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Case report

## CONGENITAL GIANT MELANOCYTIC NEVI – TIP OF THE ICEBERG

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### ABSTRACT

Congenital Giant Melanocytic Nevus (CGMN), pigmented lesion present since birth, occurs in 1 % of infants worldwide. Fifteen percent of CGMN are localized in the head and neck region and it can also have a bathing trunk distribution. It grows proportionally to the size of the body as the child matures and grows with variation in colour and surface texture. A 29 years old female presented to the Gynecology Out Patient Department for infertility. She also had multiple large nevi of varying sizes present since birth. The lesion was distributed all over the body. She also complained of sudden appearance of swellings at the back which were later excised and the histopathological examination showed the presence of neural nevus involving the dermis and subcutaneous tissue. This case is being reported for its rarity, the higher risk for melanoma transformation, its association with meningeal melanosis and few benign / malignant tumors.

**Keywords:** Congenital, Nevi, Melanoma,

### INTRODUCTION

Melanocytic nevus refers to any congenital/acquired lesion of melanocytes. They are common and they present with numerous clinical and histologic types. This variation in clinical and histologic presentations necessitates thorough knowledge to differentiate from malignant tumors. Congenital Giant Melanocytic Nevus, classified according to the size of the nevus,<sup>1-4</sup> are present since birth.<sup>1</sup> These pigmented lesions are to be followed up regularly because of their association with melanoma transformation, meningeal melanosis and few benign / malignant tumors.

### CASE REPORT

A 29 years old female presented to Gynecology Department for infertility. Incidentally, she brought to the notice of the gynecologist about multiple blackish lesions of varying sizes all over the body

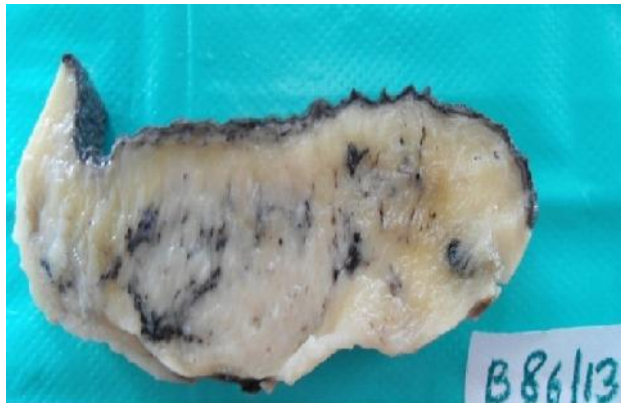
since birth. She also complained of sudden appearance of multiple nodules on her back of three months duration. On examination multiple large pigmented lesions were seen in the upper part of chest, back and face. Few areas of the pigmented lesions showed hair. The pigmented areas of the back showed three nodules measuring 5x4cm, 7x5cm and 8x5cm each with wrinkled skin (Fig 1). All other laboratory parameters were within normal limits. The clinical differential diagnosis of the nodules was neurofibroma. One of the nodules was excised and sent for histopathological examination.

**Gross** – Specimen of skin with soft tissue mass measuring 10x8x5 cm. The skin showed wrinkling and hyperpigmentation. The cut section showed many specs of black discoloration of less than 0.3 cm, involving the subcutis (Fig 2).

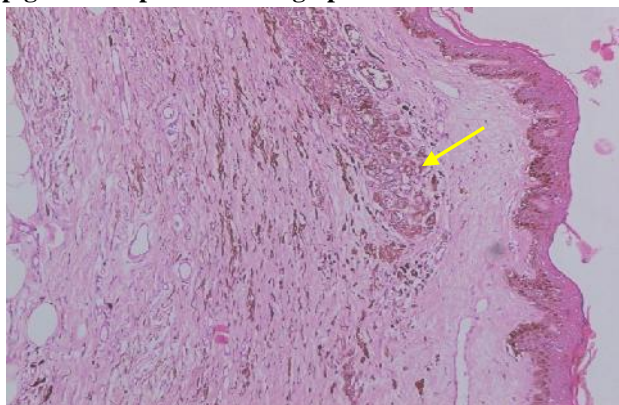
**Microscopy** -Sections showed structure of skin with dermis showing interlacing fascicles of spindle shaped cells with elongated nuclei interspersed with collagen fibers, melanophages containing melanin pigments (Fig 3). The skin appendages were also surrounded by similar cells. Melanin bleach was done and it confirmed that the pigments were melanin. Immunohistochemistry showed positivity for HMB 45 and S 100 (Fig 4). Final diagnosis of congenital giant melanocytic nevus was made.



