Imaging of Gorlin-Goltz syndrome: Series of 2 cases

Suhail Rafiq1, Farzana Manzoor2, Musaib Ahmad Dar1, Rassieq Aslam1
Departments of 1Radiodiagnosis and 2Pathology, Government Medical College, Srinagar, Jammu and Kashmir, India

Abstract

Gorlin–Goltz syndrome (GGS) is a rare autosomal dominant disorder with multisystemic involvement. It is characterized by the triad of multiple baso-cellular epitheliomas, odontogenic keratocysts (OKC), Multiple basal cell nevi and skeletal anomalies.[1-3] It has high penetrance with a variety of dermatologic or radiologic findings along with various types of neoplasms.[4,5] GGS is a rare disorder with the autosomal dominant inheritance that occurs due to mutation of tumor suppressor gene PTCH-1 located in the long arm of the chromosome 9q22.3-q31.[4] The prevalence of GGS is about 1/60,000[7] with equal affliction for males and females.[8] It is usually diagnostic in the second or third decades. Patients suffer significant morbidity from complications.

Kimonis et al. proposed that Gorlin’s syndrome can be diagnosed when 2 major or 1 major and 2 minor criteria are present.[9]

Major criteria

- Two or more basal cell carcinomas or one in persons younger than 20 years
- Histologically proven OKC 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, modeling defects of the hands and feet, or flame-shaped luencies of the hands or feet)

Keywords: Gorlin-Goltz syndrome, noncontrast computerized tomography, odontogenic keratocysts

Address for correspondence: Dr. Musaib Ahmad Dar, Resident Hostel, Government Medical College, Srinagar - 190 010, Jammu and Kashmir, India.
E-mail: drmusaib57@gmail.com
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• Ovarian/cardiac fibroma.

We present radiological findings in two cases of GGS.

CASES

Case 1
A 13-year-old boy with multiple macular lesions on the face along with jaw swelling, prognathism, frontal bossing and exophthalmos presents to the out-patient department of dental college. The patient had multiple ulcerated lesions on the chest and back. There was a family history of similar complaints in the father but was not evaluated. Noncontrast computerized tomography (NCCT) of head and brain is advised which reveals evidence of two radiolucent lesions along with unerupted tooth in the mandible [Figure 1] and single para-midline radiolucent lesion in the maxilla [Figure 2] consistent with OKCs. There was evidence of calcified falx [Figure 3] and tentorium cerebelli. There was evidence of ground glassing and expansion of greater wings of sphenoid supporting Fibrous Dysplasia [Figure 4]. Genetic analysis revealed mutation in the PTCH-1 gene consistent with GGS. Histological analysis of ulcerated lesions revealed basal cell carcinoma [Figure 5].

Case 2
A 19-year-old boy with a history of long-standing jaw swelling presents to pediatric clinic with vomiting, strabismus and headache. Physical examination reveals evidence of pectus excavatum and macrocephaly. There was no visible skin lesion. He was stabilized with antiemetics and analgesics. Previous records in form of orthopantomogram reveal evidence of two radiolucent lesions with the associated unerupted tooth in the
mandible [Figure 6]. Emergency NCCT of the head is advised. CT reveals evidence of hyperdense mass in the left cerebellar hemisphere and calcified falk. She is advised to undergo magnetic resonance imaging (MRI) for the characterization of the cerebellar lesion. MRI reveals evidence of T1 hypointense, T2/fluid-attenuated inversion recovery heterogeneous mass [Figure 7] with diffusion restriction and post contrast enhancement [Figure 8] centered upon middle cerebellar peduncle and left cerebellar hemisphere with mild compression effect on the fourth ventricle and mild lateral ventriculomegaly consistent with medulloblastoma. NCCT of the chest which was done in view of pectus carinatum revealed evidence of butterfly thoracic vertebra [Figure 9]. Enucleation of radiolucent mandible lesion was done and histopathology revealed odontogenic keratocyst [Figure 10]. In view of
the association of lytic mandible lesions, medulloblastoma, calcified falx, vertebral anomaly in form of butterfly vertebra along with pectus excavatum, and enlarged head size provisional diagnosis of GGS was made. The diagnosis was finally confirmed by genetic analysis.

