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Age-related changes of the MR appearance of CNS involvement in neurocutaneous melanosis complex

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Abstract We report a case of giant congenital melanocytic nevi (GCMN) at risk of developing neurocutaneous melanosis (NCM) with age-related changes observable on MRI of the brain. However, although the usefulness of MR imaging in NCM is well known, age-related changes on MRI have rarely been reported. The prevalence of positive MRI findings and prognosis in GCMN accompanied by epilepsy and/or mental retardation awaits clarification. This case report may suggest the importance of serial brain MRI in cases of GCMN in assessing the risk of NCM.

Introduction

Patients with giant congenital melanocytic nevi (GCMN) are generally thought to be at a risk of developing neurocutaneous melanosis (NCM). Typical cases of NCM have been thought to develop severe hydrocephalus, papilloedema and other neurological symptoms in early childhood and also to show poor prognosis. However, it has been presumed that the majority of patients with mild-to-minimal NCM are asymptomatic [1]. The real risk of NCM for patients with GCMN remains unknown. Recently, it was proposed that MRI of the brain should be performed in cases of GCMN as a means of screening for NCM [2]. We have had the opportunity to perform MRI in a child with GCMN and observed age-related changes of the brain on MRI. This paper describes the changes observed on MRI and reviews the previous reports on the risk of NCM.

Case report

A boy was born without any difficulty. Multiple nevi, accompanied by many smaller satellites, were observed on his trunk, upper extremities, head, buttocks and right thigh at birth. When he was 4 months of age, he developed apnoeic attacks accompanied by slight cyanosis. On examination, there were no remarkable findings apart from the nevi. EEG revealed sharp waves over the right central region and over both frontal regions.

MRI of the brain at the ages of 5 months and 9 months showed multiple small lesions with shortened T1 relaxation time and isointensity on T2-weighted images (Fig. 1A, B). These findings were thought to be due to deposition of melanin because of the characteristics of the signal pattern. These lesions were located in the deep cortex of the left cerebellar hemisphere, medullary part of the right cerebellar hemisphere, the pia mater of the temporal horn tips of both lateral ventricles and the pia mater of the ventral side of the pons. Post-contrast images showed no abnormal enhancement within the lesions or leptomeninges. The results of single photon emission computed tomography of the brain and scintigraphy with ^{123}I at 5 years of age were negative.

His developmental quotient at the age of 4 years was 66. He was diagnosed as having NCM and was treated with an anticonvulsant (carbamazepine). With advancing age he developed intractable complex partial seizures, presenting as a sensation of fear, followed by loss of consciousness. MRI of the brain was performed, again at the age of 5 years (Fig. 1C). The abnormal signal in the left temporal horn had disappeared while the signal changes in the lesions at the tip of the temporal horn of the right ventricle, ventral pia mater of the pons and medullary part of the right cerebellar hemisphere had decreased. The seizures gradually came under control as the lesions changed.

