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Case Report

Neurocutaneous Melanosis with Hydrocephalus and Dandy-Walker Variant

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Abstract

Neurocutaneous melanosis (NCM) is one of the rare, congenital, noninheritable phakomatoses characterized by the presence of large and/or multiple congenital melanocytic cutaneous nevi associated with intracranial leptomeningeal melanocytosis. NCM usually presents before 2 years of age. So far 302 cases have been reported in literature. We report a case of NCM presenting with obstructive hydrocephalus and Dandy-Walker Variant in a young adult.

Keywords: *Dandy-Walker variant, hydrocephalus, neurocutaneous melanosis, tuberculous meningitis*

Introduction

Neurocutaneous melanosis (NCM) is one of the rare phakomatoses. NCM is a childhood disorder which has varied presentations and is associated with other Neurocutaneous syndromes like Sturge-Weber syndrome, Neurofibromatosis type 1, Dandy-Walker syndrome. The prognosis of NCM is dismal, more so when it presented in adults and associated with Dandy-Walker syndrome, like our case.

Case Report

A 28-year-old male patient presented with history of repeated episodes of convulsion for over a year, and altered sensorium, headache, vomiting and diminished vision since a month prior to his admission. He was initially treated elsewhere with broad spectrum antibiotics and anticonvulsants. On evaluation at Emergency, his heart rate was 114/min, blood pressure was 154/110 mm of Hg, Glasgow Coma Score was E4V1M5, and pupils were sluggishly reacting to light. Fundoscopy showed bilateral post-papilloedema optic atrophy. His vision could not be assessed properly. On general examination, he was found to have congenital giant cutaneous hairy melanocytic nevus over trunk in "bathing suit" distribution associated with multiple hairy satellite cutaneous nevi over the rest of the body [Figure 1a-e]. There was no family history of similar skin lesion.

Contrast-enhanced computerized tomography (CECT) of brain revealed dural based hyperdense lesions in both frontal lobes with diffuse leptomeningeal thickening and enhancement, communicating hydrocephalus with posterior fossa cyst and partial agenesis of cerebellar vermis [Figure 2]. Lumbar cerebrospinal fluid (CSF) examination showed xanthochromia, very high protein (749.4 mg %), cell count of 16 with lymphocytes of 56%, normal sugar and elevated adenosine deaminase (ADA) of 20.18. Liver function tests were deranged with more than 300 of all hepatic enzymes and mild elevation of bilirubin. He was also found to be hypothyroid (T3-0.79 ng/ml, T4-6.63 ug/dL, and thyroid stimulating hormone - 6.77 uIU/ml). CECT of chest and abdomen were normal.

Based on CSF study, modified anti-tubercular drugs (ATD) started along with dexamethasone and broad-spectrum antibiotics (injection meropenem and injection amikacin, both having some anti-tubercular actions). His sensorium improved. MRI of brain with contrast subsequently showed lesions which were cortical based and showed T1W and T2W hyper-intensity were enhancing with contrast along with extensive leptomeningeal enhancement, obstructive hydrocephalus and a large posterior fossa extra-axial cyst, communicating with the 4th ventricle [Figures 3a-c]. MRI of spine

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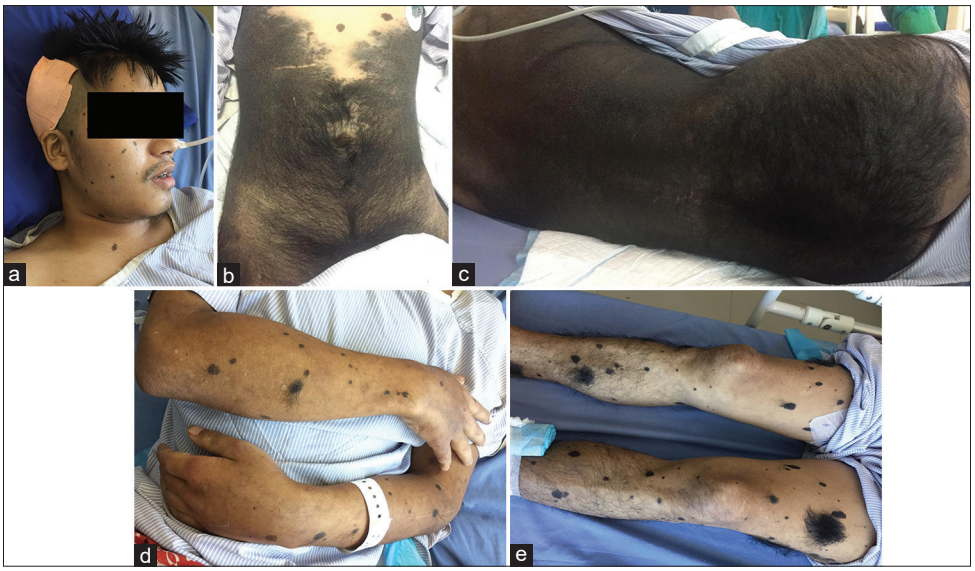

