CASE PRESENTATION

MENTOR: DR G C PATIL
KMCRI,HUBBALLI.

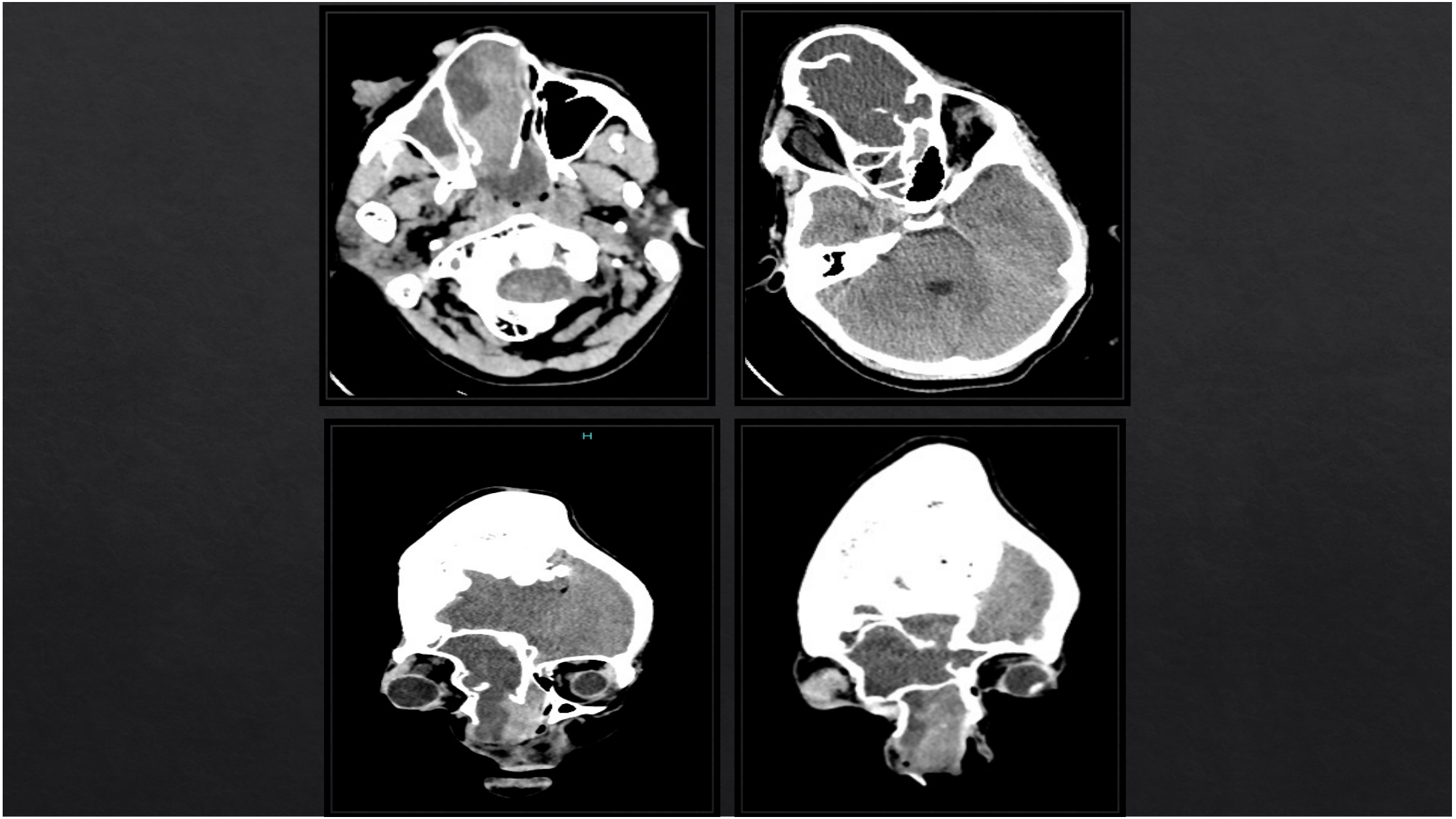
Clinical History

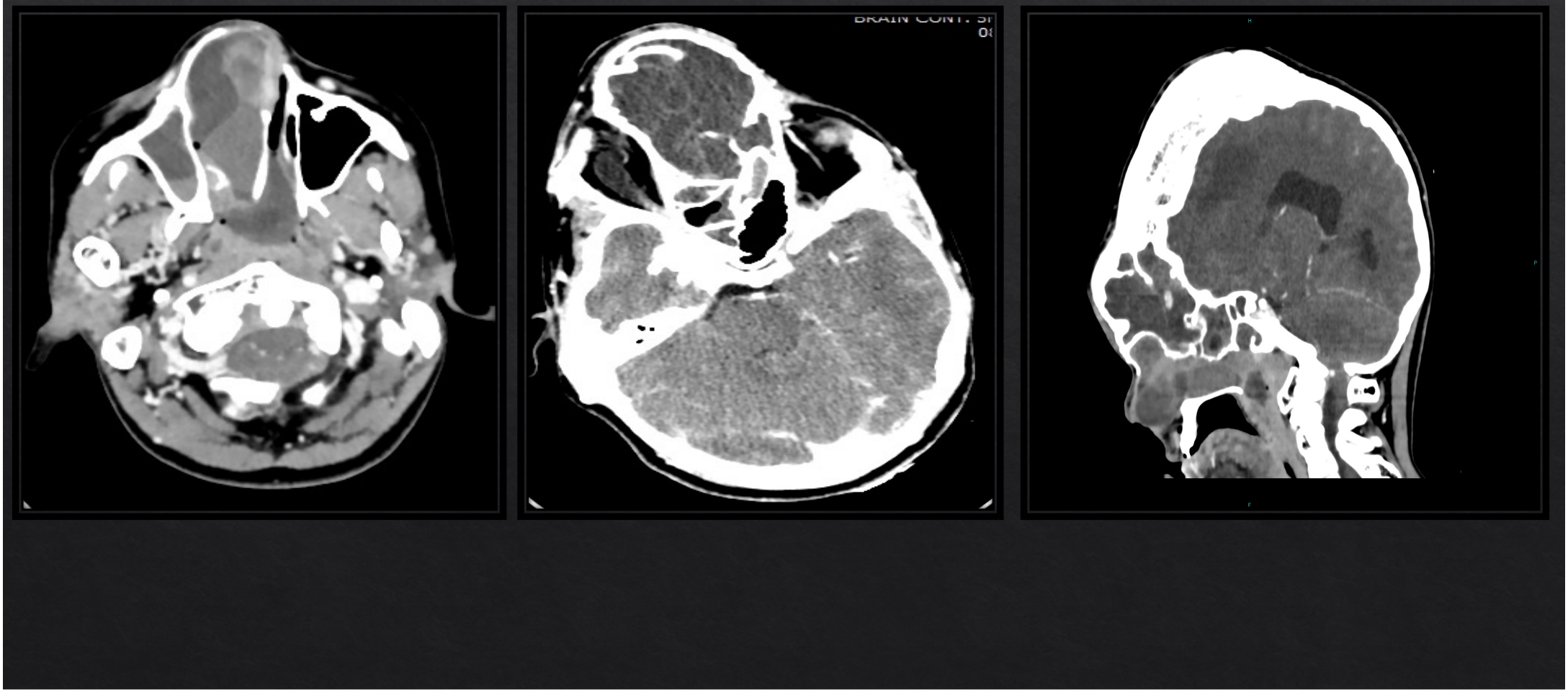
- 18year old female,
- Presenting with swelling over forehead since birth
- C/o swelling and mass like sensation in right nasal cavity with progressive breathing difficulty and mouth breathing - since 3weeks.
- No history of epistaxis/trauma.

