RADY401 Case Presentation

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June 2018

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UNC SCHOOL OF MEDICINE Radiology

A 26-year-old female presents with headache and progressive bilateral vision loss

Patient History

CC is a 26-year-old female with a PMH of rheumatoid arthritis and recently diagnosed idiopathic intracranial hypertension who presents to the ED for frontal headaches 7/10 in intensity and progressively worsened bilateral vision loss. Patient has no vision in her right eye. She reports eye pain with movement. Patient also endorses hearing static noise in quiet settings and a loss of taste and smell over the past 3 months. Denies syncope, chest pain, SOB, nausea, vomiting, or recent illness.

Physical Exam

Vitals
BP 139/72
HR 95
RR 18
T 37.1 C
SpO2 100%

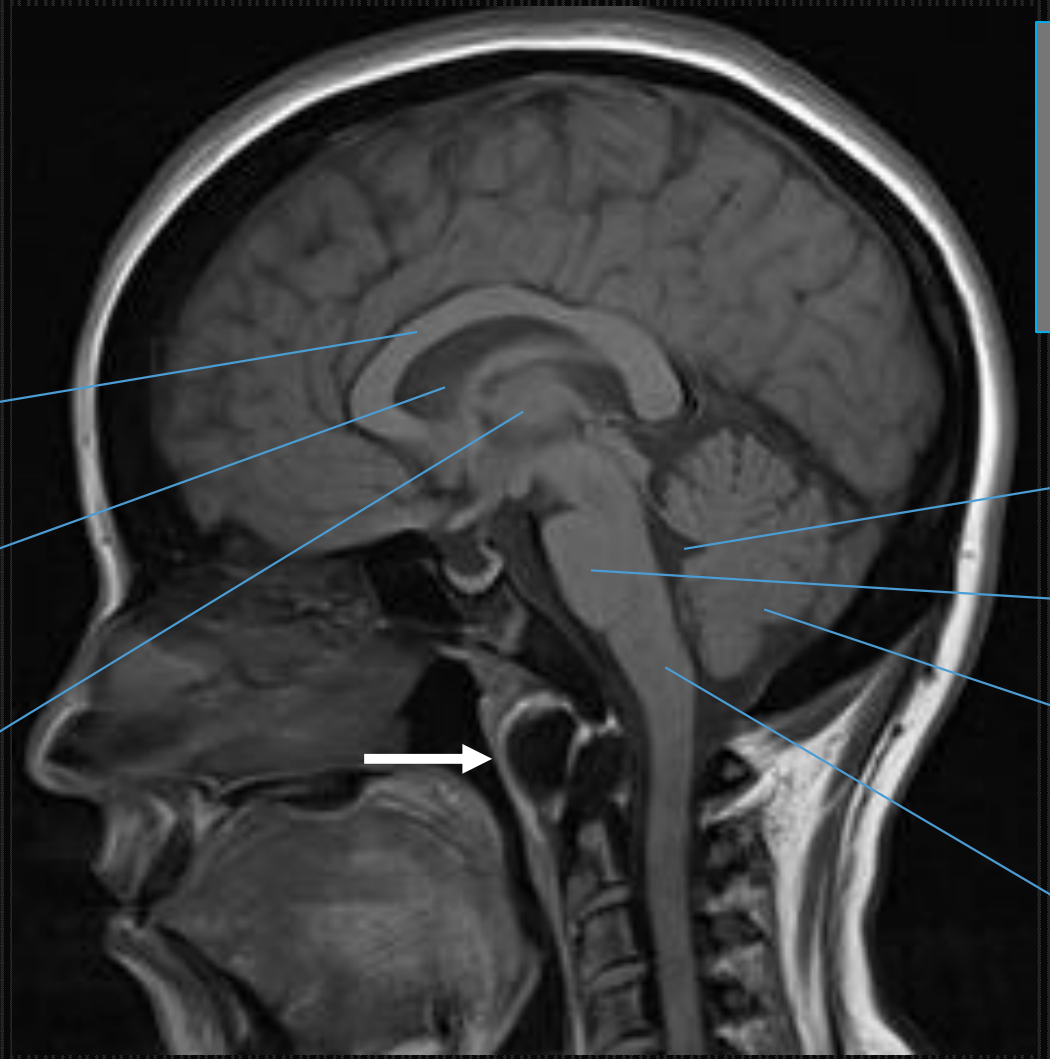
General: Well-appearing, well-nourished young female
HEENT: Normocephalic, clear conjunctivae, pupils 4mm bilaterally. L pupil briskly reactive, R pupil sluggish. No vision in R eye, 20/40 in L eye.
Resp: Clear to auscultation bilaterally
CV: RRR, no M/R/G
GI: Normoactive bowel sounds, non-distended, non-tender
Skin: Normal color, warm
Neuro: Moves all extremities with 5/5 strength throughout, sensation intact. Speech normal.

List of imaging studies

- MRI of the brain with and without contrast
- MRV of the head with and without contrast
- CT of the orbits without contrast

MRI of the Brain

Diffuse thickening and sclerosing of the bones of the calvarium, skull base, and upper cervical spine



Diffuse hypointensity of the skull bones on this T1-weighted sagittal slice indicates bone sclerosis. Note also the marked enlargement of the anterior C1 arch (arrow)

