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A Rare Presentation of Giant Congenital Melanocytic Nevus: A Case Report

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Abstract

Background: Giant congenital melanocytic nevus (GCMN) is a rare condition present at birth, with an estimated incidence of fewer than 1 in 20,000 live births. While smaller congenital melanocytic nevi are common, GCMN poses significant clinical concerns due to its potential malignant transformation into melanoma. Early recognition and long-term monitoring are crucial to mitigate risks. **Objective:** This case report aims to highlight the clinical presentation, diagnostic approach, and management challenges of GCMN to emphasize the importance of timely intervention. **Case Presentation:** A 24-year-old male presented with a progressively enlarging blackish facial lump that bled upon scratching. Laboratory tests indicated leukocytosis, and histopathology confirmed tumor growth in the epidermis with papillomatous architecture and hyperkeratosis, leading to a diagnosis of GCMN in the fronto-nasal region. **Result:** The histopathological findings confirmed GCMN, underscoring the need for its malignant potential. Despite its benign appearance, the lesion's progressive growth and bleeding tendency warranted close monitoring. **Conclusion:** GCMN, though rare, requires early detection, appropriate treatment, and lifelong surveillance to prevent malignant transformation. Delayed intervention, often due to cosmetic concerns, may increase complications. Thus, a multidisciplinary approach involving regular follow-ups is essential for optimal patient outcomes.

Keywords: Adult, Black patch, Face region, GCMN, Giant congenital melanocytic nevus,

Case Report

INTRODUCTION

Giant Congenital Melanocytic Nevus (GCMN) is a rare dermatological condition characterized by a large, pigmented skin lesion, usually evident within the first few weeks of life or at birth. Characterized by a benign proliferation of nevomelanocytes, these lesions often exhibit a distinctive appearance, with potential features such as hyperpigmentation, an irregular surface, and, in many cases, excessive hair growth, hence the term "giant hairy nevus" (Lim et al., 2020; Machado et al., 2020).

The size of GCMNs distinguishes them from other melanocytic nevi. While smaller congenital melanocytic nevi (CMNs) are relatively common, giant forms are far less frequent, occurring in fewer than 1 in 20,000 to 500,000 live births (Singh et al., 2021). The classification into giant or large CMNs

can be seen from the adult size projection of the lesion, with GCMNs typically reaching a diameter of 20 cm or more. This size classification is clinically significant, as larger nevi carry an increased risk of malignant transformation, particularly into melanoma, and pose considerable challenges in surgical management. Additionally, the psychological impact on patients due to the cosmetic disfigurement can be profound, affecting quality of life and social interactions (Mukherjee et al., 2023).

GCMNs originate during early fetal development, between the 5th and 24th weeks of gestation, due to errors in the neuroectoderm that result in abnormal growth of melanoblasts, the precursor cells to melanocytes. The pathogenesis of these lesions is complex, involving genetic mutations such as those in the N-Ras gene, which are thought to drive the excessive proliferation of melanocytic cells. Additionally, molecular factors like the overexpression of hepatocyte growth factor (HGF) and dysregulation of proto-oncogenes such as c-met and c-kit play roles in the abnormal migration and proliferation of melanocytes. Despite some familial cases, most GCMNs occur sporadically, without a clear hereditary pattern (Belysheva et al., 2019).

The significance of GCMN extends beyond its aesthetic implications. The lesion's size and depth are not merely cosmetic concerns but are also markers of potential systemic involvement, such as neurocutaneous melanosis, where melanocytic cells invade the central nervous system, leading to severe neurological complications. In the present case, however, no neurological abnormalities or additional melanotic lesions were identified, indicating the absence of clinical features suggestive of neurocutaneous melanosis or other cutaneous melanosis–related comorbidities. Moreover, the risk of malignant transformation into melanoma is a critical concern, particularly as these lesions can be challenging to monitor and manage surgically (Kim et al., 2022). The psychosocial burden on patients and their families, stemming from the lesion's often unsightly appearance, further underscores the need for effective and comprehensive management strategies (Goil et al., 2018).

