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Giant Congenital Melanocytic Nevus: A Case Report

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Abstract
Giant congenital melanocytic nevus (GCMN) is a very rare condition with an estimated incidence of between 1/20,000 and1/500,000 birth, characterized by a large brown to black skin lesions. It is caused due to genetic mutations which lead to defective proliferation/differentiation/migration of melanoblasts. The risk of transformation of GCMN to malignant melanoma varies between 0 and 3.8%. Besides malignant transformation, patients with GCMN need to be periodically assessed for neurological abnormalities and psychosocial impairment. Here we present a case of 24 years adult who presented with a progressively extensive brownish-black skin lesion of approximately 40 cm over his back. In view of the potential severe complications as well as psychological trauma we draw clinicians' attention for a timely diagnosis and management.

Introduction

Giant congenital melanocytic nevus (GCMN), defined as a melanocytic lesion which is present usually at birth or appears within first few months after birth reaching a diameter ≥20 cm in adulthood. These are also known as bathing trunk, coat sleeve or stocking nevi[1]. Congenital melanocytic nevi (CMN) are large brown-to-black skin lesions caused due to genetic mutations which lead to defective proliferation/differentiation/migration of melanoblasts. Melanoblasts are precursor cells of melanocytes[2]. Although congenital melanocytic nevi (CMN) are relatively common, GCMN is a rare entity with an incidence of 1 in 20,000 to 500,000 live births[3]. Its diagnosis is mainly clinical, but may be reinforced by the histological findings. In addition to its rarity, GCMN is of interest because of its potentially severe complications, namely melanoma, neurocutaneous melanosis (NCM), due to central nervous system involvement[4]. Here, we report a case of GCMN with giant congenital nevus in a 24 years old male patient.

Case Report

A 24 years old male patient reported to the department of dermatology with a large hyper pigmented lesion on back which was present since birth and has gradually progressed to present size. The patient was a product of non-consanguineous marriage with unremarkable prenatal, natal, postnatal, and family history. His past medical

history was also non-significant. Patient gave no history of any associated symptoms with the patch. On general physical examination, he was average built with normal adult height as per Indian standards with normal vital parameters. Systemic examination was normal. Local examination of lesion revealed an extensively pigmented patch with the largest diameter of approximately 45 cm. Tufts of coarse black hair were scattered all over the lesion. Within the pigmented nevus there were several irregularly shaped macules, papules, and plaques of different colours. On palpation the surface of the nevus was irregular. No other congenital anomaly was observed and neurological examination was normal. A clinical diagnosis of GCMN was made. Biopsy of the lesion was taken and sent for histopathological examination and the findings were consistent with those of congenital melanocytic nevus. No evidence of a malignant transformation was seen. The patient was advised frequent follow-up visits and MRI scan of the brain and spine was done, which was normal.

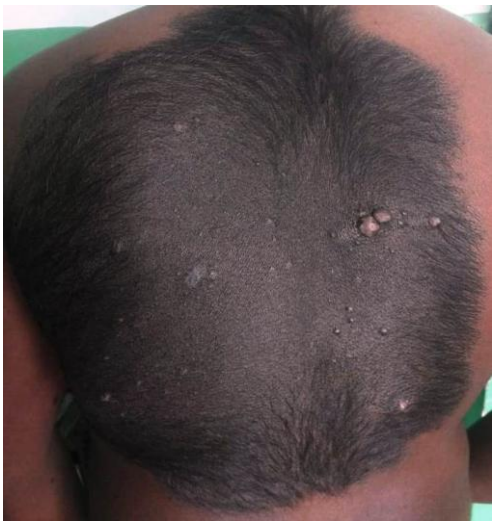

Figure 1: Giant Melanocytic Nevus

Discussion

Melanocytic nevi are benign proliferations of melanocytic cells arranged in nests in the epidermis, dermis or in other tissue[4,5]. CMN arise from gain of function somatic mutationsin either BRAF or NRAS leading to abnormal proliferation of embryonic melanoblasts. CMN are predominantly caused by sporadic de novo mutations. However, some familial cases have also been reported[2,3]. Although GCMN is considered a risk factor for the development of melanoma, the real incidence of malignancy is still a controversy and needs prospective studies of these patients[6]. However in some studies 5-10% risk of transforming into malignant melanomas has been mentioned[3]. Another complication associated with GCMN is neurocutaneous melanosis (NCM). It is characterized by abnormal melanosis of the central nervous system which is frequently asymptomatic at birth. It may manifest as hydrocephalus, lethargy, seizures, cranial nerve palsy, developmental delays, headache, and neuropsychiatric symptoms. Most of the studies reported in literature showed a slightly higher prevalence in female than males, with the ratio ranging from1.17:1 to 1.46:1[3]. However, in the present case we have reported GCMN in a male patient. These nevi have been categorized into three categories based on their size as: small (<1.5 cm), medium (1.5–19.9 cm) and giant (>20 cm)[7]. In the present case, the size measured was approximately 40 cm covering the back up to lower torso. GCMN usually begin as flat, brown, or brownish black patches but may become elevated and develop a mottled appearance and nodular surface later in life, as was seen in the present case. They are found most commonly on the trunk, followed by the limbs and the head. In the present case, the lesion involved almost the whole trunk. Although usually GCMN is an asymptomatic lesion, patients may complain of pruritus, whose mechanism is not fully understood. It is believed that symptoms may be explained by the local and intermittent stimuli of afferent sensory fibers, which would be caused by xerosis and hypohidrosis secondary to the functional impairment of adnexal structures such as sebaceous and eccrine glands[8]. Giant nevi on the scalp in a posterior axial location and those with satellite lesions are at a greater risk of malignant transformations[9]. However, in the present case, no malignant transformation was observed. Various abnormalities associated with GCMN have been reported in literature which includes limb hypoplasia, ear deformities, and angiomas[1]. No

associated congenital malformations found in this patient. The approach to GCMN represents a challenge and should be individualized taking into consideration: age of the patient, size and location of the lesion, risk of melanoma, possibility of concomitant NCM, presence of changes suggestive of malignancy on the nevus, possible functional impairments resulting from invasive procedures and the psychological impact associated with CMN or the surgical scars, often unsightly. Thus, treatment of patients with GCMN may include surgical or non-surgical procedures, psychological and / or clinical interventions, with the utmost attention in changes of colour, texture or in the surface of the lesion[10]. The size of the lesions makes their removal often dependent on the use of tissue expanders, serial interventions, use of skin flaps, grafts or a combination of more than one type of surgical procedure. More recently, synthetic skin substitutes have been used in some cases to treat the defects secondary to the removal of GCMN[11]. Taking into consideration the risk of extracutaneous melanomas, in the skin area outside the GCMN or even very deeply under the lesion, it is important to note that even patients whose nevus was completely removed must undergo lifelong, regular examinations of all skin and general medical examination, to facilitate the detection of any malignancy in its earliest stages[10]. The use of an NRAS inhibitor (trametinib), targeting the MAPK pathway, is proposed for treating patients with GCMN associated with an underlying NRAS mutation[2]. However, clinical studies to substantiate this claim are yet to be conducted. Use of endothelin-1 receptor antagonists has been proposed as a therapeutic approach for GCMN, based on evidence generated from preclinical studies[3].

Conclusion

GCMN is a very rare clinical entity which may be present in association with other congenital malformations. As it has significantly increased risk for malignant transformation patient should be kept under surveillance for the development of malignant changes. Despite its rarity, GCMN is important for its association with severe complications such as malignant melanoma and central nervous system (CNS) involvement, as well as a major psychosocial impact on the patient and his family, due to its unsightly appearance.

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