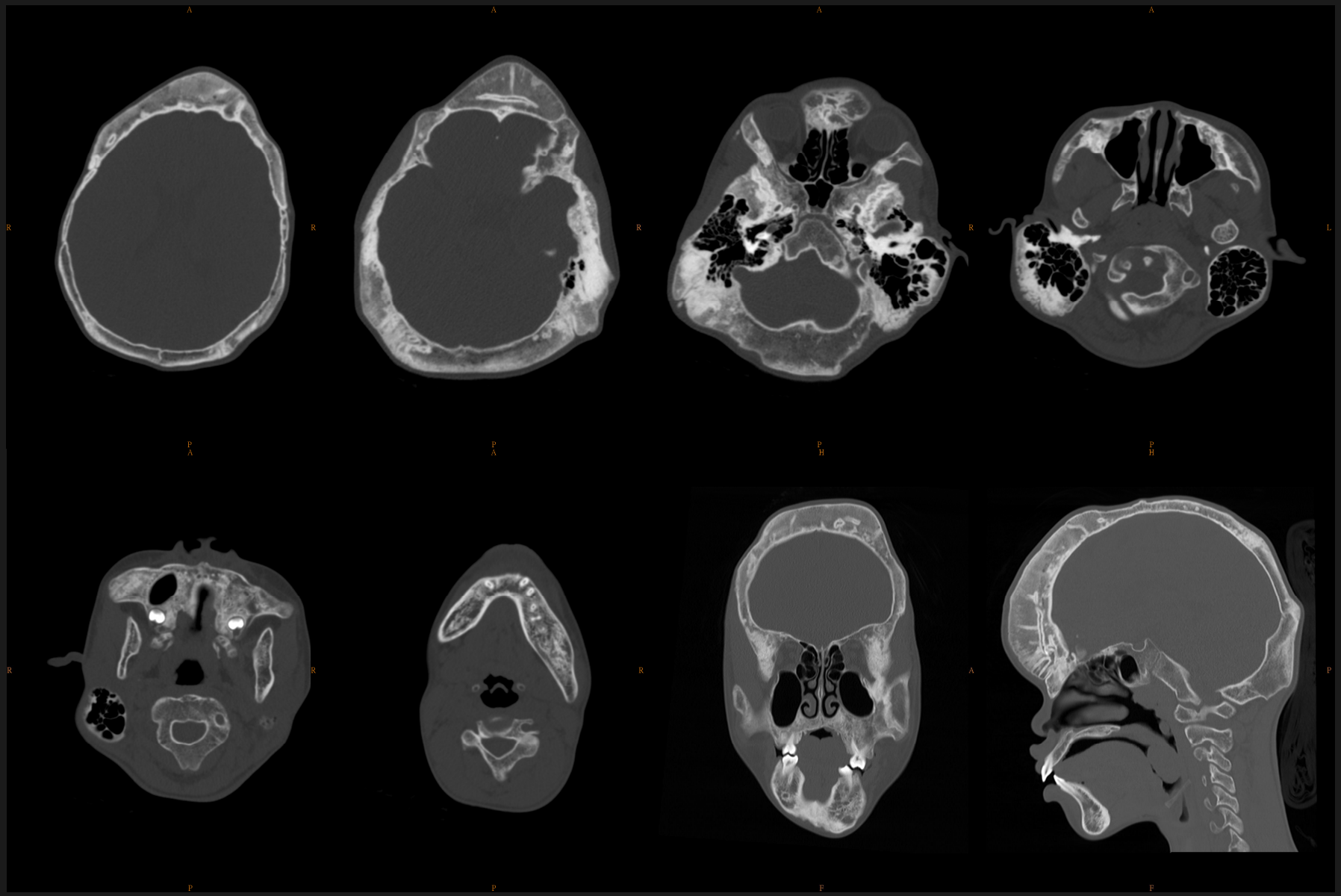
CASE 4

16572247

- 10 y/o, girl
- Chief complaint:
  - abnormal head shape since birth with bulging forehead and protrusion of upper gum


A
A
A
A
R
R
R
R
L
P
A
P
A
P
H
P
H
R
R
R
R
P
P
F
F
A
P

Differential Diagnosis

- Monostotic FD
- Polyostotic FD without MAS
- Jaffe-Campanacci syndrome
  - Non-ossifying fibromas, axillary freckling, and café au lait skin lesions, without neurofibromas
- Caffey disease: Usually < 5 months of age
  - Acute onset fever + hot, tender swelling of bones
- Cherubism
  - Familial, symmetric, bilateral fibro-osseous lesions of jaw
- Garré sclerosing osteomyelitis
  - Bony expansion, heterogeneous sclerotic pattern
- Mazabraud Syndrome
  - Polyostotic FD + intramuscular myxoma
- Craniometaphyseal Dysplasia
  - Mutations in transmembrane protein ANK on Chr 5p
  - Hyperostosis and sclerosis of craniofacial bones → facial distortion, cranial nerve compression
  - Abnormal modeling of long bone metaphyses, paranasal "bossing"
- Paget disease
  - Cotton wool CT appearance
- Hyperostosis frontalis interna
- Cemento-ossifying fibroma

Diagnosis: McCune-Albright syndrome

- McCune-Albright syndrome (MAS) is a genetic disorder characterized by the association of:
  - endocrinopathy: precocious puberty
  - polyostotic fibrous dysplasia
  - cutaneous pigmentation: coast of Maine 'cafe au lait' spots
- Genetics: MAS results from a sporadically occurring somatic *GNAS* gene mutation
- Age: FD < 10 years (~ 60%), > 10 years (40%)
- Gender: MAS: M < < F

Fibrous Dysplasia

Monostotic form

- ribs: 28%, most common
- proximal femur: 23%
- tibia
- craniofacial bones: 10-25%
- humerus

Polyostotic form

- often unilateral and monomelic: one limb
- femur: 91%
- tibia: 81%
- pelvis: 78%
- foot: 73%
- ribs
- skull and facial bones: 50%
- upper extremities
- lumbar spine: 14%
- clavicle: 10%
- cervical spine: 7%

Imaging

- Best diagnostic clues: Expanded ground-glass bone in child with precocious puberty and skin lesions
  - Intact cortex; lose of normal corticomedullary differentiation; spared inner table; unclear margin
- Locations in H&N: Skull, skull base, or facial bones
  - Bilateral and asymmetric common
  - The anterior craniofacial bones are more frequently involved than more lateral or posterior portions
  - Sphenoid, frontal, maxillary, ethmoid bones > occipital, temporal bones.
  - Extracranial involvement is rare

Signs/symptoms

- MAS **café au lait** skin lesions
- Precocious puberty
- Symptoms depend on location of FD lesion
  - Bone deformity, pain
  - Pathologic fractures primarily in childhood, peak between 6 and 10 years
  - Hearing loss with temporal bone involvement
  - Proptosis, optic canal narrowing may → decreased vision
  - Cranial nerve impingement