University Journal of Surgery and Surgical Specialities

ISSN 2455-2860 2021, Vol. 7(3)

Gorlin's Syndrome: A Case Report

Oxy TS Dharsini and Ramanan V P
Department of Plastic Surgery, Coimbatore Medical College, Coimbatore

ABSTRACT

Introduction: Gorlin's syndrome otherwise ref as Nevoid Basal cell carcinoma syndrome, is a multisystemic genetic disorder presenting with both cancerous and non-cancerous tumors. It is infrequent, and the clinical presentation vary in predominance. **Background:** Gorlin-Goltz syndrome also known as Nevoid Basal cell carcinoma syndrome, is multi system disorder involving the cutaneous, orofacial, skeletal, endocrine, ophthalmic, genital and central nervous system. The incidence of the syndrome is 1/50,000 - 1/256,000 population, with both sex having equal predominance. It is caused by the mutation occurring in the PTCH 1 gene of chromosome 9q, with autosomal dominant inheritance. Clinical diagnosis is based on minor and major criteria. **Aim & Objective:** Reporting a case with clinical presentation of various features of Gorlins syndrome. **Results :** The multiple lesions over the face were excised and skin cover was given by local advancement flaps. Histopathology report of the lesion came as Basal cell carcinoma. The pathology report of the swelling over jaw came as keratocystic odontogenic tumor of jaw. The remaining systemic examination didn't reveal any other tumors. The patient is on regular follow up. **Conclusion:** Gorlin syndrome being an infrequently occurring tumor, high degree of suspicion is required when patient presents with jaw cysts or with multiple cutaneous malignancies. The presentation may slightly differ in our Indian race in comparison with other races.

Key words: basal cell carcinoma, syndrome, lesions

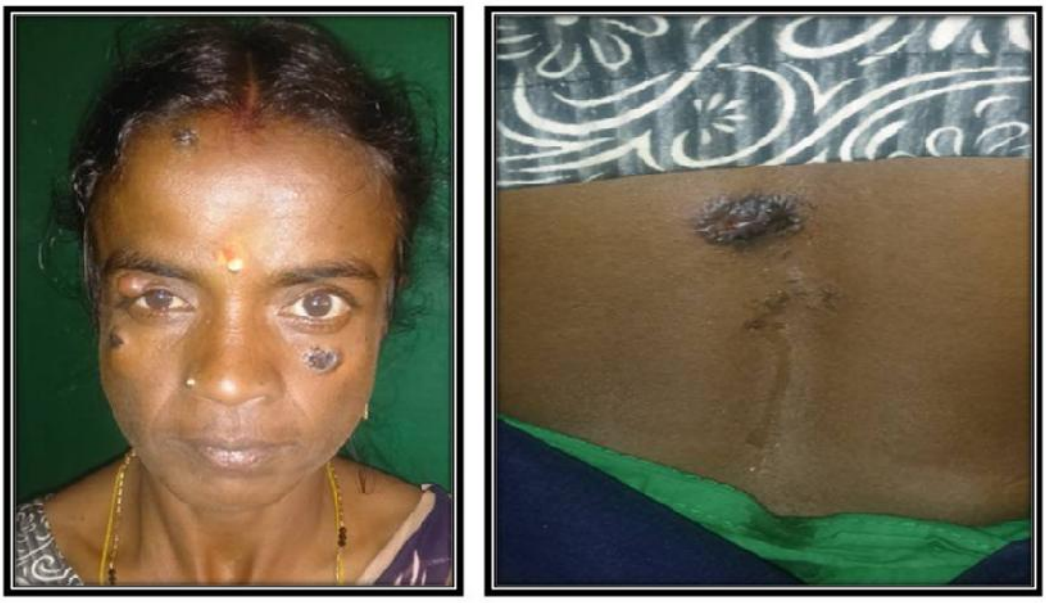
INTRODUCTION

Basal cell nevus syndrome or Nevoid basal cell carcinoma syndrome is an autosomal dominant inherited syndrome. Also referred as Gorlin syndrome characterized mainly by the presence of multiple basal cell carcinomas (BCCs), jaw cysts, and palmoplantar pits.[1-3] .Described first by Jarish and White in 1894 but its syndromic nature was defined by Gorlin and Goltz in 1960.