Corpus callosum

Lateral ventricle

Thalamus

Fourth ventricle

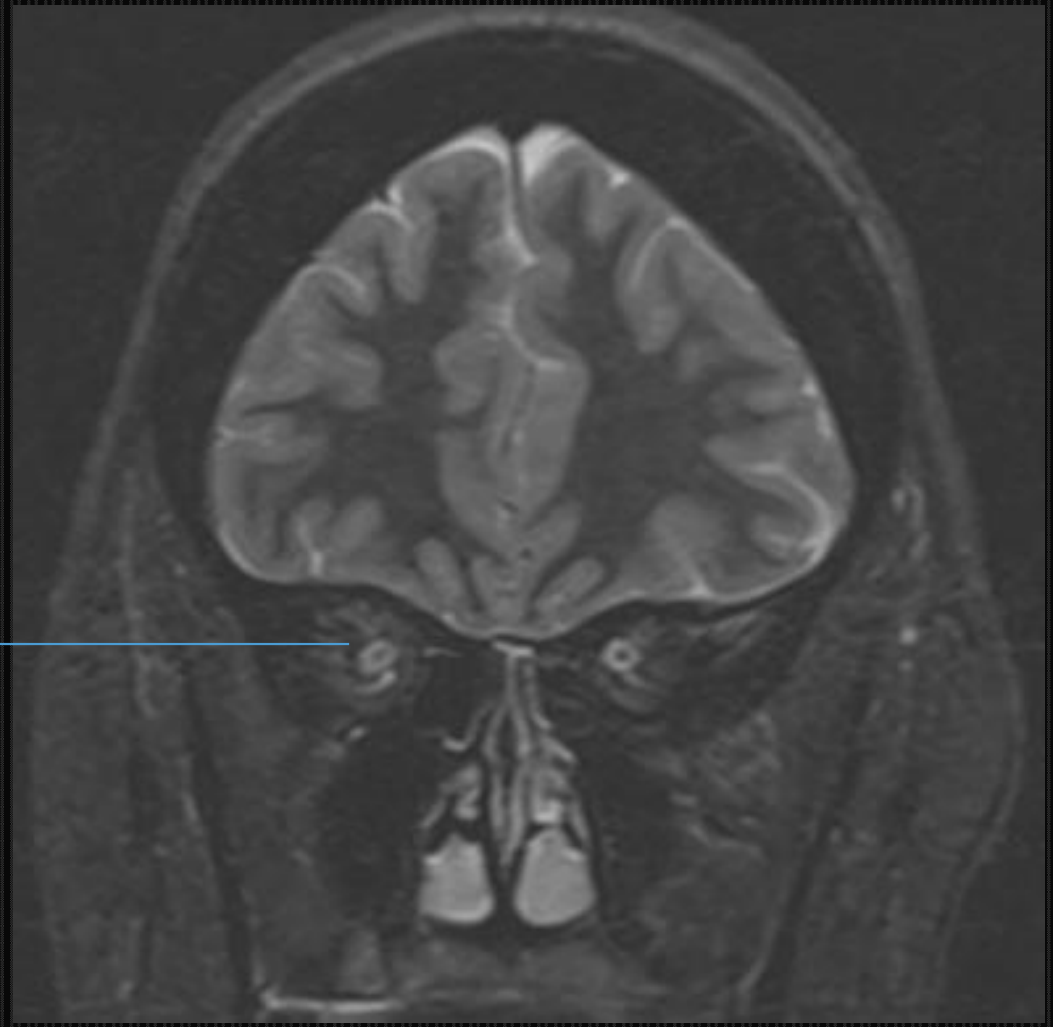
Pons

Cerebellum

Medulla

STIR Imaging

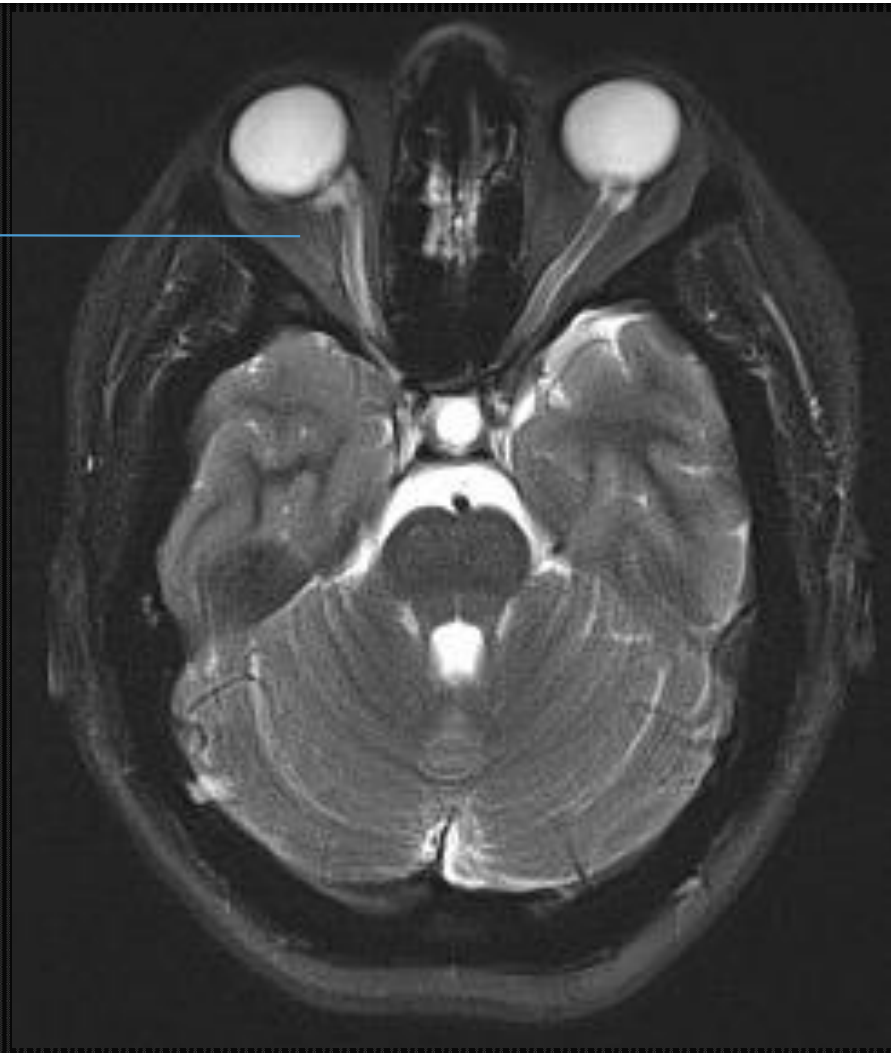
Mildly atrophic optic nerves bilaterally, right greater than left, visualized best on this coronal view using Short TI Inversion Recovery (STIR) imaging



STIR imaging is a technique that allows for increased sensitivity to malignancy and other abnormalities. It makes the effects of prolonged T1 and T2 on signal intensity additive while suppressing the signal from fat.

MRI of the Brain

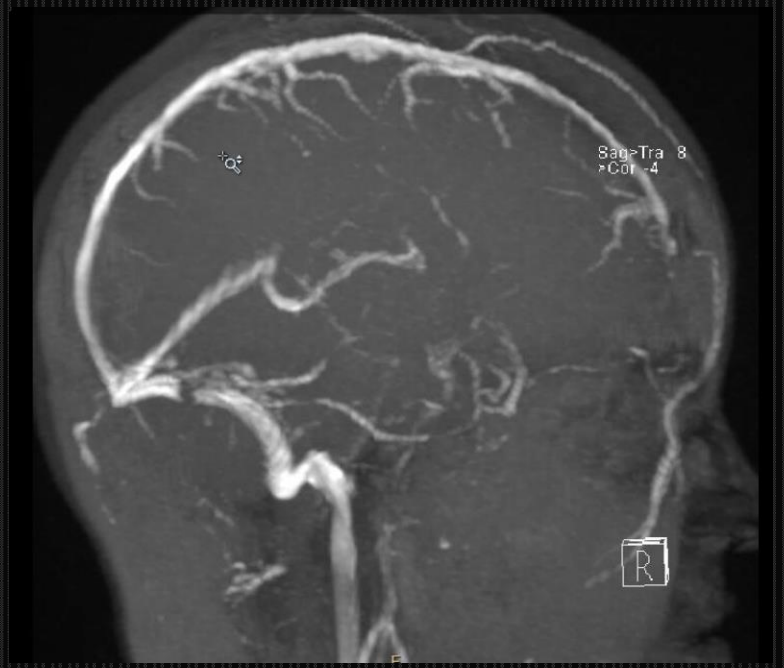
Bulging of the right optic nerve disc can be observed in this T2-weighted axial image, a finding suggestive of papilledema



