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Neurocutaneous Melanomatosis with a Rapidly Deteriorating Course

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Summary: Neurocutaneous melanosis is a rare congenital syndrome characterized by large or multiple congenital melanocytic nevi and benign or malignant pigment cell tumors of the leptomeninges. The prognosis is extremely poor for symptomatic patients, even in the absence of malignant melanoma. We present serial MR imaging findings in the brain and spine of a child with congenital giant hairy nevi who developed progressive leptomeningeal melanomatosis and whose neurologic condition rapidly deteriorated.

Neurocutaneous melanosis (NCM) is a rare congenital syndrome. This disorder is characterized by large congenital melanocytic nevus and leptomeningeal melanocytosis. Central nervous system melanomas are present in 40–60% of patients (1). Imaging findings were previously reported in neurologically asymptomatic (2) and symptomatic (3) children. The prognosis is extremely poor when patients become symptomatic or when neurologic manifestations appear. We report a case of extensive leptomeningeal melanomatosis in a 3-year-old girl who had congenital hairy nevi. She died 7 months after the initial presentation. Although fatalities do occur within 3 years of the initial presentation, this child had a progressive course that was more rapid than usual for this particular diagnosis. Serial MR imaging of the brain showed progressive pachymeningitis. This is an important constituent of the disease and might have led to the fatal complications.

Case Report

The patient was a 3-year-old girl, the only child of her family. At birth, she was noted to have extensive hairy pigmented nevi, which affected the scalp, face, and trunk in a circumferential distribution from the abdomen to the thighs (Fig 1). She was regularly examined by pediatric dermatologists (K.-c.M.). Her early neurodevelopment had been normal. At the age of 3 years 8 months, she presented with two episodes of generalized tonic-clonic convulsions and impaired consciousness. Nonenhanced CT showed dilatation of all ventricles with a hyperat-

tenuating noncalcified lesion in the left temporal lobe. MR imaging of the brain and spine was performed with a 1.5-T unit. Images showed a hyperintense lesion in the left temporal lobe in the region of the amygdala. In addition, T1-weighted images showed hyperintensities in the adjacent leptomeninges. The hyperintensity of the lesion in the left amygdala persisted on long-TR images (Fig 2A). Diffuse leptomeningeal enhancement involving the cerebral sulci, basal cisterns (Fig 3A), and spinal cord (Fig 2B) was also present. The empirical diagnosis of neurocutaneous melanomatosis was made on the basis of the radiologic findings.

Monitoring showed that the intracranial pressure was not high (about 10 cm of water). CSF biochemical tests revealed normal glucose (4.4 mmol/L) and elevated protein (8.1 g/L) levels. The white blood cell (WBC) count in the CSF was 4×10^6 per liter. Findings from the microbiological studies for bacteria, viruses, and fungi were all negative. Results of the skin biopsy were consistent with a congenital nevus without a malignant component. No evidence of primary melanoma was found elsewhere. The patient was given a course of cefotaxime and acyclovir for empirical coverage. Phenytoin was started for seizure control. The patient's condition became stable, and she was discharged home after 1 week.

The patient was readmitted 1 week later, presenting with status epilepticus. She developed frequent complex partial seizures thereafter. The seizures could not be successfully controlled with phenytoin, carbamazepine, or valproate sodium until topiramate was included in the treatment. Melanoma cells were first detected in the CSF 2 months after the patient's initial presentation. The diagnosis of NCM was established. Despite the dismal outlook, chemotherapy (four weekly cycles of intravenous [IV] vincristine 1.5 mg/m² on day 1, IV dacarbazine 200 mg/m² for 5 days on days 1–5, cyclophosphamide 750 mg/m² IC for 2 days on days 1 and 2, and intraventricular methotrexate 10 mg once) was administered with the aim of prolonging the patient's survival. The child remained in stable condition and relatively seizure-free for the next 3 months. Three courses of chemotherapy were given. Intermediate follow-up MR imaging performed at 4 months after her initial presentation revealed an increase in extent of leptomeningeal enhancement in the brain, with obliteration of the interpeduncular and suprasellar cisterns (Fig 3B).

The child's condition started to deteriorate rapidly 5 months after her initial presentation, with an increasing frequency and duration of the seizures. A third MR study performed 6 months after her initial presentation showed diffuse leptomeningeal enhancement involving the brain and spinal cord. This was more marked when compared with the initial MR findings. Furthermore, a number of new mass lesions were present in the brain (Fig 4A and B) and in the cord (Fig 4C). The appearance of these masses was indicative of malignant transformation of the melanosis (4).

The child's clinical course rapidly deteriorated after the last MR study. The patient had repeated episodes of vomiting, increased seizure activity, and decreased consciousness. She died in the 7th month after her initial presentation. The parents refused autopsy, and it was not performed.

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FIG 1. Photograph of the child showing a bathing-trunks distribution of the giant hairy pigmented nevus.

