Case Report

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Family History of Mandibular Keratocysts from Early Age: Gorlin Goltz Syndrome

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ABSTRACT

Keywords: Nevoid Basal Cell Carcinoma Syndrome; Basal Cell Nevus Syndrome; Gorlin Goltz Syndrome; Keratocystic Odontogenic Tumor

Introduction

Gorlin-Goltz syndrome or nevoid basal cell carcinoma syndrome is a rare (estimated prevalence is 1 per 40,000 to 57,000 individuals) genetically transmitted disease, which involves a tumoral predisposition due to mutations in PATCH1 gene [1]. However, 33- 60% of patients have no family history and are thought to be a result of novo germline mutations [2]. Most frequent clinical manifestations are multiple basal cell skin tumours (BCCs) and mandibular keratocysts, both appearing at early ages [3-5]. Other frequent manifestations are medulloblastomas, palmoplantar pits, and intracranial calcifications. It is an autosomal dominant disorder with high degree of penetrance (97%) but a variable degree of expression [3]. In most cases the cause lies in pathological variants of the PTCH1 gene, a tumoral suppressor gene, found on the short arm of chromosome 9. A smaller percentage of cases involve the SUFU gene, cases with less involvement by BCC and odontogenic keratocysts but with a higher incidence of medulloblastomas in childhood [6-8]. Diagnosis is clinical, genetic study is reserved for doubtful cases and screening of relatives. Its management requires multidisciplinary approach mainly oriented towards screening for cutaneous BCCs, mandibular keratocysts. Neurological evaluation is not required except for clinical suspicion or high-risk mutations such as SUFU [2].

Case Reports

Case Report 1

We first present the case of a 58-year-old female patient, first seen in our department 6 years earlier for right hemimandibular pain. Her medical history included repeated basal cell carcinomas and a left mandibular cyst operated on in childhood.

Physical Examination: Revealed no significant alterations. Imaging tests: an OPG was performed in which a bilobulated cystic radiolucent image was observed in the right angle-body and mandibular ramus up to the coronoid process, including teeth

47 and 48 (Figure a). The study was completed with a CT scan, it described a tumefactive lesion with bone cortical preservation in the angle and horizontal branch, probably benign (Figure 2).

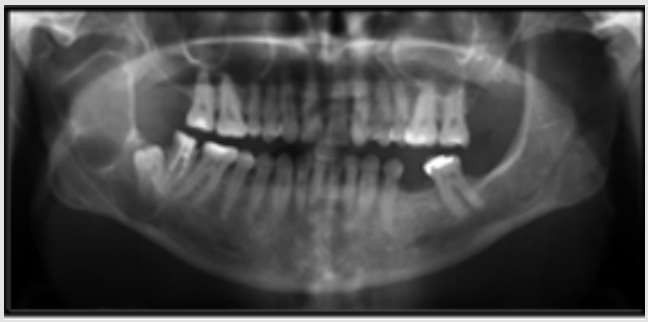

Figure 1.

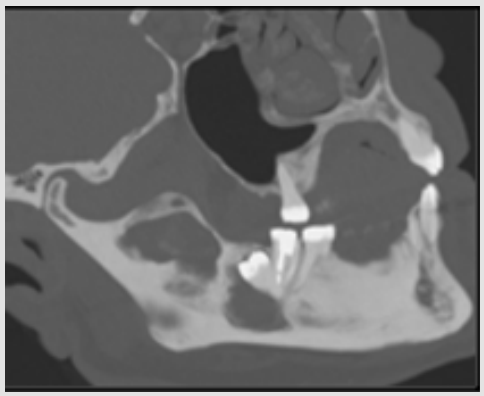

Figure 2.

Management: Surgical treatment was decided with excision of the cyst, curettage of the cavity and exodoncy of involved teeth.

Complementary Studies: Pathological study concluded that it was an odontogenic keratocyst. A genetic study was carried out on the mother confirming the mutation of the PTCH1 gene. Annual check-ups were carried out with imaging tests (the first years with CT, later with OPG) with no evidence of recurrence.

Case Report 2

Three years after first patient intervention, her 18-year-old daughter was seen in our service due to swelling in right upper maxillary region with associated ocular pain and sensation of ocular dystopia. She had no personal history of interest.

Physical Examination: Revealed right upper maxillary and periorbital region edema. Intraoral examination revealed bulging of the anterior wall of right maxillary sinus and ipsilateral nostril.

Imaging Tests: Study was completed with imaging tests (OPG + CT) in which three cystic lesions were observed in the right and left mandibular angle and right maxilla, all of them were large and involved adjacent teeth. Diffuse dural calcifications, characteristic of the syndrome, were also observed (Figure 3-5)

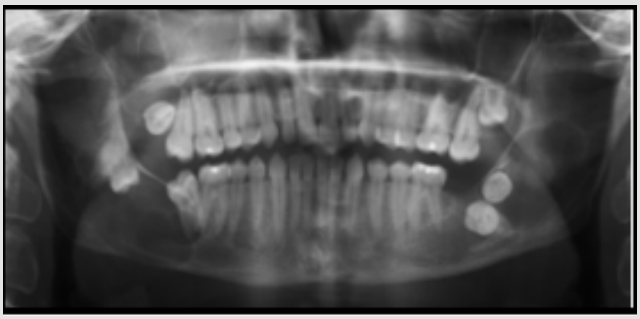

Figure 3.