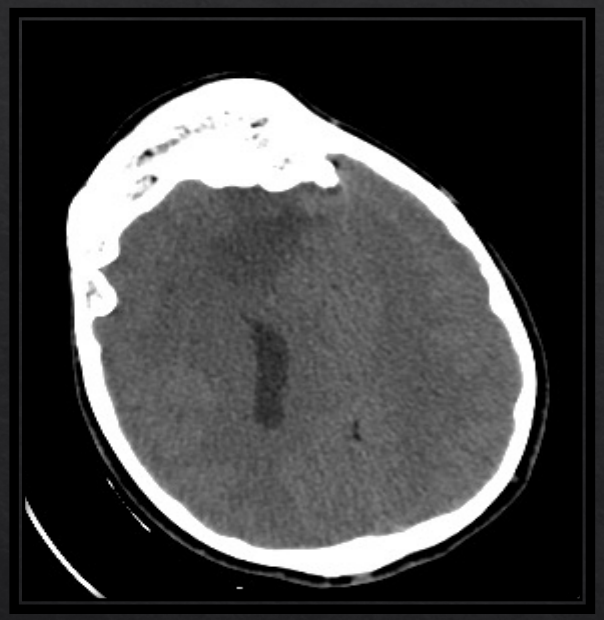
- Also c/o GTCS since 1 year- on treatment.
- Hyperpigmentation of skin on right half of the body since birth.
- Menstrual history- menarche at 17 years of age, irregular cycles.
- H/o delayed dentition and malocclusion of few of the maxillary tooth.
- No other developmental delay.
- No comorbidities.
- No mental retardation.

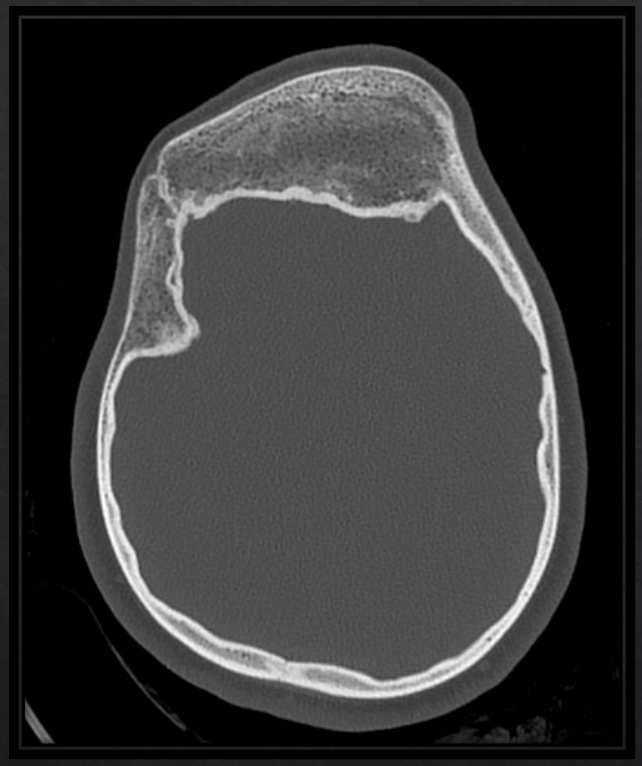
Clinical examination

- Dysmorphic face and thoracolumbar dextro-scoliosis and kyphosis.
- Hyperpigmented macule involving the right side of face , neck, body and right lower limb.







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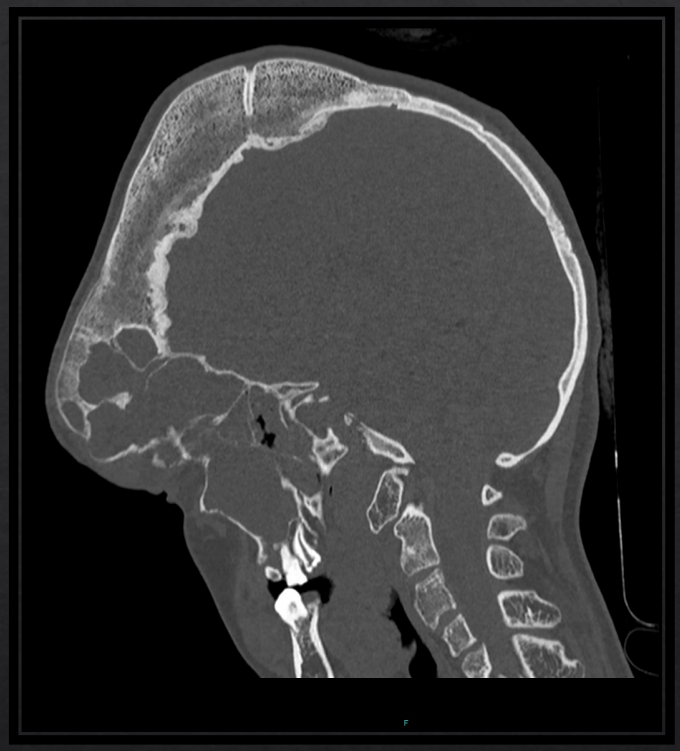

