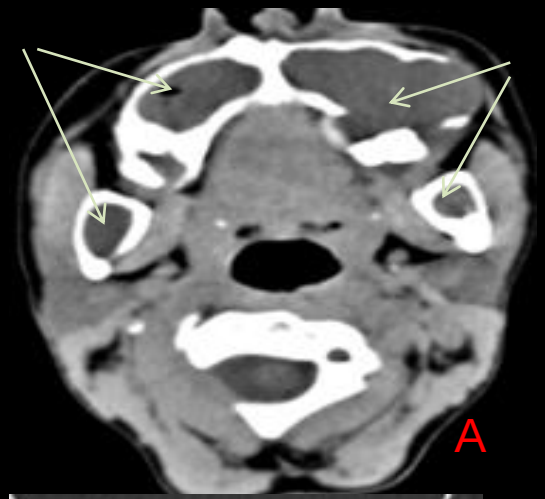
Case of the month

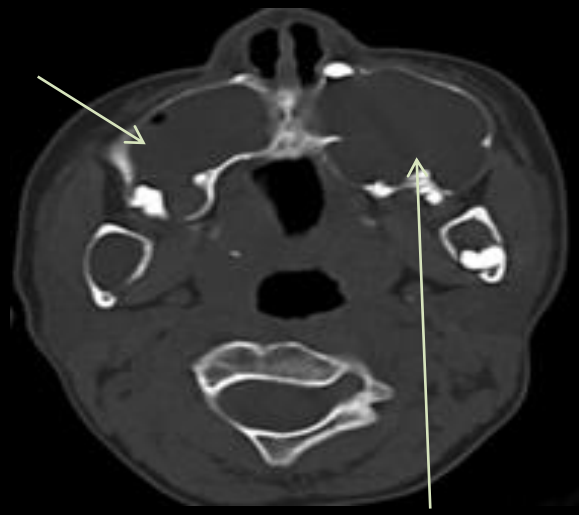
Patient presented with jaw swelling & pain.

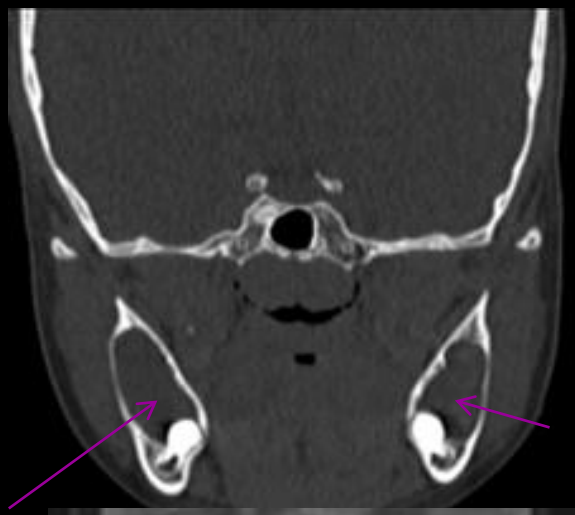
Methods

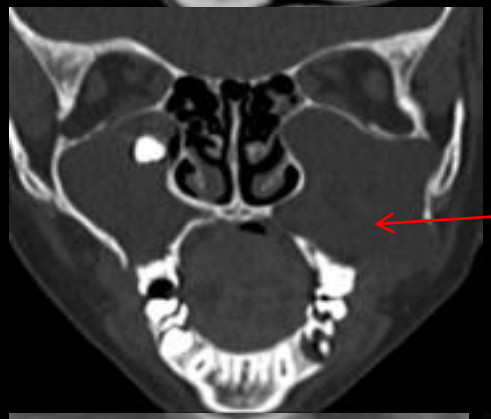
CT scan and screening MRI were performed for the same to look for characteristics of various findings.

