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Case Presentation

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Giant Congenital Melanocytic Nevus Associated with Subcutaneous Neurofibromas

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Abstract

Giant congenital melanocytic nevus (GCMN) is rare and occurs in 1/20,000-500,000 births and is usually associated with other smaller CMN. There is a significant association between bathing trunk nevus and neurofibromas and Neurofibromatosis of type 1. We describe a case of bathing trunk nevus in a twenty-year-old Korean boy associated with two fixed, hard and greyish plaques one localized in his right hip and the other in interscapular region. Histopathological examination of the biopsy taken from one plaque revealed features of subcutaneous neurofibroma. As far as we know in the current literature this variant of neurofibroma is not reported in association with GCMN. We have excluded the diagnosis of NF1 since we didn't find other clinical features of this syndrome. In addiction the nuclear magnetic resonance (MRI) with gadolinium contrast has excluded the neurocutaneous melanosis (CMN) and spina bifida occulta that could be associated with GCMN. We report this case for his peculiar clinical features in particular related to the unusual manifestation of subcutaneous neurofibromas.

Case History

We present a case of twenty-year-old Korean male who was referred to our department for evaluation of a bathing trunk nevus: a Giant Congenital Melanocytic Nevus (GCMN) localized from upper back down to lumbar region, buttocks and extended abdominal region, in particular on the right side **(Figure 1 and 2).** The lesion was present since birth and it showed continuous growth during childhood. He was otherwise healthy and there was no family

history of similar lesions. Physical examination revealed a brownish lesion with well-defined margins and maximum diameter more than 20 cm **[1,2].** The lesion showed multiple darker areas, some covered with darkly pigmented terminal hairs. Several satellite lesions were also distributed over the arms, lower trunk, thighs and legs. The patient also showed two additional lesions on his right hip and on the interscapular region. The lesions were fixed, hard in consistency and greyish plaques. The one localized on the hip was inside an area of vitiligo **(Figure 3).** No other spots of depigmentation were detected. No pigmented mucosal lesions were present. His general physical and systemic examination was within normal limits. A videodermatoscopy of nevus showed a homogeneous pattern without dermoscopic structures of malignant melanoma. Since we didn't understand the exact nature of the two addictional lesions that the patient showed we performed two deep incisional biopsies of the plaques. The histopathologic examination revealed spindle cells in superficial and mid dermis with eosinophilic cytoplasm, wavy, elongated nuclei and indistinct cell borders showing strong positive S-100 immunoreactivity consistent with diagnosis of subcutaneous neurofibroma **(Figure 4).** Although the patient didn't show any neurological signs or symptoms we have prescribed a brain and spinal Magnetic Resonance Imaging (MRI) with gadolinium contrast to detect neurocutaneous melanosis, which was negative **[1].** The Spinal MRI has also excluded a spina bifida occulta **[1].** We have looked for the presence of signs of Neurofibromatosis of type 1 (NF1): patient didn't show cafè-au-lait macules or axillary/inguinal freckling in skin and the ophthalmologic examination was within normal limits. As the pathological examination of plaques was consistent with benign lesions and the patient didn't complain any symptoms we decided not to perform surgical excision of neurofibromas.