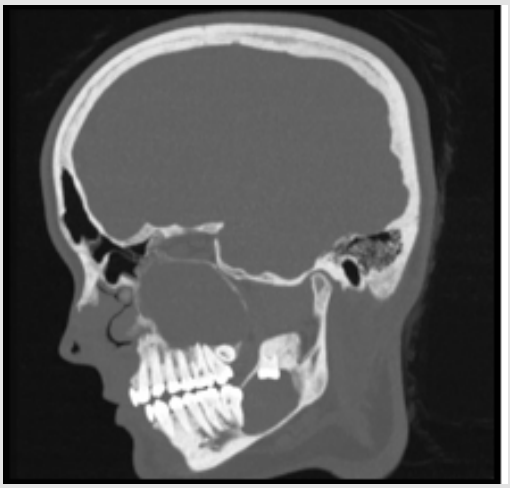

Figure 4.

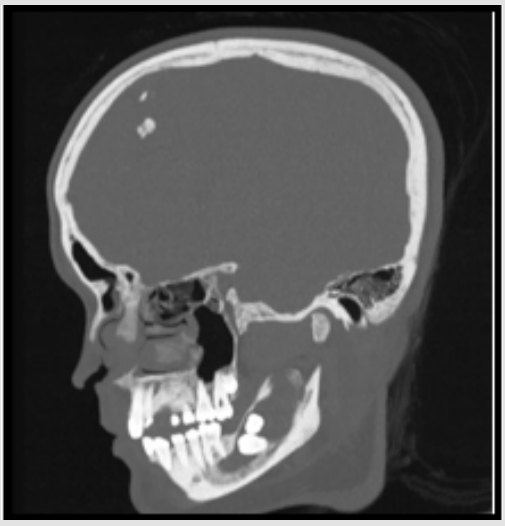

Figure 5.

Management: Surgical management was decided, performing cystectomy and curettage of the cavities together with exodoncy of involved teeth. In the case of the right maxilla, reconstruction of the defect with a Bichat ball was performed.

Complementary Studies: Pathological analysis of the three pieces concluded that they were multiple mandibular

keratocysts. Subsequent follow up two years later revealed a new left hemimandibular lesion, which required surgery. Pathological analysis concluded that it was a simple odontogenic cyst.

Results

In both clinical cases, the study was completed until enough criteria were obtained to confirm diagnosis. In the first case, the patient was diagnosed because of the histologically confirmed mandibular keratocysts. Multiple basal cell carcinomas were highly suggestive, but due to their occurrence over the age of 20 years, clinical confirmation was made by genetic study. In her daughter's case, three major criteria were observed, confirmed mandibular keratocysts, intracranial calcifications and confirmation of a first-degree relative. Close follow-up with imaging tests for early detection of recurrences was key to the correct management of the patients. The family study is also essential for genetic counselling and not to overlook other affected family members [2]. In this case, genetic testing was performed on the patient's other daughter, who tested negative.

Conclusion

Major Criteria	Minor Criteria
- Multiple BCCs before the age of 20 years	- Medulloblastoma in infancy
- Mandibular keratocyst with histological confirmation	- Craniofacial, vertebral, EESS muculoskeletal malformations
- Three or more multiple palmar or plantar pitting	- Ovarian or cardiac fibroma
- Ectopic intracranial calcification	- Pleural or lymphomesenteric cysts
- Bifid or fused ribs	- Ocular alterations (coloboma, glaucoma, cataract)
- First degree family history	

Figure 6.

Gorlin-Goltz syndrome involves a predisposition to both benign and malignant tumour pathology, as well as other multisystemic involvements. It has an autosomal dominant inheritance, so that children of an affected parent have a 50% chance of suffering from it, with a variable degree of involvement, due to its incomplete penetrance [3]. More than one hundred associated anomalies have been described, and depending on their frequency and specificity, a series of major and minor criteria are defined, with two major or one major and two minor criteria being necessary for diagnosis (Figure 6) [2]. In confirmed cases and in their first-degree relatives a follow-up at dermatological level is going to be necessary due to their high incidence of BCC from a very early age. Biopsy of all suspicious skin lesions is indicated. Periodic follow-up by OPG/CBCT of mandibular keratocysts is also recommended, since in some series they have a prevalence of 70%, in addition to a high percentage of recurrences once they have been excised [2,3]. The prognosis will be marked by the appearance of CNS involvement, and in most cases by a good diagnosis and follow-up of BCCs and keratocysts [2].

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