Patient presented with jaw swelling and pain

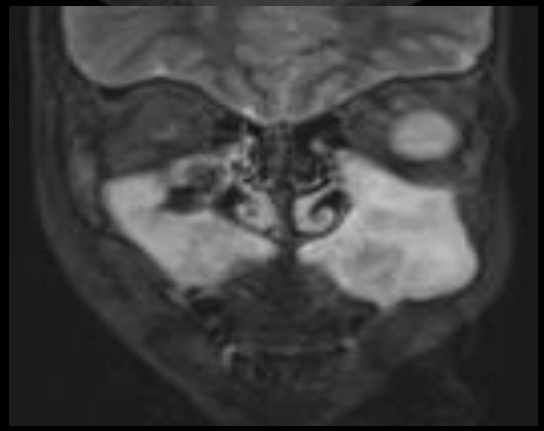


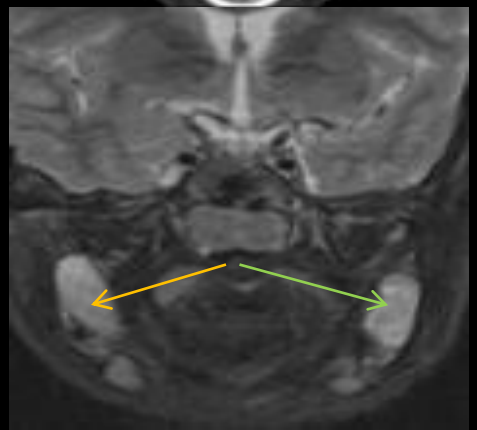


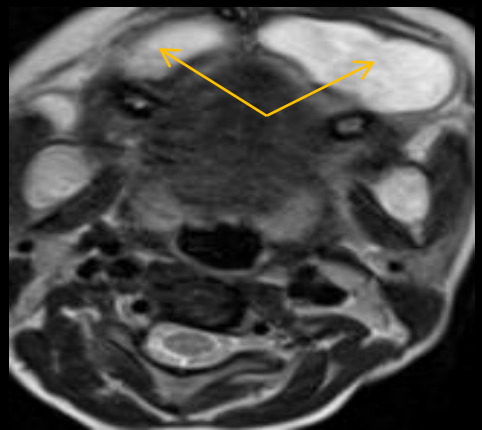




CT scan shows expansile lytic lesions involving maxillary sinus and ramus of mandible on either side with internal soft tissue density content (shown in image "A").

