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GIANT CONGENITAL MELANOCYTIC NEVI, A RARE PIGMENTARY DISORDER- A REPORT OF 3 CASES

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Abstract

A congenital melanocytic nevus (CMN) of unknown etiology is a pigmented lesion that appears at birth or during the first year of life. The giant congenital melanocytic naevi (GCMN) measures more than 20 centimeters in size. GCMN is rare, occurring in 1 per 500,000 newborns. They can lead to serious complications like malignant melanoma (MM) and neurological deficits, including neurocutaneous melanocytosis. GCMN carries around a 6% chance of turning into malignant melanoma. About half of these melanomas show up by the time a child reaches two years old, and around 80% appear by age seven. Therefore, early removal is advised.

Keywords: Giant Congenital Melanocytic Nevi, Malignant Melanoma.

INTRODUCTION

The benign proliferation of melanocytes that produce pigment results in tumour-like abnormalities of the skin or mucous membranes known as melanocytic nevi. They are a part of a spectrum of melanocytic neoplasms, which encompass malignant variants like melanoma. Melanocytic nevi are often categorized as either congenital or acquired depending on their onset. Congenital melanocytic nevus (CMN) is a skin lesion that appears at birth or develops in the first few weeks of life and is characterized by benign growth of nevomelanocytes [1]. CMNs form in utero between the fifth and the twenty-fourth week of gestation because of a localized genetic abnormality that causes an over-proliferation of melanocytes, [2]. N-Ras is a prevalent mutation in CMN. This is thought to cause hyperproliferation of melanocyte lineage cells, which in turn causes CMNs to develop [3].

This condition has also been linked to dysfunction of the cytokine hepatocyte growth factor, which controls the migration and proliferation of melanocytes [4]. While there is evidence of familial cause in certain cases, sporadic mutations are thought to be the primary cause in most cases [5]. Even though CMN develops throughout the intrauterine life, the emergence of these late congenital nevi may be due to insufficient melanin production at the beginning and/or the nevus's small size in its early stages, which makes it difficult to identify.

Melanocytic nevi are deeply pigmented, covered with moderate growth of hair, and often associated with multiple scattered satellite lesions. The predicted incidence of

giant CMN (GCMN) with a diameter larger than 20 cm is 1/20,000 to 1/500,000 births. A 3-to-2 ratio indicating a somewhat higher incidence in females compared to males has been recorded [5].Even though GCMN is uncommon, this lesion is significant because it can have serious consequences like malignant melanoma, influence the central nervous system (neurocutaneous melanosis), and, because of its ugly appearance, may have a significant psychosocial impact on the affected individuals and their families. Between 0 and 3.8% of GCMN cases will progress to malignant melanoma [6].

Case Report

Case I:

A 12-year-old male child presented with blackish, rough skin lesions on the right arm, forearm, and hand since birth. On examination, a giant melanocytic nevus was observed over the right upper limb, with pronounced thickening, wrinkling, and tufts of coarse hair (Figure 1). There were also multiple pigmented satellite lesions on the back, left forearm (Figure 2).

This child was delivered naturally by vaginal birth from a non-consanguineous relationship. There was no family history of such lesions, and the antenatal history was uncomplicated. Other congenital anomalies were absent. There was no evidence of deep CNS extension on a CT scan of the head. The fundus examination, an X-ray of the spine, and an ultrasound of the abdomen were all normal. Skin biopsy was not performed because the patient was not willing.

Figure 1: Giant Melanocytic Nevus Involving the Right Upper Limb

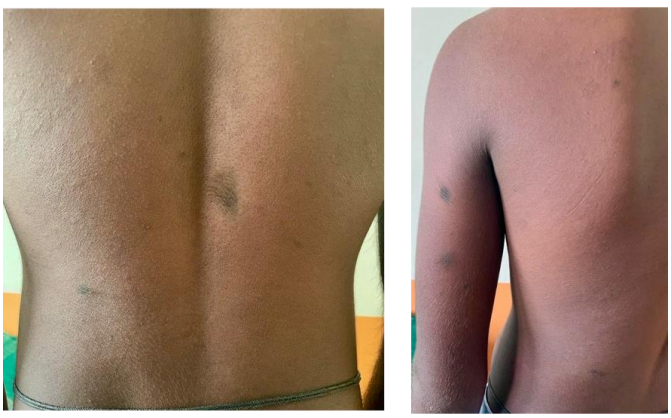

Figure 2: Multiple Pigmented Satellite Lesions on the Back, Left Forearm

Case 2:

