Hyperpigmented Papules with Palmar Pits

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A 40-year-old female presented with an ulcer over right lid with ectropion of 1 year duration with multiple pigmented skin lesions on the face, axillae, groin, and forearms since childhood, which were progressively increasing. Similar findings were present in her 20- year-old son. She also reported having seizure disorder. Physical examination revealed multiple hyperpigmented papules to ulcerating plaques of varying size about 3–6 mm present over the face, [Figure 1] neck, axillae, groins, and forearm. A few lesions over the nose had a thready border with a depressed centre. There were numerous skin colored 2–3 mm milia over the forehead [Figure 1]. She also had palmar pits [Figure 2].

Skin biopsies were taken from the pigmented lesions on the face and groin. Histopathological examination of the biopsy taken from pigmented lesion over face revealed an atrophic epidermis with multiple aggregates of cells arranged in nodules in the upper dermis, composed of basaloid cells arranged in a peripheral palisading manner [Figure 3]. The tumor cells were round to ovoid, with uniform nuclei and occasional mitosis. A contrast enhanced computed tomography (CT) scan of the head and orbit revealed an arteriovenous malformation in the left occipital region and calcification in cerebral sulcus.

Question

What's your diagnosis?

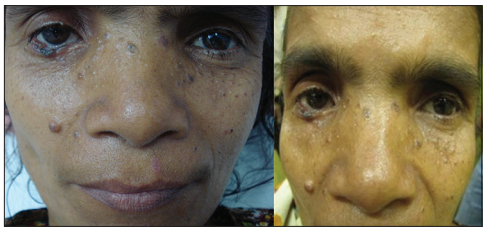

Figure 1: Hyperpigmented papules over right lower eyelid, infraorbital region with skin colored papules over forehead

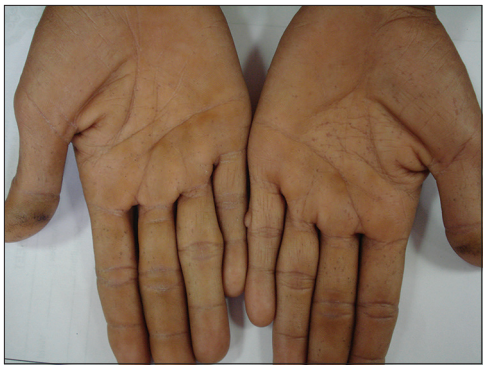

Figure 2: Palmar pits

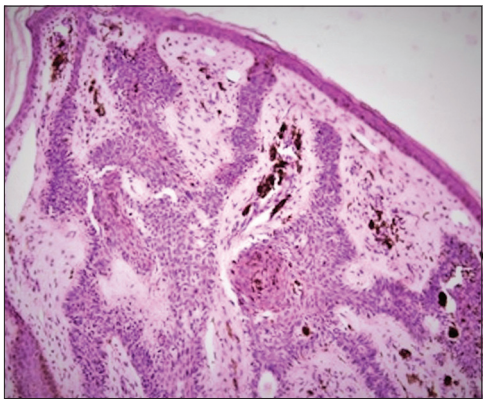

Figure 3: Upper dermis with nodules of pigment laden basaloid cells arranged in peripheral palisading manner (Haematoxylin and eosin stain original magnification ×100)

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Table 1: Diagnostic criteria for NBCCS* (*put forth by Evans and Colleagues and modified by Kimoni*[3])

Major criteria	Minor criteria
Multiple (>2) BCCs or one under 20 years	Macrocephaly
Odontogenic keratocysts of the jaws proven by histopathology	Congenital malformation
Palmar or plantar pits (3 or more)	Skeletal abnormalities
Bilamellar calcification of the falx cerebri	Radiological abnormalities
Bifid, fused, or markedly splayed ribs	Ovarian fibroma
First degree relatives with NBCCS	Medulloblastoma

*2 major or one major and two minor criteria

Answer

Nevoid basal cell carcinoma syndrome (Gorlin syndrome)

Discussion

Nevoid basal cell carcinoma syndrome (NBCCS) (Gorlin syndrome) is a hereditary condition with an autosomal dominant inheritance with high penetrance and variable expressivity.[1] It is characterized by a wide range of developmental abnormalities and a predisposition to neoplasms with a prevalence of 1/60000.[2] It is a consequence of mutations in the PTCH1 gene (chromosome 9q) that regulates cell growth. Diagnostic criteria for NBCCS are detailed below.[3] [Table 1] Our patient satisfied four major criteria. Other disorders in the differential diagnosis that can be considered include Rombo syndrome (atrophoderma, hypotrichosis, trichoepithelioma, BCCs), Bazex-Dupre-Christol syndrome (BCCs, hypotrichosis, hypohidrosis), and arsenic ingestion (BCCS, pits).

Though the mean age of onset of basal cell carcinoma (BCCs) in NBCCS is about 25 years our patient had lesions since childhood. Lesions usually appear as flesh colored or brownish papules on sun-exposed sites but non sun-exposed parts can also be involved as seen in this patient. The hyperpigmented papular lesions were initially mistaken for seborrheic keratosis (by the primary physician) though few lesions had a rolled border. Histopathology from the papular and ulcerated lesions showed classical features of BCC. The presence of milia has been described in literature and is seen in 30%–50% of patients most commonly over the infraorbital region and forehead.[4] Palmoplantar pits are seen in 85% of patients over the age of 20 years. Patients with multiple BCCs and pits in a young person should be investigated for Gorlin syndrome.

Recurrent jaw cysts, also known as odontogenic keratocysts are the most consistent and representative sign of NBCCS, seen in 90% of patients but was absent in our patient.[5,6] Ectopic calcifications of the central nervous system (CNS) have been reported, the most common being lamellar calcification of the falx cerebri, which was also seen in our patient.

Early diagnosis of NBCCS is important in order to monitor for the development of medulloblastoma and aggressive skin cancers. Treatment of BCC typically includes surgical excision and/or Mohs micrographic surgery. Other treatment modalities that have been reported include topical 5-fluorouracil, imiquimod, carbon dioxide laser ablation, and photodynamic therapy.[7] The ulcerated lesions of BCC were surgically excised and the patient was asked to regularly follow-up for periodic evaluations.

Abbreviations

NBCCS: Nevoid basal cell carcinoma syndrome

BCC: Basal cell carcinoma

CNS: Central nervous system

Declaration of patient consent

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

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Nil.

Conflicts of interest

There are no conflicts of interest.

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