittent crying and irritability complains of bilateral angulation of both lowerlimb. There were no accompanying respiratory, gastrointestinal or urinary symptoms. she was born normally at term. The birth weight was 3 kg . her immunization status was up to date. she was breast fed and was not receiving any supplements including vitamin syrup or fish oil. There was no history of recent travel or of being given unpasteurized milk and there was no major medical or social problems in her family. On examination, the baby was alert and interestingly, had an "anxious look". she was looked well nourished. She weighed 3 kg .vital signs were normal. There were no pallor, icterus, lymph node enlargement, hepatosplenomegaly or rashes. She had mild swelling on the both the tibia . The overlying skin was normal and the swellings were firm in consistency but were mildly tender. The remaining systemic examinations were normal. Over the next few days, the swelling became more prominent. The patient remained afebrile after hospitalization. Investigations showed microcytic hypochromic anaemia, thrombocytosis and neutrophil count at the upper end of the normal range. Hb was 9.3g/dl. A peripheral blood smear showed hypochromic red cells and thrombocytosis. Sickling was negative. Hence, no other abnormalities were noted. Blood culture revealed no growth and Maternal Serology for Syphilis (VDRL) was non reactive. X-rays of Bones showed forming new bone, periosteal reaction and sclerotic changes. However, there were no osteolytic lesions. Urine routine examination was normal. Her mother had a history of similar disorder or condition in the past when she was born (21 yrs back), with available radiology and treatment records. At that time she was treated with steroid .

The patient was a neonate with irritability and who on examination had multiple bony swellings which were slightly tender. Hypervitaminosis was ruled out because she was getting only infant formula and there was no excessive ingestion of the vitamin in any form. A bone infection was thought to be unlikely because of the lack of "toxicity" with normal vital signs other than a low grade temperature in a "thriving baby" despite multiple sites of involvement. The absence of significant bone tenderness and the radiological picture were against a diagnosis of multifocal osteomyelitis. Congenital Syphilis was very unlikely with a negative serology for syphilis in the mother. A bone tumour again was felt to be very unlikely because of the multiple sites of involvement (primary bone tumours are usually single) and the absence of lytic lesions on X-rays seen in most secondary bone tumours. Scurvy usually does not affect such small infants. The patient had an elevated ESR, persistently high platelet count and the x-rays showed increased bone formation and sclerosis but no lytic lesions. A diagnosis of Caffey disease, although rare, was made in this small infant based on the clinical profile and investigations.



Figure 1 x - ray of 3 day old child and clinical picture showing hyperostosis of bilateral tibia