An 11-year-old male child was brought to our OPD with complaints of blackish rough skin lesions over the back since birth. On examination, the child had a giant melanocytic nevus over the back extending up to the lateral aspect of the chest with a brownish-black hue and geographical borders. There was pronounced thickening of the involved skin. Clumps of coarse hair were scattered across the lesion. Multiple pigmented satellite lesions were noted over the chest, abdomen and lower back (Figure 3). The child was delivered naturally via vaginal delivery from a consanguineous marriage. There was no family history of such lesions, and the antinatal history was uncomplicated. Other congenital abnormalities were absent. There was no evidence of deep CNS extension on a CT scan of the head. The fundus examination, an X-ray of the spine, and an ultrasound of the abdomen were all normal. Skin biopsy was not performed because the patient was not willing .

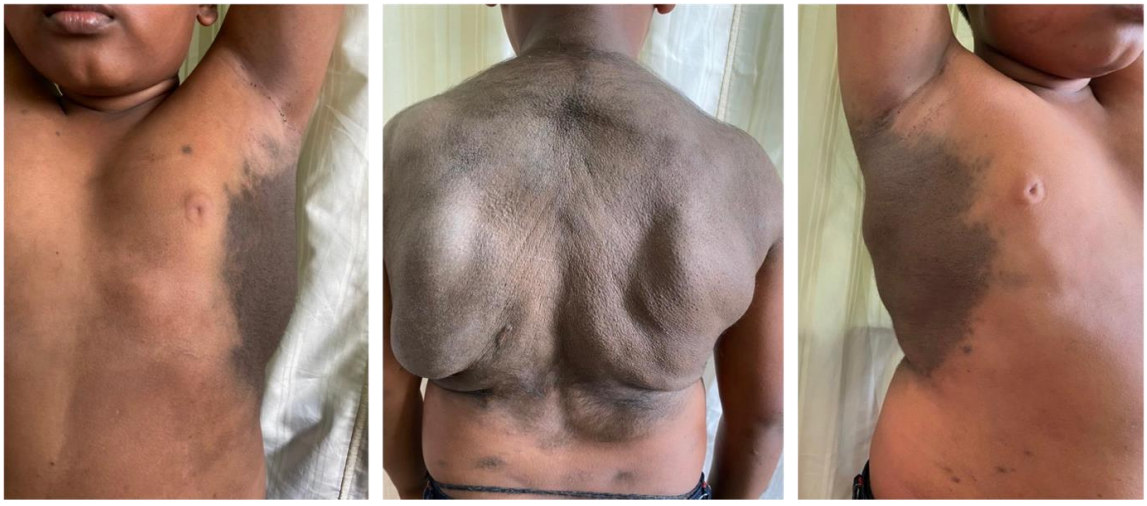

Figure 3: Giant Melanocytic Nevus over the Back Extending up to the Lateral Aspect of the Chest with a brownish-black hue and Geographical Borders with Multiple Pigmented Satellite Lesions over the Chest, Abdomen and Lower Back

Case 3:

A one-day-old male infant presented with a large pigmented patch across his body since birth. The child was delivered naturally via vaginal delivery from a consanguineous marriage there was no family history of such lesions, and the antenatal history was uncomplicated. Upon examination, a large pigmented patch was observed over the genitalia, groin, gluteal region and lower back. clumps of coarse and lusterless hair were dispersed across the back lesion, measuring 2x3 to 3x3 centimeters (Figure 4). No other congenital anomalies were detected.