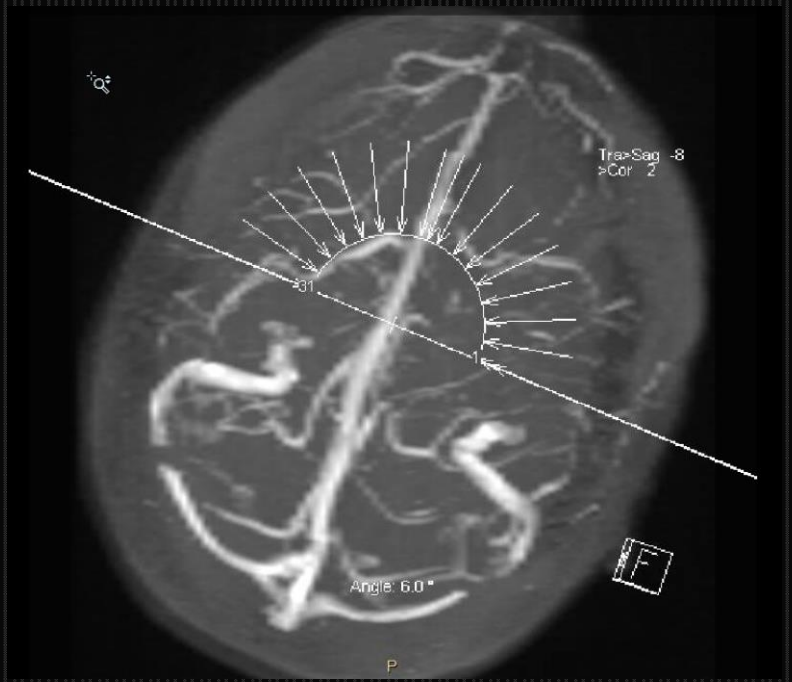
Fluid, edema, tumor, infarction, inflammation, infection, and subdural collections are all bright on T2-weighted imaging.

MRV of the head

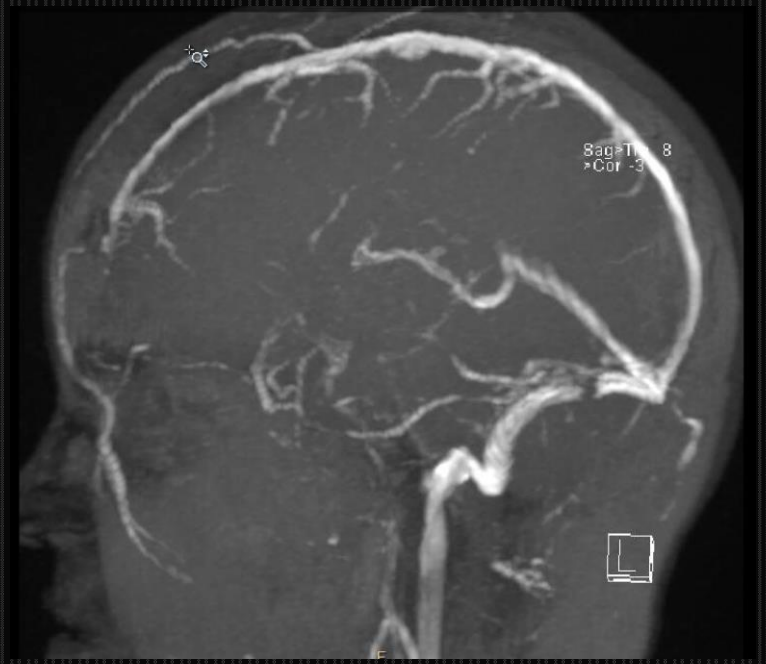
R Sagittal



Axial



L Sagittal

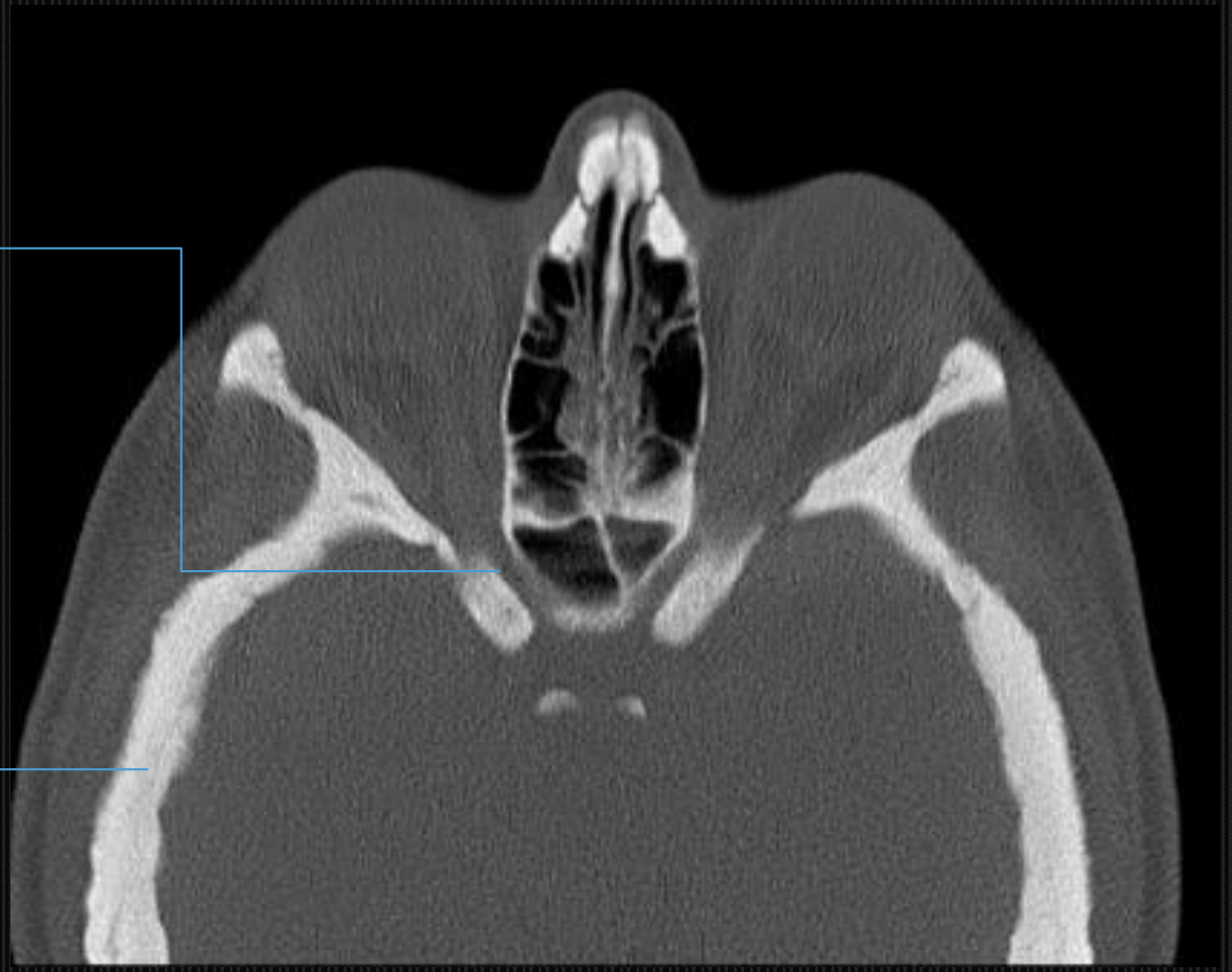


The intracranial venous circulation is unremarkable, with no evidence of venous sinus thrombosis