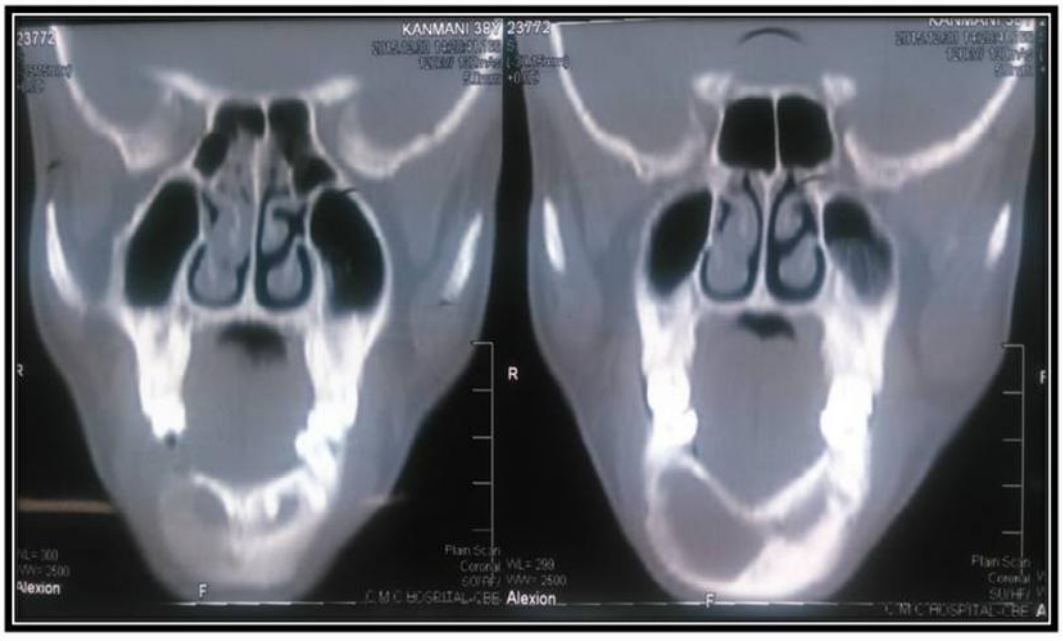
CASE REPORT

30yr old female patient presented in our hospital with multiple pigmented skin lesions and swelling over the jaw. On clinical examination, the patient was moderately built, with multiple skin lesions in various part of body measuring about 1.5x1.5cm in the maximum diameter. The lesions were pigmented, non-healing ulcerative lesion with well defined margins. The biopsy report of the skin lesion came as Basal cell carcinoma.

There was a hard swelling in the lower jaw measuring size 3x2 cm. CT scan revealed non-expansile cystic lesion arisng from the lower jaw. Curettage was done and sent for biopsy. There was minimal frontal bossing of skull along with notching of ribs and palmar-plantar pits. X ray skull showed calcification of falx cerebri. Chest x ray showed notching of ribs. Ultrasonogram of abdomen and pelvis was within normal limits.



Pre op picture showing various lesions in face and thoraco-lumbar region



CT scan face - showing cystic lesion in the mandible

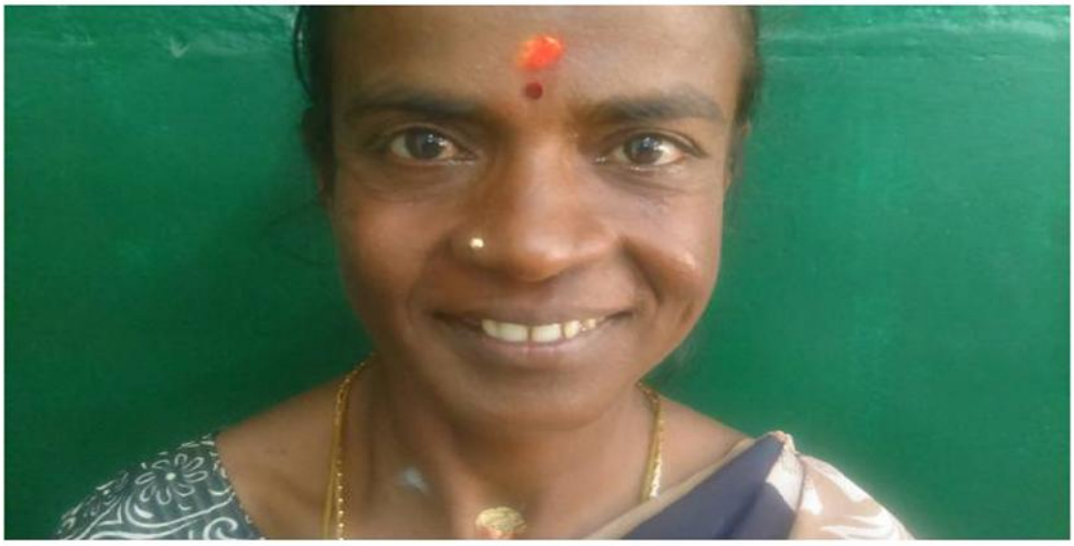
TREATMENT

The lesions in face was treated wide local excision with 5mm clearance. The resultant defect was covered by local advancement flap. The lesion in the thoracolumbar region and forehead was treated conservatively.

