

Caffey Disease Presenting as Unilateral Scapular Swelling

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ABSTRACT

Infantile cortical hyperostosis (Caffey disease) is a rare entity characterized by a triad of bone changes, soft tissue swelling and irritability. It usually presents in infants within first five months of their life with no specific gender or racial predilection. The entity is self-limited in majority of the cases and the treatment is symptomatic. It is primarily diagnosed on the basis of characteristic radiographic features and clinical data. Thus, it is important to keep the differential of Caffey disease in mind when an infant presents with the characteristic triad to prevent further unnecessary investigations.

Keywords: Caffey disease, infants, scapular swelling, self-limited, rare entity.

INTRODUCTION

Caffey disease was first described by John Caffey and WA Silverman in 1945.^[1] It presents in infants (usually in first five months) with a triad of bone changes, soft tissue swelling and irritability. It most commonly affects the flat bones, mandibular involvement being the most common (75-80% of cases), followed by clavicles, scapula, lateral aspect of ribs, calvaria and ilia. Among the long bones, ulna is the most commonly affected.^[2,3] The diagnosis is based on characteristic radiographic features. Laboratory investigations show raised ESR. It is mostly a self-limited condition.

CASE HISTORY

A five weeks old boy was admitted with chief complaints of swelling over the right scapula. There was a two weeks history of intermittent low grade fever and excessive crying. He was born by uncomplicated normal vaginal delivery, with no instrumentation being used. His birth weight was 2.9 kg. He was exclusively breast fed. No other previous medical

history was present. No major illness was present in the family. On examination, he was irritable and febrile. The respiratory rate, blood pressure and pulse rate were within normal limits. There was no pallor, icterus, cyanosis, clubbing or lymphadenopathy. No hepatosplenomegaly or rashes were seen. There was a firm swelling over the right scapula which was slightly tender. The swelling was not warm on touch and no significant erythema was noted on the overlying skin. Fullness over the mandible was also seen on examination which was previously unnoticed by the mother.

The patient was referred to our centre for further investigations. X ray right shoulder AP view was done which revealed lamellated periosteal reaction involving the right scapula with increased cortical thickness, subperiosteal new bone formation and increased soft tissue shadow in the affected region. [Figure. 1 shows X ray Right shoulder AP view showing lamellated and fusiform periosteal reaction, cortical thickening, subperiosteal new bone formation and overlying soft tissue swelling

over the right scapula. Thickening of the mandible is also seen].

A non contrast CT scan was done on 128 slice Somatom (Siemens) CT scanner with dose adjustment for paediatric age group, soft tissue and bone window reconstructions were made of the mandible and scapula region, which again showed dense cortical hyperostosis, subperiosteal new bone formation, lamellated periosteal reaction and increased soft tissue bulk in the affected regions. [Figure 2a-f show Low dose non contrast CT axial sections in bone window setting showing increased cortical thickness of mandible and right scapula. Lamellated periosteal reaction, subperiosteal new bone formation, increased soft tissue bulk and extraosseous bony masses are seen in right scapular region].



Figure 1: X ray Right shoulder AP view.

