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Caffey disease – A rare case report.

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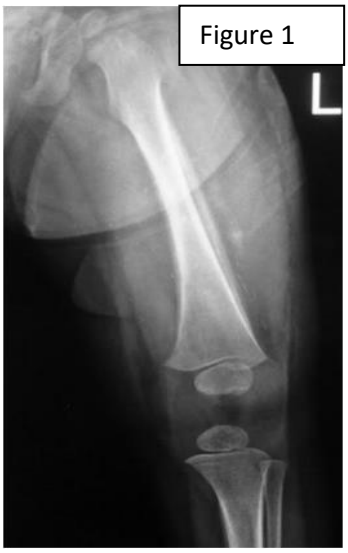
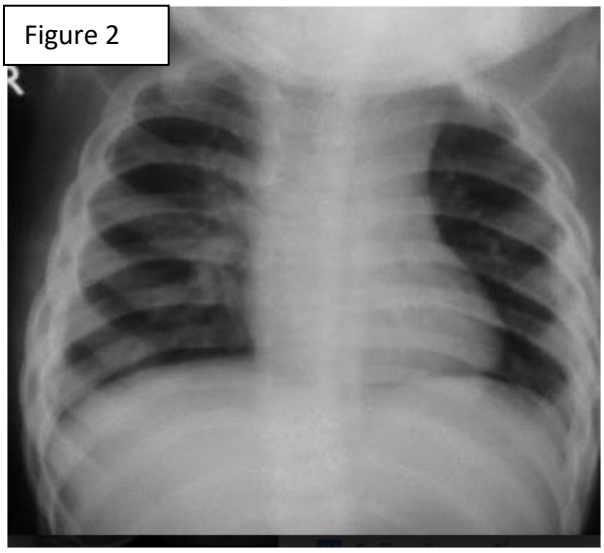
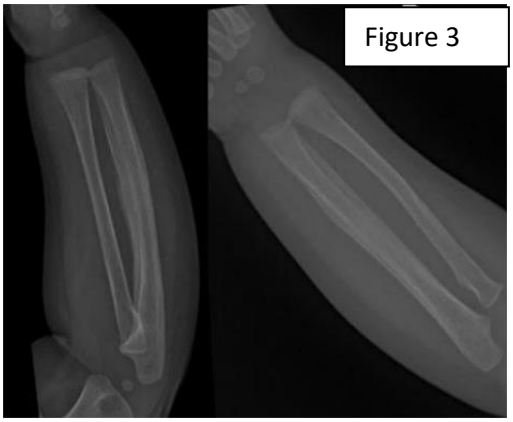
HISTORY:

8 month old male infant with localized swelling of the thigh, fever and irritability.







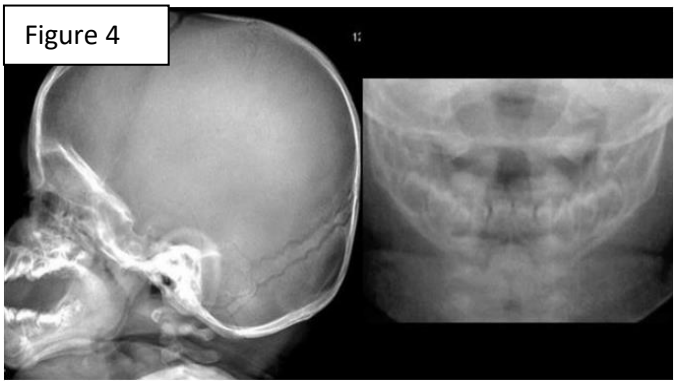


Figure 1: AP radiograph of the Left Thigh. Soft tissue swelling of the left thigh with underlying periosteal new bone formation & cortical thickening isolated to the diaphysis of left femora.

Figure 2: Chest Radiograph. Cortical hyperostosis along the inferior margin of the ribs.

Figure 3: AP and Oblique Radiographs of the left forearm.Limb edema with underlying new bone formation involving the ulnar diaphysis “spindle shaped”.

