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Gorlin-Goltz Syndrome: Case Report and Literature Review

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Abstract | **Case Report**

Gorlin-Goltz syndrome, also known as basal cell nevus syndrome, is an uncommon, autosomal dominant inherited disorder, which is characterized by numerous basal cell carcinomas, maxillary keratocysts and musculoskeletal malformations. In this article a case of Gorlin-Goltz syndrome has been reported in 28 -year-old male patient with illustration and review of literature.

Keywords: Gorlin-Goltz syndrome, CT scan, odontogenic keratocyst, falx cerebri calcification.

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INTRODUCTION

Gorlin-Goltz syndrome, also known as basal cell nevus syndrome, is an uncommon, autosomal dominant inherited disorder, which is characterized by numerous basal cell carcinomas (seen in 50–97% of people with the syndrome), maxillary keratocysts (present in about 75% of patients) and musculoskeletal malformations [1].

The incidence of this disorder is estimated to be 1 in 50,000 to 150,000 in the general population, varying by region [2].

We report a case of 28 year old patient male of gorlin goltz syndrom with illustration and review of littérature.

OBSERVATION

We present a 28 -year-old male patient that visited dermatology consultation for skin changes, predominantly on photoexposed areas, allegedly starting 1 year ago. The examination revealed numerous papules and nodes predominantly on his face, but also on the skin of the chest and back, some of wich are up to 2 cm in diameter (Fig. 1). Some of these were shiny and dome shaped, and some had pearly borders. Inspection revealed a coarse and faces frontal prominences. Histopathologic examination of a node excised from the nasal region confirmed the diagnosis of basal cell carcinoma. Our patient reported a similar case in his family.

The patient was referred to our department for cerebral and facial CT scan which revealed calcification of the falx cerebri and two mandibular kératocysts that comes into contact with the roots of the teeth 31,32,33,41,42,43 that are lysed. It also showed Intra-sinusal dental ectopy, teeth present on both sides of the maxilla, displaced by cysts.

We had foor major criteria thus the diagnostic of Gorlin-Goltz syndrome was established.

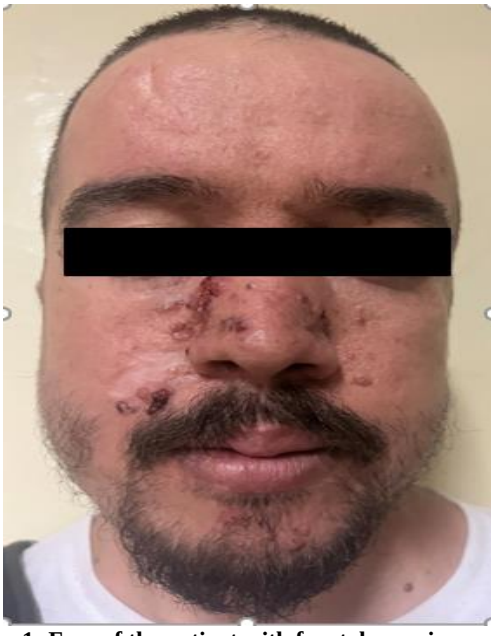

Figure 1: Face of the patient with frontal prominences and numerous facial nodes

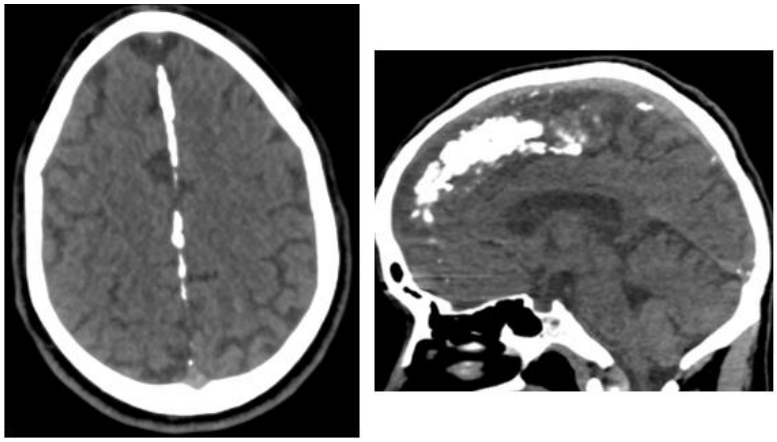

Figure 2: Cerebral CT scan in axial and sagital view showing calcification in falx cerebri