The case presented in this report is particularly noteworthy due to the unique clinical course and management challenges it posed. The patient, a 24-year-old man, had lived with a GCMN on his face for his entire life, with the lesion progressively enlarging over the years. What makes this case exceptional is the lesion's location in the nasal and frontal regions, a highly visible and functionally significant area, combined with the late onset of symptoms such as recurrent bleeding and physical discomfort. This case also presents a familial pattern of GCMN, with the patient’s mother having a similar nevus, raising intriguing questions about the genetic underpinnings of this condition.

This case highlights a rare presentation of GCMN in a high-risk facial location, emphasizing diagnostic challenges and multidisciplinary management. The patient’s late-onset symptoms (bleeding, growth) and familial history provide insights into the disease’s natural progression and potential genetic links, which are poorly understood. This report contributes to the limited literature on adult-onset GCMN complications and familial cases. It also discusses the importance of mitigating malignancy risks and improving quality of life.

This case report aims to highlight the complexities involved in managing a GCMN located in such a cosmetically and functionally critical area as the face. The unique clinical findings, including the lesion’s response to early laser treatment attempts and the challenges posed by its growth and symptomatology in adulthood, offer valuable insights into the natural history and management strategies for GCMN. Through this case, we hope to contribute to a deeper understanding of GCMN and emphasize the importance of early, comprehensive, and personalized care in improving patient outcomes (Endomba et al., 2018; Idarto, 2024).

CASE PRESENTATION

A 24-year-old male patient presented to Fatmawati Hospital with a primary complaint of a blackish facial lump that had been present since birth and exhibited bleeding when scratched. The lesion had progressively enlarged over time, causing increasing discomfort. The patient's mother reported attempting laser treatment when the patient was 6 months old at RSPAD Hospital, but the procedure was not approved by the attending physician. Consequently, the lesion remained untreated throughout childhood and adolescence.

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The patient reported significant psychosocial distress due to the visible nature of the lesion, particularly as it became more problematic in adulthood with recurrent bleeding episodes. Initial consultations at local clinics and Aulia Hospital resulted in referrals to Fatmawati Hospital for specialized care. The patient's family history revealed that his mother had similar lesions on her buttocks and thighs, suggesting a potential hereditary component. No history of asthma, allergies, hypertension, or diabetes mellitus was reported in the family.

Physical examination revealed a hyperpigmented, verrucous mass measuring 15 × 7 cm located in the fronto-nasal region. The lesion had well-defined borders and a bumpy surface texture. Laboratory tests revealed leukocytosis (19,700/µL), microcytic anemia (MCV 75.9 fL, MCH 24.3 pg), elevated absolute neutrophil count (90%), and increased lactate level (5.1 mmol/L). Other hematological parameters were within normal limits: hemoglobin 13.3 g/dL, hematocrit 41.6%, erythrocyte count 5.48 million/µL, and platelet count 240,000/µL. Electrolytes were normal (Na 137 mmol/L, K 4.3 mmol/L, Cl 101 mmol/L). Coagulation studies showed mildly shortened APTT (25.3 seconds; control 36.2), PT of 13.8 seconds (control 15.7), INR 1.00, fibrinogen 429 mg/dL, and D-dimer 319 ng/mL. Liver enzymes were mildly elevated (AST 40 U/L, ALT 57 U/L), with normal albumin level (4.06 g/dL) and random blood glucose of 142 mg/dL.

Histopathological examination of an 8.5 × 3 × 0.5 cm excised tissue specimen demonstrated papillomatous epidermal hyperplasia with hyperkeratosis. The specimen contained proliferating nevus cells with uniform, round-to-oval nuclei and cytoplasmic brown melanin pigment, interspersed with skin adnexal structures such as sebaceous glands and hair follicles, as well as cartilage tissue. Microscopy demonstrated a diffuse dermal melanocytic proliferation extending from the papillary into the deep reticular dermis, without a distinct grenz zone. The melanocytes were arranged in nests and sheets, with some dissecting between collagen bundles. In addition, melanocytic proliferation is frequently centered around pilosebaceous units, forming dense aggregates in close association with adnexal structures. These architectural and cytological features confirmed the diagnosis of Giant Congenital Melanocytic Nevus (GCMN) of the fronto-nasal region.

