ISSN: 2581-5989
PubMed - National Library of Medicine - ID: 101738774

International Journal of Dental Science and Innovative Research (IJDSIR)
IJDSIR : Dental Publication Service
Available Online at:www.ijdsir.com
Volume – 8, Issue – 1, February – 2025, Page No. : 204 - 208

Early Diagnosis and Intervention in Familial Nevoid Basal Cell Carcinoma Syndrome: A Case Report of Generational Surveillance

[1]Dr. Swathy Suresh, Junior Resident, Department of Oral and Maxillofacial Surgery, Mahatma Gandhi Post Graduate Institute of Dental Sciences, Pondicherry

[2]Dr. K Sankar MDS, Head of the Department, Department of oral and maxillofacial surgery, Mahatma Gandhi Post Graduate Institute of Dental Sciences, Pondicherry

[3]Dr. Latha Ashokan MDS, Professor Department of oral and maxillofacial surgery, Mahatma Gandhi Post Graduate Institute of Dental Sciences, Pondicherry

[4]Dr. Karuna Jothi, MDS, Assistant Professor Department of oral and maxillofacial surgery, Mahatma Gandhi Post Graduate Institute of Dental Sciences, Pondicherry

Corresponding Author: Dr. Swathy Suresh, Junior Resident, Department of Oral and Maxillofacial Surgery, Mahatma Gandhi Post Graduate Institute of Dental Sciences, Pondicherry.

Citation of this Article: Dr. Swathy Suresh, Dr. K Sankar, Dr. Latha Ashokan, Dr. Karuna Jothi, "Early Diagnosis and Intervention in Familial Nevoid Basal Cell Carcinoma Syndrome: A Case Report of Generational Surveillance", IJDSIR- February – 2025, Volume – 8, Issue – 1, P. No. 204 – 208.

Type of Publication: Case Report

Conflicts of Interest: Nil

Introduction

Familial Nevoid Basal Cell Carcinoma Syndrome (FNBCC), also known as Gorlin syndrome. The syndrome, delineated by Gorlin and Goltz in 1960 was first reported in 1894(1), is a rare autosomal dominant genetic disorder characterized by a heterogeneous array of clinical manifestations affecting multiple organ systems. Gorlin and Goltz defined the condition as a syndrome comprising the principal triad of multiple basal cell nevi, jaw keratocystic odontogenic tumors, and skeletal anomalies (2)

Keywords: FNBCC, Heterogeneous Array, Hypertelorism, Odontogenic Keratocysts

Genetic Basis: Familial nevoid basal cell carcinoma primarily arises from mutations in the PTCH1 gene, which encodes a crucial component of the Hedgehog signaling pathway. This pathway plays a vital role in regulating cell growth and differentiation throughout the body. In familial nevoid basal cell carcinoma, mutations in PTCH1 disrupt this pathway, leading to uncontrolled cellular proliferation and the development of various clinical features. While PTCH1 mutations are the most common cause, the involvement of other genes like PTCH2 and SUFU has also been identified in familial nevoid basal cell carcinoma.

Case Report

Family History

Figure 1: Clinical photographs of the family with syndrome showing frontal and temporal bossing,

The patient's family history raises suspicion for Familial Nevoid Basal Cell Carcinoma Syndrome even before the onset of any clinical manifestations. A closer look at his immediate family reveals a concerning pattern:

Mother: The patient's mother had a confirmed diagnosis of multiple odontogenic keratocysts (OKCs) through biopsy. OKCs are a hallmark feature of familial nevoid basal cell carcinoma, and their presence in the mother immediately heightened the risk for the patient.

Elder Brother: The patient's older brother presented a strong case for familial nevoid basal cell carcinoma, fulfilling several major diagnostic criteria. Firstly, he had a firstdegree relative with confirmed familial nevoid basal cell carcinoma (their mother), establishing a clear hereditary link. Secondly, he too presented with OKC, mirroring his mother's diagnosis. Finally, the presence of multiple palmar pits, syndactyly and epidermal cysts in the brother strengthened the possibility of familial nevoid basal cell carcinoma within the family.

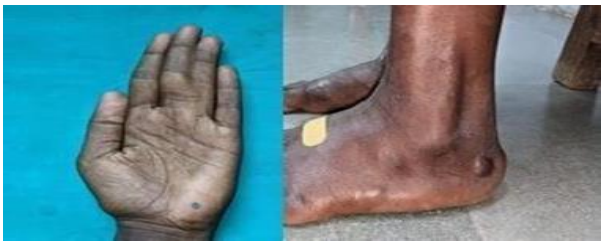

Figure 2: The elder bother showing syndactyly in the left hand along with palmar pits and, an epidermal cyst in the left leg

Clinical Features

The 5-year-old male fulfilled the major diagnostic criteria for familial nevoid basal cell carcinoma, First-degree association with a syndromic patient, with multiple palmar pits and radiologically confirmed odontogenic keratocysts (OKCs) in the jawbone. Additionally, he presented with some suggestive minor criteria, including hypertelorism and frontal bossing. A chest X-ray revealed a rib bifurcation, another potential indicator of familial nevoid basal cell carcinoma. While initial oral exams were unremarkable, the importance of radiological investigations for detecting OKC was highlighted. The patient also exhibited other findings like pectus deformity, elevated scapula, and multiple skin growths, which further led to their association with familial nevoid basal cell carcinoma.

