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Progressive brainstem compression in an infant with neurocutaneous melanosis and Dandy–Walker complex following ventriculoperitoneal shunt placement for hydrocephalus

Case report

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✓Neurocutaneous melanosis (NM) coexisting with the Dandy–Walker complex (DWC) is a rare condition, with fewer than 15 cases reported in the literature. The authors present a case of an infant with NM and DWC suffering from progressive brainstem compression following ventriculoperitoneal (VP) shunt placement for hydrocephalus. This 1-year-old boy with congenital melanocytic nevi had met normal developmental milestones until the age of 11 months, when he began regressing in ambulation and language function. Intractable vomiting had developed 1 week later. Magnetic resonance (MR) imaging of the brain revealed DWC with hydrocephalus, and spinal MR images demonstrated a proliferative process within the meninges, consistent with NM. The patient underwent right frontal VP shunt placement resulting in immediate symptom relief, but 3 weeks later became irritable, increasingly lethargic, unable to pull to stand, and unable to tolerate solid food without choking. Due to these symptoms and intractable vomiting, the patient presented to the authors' institution.

Brain MR imaging revealed a new-onset diffuse cystic process with anterior and posterior brainstem compression, marked kinking of the cervicomedullary junction, melanocyte pigmentation of the left temporal lobe, diffuse leptomeningeal enhancement, and no evidence of hydrocephalus. Consistent with these imaging findings, the degree of brainstem involvement upon gross visualization predictably deterred resection attempts beyond those necessary for biopsy. Pathological examination revealed diffuse melanocytosis, and the family decided not to pursue aggressive measures postoperatively.

This report indicates the potential for rapid intracranial manifestation of diffuse melanocytosis in NM patients. Although the prognosis is poor, early neurosurgical involvement in these patients may provide tissue diagnosis and the potential for decompression if the process is caught early in its course. *(DOI: 10.3171/PED-07/12/500)*

Key Words • brainstem compression • Dandy–Walker complex • hydrocephalus • neurocutaneous melanosis • pediatric neurosurgery • ventriculoperitoneal shunt

Neurocutaneous melanosis is a congenital, nonhereditary disorder defined by the presence of multiple and/or giant congenital melanocytic nevi associated with abnormal melanin deposits in the brain and/or leptomeninges documented by MR imaging or autopsy.[5,6,9,18] It is a rare condition, with fewer than 200 cases reported in the literature. Although there is proliferation of melanocytes in the skin and arachnoid mater, there is currently no embryological explanation for the genesis of this disorder. Approximately half of all patients with NM eventually develop melanoma of the central nervous system.[17]

A variety of developmental malformations occur in patients with NM, including cystic malformations of the posterior fossa, particularly the Dandy–Walker malformation, Dandy–Walker variant, and posterior fossa arachnoid cyst. The spectrum of these posterior fossa abnormalities has been collectively termed the DWC, and approximately 10% of patients with NM present with some variant of the spectrum.[9,12] Fewer than 15 cases of the combination of NM and the DWC have been reported in the literature.[1,5–8,10,12–14,18,19,21] We report a case of an infant with NM and DWC, who presented to us suffering from a rapidly progressive intracrani-

Abbreviations used in this paper: DWC = Dandy–Walker complex; MR = magnetic resonance; NM = neurocutaneous melanosis; VP = ventriculoperitoneal.

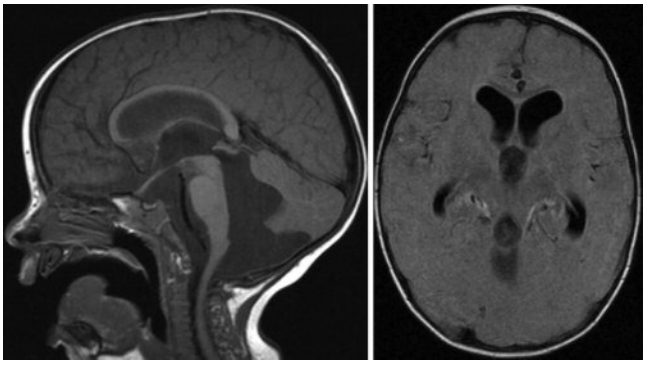

FIG. 1. Magnetic resonance images obtained at the patient's initial presentation. *Left:* Sagittal T1-weighted image demonstrating cerebellar vermian hypoplasia and gross enlargement of the fourth ventricle, consistent with DWC. *Right:* Axial T1-weighted image demonstrating hydrocephalus without aqueductal obstruction.

al process with brainstem compression following VP shunt placement for hydrocephalus.

