CASE REPORT

An Adult Case of Neurocutaneous Melanosis : MR Findings

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Neurocutaneous melanosis (NCM) is a rare nonfamilial disorder generally characterized by the development of congenital melanocytic nevi and benign or malignant melanotic tumors of the central nervous system. It has been postulated that this disorder represents a congenital error in morphogenesis of the embryonal neuroectoderm. Most patients have large nevi and display neurological symptoms within the first two years of life. Accordingly, when nevi are not large and neurological symptoms do not surface until adulthood, as in the case we now report, it is difficult to establish a diagnosis of NCM. In the present case, leptomeningeal melanosis manifested as diffuse hyperintensities in the leptomeninges of the cerebrum, cerebellum and the entire spinal cord on T_1-weighted MR images and as diffuse enhancement in the leptomeninges on Gd-DTPA enhanced T_1-weighted MR images. These MR findings enabled us to establish the diagnosis of leptomeningeal melanosis and demonstrate the extent of leptomeningeal involvement. Moreover, serial MR findings revealed malignant changes of leptomeningeal melanosis.

INTRODUCTION

Neurocutaneous melanosis (NCM) is a rare nonfamilial disorder generally characterized by the development of congenital melanocytic nevi and benign or malignant melanotic tumors of the central nervous system[1)~3)]. It is difficult to establish a diagnosis of NCM in patients that have nevi that are not large and in whom neurological symptoms do not develop until adulthood. We now report an adult case of neurocutaneous melanosis for which magnetic resonance (MR) examination was useful for establishing the diagnosis and in which serial MR findings demonstrated malignant change.

CASE REPORT

A previously healthy 22-year-old man presented with a 10-month history of repeated episodes of seizures and vomiting. Physical examination revealed some pigmented nevi on the buttocks, left upper arm and other sites. The nevi measured less than 10 mm in diameter ex-

Keywords neurocutaneous melanosis, malignant melanoma, MRI

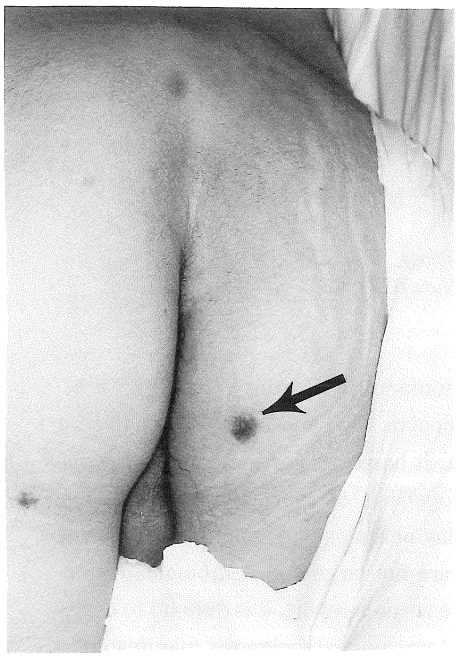

Fig. 1. There are three nevi on the buttocks. The nevus indicated by the arrow is the largest one on this patient's body.

cept for one nevus on the right buttock with a diameter of 18 mm (Fig. 1). Laboratory findings were within the normal ranges. Lumbar puncture was performed yielding xanthochroous cerebrospinal fluid (CSF) with an opening pressure of 70 mm of water. CSF examination revealed a leukocyte count of 9/mm^3, glucose of 19 mg/dl, and protein of 343 mg/dl. Cytological examination of the CSF revealed large cells with pigmented granules.

On MR imaging of the head and the entire spine (Figs. 2–4), T_1-weighted images showed mild dilatation of the lateral ventricles and diffuse hyperintensities in the leptomeninges of the cerebral cortex, cerebellar cortex and the entire spinal cord and septum pellucidum. T_2-weighted images showed diffuse hypointensities in the leptomeninges. Gd-DTPA enhanced T_1-weighted images showed diffuse enhancement in the leptomeninges. On the basis of these studies, leptomeningeal melanosis was suspected.