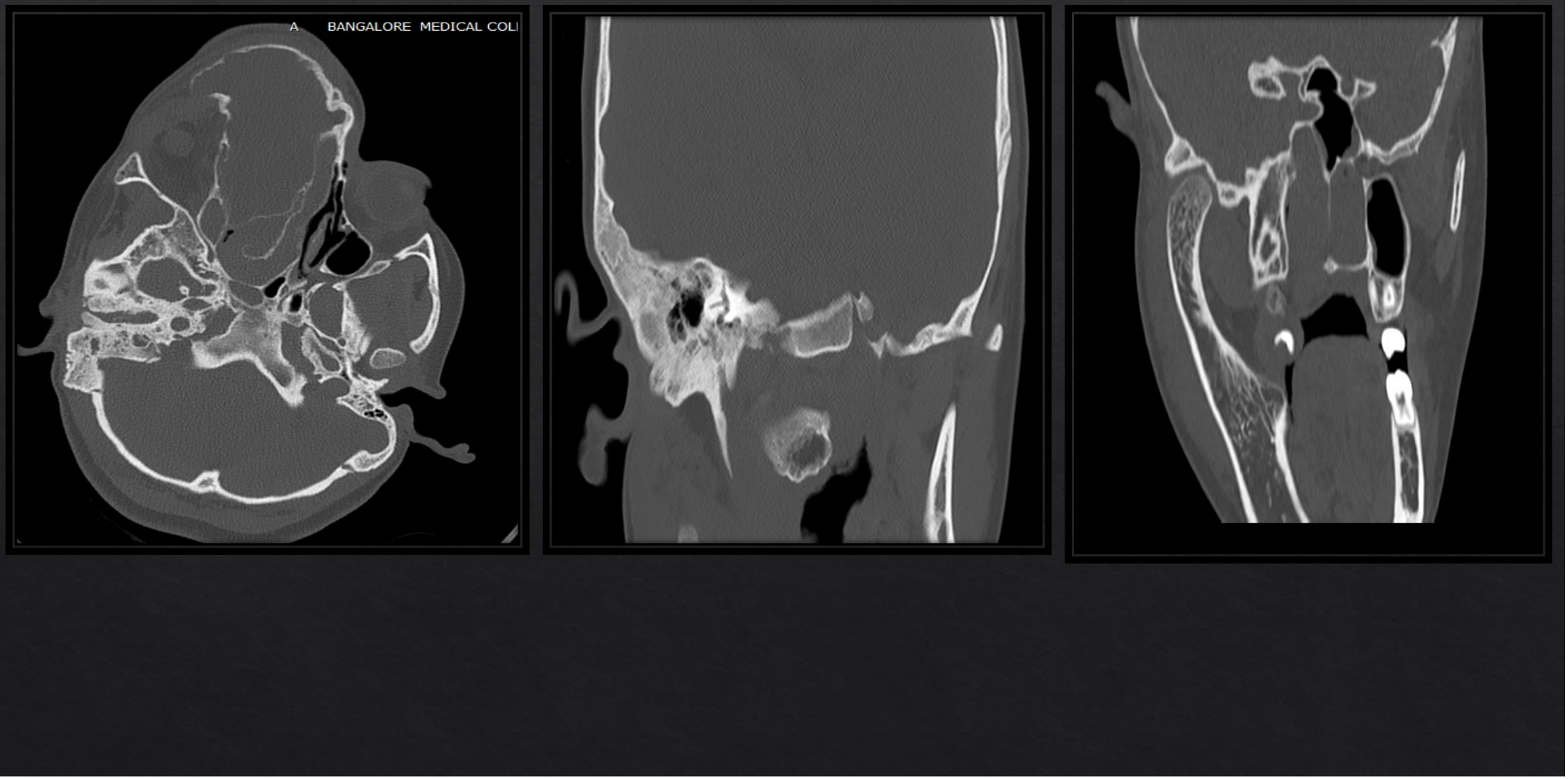
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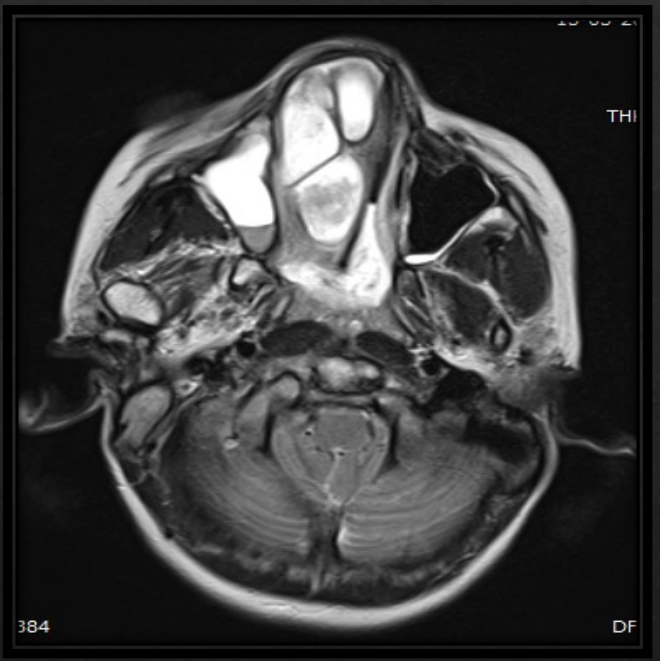
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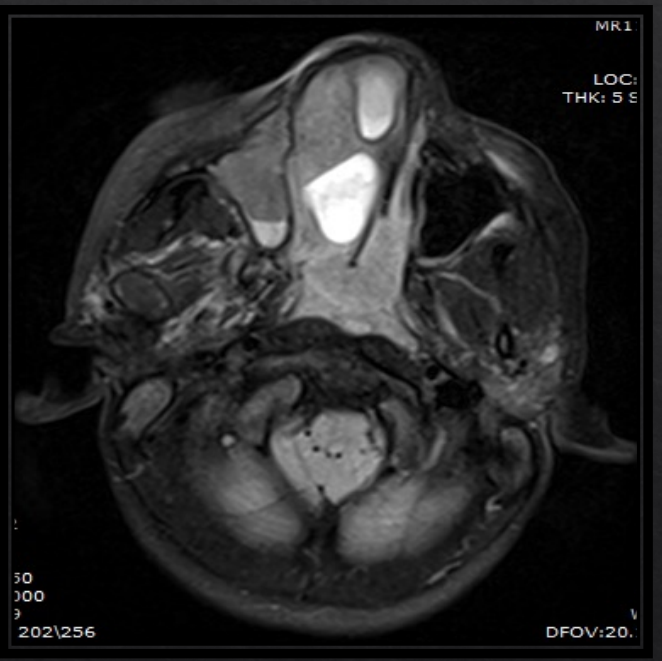
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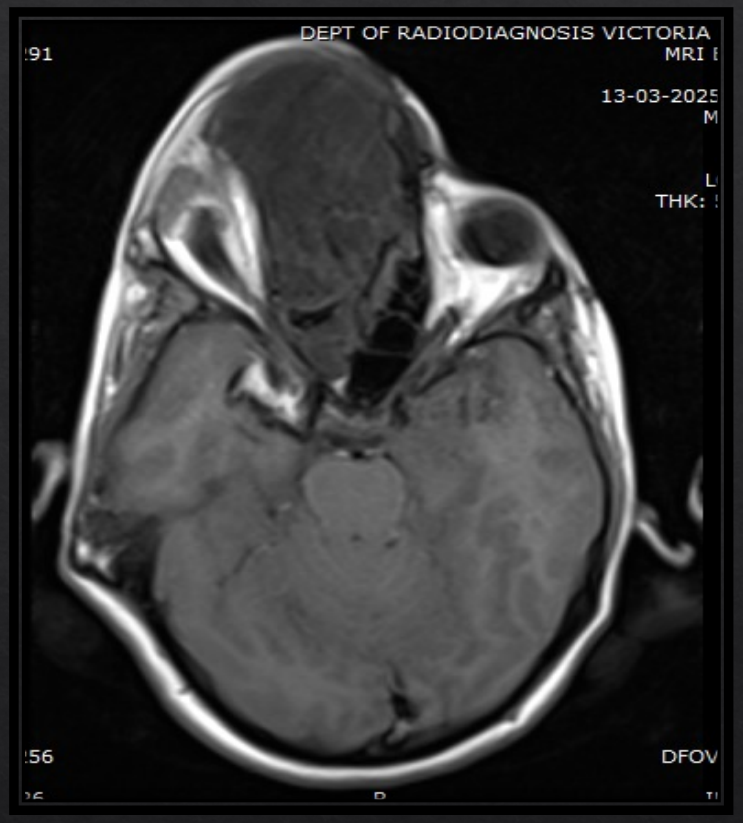
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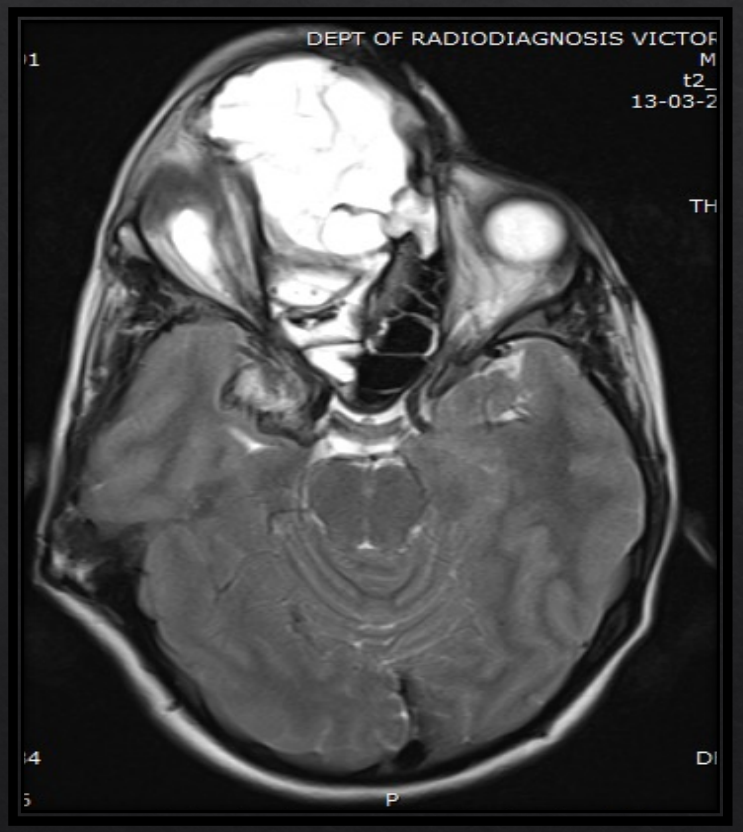