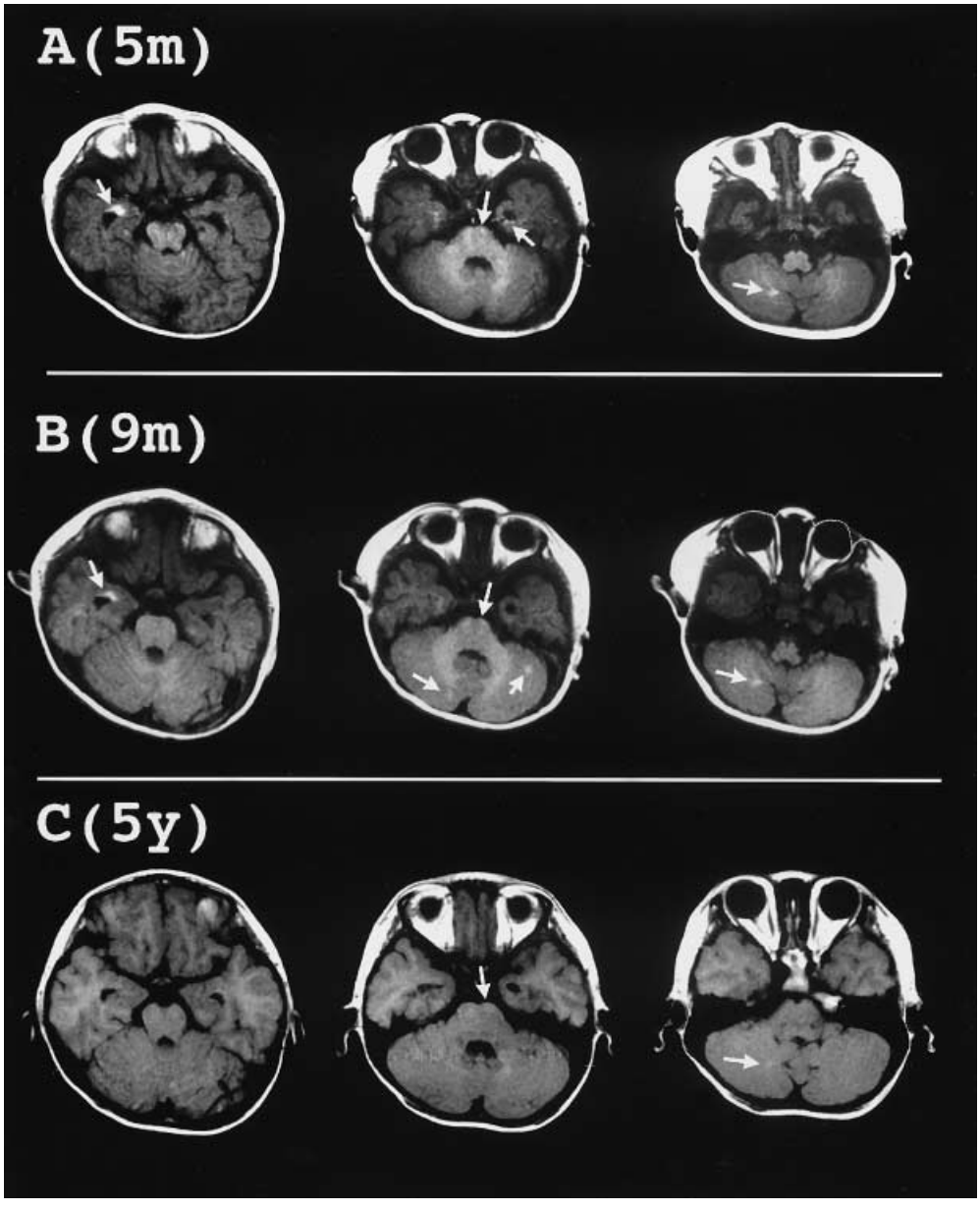

Fig. 1 A–C Axial T1-W MRI showing the locations of melanin deposition in the brain at the ages of 5 months (**A**), 9 months (**B**) and 5 years (**C**). Slices at three levels at each age are selected to demonstrate the changes in the lesions. *Arrows* indicate the sites of deposition of melanin. **A** At 5 months the lesions are located in the medullary part of the right cerebellar hemisphere, the tips of the temporal horns of both lateral ventricles and the pia mater of the ventral side of the pons. The lesions in the left cerebellar hemisphere are not detectable. **B** At 9 months of age the lesions are located in the tip of the right temporal horn, the medullary part of the right cerebellar hemisphere, the deep cortex of the left cerebellar hemisphere and the pia mater of the ventral side of the pons. **C** At 5 years there is a less intense lesion in the medullary part of the right cerebellar hemisphere. A poorly defined lesion is also seen in the pia mater of the ventral side of the pons

Discussion

Rokitanski reported the first case of NCM, an autopsy case of a 14-year-old girl, in 1861. Because in those days pathological investigation of the brain was necessary for the diagnosis of NCM, many cases of NCM that were reported had been autopsied or had (malignant) melanomas [1]. In 1991, Kadonaga and Frieden [1] revised the former criteria for the diagnosis of NCM: (1) large or multiple (three or more) congenital nevi, large being defined as equal to or greater than 20 cm in an adult, 9 cm on the scalp of an infant, or 6 cm or greater on the body of an infant, (2) no evidence of a cutaneous melanoma and (3) presence of a nevus or nevi on the scalp or neck, or in a posterior midline location. They noted that histological confirmation of CNS lesions is necessary for a definitive diagnosis and that all other diagnoses are labelled provisional.

Therefore, it was assumed that many cases of mild-to-minimal NCM were undiagnosed due to lack of histological confirmation of CNS lesions [3]. However, since the first case report of NCM diagnosed by MRI in 1991, childhood cases of mild GCMN showing abnormal MRI findings have been reported and considered to be at risk of NCM [2–4]. Frieden et al. [3] also reported 9 patients with abnormal MRI findings among 20 with

GCMN and who were at risk of NCM. However, Ruiz Maldonado et al. [5] reported no patients with abnormal MRI findings among 13 with GCMN. They attributed this difference to the different age distribution between the two studies. The average age of the patients of Frieden et al. was 8.5 months, and that in the study of Ruiz-Maldonado et al. study was 11.6 years. Ruiz-Maldonado et al. speculated that in older patients with GCMN a minimal amount of melanin may not be detected on MRI, while in newborns and infants larger amounts of melanin may be easier to detect. However, so far, such cases of GCMN have been rarely reported. Not only was there a difference in the mean age of the two groups, but there may also have been a difference in the MR techniques, which were not mentioned in detail in those papers.

Our case did not show any progressive manifestations like those severe cases described in other reports. Abnormal deposition of melanin in the meninges has not been surgically confirmed either. He was thought to be a mild case of GCMN at risk of NCM. The two lesions observed in the cerebellar hemispheres at the age of 9 months were not observed at the age of 5 months or 5 years. This may be due to different slice levels through the cerebellar hemispheres. However, the signal from other lesions in the temporal horns, the ventral side of the pons and the right cerebellar hemisphere gradually became less intense. Thus, in our case, the mechanism underlying the indistinctness of lesions on MRI may be diffusion of the melanin or further myelination of the white matter, not reduction of melanin as Ruiz-Maldonado et al. inferred. The progress of our case suggests variability of the MRI findings in NCM according to age or brain development. This observation may also explain the prevalence difference between the reports of Frieden et al. and Ruiz-Maldonado et al.

NCM is thought to be a severe disease with a poor prognosis. It has been suggested that some patients with GCMN may develop NCM. Thus the actual risk of NCM in patients with GCMN should be clarified, and it is preferable that patients with GCMN at risk of NCM undergo repeated MRI of the brain.

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