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Case Report

Giant congenital melanocytic nevus in an 8-year-old female: a case report

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ABSTRACT

Giant congenital melanocytic nevus are melanocytic lesions secondary to abnormal melanoblast migration during the embryogenesis. The incidence reported is in 1 case for every 20 000 newborns and it is present since birth. The importance of its detection relies on the associated complications, such as melanoma development or neurocutaneous melanosis, as well as the psychological and social impact they generate; patients will require a long-term multidisciplinary attention. There is no elective treatment, management is still controversial, it must be individualized according to the characteristics of the nevus and the age of the patient. We present the case of an 8-year-old patient where the diagnosis of giant congenital melanocytic nevus is determined for the first time.

Keywords: Metastatic melanoma, Giant congenital melanocytic nevus

INTRODUCTION

Congenital melanocytic nevus (CMV) is a benign skin lesion caused by nevomelanocytic proliferation, present since birth; it is considered giant when its diameter is greater than 20 cm; the incidence reported is in 1 case for every 20 000 newborns.[1-7]

Its etiology is still unknown; nevertheless, new studies show that there is an association with somatic mutations on genes BRAF and NRAS.[8-13] Silva et al also reported mutation in KRAS, APC, and MET.[11] Other genetic alterations described for the etiology on the giant congenital melanocytic nevus (GCMN) are multiple segmental clonal chromosomal aberrations, chimeric gene fusions and TERT promoter hypermethylation.[14]

GCMN is mostly found on the trunk, followed by limbs, head, and neck.[5,15] Due to its localization, it is also known as bathing trunk nevi.[15] Generally, on the neonate the nevus is described with a lighter color, over the years it develops hyperpigmentation and coarse terminal hair.[1,15] This lesion may acquire a cerebriform or wart - like (verrucous) appearance and is also related to ulceration.[5,16] Nodules may be observed, which lead to the benign neurotization of the nevus.[1,5]

It is described that in 80% of the cases satellite solitaire nevus are present, also, multiple may be spread across the limbs, trunk, head, and neck (or globally around the body).[5,15]

Melanoma risk development is estimated to be around 8.5–10%.[4,5,14] Risk factors include the presentation of more than 3 lesions, size greater than 20 cm and presentation age between 3–5 years.[4,5]

Patients with GCMN and multiple satellite lesions present 10–15% risk in developing melanoma.[16]

There are reports of association with neurocutaneous melanoma, where there is an accumulation of melanocytes on the brain (temporal lobe, amygdalae, thalamus, and

cerebellum).[7,17] It is observed on the magnetic resonance as a highlight of the leptomeninges, which present a thickening on the brain and the vertebral spine.[7,18] Other clinical manifestations associated are seizures, hydrocephalus, or brain malformations (like Dandy Walker disease).[4,5,7,18,19] Bhagwat et al and Sarkar et al have reported cases of GCMN associated with neurofibromas, which could be explained due to the cellular proliferation control of BRAF and NRAS on the Ras channel.[20,21]

CASE REPORT

We present an 8-year-old female who attends for the first time to a paediatric evaluation, where congenital melanocytic nevus with multiple satellite diagnosis is determined (Figure 1).

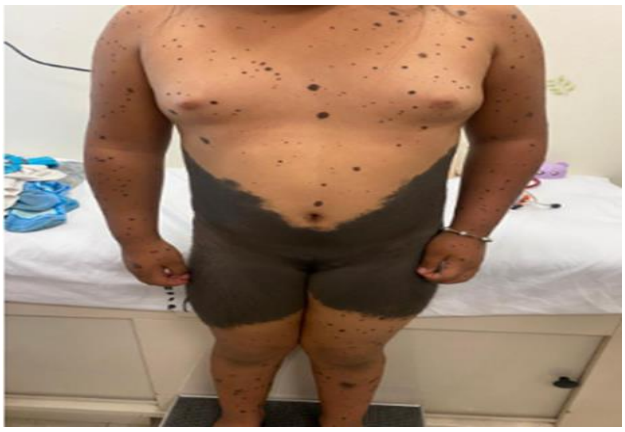

Figure 1: Giant congenital melanocytic nevus "bathing trunk nevi".