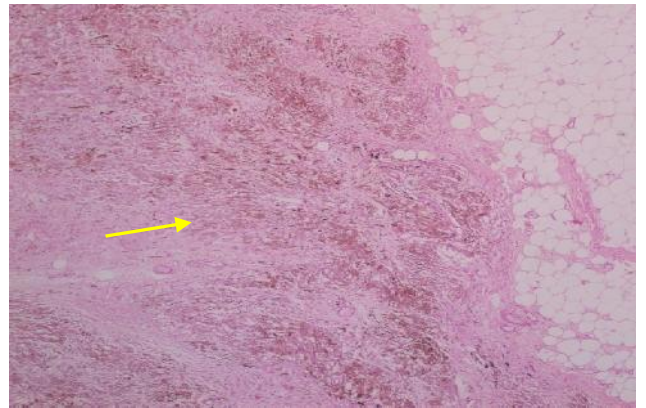
**Fig 1: CGMN involving the back, is hair borne. Nodules are seen (arrow).**



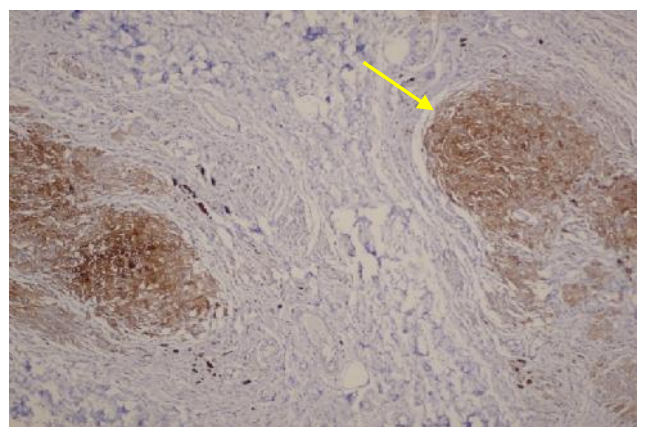
**Fig 2: GROSS- Cut section shows multiple black pigmented specs extending upto the subcutis.**



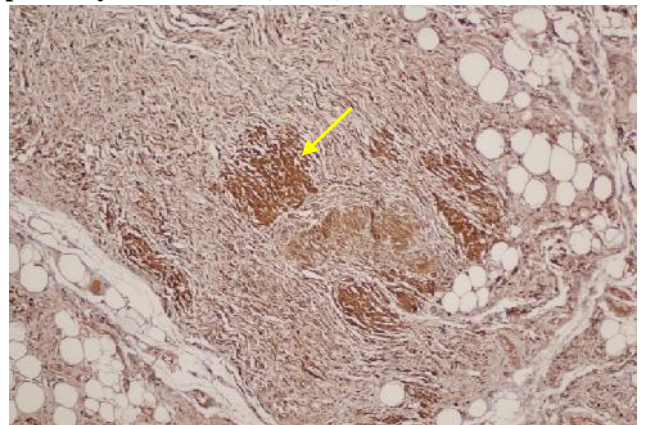
**Fig 3: a) epidermis and underlying dermis showing melanocytes (100 X)**



**Fig 3 b): shows dermis with interlacing fascicles of spindle shaped cells interspersed with collagen fibres and theques of melanocytes (40 X)**