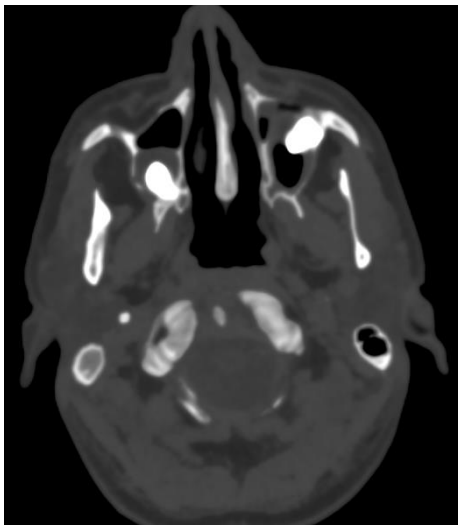

Figure 3: **Facial CT scan in axia view showing bilateral Intra-maxillar dental ectopy**

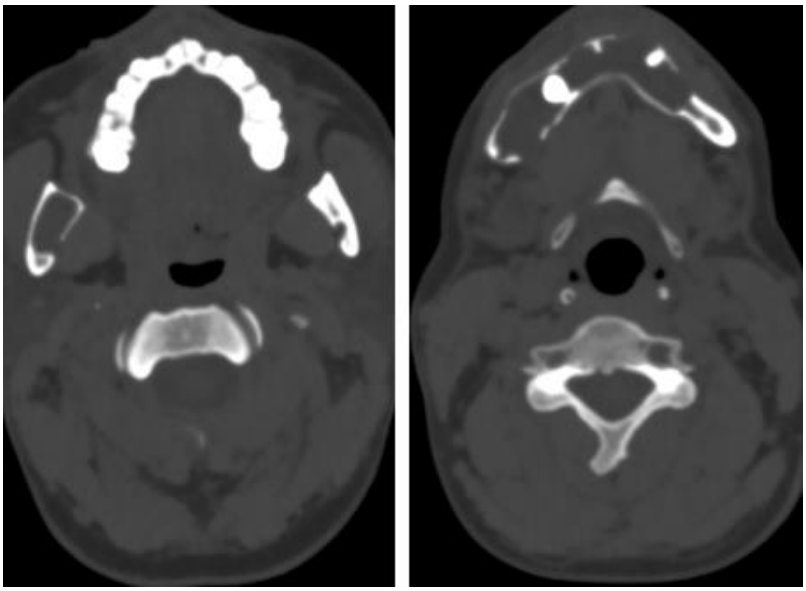

Figure 4: Facial Ct scan in axial view showing two mandibulars kératocysts that comes into contact with the roots of the teeth 31,32,33,41,42,43 that are lysed.

DISCUSSION

Gorlin-Goltz syndrome is autosomal dominant with a high penetrance and variable expressivity. It is caused by mutations in the patched tumor suppressor gene (PTCH), a human homologue of the Drosophila gene mapped to chromosome 9q21-23 [1, 4].

It was first reported by Jarisch and White in 1894. Binkley and Johnson in 1951, and Howell and Caro in 1959 suggested a relationship between basal cell epitheliomas and developmental malformations. Robert J. Gorlin and Robert W. Goltz described the distinct syndrome, consisting of the presence of multiple nevoid basal cell epitheliomas, jaw cysts, and bifid ribs [1].