Upon dural opening during right temporal craniotomy, the leptomeninges appeared darkly pigmented diffusely. Cerebral and meningeal biopsies were performed. Subsequent microscopic examination of the tissue specimens thus obtained revealed a proliferation of cells, some containing melanin, but with no gross atypia in the thickened leptomeninges. These findings were consistent with benign melanosis. The patient's past history indicated that he had three nevi on the buttocks and one nevus on the upper arm at birth. Pathologic examination of a nevus from the buttocks demonstrated an intradermal benign pigmented nevus. Therefore, we established the diagnosis of neurocutaneous melanosis.

Thereafter, we observed the patient and instituted therapy with antiseizure medication. Follow up Gd-DTPA enhanced T_1-weighted images (Fig. 5) 14 months after initial presentation showed increased diffuse thickening of the leptomeninges and septum pellucidum relative to earlier studies and masses in the frontal, suprasellar region and cerebellar vallecula, upon which basis we suspected malignant change of leptomeningeal melanosis. After 25 months from the onset of neurological symptoms, the patient developed respiratory failure and then died, the cause of which we suspect to be increased intracranial pressure. Upon au-

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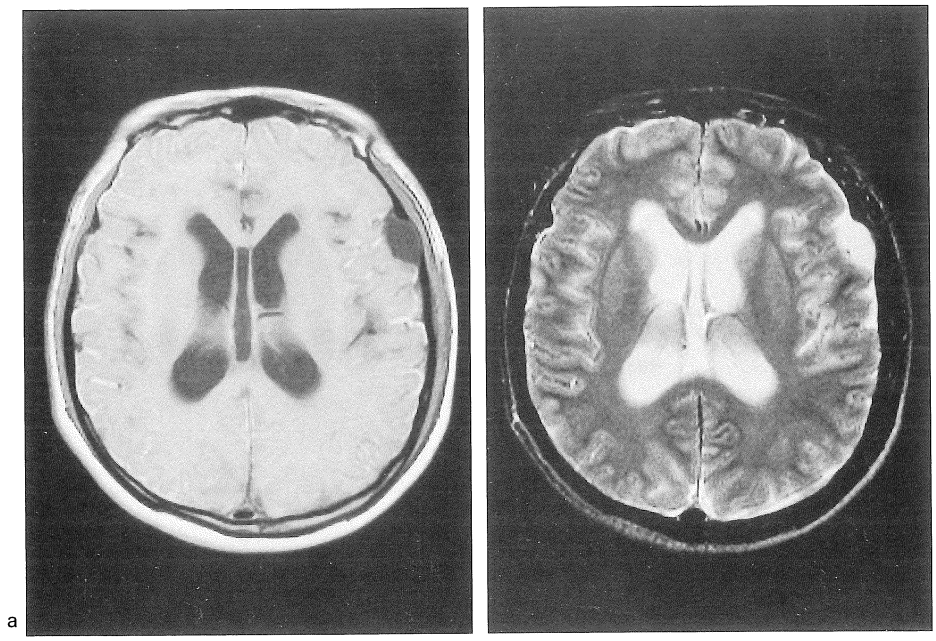

Fig. 2. (a) T_1-weighted axial image of the brain (SE : TR/TE=600/15 ms) showing diffuse hyperintensities in the leptomeninges of the cerebral cortex, mild dilation of the lateral ventricles and an arachnoid cyst in the left fronto-parietal region. (b) T_2-weighted axial image of the brain (SE : TR/TE=2500/90 ms) showing diffuse hypointensities in the leptomeninges.

