Bulky Perineal Naevocytoma with a Rare Variant of Giant Congenital Melanocytic Nevus in a Female Neonate

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ABSTRACT

Background: Congenital melanocytic nevi are common and vary in size. Some can be quite extensive but are usually benign. However, a giant congenital nevus is rare and has the potential for malignant transformation. Giant nevus could occur in any part of the body; though it is most common on the trunk, it rarely covers over three-quarters of the skin. It is known to be associated with other congenital malformations such as spina bifida and foot deformity. Very few cases of bulky perineal naevocytoma associated with giant congenital melanocytic naevi have been reported in the literature. **Case summary:** We report a case of a 2 day old female neonate with a giant melanocytic nevus that covers about 85% of the whole body associated with a bulky perineal naevocytoma arising within the lesion. **Conclusion:** Such extensive lesion is at high risk of malignant transformation and has a major psychosocial impact on the caregivers.

Keywords: perineal naevocytoma, congenital, melanocytic, nevus, neonate.

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Introduction

Congenital melanocytic naevi are brown or black moles present at birth in 1 - 2% of newborns or may develop in the first year of life.[1,2] They vary in size and are formed by the overgrowth of the melanocytes. Giant congenital melanocytic nevus (GCMN), giant hairy nevus or nevocellular nevus represents a special group of melanocytic lesions that are rare in newborns (1:500,000 newborns). These generally cover large areas of the body [>9cm in neonates] exceeding 2% of body surface area and are usually seen in the lower trunk, lower limbs, scalp, and neck but may involve any skin surface.[3] The GCMN is known by various names such as; bathing trunk, cape, vest, coat-sleeve or stocking naevus, depending on regional distribution.[4]

Various patterns and extents of melanocytic naevi have been reported in the literature. Based on this, *Martins de Silva* and Colleagues observed that all reported patterns fit into six groups. The "6B" rule was suggested by the workers as follows: 1. Bolero (upper back), 2. Back, 3. Bathing trunk, 4. Breast/Belly, 5. Body extremity, 6. Body. If satellite lesions are excluded.[5]

Despite its low incidence, GCMN is important because of the severe complications that may develop. These include malignant transformation, central nervous system involvement and significant psychosocial impact on the family, especially in those with large lesions.

The potential risk for developing malignant melanoma in infancy or childhood often involves scalp or posterior axial distribution and is reported to be about 5-15%.[3,6] Neurocutaneous melanosis (NM), characterised by the proliferation of melanocytes in the central nervous system, is seen in about 3-12% of patients before the age of 2 years.

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When NM occurs, it has an unpredictable course and a poor prognosis. The GCMN may be associated with neural tube defects, foot deformities, and hypertrophy or atrophy of a limb affected by the nevus. There are very few reports of bulky perineal naevocytomas and to the best of our knowledge, none has been reported in a neonate.[7,8] The presence of additional congenital malformation(s) can lead to social stigma on the patient and the family.

We report a 2 day's old female neonate with a very rare GCMN with satellite naevi and bulky perineal naevocytoma.

Case report

BAF, 2 day old female neonate was seen at Federal Medical Centre, Nguru, Yobe State in December 2018. She was delivered via a normal vaginal delivery at term in a non-consanguineous marriage to a 30 year old multigravida. The pregnancy was supervised with uneventful antenatal history; the mother was not a known diabetic and did not take alcohol. She received routine ante-natal care and had no history of use of unprescribed medication; no previous congenital malformation in any family member.

The patient presented with a hyperpigmented patch all over the trunk, head, neck and most of the limbs at birth, sparing the face and distal extremities (Figure 1). The lesion on the trunk has a verrucose surface but is not hairy. Some satellite lesions were found on the forehead and left cheek in what was left of normal skin (Figure 2). The extensively pigmented patch covers approximately 85% of the entire skin surface, sparing only part of the face, right wrist and hand (Figure 3). She has normal weight, Length and occipitofrontal circumference.

