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Letter to Editor

Gorlin syndrome – A case report

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Sir,

Nevoid basal cell carcinoma syndrome (NBCCS) or Gorlin syndrome results from a series of abnormalities affecting various organs. Mutations in the patched gene (PTCH 1) on chromosome arm 9q are identified as the cause of abnormalities. It is an autosomal dominant syndrome, delineated by Gorlin and Goltz in 1960.[1] Here, we report a case of Gorlin syndrome.

A 55-year-old lady presented with multiple hyperpigmented plaques ranging in size from 2 × 2 to 5 × 5 cm over face and trunk since the age of 20 years. There was progressive increase in size and number of the lesions and some of them subsequently ulcerated [Figure 1]. None of the family members were similarly affected. Macrocephaly, hypertelorism, frontal bossing, marfanoid habitus with positive thumb and wrist signs, multiple palmar pits [Figure 2], and a cystic palmar swelling of size 2 × 2 cm were the other findings.

X-ray skull showed calcification of falx cerebri [Figure 3] and jaw cyst. Skin biopsy was typical of basal cell carcinoma (BCC) [Figure 4].

We arrived at a diagnosis of Gorlin syndrome as she satisfied Kimonis *et al.* criteria for diagnosis.[2]

NBCC is a rare genodermatoses with an approximate prevalence of 1 case/50,000–150,000 population.[3]

A diagnostic criteria was put forward by Kimonis *et al.*[2]

Major criteria

- >2 BCCs or 1 BCC in those <20 years
- Odontogenic keratocysts of the jaw (diagnosed by histopathology)
- Palmar or plantar pits (three or more in number)
- Bilamellar calcification of the falx cerebri
- Bifid, fused, or markedly splayed ribs
- First-degree relative with NBCCS.

Minor criteria

- Macrocephaly
- Congenital malformations such as cleft lip or palate, frontal bossing, coarse facies, and moderate or severe hypertelorism
- Other skeletal abnormalities – Sprengel deformity, marked pectus deformity, and syndactyly of the digits

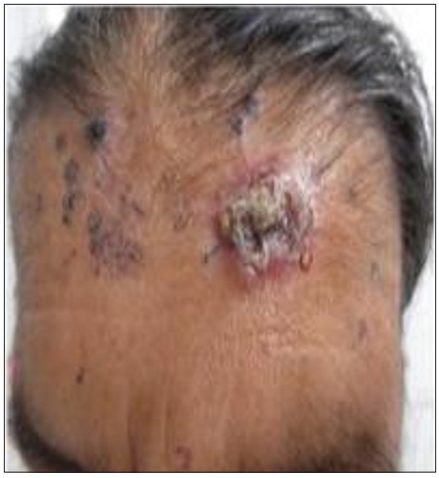

Figure 1: Ulcerated plaque on the forehead of a patient with Gorlin's syndrome.

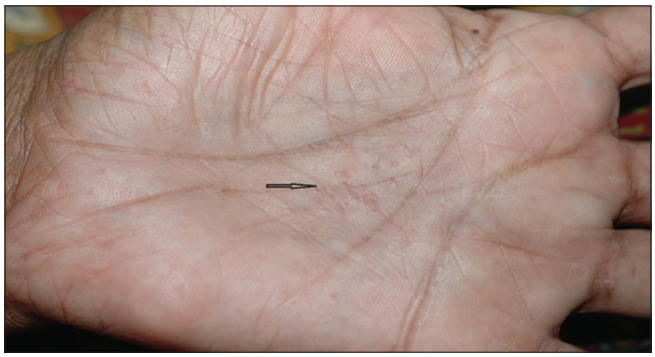

Figure 2: Multiple palmar pits in Gorlin's syndrome.

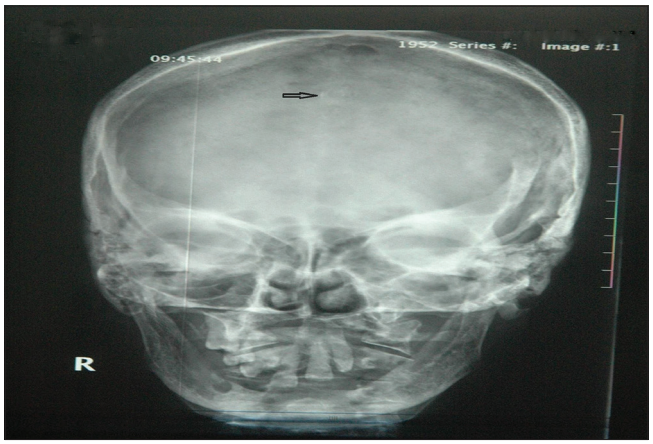

Figure 3: Calcification of falx cerebri in Gorlin's syndrome.