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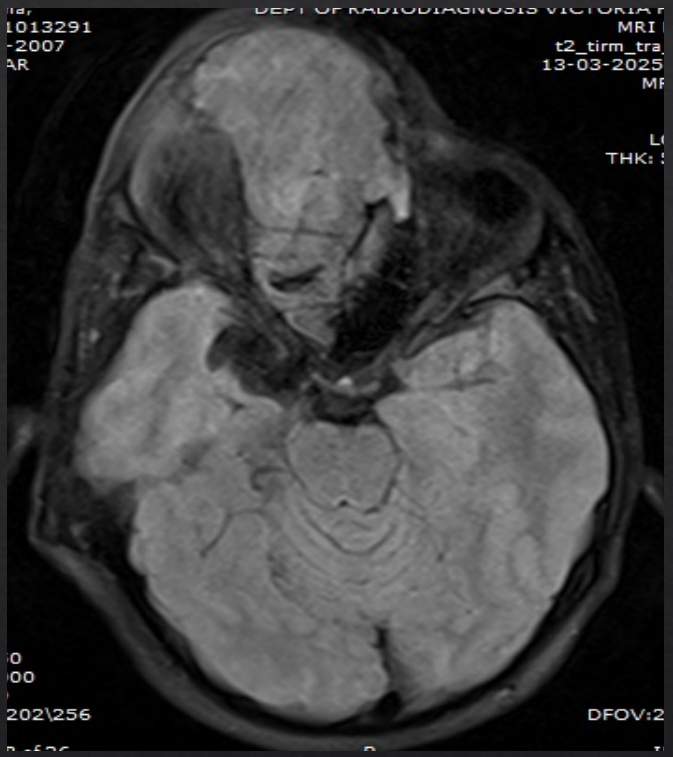
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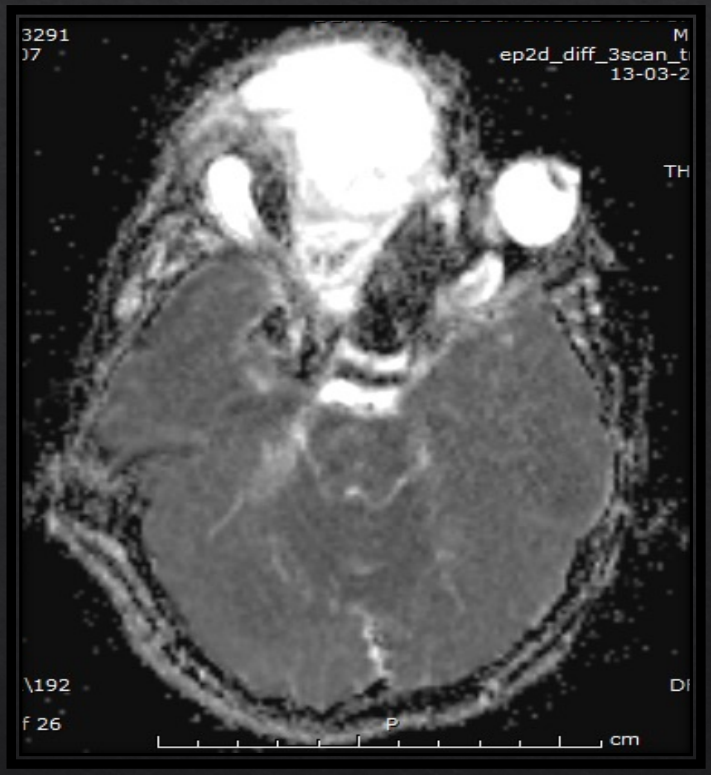
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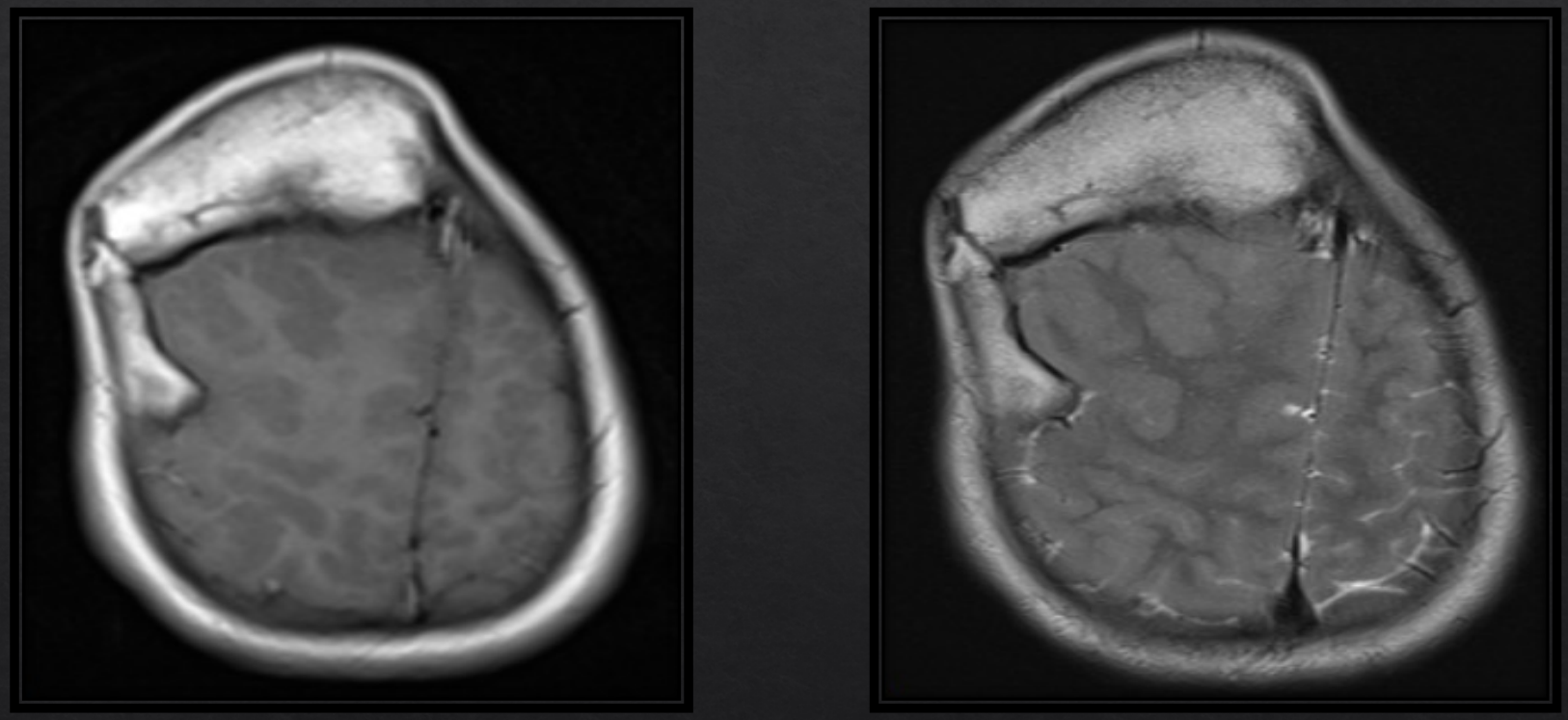
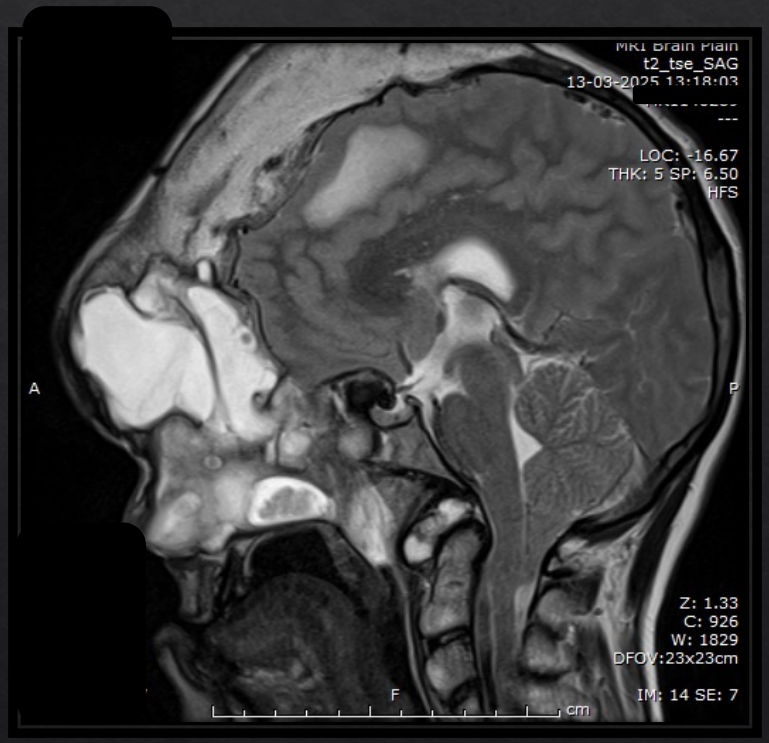
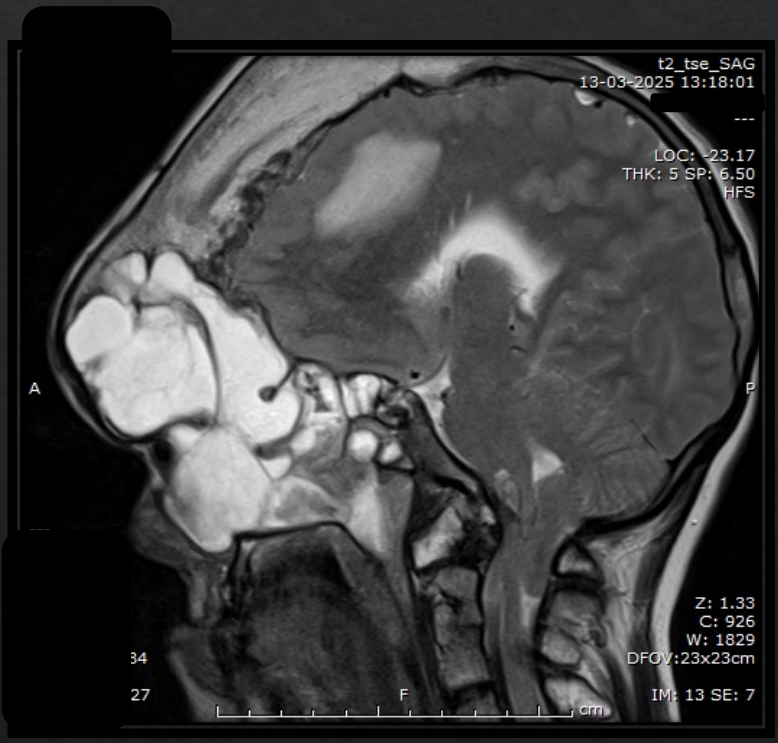



