Caffey Disease Masquerading as Osteomyelitis: A Case Report and Review of the Literature

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We present a case of a 4 month old female presenting with arm swelling and pain, whose clinical presentation and work-up led to an unexpected diagnosis of infantile cortical hyperostosis.

Abstract

Infantile cortical hyperostosis (Caffey Disease) was first described by Caffey and Libman in 1947. It is a rare bone disease that begins in early infancy presenting with some form of irritability, 2-10 bone swellings, and cortical hyperostosis of the skull, commonly including the mandible, skull, clavicle, or rib. The mandible is the most frequently involved bone, present in 75-85% of cases, and it's involvement is considered pathognomonic. Imaging findings include periosteal new bone formation with cortical thickening and underlying soft tissue swelling. Theiology of Caffey Disease is unknown, but it has been recently associated with type 1 collagen mutations.

Case Report

History of present illness

Our patient is a 4 month old Hispanic female presenting with a two month history of pain and swelling of the left forearm. She was born at 36 weeks to a G1P1 mom via C-section due to fetal distress. She was diagnosed with a left forearm fracture, splinted, and discharged home.

Physical exam:

VS: T 99.2, HR 138, RR 36, BP 81/53, Weight 6.23kg

PH: Full Savvy

Genitalia: Female

Abdomen: Soft, no hepatosplenomegaly

Gen: Awake, alert, fussy during exam

Family history: No bone, connective tissue disease, no immunodeficiencies.

Social history:

No daycare exposure.

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Primary care providers:

Birth: Full term; no complications

Elective cesarean section due to fetal distress.

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Figure 2: Bone biopsy findings

Figure 3: Images from the literature

Figure 1: Imaging findings

Left forearm plain film showing diffuse periosteal reaction with periosteal thickening and cortical fracture.

MRI of the left forearm showing metaphyseal and epiphyseal inflammatory changes with periositos and abnormal marrow signal throughout the left radius.

AC-CT scan of the head was performed as part of the non-accidental trauma work-up and revealed symmetric, chronic-appearing periosteal reaction consistent with the mandibular ram.

Differential diagnosis:

- Infections etiology
- Osteomyelitis caused by Staphylococcus aureus, Group A β-hemolytic streptococci, Haemophilus influenzae, and Neisseria meningitidis
- Congenital infections such as syphilis
- Metabolic disorders such as congenital infections
- Hereditary storage disorders such as I cell disease
- Inherited metabolic disorders such as urea cycle disorders, tyrosinemia type 1, inborn errors of purine and pyrimidine metabolism

Conclusions

We present an unusual case of Caffey Disease seen at Texas Children’s Hospital in Houston, Texas, and perform a review of recent literature. Caffey Disease is an inflammatory disorder that presents with a characteristic triad of irritability, soft tissue swelling in the bone, and an involucrum or thickening of the cortical bone. It has been recently associated with bone collagen mutations.

References