CT of the orbits without contrast

Enlargement of the anterior clinoid processes causing narrowing of the optic nerve canals is seen here on unenhanced CT

Diffuse sclerosis and thickening of the bones of the face and skull

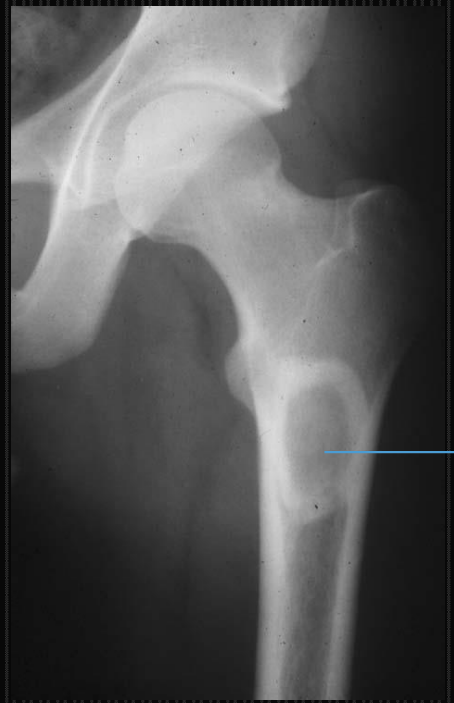


Differential Diagnosis

- Fibrous Dysplasia
- Paget's Disease
- Sclerosing metastases
- Osteopetrosis
- Van Buchem Disease

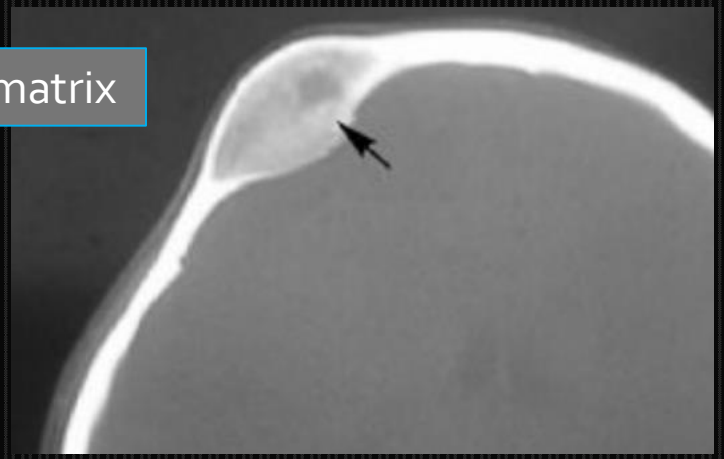
Differential Diagnosis: Fibrous Dysplasia

- Tends to present as a well-circumscribed, expansile lesion with ground-glass matrix and intact overlying bone
- Most common locations include the diaphysis or metaphysis of the proximal femoral shaft, tibia, humerus, and radius
- Fibrous dysplasia of the calcaneus is extremely rare



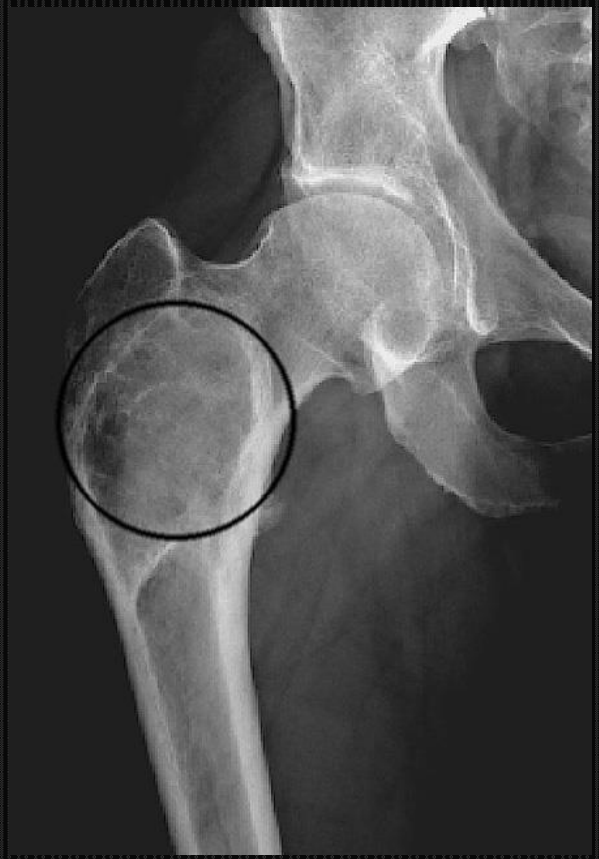
http://nagendraradiology.blogspot.com/2013/11/fibrous-dysplasia-fd-fibrous-dysplasia.html