**Fig 4: Immunohistochemistry – a) Melanocytes showing positivity for HMB 45 (100 X)**



**Fig 4: IHC – b) Melanocytes showing positivity for S100 (100 X)**

## DISCUSSION

Melanocytes are of neural crest origin and they migrate along nerves that emerge from the spinal cord and merge with the skin. As they reach the skin, they spread out evenly among the epidermal cells. These melanocytes produce pigments, which protect the skin from damage by the ultraviolet rays from the sunlight. Congenital Melanocytic Nevi (CMN)

reflects a failure of the normal process of migration of melanocytes into the skin. Instead of spreading out evenly into the skin, many cells collect at the same spot.

Majority of the CMN are sporadic as our case, but few familial cases have also been reported. Congenital melanocytic nevi are present at birth in 1% to 2% of newborns.<sup>1-3</sup> Large CGMN are rare occurring 1 in 20,000 to 1 in 500,000 newborns.<sup>1</sup>

CMN are classified according to their size, as small-less than 1.5cm, medium 1.5-19.9cm and large - more than 20 cm.<sup>1,3,4</sup> Zeal LH et al recommended defining CGMN as nevi covering 1% body surface area in the face and neck and 2% elsewhere in the body. Hence our case according to this can be classified as CGMN. The size is significant as it determines the therapeutic options and risk for malignancies.

CGMN can occur in both sexes<sup>4</sup> with a slight female predilection<sup>5</sup>. Majority of Congenital melanocytic nevi, increase in size during first trimester of pregnancy.<sup>5,6</sup>

Clinically the CMN are tan to brown, small, uniformly pigmented, flat to elevated with well defined, round borders. Giant melanocytic nevus is darkly colored and well delineated from the normal skin. Giant CMN are often covered with hair or proliferative nodules.<sup>7</sup> CGMN can occur at any site, may involve whole extremity, scalp or the trunk and may extend into the placenta. CGMN is associated with meningeal or cerebral melanosis.

The majority of these patients lead normal life without any complications. CGMN is at an increased risk for the development of melanoma and is as high as 5-7 % by age 60 years, and Arif et al suggested the incidence as 2% to 31 % for melanoma transformation.<sup>5,8</sup> In another study, 70% of melanomas occur in patients with giant CMN before puberty.<sup>5</sup> Hence there is no age limitation for the risk of melanoma transformation. The risk of melanoma may be greater in those with giant congenital melanocytic nevi with larger diameter.<sup>1,9</sup> Another study suggests multiple nevi alone or with associated posterior midline location of large congenital melanocytic nevi may be complicated by underlying cranial or spinal leptomeningeal melanocytosis.<sup>10</sup>

Crowe et al<sup>11</sup>, in their study of 223 patients with neurofibromatosis found that 3 patients had extensive CMN. Von Recklinghausen in his monograph

described 1 of 28 patients as having giant CMN. Few benign conditions like diffuse lipomatosis, hamartomas, hemangiomas, lymphangiomas, mastocytomas, schwannomas, Von-Recklinghausen's disease, vitiligo, structural brain malformations, hypertrophy of skull bones, skeletal asymmetry, hydrocephalus are associated with CGMN. The explanation for these mixed neoplasms is that CMN precursor cell, at least in some cases, is pluripotent stem cell which has the capacity to give rise to multiple cell types.<sup>12</sup> Similarly malignant conditions associated with CGMN are neuroectodermal tumors, malignant melanoma (6% to 12%), neurocutaneous melanosis, rarely rhabdomyosarcoma. One of the syndromes known as Epidermal Nevus syndrome, also known as Feuerstein syndrome/ Solomon's syndrome, consists of extensive congenital nevi with abnormalities of central nervous system (CNS), musculoskeletal system, cardiovascular, genitourinary and eyes.<sup>12</sup>

