RADY401 Case Presentation

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Ed. John Lilly, MD
A 26-year-old female presents with headache and progressive bilateral vision loss
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
Diffuse thickening and sclerosing of the bones of the calvarium, skull base, and upper cervical spine

Corpus callosum
Lateral ventricle
Thalamus

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)

Fourth ventricle
Pons
Cerebellum
Medulla
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.
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.
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
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.

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

Characteristic “cotton-wool” thickening is due to irregular areas of sclerosis

Presented 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.

Multiple round, well-circumscribed sclerotic lesions of the calvarium on CT.
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
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

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

The prominent mandible is a key feature of VBD
Patient underwent lumbar puncture, which yielded an opening pressure of 41 mmH2O. 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.
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


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

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<tr>
<th>Procedure</th>
<th>Appropriateness Category</th>
<th>RRL</th>
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<tr>
<td>MRI orbits without and with IV contrast</td>
<td>Usually Appropriate</td>
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<td>CT orbits with IV contrast</td>
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<td>Arteriography cervicocerebral</td>
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<td>X-ray orbit</td>
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**Variant 6:** Headache of skull base, orbital, or periorbital origin.

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<tr>
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*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.
Endosteal cortical thickening in the diaphysis of long bones

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

Thickening may extend to the metaphyses but consistently spares the epiphyses

Discussion: Classic Radiographic Findings 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.
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.


