Case Study

Caffey disease with acute sialadenitis: A diagnostic challenge!

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Abstract:

Background: Caffey disease or Infantile Cortical Hyperostosis (ICH) is a rare and self-limiting condition affecting young infants. It is characterized by acute inflammation of the periosteum and the overlying soft tissue and is accompanied by systemic changes of irritability and fever. We report a unique case of Caffey disease with acute sialadenitis which led to a significant diagnostic challenge.

Aim: The aim of the case report is to highlight this self-limiting disease entity to avoid unnecessary and invasive investigations.

Case description: We report a one and a half month old male child, with bilateral neck swelling with fever who had features suggestive of acute sialadenitis. He was given an adequate antibiotic course for 14 days but continued to have a significant underlying jaw swelling with high counts, inflammatory markers and alkaline phosphatase which led to a suspicion of Caffey disease. X-ray of mandible confirmed the diagnosis. Child was given oral ibuprofen and discharged with significant improvement

Conclusion: The diagnosis of this disease needs an awareness of this condition along with a high index of suspicion.

Keywords: Caffey disease, cortical hyperostosis, inflammation, periosteum

Introduction:

Caffey disease is a rare self-limiting genetic disorder which usually affects young infants with an average age of onset of 9 to 11 weeks of life 1. It is characterised by an episode of massive subperiosteal new bone formation involving the diaphysis of the mandible, scapula, clavicles, long bones and is associated with increased inflammatory markers, increased alkaline phosphatase levels and systemic changes of fever and irritability1. Most cases are sporadic, but a few familial cases with both autosomal dominant and recessive patterns have been described 2. In the familial form, a novel missense mutation COL1A1, which is the gene encoding for the alpha-1 chain of Type 1 collagen has been found 3. This has led to some authors placing Caffey disease in the same family as type I collagen-related diseases such as osteogenesis imperfecta I-IV, Ehlers-Danlos syndromes type I and VII, idiopathic osteoporosis, and dermatofibrosarcoma protuberans 3, 4.

We report a case of acute sialadenitis with underlying Caffey disease which led to a significant diagnostic dilemma. We highlight this self-limiting disease entity to avoid unnecessary investigations.

Case Description:

A one and a half month old male child, first issue of a non consanguineous marriage was admitted with bilateral neck swelling since day of life fifteen associated with fever. He was exclusively breast fed and his birth history was uneventful. Anthropometry parameters were within normal limits. Systemic examination was normal. Local examination revealed firm tender mobile swelling with normal overlying skin on both sides of jaw. The laboratory investigations were as follows: Hemoglobin 6.9 gm/dl, WBC count 21,300/cmm with neutrophilic dominance, platelet count 10.2 lakhs/cmm, CRP 62.7 mg/dl, salivary amylase 329 U/L (25-125), alkaline phosphatase 1024 IU/L (98-279), immunoglobulins levels [IgG 817 mg/dl (35-1620), IgA 73.2 mg/dl (1-91), IgM 95.2 mg/dl (6-66)]. Ultrasonography (USG) neck was suggestive of bulky and hypoechoic parotid and submandibular glands suggestive of acute sialadenitis with bilateral cervical lymphadenopahy. Hence, child was treated with intravenous ceftriaxone and cloxacillin after sending blood culture. However, child did not show any improvement even after 48 hours and hence suspecting methicillin resistant staphylococcus (MRSA) infection, antibiotics were escalated to intravenous Vancomycin

and Piperacillin-Tazobactam. Serial blood cultures were negative. Although the fever subsided, high WBC counts and raised CRP were persistent throughout the course of the hospital stay. Intravenous antibiotics were continued for 14 days. At the end of his antibiotic course, child was afebrile, less irritable with persistence of high counts, thrombocytosis and elevated CRP (Hemoglobin-6.9 gm/dl, WBC count-25,900/cumm; platelet count-12·74 lakhs/cumm, CRP-74 mg/dl). Repeat USG showed reduction in the size of both the glands (parotid & submandibular) and the cervical lymph nodes. However, persistence of underlying jaw swelling, and raised WBC counts, platelets, CRP, alkaline phosphatase levels led us to suspect an underlying Caffey disease, X-ray of mandible revealed gross subperiosteal spongy osseous thickening suggestive of osetitis [Figure 1, Figure 2]. X-rays of all the other bones were normal. The child was commenced on oral Ibuprofen and discharged with significant improvements after six months of follow up.

Discussion

The clinical features of Caffey disease described by Caffey and Silverman include irritability, swelling of the overlying soft tissue which usually precedes the cortical thickening of the underlying bones, fever and anorexia 1. The swelling is painful, firm with no evidence of redness or suppuration. Common sites include mandible followed by scapula, clavicle, ribs and long bones and rarely the ilia, parietal bones, and metatarsals1. Our patient had an early onset at one and a half months with isolated mandible involvement. An infantogram to look for other bone involvement was normal. The laboratory findings seen in Caffey disease include elevated acute phase reactants, high alkaline phosphatase, thrombocytosis, anaemia and raised immunoglobulin levels1. Our patient had all of the above lab features. Serum immunoglobulin levels were normal in this patient except for high IgM. Co-existence of acute sialadenitis with Caffey disease is not known. This is the first case reported in the literature and led to a significant diagnostic challenge. Our patient presented with features of acute sialadenitis involving both the parotid and submandibular gland which partially responded to antibiotics as demonstrated clinically by fever remission and reduction in the size of the glands (parotid & submandibular) and lymph nodes. However, persistence of underlying jaw swelling with elevated acute phase reactants and alkaline phosphatase levels led us to a suspicion of Caffey disease which was confirmed by X

ray of the mandible. Other differential diagnosis (osteomyelitis, hypervitaminosis A, malignancy, scurvy, bone tumours and child abuse) were excluded based on history and physical examination.