Figure 1: (a-e) Congenital giant cutaneous hairy melanocytic nevus over trunk in "bathing suit" distribution associated with multiple hairy satellite cutaneous nevi over the rest of the body

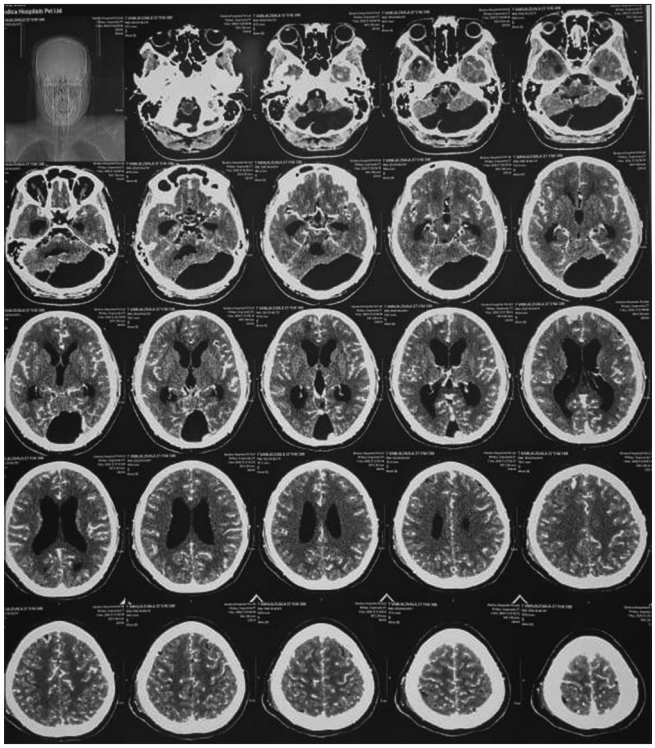

Figure 2: Contrast enhanced computerized tomography of brain revealed dural based hyperdense lesions in both frontal lobes with diffuse leptomeningeal thickening and enhancement, communicating hydrocephalus with posterior fossa cyst and partial agenesis of cerebellar vermis

showed multiple extramedullary intra-dural septations and loculations in the dorsal spine and thickened cauda equine nerve roots [Figure 4a and b]. A Ventriculo-Peritoneal shunt (VP shunt) was done after a week of the above treatment. Ventricular CSF showed protein of 98 mg%, 6 cells and ADA of 1.54. Acid Fast Bacillus was not found in the smear. Gene Xpert for MTB/RIF from CSF did not detect tubercle bacilli. CSF cytology from cytocentrifuge deposit showing atypical melanocytes with brown pigment and vesicular nuclei with prominent eosinophilic nucleoli. Cell block from CSF centrifuged deposit showed many cells with vesicular nuclei having prominent nucleoli and brown pigment [Figure 5a and b]. Presence of melanocytes confirmed by immunohistochemicalstains specific melanocytes HMB45 and Melan A which target proteins gp100 and Melan A on the cells.

Discussion

NCM is a rare, congenital, noninheritable disorder characterized by the presence of multiple and or large congenital melanocytic nevi and are associated with benign and or malignant melanocytic tumors of the leptomeninges.[1] First case of NCM was described and reported by Rokitansky in 1861 in 14 years old boy with a congenital nevus and mental retardation and hydrocephalus.[2] Since then over 300 cases have been reported in literature.[3] Most cases are sporadic, with an equal gender predilection and they usually present before the age of 2 years.[4,5,6] Rarely NCM presents in adulthood. Some remain asymptomatic.[7]

Pathogenesis of NCM has been stated as a neuro-ectodermal defect during morphogenesis involving melanoblasts of skin and pia mater originating from neural crest cells. Two-thirds of patients of NCM have giant congenital melanocytic nevus. A third shows multiple small lesions. Our case had features of both.