Figure 3: Showing palmar pits, multiple skin growths, elevated scapula Clinical Course:

During a routine follow-up visit, the patient presented with mild swelling in the upper left front tooth area. An orthopantomography revealed multiple radiolucencies in the jawbone, identified in the left maxillary arch, radiolucencies were detected in the bilateral second mandibular molar regions (teeth 36 & 46) extending to the mandibular body.

FNAC was performed which gave a cheesy white fluid suggestive of OKC.

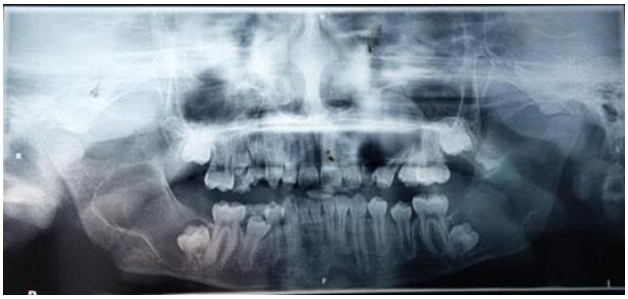

Figure 4: Pre-operative radiograph (OPG)

Management

To address the OKCs, the patient underwent surgical enucleation under generalanesthesia. This procedure involves carefully removing the cyst wall while minimizing disruption to surrounding healthy tissue.

Additionally, chemical cauterization with Carnoy's solution was performed to reduce the risk of recurrence. Due to their proximity to the cysts, teeth 63, 35 & 36, and developing tooth buds 37 & 38 were extracted. Closure was done with 3-0 vicryl. The specimen obtained was sent for histopathological examination. Following surgery, the patient recovered well and remains under close follow -up for monitoring and early detection of any new lesions.

Histopathologic examination revealed an epithelial lining of 6-8 cells thick lacking rete ridges with palisading hyperchromatic basal cell layer composed of cuboidal to columnar cells. The luminal surface had corrugated parakeratotic epithelial cells all suggestive of OKC.

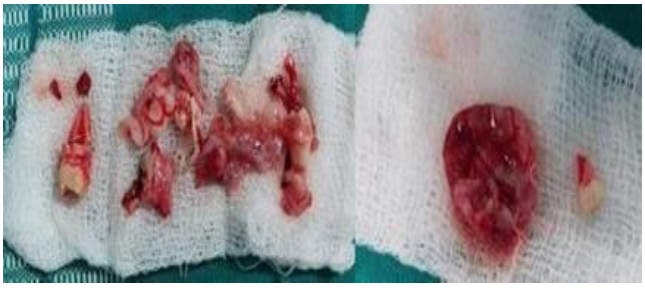

Figure 5: Enucleated cyst with extracted teeth

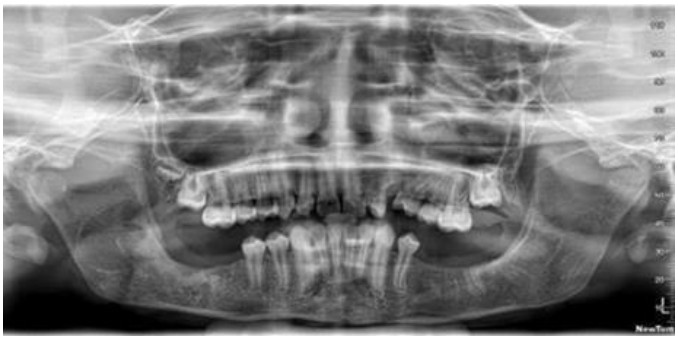

Figure 6: Post-operative radiograph after a span of 1 year

Outcome and follow-up

The patient recovered well from the surgery and did not experience any major complications. This highlights the effectiveness of early intervention and appropriate surgical management for OKCs in familial nevoid basal cell carcinoma patients.

Following the surgery, the patient remains under close and regular follow-up with a multidisciplinary team, typically consisting of a dentist, physician, and potentially other specialists as needed.

Discussion

The disease is an autosomal dominant disorder mainly characterized by the presence of multiple basal cell carcinomas, odontogenic keratocysts of the jaw, and palmar pits. It is seen in males and females equally. It is caused by mutations in PTCH, a tumor suppressor gene, a human homolog of a Drosophila segment polarity gene PTCH located in a long arm of chromosome 9q22.3. The prevalence of Gorlin syndrome is estimated to be about 1 in 60,000.