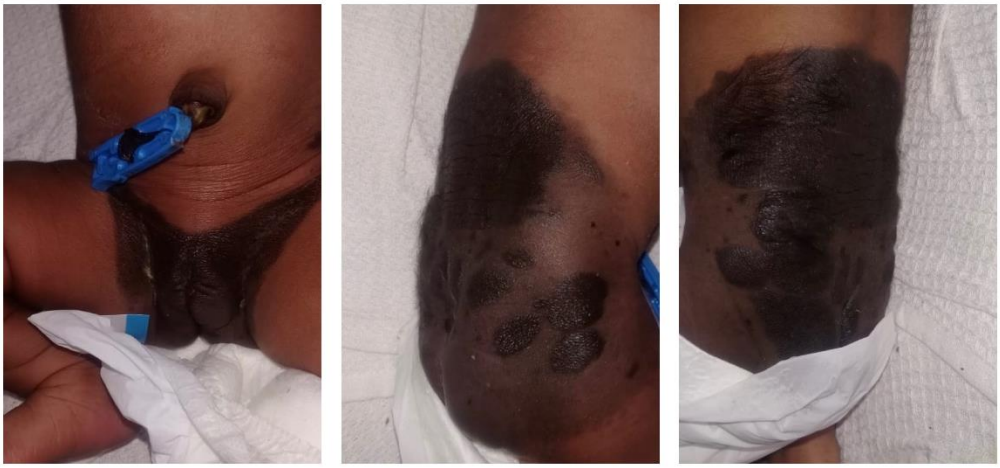

Figure 4: Giant Melanocytic Nevus Over the Genitalia, Groin, Gluteal Region and Lower Back with Tufts of Coarse and Lusterless Hair were Dispersed across the Back Lesion

DISCUSSION

Congenital melanocytic nevi, which affect around 1% of infants, are pigmented cutaneous lesions that are generated by a mix of naevus cells originating from the epidermis and dermis [7]. The occurrence of CMN at birth, its capacity to enlarge, and its potential for malignant transformation set it apart from the acquired form of melanocytic nevus. CMNs are primarily classified according to size: small, medium, large, or giant. This is determined by estimating the maximum diameter of the lesion at adult size. Projected diameters for small, medium, and large/giant CMNs are <1.5 cm, 1.5 to 19.9 cm, and >20 cm, respectively [8]. One in every 100 live births is affected by the small form, 6 per 1000 by the medium form, and one in 20,000 to 500,000 live births worldwide is thought to be affected by the large form [4, 9]. The margins of GCMN are asymmetrical, with a color ranging from dark brown to black, verrucous surfaces, and satellite lesions extending beyond the main lesion's boundaries. They could be dispersed across the entire surface of the skin. Based on their geographic distribution, these lesions are referred to as bathing trunk, coatsleeve, or stocking naevi [10]. GCMN may darken or lighten over time, become more heterogeneous or homogeneous in pigmentation, exhibit increased hair growth, develop a more uneven surface, or, less frequently, spontaneously regress. Although GCMN is mostly asymptomatic, some patients may experience xerosis and pruritus.

Patients with GCMN should be concerned about the potential for melanoma. Suspect malignancy if there is localized growth, discomfort, bleeding, ulceration, noticeable pigmentary changes, or pruritus. The likelihood of developing malignant melanoma is high.

Individuals with giant naevi are around 6% [11]. Neurocutaneous melanosis (NCM), a rare disease with fewer than 100 symptomatic instances reported in the literature, is one of the serious complications of GCMN. It is distinguished by the existence of congenital melanocytic lesions in conjunction with benign or malignant melanocytic proliferations in the central nervous system. Melanocytic cells are abundant in the leptomeninges of the brain and/or spinal cord, either in nodules or widely dispersed. An MRI or other radiographic imaging is required to assess the amount of melanocytic

deposits in the central nervous system. When the patient is between four and six months old, a baseline MRI should be taken. While the diagnosis of GCMN primarily relies on clinical assessment, histopathological examination, when feasible, is essential to confirm the diagnosis and exclude any potential malignant changes. The most typical histopathological findings in this regard are hyperkeratosis and hyperplasia, elongation of rete ridges, and increased melanocyte count [4].

The approach to managing GCMN involves providing symptomatic relief and palliative care. Treatment options for patients with GCMN may encompass both surgical and non-surgical approaches. Surgical treatment for GCMN begins at six months of age. Surgical techniques utilized in the treatment include serial excision and reconstruction with secondary skin grafting, tissue expansion, local rotation flaps, and free tissue transfer. Excisions may not completely remove the chance of developing melanoma due to the depth of some lesions, particularly if the leptomeninges are involved [12]. Recently, Er: YAG laser, Q-switched ruby laser, and carbon dioxide laser have been utilized for resurfacing and targeted management of deep pigmentations. [13]. Nonsurgical methods for GCMN include curettage, laser ablation, dermabrasion, and chemical peel [14].

CONCLUSION

GCMN is an uncommon disease with an unclear cause that may present at birth or in the first year of life. Its risk of developing malignant melanoma necessitates an accurate diagnosis and appropriate treatment. It is important to look for further congenital defects as soon as possible after birth. Furthermore, patient follow-up is a crucial component of management.

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