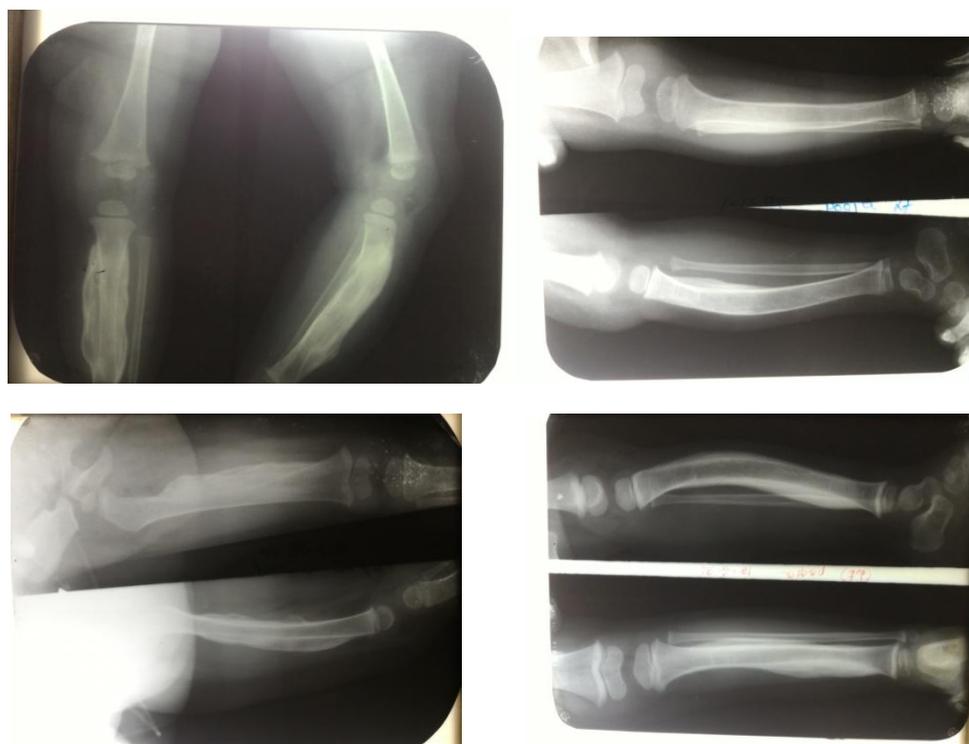


Figure 2 x ray showing hyperostosis of bilateral tibia and femur 21 years back of the mother

The patient was commenced on Ibuprofen at a dose of 50mg three times a day, and the dose was reduced to twice daily after 3 weeks and was discontinued at week 6 and treated with steroid. The child became asymptomatic with the tibia swellings significantly reduced in 6 weeks

III. Discussion

Caffey disease, also known as Infantile Cortical Hyperostosis is a self limiting disorder. It is characterized by a triad of systemic symptoms (irritability and fever), soft tissue swelling and underlying cortical bone thickening. It was first reported as a disease entity by Caffey and Silverman in 1945. The exact aetiology of this condition is still unknown. 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. The discovery of a gene locus in 3 unrelated families with autosomal dominant inheritance (gene COL1A1, 17q21) which encodes Alfa-1 chain of Type I collagen, has raised some doubts whether some cases are a type of Collagenopathy, like Osteogenesis imperfecta. Similar conditions have also been reported following prolonged treatment with Prostaglandin E1 for maintaining ductal patency in infants with cyanotic heart disease. The existence of two forms of Caffey disease has been suggested, a classical mild infantile form (ICH) delineated by Caffey and Silverman and a severe form with prenatal onset. The condition has been described as rare with no sex or racial predilection. The incidence of the disease appears to fluctuate and other environmental effects may exert an influence. Although the exact incidence of the more common classic form of ICH is unknown, there is a world wide decrease in the number of cases particularly the non-familial form. The classic form has an onset within the first 6 months of life. The manifestations include irritability, swelling of the overlying soft tissue that precedes the cortical thickening of the underlying bones, fever and anorexia. The swelling is painful with a wood like induration 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. There are usually no other signs and symptoms. Isolated cases of facial nerve palsy and Erb's palsy have been reported in the literature. The pain can be severe and can also result in pseudo paralysis. Other rare clinical findings include dysphagia, nasal obstruction and proptosis. The study patient had none of these uncommon features. Laboratory findings include elevated ESR, and in some patients high alkaline phosphatase, thrombocytosis, anaemia and raised immunoglobulin levels. The studied patient had elevated ESR, thrombocytosis and anaemia but the alkaline phosphatase was not elevated. Serum immunoglobulin test was not performed. The severe prenatal onset form is characterized by extensive hyperostotic bone involvement, angulations and shortness of long bones, as well as polyhydramnios and fetal hydrops, which may lead to the incorrect diagnosis of lethal form of Osteogenesis imperfecta. But absence of other signs like a blue sclera, delicate skin and total absence of fractures and typical histopathological features differentiate this condition from Osteogenesis imperfecta. Radiography is the most valuable diagnostic study in ICH. Cortical new bone formation (Cortical Hyperostosis) beneath the regions of soft tissue swelling, which is the characteristic feature. While no laboratory tests are specific for diagnosis of ICH, the important differential diagnosis that are to be excluded are osteomyelitis, chronic hypervitaminosis A, bone tumour, scurvy, child abuse and prolonged PGE1 infusion. Awareness of the existence of this rare condition and its typical clinicoradiological profile will avoid the patient being subjected to unnecessary investigation. Caffey disease is mostly self-limiting and resolves within six months to one year and may not need any treatment. However, Indomethacin or Naproxen could be used in really symptomatic cases. Steroids can be administered if there is poor response to Indomethacin. In this case, Ibuprofen followed by steroid 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.

IV. Conclusion

The aim of this short report is to highlight this disease entity to avoid unnecessary and invasive investigations. The diagnosis of this disease needs an awareness of this condition along with a high index of suspicion. A good history, clinical examination, basic laboratory studies, and plain radiographs are sufficient enough to confirm a diagnosis of this entity in most cases.

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