

GORLIN-GOLTZ SYNDROME - INCIDENTAL FINDING IN PATIENT WITH BILATERAL CHEEK SWELLING

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ABSTRACT

Gorlin-Goltz syndrome, is a rare hereditary autosomal dominant disorder characterized by a wide range of developmental abnormalities and a predisposition to neoplasms. It is characterized by multiple basal cell carcinoma, odontogenic keratocysts of the jaw, hyperkeratosis of palms and soles, skeletal abnormalities, intracranial ectopic calcifications, bifid ribs and facial dysmorphism. We report a case of young male who presented with complaints of bilateral cheek swelling and progressive decreased mouth opening, who was presumptively diagnosed as a case of chronic osteomyelitis. Subsequently, imaging features fitted into the syndromic features of Gorlin-Goltz syndrome.

Keywords: Basal cell nevus, odontogenic keratocysts, skeletal anomalies, falx calcification.

Introduction

Gorlin-Goltz syndrome, also known as basal cell nevus syndrome, is a rare, autosomal dominant inherited disorder, which is associated with numerous basal cell carcinomas, maxillary and musculoskeletal malformations. These patients also have other manifestations that include prognathism, bifid ribs, calcification of tentorium and falx. These patients are predisposed to various anomalies and neoplasms.^{1,2} Basal cell nevus syndrome is caused by mutations in the *PTCH1* gene and is transmitted as an autosomal dominant trait with complete penetrance and variable expressivity.³ Its Clinical diagnosis relies on specific criteria.

Case Report

A 25-year-old male patient reported to outpatient department of our hospital with complaints of swelling and dull pain over bilateral cheeks. The patient had

similar complaints on the right side 10 years back for which he had undergone some surgical intervention. No previous surgical records were available with him, for the past surgery. Our patient denied any similar changes in his family members or closer relatives.

Physical examination of the patient revealed tall stature, frontal bossing, hypertelorism and left cheek swelling with progressive decreased mouth opening. Dermatological examination was normal. Ultrasound examination of the abdomen did not show any abnormalities.

Computed tomography(CT)of face and neck with contrast enhanced MRI corroboration revealed a large well defined expansile non-enhancing cystic lesions involving left hemimandible. An expansile lesion was also seen arising from the posterior aspect of right maxillary alveolar arch involving the tooth sockets of 2nd and 3rd molar (Fig. 1). Another similar imaging morphology lesion was noted involving alveo-

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lar arch of maxilla on the left side. On MRI corroboration, the lesions were predominantly hyperintense on T2W and isointense on T1W images (Fig. 1). There was associated widening of right hiatus semilunaris.

