CASE REPORT

Neurocutaneous Melanosis and Dandy-Walker Malformation in a Newborn: A Case Report

Negin Armide, MD[1]; Bita Pourmohammadi, MD[2]; Meisam Babaei, MD[3,4]

[1]Department of Medicine, North Khorasan University of Medical Sciences, Bojnurd, Iran.
[2]Student research committee, North Khorasan University of Medical Sciences, Bojnurd, Iran.
[3]Rare Pediatric Neurological Diseases Research Center, Mashhad University of Medical Sciences, Mashhad, Iran.
[4]Department of Pediatrics, North Khorasan University of Medical Sciences, Bojnurd, Iran.

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ABSTRACT

Neurocutaneous melanosis (NCM) and Dandy-Walker malformation (DWM) are both uncommon congenital condition, involving the central nervous system (CNS). Combined NCM with DWM have been reported in a few studies. Potential consequences, including hydrocephalus and CNS malignancies make these illnesses extremely challenging to diagnose and treat. Occurrences of NCM and DWM together, can result in progressive neurological consequences and patients with this condition seems to have really poor prognosis.

This case report describes a 5-day-old girl who was born with several large pigmented cutaneous nevi all over her body. She was born at term to non-consanguineous parents who had no family history of similar conditions. Imaging studies, such as brain CT scan, showed cystic dilatation of the fourth ventricle, hypoplasia of the cerebellar vermis, enlargement of the posterior fossa, and possibly secondary hydrocephalus—all of which are indicative of DWM. A biopsy of the nevi confirmed NCM. The patient had no neurological symptoms or systemic complications at the time of presentation.

The combination of NCM and DWM is extremely rare, with fewer than 40 reported cases so far. While the exact connection between these two conditions is not fully understood, their coexistence of these conditions might be linked to their shared embryonic origin. Both melanocytes and parts of the CNS come from the neural crest during early development. Since these conditions can have serious neurological and systemic effects, managing them requires multidisciplinary care because of potential consequences of these disorders.

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Introduction

Multiple melanocytic lesions on the skin are a hallmark of neurocutaneous melanosis (NCM), an uncommon congenital condition that is frequently linked to involvement of the central nervous system (CNS). Due to the possibility of CNS cancer, specifically melanoma, this illness is most commonly detected in infants or toddlers and has been shown to have potentially serious consequences (1,2). Even though NCM is a rare condition, reported in fewer than

Corresponding Author:
Meisam Babaei
Email: babaee1359@gmail.com

200 studies, it poses significant clinical challenges, particularly in evaluation, management, and prognosis.

Another rare congenital abnormality is the Dandy-Walker complex (DWC) represented by partial or total agenesis of the cerebellar vermis, a cystic dilatation of the fourth ventricle, and an expanded posterior fossa. Different developmental delays and neurological disabilities can result from DWC, depending on how severe the deformity is (3).

Despite the rarity of both NCM and DWC, few research has examined their co-occurrence, this co-occurrence has been reported in fewer than 40 cases. DWC has been linked to eight to ten percent of NCM cases, and the prognosis appears to be extremely poor when it is linked to NCM. While many reported cases of NCM with Dandy-Walker malformation (DWM) have poor prognosis and early mortality, recent reports suggest that some patients may survive beyond early childhood, although often with severe neurological disability. This reflects a heterogeneous prognosis (4) . Both melanocytes and parts of the CNS originate from the neural crest during early development, which suggests a possible association between these conditions (5). However, the exact pathophysiological mechanism underlying their coexistence remains unclear. Disturbances in early neuroectodermal development may account for the coexistence of NCM and Dandy-Walker deformity. During development, the neural crest gives rise to both melanocytes and components of the CNS. Proposedly, abnormal neural crest cell migration or proliferation may disrupt the inductive function of primitive meningeal tissue in posterior fossa development in addition to causing excessive melanocytic deposits. DWM could eventually result from such disruption, which could also affect normal vermian formation and fourth ventricle fenestration. The uncommon but acknowledged correlation between these two disorders can be explained by their similar embryological beginnings (6,7).

Accordingly, this case report aims to highlight the clinical presentation, diagnostic workup, and therapeutic considerations of an individual with both conditions. The pathophysiological reasons behind the simultaneous occurrence of NCM and DWC are still poorly understood. Understanding the complexities of co-occurring NCM and DWC can guide clinicians in providing appropriate care and improving long-term outcomes for affected patients.

