CASE 4
13 Y/O GIRL

Brief history

- **Past history**
  Abnormal craniofacial appearance

- **Chief complain**
  Recent developing hearing impairment

Imaging studies

Head CT (no C) (2020.08.04)

Imaging findings

- ✓ **Hyperostosis with increased osteosclerosis** involving multiple craniofacial bones, also with hyperostosis of multiple cervical vertebrae
- ✓ **Hyperostotic deformity of the skull base and temporal bones** with concomitant absence of bilateral foramen spinosum, stenosis of bilateral interal acoustic canals, and stenosis of the left jugular foramen
- ✓ **Chronic otitis media, left**

Differential Diagnoses

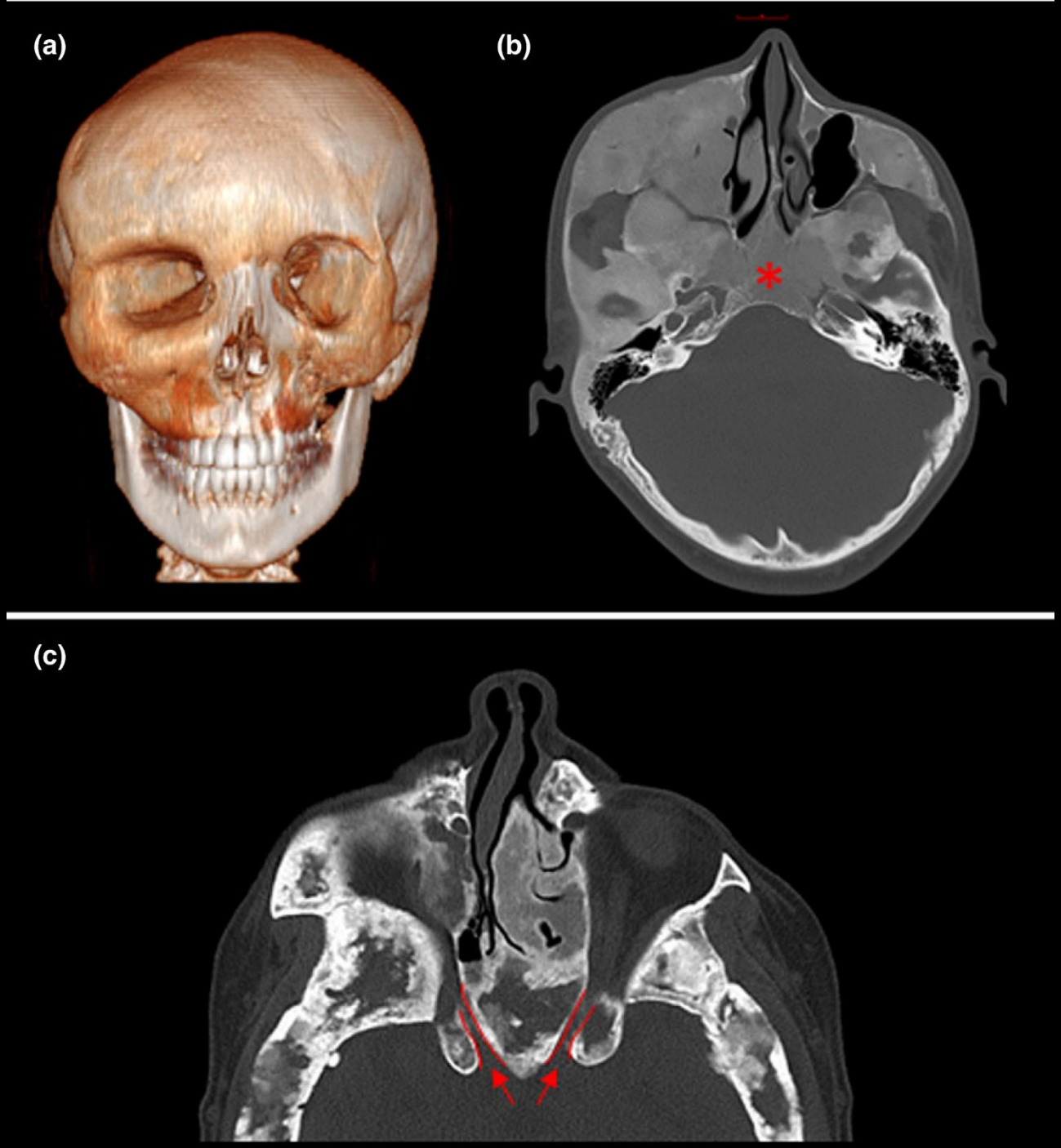
- **Fibrous Dysplasia/McCune-Albright syndrome (FD/MAS)**
- Paget disease
- Jaffe-Campanacci syndrome
- Garré sclerosing osteomyelitis