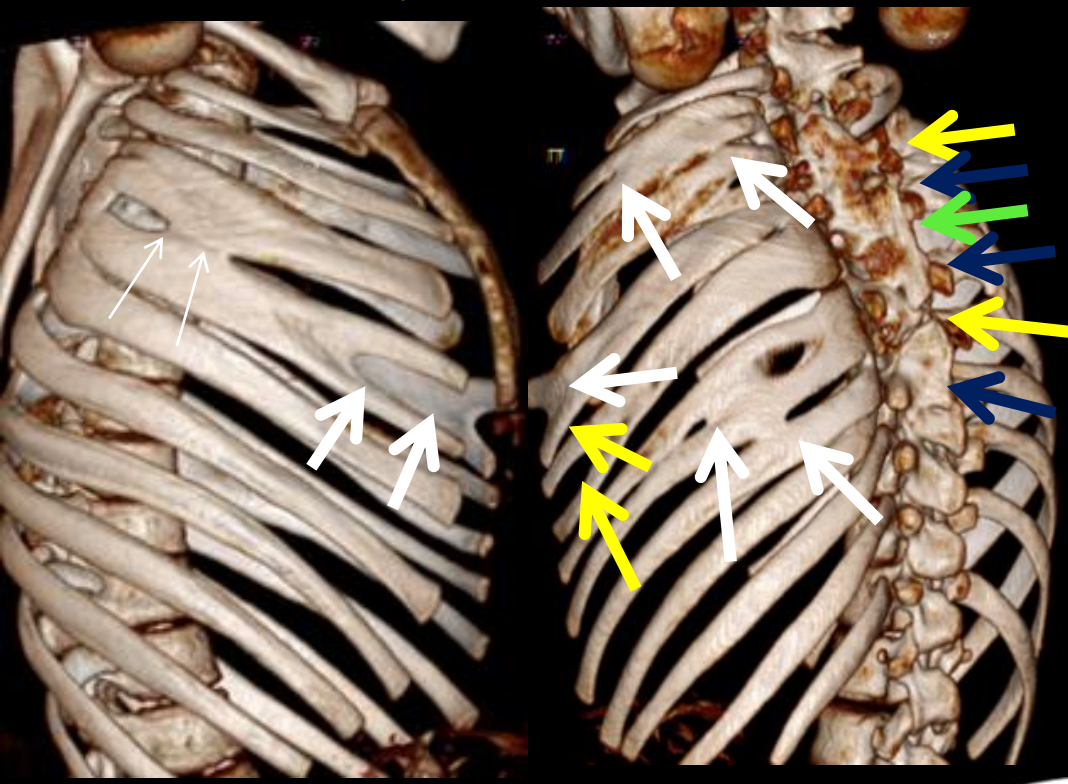






MRI scan of same patient showing similar features involving maxillary sinus and Ramus of mandible on either side suggestive of odontogenic keratocysts in mandible and in maxilla on either side

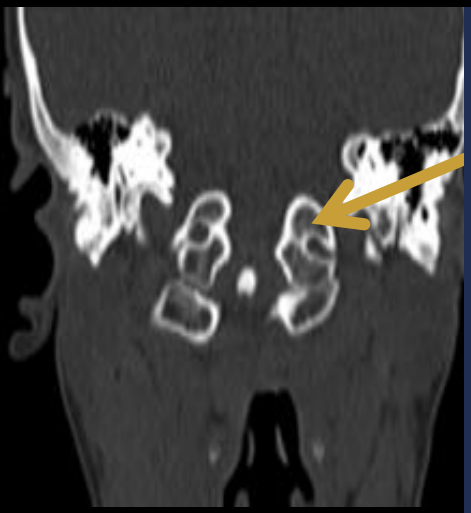
Various other features are also seen as shown



CT thorax reveals anomalies involving multiple ribs with fusion of ribs and presence of multiple bifid ribs on either side.

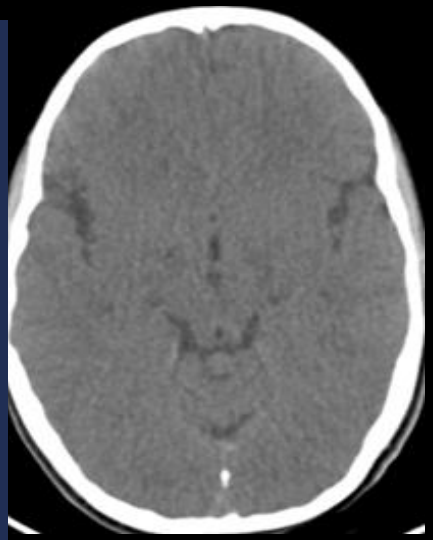
Incomplete fusion of multiple dorsal vertebral bodies.

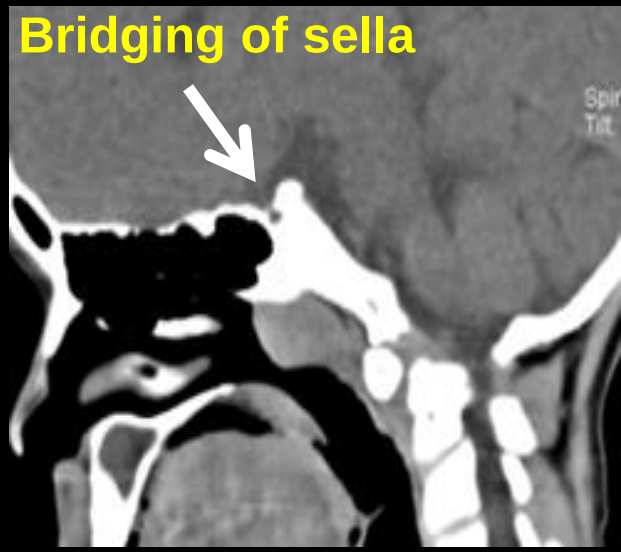
Osseous defects involving posterior arch and bifid spinous process at multiple dorsal levels.