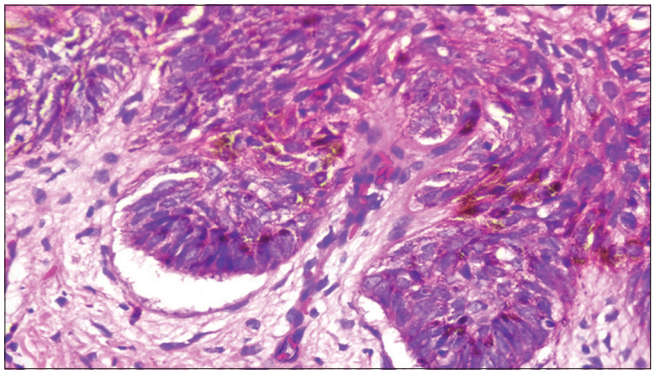

Figure 4: Skin biopsy from ulcerated plaque showing aggregates of basaloid cells with hyperchromatic nuclei, palisading, and cleft formation (hematoxylin and eosin, × 400).

- Radiologic abnormalities such as bridging of the sella turcica, hemivertebrae, fusion or elongation of the vertebral bodies, modeling defects, and flame-shaped lucencies of the hands and the feet.
- Ovarian fibroma
- medulloblastoma

To make a diagnosis of NBCCS, the patient should satisfy at least two major or one major and two minor criteria. Our patient had multiple BCCs, palmar pits, calcification of falx cerebri, macrocephaly, frontal bossing, and hypertelorism.

Enlarged calvarium and increased head circumference, widening of root of nose, frontal and biparietal bossing, mild hypertelorism, and increase in mandibular length contribute to the characteristic facies, all of which were seen in our patient.[4] Odantogenic keratocysts can be seen in 74-100% of affected patients.[5] Odontogenic keratocysts linked with NBCC are now termed as "keratocystic odontogenic tumor."[5]

In most cases, tumors appear between the adolescent period and 35 years. Kimonis *et al.* reported that by the age of 21.5 years, half of the affected develop their first BCC and by 35 years, the chance of developing the malignancy reaches 90%.[6] Patients do not show predilection for any particular clinical or histological variant of BCC; at times, they may be small, mimicking milia, small nevi, tags, or hemangiomas. Only a small fraction of tumors becomes invasive.[7] Asymmetric palmar pits, more than 3 in number, are found in about 80% of patients <10 years and when present may serve as an early diagnostic criterion.[8]

Our patient had disease onset at the age of 20. She had annular ulcerated and keratotic plaques.

Other tumors described in Gorlin syndrome such as medulloblastoma, meningiomas, bilateral calcified ovarian fibromas, fibrosarcoma, rhabdomyosarcoma, and cardiac fibroma were absent in our patient and her ultrasonogram was normal.[9] Cryptorchidism, gynecomastia, and reduced body hair are occasionally reported in affected males.[4]

Kyphoscoliosis and short fourth metacarpal may also be more common.[4] Corneal opacity leading to congenital blindness, cataract, glaucoma, milia-like lesions on the

palpebral conjunctiva, and strabismus are the ocular features described.[4]

Risk of BCCs shows a strong positive correlation with exposure to ultraviolet radiation. Thus, these patients need to avoid excess sun exposure.[7]

In the absence of follicular involvement, superficial BCCs may be treated by application of topical agents such as 0.1% tretinoin, imiquimod, 5 fluorouracil (FU), or tazarotene.[9] Examination at 3 monthly intervals for prompt excision of lesions showing morphological changes is recommended.[9] Isotretinoin 0.5–1.0 mg/kg/day is advocated as medication that can cause regression of lesions smaller than 1.0 cm in size and that may prevent the development of new lesions.[9]

In patients with multiple lesions, electrodesiccation, cryosurgery, laser ablation, photodynamic therapy, and topical chemotherapy can be tried.[9] There are contradictory reports regarding intralesional Interferon for the treatment of BCCs.[9] Radiotherapy should be avoided.[10]

Surgical excision with a margin of 3–4 mm is advised for patients with BCC if lesions are ulcerated or if the number of lesions is limited.[10]

The US Food and Drug Administration has approved hedgehog inhibitor, Vismodegib, for the treatment of locally advanced or metastatic BCC.[11]

Our patient was treated with topical 5 FU and oral isotretinoin. Ulcerated lesions on face and trunk were treated surgically. Strict sun avoidance was advised and physical sunscreens were given and she is under regular follow-up.

Patient's with Gorlin syndrome should be kept under close monitoring since they are at risk for early-onset BCC.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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