Intra op

Immediate post op

One year follow up revealed no recurrence of the lesion in any part of the body.



6 months follow -up

DISCUSSION

Basal cell carcinoma syndrome is inherited as an autosomal dominant disorder . It is mapped to chromosome 9q22.33.1 exhibiting high penetrance and variable expressivity.[4] in the drosophila gene. It has variable expressivity, so all findings are not present in same patient. It has familial inheritance and commonly occurs in both sexes, but around 30% of the people have sporadic mutations.

The cutaneous manifestations are multiple BCCs, milia, epidermal cysts, sebaceous cysts, lipoma and fibromas. In our patient there was multiple basal cell carcinomas with one epidermal cyst in the right upper eyelid.

Multiple odontogenic keratocysts arises from the dental lamina of the mandible and occasionally in the maxilla are common in this disorder. These are unilocular or multilocular lined by stratified squamous epithelium. These cysts may be complicated by the development of pathological fractures, amelobastomas, and squamous cell carcinomas and have a high rate of recurrence. Small cysts may be asymptomatic and can be identified by radiological investigation. Pits on hands and feet are characteristic features of the syndrome. They more frequent with advancing age. the pits are a result of premature desquamation of most of the horny layer.

In the skull there is early onset of calcification of Falx cerebri, tentorium cerebri, dura, and choroids. Bridging of sella turcica due to calcification of the diaphragm sellae is seen in 60-80% of patients. Neurological abnormalities include agenesis of the corpus callosum, congenital hydrocephalus, mental retardation, medulloblastomas, and meningiomas. Ophthalmological abnormalities seen are internal strabismus, congenital blindness, and hypertelorism. Calcification and hypertelorism was seen in our patient.

Other skeletal abnormalities include rib anomalies (splaying, synostosis, bifid, and cervical ribs), vertebral anomalies (block vertebra, hemivertebra, spina bifida occulta, and kyphoscoliosis), and shortening of the metacarpal and phalanges.

Abnormalities of the reproductive system are ovarian and uterine fibromas in females and cryptorchidism and hypogonadism in males. Other findings in BCNS are mesenteric cysts, renal calculi, cardiac fibromas and a tendency to develop various other neoplastic lesions such as melanomas, neurofibromas, and rhabdomyosarcomas. Our patient had none of these findings.

The major diagnostic criteria include multiple BCCs, odontogenic keratocysts, palmar and plantar pits, flare calcification, and a positive family history. The minor criteria are congenital skeletal anomalies (ribs, vertebra), cardiac or ovarian fibromas, medulloblastomas, lymphomesenteric cysts, and congenital malformations (i.e., cleft lip/palate, polydactyly, eye anomalies).[5]

The presence of 2 major or 1 major or 2 minor criteria is diagnostic of Gorlin syndrome.[5] Our patient had four major (BCCs, Odontogenic cysts, palmoplantar pits, and falx cerebri calcification) and one minor criteria (frontal bossing).

As it has autosomal dominant inheritance with good penetrance and any child of an affected family is at 50% risk of carrying the affected gene makes genetic counseling very mandatory. Regular follow up is necessary for assessing progression of BCCs.[6] In conclusion, we need to have a high index of suspicion to diagnose this rare syndrome as early diagnosis and genetic counseling could prevent consequences.

Conflict of Interest: Nil.

REFERENCES

1. Gorlin RJ, Vickers RA, Kelln E, Williamson JJ. The multiple basal cell naevi syndromes: Analysis of a syndrome consisting of multiple naevoid basal cell carcinoma, jaw skeletal cyst, skeletal anomalies, medulloblastoma, hype responsiveness to parathormone. Cancer. 1965;18: 89–104. [PubMed]

2. Gorlin RJ. The Naevoid basal cell carcinoma syndrome. Medicine (Baltimore) 1987;66:98–113. [PubMed]

3. 7th ed. USA: McGraw-Hill Companies; 2008. Fitzpatrick's Dermatology in General Medicine; pp. 1042–3.

4. Quinn AG. Molecular genetics of human non–melanoma skin cancer. Cancer Surv. 1996;26:89–114.[PubMed]

5. Kimonis VE, Goldstein AM, Pastakia B, Yang ML, Kase R, DiGiovanna JJ, et al. Clinical manifestations in 105 persons with naevoid basal cell carcinoma syndrome. Am J Med Genet. 1997;69:299–308. [PubMed]

6. Chavan RG, Phadke VA, Joshi R, Joshi K KU, Wadhwa SL. Gorlin's syndrome. Indian J Dermatol. 1998;43:175–8.