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Review Article

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SYNDROME DE GORLIN-GOLTZ: A PROPOS D'UN CAS

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ABSTRACT

zGorlin-Goltz syndrome is a hereditary condition, with autosomal dominant transmission, due to a mutation in the PTCH gene with chromosomal location 9q 22.3-q31. It is a rare syndrome characterized by a spectrum of developmental abnormalities and a predisposition to different cancers. The complexity of clinical signs has led to the establishment of specific criteria to facilitate diagnosis. Therapeutic management is symptomatic, with rigorous oncological monitoring. We report a new observation of Gorlin-Goltz syndrome in a 45-year-old man and, through a literature review, we discuss the main characteristics of this rare condition.

KEYWORDS: Gorlin-Goltz syndrome, odontogenic keratocyst, basal cell nevomatosis.

INTRODUCTION

Gorlin and Goltz syndrome, also called basal cell naevomatosis (NBC), is a rare hereditary disorder with autosomal dominant transmission, complete penetrance and variable expressivity. It is characterized by a spectrum of developmental anomalies and a predisposition to different cancers. This syndrome is due to mutations in a tumor suppressor gene with chromosomal location 9q 22.3-q31.[1,2] This ground justifies early detection and prolonged monitoring of patients and their descendants.

OBSERVATION

A 45-year-old man, with no particular history, who presented with a left cheek swelling and another fistulized right one bringing back pus.

Maxillofacial examination: a slender appearance with protrusion of the frontal bumps, hypertelorism and multiple small nevi scattered across the face (fig1).

The intraoral examination: found a mandibular mass filling the vestibule of the 34th tooth up to the retromolar trigone. The oral mucosa was erythematous with chronic periodontitis in a very poor oral condition (fig2).

The rest of the somatic examination; neurological, ophthalmological and orthopedic was unremarkable.

He had no mental deficit, the medical investigation did not reveal any family history.

The radiological assessment (orthopantomogram, CT scan of the facial area) revealed voluminous clear mandibular and maxillary radio images with a multilocular cystic appearance of variable sizes with clear boundaries pushing back the adjacent teeth (figs. 3, 4 and 5).

All of its signs (multiple cysts, basal cell nevi, facial abnormalities) supported the diagnosis of Gorlin-Goltz syndrome.

The treatment was conservative consisting of enucleation of the mandibular and maxillary cysts with fistulectomy under general anesthesia, excision of the basal cell nevi was also carried out.

Pathological examination of the cysts confirmed the diagnosis of keratocysts. a control orthopantomogram, carried out after 6 months, showed the absence of recurrences and good bone reconstruction.

Figure 1: Clinical appearance seen from the front and in profile.

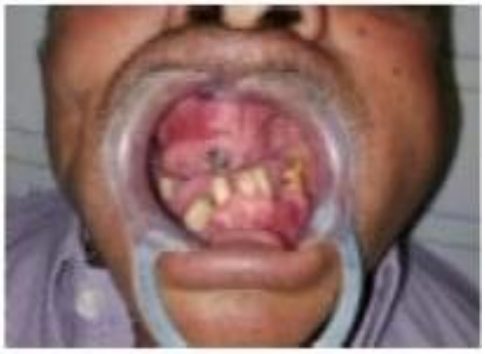

Figure 2: oral condition.

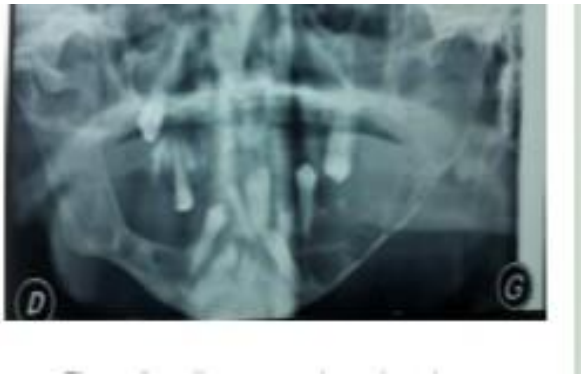

Figure 3: dental panoramic x-ray.

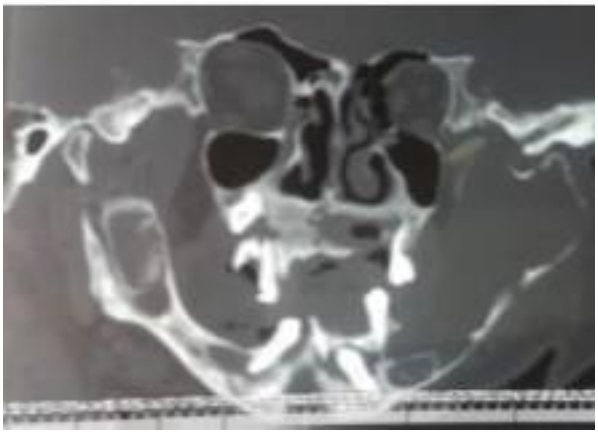

Figure 4: Massive facial CT (coronal section).

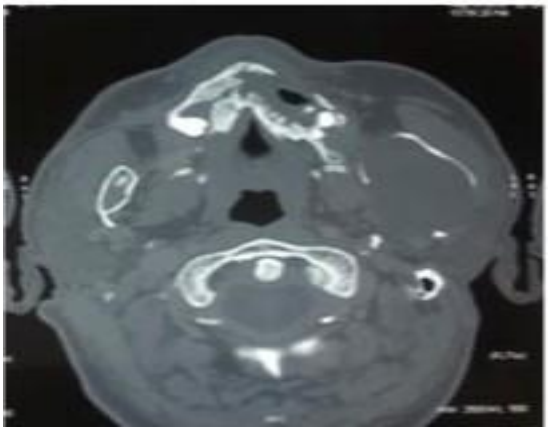

Figure 5: Massive facial CT (axial section).