Fusion of occipital condyle and condylar process of C1 on left side.

Focal calcifications involving interhemispheric fissure.





Results

CT and MRI findings revealed multiple odontogenic keratocysts in mandible and maxilla.

Associated with other features like bridging of sella, premature calcifications involving falx, multiple rib and vertebral anomalies.

In view of above described radiological findings and given clinical details – suggest Gorlin-Goltz syndrome aka Basal cell naevus syndrome.

It's a rare phakomatosis characterized by multiple odontogenic keratocysts, multiple basal cell carcinoma, skeletal & other abnormalities.

Discussion

Gorlin's-Goltz syndrome is an autosomal dominant syndrome with high penetrance & variable expressivity, such that not all findings are present in each patient.

It's known to run in families, with an equal frequency in both sexes.

Diagnosis is made by having two major criteria or one major and two minor criteria.

Major criteria	Minor criteria
➢ Basal cell carcinoma: >2 or 1 under age 20	➢ Macrocephaly
➢ Odontogenic keratocysts	➢ Frontal bossing, cleft lip or hypertelorism
➢ Palmar pits: 3 or more	➢ Sprengel deformity, pectus excavatum or pectus carinatum, syndactyly
➢ Bilamellar calcification of the falx cerebri	➢ Bridging of the sella turcica, hemivertebrae, flame shaped ossesous radiolucencies
➢ Rib anomalies: bifid rib, fused, splayed.	➢ Ovarian fibroma
➢ First degree relative with Gorlin syndrome	➢ Medulloblastoma

Discussion

Multiple odontogenic keratocysts, arising from the rests of dental lamina of the mandible and occasionally the maxilla are common in this disorder. These are unilocular or multilocular, lined by stratified squamous epithelium and may contain displaced teeth. These cysts may be complicated by the development of pathological fractures, ameloblastomas and squamous cell carcinomas, and have a high rate of recurrence.

It is important to differentiate odontogenic keratocysts from cystic lesions. CT enables the detection of small cysts not evident on radiographs; erosion of the cyst walls; displacement and resorption of adjacent teeth and can better define the extent of these cysts and their complications.

Rib anomalies could be unilateral or bilateral, alterations in 1st to 4th ribs are most typical and easily depictable on CT scan.

Conclusion

- In any patient with multiple OKCs, the possibility of GGS must be considered.
- It is great importance to make an early diagnosis because of its malignant potential, since the severity of complications, such as malignant skin and brain tumors can be reduced, and maxillofacial deformities related to the jaw cysts can be avoided. Regular follow-up should be offered.
- Its transmission is autosomal dominant with good penetrance implies the need of genetic counseling.
- This syndrome has a number of skeletal and systemic radiological manifestations.
- Awareness of the pathognomonic radiologic findings (odontogenic cysts in the jaw, rib anomalies, calcifications of the falx cerebri) will allow early detection and definite diagnosis of GGS. These imaging findings are accurately recognizable in a cross sectional imaging.

References

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3. Gorlin-Goltz syndrome: a rare case report Ravi Prakash Sasankoti Mohan, Sankalp Verma, Neha Agarwal, Udita Singh, BMJ Case Rep. 2013; 2013: bcr2013010409. Published online 2013 Jun 27. doi: 10.1136/bcr-2013-010409,PMCID: PMC3702894.

4. Imaging findings of Gorlin-Goltz syndrome, Parisa Hajalioghli, Ali Ghadirpour, Reza Ataie-Oskuie, Acta Radiol Short Rep. 2015 Jan; 4(1): 204798161455294. Published online 2015 Jan 6. doi: 10.1177/204798161455294, PMCID: PMC4299365

Final Diagnosis

GORLIN GOLTZ SYNDROME

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