Caffey's Disease in an Infant
Afsheen Batool Raza, Iftikhar Ijaz, Farrah Naz and Taeed Ahmed Butt

INTRODUCTION
Infantile cortical hyperostosis, also known as Caffey's disease, was first reported in 1945 by Caffey and Silverman.1 It is an episode of massive subperiosteal new bone formation typically involves the diaphysis of the long bones, mandible and clavicles.2,3 It is characterized by irritability, fever, bone pain, pseudoparalysis of the affected limbs and soft tissue swellings.4,5 The disease may be present at birth or occur shortly thereafter. All races are affected with equal ratio in males and females. The cause is unknown. Familial incidence is compatible with the dominant inheritance pattern with incomplete penetrance. Both familial and sporadic forms occur.

We report here an infant boy, 4 months of age, with similar presentation.

CASE REPORT
An infant boy aged 4 months was brought to the medical outpatient department of Children's Hospital Lahore by his mother with a 10 days history of fever, irritability and swelling of forearms and right cheek. There was no history of trauma, joint swellings, pallor or bleeding from any site. Also no history of loose motion, vomiting, skin rash or any urinary complaints. He was the first child born full term to the consanguineous parents with no history of birth trauma and his past medical and family history were insignificant. He was developmentally a normal child. During the course of illness, initially he had tender swelling of the right forearm with accompanying symptoms of irritability and fever upto 101°F. Parents sought advice from a local doctor where a diagnosis of osteomyelitis was made and treated with intravenous antibiotics without improvement. Four days later, he was found to have similar swelling of the left forearm as well. His blood counts revealed neutrophilic leukocytosis (Total leukocyte count of 15,300* 10^9/L and polymorphonuclear cells of 70%) with ESR 35 mm at the end of 1 hour. His serum calcium and phosphate was normal but alkaline phosphatase was high (870 IU/L mg/dl).

After completing 7 days of parenteral antibiotics, he was referred to us. During evaluation, the child was alert and playful. He was febrile (101°F) with heart rate of 120 beats per minute, blood pressure of 90/60 mmHg and respiratory rate of 48 breaths per minute. He weighed 7.5 kg. The local examination revealed swelling of both forearms with wood-like induration and tenderness but no redness, warmth and joint swelling (Figure 1). There was normal movement at joints. His systemic examination revealed no rash, lymphadenopathy or any other positive findings. Based on history and examination findings, a provisional diagnosis of Caffey's disease was made.

Child was treated with syrup ibuprofen and indomethacin. There was remarkable improvement in the irritability, tenderness and swelling over 10 days and child was discharged.
Caffey's disease is a rare condition presents in infants, most commonly in early infancy under 5 months of age. Gene map locus 17q 21.31-922 (COL 1A1,120150). It has two forms. The prenatal is rare and occurs as early as 24th week of intrauterine life. It is severe with marked angulation of the long bones. The postnatal form is common and usually occurs before 5 months of life. Proposed mechanism includes hypoxia due to inherited defects of the arterioles of the periosteum resulting in hypoxic damage, which acts as an exciting event for periosteal reaction. Theories describing allergy and infective aetiologies have also been postulated. Whatever the aetiology, initial event is inflammation of periosteum and surrounding soft tissue followed by subperiosteal bone formation. Original cortical bone is replaced with new lamellar immature bone followed by final phase of resolution.

Blood tests may show anaemia, moderate leukocytosis, thrombocytosis, increased ESR, CRP and elevated alkaline phosphatase. One study showed thrombocytosis, raised Ig M and vitamin E deficiency in patient with Caffey's disease. X-ray shows periosteal hyperostosis confined to diaphysis of long bones. This patient's bone scan showed 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 of the Caffey's disease as its findings precede the characteristic findings of plain radiography. There is no specific treatment. NSAID especially Indomethacin and ibuprofen are effective. Steroids can be used for extensive disease, recurrences may be experienced for months and years. Indomethacin has been used to control flare-ups. As a result of extensive periosteal reaction, patient may develop facial asymmetry, exophthalmos. Orthopedic intervention like osteotomies may be required in complications.

The clinical course is variable and unpredictable but usually the acute symptoms resolve over the course of a few months and the outcome is good with spontaneous resolution. Sometimes, relapses may occur several years later. Closer differential diagnosis includes osteomyelitis, which usually affects one bone at a time and has bone destruction and sclerotic bony changes. Possibility of trauma, which resemble Caffey's disease, can be differentiated by the presence of fracture line, irregular metaphyseal, subperiosteal hemorrhages, bruises and head injury. Scurvy is differentiated by subperiosteal hemorrhages, irregular metaphyseal, pencil thin cortex, marked osteopenia and decreased alkaline phosphatase. Moreover, its presentation before 4 months of age is rare. Other possible differentials include leukemia, osteogenesis imperfecta, bone tumors, hyperphosphatasia and syphilis.

REFERENCES