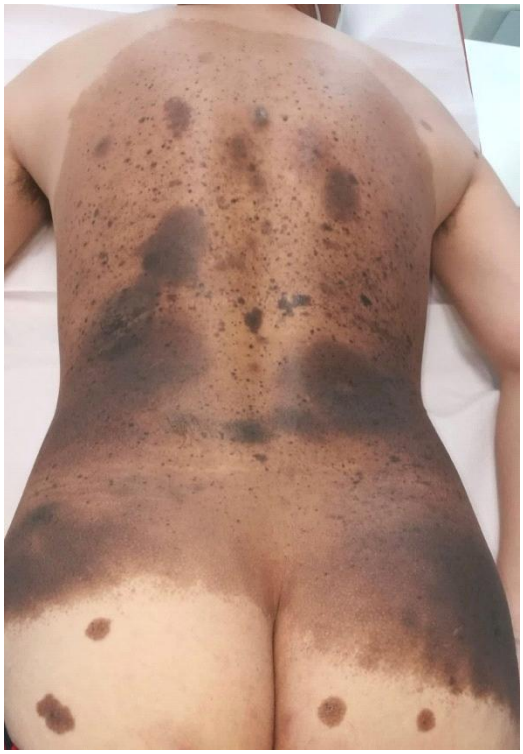


Figure 1: Bathing trunk melanocytic nevus with some darker areas and satellite lesions scattered over the skin of buttocks.

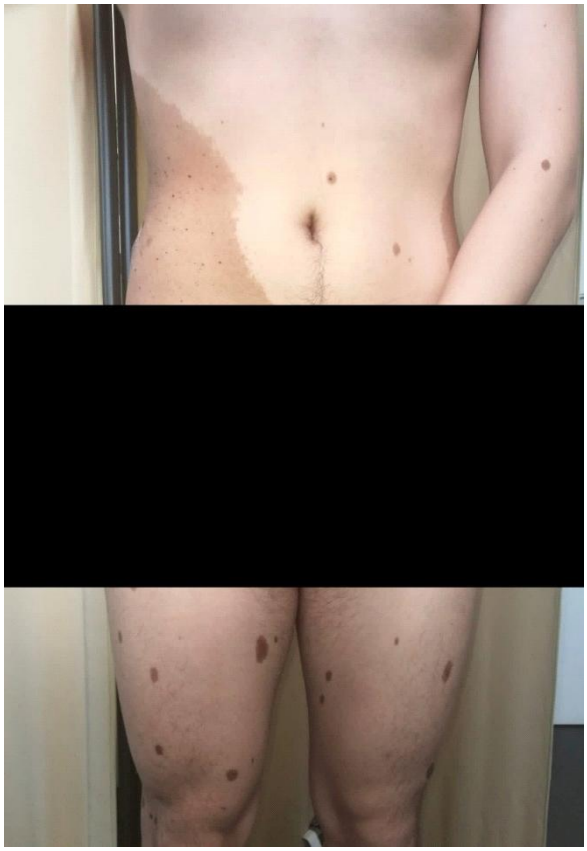


Figure 2: Abdominal distribution of bathing trunk nevus associated with satellite lesions distributed in the abdomen and upper and lower extremities.