There was an associated spherical mass arising within the lesion at the perineum protruding through the vulva (Figure 4). The right lip of the labia was separated from the left by the vagina; the left vulval lip appears grotesque. The child had a normally sited urethral opening and was making urine normally. She passed meconium within 24 hours of life and the anus is puckered. No other abnormal opening was seen in the perineal area, and the beddings were not soiled with faeces. No gonadal mass was palpated in the perineal area.

She has rocker-bottom feet bilaterally, but no spina-bifida or asymmetry of the limbs was seen (Figure 5). The bulky, spherical, heavily pigmented mass in the left lip of labia majora measured 8cm x 6cm in diameter. Ultrasound scan of the perineal mass revealed a homogenous mass of soft tissue density measuring 47.2 by 41.1mm in diameter with no intra-abdominal extension/connection (Figure 6). She had normal visceral situs, the uterus and both kidneys are normally situated and appear normal.

The histology of the biopsy of the skin showed a compound naevus with intradermal and junctional activity and deep tissue involvement (Figures 7 and 8). In addition, the presence of melanocytes surrounding sweat glands further demonstrates adnexocentricity (Figure 9); which is consistent with the diagnosis of GCMN. The histological section of the skin overlaying the mass shows pigmented compound naevus with junctional and dermal components. Section of the labial mass showed melanocytes in nests, cords and strands within a fibrocollagenous stroma with no atypical features; this is consistent with the diagnosis of naevocytoma (Figures 10). Thus, we diagnosed GCMN with facial satellite naevi and a bulky perineal naevocytoma.

These collagen bundles, seen in the histology of naevocytoma, however, did not fit into a row called 'Indian-file' pattern. Several slides made did not reveal mitosis or cytological atypia in the biopsy. The absence of extensive areas with a neural appearance called 'lames foliacees' and lack formation of a pseudofollicular structure limits the diagnosis of malignant transformation (Figure 10).

After the diagnosis, a multidisciplinary team was assembled to develop and implement the care plan. The parents were counselled on the need for staged surgeries to excise the lesion and the need for rotation flaps and have been prepared for the rigours of several follow-up visits anticipated in the provision of multidisciplinary care.

Figure 1: Naevi on the head, trunk and limbs with verrucose surface.

Figure 2: Naevus on the scalp and cheeks with satellite lesions on the face

Figure 3: Naevus involving most the left hand.

Figure 4: The bulky Vulvar mass in the perineum.

Figure 5. Rocker-bottom feet and variable involvement of both feet.

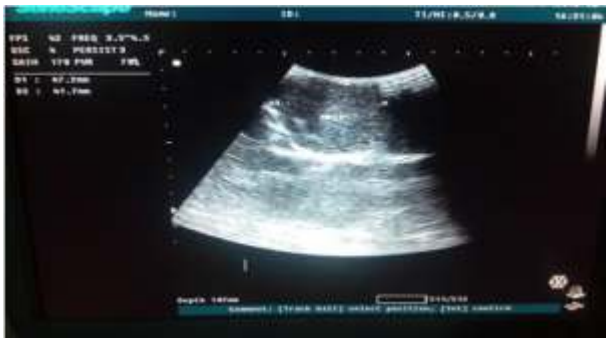

Figure 6: the Bulky perineal mass showing soft tissue consistency.