Radiography is the most critical diagnostic study in Caffey disease 5. The presence of gross subperiosteal spongy osseous thickening of the mandible along with no evidence of any osteolytic lesion confirmed the diagnosis of Caffey disease in our patient. Magnetic resonance imaging (MRI) does not add any significant information to the results obtainable with plain radiography 6.

Caffey disease is usually self-limiting and resolves within six months to one year 7. However, non-steroidal anti-inflammatory drugs could be used in symptomatic cases as discussed by Varma et al 8. Throughout the disease, the bone lesions can recur at their original sites or at newer sites at a later stage 9. Hence, follow up of these children is critical.

Conclusion

We present a rare case of Caffey disease with acute sialadenitis which led to a perplexing diagnostic dilemma. Caffey disease is a clinical diagnosis, and our awareness of the existence of this condition and its typical clinicoradiological profile avoided the child being subjected to unnecessary investigations. The aim of this case report is to alert paediatricians of the occurrence of acute sialadenitis in a child with underlying Caffey disease which can lead to challenging diagnostic dilemma.

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Figure-1

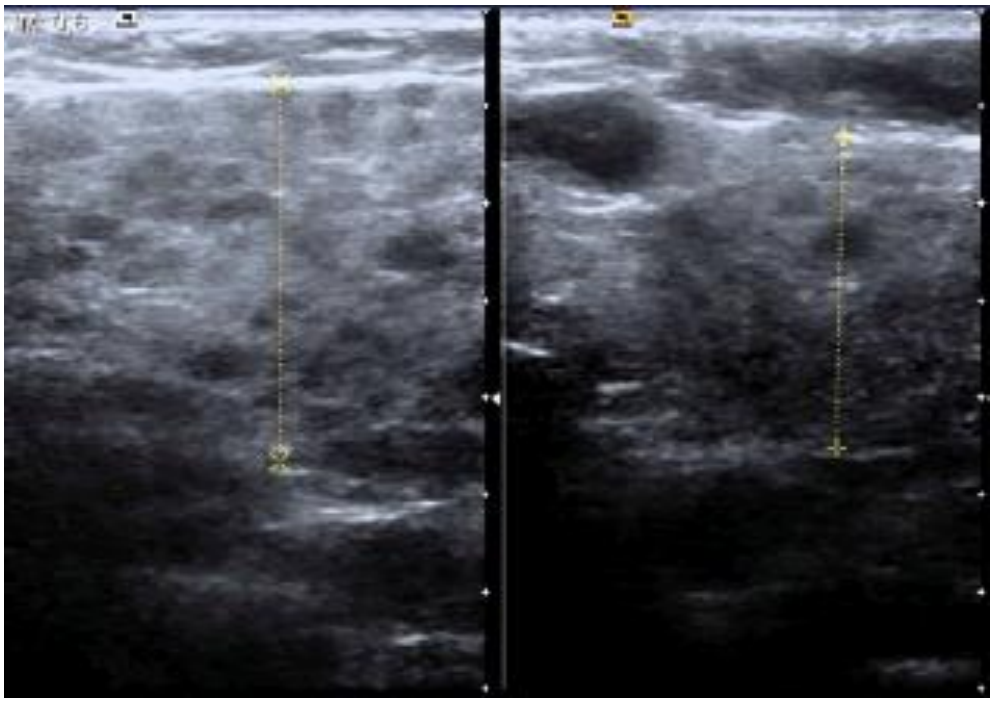

Fig 1: USG showing bilateral parotid enlargement with heterogeneous echogenicity

Figure 2

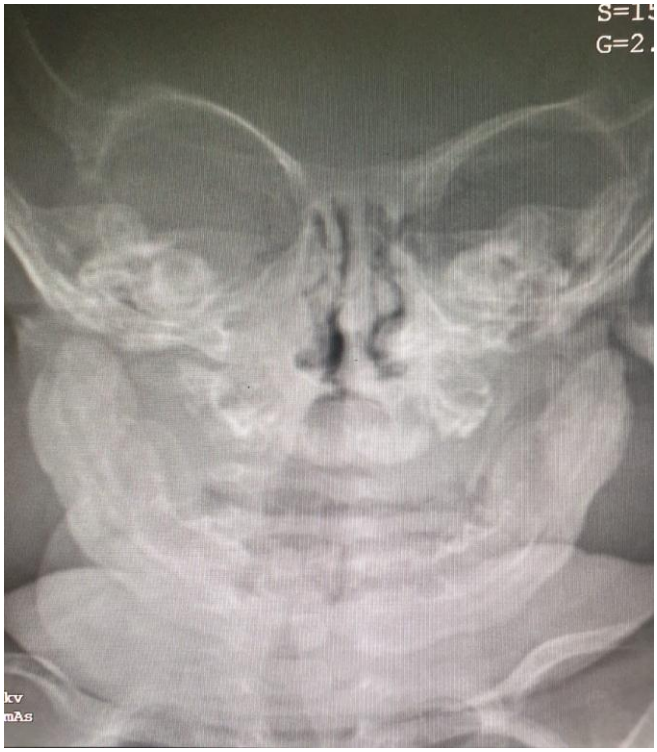

Fig 2: Xray mandible (AP view): Gross subperiosteal spongy osseous mandibular thickening suggesting osteitis

Figure 3

Fig 3: Xray mandible (lateral view): Gross subperiosteal spongy osseous mandibular thickening suggesting osteitis