Ground-glass matrix



http://academic.hep.com.cn/fmd/EN/10.1007/s11684-009-0024-7

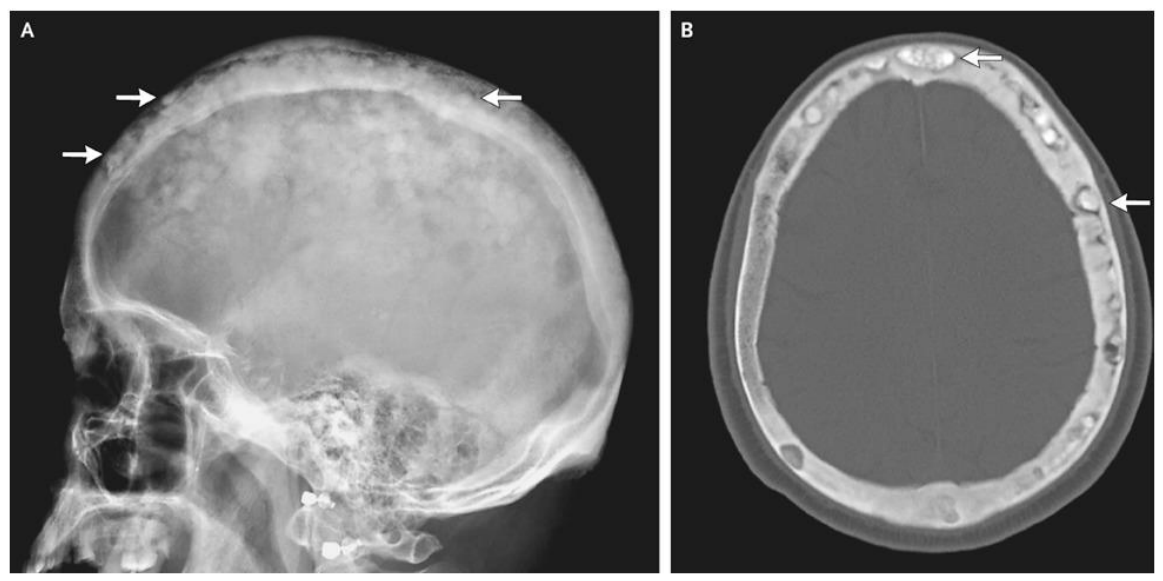
The "**rind sign**" refers to the sclerotic margin surrounding the lesion, and is highly suggestive of fibrous dysplasia



https://orthoinfo.aaos.org/en/diseases--conditions/fibrous-dysplasia/

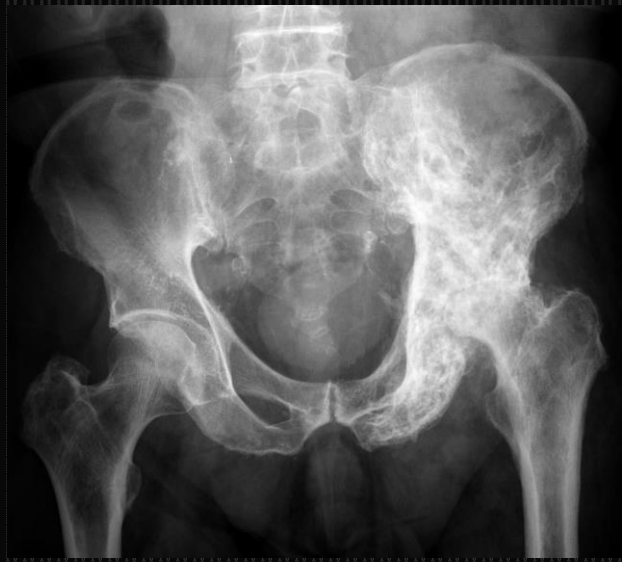
Differential Diagnosis: Paget's Disease

- Typically affects older patients, with characteristic thickening of the cortical skull
- Commonly affects spine, skull, pelvis, and long bones
- Consists of osteolytic and osteoblastic phases with accelerated bone turnover resulting in bone deposition and expansion



https://www.nejm.org/doi/full/10.1056/NEJMicm0912945

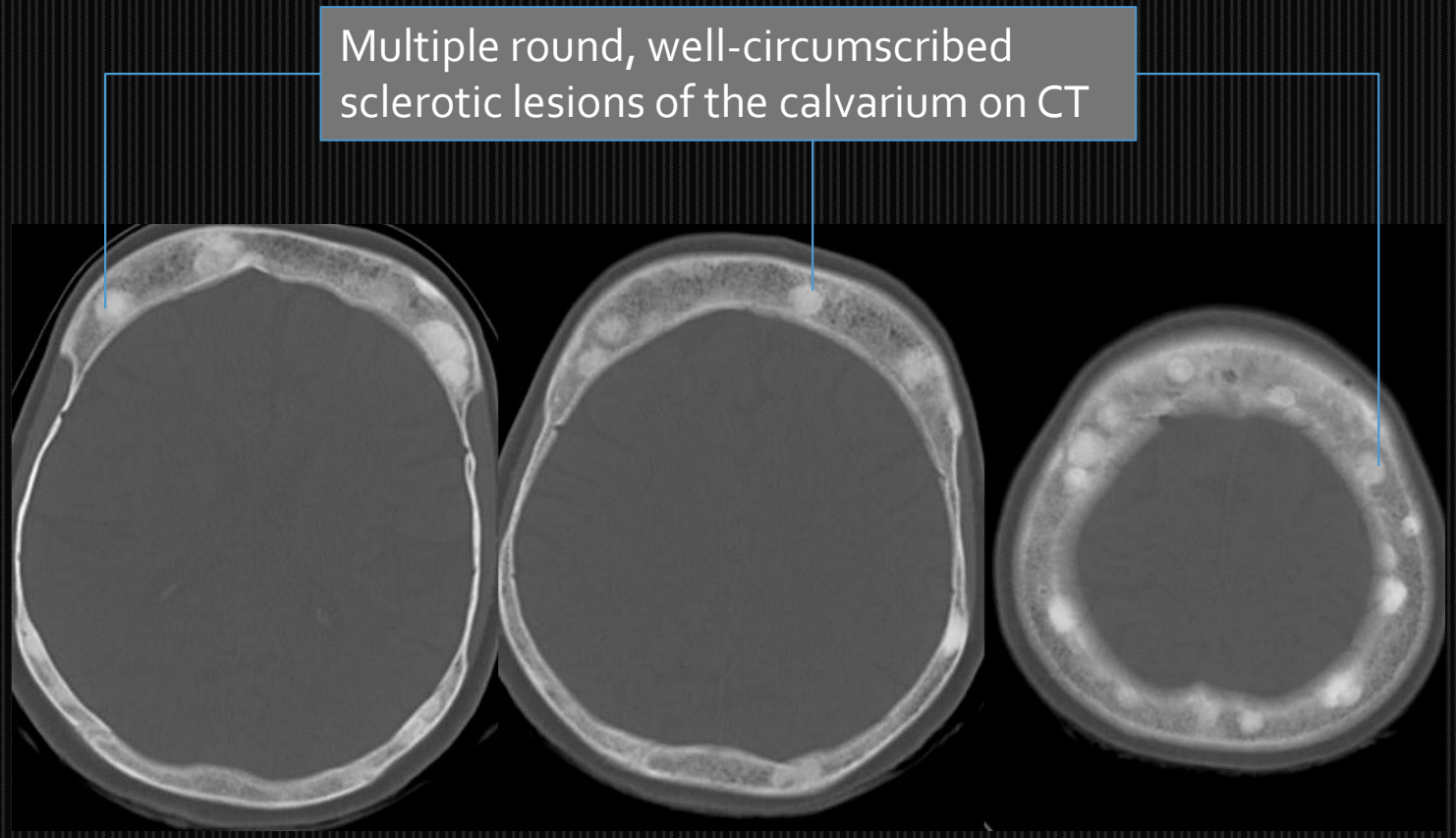
Characteristic "**cotton-wool**" thickening is due to irregular areas of sclerosis



