Neurocutaneous melanosis in a newborn with giant congenital melanocytic nevus

Mélanose neurocutanée chez un nouveau-né présentant un nævus mélanocytique géant congénital

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Abstract Neurocutaneous melanosis (NCM) is known as a rare phakomatose characterised by large or numerous pigmented congenital nevi associated with leptomeninges melanin-containing deposits. We report a case of a newborn presenting at birth with a giant nevus covering about 40% of the total body surface. MRI showed T1 hyperintensities in the right amygdala and predominantly in the cerebellum corresponding to melanocytic cells.

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Résumé La mélanocytose neurocutanée (MNC) est classée parmi les phacomatoses rares. Elle se caractérise par l’association de plusieurs nævi ou d’un large nævus pigmenté congénital avec des dépôts leptoméningés de mélanocytes. Nous rapportons le cas d’un nouveau-né présentant un nævus géant congénital couvrant environ 40 % de l’ensemble de sa surface corporelle. L’imagerie par résonance magnétique montre des dépôts de mélanocytes au sein de l’amygdale hippocampique droite et, majoritairement au niveau cérébelleux, apparaissant en hypersignal spontané T1.

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Introduction

Neurocutaneous melanosis (NCM) is a rare non familial syndrome, classified as a melanophakomatose with about 100 cases reported [4]. NCM also named Touraine syndrome is defined by large or multiple congenital cutaneous nevi in association with meningeal melanosis or melanoma. Giant congenital melanocytic nevi (GCMN) occur in approximately 1 per 20 000 live births, corresponding to two-thirds of NCM [2]. The incidence of central nervous system (CNS) involvement in GCMN is unknown. When the CNS is affected, the most commonly reported findings include: melanocytic accumulation in the brain (temporal lobe, amygdala, thalamus, and cerebellum) enhancement of thickened leptomeninges in brain and spine [9], associated Dandy Walker complex [10] or other brain malformations. We report a case of a child presenting at birth with a GCMN. We describe and discuss the findings on brain magnetic resonance imaging (MRI) performed 2 days after birth.

Case report

The child was born at 40 weeks by normal delivery. He was the fourth child of his family. Second trimester prenatal ultrasound showed peritoneal calcifications and no central nervous system abnormality. Because of peritoneal calcifications some more specific prenatal investigations were performed: an infection assessment resulted negative, karyotype and mucoviscidosis assessment showed no alterations. Apgar scores resulted 9/10/10 respectively at 1, 5 and 10 min. Physical examination at birth revealed a giant hairy nevus covering the back and extending to the abdomen until 2 cm to the median line. Multiple nevi were found on the proximal and distal extremities, also on the scalp and high face. The rest of the face and the neck were spared. In total the estimated surface of the nevi was 40% of the total body surface (Fig. 1). Neurological examinations were within normal limits. Biopsy of the nevus was performed at 1 day of age; it confirmed the presence of melanocytic cells without signs of malignancy (Fig. 2). Afterbirth biological assessment showed negative result. Transfontanellar ultrasound was performed at 2 days of age in routine evaluation of every newborn showed stripping aspect of thalamo-striated vessels. In order to investigate the brain and spine, MRI was required. The examination was performed on a 1.5-T magnet (Achieva R1.2; PHILIPS, Best, The Netherlands). MRI showed hyperintensities on T1-weighted images (WI), low signal on T2-WI in the cerebellum underlining the cortex (leptomeninges), bilaterally (Fig. 3a-c). In supratentorium, same signal abnormalities were found in the amygdala of the right hippocampus (Fig. 3d). On the basis of the presence of GCMN, these findings were interpreted as melanosis deposits and the diagnosis of neurocutaneous melanosis was made. No abnormality was found on the spinal cord. At 6 days of age the giant congenital nevus was treated by dermabrasion to decrease the risk of melanoma. The child went few months later to paediatric routine consultation and had normal neurological examination.