2 major criteria or 1 major and 2 minor criteria are obligatory to diagnose GorlinGoltz syndrome.

The Major and Minor Criteria are as Follows

Major Criteria Consist of

- More than two basal cell carcinomas or one inpatient less than 20 years old
- Odontogenic keratocysts of the jaw

- Bilamellar calcifications of falx cerebri and tentorium
- Three or more palmar or plantar pits
- Bifid or fused, or markedly splayed ribs
- First-degree relative with Gorlin-Goltz syndrome.

Minor Criteria Consist of

- Macrocephaly
- Congenital anomalies (cleft lip or palate, frontal bossing, coarse facies, and moderate or severe hypertelorism)
- Other skeletal anomalies (Sprengel deformity, marked pectus deformity, and marked syndactyly of the digits)
- Radiologic anomalies (such as bridging of the sella turcica, vertebralanomalies, modeling defects of the hands and feet, or flame-shaped radiolucencies of the hands and the feet)
- Medulloblastoma, seizures, mental retardation, meningioma.
- Ovarian fibroma (3)

This case report delves beyond the surface of a successful Nevoid basal cell carcinoma syndrome management strategy. It offers valuable insights into the intricate interplay between early detection, meticulous surgical intervention, and long-term vigilance for optimal patient outcomes.

This case highlights the importance of family history in the early detection of Nevoid basal cell carcinoma syndrome. Regular follow-up and comprehensive screenings are vital for early diagnosis and timely intervention.

OKCs are usually the first sign and most constant finding of this syndrome. (2) The literature contains references to various treatment modalities for OKC, for example :(4)

- Simple curettage
- Enucleation (intact shelling with or without the use of Carnoys solution or cryotherapy)
- Radical enucleation
- Decompression and Marsupialization
- Resection
- Endoscopic enucleation

Brondum reported a low recurrence rate in OKC following the use of a polyethylene drainage tube placed at the operation site and biopsied months before primary cystectomy (6). This case demonstrates the effectiveness of surgical enucleation combined with chemical cauterization with carnoy's solution in managing OKCs and preventing recurrence. It has been reported that the application of Carnoy's solution into the cyst cavity for 3min after enucleation results in the lowest rate of recurrence (0–2.5%) without damage to the inferior alveolar nerve in (5)

Genetic counseling and regular screening of at risk family members are critical components of managing Nevoid basal cell carcinoma syndrome.

Preventive measures

Long term follow-up is critical in managing Nevoid basal cell carcinoma syndrome due to the high risk of recurrence of OKCs and the potential for developing new lesions. Patients require regular dental check-ups and periodic radiographic evaluations to detect any new cysts early. Additionally, monitoring for other manifestations of Nevoid basal cell carcinoma syndrome, such as Basal Cell carcinoma and medulloblastomas, is essential.

Preventive strategies should also focus on minimizing exposure to factors that can exacerbate the condition, such as ultraviolet radiation, which can increase the risk of developing Basal Cell carcinoma. Educating

patients and their families about the importance of sun protection and regular skin examinations is vital.

Conclusion

This case report demonstrates that early diagnosis, prompt surgical intervention, and a comprehensive follow-up plan are crucial for managing Nevoid basal cell carcinoma syndrome effectively. By implementing preventive strategies and promoting patient education, healthcare professionals can significantly improve the quality of life and long-term outcomes for Nevoid basal cell carcinoma syndrome patients.

References

1. Bala Subramanyam S, Naga Sujata D, Sridhar K, Pushpanjali M. Nevoid Basal cell carcinoma syndrome: a case report and review. Journal of Maxillofacial and Oral Surgery. 2015 Mar;14:11-5.
2. Baliga SD, Rao SS. Nevoid-basal cell carcinoma syndrome: a case report and overview on diagnosis and management. Journal of maxillofacial and oral surgery. 2010 Mar;9:82-6.
3. Muhammed AH, Ali SM, Delan JA, Payman Kh M. Early diagnosis Gorlin-Goltz syndrome: A Rare Case Report. Sur Cas Stud Op Acc J. 1 (3)- 2018. SCSOAJ. MS. ID.;115.
4. Kalia V, Kaushal N, Kalra G. The syndromic multiple odontogenic keratocyst in siblings: A familial study. Ann Maxillofac Surg. 2011 Jan;1(1):77-82. doi: 10.4103/ 2231-0746.83156. PMID: 23483828; PMCID: PMC3591029.
5. Manfredi M, Vescovi P, Bonanini M, Porter S. Nevoid basal cell carcinoma syndrome: a review of the literature. International journal of oral and maxillofacial surgery. 2004 Mar 1;33(2):117-24.
6. Belgal PG, Pathak B, Shastry L. Updates in the surgical management of odontogenic keratocyst. Journal of Advanced Clinical and Research Insights. 2019 Jul 1;6(4):116