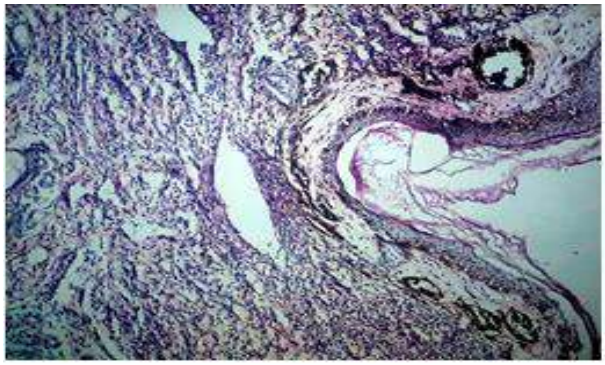

Figure 7 Junctional and interdermal melanocytes extending to subcutaneous tissue

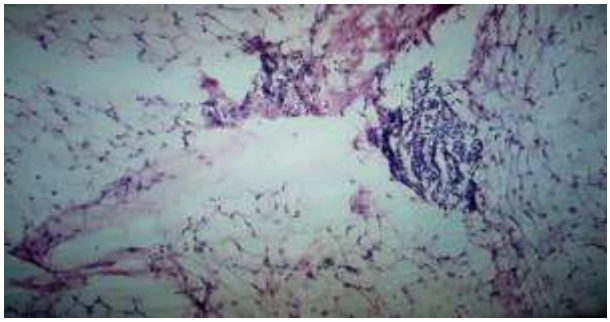

Figure 8: Melanocytes infiltrating fibrous septae of subcutaneous adipose tissue

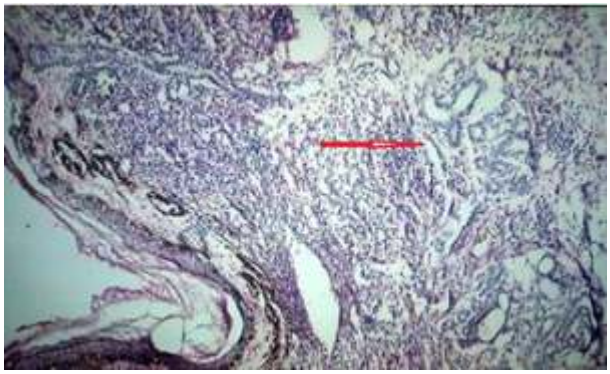

Figure 9. Adnexocentricity; Melanocytes surrounding sweat glands (Red arrow)

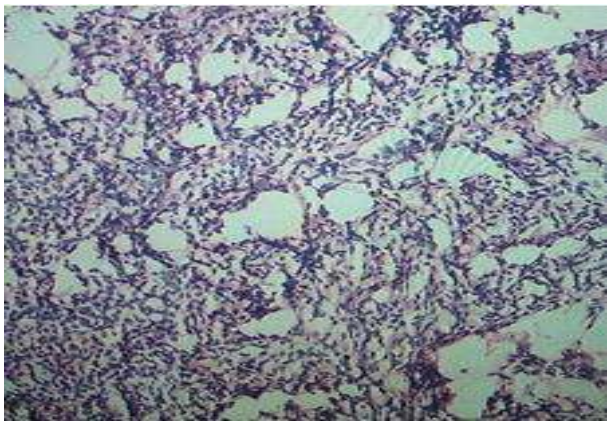

Figure 10: Melanocytes in nests, cords and strands within a fibrocollagenous stroma.

The costs of investigations were borne by the authors due to socioeconomic constraints in the family. The patient was discharged after two weeks to enable the family to prepare for the commencement of serial skin grafting. Unfortunately, we lost to the patient to follow up and therefore, further care could not be instituted.

Discussion

Giant congenital melanocytic nevus mostly involves the lower back and thighs, but our patient had it on about 85% of the body surface area. The only body part spared is the distal half of the right forearm, part of the soles of both feet and the face with satellite lesions. The distribution of the lesion we described did not fit into any of the six known patterns reported but rather summated all the six known patterns of distribution described by the "6B" rule.[5] This, we believe, might be the most extensive GCMN reported in the literature.

Though the posterior neck was involved, the girl did not manifest any neurologic features and had normal (tone and) primitive reflexes. Brain MRI is essential to confirm/rule out neurocutaneous melanosis but was not available in our centre. Where available, an MRI of the brain with intravenous administration of the contrast agent should be performed in all patients with GCMN. In symptomatic patients, MRI may demonstrate thickening of the leptomeninges with strong contrast enhancement after gadolinium administration; such cases are usually fatal.[9] High urine dopamine concentration in GCMN children (especially with neurocutaneous melanosis) can indicate patients with a more serious neurological disease where neuroimaging may not be available. [9] This too, has not been assayed in the patient due to non-availability.