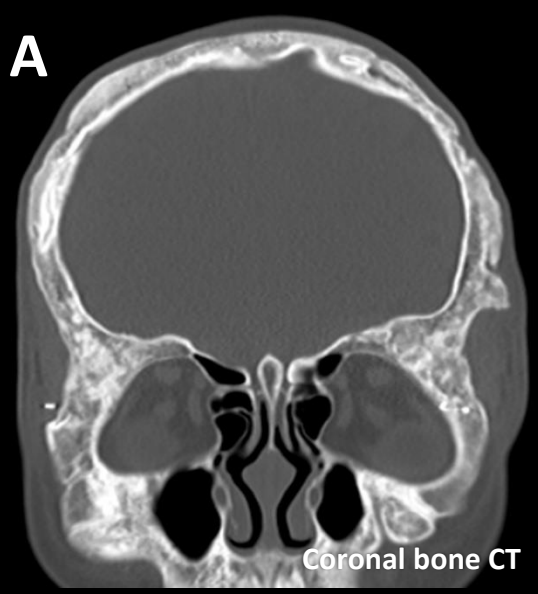
McCune-Albright syndrome (MAS)

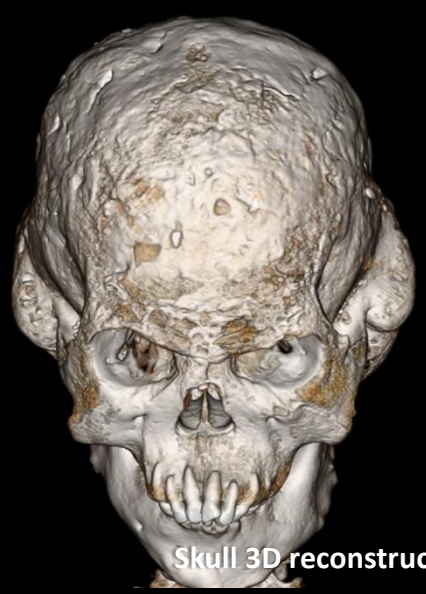
- Rare genetic disorder resulting from postzygotic somatic mutations in the **GNAS gene** and is usually diagnosed during **childhood or adolescence**

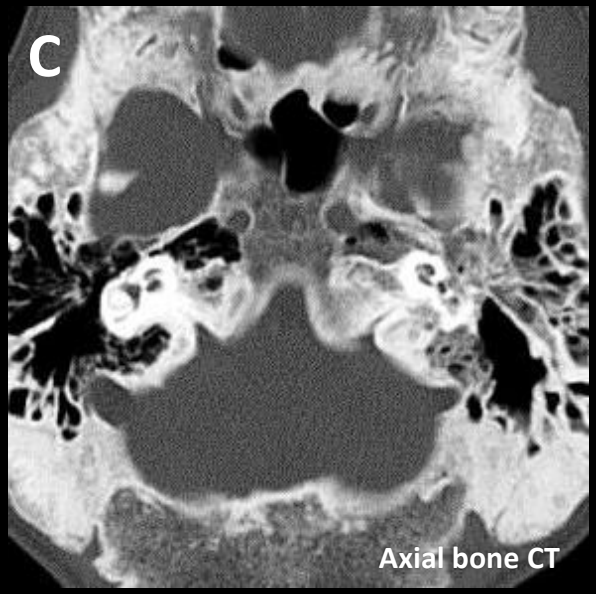
- Clinical triad:
  - **Monostotic or polyostotic fibrous dysplasia**
  - **Precocious puberty**
  - **Café-au-lait skin macules**

- Imaging features:
  - **Uni- or multifocal expansile bone lesion(s)** with variable fibrous stromal matrix (ground glass/mixed/cystic)
  - **Craniofacial involvement** can lead to calvarial deformity and facial disfigurement ("**Leontiasis ossea**" is severe cases)
  - **Entrapment syndromes** (vascular or neurological)


(a)
(b)
(c)


A
Coronal bone CT


B
Skull 3D reconstruction


C
Axial bone CT