Case Report

History and Presentation. This 1-year-old Caucasian boy had congenital melanocytic nevi covering more than 30% of his body but had experienced an otherwise uncomplicated birth. He had met developmental milestones normally until the age of 11 months, when he began regressing in ambulation and language function. Decreased feeding was also noted. These symptoms were followed 1 week later by intractable vomiting. An MR imaging study of the brain (Fig. 1) performed at that time revealed a grossly abnormal posterior fossa with agenesis of the cerebellar vermis, enlargement of the fourth ventricle, and hydrocephalus of the lateral and third ventricles, all consistent with DWC. Spinal MR imaging demonstrated a proliferative process within the meninges, consistent with the diagnosis of NM. The patient underwent placement of a right frontal VP shunt, which resulted in correction of his hydrocephalus and immediate symptom relief. During the 3rd postoperative week, however, he became irritable, increasingly lethargic, unable to pull to stand, and unable to tolerate solid food without choking. Due to these symptoms and intractable vomiting, the patient presented to our institution, and a neurosurgical consultation was requested to rule out obstructive hydrocephalus.

Examination. Clinical examination revealed the patient to be afebrile, with multiple melanotic lesions on his body. The results of neurological examination were remarkable for mild lethargy with evidence of ninth and 10th cranial nerve dysfunction; the patient had no signs or symptoms of hydrocephalus.

An MR imaging study of the brain (Fig. 2A–D) revealed diffuse leptomeningeal enhancement with melanocytic pigmentation deposits in the left temporal lobe, large cystic-appearing masses in the right cerebellopontine angle, and associated anterior and posterior compression of the brainstem with kinking of the cervicomedullary junction. There was no evidence of hydrocephalus or shunt malfunction (Fig. 2E).

Operation and Postoperative Course. On the basis of the correlation of these imaging findings with the patient's clinical symptoms, surgery was considered. Preoperative discussion with the family emphasized the expectation that little more than a biopsy could be performed due to the degree of brainstem involvement evident on MR images. Because