The combination of a bulky perineal naevocytoma and giant congenital melanocytic nevus is very rare. Huh *et al*[7] and Reyes-Mugica *et al*[8] reported 3 cases in older children, though the lesions were present at birth in two. No similar case was reported in a neonate.

Early excision with histological examination of suspicious lesions speeds up the diagnosis of melanoma, a known commonly dreaded complication.[10] Given its natural history, the sheer extent of the lesion puts the patient at high risk of malignant transformation[10], despite the lack of histological evidence for it.

Both clinical management and histopathologic interpretation of atypical proliferations in congenital melanocytic nevi pose significant challenges to dermatologists and pathologists.[5] The management plan for this patient must be uniquely designed given the extent of distribution in order to optimally achieve the two major aims of care: viz; cosmesis and malignancy risk reduction. Unfortunately, our patient was lost to follow-up.

Conclusion

Giant congenital melanocytic naevi have been rarely reported among neonates in the literature. We report a case of a 2 day old female neonate with a giant melanocytic nevus that covered about 85% of the whole body associated with a bulky perineal naevocytoma arising within the lesion. The management plan for children with GCMN must consider the extent of the lesion, risk of malignant transformation, social stigma and concurrent high costs of care, especially in a resource-limited setting such as ours.

Consent: We obtained informed consent from the parents. The hospital ethics committee of Federal Medical Centre, Nguru, approved the request to report the case.

Contributions: All the authors participated in managing the patient, writing the manuscript and agreed to take responsibility for the integrity of this contribution.

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References

1. Walton RG, Jacobs AH, Cox AJ. Pigmented lesions in newborn infants. *Br J Dermatol* 1996; 95: 289- 290.
2. J. C. Alper and L. B. Holmes, "The incidence and significance of birthmarks on a cohort of 4,641 newborns," *Pediatric Dermatology*, 1983; 1(1): 58–68,
3. Sawicka E, Orest Szczygielski O, Żak K, Pęczkowski P, Michalak E, Bekiesińska-Figatowska M. Giant Congenital Melanocytic Nevi: Selected Aspects of Diagnostics and Treatment. *Med Sci Monit*, 2015; 21: 123-132 doi: 10.12659/msm.891279
4. Caro William A. Tumours skin, In: Dermatology, Edited by Moschello SL, Pillshury DM, Hurley HJ, WB Saunders Company, Philadelphia, 1978, p. 1323-1407.
5. Martins da Silva, VP, Marghoob A, Pigem, R, Carrera C, Aguilera P, Puig-Butill JA. *et al*. Patterns of distribution of giant congenital melanocytic nevi (GCMN): The 6B rule. *J Am Acad Dermatol* 2017; 76(4): 689-694
6. Mackie RM. Melanocytic nevi and malignant melanoma. In: Rooks textbook of dermatology, 5th edition. Eds Champion RH, Burton JL, Ebling FJG, Oxford; Blackwell scientific publications 1993; pp 1525-1560.
7. Huh JW, Yoo J, Kim MS, Choi KH, Jue MS, Park HJ. Late-onset bulky naevocytoma of the perineum masquerading as a malignant melanoma. *Clin Exp Dermatol.* 2017; 42(2):178-181. doi:10.1111/ced.12971.
8. Reyes-Mugica M, Gonzalez-Crussi F, Bauer BS, Medina-Escobedo. Bulky Naevocytoma of the perineum: a singular variant of congenital giant pigmented naevus. *Virchows Archiv A* 420(1), 87-93, 1992.
9. Bekiesinska-Figatowska M, Szczygielski O, Boczar M *et al*: Neurocutaneous melanosis in children with giant congenital melanocytic nevi. *Clin Imaging*, 2014; 38(2): 79–84
10. Ammed H, Pindiga UH, Onuora CU, Abimiku A, Dikko AA. Giant congenital pigmented naevus with unusual presentation and early malignant transformation in a Nigerian infant: Case report and review of the literature. Niger Postgrad Med J 2001; 8:26-31

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