Clinical presentations are usually with signs of intracranial hypertension, focal seizures, motor deficits or aphasia. Hydrocephalus is present in two-thirds of patients due to obstruction of CSF flow or reduced absorption as a result of thickened leptomeninges.

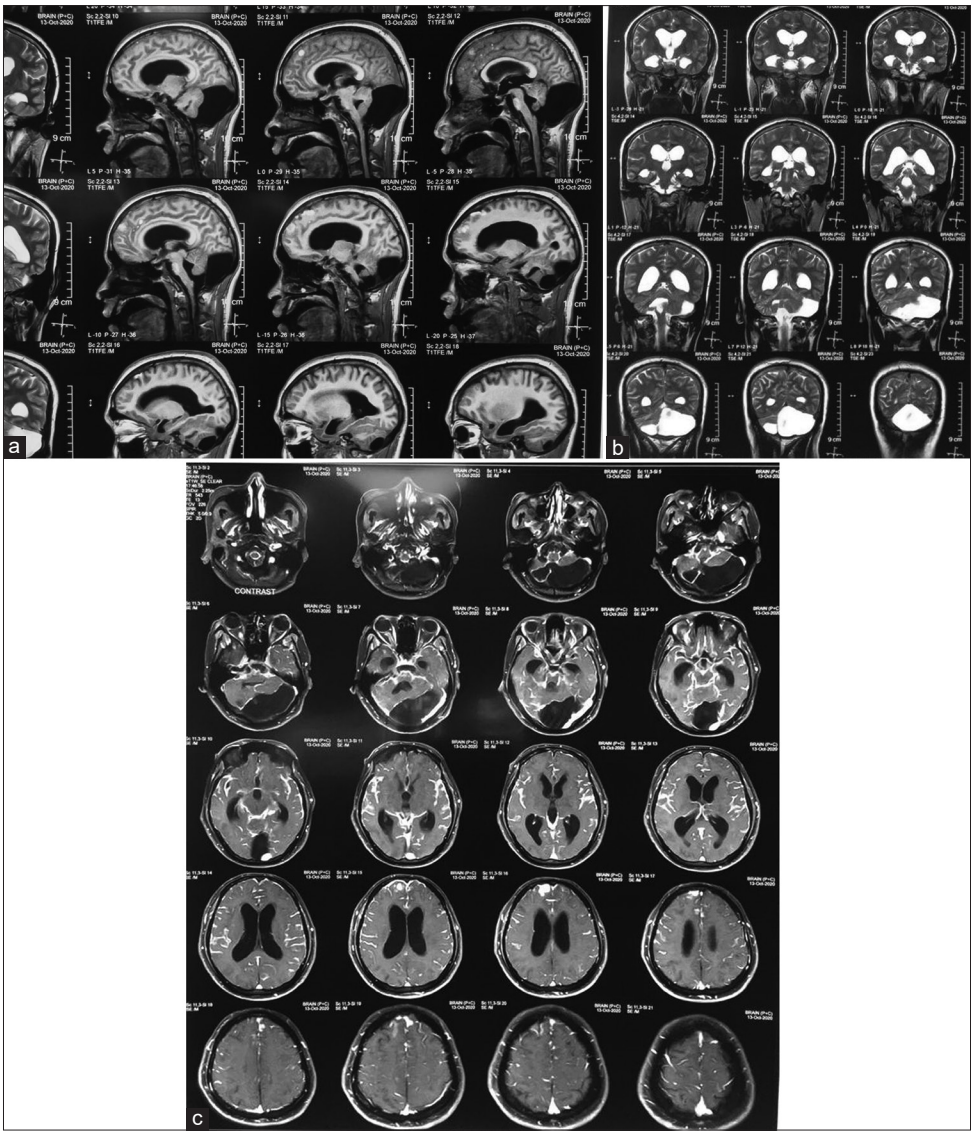

Figure 3: (a) MRI of brain showing linear cortical based (b) T1 hyperintense and (c) T2 hypointense signal in bilateral cerebral hemisphere showing no obvious enhancement or any other abnormal enhancing focus in post contrast

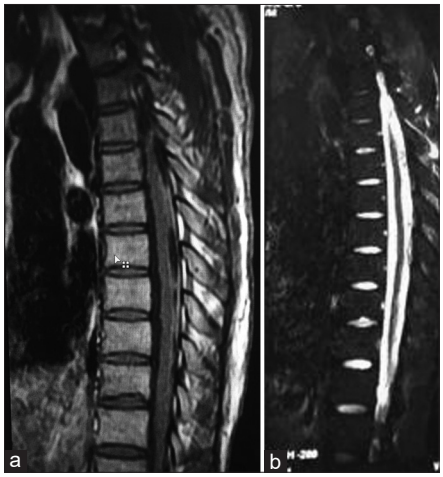

Figure 4: MRI of spine showing (a) T1 hyperintense and (b) T2 hypointense linear extramedullary intradural septations and loculations in the dorsal spine and thickened cauda equina nerve roots.