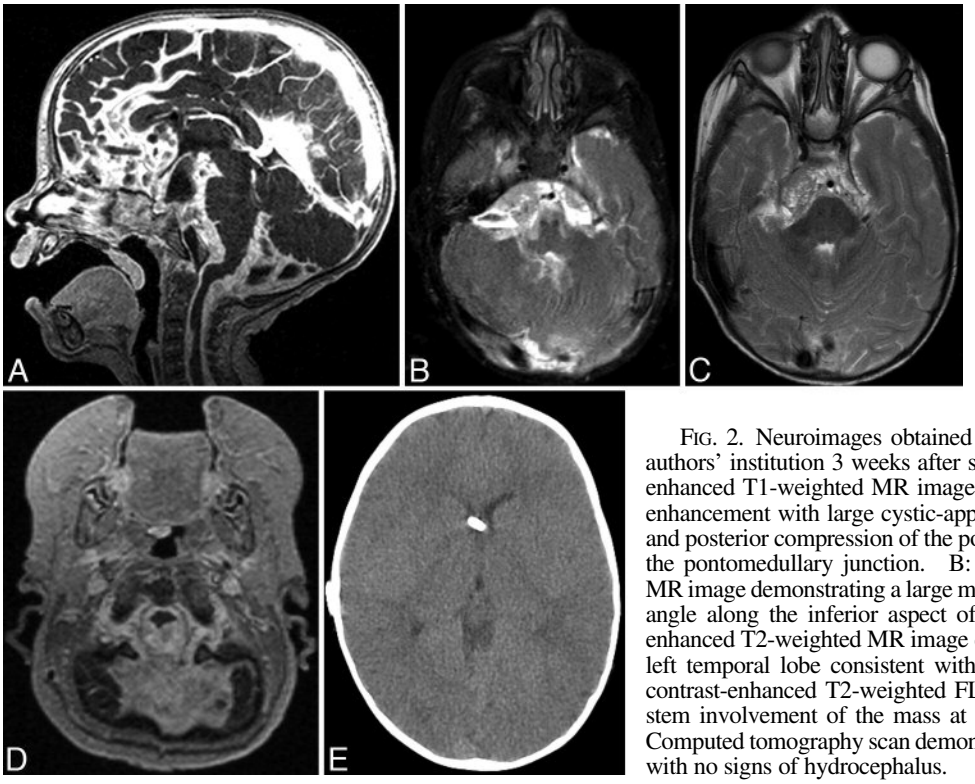

FIG. 2. Neuroimages obtained upon the patient's presentation to the authors' institution 3 weeks after shunt placement. A: Sagittal contrast-enhanced T1-weighted MR image demonstrating diffuse leptomeningeal enhancement with large cystic-appearing masses associated with anterior and posterior compression of the pons, medulla, and upper cervical cord at the pontomedullary junction. B: Axial contrast-enhanced T1-weighted MR image demonstrating a large mass involving the right cerebellopontine angle along the inferior aspect of the frontal lobe. C: Axial contrast-enhanced T2-weighted MR image demonstrating a hyperdense area of the left temporal lobe consistent with melanocyte pigmentation. D: Axial contrast-enhanced T2-weighted FLAIR MR image demonstrating brainstem involvement of the mass at the level of surgical intervention. E: Computed tomography scan demonstrating optimal placement of VP shunt with no signs of hydrocephalus.

further therapy could not be offered without definitive histopathological findings, the patient underwent posterior fossa craniectomy for neurosurgical evaluation of the disease process.

Surgical intervention consisted of a posterior laminectomy from C-1 to C-3 and small midline suboccipital craniectomy. A small midline opening in the dura mater was made at the level of C-1 and the foramen magnum, revealing a highly vascular reddish-brown tumor protruding outward under pressure. Although an excisional biopsy was performed, due to the tumor's extensive nature, prominent hypervascularity, and degree of protrusion under pressure, the risks of brainstem injury were deemed to greatly outweigh any potential benefits of additional resection. Consequently, the dura was closed following the biopsy.

Postoperatively, the patient was transferred back to the pediatric intensive care unit for close monitoring. Given the extremely poor prognosis of the patient's disease, an extensive discussion between the physicians and the family concluded with the decision not to pursue aggressive measures. The patient died shortly thereafter.

Histopathological Findings. Microscopic evaluation of the intradural cervical mass (Fig. 3) revealed a relatively monomorphic population of small round cells, some of which had more abundant cytoplasm and a few of which had melanocytic pigment. Necrosis was not identified, and only a few mitotic figures and mild cellular pleomorphism were noted. Cells demonstrated strong immunophenotypic evidence of melanocytic differentiation with positivity for melanocytic markers HMB-45, Melan A, S100 protein, and tyrosinase. The tumor was highly vascular with the most well-differentiated tumor cells clinging to blood vessels. The Ki 67 labeling index was greater than 30%, correlating with the clinically apparent strong proliferative capacity of the lesion. The constellation of these findings was consistent with the diagnosis of diffuse melanocytosis.