Discussion

Neurocutaneous melanosis is a rare sporadic neuroectodermal dysplasia defined by large or multiple congenital cutaneous nevi in association with meningeal melanosis or melanoma. Revised criteria for diagnosis proposed by Kadonaga and Frieden in 1991 [9] include:
• large nevi (that means > 20 cm in adults, > 9 cm on the infant scalp, or > 6 cm on the infant body);
• multiple nevi (greater than or equal to 3) [4];
• no evidence of cutaneous melanoma, except in cases where the meningeal lesions are histologically benign;
• no evidence of meningeal melanoma except in cases where the cutaneous lesions are determined benign.

Definite diagnosis is based on histological findings. In case of no histological confirmation of brain lesions, as in our case, diagnosis is considered provisional. In our case, there were numerous satellite nevi with a giant one covering the entire back, more pronounced in upper back, neck and occipital areas, a “cape-like” nevus, as in one-third of GCMN. There is a complex relationship between GCMN, NCM and malignancy. It depends partly on the location of the giant nevus, but the risk for developing NCM is unknown [4]. Hale et al. have shown that the risk for developing melanomas and NCM increases with the number of satellite cutaneous lesions [8]. Neurological manifestations of NCM occur early in life, with a median age 2 years, but can also be observed in the second decade or later; the most frequent cause of neurological symptoms is hydrocephalus and that often lead to MRI of the brain [11].

Our case is one of the earliest detection of brain lesions (day 2 after birth) reported in NCM. The MR presentation was similar to the others published; showing high signal intensities on T1 in the right amygdala (parenchymal deposition) and cerebellum (leptomeningeal deposition), which are common locations of melanin cell content deposits [2]. Leptomeningeal melanosis or parenchymal melanin deposits is T1 shortening on magnetic resonance (MR) imaging, usually seen as high signal in the convexities, subar-

Figure 3 Non enhanced MR images: axial (a) and coronal (b) T1-WI showing hyperintensities underrining cerebellar fissures, low signal on coronal T2-WI (c) (arrows), suggestive of melanocytic cells deposits in subarachnoid spaces; (d) on coronal T1-WI a hypersignal on the right amygdala indicative of melanotic cells.

Figure 3 Imagerie par résonnance magnétique sans contraste : images pondérées T1 dans les plans axial (a) et coronal (b) montrant des hypersignaux surlignant le cortex cérébelleux, apparaissant en hyposignal sur la coupe coronale pondérée T2 (c) (flèches) et suggérant des dépôts de mélanocytes dans les espaces sous-arachnoïdiens ; l’image coronale en pérération T1 montre un hypersignal amygdalien droit correspondant à des dépôts de mélanocytes.
achnoid spaces, or brain parenchyma as cerebellum, temporal lobes, pons and medulla on noncontrast images [6]. The detection of CNS melanin pigment is possible using MRI, because the paramagnetic effect of melanin leads to a decrease in both T1 and T2 relaxation times with a highly characteristic appearance on imaging [1]. The leptomeningeal involvement appears more obvious after gadolinium injection that allows seeing thickened highly enhanced leptomeninges in basal cisterns and on the surface of the spinal cord [3]. In our case contrast media was not injected because the newborn was not prepared to receive injection before MR examination. On non enhanced MRI there was no high signal on T1-weighted images suggesting involvement neither of basal cisterns nor of the spinal cord. Unless that, leptomeningeal involvement could not be definitely ruled out. For our patient a clinical and MRI follow-up was considered. The time course of this disease is ill known and may develop worse to result in neurological symptoms. Thus, repeated MR studies are recommended. The aim is to detect CNS lesions or complications, but also the malignant transformation of both cutaneous or CNS lesions [7]. This attitude is not well documented and may be debatable. In Forster et al. series, only one of the 46 non symptomatic patients with GCMN developed neurological symptoms on clinical follow-up averaging 5 years (range, 2-8 years) and, no patient has developed a cutaneous or central nervous system melanoma [5]. In symptomatic cases with malignant leptomeningeal involvement, chemotherapy is described to have a little effect on the rapid course of NCM. Shunting hydrocephalus if present could lead to dissemination of melanoma in the peritoneal cavity [11]. Dermabrasion was performed in our case to decrease the risk of malignant transformation.

In conclusion, we report one of the earliest observations of non symptomatic neurocutaneous melanoma with typical MR appearance of amygdala and cerebellum involvement, associated with a giant congenital nevus. Our report adds some more information to be considered in the spectrum of this rare disease.

References