

Radiological Findings of Gorlin Syndrome-A Case Report

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Abstract

Basal cell nevus (Gorlin-Goltz) syndrome is a rare autosomal dominant disorder with multiple developmental anomalies and predisposition to various neoplasms. We present a 60 year old male with pigmented, ulcerated skin lesions in face, neck and trunk, histologically proved to be basal cell carcinomas. Mild exophthalmos, hypertelorism, chest wall deformity & scoliosis were noted. Radiological imaging showed calcification of falx cerebri & tentorium cerebelli, bridging of sella turcica, right third, fourth and fifth bifid ribs, scoliosis of lumbar spine, odontogenic keratocyst of mandible and flame shaped lucencies in hands.

Keywords: Bifid rib, Gorlin-Goltz syndrome, Odontogenic keratocyst

Introduction

Basal cell nevus (Gorlin-Goltz) syndrome is a rare autosomal dominant disorder with high penetrance and variable expressivity caused by mutation of tumor suppressor gene PTCH-1 located in long arm of chromosome 9q22.3-q31,¹ leading to multiple developmental anomalies and predisposition to various neoplasms at an early age. It presents with multiple basal cell carcinomas especially at a younger age. Wide spectrum of radiological manifestations is seen involving various systems.

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Case Report

A 60 years old male, presented with skin lesions in face, neck and trunk. There is history of sibling having similar skin lesions who died at the age of 15 years due to some unknown cause. On examination multiple pigmented ulcerated skin lesions were seen in the face neck & trunk. Mild exophthalmos, hypertelorism, chest wall deformity & scoliosis were noted. Histologically, the pigmented lesions were shown to be Basal cell Carcinoma.

Skull radiograph AP view shows calcification of falx (Fig. 1a). Axial NCCT scan shows heavily calcified falx cerebrii & tentorium cerebellii (Fig. 1b and 1c). Lateral view of skull shows bridging of sella turcica (Fig. 1d). Chest radiograph PA view shows bifid right fourth and fifth ribs & splaying of