DISCUSSION

Gorlin-Goltz syndrome is an autosomal dominant genetic disorder.[1,2] The first description was made in 1894 by Jarisch, but it was Gorlin and Goltz who, in 1960, defined the condition that bears their name. And since then, more than 700 cases have been reported in the literature.[1,2,3] The prevalence of this syndrome has been variously estimated, it is approximately 1/60,000 in the general population; it is found in 0.5% of patients with basal cell carcinoma.[4] The risk of having an affected child is 50% in the offspring.[4] Men and women are affected in the same proportions. Transmission is autosomal dominant, with a penetrance of 97% and variable expressivity.[1,2,4] However, sporadic cases have been reported. Between 5 and 50% of patients present with neomutations and in more than 60% of cases, no other family member is affected.[4,5,8]

The gene responsible for the syndrome was located on chromosome 9 at 9q22.3-q31 in 1996.[4,5,9]

This is the Patched (PTCH) gene, believed to have an anti-oncogenic role. According to Knudson in 1971, there is a loss of the two alleles of this gene (loss of heterozygosity) with a first germline mutation, responsible for the malformation syndrome, which predisposes to the occurrence of tumors, and the second mutation (somatic) which leads to the development of the tumor.

Clinically, this condition is characterized by a spectrum of developmental abnormalities and predisposition to different cancers.[1,2,4] It results in numerous basal cell nevi, Ontogenic keratocysts of the maxillae, palmoplantar hyperkeratosis, abnormalities of skeleton, ectopic intracranial calcifications and suggestive facial dysmorphism; A mental retardation is observed in 5% of patients.[2,3,7] These clinical manifestations are age-dependent.[2,3,4] Some signs are present from birth, others appear gradually during the second decade.

On the skin covering, basal cell nevi are found throughout the body, except the lower extremities. They transform into basal cell carcinomas usually between the age of puberty and 35 years.[4,9] Odontogenic keratocysts are present in 75% of patients, sometimes as the only manifestation during the first decade. They are often bilateral or multiple, large and recurrent.[4,9,12]

The complexity of the clinical signs found in this syndrome has led to the establishment of specific criteria to facilitate the diagnosis. It involves finding in a patient at least two of the four major criteria or one major criterion and two minor criteria.[2,4]

Major Criteria

– Odontogenic keratocysts
– Palmar-plantar porokeratosis (pitts)
– Basal cell carcinomas
– Intracranial calcifications (false brain)

Minor Criteria
– Macrocephaly, frontal bumps, hypertelorism
– Prognathism, ogival palate, milium grains
– Epidermal cysts
– Bone anomalies: costovertebral, thoracal
– Shortening of the 4th metacarpals
– Ovarian fibroids, medulloblastoma

The differential diagnosis is rarely raised apart from a few rare dermatological syndromes, such as Bazex syndrome which associates basal cell carcinomas with hypotrichosis, hypohidrosis and follicular atrophoderma.

The treatment of odentogenic keratocysts is surgical, the objective of which is the eradication of the entire lesion as well as the reduction of the potential for recurrence.

Keratocysts present an increased risk of malignant transformation into squamous cell carcinoma and a significant potential for recurrence even after primary treatment (up to 10 years later).

It is recommended to carry out annual control examinations, low recurrence rates are observed in radical interventions, the highest are recorded after simple enucleation.

CONCLUSION

Gorlin-Goltz syndrome is a rare genetic disorder. It is classically defined by the triad composed of basal cell nevi, maxillary keratocysts and skeletal malformations. The oncological potential of this syndrome makes it serious, justifying early detection and regular and prolonged monitoring of patients and their descendants. Treatment remains simply symptomatic.

REFERENCES

1. Compton JG, Goldstein AM. Fine mapping of the locus retrospective clinical and histologic study. Int J Oral Maxillofac Surg, 1996; 25: 124. for nevoid basal cell carcinoma syndrome on chromo-some 9q. J Invest Dermatol, 1994; 103: 178-81.
2. Bale AE, Gailani MR, Leffel DJ. Nevoid basal cell carci-noma syndrome. J Invest Dermatol, 1994; 103: 126S-130S.
3. Lambrescht JT, Kreusch T. Examine your orofacial cleft patients for Gorlin-Goltz syndrome. Cleft Palate Craniofac J., 1996; 34: 342-50.
4. Lacombe D. Le syndrome de Gorlin. Encyclopédie Orphonet, février, 1997.
5. Daley TD, Wysocki GP, Pringle GA. Relative incidence of odontogenic tumors and oral jaw cysts in a canadian population. Oral Surg Oral Med Oral Patol, 1994; 77: 276-80.
6. El-Hajj G, Anneroth G. Odontogenic keratocysts-a retrospective clinical and histologic study. Int J Oral Maxillofac Surg, 1996; 25: 124-9.
7. Rivet JV, Servant M, Monteil JP. Syndrome de Gorlin-Goltz à propos d'un kyste maxillaire. Rev Stomatol Chir Maxillofac, 2000; 101: 194-6.
8. Gorlin RJ, Goltz RW. Multiple nevoid basal cell epithe-lioma, jaw cysts and bifid rib syndrome. N Engl J Med., 1960; 262: 908-12.
9. Panté P, Beauvais P, Marie MP. Un nouveau cas de nae-vomatose basocellulaire (syndrome de Gorlin-Goltz). Rev Stomatol Chir Maxillofac., 1978; 79: 411-8.
10. Amlashi SFA, Riffaud L, Brassier G, Morandi X. Nevoid basal cell carcinoma syndrome: relation with desmo-plastic medulloblastoma in infancy. A population-based study and review of the literature. Cancer, 2003; 98: 618-24.