Available online at www.sciencedirect.com

ScienceDirect

journal homepage: http://Elsevier.com/locate/radcr

Case Report

Sonographic and magnetic resonance imaging findings of neurocutaneous melanosis

Yingming Amy Chen MD[a,*], *Joel Woodley-Cook MD*[a], *Michael Sgro MD*[b], *Aditya Bharatha MD*[a]

[a] *Department of Medical Imaging, St. Michael's Hospital, University of Toronto, 30 Bond Street, Toronto, ON M5B 1W8, Canada*
[b] *Department of Pediatrics, St. Michael's Hospital, University of Toronto, 30 Bond Street, Toronto, ON M5B 1W8, Canada*

ARTICLE INFO

Article history:
Received 3 November 2015
Accepted 10 December 2015
Available online 12 January 2016

Keywords:
Neurocutaneous melanosis
Congenital nevus
Neonatal brain ultrasound
MRI

ABSTRACT

Neurocutaneous melanosis is a rare nonfamilial phakomatosis characterized by large or multiple congenital melanocytic nevi plus the presence of central nervous system melanosis or melanoma. We report a case of a male infant with a giant posteroaxial nevus and evidence of intracranial melanosis on ultrasound and magnetic resonance imaging.

Case report

Clinical presentation

A male infant with normal prenatal investigations was born at term by uneventful vaginal delivery. Physical examination at birth showed a giant hairy nevus and multiple satellite nevi covering most of his back and buttocks (Fig. 1). Neurologic examinations were normal. Family history revealed large nevi in his paternal great aunt and paternal grandfather.

Imaging findings

Transfontanellar ultrasound was performed on the day of birth, which demonstrated a few small echogenic foci in the left thalamus and left choroidal fissure, without mass effect (Fig. 2). Brain magnetic resonance imaging (MRI) confirmed the presence of those foci, which were T1 hyperintense. Additional foci were noted in the right thalamus (not shown), bilateral inferior basal ganglia, and right cerebellum (Figs. 3A-C). The lesions appeared hypointense on T2-weighted and susceptibility-weighted images. There was no evidence of leptomeningeal

Disclosures: Dr. Yingming Amy Chen, Dr. Joel Woodley-Cook, Dr. Michael Sgro, and Dr. Aditya Bharatha: None.
Competing Interests: The authors have declared that no competing interests exist.

* *Corresponding author.*
E-mail address: yingming.chen@utoronto.ca (Y.A. Chen).

http://dx.doi.org/10.1016/j.radcr.2015.12.004

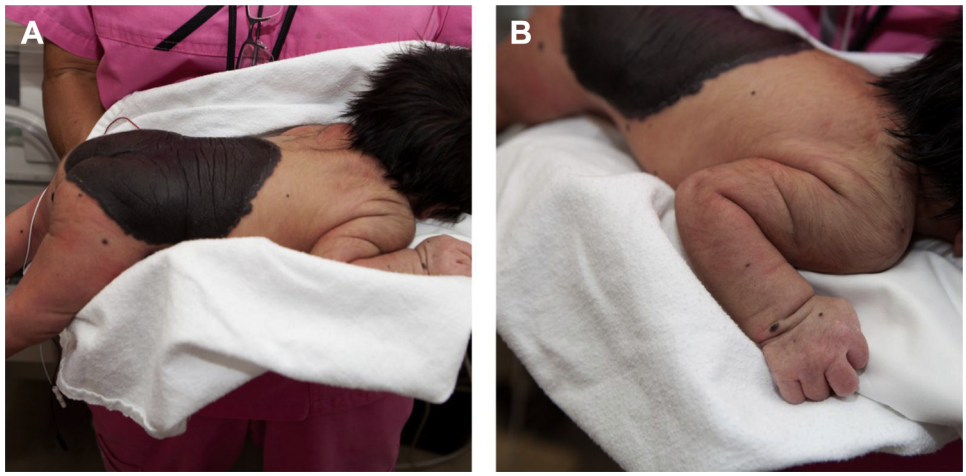

Fig. 1 – Congenital giant nevi on the back of a term baby, with dominant "cape-like" lesion on his back and buttocks (A), and multiple surrounding small lesions extending to both upper and lower extremities (B).

enhancement. Given the presence of the giant nevi, the provisional diagnosis of neurocutaneous melanosis (NCM) was established.