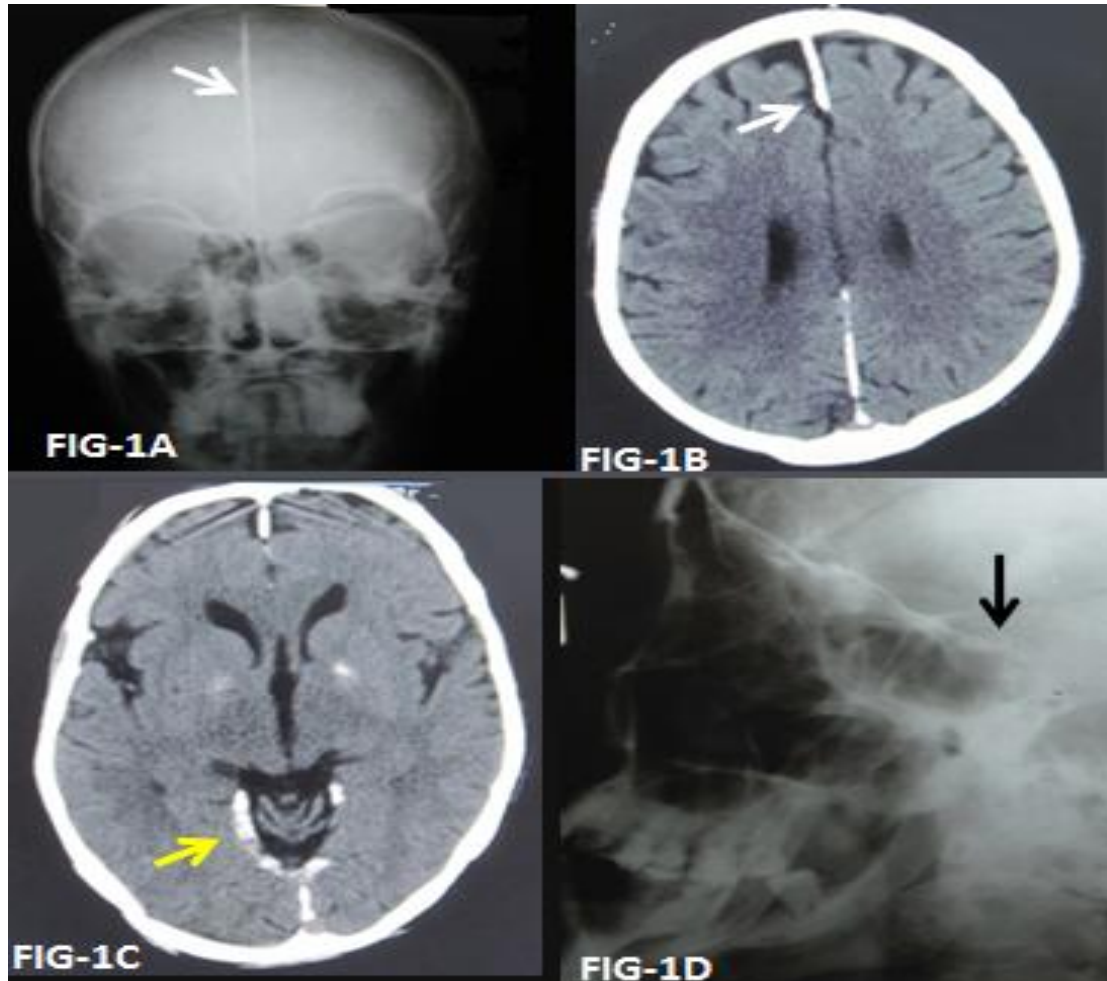


Fig. 1a: AP Radiograph of skull of a 60 years old male with Gorlin’s syndrome shows calcification of falx cerebri (arrow). **Fig. 1b:** Axial NCCT of brain shows heavily calcified falx cerebri (arrow). **Fig. 1c:** Calcified tentorium cerebelli (arrow). **Fig. 1d:** Lateral view of skull shows bridging of sella turcica (arrow).

right third rib with narrowed right hemithorax (Fig. 2a). Radiograph of LS spine AP view shows scoliosis of the lumbar spine (Fig. 2b). A panoramic radiograph shows an edentulous mandible with a small odontogenic keratocyst in the right side (Fig. 3a). X-ray of bilateral hands shows flame shaped cortical lucencies in the metacarpals and phalanges (Fig. 3b).

Discussion

Basal cell nevus (Nevoid Basal cell carcinoma/Gorlin/ Gorlin-Goltz) syndrome is inherited as an autosomal dominant trait with high penetrance & variable expressivity. It is

the fifth phakomatosis. It is a rare condition with a prevalence of 1 in 50,000 to 150000 with regional variation.² The male: female ratio is 1:1. It is difficult to diagnose this syndrome in childhood. The median time of diagnosis is second and third decades and it may go undiagnosed in many patients.

Jarisch and White described it for the first time in 1894. In 1960, Robert James Gorlin and William Goltz gave a vivid description of the syndrome.

It involves multiple systems including skin, skeletal system, genito-urinary system,

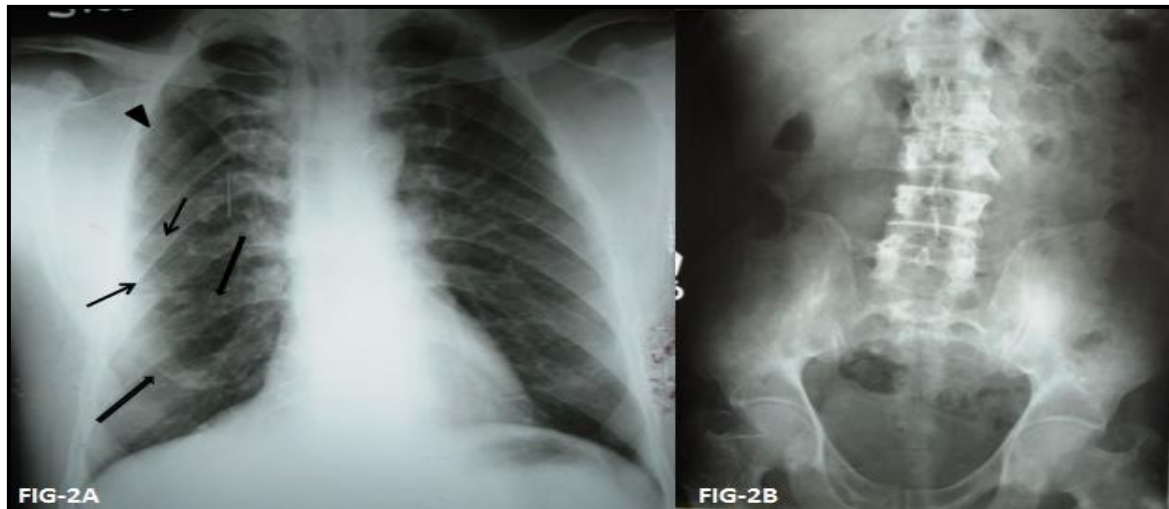


Fig. 2a: PA chest radiograph showing bifid right anterior 4th (thin arrows), 5th (thick arrows) ribs and posterior 3rd rib (arrowhead). **Fig. 2b:** LS spine AP view of the lumbar spine showing scoliosis.