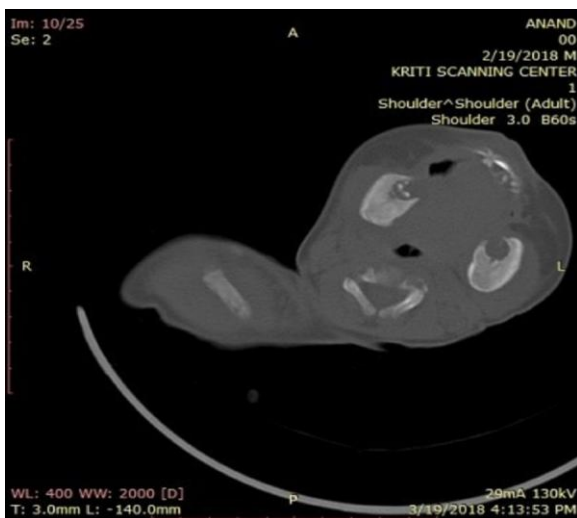


Figure 2a

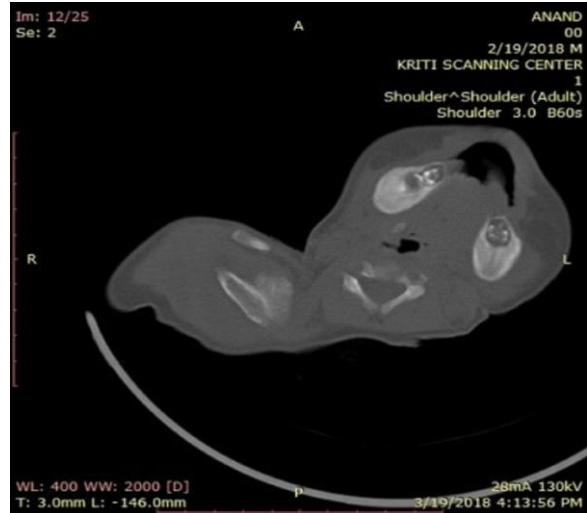


Figure 2b



Figure 2c



Figure 2d



Figure 2e



Figure 2f

Laboratory investigations revealed increased ESR.

Based on history, classical imaging features and laboratory investigations a diagnosis of Caffey disease was made. Other differentials which need to be excluded are

- Osteomyelitis: ruled out due to absence of toxic signs in the child, lack of local signs over the swelling, no subperiosteal/intramuscular collection, no lytic areas on radiograph and CT.
- Trauma/ Non Accidental injury: ruled out due to absence of fracture or focal callus formation.
- Myositis ossificans: ruled out by absence of history of significant trauma and lack of typical

peripheral calcification in increased muscle bulk.

- Sclerotic skeletal dysplasias: ruled out by lack of familial history, unilateral focal asymmetric involvement, onset and course of development.
- Hypervitaminosis A: ruled out as child was exclusively breast fed with no added supplementation.
- Scurvy: usually does not present before six months of age.
- Neoplastic (Ewing sarcoma/metastatic neuroblastoma): multifocal involvement and age unusual for Ewing sarcoma, lack of spiculated (sunburst pattern) periosteal reaction as seen in metastatic neuroblastoma.

The child was started on Ibuprofen 50mg, three times a day for three weeks and reduced to two times a day till six weeks. Significant improvement was noted after two weeks.

DISCUSSION

Caffey disease is a rare self-limiting entity presenting in first five months of life without any gender or racial predilection.^[4] The exact etiology is still unknown. It has both familial and sporadic forms. Most of the cases are sporadic. Recent studies suggest that it is caused by a mutation in the COL1A1 gene on chromosome 17q21 which encodes for Alfa-1 chain of type I collagen. It is inherited in autosomal dominant pattern with incomplete penetrance.^[5] Another proposed mechanism is a disturbance in endogenous prostaglandin regulation, as similar features have been reported in infants who receive prostaglandin therapy for maintaining ductal patency in congenital cyanotic cardiac conditions.^[6]

A rare variant of the disease is prenatal onset infantile cortical hyperostosis, which is a lethal form resulting in early death of the newborn postnatally.^[7] It is known to have a more

severe course if the onset starts before 35 weeks of gestation. It has both sporadic and inherited forms. The inherited form is known to have an autosomal recessive pattern of inheritance. It can be diagnosed on antenatal ultrasonography. It shows short angulated bones and irregular diaphysis. Other associated features include hydrops fetalis, polyhydramnios, pulmonary hypoplasia and prematurity.^[8,9] Other differentials for short angulated bones include Osteogenesis imperfecta, camptomelic dysplasia and hypophosphatasia. However, the absence of fractures, blue sclera, delicate skin in prenatal Caffey disease helps exclude other entities.

The course of the postnatal disease has been classified into three phases: acute, subacute and chronic. The acute phase is characterized by periostitis and adjacent soft tissue inflammation. In the subacute phase there is resolution of inflammation, periosteal thickening is seen which results in ossifying periostitis, lamellated new bone formation and osseous deposition may occur in adjacent soft tissues. In late phase there is resolution. There is removal of peripheral bone starting from inner to outer surface. Caffey disease is mostly self-resolving and complete resolution is seen up to 1 year of age. Symptomatic treatment by NSAIDs is given, sometimes steroids are added in refractory cases. Some long term sequelae may be seen in the form of mandibular asymmetry, persistent synostosis of the affected bones, bowing of long bones, limb

length discrepancy. In some cases relapse at same or different sites has been documented in later age.^[4]

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