MRI Brain Plain
t2_tse_SAG
13-03-2025 13:18:03

LOC: -16.67
THK: 5 SP: 6.50
HFS
A
P
Z: 1.33
C: 926
W: 1829
DFOV:23x23cm
F
cm
IM: 14 SE: 7


t2_tse_SAG
13-03-2025 13:18:01

LOC: -23.17
THK: 5 SP: 6.50
HFS
A
P
Z: 1.33
C: 926
W: 1829
DFOV:23x23cm
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27
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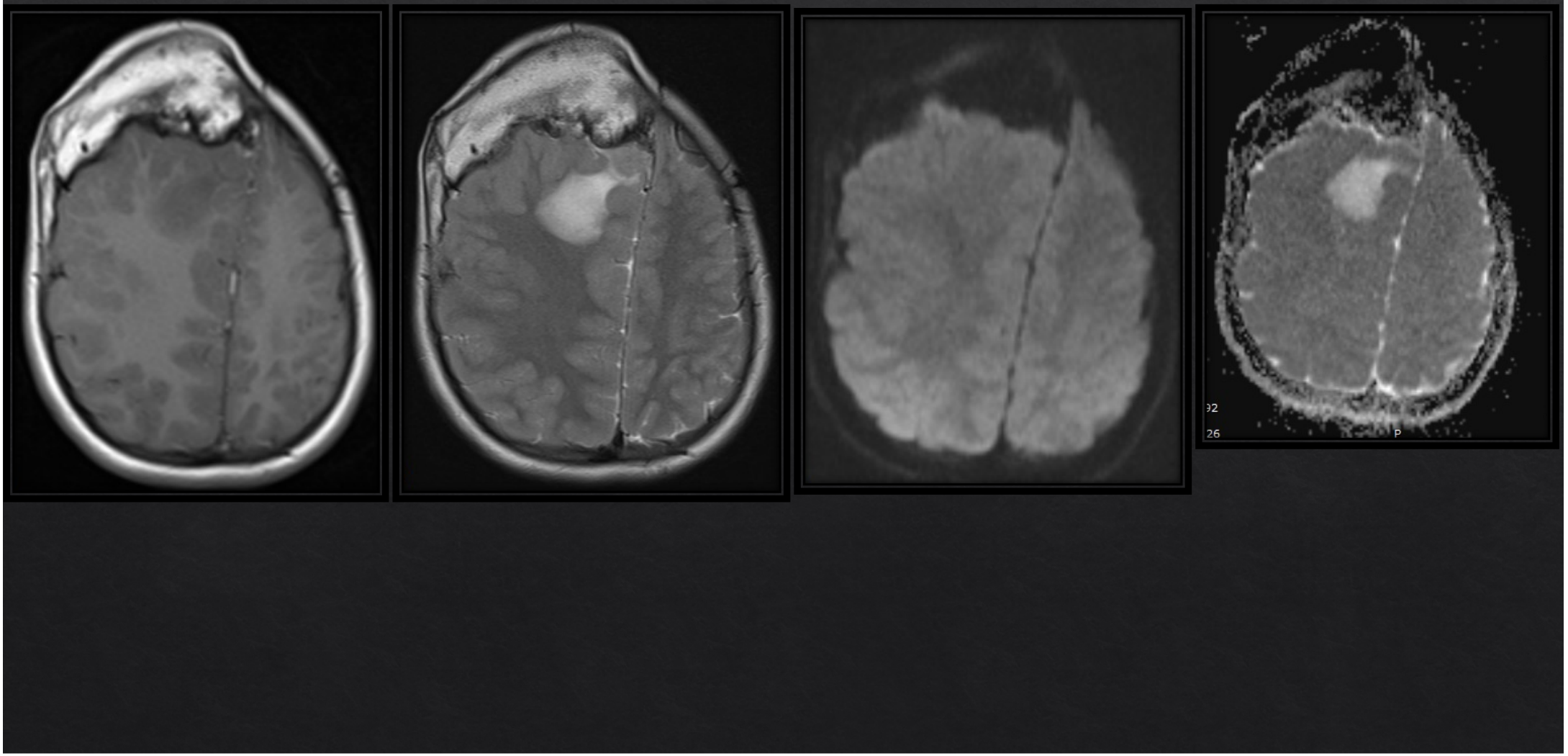
cm
IM: 13 SE: 7



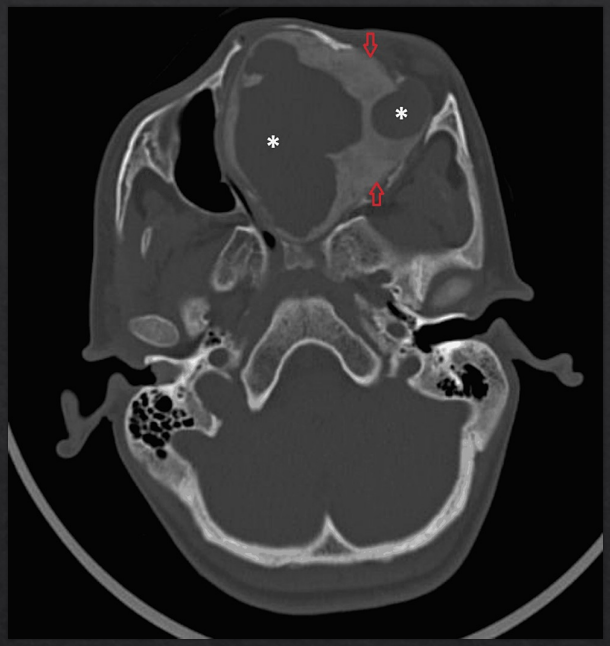
Summary

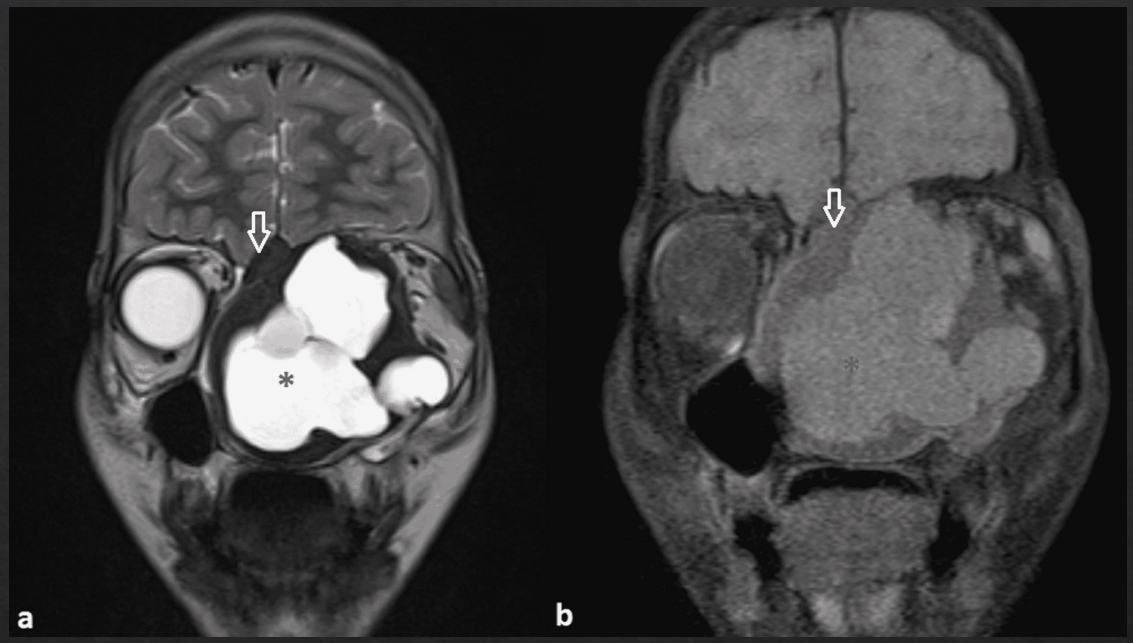
- Multiloculated cystic lesion with fluid-fluid levels in right nasal cavity involving ipsilateral maxillary, sphenoid and frontal sinuses extending into posterior choanae causing significant airway narrowing.
- Craniofacial Fibrous cortical dysplasia involving frontal bone, frontal sinuses right hemi mandible , right temporal bone and right zygomatic bone.
- Right proptosis and stenosed optic canal.
- Arnold Chiari malformation.
- Right Parafalcine meningioma and falx lipomas.