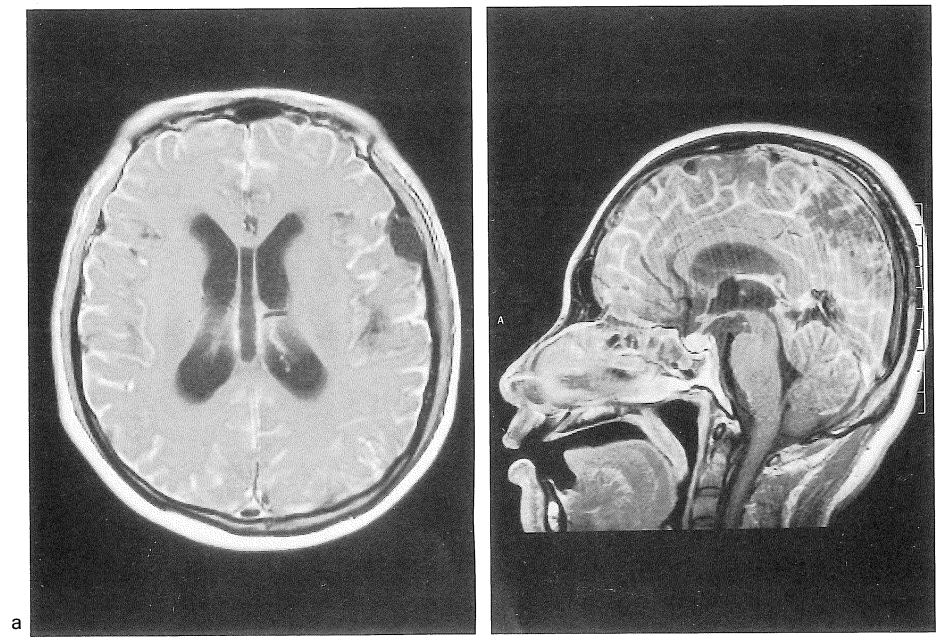

Fig. 3. (a) Brain Gd-DTPA enhanced T_1-weighted image (SE : TR/TE=600/15 ms) showing diffuse enhancement in the leptomeninges of the cerebral cortex. (b) Brain contrast-enhanced T_1-weighted sagittal image showing diffuse enhancement in the leptomeninges of the cerebral cortex, cerebellar cortex and brain stem.

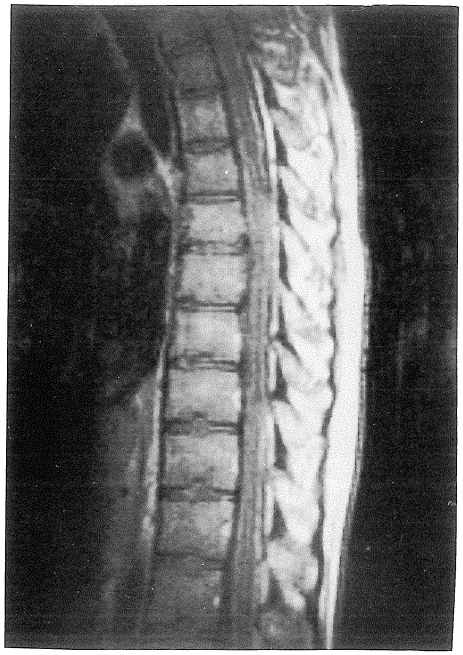

Fig. 4. (a) Thoracic spine Gd-DTPA enhanced T_1-weighted sagittal image (SE : TR/TE=600/20 ms) showing diffuse enhancement in the leptomeninges of the spinal cord.

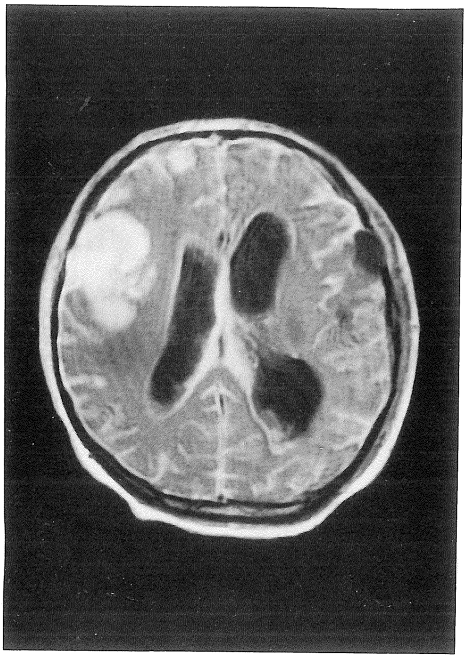

Fig. 5. Brain Gd-DTPA enhanced T_1-weighted axial image (SE : TR/TE=600/14 ms) 14 months after the first examination showing diffuse, thicker enhancement in the leptomeninges and septum pellucidum and homogeneous-solid in the frontal region.

topsy, solid masses were found in the subarachnoid space, microscopic examination of which displayed malignant melanoma.

