Diseases of Bone manifested in the jaws

M. Mupparapu, DMD
Division of Oral & Maxillofacial Radiology
Department of Diagnostic Sciences
m.mupparapu@umdnj.edu

• Fibrous dysplasia
• Periapical cemental dysplasia (PCD)
• Florid cemento-osseous dysplasia (FCOD)
• Cemento-ossifying fibroma
• Aneurysmal bone cyst
• Cherubism
• Paget’s disease
• Langerhan cell histiocytosis

Benign
1-5 yrs.
1. Ewing’s Sarcoma
2. Rhabdomyosarcoma
3. Neuroblastoma
4. Lymphoma
5. Retinoblastoma
6. Wilms tumor
7. Eosinophilic granuloma
8. Neurofibromatosis

5-10 yrs.
1. Ewing’s Sarcoma
2. Rhabdomyosarcoma
3. Neuroblastoma
4. Lymphoma
5. Retinoblastoma
6. Wilms tumor
7. Eosinophilic granuloma
8. Neurofibromatosis

10-15 yrs.
1. Ewing’s Sarcoma
2. Rhabdomyosarcoma
3. Neuroblastoma
4. Lymphoma
5. Retinoblastoma
6. Wilms tumor
7. Eosinophilic granuloma
8. Neurofibromatosis

16-25 yrs.
1. Ewing’s Sarcoma
2. Rhabdomyosarcoma
3. Neuroblastoma
4. Lymphoma
5. Retinoblastoma
6. Wilms tumor
7. Eosinophilic granuloma
8. Neurofibromatosis

Fibrous dysplasia
• Results from localized change in normal bone metabolism
• Results in replacement of all components of cancellous bone by fibrous tissue containing varying amounts of abnormal appearing bone
• Monostotic, polyostotic (Jaffe type) and associated with McCune-Albright syndrome
• Monostotic form accounts for 70% of all cases
• The occurrence in the order of decreasing frequency
  – Rib
  – Tibia
  – Femur
  – Maxilla
  – Mandible
• Maxilla:mandible 2:1 occurs in the posterior aspect
• Unilateral involvement

Fibrous dysplasia
• Current definition: It is a benign neoplasm with a low grade malignant potential
• Somatic mutations within the GNAS 1 gene
• Patients with Albrights syndrome – associated with pituitary adenomas, have malignant potential

Polyostotic fibrous dysplasia
• One or more endocrine abnormalities
• Café au lait pigmentation

Café au lait pigmentation
Radiographic features

1. Affects maxilla more than mandible 2:1
2. Ill-defined periphery with gradual blending of normal trabecular bone into an abnormal pattern
3. On occasion, the boundary appears sharp and even corticated
4. The abnormal trabeculae are shorter, thinner, irregularly shaped and more numerous, which creates
5. Radiopaque appearances varying from "ground glass" to "orange peel" to a wispy "cotton wool" appearance
6. Small lesions do not show any effect on surrounding structures
7. Thinning of outer cortex is observed
8. Displaces mandibular canal in a superior direction
9. 0.01% of patients with FD may develop osteosarcoma
10. 4% of patients with McCune Albright syndrome may develop osteosarcoma

Case report

- A.R., a 40-yr-old Nigerian male
- Referred to the NJDS-OMFR clinic for radiographic exam of the mandible
- Chief complaint: occasional pain in the left jaw and shoulders for approximately 3 yrs
Panoramic view

Affect on adjacent structures

D/D
- Hyperparathyroidism. Polyostotic, bilateral and do not cause bone expansion [Exception - Secondary HP]
- Paget’s disease. May produce similar bone pattern, may cause expansion but occurs in an older age group. The entire mandible is involved
- PCD. Occasionally may show a similar bone pattern + occurs in older age group
- Osteomyelitis. May result in the enlargement of the jaws, but the additional bone is laid down on the surface of the outer cortex
- Osteogenic sarcoma. May have a similar appearance but should show malignant radiologic features.