- **Multiloculated cystic lesion with fluid-fluid levels in right nasal cavity extending into ipsilateral maxillary, sphenoid and frontal sinuses.**

Differentials are

- Aneurysmal bone cyst in the background of fibrous dysplasia
- Giant cell reparative granuloma of the nasal cavity





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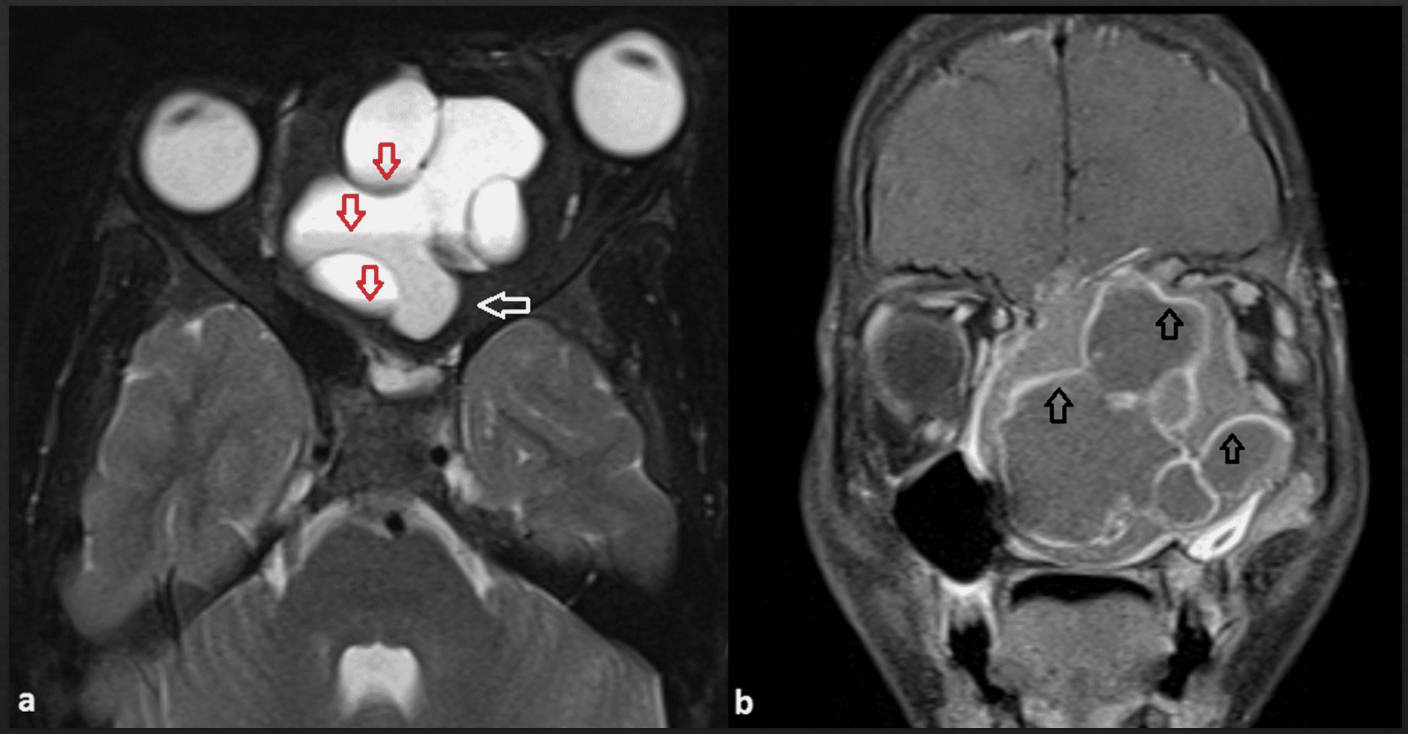
Sharma G, Sharma P, Shankar S, et al. (April 25, 2022) Fibrous Dysplasia With Aneurysmal Bone Cyst Presenting as Sinonasal Mass. Cureus 14(4): e24485. doi:10.7759/cureus.24485



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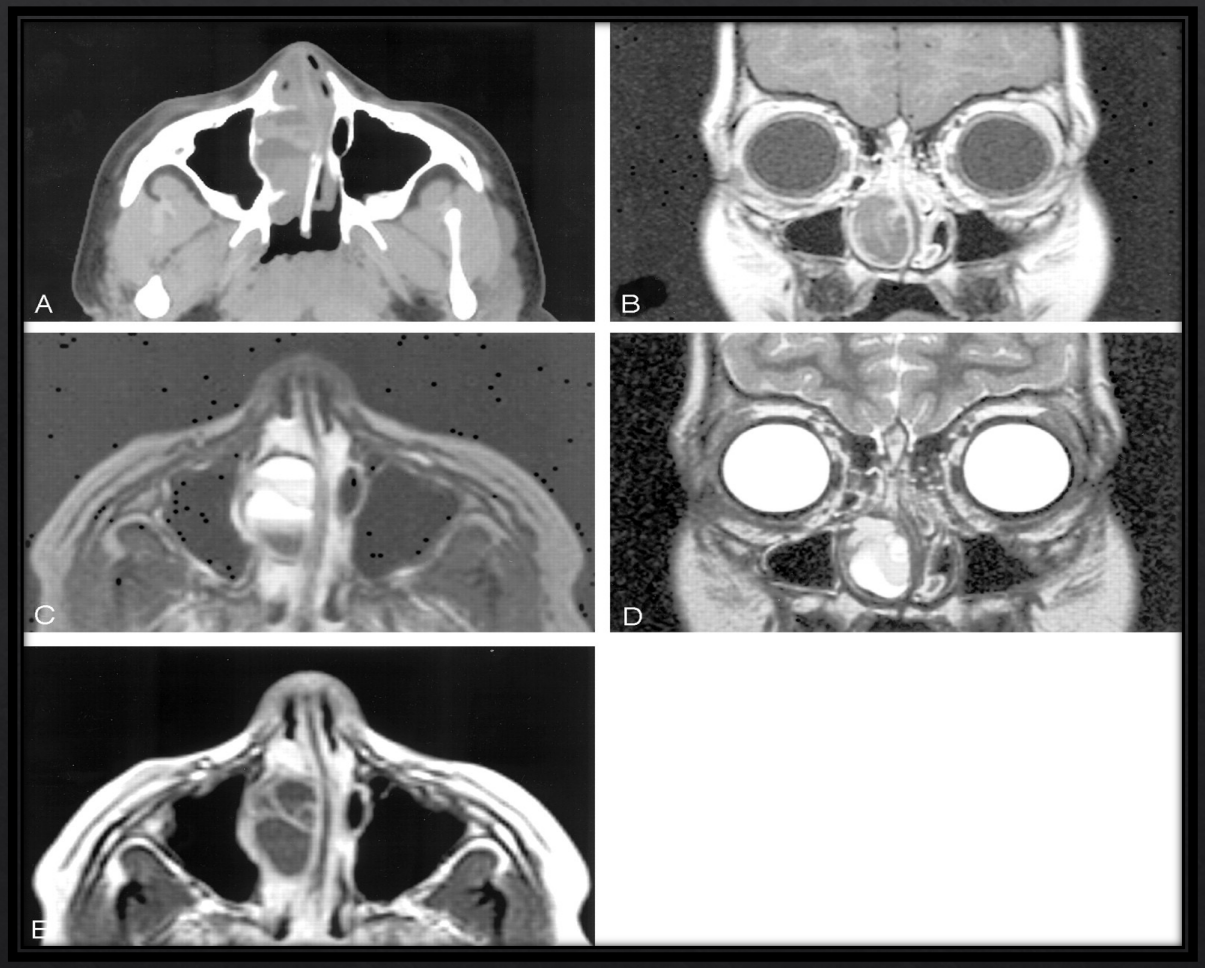
Giant cell reparative granuloma of the nasal cavity