https://orthoinfo.aaos.org/en/diseases--conditions/pagets-disease-of-bone/

Differential Diagnosis: Sclerosing metastases

- Presents as asymmetric, irregular foci of the skull, commonly associated with soft tissue masses
- Bone is the third most common organ affected by metastasis (behind lung and liver)
- Most common sites of metastases are the vertebrae, pelvis, ribs and the ends of long bones
  - Due to high red marrow content



http://www.neuroradiologycases.com/2012/01/osteoblastic-metastasis.html

Differential Diagnosis: Osteopetrosis

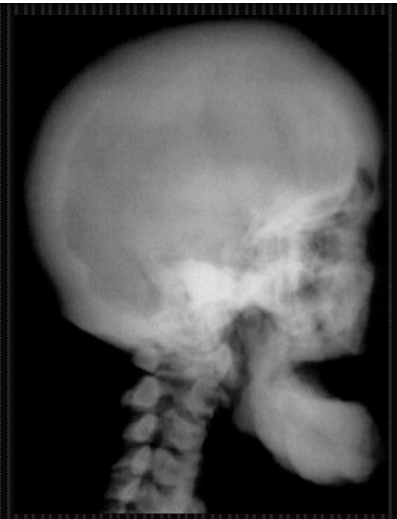
- Disorder characterized by increased bone density due to failure of osteoclast development or function
  - Comprised of the autosomal dominant (ADO) and more severe autosomal recessive (ARO) subtypes
- Common at sites of endochondral ossification, widening of metaphyses of long bones
- Complications include cranial nerve compression, brittle bones, and bone marrow failure in children

"**Sandwich vertebrae**" finding: vertebral endplate sclerosis seen on lateral XR with well-demarcated margins between peripheral sclerotic and central lucent bone

"**Bone within a bone**" finding: sclerosis along the inner margins of the lateral aspect of the vertebral bodies

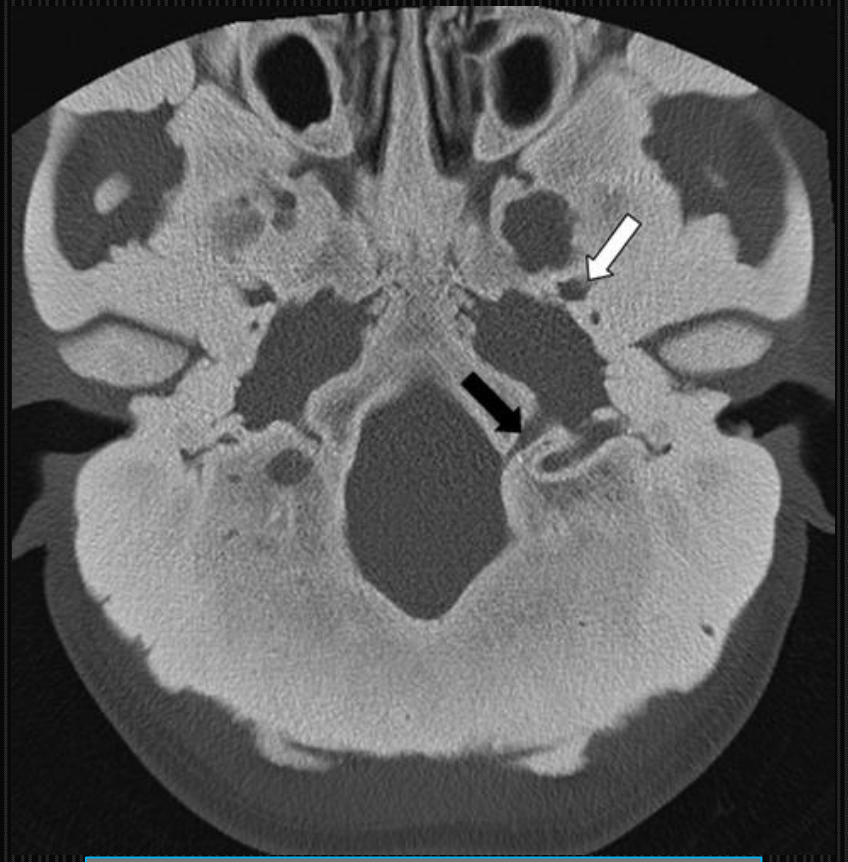
Differential Diagnosis: Van Buchem Disease (VBD)

https://www.iofbonehealth.org/osteoporosis-musculoskeletal-disorders/skeletal-rare-disorders/hyperostosis-corticalis-generalisata



https://www.nejm.org/doi/full/10.1056/NEJMicm1609871

The prominent mandible is a key feature of VBD



Severe hyperostosis of the skull, orbital walls, and petrous bones

- Also known as hyperostosis corticalis generalisata
- Autosomal recessive skeletal dysplasia characterized by generalized bone overgrowth, mainly in the skull and mandible
- Presents with large, overbearing mandible; less likely to manifest as circumferential skull enlargement, as seen in our patient
- Complications include facial nerve palsy, optic atrophy, and impaired hearing

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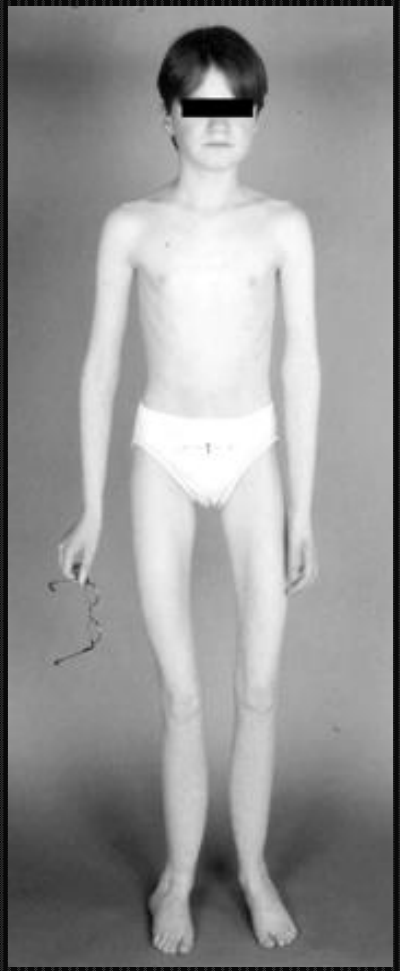
Treatment

Patient underwent lumbar puncture, which yielded an opening pressure of 41 mmH_2O. She was treated with 2000 mg of Diamox. Neurosurgery was consulted and recommended shunt placement for relief of cranial pressure. Patient was advised that repeat surgery may be necessary in the future if vision loss continued to worsen after shunt.

Discussion: Camurati-Engelmann Disease (CED)

- Also known as Progressive Diaphyseal Dysplasia (PDD)
- Rare, autosomal dominant bone dysplasia due to mutation of TGFB1
- Typically presents in childhood and almost always develops before the age of 30
- Manifests most commonly as pain in the extremities
- Involvement of the long bones is most common, although changes in the skull or pelvis are also frequent
  - Optic nerve compression is attributed to bony overgrowth of the orbit, optic canal stenosis, and increased ICP



https://jmg.bmj.com/content/43/1/1

Discussion: Was our imaging appropriate?

