Infantile Cortical Hyperostosis

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Case details

- 5mon old male infant
- Brought with c/o episodes of
  - Irritability
  - Swelling over right mandible
  - Episodes of crying

- Possibility of seizure equivalent considered hence treated for the same
- MRI Mandible: Chronic OM of Mandible
- Ref. to us as Recurrent osteomyelitis of mandible
On examination

- Afebrile, non-toxic
- Swelling over Rt. Manible, Hard in consistancy, Tender
- Systemic examination: Normal
Investigations:

- **CBC:**
  - TC: 16800
  - PMN: 56; L: 36; St: 6; M: 2
  - Hb: 8.2 gm/dl
  - Platelet: 6.7 lack/cm²
- **CRP:** 59 mg/lit
- **Ca:** 9.1 mg/dl; **Po4:** 6.9 mg/dl
- **SAP:** 758 IU/lit
- X ray mandible: **soft-tissue swelling and calcification and cortical hyperostosis**

**DIAGNOSIS:** CAFFEY’s disease
Treated

NSAIDs (Ibuprofen) for 1wks

Complete resolution of swelling

Reviewed after 1wk with recurrence of swelling

Repeated Ibuprofen …..,
CAFFEY’S DISEASE
Etiology

- Caffey and Silverman first reported this disease as a distinct entity in 1945

- unknown etiology.

- It is often sporadic, but both autosomal dominant and autosomal recessive inheritance have been reported.

- In three unrelated families with autosomal dominant inheritance, a linkage to mutations of the \textit{COL1A1} gene (codes for the $\alpha_1$ chain of type I collagen) has been reported.
Clinical features:

• The typical clinical triad includes fever of abrupt onset, hyperirritability, and soft-tissue swelling (especially over the mandible)

• Onset in infancy (<6 mo, average 10 wk) is most common

• The swelling is painful with a woodlike induration but with minimal warmth or redness; suppuration is absent. The soft-tissue swelling precedes the bony change

• The bones most commonly affected are flat bones: mandible (75% involvement), clavicle, rib (especially the lateral arches), scapula, skull, and ilium.
• The tubular bones most commonly affected are the ulna bones, which usually show asymmetric involvement.

• There are unpredictable remissions and relapses; an episode may last 2 wk to 3 mo.
• **Laboratory data**
  
  • elevated erythrocyte sedimentation rate (ESR),
  • an elevated serum alkaline phosphatase level,
  • moderate leukocytosis, thrombocytosis,
  • iron-deficiency anemia, Anemia is thought to be due to widespread myelofibrosis.
• **X ray:**
  - Plain radiographs may show soft-tissue swelling and/or cortical hyperostosis (with doubling or tripling of the normal width of the bone).
  - The periosteal reaction progresses to subperiosteal new bone formation.

• **MRI:**
  - Compared with plain radiography overall, MRI adds little important additional information for the clinical evaluation of Caffey disease, but is useful when infection or neoplasia are considered more likely diagnoses.
  - MRI may be used to exclude subperiosteal hemorrhage; MRIs depict hemorrhage with subsequent new bone formation, as seen with differential diagnoses (e.g., trauma, scurvy).

• **Bone scan:**
  - The distribution of radiotracer accumulation is similar with bone and gallium scans. Accumulation of the radiopharmaceutical in the involved bones is markedly increased during the active phase of the disease.
  - The characteristic "bearded-child" appearance is due to the intense and diffuse abnormal accumulation of radiotracer in the mandible.
<table>
<thead>
<tr>
<th>Differential Diagnosis</th>
<th>Features Resembling those of Caffey Disease</th>
<th>Features Distinct from those of Caffey Disease</th>
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<tbody>
<tr>
<td>Osteomyelitis</td>
<td>Similar MRI findings of soft tissue and marrow edema, periosteal reaction</td>
<td>Usually only affects 1 bone for a given clinical period, bone destruction and sclerotic bony changes</td>
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<td>Leukemia</td>
<td>Pronounced periosteal bone formation</td>
<td>Lytic bone lesions, radiolucent metaphyseal bands</td>
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<td>Hypervitaminosis A</td>
<td>Periosteal new bone formation typically along the diaphysis of long bones</td>
<td>Characteristic clinical/radiographic findings at the end of the first year, mandible not involved, increased blood level of vitamin A</td>
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<td>Healing rickets</td>
<td>Striplike density that parallels the outer cortical margin of long bones, resembling a periosteal reaction</td>
<td>Splaying and irregularity of the metaphysis, slower resolution of clinical and radiographic findings</td>
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<tr>
<td>Bone tumor</td>
<td>Periosteal new bone formation, similar appearance of microscopic proliferation of subperiosteal cells</td>
<td>Malignant features of bone tumors including tumor mass; solitary lesion</td>
</tr>
</tbody>
</table>
• **Treatment includes:**
  
  • Mainly palliative, Pain relief
  • indomethacin
  • prednisone if there is a poor response to indomethacin.
Thank you