GORLIN SYNDROME
(NEVOID BASAL CELL CARCINOMA SYNDROME)

SÍNDROME DE GORLIN
(SÍNDROME NEVOIDE BASOCELULAR)

DE-DOMINGO B¹, GONZÁLEZ F², LORENZO P³

ABSTRACT

Clinical case: A 77 year-old male patient with Parkinson’s disease and senile dementia had many facial basal cell carcinomas and an ectropion of the left eye. When he experienced respiratory difficulty he was diagnosed to have an ameloblastoma in left nostril requiring surgery.

Discussion: Gorlin syndrome is an autosomal dominant condition characterized by basal cell carcinomas, and skeletal and neurological anomalies. The presence of multiple basal cell carcinomas on the eyelids in a child or in a young patient should alert ophthalmologists to the possibility of this syndrome (Arch Soc Esp Oftalmol 2008; 83: 321-324).

Key words: Gorlin syndrome, nevoid basal cell carcinoma syndrome, basal cell carcinoma, ameloblastoma, PTCH gene.

RESUMEN

Caso clínico: Varón de 77 años con enfermedad de Parkinson y demencia senil. Presentaba múltiples carcinomas basocelulares faciales y ectropión en ojo izquierdo. Comenzó con insuficiencia respiratoria y fue diagnosticado de ameloblastoma en fosa nasal izquierda e intervenido quirúrgicamente.

Discusión: El síndrome de Gorlin es una enfermedad autosómica dominante caracterizada por carcinomas basocelulares, anomalías esqueléticas y del sistema nervioso. Su pronóstico depende de la evolución de las lesiones malignas.

Es importante sospechar un síndrome de Gorlin en pacientes jóvenes con múltiples carcinomas basocelulares o en pacientes que acuden al oftalmólogo con estas lesiones a nivel palpebral, ya que su seguimiento es fundamental.

Palabras clave: Síndrome de Gorlin, síndrome basocelular nevoide, carcinoma basocelular, ameloblastoma, gen PTCH.

INTRODUCTION

Gorlin syndrome is a dominant autosomal disease that affects both men and women similarly. These patients develop multiple basal cell carcinomas, even in areas that are not exposed to the sun.

Given that it also has skeletal and nervous system abnormalities, it is known as «fifth phacomatosis». This syndrome is relevant in ophthalmology because the cutaneous lesions frequently affect the eyelids. We describe the case of a patient with this syndrome.
CASE REPORT

A 77 year old male diagnosed with the Gorlin syndrome who for over twenty years exhibited basal cell carcinomas in the facial area. In 1995, he went to the ENT practice because of breathing difficulties and was diagnosed with ameloblastoma of the left maxillar sinus and surgically intervened (fig. 1). In 2004, he exhibited a relapse of this process, but due to his neurological state (sub cortical dementia of vascular component and Parkinson’s disease) it was decided not to reintervene. This same year he was again sent to the ophthalmology unit exhibiting an ectropion and 2mm tumor on the free edge of the left eye inferior eyelid (fig. 2). Under local anesthesia the ectropion was corrected by performing a suspension of the inferior eyelid with tarsal bandelette and exeresis of the lesion at the free edge. The report form the pathologist describes the lesion as a basal cell carcinoma that respects the surgical margins, and thus no extra treatment was carried out. At present the clinical situation of the patient is stable.

DISCUSSION

Publications on the Gorlin syndrome highlight the existence of a genetic alteration linked to skeletal and development abnormalities where basal cell carcinomas appear. Tumorations such as maxilar keratocysts, meduloblastomas and ameloblastomas can appear. This is due to the alteration of the PTCH gene, located in the 9q22.3-q31 chromosome (1). The molecular biology of the tumors that appear in this syndrome is similar to that of retinoblastoma, characterized by a mutation of a recessive oncogene. The PTCH gene acts both in the development of the individual and in tumor suppression. The congenital abnormalities are a consequence of the mutation in one of the copies of this gene, while the appearance of tumorations requires the mutation of these copies. Our patient exhibited a recur-
rring maxilla ameloblastoma, a frequent tumoration in this syndrome (11%), with the existence of multiple keratocysts (24%) being more prevalent.

The incidence of Gorlin syndrome is 1 per 50,000-150,000 inhabitants, and is highest in certain areas of Australia, where exposure to the sun is higher. The most normal form of presentation is the appearance of dental or basal cell carcinomas in the third decade of life. It is important to suspect this syndrome in child patients with basal cell carcinomas. Diagnosis is mainly clinical with the Kimonis criteria (1997) being important as it considers the presence of 2 major criteria or of 1 major and 1 minor criteria (table I) necessary (2, 3). In our patient, similar to the majority of cases, the multiple basal cell carcinomas present in different regions and the maxilla ameloblastoma were enough for its diagnosis. Genetic tests were not necessary for confirmation.

From the ophthalmological point of view, in addition to the basal cell carcinomas of the eyelids, present in the adnexa oculi of our patient in a multiple manner, dysgenesis in the anterior segment with cataracts and Peters’ anomaly have been described, as well as myelinization of the nerve fibers and persistent primary vitreous, and recently vitreoretinal alterations with epiretinal membrane formations (4). Hypertelorism increases the separation of the orbits and has great diagnostic value in this syndrome (2).

The treatment of this syndrome is multidisciplinary, depending on the systems that are affected. In our case we needed the collaboration of the services of dermatology, maxillofacial surgery, ENT, neurology, ophthalmology and pathology for its diagnosis and later treatment. Prognosis depends on the evolution of the malignant lesions as well as the associated or secondary problems.

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<tr>
<th>Major criteria</th>
<th>Minor criteria</th>
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<tr>
<td>2 or more basal cell carcinomas in &lt; 20 years</td>
<td>Macrocefalia</td>
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<tr>
<td>Maxillar keratocysts confirmed with histopathological study</td>
<td>Congenital malformations: lip or palatine crack, frontal prominence, broad facies, moderate/severe hypertelorism</td>
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<td>3 or more palm-sole pits</td>
<td>Other skeletal alterations: Sprengel’s deformity, marked chest deformation, marked syndactilia</td>
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<td>Bilamellar calcification of the cerebral sickle</td>
<td>X-ray anomalies: bridge in sella turca, vertebral anomalies (hemivertebrae, fusion or elongation of vertebral bodies), modeling defects in hands or feet and flame-shaped X-ray transparencies in hands or feet</td>
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<td>Ribs that are bifid, fused or markedly expanded</td>
<td>Ovarian fibroma</td>
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<td>First degree relative with Gorlin syndrome</td>
<td>Medulloblastoma</td>
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Fig. 3: Ameloblastoma: neoplastic proliferation made up by high cylindrical cells with nuclear polarization in the surface opposite the basal membrane. The central portion of these islets is composed of a lax mesh of cells reminiscent of the starred mesh.
lesions, for later resection, as we have done in our case.

REFERENCES