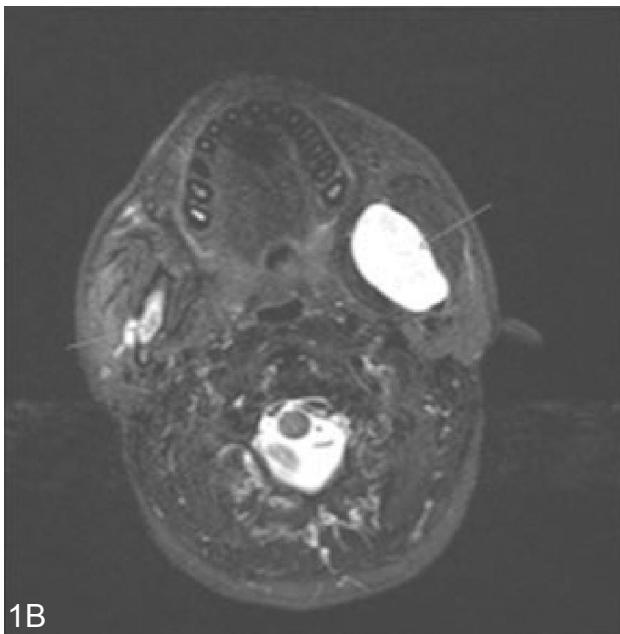
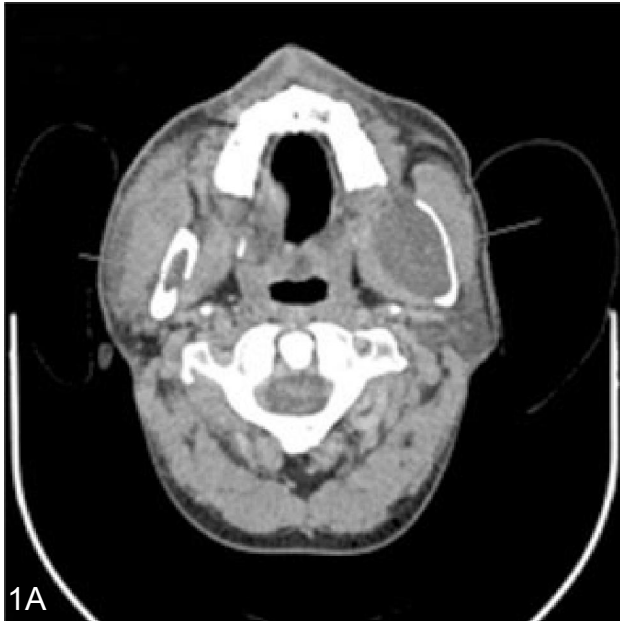


Figure 1: (A) Contrast enhanced CT shows evidence of non enhancing expansile lesion in ramus of left hemimandible. (B) T2W axial MRI showing hyperintense exapnsile lesions involving ramus of the mandible bilaterally, with larger lesion seen on the left side.

Sections through brain show calcification of falx cerebri and tentorium cerebelli (Fig. 2a). Spina bifida occulta

was seen at C6, C7 and D1 vertebrae with cervical scoliosis (Fig. 2b). Incidental note was also made of partial C5-6 block vertebra (Fig. 3).



Figure 2: (A) Contrast enhanced axial CT showing evidence of spina bifida occulta at C6 vertebra. (B) Non contrast enhanced axial CT head shows calcification of falx cerebri and tentorium cerebelli

Surgical excision and enucleation of the maxillary and mandibular cysts were done and sent for histopathological examination which revealed epithelial lining with parakeratotic layer, spinous cells and

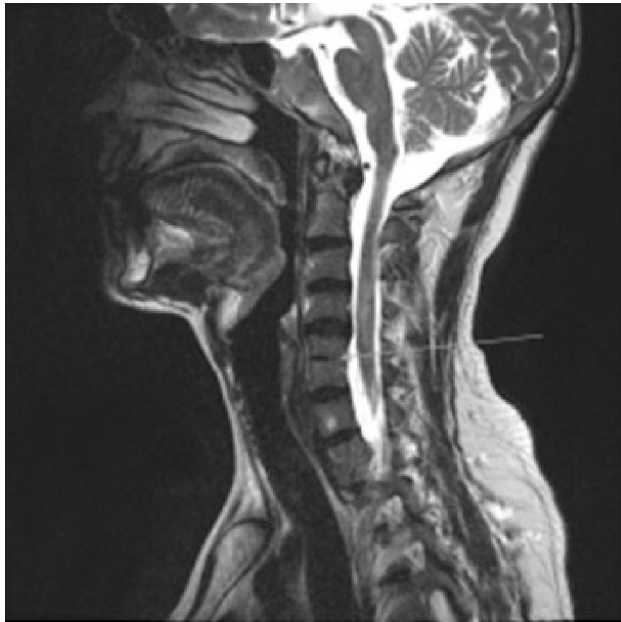


Figure 3: T2W sagittal MRI neck showing partial block vertebra at C5-6 level.

hyperchromatic columnar cells in basal layer, consistent with odontogenic keratocysts (Fig. 4).