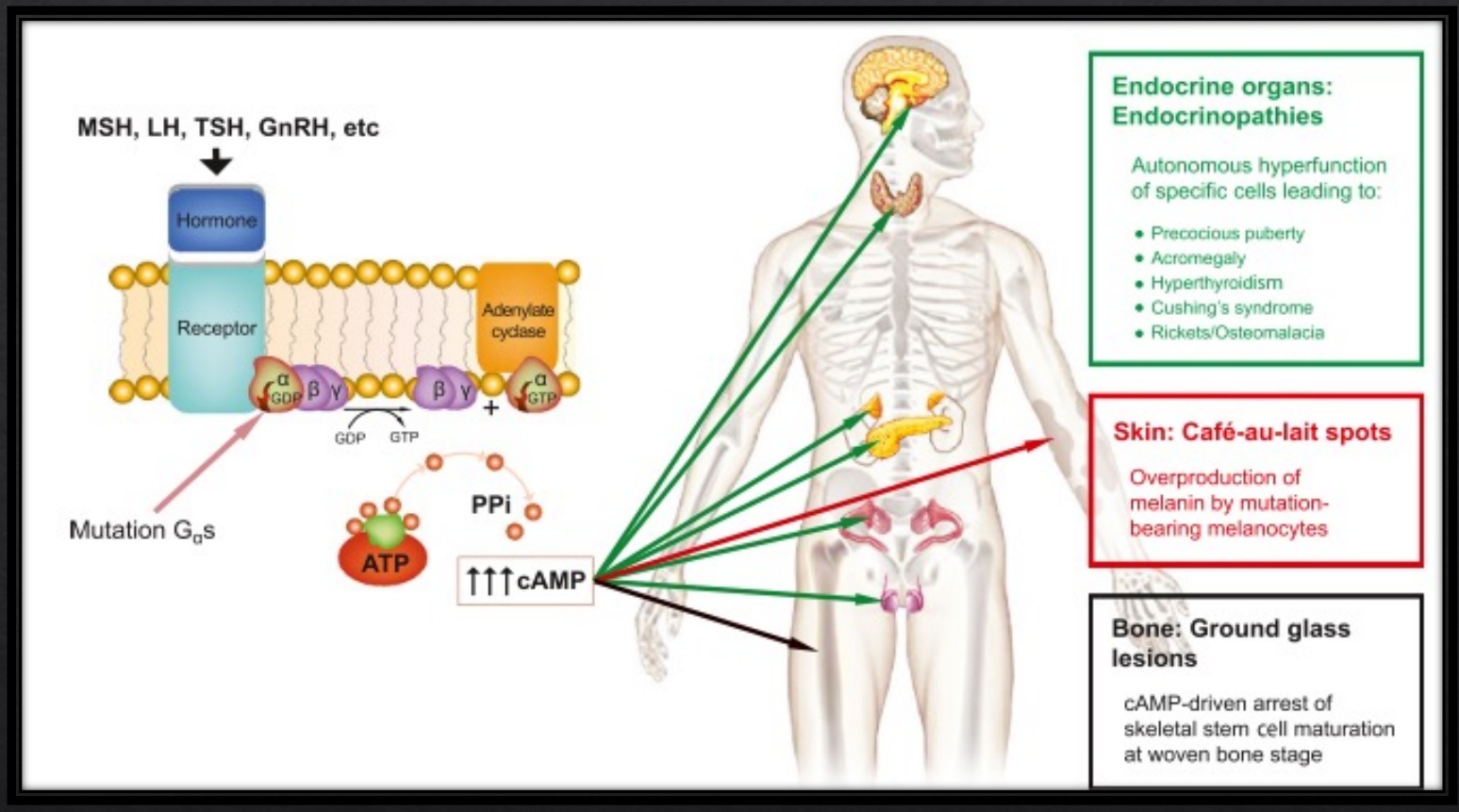


Morris JM, Lane JI, Witte RJ, Thompson DM. Giant cell reparative granuloma of the nasal cavity. AJNR Am J Neuroradiol. 2004 Aug;25(7):1263-5. PMID: 15313721; PMCID: PMC7976521.

Fibrous dysplasia

- Mono-ostotic FD- FD of single bone
- Poly-ostotic FD- FD of multiple bones
- McCune-Allbright syndrome
- Mazabraud syndrome- Myxomas- intramuscular

- FD arises sporadically, and there are no confirmed cases of vertical transmission.
- Morphological changes –related to post-zygotic mutations of the α-subunit of the G-s stimulatory protein (GNAS mutations).
- Leading to activation and inappropriate cyclic adenosine monophosphate (cAMP) overproduction


MSH, LH, TSH, GnRH, etc
Hormone
Receptor
Adenylate cyclase
α GDP
β γ
β γ
α GTP
+
GDP
GTP
Mutation $G_\alpha s$
PPi
ATP
↑↑↑cAMP
Endocrine organs: Endocrinopathies
Autonomous hyperfunction of specific cells leading to:
• Precocious puberty
• Acromegaly
• Hyperthyroidism
• Cushing's syndrome
• Rickets/Osteomalacia
Skin: Café-au-lait spots
Overproduction of melanin by mutation-bearing melanocytes
Bone: Ground glass lesions
cAMP-driven arrest of skeletal stem cell maturation at woven bone stage

Monostotic FD

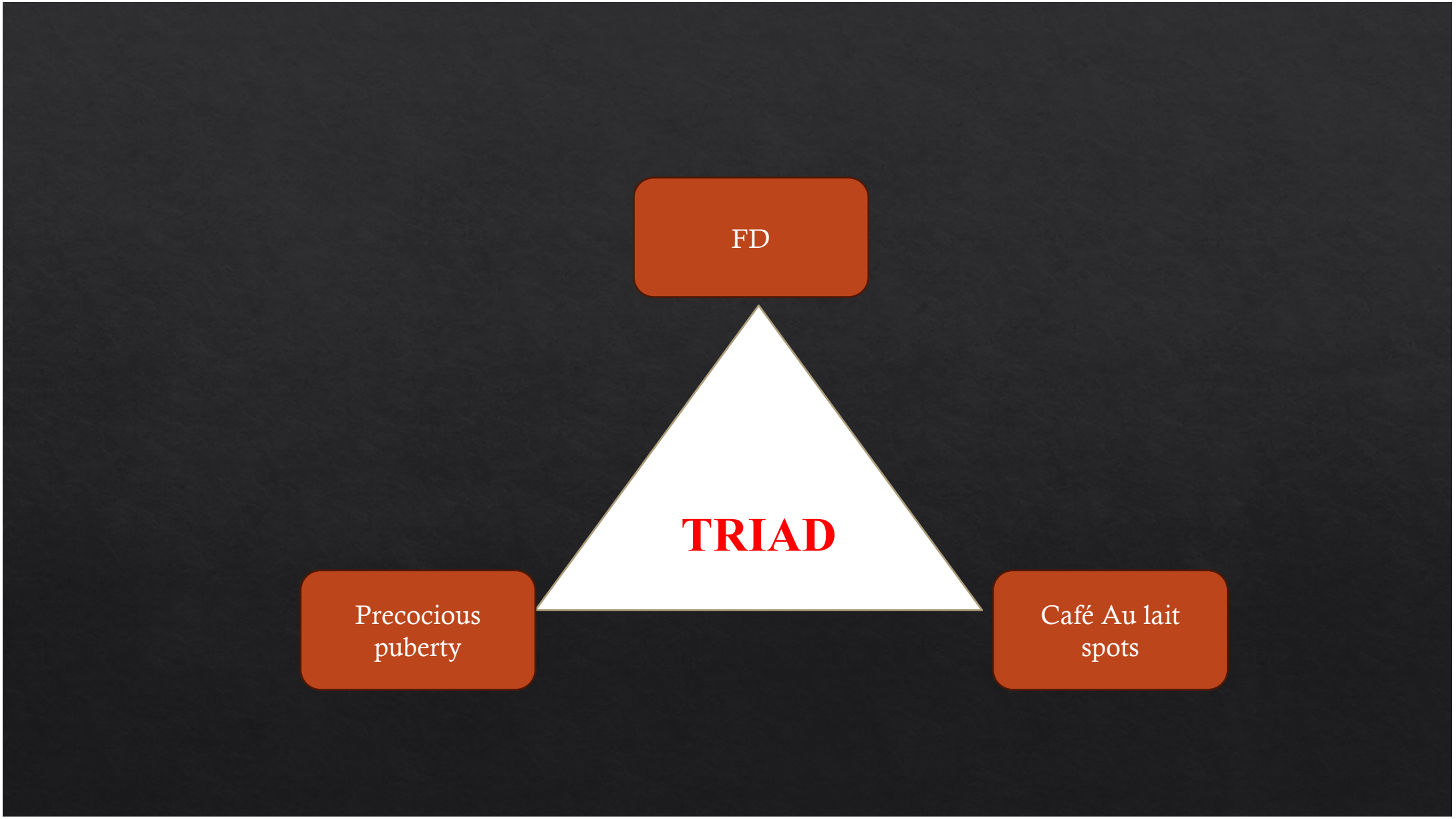
- The most common location - is rib, skull and femur.
- FD lesions are not static morphological abnormalities.
- They are characterized by age-related , histological, radiological and clinical transformations.
- In early childhood, lesions are metabolically active and expand during linear growth.
- The lesions typically become static in size after puberty and metabolic activity may decrease throughout adulthood.