Variant 6: **Visual loss. Intraocular mass, optic nerve, or pre-chiasm symptoms. Initial imaging.**

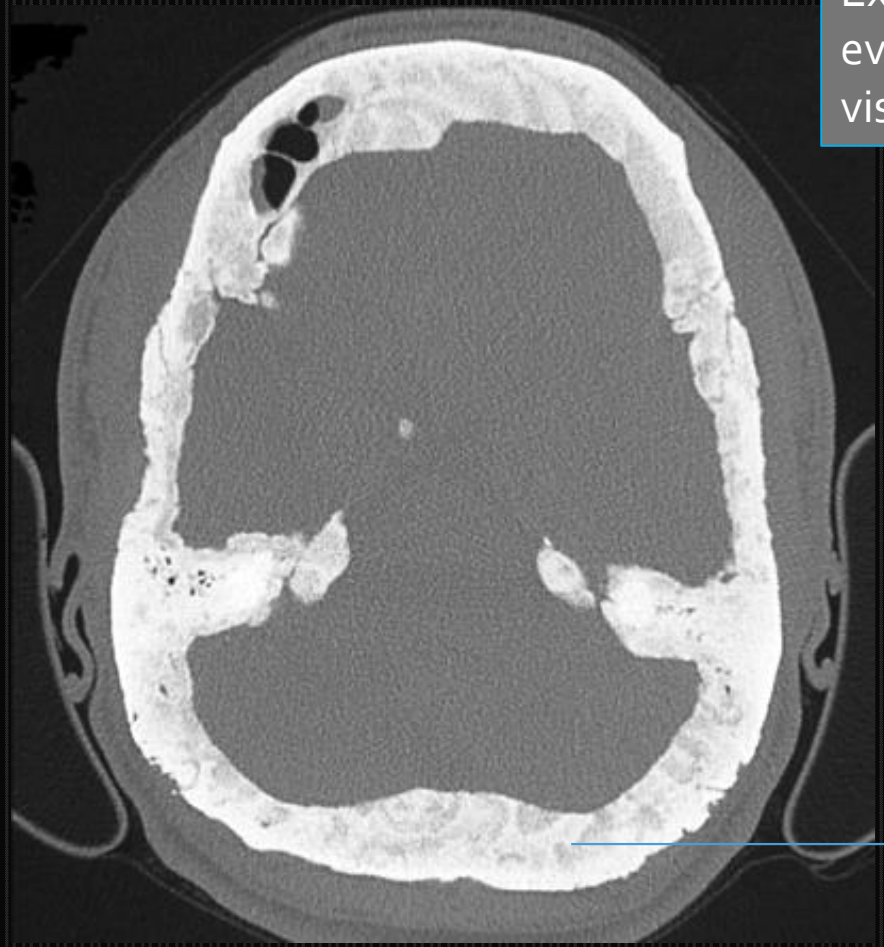
Procedure	Appropriateness Category	RRL
MRI orbits without and with IV contrast	Usually Appropriate	O
CT orbits with IV contrast	Usually Appropriate	☢☢☢
MRI orbits without IV contrast	Usually Appropriate	O
CT orbits without IV contrast	May Be Appropriate	☢☢☢
MRI head without and with IV contrast	May Be Appropriate	O
CT head with IV contrast	May Be Appropriate	☢☢☢
MRI head without IV contrast	May Be Appropriate	O
CT head without IV contrast	May Be Appropriate	☢☢☢
CTA head and neck with IV contrast	May Be Appropriate	☢☢☢
MRA head and neck without and with IV contrast	May Be Appropriate	O
MRA head and neck without IV contrast	May Be Appropriate	O
Arteriography cervicocerebral	Usually Not Appropriate	☢☢☢
CT head without and with IV contrast	Usually Not Appropriate	☢☢☢
CT orbits without and with IV contrast	Usually Not Appropriate	☢☢☢
X-ray orbit	Usually Not Appropriate	☢

Discussion: Was our imaging appropriate?

Variant 6: **Headache of skull base, orbital, or periorbital origin.**

Radiologic Procedure	Rating	Comments	RRL*
MRI head and orbits without and with IV contrast	8		O
MRI head and orbits without IV contrast	7		O
CT head and orbits without and with IV contrast	7		☢☢☢
CT head and orbits with IV contrast	7		☢☢☢
CT head and orbits without IV contrast	6		☢☢☢
MRA head without and with IV contrast	5		O
MRA head without IV contrast	5		O
CTA head with IV contrast	5		☢☢☢
Arteriography cervicocerebral	2		☢☢☢
Rating Scale: 1,2,3 Usually not appropriate; 4,5,6 May be appropriate; 7,8,9 Usually appropriate			*Relative Radiation Level

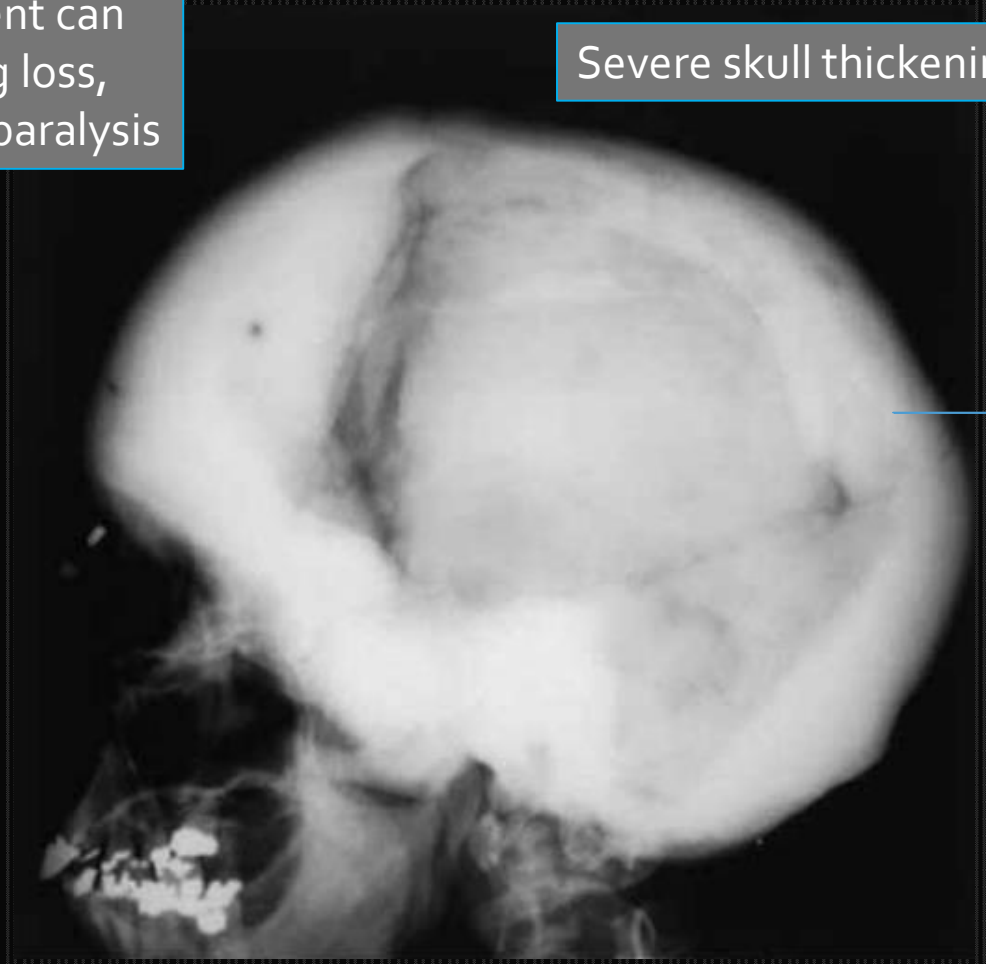
Discussion: Classic Radiographic Findings of CED