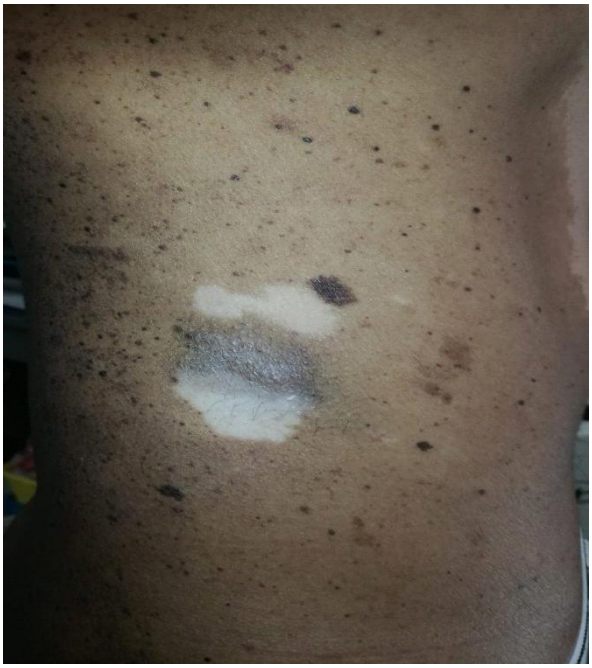
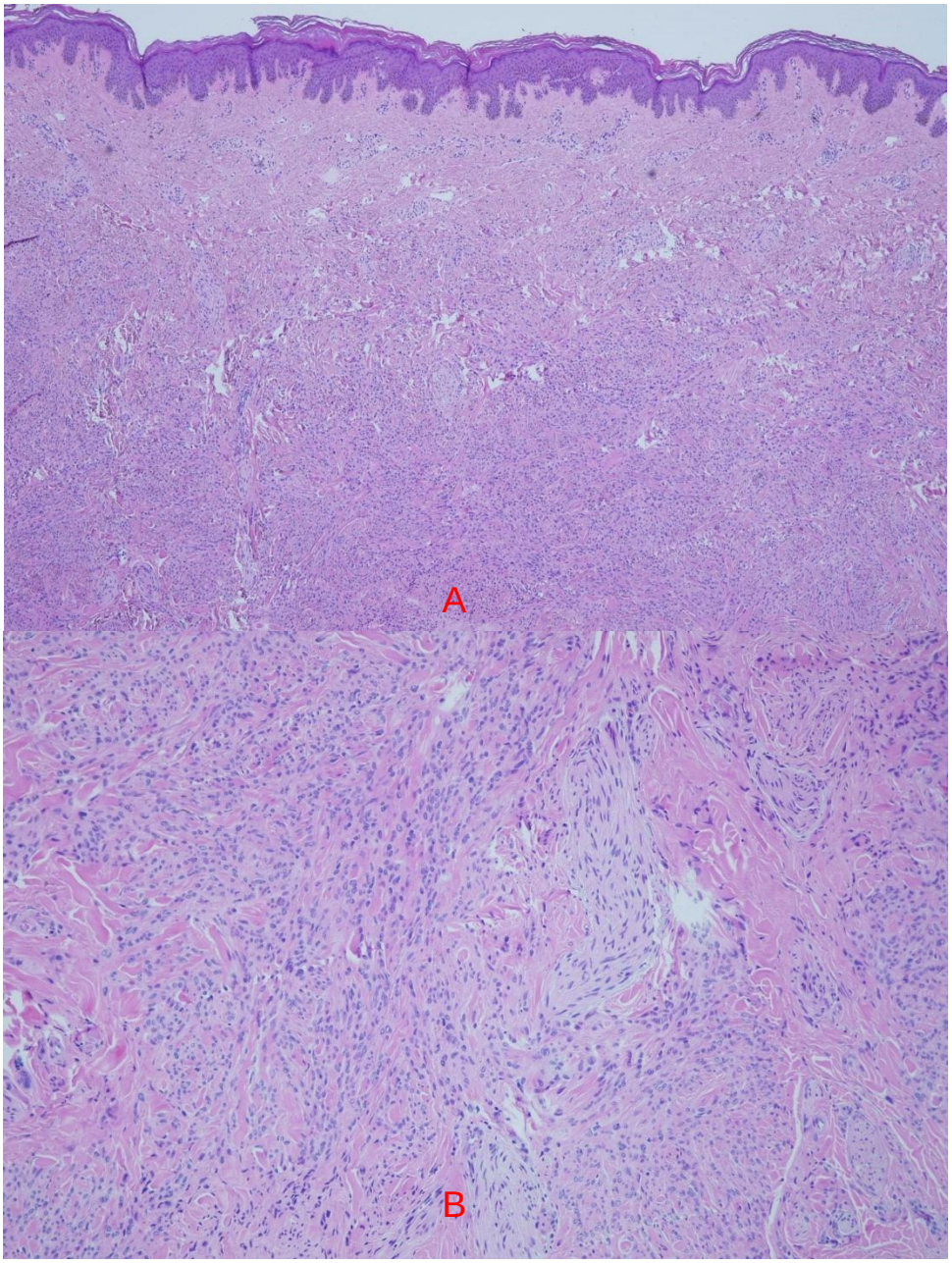


Figure 3: Greyish, hard, firm plaque within an area of vitiligo consistent with subcutaneous neurofibroma localized in the right hip of patient.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B