Case Presentation

A 5-day-old female neonate was evaluated for the presence of multiple, large pigmented cutaneous nevi that were noted at birth. She was born from non-consanguineous parents, with no history of any congenital disorder. She was born at full term via vaginal delivery with stable vital signs and no immediate complications or abnormalities.

On examination, the patient's head circumference was normal, with a non-syndromic appearance, and ophthalmologic examination, including fundoscopy, was unremarkable. However, the patient was found to have several large, pigmented cutaneous nevi, both hairy and hairless, distributed across different areas of her body (Figure 1). Given the widespread nature of these nevi, evaluation was initiated to assess the possibility of associated CNS involvement. Since the patient's neurological condition was stable, no CSF analysis was done.

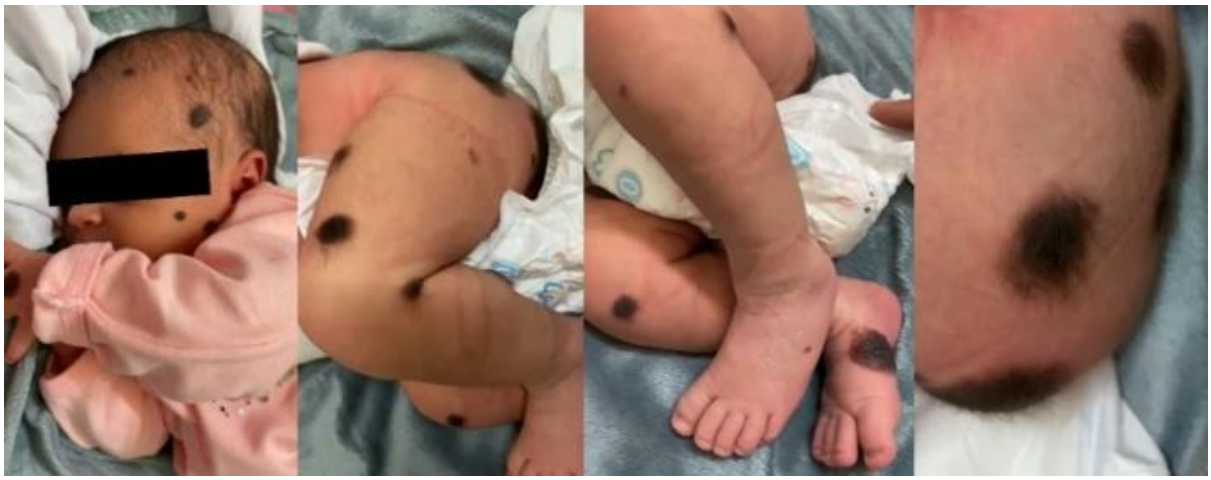

Figure 1. Multiple congenital melanocytic nevi on whole body

The nevi biopsy confirmed NCM and subsequent imaging studies, brain CT scan and MRI with contrast revealed the presence of a cystic dilation of the fourth ventricle, hypoplasia or aplasia of the cerebellar vermis, enlargement of the posterior fossa and possible secondary hydrocephalus, leading us to diagnosis of DWM (Figure 2,3). This finding confirmed the co-occurrence of DWC with NCM in this neonate. Small amount of meningeal enhancement was observed on brain MRI after contrast injection in superior sinus, and interpretation was confirmed by a pediatric neuroradiologist. The patient exhibited no apparent neurological symptoms at the time of evaluation, and no evidence of organ involvement or systemic complications existed. The co-existence of these two conditions in a neonate underscores the need for careful monitoring for potential neurological and developmental complications as the child grows.