She presents an 80 cm long bathing trunk nevus distributed along the trunk, tights, and gluteus (Figure 1); along wart – like surface, the presence of nodules, (Figure 2) and multiple satellite lesions (Figure 3).

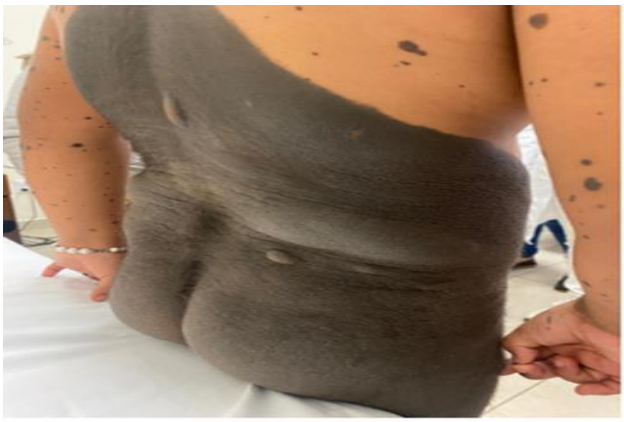

Figure 2: Color heterogeneity over the nevus extension, superficial roughness, extensive dermal nodules, hypertrichosis in the posterior midline.

She was sent to the area of oncodermatology on a third level unit, for integral follow-up due to the risk factors for developing melanoma.

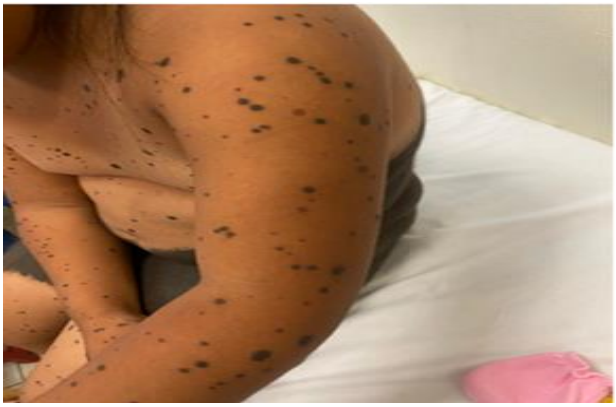

Figure 3: Multiple satellite nevi (>50 nevus), of different shade, pigmentation and extension.

DISCUSSION

Multidisciplinary management should be provided to patients with GCMN, including paediatric, dermatology, psychology, plastic surgery and neurology attention.

Treatment decisions should be individualized, depending on nevus size, clinical appearance, topography, follow up accessibility, and malignancy probability, having as objective satisfactory cosmetic results, in accordance with malignancy risk and keeping an adequate function.

Treatment options are divided into surgical excision and non-surgical excision. Non-surgical excision techniques include laser abrasion, dermabrasion, curettage, and chemical peel.[4] Reconstructive surgical techniques as part of excisional treatment have been reported since 1980.[17]

Recent studies show promising results for the treatment of GCMN and melanoma prevention with immunotherapy agents. Choi et al demonstrated in a murine model that the topical use of squaric acid dibutyl ester (SADBE) may induce a depigmentation of the melanocytic lesions, as well as bringing a protector factor for melanoma development.[2] Its mechanism of action is by macrophage recruitment. Also, the use of MEK inhibitors (binimetinib, trametinib) and PI3K (omipalisib), may induce hypopigmentation of melanocytic lesions, although no complete ablation.[2,22-24]

CONCLUSION

The GCMN is considered as a benign lesion, although, it is reported a higher risk for melanoma development, therefore, it is of vital importance to make an early diagnosis, multidisciplinary clinical approach and immediate follow up to improve life quality and expectancy.

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