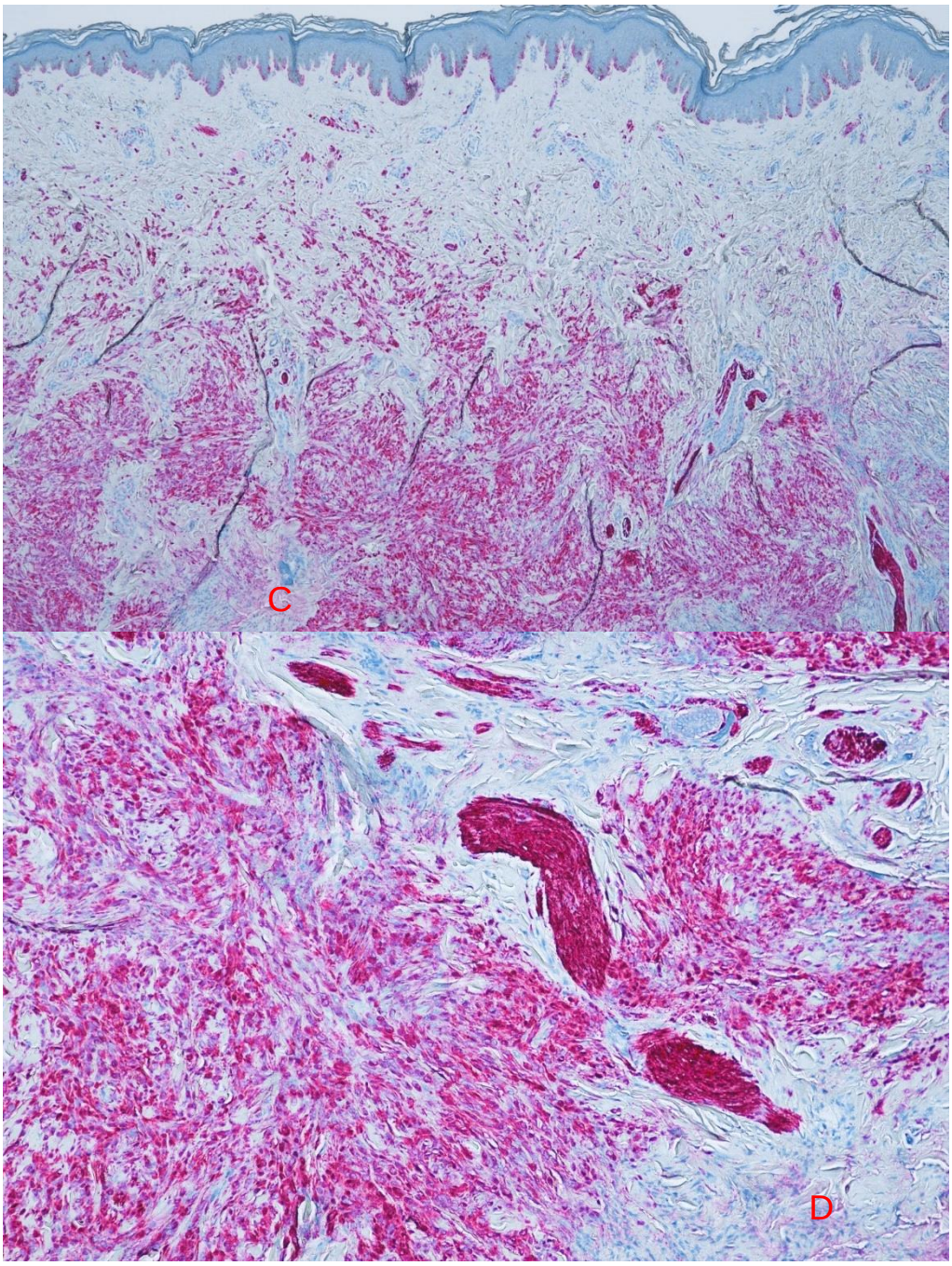

Figure 4: A, B: Histopathological examination with haematoxylin and eosin stain of a skin biopsy shows spindle cells in dermis with eosinophilic cytoplasm, elongated nuclei and indistinct cell borders. A large nerve fiber is present in the centre of field. C, D: The cells show strong positive S-100 immunoreactivity.