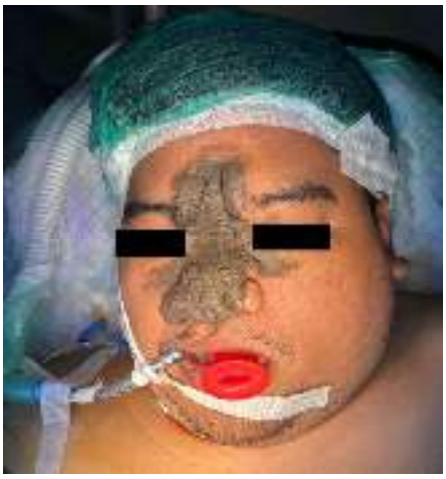

Figure 1. 15x7 cm lesion with a clearly defined bumpy surface on the fronto-nasal region (front view).

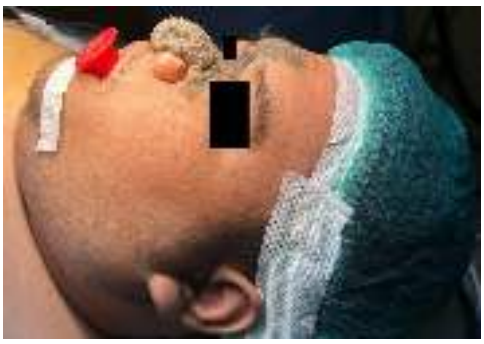

Figure 2. 15x7 cm lesion with a clearly defined bumpy surface on the fronto-nasal region (left view).

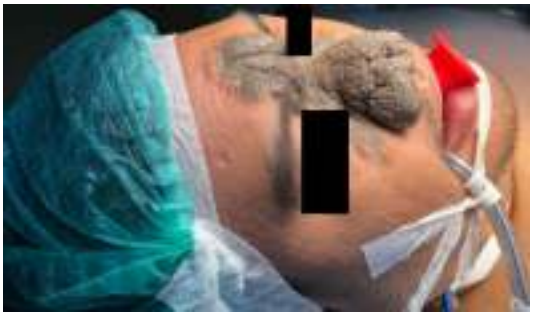

Figure 3. 15x7 cm lesion with a clearly defined bumpy surface on the fronto-nasal region (right view).

The patient was evaluated by a multidisciplinary team, including plastic surgery and oncology specialists, who recommended surgical excision. Preoperative antibiotics were administered due to the observed leukocytosis. Postoperative recovery was uncomplicated, and the patient was advised to undergo regular dermatologic and oncologic follow-up to monitor for potential recurrence or malignant transformation. Informed consent was obtained from the patient for publication of this case, including use of clinical photographs. Institutional ethics approval was waived for this case report.

DISCUSSION

This case report contributes to the growing body of literature on giant congenital melanocytic nevus (GCMN) by presenting a 24-year-old male with a fronto-nasal lesion exhibiting characteristic clinical and histopathological features. The findings align with existing reports while highlighting unique aspects of disease progression and management challenges. Recent case studies have documented diverse presentations of GCMN, including a 14-year-old girl with truncal involvement (Saribulan & Ennesta Asri, 2023), a 35-year-old male with abdominal lesions showing benign melanocytic infiltration (Agarwal et al., 2019), and a 6-year-old boy with extensive involvement of 20% body surface area (Merchan-Cadavid et al., 2021). Notably, our patient's late presentation with bleeding complications mirrors the malignant potential demonstrated in a 55-year-old male with metastatic transformation (Lim et al., 2020), underscoring the importance of long-term surveillance.

A melanocytic nevus is called congenital if it appears within the first year of life or at birth (Salgado et al., 2025). Congenital Melanocytic Nevus (CMN) is also known as a hairy nevus because it often has a lot of hair. CMN can change in surface, color, hair growth, and size. Nevi are small if they are 1.5 cm or less, medium if they are 1.5 to 19.9 cm, and giant if they are 20 cm or more. Patients with giant congenital melanocytic nevus may feel embarrassed due to a large pigmented spot on their face. Research shows that people with large CMN have a higher risk of cancer and a 5–10% chance of developing melanoma if untreated. Therefore, treatment is important (Reiter et al., 2022).

Lifelong surveillance of giant congenital melanocytic nevi (GCMN) is essential for the early detection of malignant transformation. Clinical assessment should focus on features such as altered pigmentation, increased thickness, nodularity, ulceration, or bleeding, as these may indicate progression. While GCMN typically undergoes age-related changes in color and size, the occurrence of pain, ulceration, bleeding, or disproportionate growth warrants immediate biopsy. Given that malignant transformation has been reported across all age groups, lifelong surveillance remains imperative. Notably, even patients who have undergone complete excision require continued dermatologic and systemic evaluation for early detection of malignant transformation (Mologousis et al., 2024).