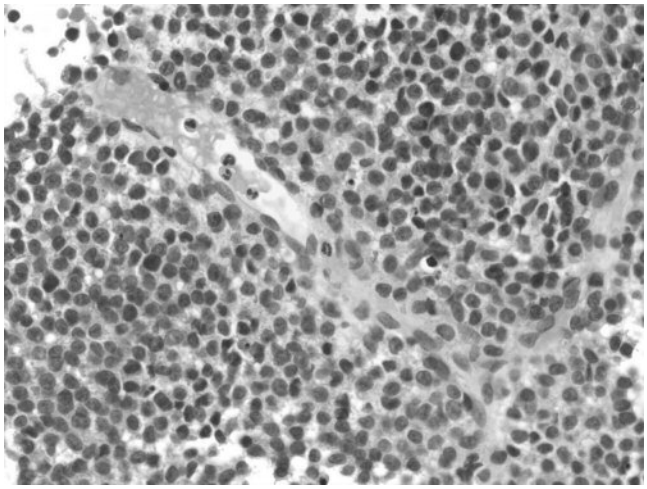

FIG. 3. Photomicrograph of stained section of tumor showing closely packed cells with small amounts of cytoplasm. Occasional cells contain melanocytic pigment. H & E, original magnification × 40.

Discussion

First described by Rokitansky[20] in 1861, NM is characterized by the following diagnostic criteria: 1) large or multiple congenital melanocytic nevi with meningeal melanosis or melanoma, 2) absence of cutaneous melanoma (except for meningeal lesions), and 3) no evidence of meningeal melanoma except in patients with benign cutaneous lesions.[12,20] Typically seen in Caucasians with no sex predilection, the prognosis of NM diminishes greatly once neurological symptoms occur (regardless of the presence or absence of neoplasm), as the rapidity of the disease often makes it refractory to chemotherapy and radiation.[6,11,13,15,18] Patients usually die due to events related to increased intracranial pressure.[17]

The DWC classically occurs during the embryological stage prior to the period during which the fourth ventricle foramina typically become patent (between 28th and 50th day of gestation).[16] Originating as a failure of normal posterior medullary velum regressive changes, the complex classically manifests as vermian agenesis, cystic dilatation of the fourth ventricle, and separation of the cerebellar hemispheres.[4,6] Although the mechanism of the association between DWC and NM is currently unknown, the most widely accepted hypothesis is excessive meningeal melanosis resulting in defective ectodermal–mesodermal interaction and consequently leading to maldevelopment of the cerebellum and the fourth ventricle.[2,3]

Our patient was an infant with NM and DWC who presented with hydrocephalus at 11 months of age. A VP shunt was placed with apparent success, but shortly thereafter, the patient experienced symptoms that were initially believed to be recurrent hydrocephalus. Following clinical examination and repeated imaging studies, however, it became clear that the new symptoms (particularly the impaired swallowing) were due to a new lesion that had rapidly progressed in the 3 weeks since the shunt placement, resulting in rapid brainstem compression unrelated to the patient's previous hydrocephalus. Although a tissue biopsy could be performed, the degree of brainstem compression and bulk of the melanocytosis mass made it unfeasible to attempt gross total resection without a high likelihood of brainstem injury. Histopathological examination of the biopsy specimen revealed diffuse melanocytosis (diffuse melanosis), which although categorized along with malignant melanoma in the same World Health Organization classification as primary melanoma, carries distinct clinical and pathological differences, both of which were present in this patient. Diffuse melanocytosis is much more likely than malignant melanoma to be associated with congenital melanocytic nevi, and diffuse melanocytosis is distinguished histopathologically by the absence of malignant cells. Sadly, both diffuse melanocytosis and malignant melanoma are associated with a very poor prognosis. This report illustrates the rapidity with which diffuse melanocytosis can progress intracranially in patients with NM, and shows that even in the setting of surgically treated hydrocephalus, recurrent neurological symptoms in patients with NM and DWC can rapidly occur in a manner unrelated to the original hydrocephalus.

Conclusions

This is the first report of brainstem compression following successful VP shunt placement in an infant with NM and DWC, and it indicates the potential for rapid intracra-

nial manifestation of diffuse melanocytosis in patients with NM even in the setting of surgically treated hydrocephalus. Expedient neurosurgical involvement in such cases may provide tissue diagnosis and allow for treatment of hydrocephalus with the potential for palliative operative decompression if the process is caught early in its course. We hope that in the future neurosurgical intervention could be augmented by adjuvant treatment that could make a meaningful impact on the prognosis of this disease.

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