Radiographic features

- Ground-glass opacities: 56%.
- Homogeneously sclerotic: 23%.
- Cystic: 21%.
- Well-defined borders
- Expansion of the bone, with intact overlying bone
- Endosteal scalloping may be seen

McCune-Albright syndrome

- Approximately 2–3% of patients with FD have extra-skeletal disease, known as McCune-Albright syndrome.
- Usually polyostotic, can present as monostotic FD.
- Aggressive course than polyostotic FD.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Precocious puberty
Café Au lait spots

Café au lait Spots

Coast of Maine

Coast of California


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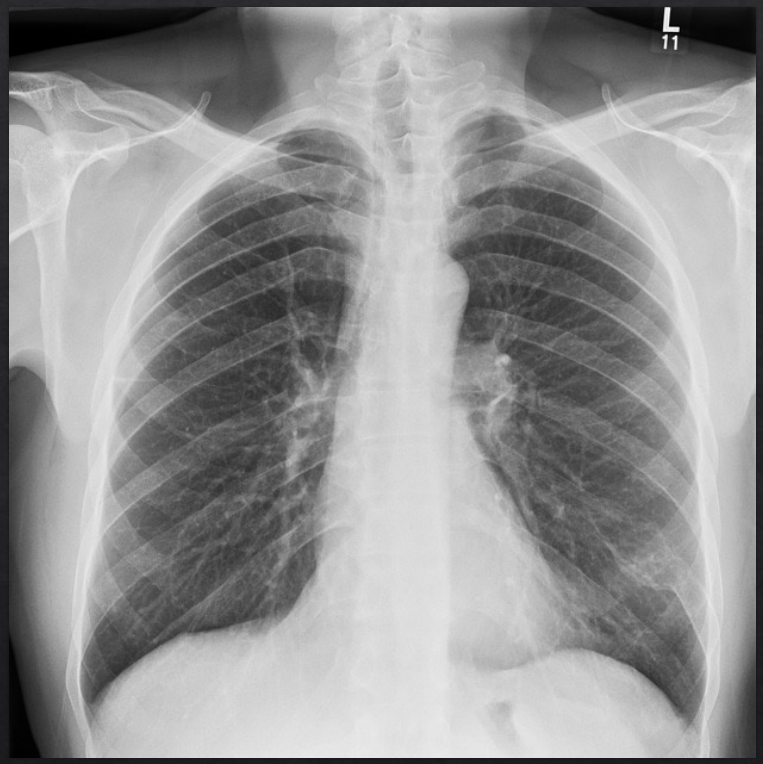
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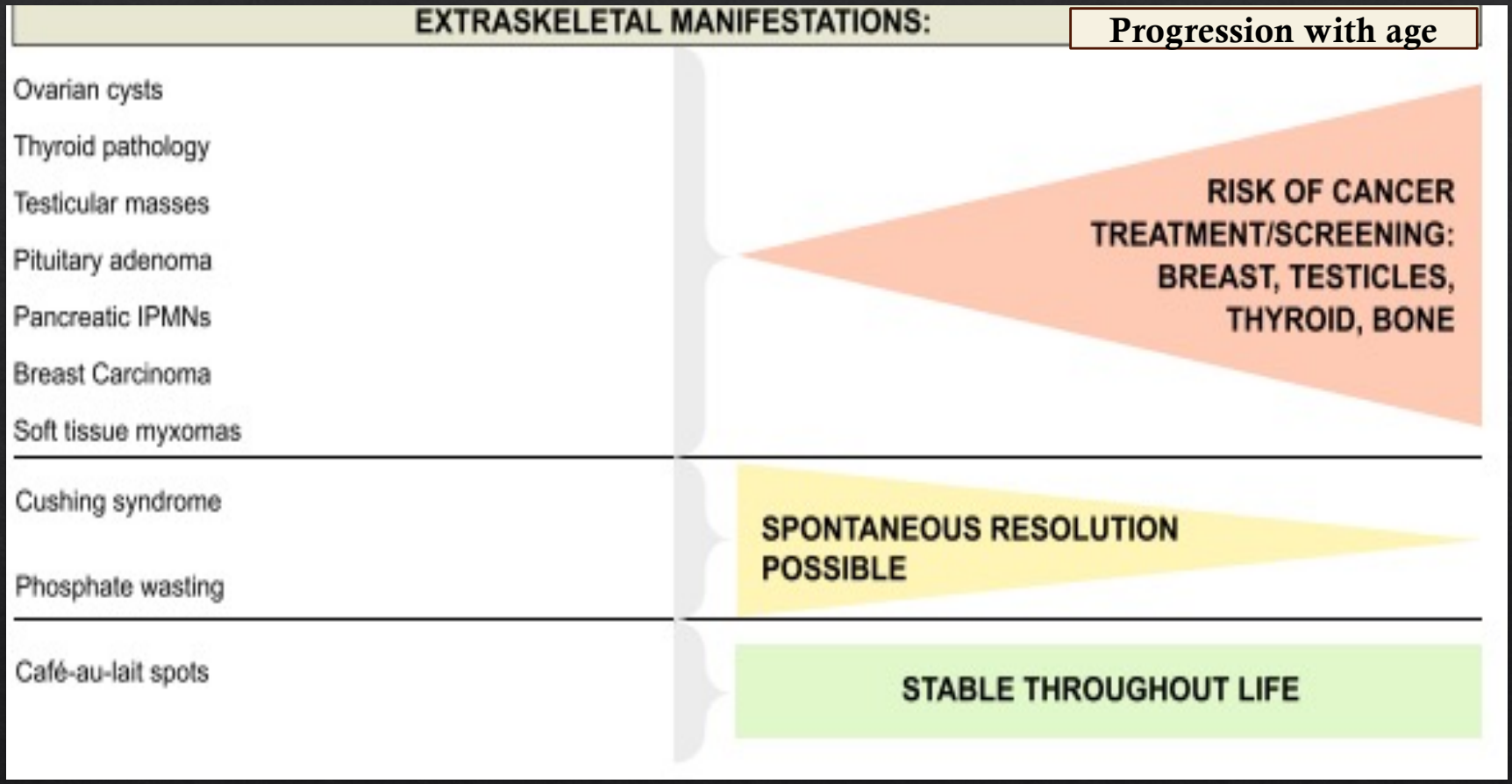
EXTRASKELETAL MANIFESTATIONS:
Progression with age
Ovarian cysts
Thyroid pathology
Testicular masses
Pituitary adenoma
Pancreatic IPMNs
Breast Carcinoma
Soft tissue myxomas
RISK OF CANCER
TREATMENT/SCREENING:
BREAST, TESTICLES,
THYROID, BONE
Cushing syndrome
Phosphate wasting
SPONTANEOUS RESOLUTION
POSSIBLE
Café-au-lait spots
STABLE THROUGHOUT LIFE

Thank you