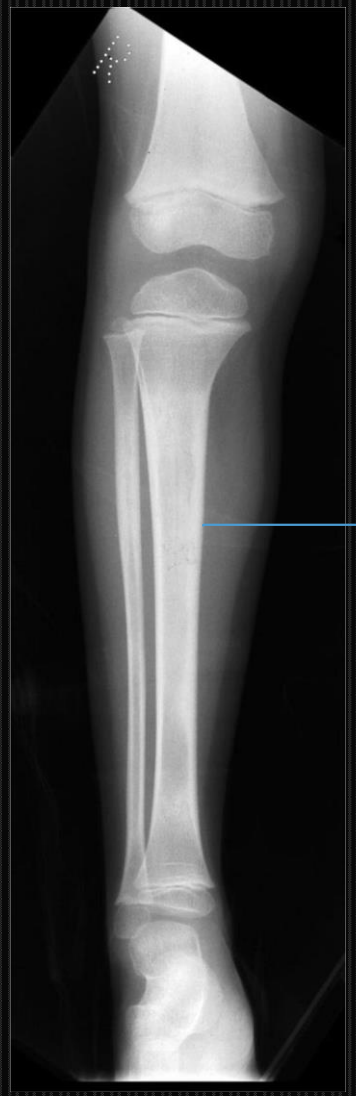
Extensive skull involvement can eventually lead to hearing loss, vision deficits, and facial paralysis

Axial CT (bone window) showing extensive sclerosis and thickening at the calvaria and petrous bones

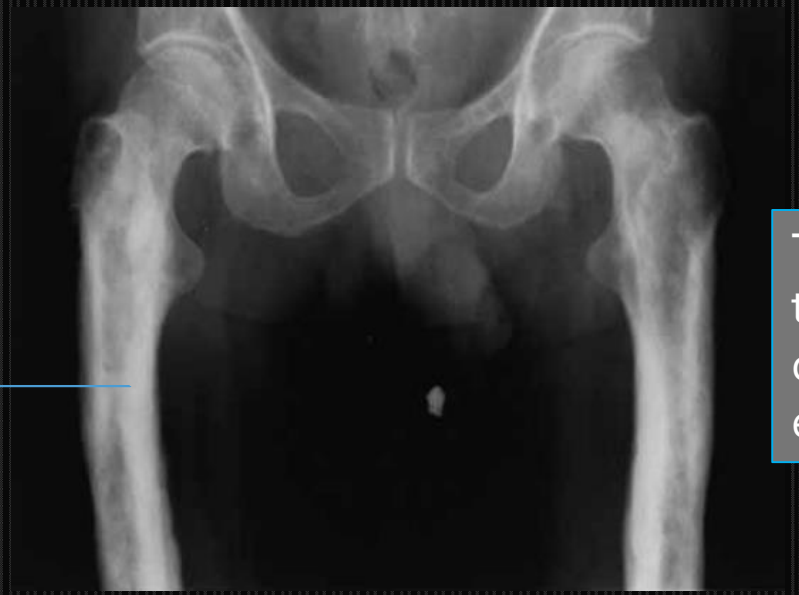
Severe skull thickening



Discussion: Classic Radiographic Findings of CED

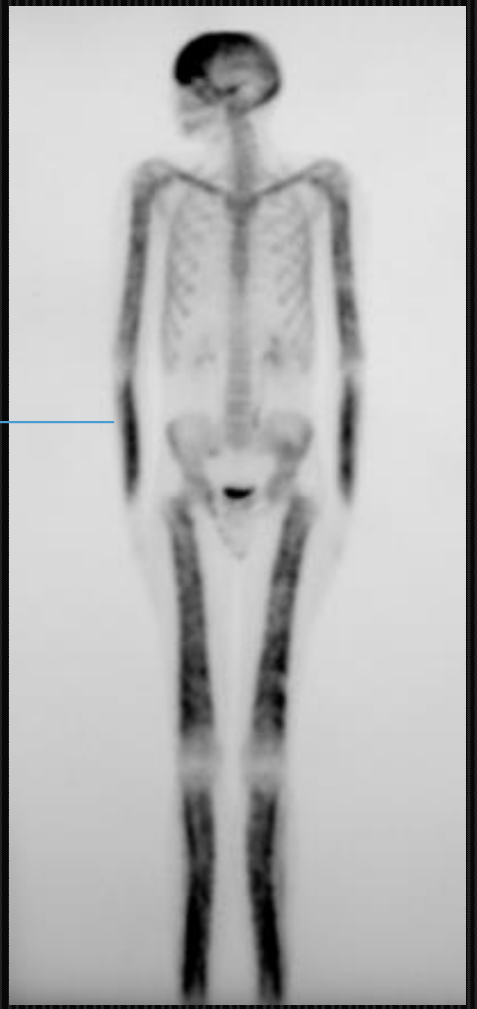


Endosteal cortical thickening in the diaphysis of long bones



Thickening may extend to the metaphyses but consistently spares the epiphyses

Whole body bone scintigraphy shows symmetric uptake in areas of increased osteoblastic activity



Discussion: Management of CED

- **Glucocorticoids** and **bisphosphonates** are the mainstay of pharmacologic therapy, although efficacy has yet to be established through randomized controlled trials
- **Surgical decompression** is the definitive method of management for progressive symptomatic skull base involvement
- **Gene therapy** may present an alternative method of treatment for some patients

https://www.massagetoday.com/mpacms/mt/article.php?id=14602

In Summary…

- Camurati-Engelmann Disease (CED) is a rare, progressive bone dysplasia that primarily affects the long bones
- Differential diagnosis includes fibrous dysplasia, osteopetrosis, Paget's Disease, sclerosing metastases, and Van Buchem Disease
- Progression of CED can cause disabling neurologic symptoms and cranial nerve deficits

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