A study of 57 patients with CMN, suggests that somatic mosaicism for NRAS codon 61 mutations in a progenitor cell within the neuroectoderm cause multiple CMN and neuromelanosis (including nonmelanocytic CNS lesions).<sup>13</sup>

Treatment is usually to obtain an acceptable cosmetic result to decrease the psychosocial inconvenience to the patient and to minimize the risk of malignancy. Curettage is an alternative to surgical excision if performed in the first 2 weeks of life.<sup>14</sup> Even after complete removal of the nevi, the risk of malignancy persists as the melanoma can occur at extracutaneous sites, especially in CNS.<sup>5, 15</sup>

## CONCLUSION

Long term follow up of the CGMN patients helps in early diagnosis of malignant melanoma and various benign/malignant tumors. Regular follow up of the patient with magnetic resonance imaging is essential for early detection of CNS complications. Anxious couples need to be counseled, as there is no risk associated with pregnancies. This case reported for its rarity and its associated complications.

## REFERENCES

1. Mary Wu Chang, Vourc'h-Jourdain M. The Risk for Melanoma in large, congenital melanocytic nevi, *J Am Acad Dermatol* 2012,

www.jwatch.org/./risk-melanoma-large-congenital-melanocytic-nevi.

2. Alper JC, Holmes LB. The incidence and significance of birthmarks on a cohort of 4641 newborns, *Pediatric Dermatology*. 2012;1(1):58-68
3. James WD, Berger TG. *Andrews' Diseases of the skin: clinical Dermatology*, Saunders Elsevier. 2006; 10<sup>th</sup> edn ; Philadelphia, pg 678-79
4. Karthik Natarajan. Congenital Melanocytic Nevi : Catch Them Early!. *J Cutan Aesthet Surg*. 2013;6(1) :38-40.
5. Bhagyalakshmi A. Giant Congenital Melanocytic Nevus of Scalp: a rare case. *International Journal of Research in Medical Sciences*. 2013;1(3):317-19
6. Timothy McCalmont, Dirk M Elston. Melanocytic Nevi. Updated Jun 3, 2013, [emedicine.medscape.com/article/1058445](http://emedicine.medscape.com/article/1058445).
7. Price HN, Schaffer JV. Congenital Melanocytic Nevi-when to worry and how to treat: Facts and controversies. *Clinics in Dermatology*. 2010;28(3):293-302
8. Arif Turkmen, Ebopras, DaghanIsik, Mehmet Bekerecioglu. Comparison of Classification Systems for Congenital Melanocytic Nevi. *Dermatologic Surgery* 2010;36:1554-62
9. Hale EK, Stein J, Ben- Porat L. Association of melanoma and neurocutaneous melanocytosis with large congenital melanocytic nevi- results from the NYU-LCMN registry. *Br J Dermatol*. 2005;152(3): 512-17
10. Lovett A, Maari C, Decarie JC. Large congenital melanocytic nevi and neurocutaneous melanocytosis: one pediatric center's experience. *J Am Acad Dermatol*. 2009;61(5):766-74
11. Crowe FW. A Clinical, Pathological and Genetic study of multiple Neurofibromatosis. Springfield, IL, Charles C Thomas, 1956, p 181.
12. Fitzpatrick's *Dermatology in General Medicine*. Klaus Wolff. Melanocytic tumors, Benign neoplasia and hyperplasia of melanocytes, 1;7th ed: C1099-1101.
13. Veronica A. Multiple Congenital Melanocytic Nevi and Neurocutaneous Melanosis Are Caused by Postzygotic Mutation in codon 61 of NRAS, *Journal of Investigative Dermatology*. 2013;133, 2229-36
14. Linda E . De Raev. Curettage of Giant Congenital Melanocytic Nevi in Neonates. *Arch Dermatol*. 2002;138(7) 943-47
15. Marghoob AA, Schoenbach SP, Kopf AW. Large Congenital nevi and the risk for the development of malignant melanoma. *Arch Dermatol* 1996;132:170