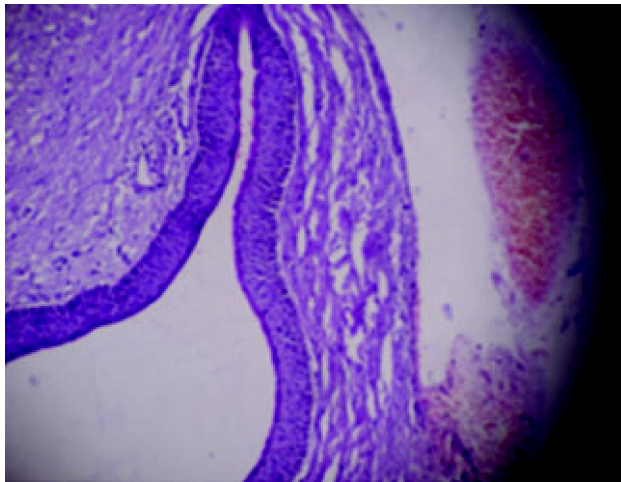


Figure 4: Histopathological examination shows epithelial lining with parakeratotic layer, spinous cells and hyperchromatic columnar cells in basal layer, consistent with odontogenic keratocysts

Discussion

Basal cell nevus syndrome, also known as Gorlin-Goltz syndrome or fifth phacomatosis, is a hereditary condition characterized by a wide range of developmental anomalies and predisposition to neoplasms. The estimated prevalence varies from 1/57000 to

1/256000 with male: female ratio 1:1.4 It is seen in males and females equally.⁵ This disease was first reported by Jarisch and White in 1984 but completely described in 1960 by Robert James Gorlin and William Goltz. This disease is caused by mutations in the tumoursuppressor gene *PTCH1* and is transmitted as an autosomal dominant trait with complete penetrance and variable expressivity. However, 35-50% cases occur due to new mutations.⁶

The major and minor criteria for diagnosis of Gorlin-Goltz syndrome was described by Evans et al. and Kimonis et al. in 1997. To establish the diagnosis two major or one major and two minor criteria are required, which are described below.^{5,7}

Major criteria

1. Multiple (>2) BCCs or one under 20 years
2. Odontogenic keratocysts of the jaws proven by histopathology
3. Palmar or plantar pits (3 or more)
4. Bilamellar calcification of the falx cerebri
5. Bifid, fused or markedly splayed ribs
6. First degree relatives with NBCCS

Minor criteria

1. Macrocephaly determined after adjustment for height
2. Congenital malformation: cleft lip or palate, frontal bossing, moderate or severe hypertelorism
3. Other skeletal abnormalities: Sprengel deformity, marked pectus deformity, marked syndactyly of the digits
4. Radiological abnormalities: bridging of the sella turcica, vertebral anomalies such as hemivertebrae, fusion or elongation of the vertebral bodies, modelling defects of the hands and feet, or flame shaped lucencies of the hands or feet
5. Ovarian fibroma
6. Medulloblastoma

Most commonly seen intracranial abnormality in Gorlin syndrome is lamellar calcification of the falx, which occurs in up to 67% of patients.⁸ Calcification has also been described in the tentorium, petroclinoid ligaments, dura, pia, choroid plexus, and basal ganglia. The odontogenic keratocysts seen in Gorlin-Goltz syndrome usually consist of unilocular or multilocular radiolucencies in the body of the mandible,

angle or ramus.⁹ The mean height is much higher in these patients and about 15% of patients are extremely tall, as seen in our patient.¹⁰ Other skeletal abnormalities include spina bifida occulta of cervical and thoracic vertebrae, scoliosis, fused vertebrae.⁹

Conclusion

Early diagnosis and proper treatment with multi-disciplinary approach for Gorlin-Goltz syndrome is required due to its higher incidence of recurrence, biological aggressiveness and susceptibility for development of malignancy. It also helps in reducing long term sequel of the disease. Genetic counselling is suggested for these patients as the syndrome is autosomal dominant.

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