The lesion of the patient was still small at 6 months of age, so permission was not given for laser treatment. This follows several studies stating that melanocytic nevi change in size from small to large. Research conducted by Abbott et al. (2015) reported that during 12 months of follow-up, there were 16% cases of nevi changes, with 36% experiencing diameter development. In addition, research conducted by Banky et al. also reported that out of 1000 patients, there were 329 nevi changes, with 67% of them experiencing changes in size. The latest study also found that nevi usually experience an

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increase in diameter over time, but this will decrease with aging (El-Rayes et al., 2025; Mologousis et al., 2024).

Therefore, the patient is suspected of experiencing an increase in the size of melanocytic nevi, which would cause the patient to need further management. In the Health Belief Model theory, this is related to perceived severity, which states that a person needs to seek health care when feeling a high level of severity and consequences if the condition continues. Initially appearing as flat, brown, or brownish black patches, GCMN might later rise, take on a mottled appearance, and develop a nodular surface. They are most commonly found in the trunk, then the limbs, and the head. The mechanism underlying the pruritus that patients may experience from GCMN, an asymptomatic lesion, is not fully understood; however, it is thought that local and intermittent stimuli of afferent sensory fibers, which would be caused by xerosis and hypohidrosis secondary to the functional impairment of adnexal structures like sebaceous and eccrine glands, could explain the symptoms.

Melanocytic nevi are non-cancerous skin growths. Congenital melanocytic nevi (CMN) happen due to genetic changes that cause abnormal growth of skin cells. Most CMN occur randomly, but some can run in families. More research is needed to see how often CMN becomes cancerous, though giant congenital melanocytic nevi (GCMN) may raise the risk of melanoma, estimated at 5-10% (Wei et al., 2025). The risk is higher for those with more lesions and large nevi on the scalp. However, no cancer was found in this case. GCMN can also lead to other issues like blood vessel tumors, underdeveloped limbs, and ear deformities. Patients often seek help due to concerns about their appearance as the nevi grow larger over time, which can affect their self-esteem and lead to feelings of stigma, especially if the lesions are on visible areas like the face (Verma, 2020).

This patient has no related birth defects. GCMN is a complex condition, and treatment should be customized based on age, lesion location and size, melanoma risk, possible NCM, signs of cancer in the nevus, potential functional issues from surgery, and psychological effects from CMN or visible scars. Treatment for GCMN may include clinical, psychiatric, and/or surgical options, as well as non-surgical methods. Surgical management of giant congenital melanocytic nevi (GCMN) may involve tangential excision, curettage, bloc or serial excision with direct closure, or excision with reconstruction using tissue expansion, skin grafts, or flaps. Non-surgical approaches, including dermabrasion, chemical peeling, cryotherapy, electrosurgery, and ablative laser therapy, can be considered to reduce pigmentation and improve cosmetic outcomes, although nevus cells are not completely removed. Children with GCMN are also prone to social, behavioral, and emotional difficulties, such as isolation, embarrassment, and low self-esteem, which increase the risk of depression, anxiety, and impaired social adaptation due to stigma. Thus, early interventions such as neonatal curettage within the first two weeks of life under local anesthesia, alongside psychosocial support, are recommended to improve quality of life (Dong & Wang, 2022; Soong et al., 2022). In this patient, however, no early treatment was provided during childhood or adolescence, and management consisted solely of surgical procedures, specifically Radial Forearm Free Flap and Split-Thickness Skin Graft. Changes in color, texture, or surface of the lesion should be closely monitored. Due to the size of the lesions, removal often needs tissue expanders and various surgical techniques like skin flaps or grafts (Mutti et al., 2017).