DISCUSSION

GGS was first reported by Jarish in 1894. The syndrome has been designated by a variety of different terms including Gorlin syndrome, nevoid basal cell carcinoma syndrome, basal cell nevus syndrome, syndrome of jaw cysts and jaw cyst-basal cell nevus-bifid rib syndrome. There have been case reports of Gorlin Goltz with fibrous dysplasia earlier also. After typical clinical and radiological features of GGS final diagnosis was confirmed by genetic mutation analysis.

In case no. 2, presence of mandibular radiolucent lesions with the associated unerupted tooth, calcified falx, medulloblastoma and vertebral anomaly in form of butterfly vertebra, diagnosis of GGS was made. Later, enucleation of the radiolucent lesion revealed OKC. 5%–10% of the patients may develop brain medulloblastoma, a potential cause of early death, thus requiring intervention by a neurologist.

Learning points
• Proper knowledge of different Radiological findings in GGS can help in earlier diagnosis of this syndrome
• Sometimes, syndrome can present without classical features.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initial(s) will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

REFERENCES

1. Gorlin RJ, Goltz RW. Multiple nevoid basal-cell epithelioma, jaw cysts and bifid rib. A syndrome. N Engl J Med 1960;262:908-12.
2. Manfredi M, Vescovi P, Bonanini M, Porter S. Nevoid basal cell carcinoma syndrome: A review of the literature. Int J Oral Maxillofac Surg 2004;33:117-24.
3. Casarotto AR, Loures DC, Moreschi E, Veltrini VC, Trento CI, Gottardo VD, et al. Early diagnosis of Gorlin-Goltz syndrome: Case report. Head Face Med 2011;7:2.
4. Snoeckx A, Vanhoenacker FM, Verhaert K, Chappelle K, Parizel PM. Gorlin-Goltz syndrome in a child: Case report and clinical review. JBR-BTR 2008;91:235-9.
5. Daneswari M, Reddy MS. Genetic mutations in Gorlin-Goltz syndrome. Indian J Hum Genet 2013;19:369-72.
6. Matsuzawa N, Nagao T, Shimozato K, Niikawa N, Yoshiura KI. Patched homologue 1 mutations in four Japanese families with basal cell nevus syndrome. J Clin Pathol 2006;59:1084-6.
7. Lee BD, Kim JH, Choi DH, Koh KS, Lee SR. Recurrent odontogenic keratocysts in basal cell nevus syndrome: Report of a case. Korean J Oral Maxillofac Radiol 2004;34:203-7.
8. Kimonis VE, Goldstein AM, Pastakia B, Yang ML, Kase R, DiGiovanna JJ, et al. Clinical manifestations in 105 persons with nevoid basal cell carcinoma syndrome. Am J Med Genet 1997;69:299-308.
9. Kimonis VE, Mehta SG, Digiovanna JJ, Bale SJ, Pastakia B. Radiological features in 82 patients with nevoid basal cell carcinoma (NBCC or Gorlin) syndrome. Genet Med 2004;6:495-502.
10. Jarish W. Zur lehre von den autgeschwulsten. Arch Jur Dermatol Syphilolog 1894;28:163-222.
11. Markel J. Implant prosthodontic rehabilitation of a patient with nevoid basal cell carcinoma syndrome: A clinical report. J Prosthet Dent 2003;89:436-42.
12. Honavar SG, Shields JA, Shields CL, Eagle RC Jr, Demirci H, Mahmood EZ. Basal cell carcinoma of the eyelid associated with Gorlin-Goltz syndrome. Ophthalmol 2001;108:1115-23.
13. Doede T, Seidel J, Riede FT, Vogt L, Mohr FW, Schier F. Occult, life-threatening, cardiac tumor in syndactylism in Gorlin Goltz syndrome. J Pediatr Surg 2004;39:e17-9.
14. Deepa MS, Paul R, Balan A. Gorlin Goltz syndrome: A review. J Indian Acad Oral Med Radiol 2003;15:203-9.
15. Gorlin RJ, Cohen MM, Levin LS. Syndromes of the Head and Neck. 3rd ed. New York: Oxford University Press; 1990. p. 372-8.
16. Pandeshwar P, Jayanthi K, Mahesh D. Hindawi Publishing Corporation Case Reports in Dentistry; 2012:4. doi: 10.1155/2012/247239. Article ID 247239.