Ethmoid sinuses

Sphenoid sinus involvement
Genetics/Diagnosis

GNAS 1 +ve. GNAS 1 mutations are also seen in thyroid adenomas, osteosarcoma, breast ca and some other malignancies

High levels of C-Fos proto-oncogene expression. This is positive in both fibrous dysplasia and osteosarcoma

Secondary Hyperparathyroidism [Alport Syndrome]

- Genetic cause of renal failure. X-chromosome linked. Gene mutation causes abnormalities in type IV collagen leading to abnormal accumulation of type V and type VI, that in turn leads to permeability problems within the kidney leading to SCLEROSIS
- Renal sclerosis>>> Chronic Renal Failure
- Decreased production of vitamin D→lower calcium levels and increased serum phosphate levels
- Eventually leading to HYPERPARATHYROIDISM
- Compensatory anemia, osteomalacia, osteitis fibrosa cystica and secondary hyperparathyroidism
Periapical Cemental Dysplasia

PCD Vs Cementoblastoma & Osteoblastoma

Simple bone cyst and FCOD

Florid Cemento-osseous dysplasia

- FOD lesions are bilateral and present in both jaws.
- Lesions in the mandible occur above the inferior alveolar canal
- Periphery is very similar to PCD
- May have cotton wool appearance
- FOD lesions can displace infra alveolar canal inferiorly and the floor of the maxillary sinus superiorly

Florid cemento-osseous dysplasia: a systematic review

Dentomaxillofac. Radiol., May 1, 2003; 32(3): 141 - 149
Cemento-ossifying fibroma

- Highly cellular, fibrous tissue that contains varying amounts of abnormal bone or cementum-like tissue
- Soft tissue capsule is present around the lesion
- Juvenile ossifying fibroma, an aggressive form of COF occurs in the first two decades of life
- Teeth are displaced
- Well defined borders, separated from the normal bone by a thin radiolucent line representing a fibrous capsule
- Advanced Imaging: CT

Aneurysmal Bone Cyst

- A reactive lesion of bone
- Resembles CGCG due to the histologic presence of the giant cells
- ABCs may develop in association with other primary lesions such as fibrous dysplasia, central hemangioma, giant cell granuloma and osteosarcoma.
- Occurs in individuals <30 yrs, mostly females
- Rapid bony swelling, painful
- Mandible to maxilla 3:2, molar region > anterior region
- Well defined periphery, circular
- Multilocular and septate resembling Central Giant Cell Granuloma
- Extreme expansion of outer cortical plates
- ABCs can displace and resorb teeth
- A hemorrhagic aspirate favors the diagnosis of ABC
- Advanced Imaging: CT
Cherubism

- A rare inherited developmental abnormality that causes bilateral enlargement of the jaws giving the child a cherubic facial appearance.
- Researchers isolated the gene responsible – chromosome 4p16
- Lesions grow in the postero-anterior direction
- Bilateral multilocular lesions, well defined periphery
- Ramus/hyperviscosity region is epicenter of the lesions
- Lesions get filled in with granular bone after the active phase ends
- Cosmetic recontouring recommended


Case courtesy: Dr. Art Nouel, Santo Domingo
Paget’s disease of bone (osteitis deformans)

- Bone pain
- Headaches and hearing loss
- Pressure on nerves
- Increased lead call, leaning of limb, or curvature of spine
- Softening of bone
- Damage to cartilage of joints

Appearance of internal structure depends on the radiographic stage:

1. Phase I: Radiolucent resorptive stage
   a. Bone resorption predominates
   b. Bone turnover is 20 times normal rate
2. Phase II: Granular/ground glass appearing stage
   a. Woven bone formation
   b. Ineffective mineralization
3. Phase III: Dense bone deposition
   a. Bone is disorganized and sclerotic
   b. Weaker than normal bone

Osteoporosis Circumscripta

- Always exhibits bone enlargement
- Kidney stones are common in patients with Paget’s
- Skull bones may enlarge 3-4 times their normal thickness
- Outer cortex may remain the same or slightly thinned
- Bone scan reveals the activity of the lesion (increased uptake)
- Extreme elevation of serum Alkaline Phosphatase levels aid in the diagnosis

Langerhans cell Histiocytosis

- Abnormal proliferation of Langerhans cell or their precursors (skin derived)
- 10% of all patients with LCH have oral lesions
- Eosinophilic granuloma commonly appears in the skeleton (ribs, pelvis, long bones, skull, jaws) and occasionally in the soft tissues
- Swelling, pain, bleeding, and loosening of teeth intraorally
- Well-defined periphery of the lesions radiographically, sometimes punched-out appearance
- Usually no root resorption, may stimulate new periosteal bone formation
- The epicenter of bone destruction starts at midroot level as opposed to the periodontal lesions where destruction starts at the crestal level
- Letterer-Siwe disease is the most severe form, fatal outcome
Eosinophilic granuloma

- Diagnosis by positive S100 protein staining of Langerhans cells
- Detection of rod shaped Birbeck granules of Langerhans cells
- Radiographic features consistent with the Histiocytosis

Cleidocranial dysplasia

- RUNX2 gene is responsible for ossification and was demonstrated in knockout mouse models where only cartilage was seen and no ossification was found.
- RUNX2 mutations are responsible for changes that are seen in cleidocranial dysplasia.
The End