NCM has been reported to be associated with other neuro-cutaneous syndromes such as Sturge-Weber Syndrome and von Recklinghausen's neurofibromatosis 1. NCM has also been reported to be associated with posterior fossa cystic malformations like Dandy Walker malformation (DWM), like in our case, in about 10% cases.[8-10] The prognosis of patients of NCM with DWM is extremely poor. Children die early in life from malignant transformation of the melanosis. NCM and DWM concurrence also suggests common etio-pathogenesis. There are case reports of NCM masquerading as neurofibromas.[11] NCM with multiple intracranial calcifications are also described in literature.

The diagnostic criteria were first given by Fox[12] in 1972, and later modified by Kadonaga and Frieden[13] in 1991, which were as follows: (1) large nevus (>20 cm in adults and lesions which are approximately 9 cm of diameter on

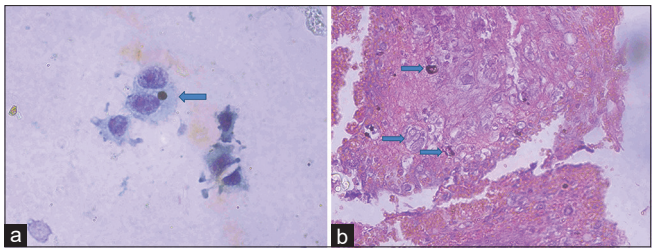

Figure 5: (a) cerebrospinal fluid cytology from cytocentrifuge deposit showing atypical melanocytes with brown pigment and vesicular nuclei with prominent eosinophilic nucleoli. (b) Cell block from cerebrospinal fluid centrifuged deposit showed many cells with vesicular nuclei having prominent nucleoli and brown pigment

the head or 6 cm on the body in infants), (2) multiple (≥3) nevi, (3) no evidence of cutaneous melanoma, except in cases where meningeal lesions are histologically benign, (4) no evidence of meningeal melanoma, except in cases where the cutaneous lesions are benign. Our case was compatible with the diagnosis of NCM (large nevus, multiple nevi, no evidence of cutaneous or meningeal melanoma).

The treatment with radiation therapy or chemotherapy is dismal if there is the benign melanocytic proliferation of the leptomeninges.Patients of NCM may develop malignant melanoma in 40%–60% of cases, and malignant transformation is heralded by development of intra-parenchymal invasion or intracranial or intraspinal masses.[14]

Increased CSF ADA level is an important diagnostic clue in tubercular meningitis (TBM). It has sensitivity of 82.14% and specificity of 90.91% in diagnosing TBM.[15] Raised CSF ADA level has been reported in cryptococcal meningitis,[16] listeria meningitis,[17] sarcoid meningitis,[18] meningeal involvement with leukemia or lymphoma,[19] toxoplasmosis, cerebral infarction, neurosyphilis, and other aseptic meningitis. Among these, TBM has highest ADA activities (median 21.3 U/l, range 20.0–23.0), followed by lymphoma (median 13.0, range 4.0–25.0). The sensitivity and specificity of the test for diagnosing TBM is 100% and 99% respectively when a cut-off value of 20.0 is used. In our case, as the CSF ADA as well as the protein was very high and suggestive of TBM and after a week of treatment with ATD and steroids, both values came down significantly, we proposed a full course of ATD (12 months).

We may also postulate that the high ADA and high CSF protein are associated with NCM because of the CSF flow obstruction by the melanin pigments which could have been resolved so quickly with Dexamethasone rather than associated TBM (in the absence of tubercle bacillus isolation). However, there are no such references in the available literature. We tend to regularly follow-up the patient regarding the neurological outcome as well as cutaneous lesions at 3–6 months interval.

Conclusion

Our case is unique because of adult presentation, with giant as well as multiple nevi, extensive intracerebral and spinal cord involvement, hydrocephalus, Dandy-Walker Variant complicated by possible TBM.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understand that name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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