DISCUSSION

Neurocutaneous melanosis (NCM) is a rare nonfamilial disorder generally characterized by the development of congenital melanocytic nevi and benign or malignant melanotic tumors of the central nervous system. It has been postulated that this represents a congenital error in morphogenesis of the embryonal neuroectoderm[1]. Melanocytes are derived from melanoblasts arising from neural crest cells and produce the pigment melanin[1)~4)]. Melanocytes are found normally in the CNS, most commonly in the pia matter covering the cerebral and cerebellar hemispheres, the anterior surface of the brain stem, the basal surface of the brain and in the anterior surface of the spinal cord[4]. In NCM, however, there is focal and diffuse proliferation of sheets and nests of melanocytes[2)~4)]. Congenital skin lesions may occur as multiple or large pigmented cutaneous nevi distributed over the head, neck, trunk or show a bathing suit distribution (lower abdomen, pelvis, buttocks and upper thighs) over the body[1)~3)]. Kadonaga proposed the following criteria for a diagnosis of NCM : 1) large or multiple congenital nevi associated with meningeal melanosis or melanoma ; 2) no evidence of cutaneous melanoma except in patients with be-

nign meningeal lesions ; 3) no evidence of meningeal melanoma, except in patients with cutaneous benign lesions[1]. In the above, large is defined as a lesion equal to or greater than 20 cm in maximal diameter in an adult. Lesions in neonates and infants that measure approximately 9 cm in diameter on the head or 6 cm on the body are also included. Multiple is defined as three or more lesions. The patient of the present case report met all three criteria.

Neurological symptoms of NCM generally appear within the first 2 years of life, or less frequently, in the teens or twenties[1]. These symptoms, which include irritability, lethargy, headache, recurrent vomiting, seizures and photophobia and the physical sign of increased head circumference, are usually the result of increased intracranial pressure. As the disease progresses, the patient may develop ataxia, aphasia, dysarthria, loss of the ability to walk and show evidence of a psychiatric disorder[1]. Increased intracranial pressure is thought to be due to obstruction of CSF flow at the cisterna magna or at outlets of the fourth ventricle or due to interference with CSF reabsorption by the arachnoid villi secondary to melanocytic infiltration[5]. In the present case, it was thought that the neurological symptoms developed in adulthood due to a low level increase of intracranial pressure. Leptomeningeal melanosis is malignant in 40 to 50% of patients with NCM[6),7]. Various therapeutic measures such as chemotherapy, radiotherapy and surgery have been attempted as treatment for NCM, although no major success has been reported for reducing mortality in patients with this disease[1),8),9]. The prognosis for symptomatic NCM is poor; most patients die within three years after the initial onset of neurological symptoms, either from malignant melanoma or progressive growth of benign melanotic cells[1]. The patient of the present case report died 25 months after the onset of neurological symptoms.

NCM should be clinically suspected in patients with large nevi in whom neurological findings develop within the first two years of life. Leptomeningeal involvement should be suspected in patients found to have melanocytosis in the CSF and can be confirmed by brain or spinal cord biopsy showing marked proliferation of melanocytes in the leptomeninges, with or without brain and/or spinal cord invasion. There have been a few reports of MR findings in NCM which generally have consisted of leptomeningeal hyperintensities in T_1-weighted images. These hyperintensities in T_1-weighted images can be explained by the paramagnetic effect of free radicals in intracellular melanin deposits[7),10]. In adult cases lacking large nevi and in which neurological symptoms do not present until adulthood such as in the present case report, MR studies are particularly useful for establishing the diagnosis of NCM without resorting to invasive diagnostic procedures. CSF cytology and biopsy findings are not of value for establishing the extent of leptomeningeal involvement, whereas MR studies can be very useful for establishing the extent of involvement and for detecting the complication of hydrocephalus. Moreover, serial MR studies can be used to demonstrate malignant changes in leptomeningeal melanosis.

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