Table 1. Previous reported cases

Author	Patient	Chief Complaint	Accompanying Symptoms	Physical Examination	Additional Examination	Diagnosis	Management
Saribulan & Ennesta Asri (2023)	14-year-old girl	Large hard patches on the trunk that were blackish in colour and had not hurt or itched since birth	On the trunk, hypertrichosis, macula hyperpigmentation, and plaque hyperpigmentation were discovered.	Location with efflorescence plaques, hypertrichosis, and hyperpigmentation on the back, neck, shoulders, left arm, and lower limbs Hyperpigmented tumor on the middle to upper back	Histology shows diffusely distributed brown-pigmented nevus cells, some of which encircle the skin of the adnexa On the patient, a large congenital melanocytic nevus was noticed. The rating of the dermatology life quality index score was 10.	Giant Congenital Melanocytic Nevus	This progress could not be tracked since the patient did not show up for follow-up.
Agarwal et al. (2019)	35-year-old male	The patient had a trunk nevus since birth.	Soft tissue mass on both sides of the flanks has been gradually growing since late childhood	Bathing or garment trunk with large hyperpigmented lesions of a nevus	Histopathological examination shows that the epidermis contains lobules and nests of oval to spindle cells with brownish melanin pigment that extend deeply into the subcutaneous tissue around blood vessels.	Giant Congenital Melanocytic Nevus	Following an uneventful surgical phase, the patient was discharged.
Merchan-Cadavid et al. (2021)	6-year-old male	Hyperpigmented, elevated lesions on the back and belly that had been present since birth	This patient had no accompanying symptoms	The lesions grew into a tumoral mass that covered around 20% of his body's surface and 28% of its weight	The predominant melanocytic cell presence and architecture, which were hyperpigmented and primarily confined to the dense, reticular superficial dermis, were the characteristics of the histological pattern.	Giant Congenital Melanocytic Nevus	A good level of healing following surgery, with over 90% of the skin grafts integrated and donor areas adequately epithelized.
Lim et al. (2020)	55-year-old male	Giant CMN with satellite lesions on the lower left leg.	The giant CMN had acquired cutaneous melanoma. Local lymph node metastasis was observed.	Intradermal nevus with noticeable pigmentation was seen in the CMN area. Malignant melanoma with nodular growth was revealed by the nodular lesion.	Punch biopsy for the surrounding CMN area and incisional biopsy for nodular lesions.	Giant Congenital Melanocytic Nevus	Following a month of nivolumab treatment, there were several metastases to other organs, including the skin.

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CMN can be treated in different ways. Surgery is the most common treatment and depends on the size and location of the CMN (Yacine et al., 2022). It can be done in one session, which is easier than using lasers for smaller CMNs. However, surgery can be invasive, requires anesthesia, has infection risks, and may leave scars. Lasers are also used to treat CMN, but their effectiveness is debated. They work well for small and medium CMNs, but larger ones might need more treatment.

Several procedures, such as curettage, cryotherapy, and dermabrasion, can also be performed to treat CMN (Mateuszczyk et al., 2023). In order to take advantage of the temporary division plane between the superficial and deep dermis, curettage is typically done within two weeks of birth. Skin grafting after the treatment has been proposed as a way to reduce scarring and infection, and it has demonstrated aesthetic improvement. On the other hand, two-thirds of the 42 patients (28 out of 42) with CMN of varied diameters treated with cryotherapy in a recent research by Elmelegy (2021) demonstrated an outstanding response. Dermabrasion can also be performed to remove the nevus but not to treat hypertrichosis (Hafezi et al., 2021).

The main limitation of this report is the absence of microscopic photographic documentation of the histopathological specimen, which may restrict independent appraisal and slightly undermine the robustness of the findings. Although the pathology report demonstrated features consistent with giant congenital melanocytic nevus (GCMN), future case reports should include histopathological images to provide stronger visual corroboration and enhance diagnostic transparency.

CONCLUSION

A giant congenital melanocytic nevus is an uncommon occurrence presenting at birth or shortly thereafter with massive hyperpigmented lesions, often exceeding 20 cm in diameter. This case highlights the clinical challenges of giant congenital melanocytic nevus (GCMN), particularly when involving cosmetically sensitive facial regions. Key findings confirm the progressive nature of GCMN and its significant psychosocial impact, reinforcing the need for early multidisciplinary intervention. Due to its rarity and associated stigma, many patients delay seeking treatment despite its malignant potential. Future research should optimize surgical approaches, monitoring protocols, and targeted therapies while improving awareness among healthcare providers.

CONFLICT OF INTEREST

The authors declare no conflicts of interest.

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