Fig. 3a: Panoramic view showing small odontogenic keratocyst (arrow) in the body of mandible on right side. **Fig. 3b:** Radiograph of hand showing flame shaped lucencies in the metacarpals and phalanges.

CNS, heart, and ovaries. Classic triad of multiple basal cell carcinomas of the skin, odontogenic keratocyst of the jaw and bifid ribs is seen. Other rib anomalies include splaying, synostosis and cervical ribs. Involvement of first four ribs is typical and it may be unilateral or bilateral. Other skeletal anomalies include kyphoscoliosis, blocked vertebrae, hemivertebrae and spina bifida occulta, short fourth metacarpals, pectus carinatum or pectus excavatum and Sprengel's deformity. Flame shaped lucencies in phalanges defined by Dunnick et al which are small, pseudocystic, lytic bone

lesions are seen on radiographs of the hands (30%) and feet (17%).^{3,4}

Facial dysmorphism (due to cleft lip or cleft palate, frontal bossing and moderate to severe hypertelorism), pitting of palms and soles and macrocephaly are commonly seen.

The odontogenic keratocysts are one of the most consistent (appears in more than 90% of the cases)⁵ and early manifestations (usually in the second decade) of this syndrome. The mandible is more commonly involved than maxilla and posterior involvement is more common than anterior.

Surgical enucleation is the treatment offered with a very high rate of recurrence. There is risk of development of ameloblastoma or squamous cell carcinoma in these cysts.⁶

Ectopic calcification of the CNS in the form of lamellar calcification of the falx cerebri(70-80%),calcification of the tentorium(20-40%), diaphragm sellae (60 to80%) with bridging of sella turcica are very common abnormalities.⁷

It is associated with CNS neoplasms, mostly medulloblastoma (in 10% of cases). However, other CNS neoplasms like meningioma, astrocytoma and craniopharyngioma and cardiac and ovarian fibromas have also been reported with Gorlin's syndrome.

Kimonis et al proposed that Gorlin's syndrome can be diagnosed when 2 major or 1 major and 2 minor criteria are present.⁸

• **Major criteria**

- Two or more basal cell carcinomas or one in persons younger than 20 years.
- Histologically proven odontogenic keratocysts of the jaw.
- Three or more palmar or plantar pits.
- Bilamellar calcification of the falx cerebri.
- Bifid, fused, or markedly splayed ribs.
- First-degree relative with Gorlin syndrome.

• **Minor criteria**

- Macrocephaly.
- Congenital malformations (cleft lip or palate, frontal bossing, coarse face, hypertelorism).

- Other skeletal abnormalities (sprengel deformity, marked pectus deformity, or syndactyly of the digits).
- Radiologic abnormalities (bridging of the sella turcica, vertebral anomalies like hemivertebrae, fusion or elongation of the vertebral bodies, modelling defects of the hands and feet, or flame-shaped lucencies of the hands or feet)
- Ovarian /cardiac fibroma.
- Medulloblastoma.

In our case, the patient presented at a very late age and has more than two major criteria (basal cell carcinoma, falcine calcification, bifid ribs, odontogenic keratocyst of mandible, death of first degree relative having similar lesions in childhood) & few minor criteria like hypertelorism, scoliosis, bridging of sella turcica and flame shaped lucencies of hands & hence a case of Gorlin's syndrome.

The need to recognize Gorlin's syndrome is its cancer predisposition at a very early age. Exposure to UV rays predispose to basal cell carcinoma. The diagnosis can be made by a thorough family history, dental and dermatological examination and radiological investigations like radiographs, CT scan, MRI brain to look for CNS neoplasms (medulloblastoma) and pelvic ultrasonography in case of females to look for ovarian fibromas.

The diagnosis is confirmed by gene mutation analysis. Antenatal diagnosis is possible with DNA analysis of fetal cells after amniocentesis or chorionic villus sampling.

Conclusion

Basal cell nevus (Gorlin-Goltz) syndrome is a rare genetic condition characterized by

multiple basal cell carcinomas, odontogenic keratocysts, palmar and plantar pitting, rib anomalies, lamellar dural calcification, bridging of sella turcica, kyphoscoliosis, hypertelorism, exophthalmos, CNS tumors like medulloblastoma and ovarian fibroma. Although patient survival is not noticeably affected, morbidity can be significant. Early diagnosis of this condition is important for early detection of cancers associated with it and their management. Radiological imaging is sine qua none for its diagnosis and management.

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