Discussion

NCM is a rare congenital syndrome characterized by the proliferation of melanin-producing cells in both the skin and the leptomeninges. Rokitansky first described this nonfamilial neuroectodermal dysplasia in 1861 (5). The criteria for diagnosis of NCM, as proposed by Fox (6), include the following: 1) unduly large or unusually numerous pigmented nevi in association with leptomeningeal melanosis or melanoma, 2) no evidence of malignant change in any of the cutaneous lesions, and 3) no evidence of malignant

melanoma in any organ apart from the meninges. Cases with leptomeningeal involvement have a potential for malignant degeneration, with the estimated prevalence of 40–60% according to different reports (1, 6, 7). Even in the absence of melanoma, symptomatic NCM has a poor prognosis (1). Treatment such as irradiation or chemotherapy has little effect on the symptoms.

Previously, the diagnosis of leptomeningeal melanosis has been difficult to establish because malignant cells or melanin-containing cells were rarely found (1). However, with advances in imaging, the diagnosis of leptomeningeal melanosis can be made radiologically in patients with suspected NCM, especially when they become symptomatic or when they have neurologic signs such as epilepsy, hydrocephalus, cranial palsies (7–9), and myelopathy (10).

Previous reports (8) have demonstrated the usefulness of contrast-enhanced MR imaging in the detection of leptomeningeal involvement in patients with NCM. In rare cases, patients with NCM can present with intraparenchymal mass without leptomeningeal involvement (9).

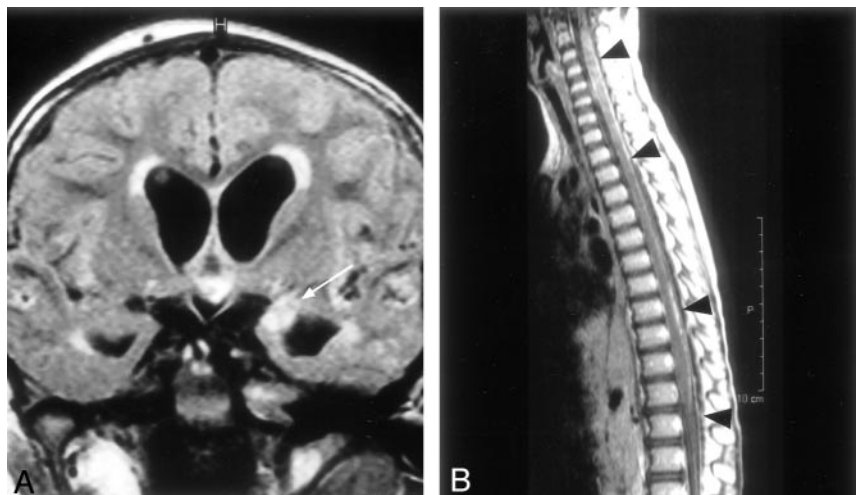
Frieden et al (2) reported that the most common MR finding in asymptomatic children with NCM was T1 shortening in the infratentorial structures rather than leptomeningeal thickening. Faillace et al (4) suggested that the malignant transformation was heralded by the appearance of intracranial or intraspinal masses or by direct parenchymal invasion. All of these features were illustrated in our single case in which serial MR images demonstrated a rapid progression of the disease along with rapid worsening of the patient's neurologic condition.

At presentation, when the child experienced her first seizure, both hyperintensities and diffuse leptomeningeal enhancement were present on T1-weighted images from the first MR study. The high signal intensity before contrast enhancement was most prominent at the amygdala in the left temporal region. This is one of the common locations of involvement in patients with asymptomatic NCM, as described in the report by Frieden et al (2). The high signal intensity is

FIG 2. MR images.

A, Coronal fluid-attenuated inversion recovery MR images (TR/TE/T1, 8000/110/2400). The lesion at the left amygdala is slightly hyperintense (arrow).

B, Sagittal contrast-enhanced spin-echo T1-weighted MR image (TR/TE, 425/14) obtained at presentation shows enhancement of leptomeninges covering the entire cord (arrowheads).



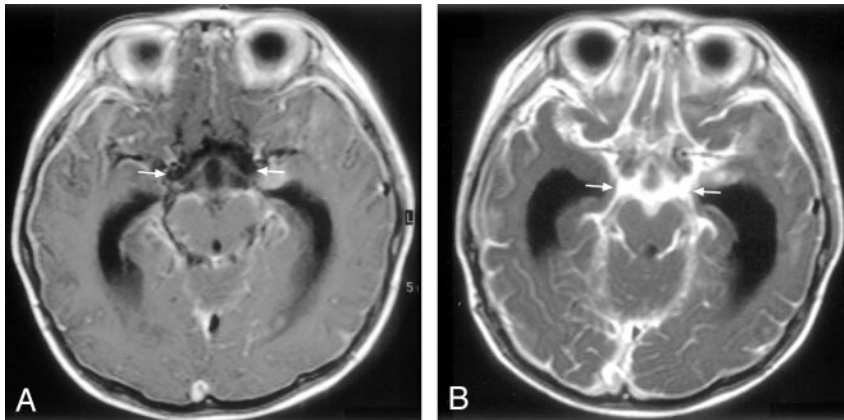


FIG 3. Axial contrast-enhanced spin-echo T1-weighted MR images (500/15). Leptomeningeal enhancement has progressed between the two studies, with enhancement of the interpeduncular and suprasellar cisterns (between arrows) after the administration of contrast material.