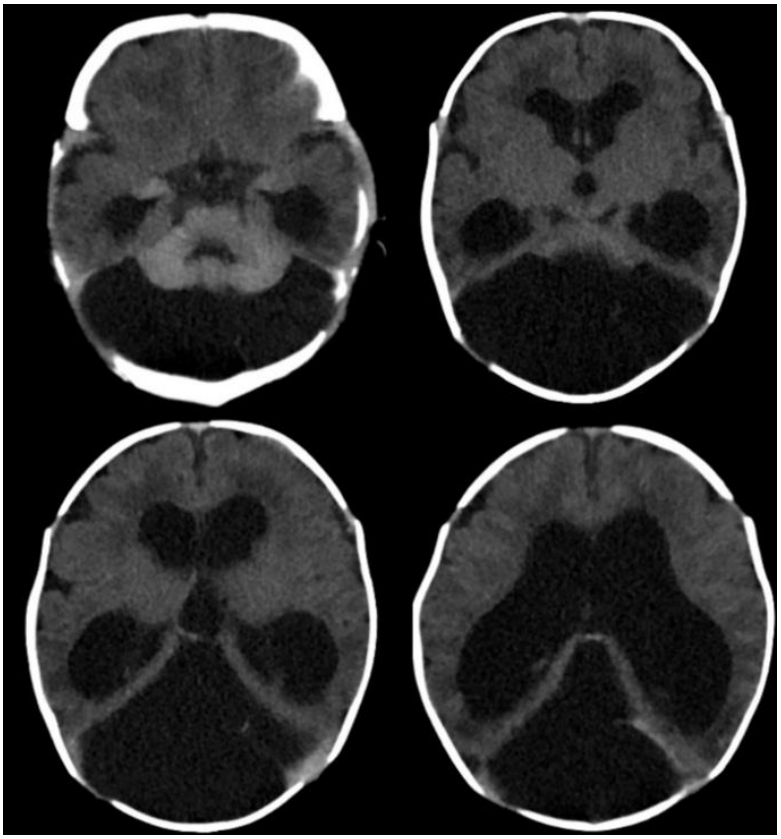

Figure 2. Brain CT scan showed small hyperdense area in the anterior right temporal lobe and to a lesser extent on the left side. The cerebellum and pons also appear slightly hyperdense. Mild to moderate dilatation of the lateral and third ventricles. Small cerebellum with a large cystic lesion posteriorly. The inferior vermis appears to be present

Discussion

NCM is an uncommon, congenital nonheritable neuroectodermal dysplasia, characterized by the presence of big, often several melanocytic lesions on the skin, alongside CNS involvement (3).The following criteria applied to this lesion: 1) several nevi (three or more lesions); 2) large nevus (bigger than 20 cm in adults and lesions that are about 9 cm in diameter on the head or 6 cm in diameter on the body in infants); 3) no evidence of cutaneous melanoma, unless the meningeal lesions are histologically benign; 4) no evidence of meningeal melanoma, unless the cutaneous lesion is benign. The present patient met the diagnostic criteria for NCM, including several nevi, a big nevus larger than 6 cm in size, and no signs of meningeal or cutaneous melanoma (4).

Only a small number of case reports in the literature describe the extremely rare co-occurrence of NCM with DWC (8). As previously stated, the DWM manifests as a large posterior fossa with high tentorium insertion, cerebellar vermis hypoplasia or aplasia, and cystic dilatation of the fourth ventricle, communicating with the posterior fossa (9).

Depending on the severity of the deformity, it may cause severe neurological impairments, such as delays in motor and developmental abilities. The majority of patients with DWM exhibit symptoms and indicators of elevated intracranial pressure, typically associated

with hydrocephalus and posterior fossa cysts. While some patients may come with different comorbidities that helps physician to an early diagnosis of illness, many patients do not show special clinical symptoms for years (10).