Outcome and follow-up

The patient remained neurologically asymptomatic at 3 months. A follow-up MRI demonstrated stable T1-hyperintense lesions (Figs. 3D-F).

Discussion

NCM is a rare nonfamilial phakomatosis first described in 1861 [1], with approximately 100 cases reported in the literature. Patients usually present with giant nevus in the lumbosacral region at birth, with normal neurologic examination [2]. Diagnostic criteria involves a large nevi (>20 cm in diameter) or multiple (>3) congenital melanocytic nevi, plus the presence of benign central nervous system (CNS) melanosis or malignant CNS melanoma [2]. Patients with large congenital melanocytic nevus in the posterior axial location [3] especially when associated with satellite nevi, are at higher risk for developing CNS melanosis [4,5]. The condition is associated with other brain abnormalities, including Dandy–Walker malformation [6–9], Chiari I malformation [10], and arachnoid cyst [10].

Normal melanocytes originate from the ectodermal cells of the neural crest and are found in the reticular formation of medulla and pons, substantia nigra, and the leptomeninges [11]. In NCM, there is an over-proliferation of melanocytes in the leptomeninges at the base of the brain [2,12], as well as in the parenchymal perivascular spaces of the anterior temporal lobes, thalami, basal ganglia, cerebellum, pons, and medulla [13,14]. The proliferation of CNS melanocytes may be either benign or malignant and remain difficult to determine even on histologic examination [15].

Despite the presence of CNS melanosis, most patients are asymptomatic at birth [4,10]. Neurologic symptoms if present, occur within the first 2 years of life, and portend a poor prognosis [2]. The most common symptoms are hydrocephalus, lethargy, seizures, and cranial nerve dysfunction. Hydrocephalus is believed to occur secondary to melanocytic

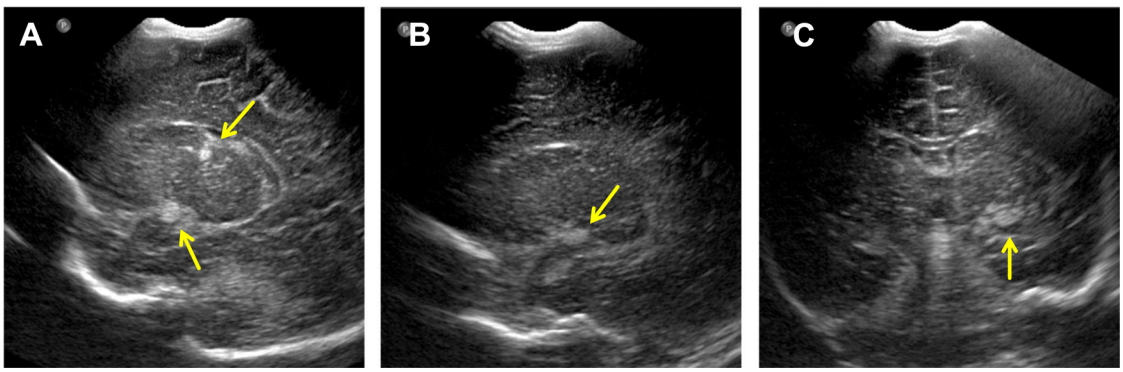

Fig. 2 – Neonatal transfontanellar ultrasound images in left parasagittal (A, B) and coronal (C) planes demonstrate echogenic lesions in the left thalamus and left choroidal fissure (arrows).