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Retroauricular dermoid cyst associated with Gorlin syndrome. A case report

A.A. Henares Chavarino [a,*], C.P. Bérniz Laborda [a], M. Estiragués Cerdá [a], A. Ros Magallón [a], M. Vicente Ruiz [a], A. Bazán Álvarez [b]

[a] Department of Plastic, Aesthetic and Reconstructive Surgery, Clínica Universidad de Navarra, Spain
[b] Head of Department of Plastic, Aesthetic and Reconstructive Surgery, Clínica Universidad de Navarra, Spain

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ABSTRACT

Background: Gorlin syndrome is an autosomal dominant genodermatosis characterized by the early appearance of multiple basal cell carcinomas, odontogenic keratocysts and skeletal abnormalities. It is caused by mutations in the hedgehog signalling pathway, primarily in the tumour suppressor gene PTCH1 (9q22.1-q31).
Case summary: We present a 14-year-old girl who consulted for asymmetrical ears, in the context of multiple disorders such as mental retardation, snoring, non-specific coagulation abnormalities, retrognathia, *pectus excavatum* and scaphoid duplication. During the intervention, a retroauricular cystic tumour was found incidentally, reported by Pathology as a dermoid cyst. The syndrome is confirmed by a genetic study with the result of a new pathological variant in PTCH1.
Conclusion: We describe the coexistence of this entity with a dermoid cyst. Furthermore, it is exceptional in its retroauricular location and the pathological point mutation in the PTCH1 gene, consisting of the pCys56Gly variant.

Core tip

Although the dermoid cyst and Gorlin syndrome are well-known entities, we present the case of a 14-year-old patient in which the coexistence of both pathologies occurs. We propose the association of this type of tumour with this genodermatosis, and we describe a new pathological variant of the mutation. In addition, it's exceptional the location of the cyst in the retroauricular region and not in the midline.

1. Background

Gorlin syndrome (GS), also known as nevoid basal cell carcinoma syndrome (ICD-10: C44.9; ORPHA: 377), is a rare autosomal dominant genodermatosis [1,2]. It is characterized by a predisposition to the development of neoplasms including basal cell carcinoma (BCC) and the presence of developmental defects. GS is caused by different types of mutations (deletions, insertions, nonsense mutations or missense mutations) in the tumour suppressor gene PTCH1, located on chromosome 9q22.3 and coding for the homonymous transmembrane receptor, which is involved in the intracellular hedgehog signalling pathway [3,4]. This pathway is involved in tissue

* Corresponding author. Clínica Universidad de Navarra, Department of Plastic, Aesthetic and Reconstructive Surgery, Avda. Pío XII 36, 31008, Pamplona, Navarra, Spain.
E-mail addresses: ahenarescha@unav.es (A.A. Henares Chavarino), cberniz@unav.es (C.P. Bérniz Laborda), mestiragues@unav.es (M. Estiragués Cerdá), arosm@unav.es (A. Ros Magallón), mvicenter@unav.es (M. Vicente Ruiz), abazana@unav.es (A. Bazán Álvarez).

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polarity and stem cell population, modulating proliferation, migration, and differentiation [5]. Its clinical manifestations are manifold, although no clear correlation between phenotype and genotype has been established [2]. BCCs appear almost constantly at an early age, particularly before the age of 20, as well as odontogenic keratocysts and palmoplantar pits. It is variably accompanied by developmental anomalies such as costal and craniofacial abnormalities, e.g., hypertelorism, macrocephaly, cleft lip and palate, polydactyly, syndactyly, or spina bifida [3,6]. The most important cause of premature death in these patients, however, is medulloblastoma. Other tumours associated with this entity include ovarian or cardiac fibromas, mesenteric fibromas or meningiomas [2,7].

In this clinical note we present the case of a patient suffering from Gorlin's syndrome associated with a retroauricular dermoid cyst and propose the occurrence of dermoid cysts as a further manifestation within the clinical manifestation associated with this disease.

Dermoid cysts are developmental tumours composed histologically of ectoderm and mesoderm, including epithelium-lined cavities and cutaneous appendages such as hair, hair follicles or sebaceous glands [8]. Being benign and occurring mainly in the gonads (only 7% of these tumours are found in the head and neck), they rarely malign [9]. They differ from epidermoid cysts and cholesteatomas in that these two only consist of squamous epithelium, and from teratomas in that these tumours include derivatives of the endoderm, such as gastrointestinal tissue, respiratory mucosa or sweat glands [8,10].

2. Case summary

We present the case of a 14-year-old female patient who came to our clinic for evaluation of an asymmetry between the two ears present since birth, with one left ear having greater projection than the contralateral ear. The physical examination revealed ear asymmetry due to a constricted left ear at the level of the middle third, transversally, with a smaller anterior-posterior diameter than the contralateral ear (distance from the tragus notch to the lateral edge of 3 cm on the right and 2.5 cm on the left). There was also a prominent fold of the antihelix in the left ear and an increased angle between the pinna and the mastoid. All this forms a procident ear or *helix valgus* (Fig. 1).

The patient also presented retrognathia with dental malocclusion, flat valgus feet and *pectus excavatum*. Her medical history included frequent snoring, mild mental retardation, supernumerary scaphoid, and a non-specific alteration of the prothrombin gene.