A, At initial presentation.
B, At 4-month follow-up.

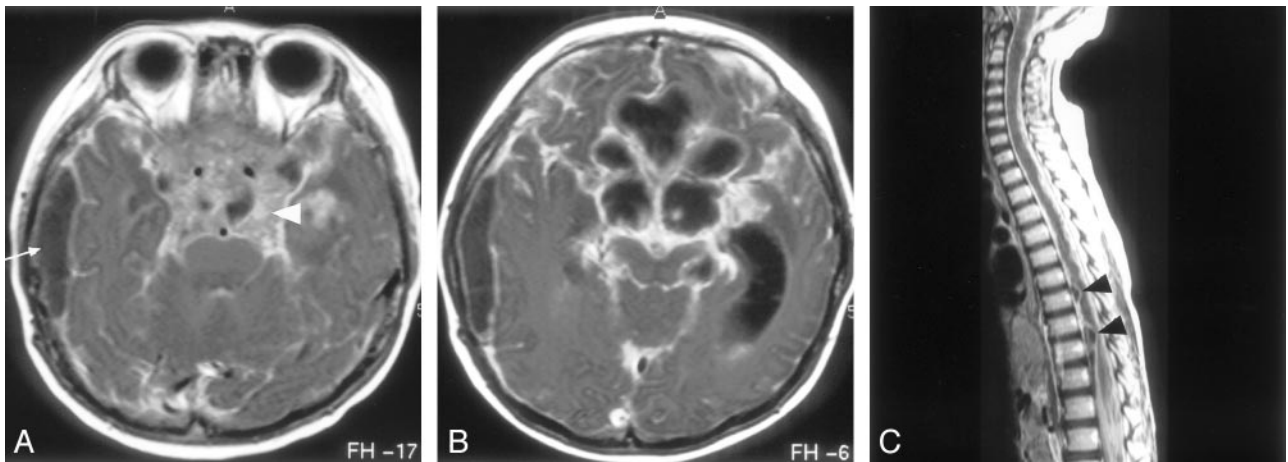


FIG 4. MR images obtained at 6-month follow-up.

A and B, Axial contrast-enhanced spin-echo T1-weighted MR images (500/15) show a new heterogeneously enhancing mass lesions (arrowhead in A) at the interpeduncular and suprasellar cisterns on top of the leptomeningeal enhancement. A subdural collection (arrow in A) is also present at the right temporoparietal region. Cystic components are present within the mass (B). These likely represent tumor necrosis.

C, Sagittal contrast-enhanced spin-echo T1-weighted MR image (425/14) shows progressive leptomeningeal enhancement with a few intradural nodules (arrowheads).

thought to be attributed to the presence of melanin, which elicits paramagnetic effects by shortening the T1 relaxation time (11). However, the presence of this hyperintensity on T1-weighted images is not universal to all patients with NCM, even those with the evidence of the proliferation of pigmented cells over the leptomeninges (8). Other reports of metastatic melanotic melanoma also fail to demonstrate the characteristic signal intensity on MR images (11). In the case that Sebag et al (9) reported, the characteristic finding of decreased T1 and T2 relaxation was present in a temporal lobe lesion. In our case, the nodule at the amygdala was hyperintense on T1-weighted images and also slightly hyperintense on T2-weighted images. Ko et al (12) has described the same observation of hyperintensity on long-TR images in the two cases. In one of the patients, histologic findings confirmed the presence of melanoma. The nature and effects of melanin on MR signal intensity in patients with NCM are complex. The increase in signal intensity on T2-weighted images in our case might be explained by the malignant transformation and the ensuing necrosis.

Our child developed a very rapid progression of the disease, with evidence of increasing extent of leptomeningeal enhancement on the second image (Fig 3B) and with multiple mass lesions in the brain and cord on the third (Fig 4). The appearance of these masses was a definite sign of malignant transformation (4), which was confirmed with the isolation of melanoma cells in the CSF. The presence of necrosis within the masses (Fig 4B and C) was additional supporting evidence for malignant transformation (3).

Involvement of spinal meninges in NCM has been reported in as many as 20% of cases (6). van Heuzen et al (10) described a focal intradural mass in the thoracic region in a patient with NCM. Diffuse spinal involvement is unusual in this diagnosis, though Rhodes et al (8) described this finding in one patient. However, no progression of the leptomeningeal enhancement in the cord was observed during follow-up imaging in that particular case.

To our knowledge, our patient had the most bizarre MR appearance among those of all the reported cases. Clinically, she also had a more rapidly progres-

sive course than that usually seen in this condition. The rapid proliferation of pachymeningitis within a relatively short period of 6 months accounts for the fast increase in intracranial pressure, the repeated episodes of vomiting, the increased seizure activity, and the state of decreased consciousness in the final stage.

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