Discussion

Congenital Melanocytic Nevus (CMN) is usually defined as melanocytic lesion present at birth. Some authors also include in this definition melanocytic nevi that develop during the first few years of life, these are defined as tardive

congenital nevi [2]. Giant Melanocytic Nevus (GCMN) is generally defined as a congenital melanocytic lesion of 40 cm or larger [2]. GCMN can affect any region of the skin. Sometimes it shows a "garment-like" distribution. The "bathing trunk" nevus is a special type of GCMN, in the shape of "garment-like" distribution in which nevus covered the cutaneous area from upper back to the buttocks [3]. The CMN is seen in 1-6% of all live births [4]. The incidence of GCMN is estimated at less than 1:20000 live births and the variety "garment-like" is even lower: less than 1:500000 live births [4].

GCMN is associated with the possibility of malignant transformation in melanoma. Patients with GCMN and multiple congenital satellite lesions and /or GCMN with paravertebral or axial location, such as back, neck or head are associated with a higher risk of developing melanoma [5,6]. The probability that a patient diagnosed with GCMN develops melanoma is increased in childhood, before puberty [7]. However recent studies point out that the risk does not disappear during adulthood [8]. The melanoma arises within a GCMN has an unfavourable prognosis because usually occurs in the dermis or in deeper layers of skin and it makes it difficult to identify. The deeper location also facilitates earlier spread through lymphoid and blood vessels, leading to early metastasis. Additionally the rough and nodular surface of GCMN masks the early observation of the tumour [9,10].

The treatment of giant congenital nevi should be chose considering size and location of the lesions, the risk of developing melanoma and the possible functional impairments resulting from invasive procedures [1]. According to the patient, we didn't perform a surgical excision of GCMN. It was planned a clinical and videodermatoscopic follow-up, every six months [1], to detect the possible early onset of melanoma.

Another serious association with GCMN is Neurocutaneous Melanosis (NCM).

It is characterized by the presence of benign or malignant melanocytic proliferations in the Central Nervous System (CNS) associated with the occurrence of congenital melanocytic lesions in the skin. The leptomeningeal/nervous system infiltration is usually benign, more rarely may progress to melanoma [11]. The clinical findings of NCM are mostly related to increased cerebrospinal fluid pressure and the prognosis of symptomatic NCM is poor with more than half of death occurring within 3 years of diagnosis [1]. Since our patient didn't complain any symptoms we have performed a MRI to exclude an asymptomatic form of NCM that was negative.

Bathing trunk nevus may be also associated with spina bifida occulta [12]. The spinal MRI in our patient didn't show any abnormalities and we have ruled out this condition.

Some cases of vitiligo- like depigmentation have been described in nevus [13]. Our patient had developed a subcutaneous neurofibroma in area of hypopigmentation.

Several cases have described the association of GCMN with benign tumours such as neurofibroma (not only in setting of neurofibromatosis) [14-18], lipomas [15-18] and the association of the two. In all previous cases neurofibromas associated with GCMN showed different clinical aspects [14,18].

For this atypical appearance we needed to perform two biopsies in order to make the correct diagnosis. To our knowledge that aspect has never been described before, in literature, in patient with GCMN.

Since the neurofibromas are lesions related to NF1 and the possible association between GCMN and NF1 [19] we have ruled out other signs related to this condition to exclude the diagnosis.

By presenting this case we would like to help other physicians in the identification of cases of subcutaneous neurofibroma in patient with GCMN. Finally, we want to highlight the possible association between NF1 and GCMN and we recommend excluding this syndrome in patient with multiple subcutaneous neurofibromas and bathing trunk nevus.

Conflicting Interests: The authors declared no potential conflicts of interest.

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