It was decided to perform a unilateral otoplasty under general anaesthesia. A surgical incision was made centred in the retroauricular sulcus, and a 3 × 2 cm subcutaneous tumour was incidentally found in close relation to the mastoid, but without infiltrating it. The section of the piece, rough and greyish in external appearance, showed a semi-solid whitish content, and was referred to the pathology department for study (Fig. 2). The otoplasty was completed using the Furnas technique. Histological analysis revealed skin showing a cyst in the dermis lined by a flat stratified keratinising epithelium, in direct contact with occasional mature looking pilosebaceous follicles. The lumen of the cyst contains keratin in flakes and no cytoarchitectural atypia can be seen, and it is diagnosed as a dermoid cyst.

It is in this context that a genetic study is requested for the patient. This study resulted in a pathological point mutation in the PTCH1 gene consisting of the pCys56Gly variant, related to the nevoid basal cell carcinoma syndrome.

After analysis of the data, it is considered that there must be a link between Gorlin's syndrome and the occurrence of dermoid cysts.

3. Discussion

Despite the low prevalence of dermoid cysts, this entity should be included in the differential diagnosis of tumours in the head and neck and, as in our case, also specifically in those appearing in the mastoid or retroauricular region. However, it is difficult to differentiate them from adenopathies, lipomas or gill cysts prior to surgical resection or histological analysis of the specimen.

Their removal is not only for aesthetic purposes, but is recommended due to the theoretical, albeit minimal, risk of malignancy. It should be complete to avoid recurrence or superinfection.

Although an association with Gorlin's syndrome has previously been proposed in the literature [11], the authors only describe this correlation with midline appearance. This can easily be explained by the fact that this is the most frequent location of these tumours in the head and neck. It is therefore rare to find dermoid cysts in the mastoid or retroauricular region, with only a few cases described in

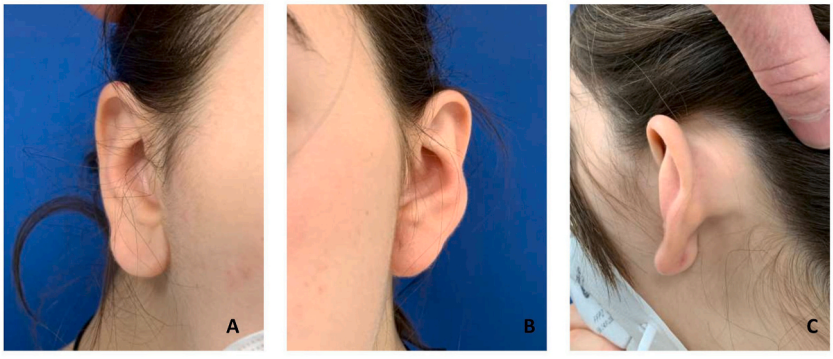

Fig. 1. Left: frontal view of the right ear. **Middle:** frontal view of the left ear, constricted at the level of the middle third transversely and with a prominent fold of the antihelix. **Right:** posterolateral view of the left ear, with an increased angle between the pinna and the mastoid.

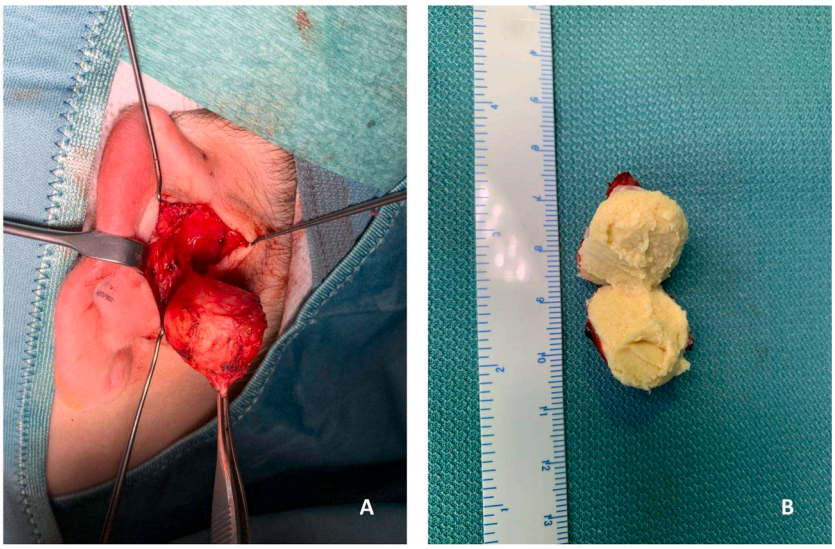

Fig. 2. Left: an intraoperative image showing an encapsulated cystic tumour in the retroauricular region at the time of removal. **Right:** the surgical specimen is exposed after its section through its midline. The whitish semi-solid content of its interior can be appreciated.

the literature [10,12–16], and even more so in relation to GS. This communication corroborates the proposal of Pivnick et al. (1996), broadening the spectrum of clinical presentation and providing new evidence on dermoid cysts in the retroauricular region.

In addition, there is a clear association between this developmental cyst and alterations in hedgehog signalling [17], which is implicated in Gorlin syndrome, which reinforces our proposal.

4. Conclusion

We describe the coexistence of this entity with a dermoid cyst and a new pathological point mutation in the PTCH1 gene, consisting of the pCys56Gly variant. Furthermore, it is exceptional in its retroauricular location.

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Consent to participate

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Consent for publication

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The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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