Figure 4: Lateral and AP Radiographs of the mandible. Diffuse thickening and increased density of maxilla and mandibles. Note the thick layer of periosteal new bone formation along the inferior surface of the mandible.

FINAL DIAGNOSIS: CAFFEY DISEASE

CAFFEY DISEASE.

Aka: Infantile Cortical Hyperostosis (ICH) / Caffey-Silverman Syndrome / de toni Caffey disease.

First reported as a disease entity by Caffey and Silverman in 1945,

- **Described two types**, a classical mild infantile form (ICH) delineated by Caffey and Silverman and a severe form with prenatal onset.
- ICH is self-limiting disease begins in early infancy, usually before 6 months of age and is characterized by clinical triad of fever, irritability, soft tissue swelling.
- Severe prenatal onset form is characterized by extensive hyperostotic bone involvement, angulations and shortness of long bones, as well as polyhdramnios and fetal hydrops [DD: Lethal form of Osteogenesis imperfecta].

Etiology and Epidemiology:

- Self limiting disorder of unclear etiology.
- Mostly sporadic but a few familial cases with autosomal dominant and recessive patterns have been described. Mutation in the COL1A1 gene (type1 collagen) is associated with Caffey's, though the exact etiopathogenesis remains largely unknown.
- No sex or racial predilection.
- Estimated occurrence approx 3 per 1000 infants, however an interesting and unexplained fact is the gradual decrease in the number of cases over the last couple of decades.

Diagnosis: Clinical triad of symptoms in an infant, Radiological features (enumerated below) and Lab investigations. ESR and alkaline phosphatase levels are often elevated. Histologically at its inception, the lesion is intraperiosteal with formation of osteoid trabeculae within the inflamed periosteum, subsequently the process extends to involve the juxtaperiosteal soft tissue, muscles and connective tissue and eventually replaced with new bone formation.

Radiology of Caffey's Disease:

- Multifocal and asymmetric with a predilection for tubular bones.
- Hallmark: Periosteal new bone formation and cortical thickening that underlies the soft tissue swelling.

<u>Plain Radiograph:</u>

- Ulna and Clavicle are the most common sites. Involvement of the mandible & the ribs also frequently occur. Other long bones, scapula, skull may also be involved. All the bones of the body are implicated except phalanges, vertebral bodies and cuboidal bones.

- Tubular bones: Only the diaphysis is involved causing spindle shape of bones. Massive cortical thickening & widening of bones is a noted feature. Bridging of bones across interosseous membrane (local fusion of cortices) and enlargement, marginal hyperostosis and sclerosis of flat bones occur.

Skeletal Scintigraphy: Increased non-focal uptake indicative of new bone formation, correspond to the areas of skeletal hyperostosis.

MRI: Periostitis with soft tissue edema. In isolation, MR findings can lead the radiologist to give alternate diagnosis. Hence radiography should be the primary modality of investigation and follow up.

Course of the disease:

- Usually a self - limiting disease. Choice of steroids is empirical to ameliorate pain and the child's suffering.
- Rarely, the active disease may persist and recur intermittently for years with crippling deformities in the extremities and markedly delayed muscular and motor development.
- In long standing cases, the hyperostosis appears to be reamed out from inside producing a thin walled bone with large medullary cavity.

Differential diagnosis: Osteomyelitis, Battered baby syndrome, Hypervitaminosis A, Hyperphosphataemia, Prostaglandin E1 and E2 administration, Congenital syphilis & Metastatic Neuroblastoma.

References:

1. Frana L, Sekanina M. Infantile cortical hyperostosis. Archives of Disease in Childhood 1976;51(8):589-95.
2. Newberg AH, Tampas JP. Familial infantile cortical hyperostosis: an update. AJR. American Journal of Roentgenology 1981;137(1):93-6.
3. Phatak S V, Kolwadkar P K, Phatak M S. Pictorial essay: Infantile cortical hyperostosis (Caffey's disease). IJRI 2004; 14:185-6