Case Report: Giant Congenital Melanocytic Nevus and Neurofibroma

Sophia Sangar, BA; Preeti Jhorar, MD

David Geffen School of Medicine, UCLA[1]; UCLA Department of Dermatology, UCLA[2]

Background

- A 4 year old Caucasian male with a history of Neurofibromatosis 1 (NF-1) confirmed by mutational analysis and a giant congenital melanocytic nevus (CMN) involving the lower trunk, presented with 3 subcutaneous nodules in the region of the nevus. History was obtained from patient's mother.
- The patient's gestational history was unremarkable and he was born full term. Patient was developmentally up to date without any history of seizures or hearing impairment. MRI of the brain and spine in August 2015 was unremarkable and ruled out any neurocutaeneous melanosis.

Physical Exam

- A 4 year old Caucasian male with a history of Neurofibromatosis 1 (NF-1) confirmed by mutational analysis and a giant congenital melanocytic nevus (CMN) involving the lower trunk, presented with 3 subcutaneous nodules in the region of the nevus. History was obtained from patient's mother.
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DDx and Histopathology

- The differential diagnosis included lipoma, fibrous tumor, cyst, and nodular melanocytic growth in giant congenital nevus. Two 4 mm punch biopsies were obtained.
- Histopathological evaluation of the skin biopsy specimens revealed subcutaneous neurofibromas (figure 1 and 2) without any evidence of malignancy. Immunohistochemical staining of lesional cells was positive for S100 and patchy positive for Melanoma Antigen Recognized by T cells 1 (MART-1) but negative for tyrosinase.

Figure 1

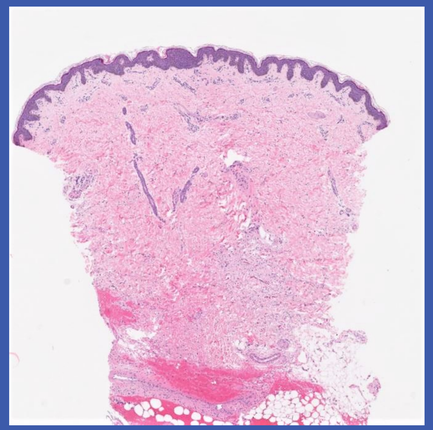
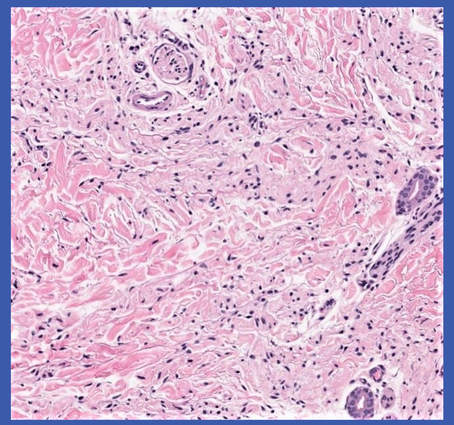


Figure 2



Discussion

- This case reports a rare occurrence of subcutaneous neurofibromas within a CMN in a patient with NF-1. The diagnosis of NF-1 is characterized by the presence of six or more café-au-lait macules, two or more cutaneous/subcutaneous neurofibromas or one plexiform neurofibromas, freckling of the groin or axillary regions (Crowe's sign), optic pathway gliomas, two or more Lisch nodules, bony dysplasia, and a first degree relative with NF1.[1]
- Previous reports have confirmed an increased incidence of giant congenital melanocytic nevi (GCMN) and melanoma in NF-1.[2,3] Giant pigment nevi are also reported to be present in up to 5% of patients with NF-1.[2] Both melanocytes and Schwann cells are derived from the neural crest lineage, which may explain their co-occurrence. Neurofibromas in NF-1 typically present as soft papules during puberty. However, there are just two case reports of subcutaneous neurofibromas developing in CMNs at an earlier age as in seen our patient, which is an unusual presentation.[2,4]
- CMN is a benign lesion and occurs in about 1%-6% of newborns. GCMN also known as bathing trunk nevus, occur in about 1 to 12,000-20,000 live births and has a multifactorial inheritance.[5,6] GCMNs increase an individual's lifetime risk of malignant melanoma, especially early in life, and neuromelanosis.[7] Greater size (diameter of 40 to 60cm), numerous satellite nevi (>20), and truncal location (2.9% on trunk vs 0.3% on head or limb) contribute to the increased risk of the malignant transformation.[6]
- GCMNs are frequently associated with benign melanocytic growths also known as proliferative nodules which may be confused clinically and pathologically with malignant melanoma.[8] When GCMS overlay the vertebral column, they can be associated with spina bifida, meningocele, vascular nevi, lipomas, Dandy-Walker malformation, arachnoid cysts, and Chiari 1 malformation.[2]