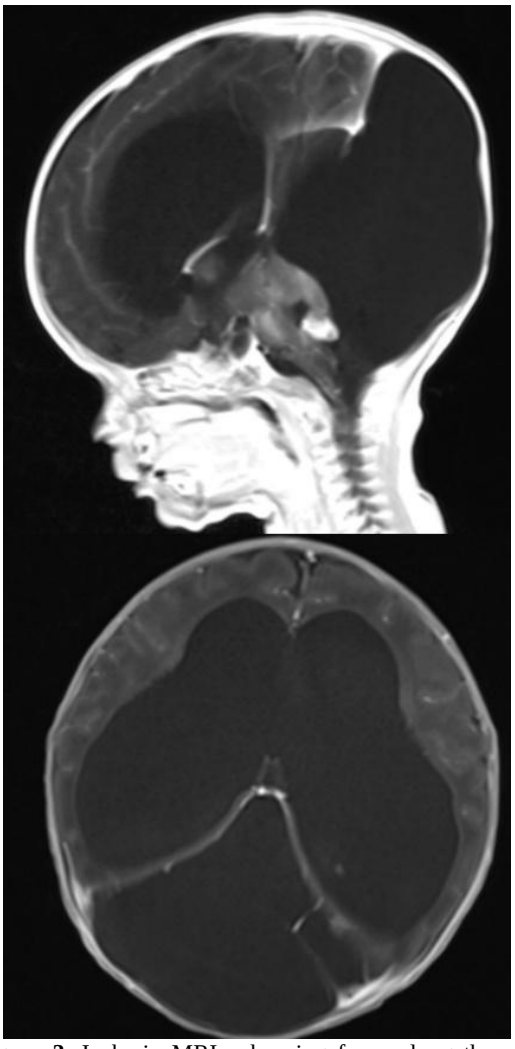

Figure 3. In brain MRI enhancing focus along the anterior aspect of the bilateral temporal lobe and an enhancing lesion at the foramen Magendi is consistent with leptomeningeal melanosis, appearing to be the reason for hydrocephalus. A large dorsal cystic lesion in the posterior fossa also seen with enhancement of bilateral cerebellar cortex and pons. Leptomeningeal melanoma in the foramen magnum region appears to be the cause of obstructive hydrocephalus.

In the present case, the patient met the diagnostic criteria for NCM but had no neurological symptoms at the time of evaluation. Since cutaneous manifestations in neonates can be associated with underlying CNS abnormalities, brain imaging was performed. Besides, brain MRI and CT scan revealed the presence of DWM. Unlike some reported cases where neurological symptoms are present at birth or develop early, the studied patient remained asymptomatic, this issue highlights the variability in clinical presentation and the importance of early evaluations for accurate diagnosis. The main limitation of this report is the absence of autopsy and genetic testing, limiting our ability to fully understand the underlying causes.

Although the etiology of NCM is still not fully known, it is believed to be the result of an error in the embryonal neuroectoderm's development (9). It is thought that two distinct pathophysiological mechanisms may cause the complex malformation known as DWM; the arrest of vermian development and the failure of fourth ventricle foramina fenestration, resulting in an enlarged Blake’s pouch and compresses the vermis (11).

Recent molecular research has shown that somatic mutations in the NRAS gene, and less commonly in BRAF, are essential in the pathophysiology of leptomeningeal melanosis and congenital melanocytic nevi. The neurological issues associated with NCM may be explained by these mutations, which cause abnormal melanocyte survival and proliferation. These results imply that common signaling pathways, particularly those involving the MAPK pathway, may be responsible for the cutaneous and CNS manifestations of NCM, as well as its correlation with developmental abnormalities like the DWC, even though we did not conduct molecular genetic testing on the present patient (12,13).

Since very few examples of these two illnesses co-occurring are available in the literature, the exact mechanisms that might connect them are still open to investigation. However, the data from this case and other comparable documented cases raise the notion that NCM disrupts mesenchymal development , as well as the normal inductive actions of primitive meningeal tissue, possibly leading to concurrent Dandy-Walker deformity (13).

This fact suggests that there may be a shared origin for these developmental defects. Irritability, seizures, fatigue, nausea and vomiting, bulged fontanels, stiff neck, papilledema, and cranial nerve palsies are the typical neurological manifestations of these two disorders. These symptoms are indicators of elevated intracranial pressure. Because of primary CNS melanosis, the course is typically progressive and fatal once the neurological symptoms appear (14,15).

Early detection of DWM and NCM is essential for managing potential consequences such hydrocephalus or malignant transformation of CNS melanotic lesions, as well as for monitoring neurological development. patients with NCM should have routine neuroimaging and neurological evaluations due to their higher risk of neurological sequelae, such as seizures and cognitive impairments (16).

Furthermore, DWM suggests possible surgical procedures to treat hydrocephalus or other related disorders, as well as continuous monitoring for

problems with motor development. Cystoperitoneal (CP) or ventriculoperitoneal (VP) shunts are two possible surgical treatments. Endoscopic third ventriculostomy (ETV) and other endoscopic treatments may be appropriate for other patients (1).

While no standard treatment protocols are available for the co-occurrence of NCM and DWM, a multidisciplinary approach involving pediatric neurology, dermatology, and neurosurgery is essential to ensure comprehensive care for affected infants.

In Conclusion

This case demonstrates the uncommon but crucial clinical correlation between DWM and NCM. The need of close observation for neurological and developmental conditions, shows importance of early detection of these two disorders in a newborn. Although the pathophysiological link between these conditions remains unclear, the co-occurrence necessitates careful, long-term management to optimize outcomes for affected children. To improve our comprehension of the relationship between NCM and DWC and to provide more effective management techniques for these cases, more research and case studies are required.

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Author's Contribution

All authors had full access to the data, contributed to the study, approved the final version of the manuscript, and took responsibility for its accuracy and integrity

Conflict of Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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