It appears in all ethnic groups, but most often in whites. Males and females are equally affected [3]. Along with multiple basal cell carcinomas (BCC), jaw cysts and musculoskeletal anomalies are lesserknown manifestations of this disorder involving the skin, central nervous system, ophthalmic, endocrine, urogenital system [4-8].

The diagnostic criteria for nevoid basal cell carcinoma were established by Evans *et al.,* and modified by Kimonis *et al.,* in 1997 [2, 3]. According to them, diagnosis of Gorlin-Goltz syndrome can be established when two major or one major and two minor criteria are present as described below (Table 1) [9, 11].

Table 1: Table of major and minor criteria

Major criteria	**Minor criteria**
More than two basal cell carcinomas or one basal cell carcinoma at younger than 30 years of age or more than 10 basal cell nevi.	Congenital skeletal anomalies; fused, splayed, missing, or bifid ribs, wedged or fused vertebrae
Any odontogenic keratocyst (proven on histology) or polyostotic bone cyst.	Occipital-frontal circumference more than 97%
Three or more palmar or plantar pits (present in about 65% of patients).	Cardiac or ovarian fibroma
Ectopic calcification: Lamellar or early at younger than 20 years of age.	Medulloblastoma
Falx cerebri calcification.	Lymphomesenteric cysts
Positive family history of nevoid basal cell carcinoma.	Congenital malformations such as cleft lip or palate, polydactylism or eye anomalies (cataract, coloboma, microphthalmus).

Some authors take plurilamellar appearance of the falx cerebri calcification as a pathognomonic symptom of Gorlin-Goltz syndrome

Other diagnostic findings in adults with Gorlin-Goltz syndrome are skeletal, craniofacial, neurological and oropharyngeal anomalies (Table 2) [10, 11].

Table 2: Table presenting other diagnostic findings in Gorlin-Goltz syndrome

Skeletal anomalies	**Hemivertebrae, scoliosis, syndactyly, polydactyly, shortened 4th metacarpal.**
Craniofacial anomalies	Frontal bossing: increased size of calvaria (occipitofrontal circumference 60 cm or more in adults) brachycephaly; macrocephaly, coarse face heavy fused eyebrows; broadened nasal root calcification of the falxes; tentorium cerebelli calcification; bridged sella turcica; low positioning of occiput
Ophtolmological anomalies	congenital blindness due to corneal opacity, congenital or precocious cataract or glaucoma coloboma of iris, choroids, or optic nerve; convergent or divergent strabismus; nystagmus
Neurological anomalies	Agenesis/dysgenesis of corpus callosum congenital hydrocephalus meningioma; mental retardation; schizoid personality
Oropharyngeal anomalies	Cleft lip/palate; high arched palate or prominent ridges.

Treatment involves removal of tumors by surgical excision, laser ablation, photodynamic therapy, or topical chemotherapy, while radiotherapy is a contraindication. Chemoprevention involves use of vitamin A analogs. Recurrent odontogenic cysts (up to 60% of cases) require repeated surgical excisions. 5–10% of the patients may develop brain medulloblastoma, a potential cause of early death, thus requiring intervention by a neurologist [12].

Though survival in Gorlin-Goltz patients is not affected significantly, morbidity from complications can be considerable. Nowadays gene mutation analysis, if feasible, can confirm diagnosis. Antenatal diagnosis is possible with ultrasound scans and DNA analysis. Thus, a genetic counsellor is of importance in the ongoing care of the patient [12].

CONCLUSION

Gorlin and Goltz syndrome is a rare, multisystem, genetic disorder, with autosomal dominant transmission. It is classically defined by the triad of basal cell nevi, maxillary keratocysts and skeletal malformations.

The carcinological potential of this syndrome makes its seriousness, justifying early detection and regular regular and prolonged monitoring of patients and their offspring. The treatment remains simply symptomatic [1, 2].

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