# Diffuse Meningeal Melanomatosis with Congenital Facial Nevus in an Adult

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## Clinical Description

A 43-year-old man presented with a 4-month history of headache, which was diffuse and progressive in intensity and was associated with recurrent vomiting. He was evaluated elsewhere with a computed tomography (CT) head and was detected to have hydrocephalus. He was treated with a ventriculoperitoneal shunt and antitubercular treatment with steroids. His symptoms did not improve; rather, he continued to deteriorate and presented to us with quadriparesis, bulbar dysfunction, and behavioral disturbances. He had a large nevus that was present over the left half of the face [Figure 1a] and had been noticed since infancy. Abnormal nevi were not detected elsewhere on the body. Magnetic Resonance Imaging (MRI) imaging findings are detailed in Figure 1b-h. The lumbar puncture was repeated thrice. On two occasions, the cell count was less than 4 with a protein of 13 mg/dL and normal glucose. Cerebrospinal fluid (CSF) was also subject to gram stain, CBNAAT for mycobacterium, Enzyme linked immunosorbent assay (ELISA) for brucella, and Neurocysticercosis (NCC), Venereal disease Research Laboratory (VDRL), and India ink staining, which were negative. The third CSF's cytological analysis revealed abnormal large cells containing pigment. These cells were positive for Human Melanoma Black 45 (HMB45), indicating a melanomatous origin [Figure 2]. A diagnosis of diffuse meningeal melanomatosis was made.

## Discussion

Diffuse meningeal melanomatosis is a rare and aggressive condition characterized by the infiltration of melanoma cells into the meninges.[1] Rarely, this condition may be associated with large nevi over the face or trunk, especially in children with a distinctive phakomatosis known as neurocutaneous melanosis. These nevi are usually benign; however, there is a minute risk of malignant transformation.[2] Such a phenomenon occurring in the fifth decade of life such as in our case is exceptionally rare. The relationship between large nevi and diffuse meningeal melanomatosis is not fully understood, but it is believed that the melanoma cells may originate from the nevi and then spread to the meninges.

We also wish to highlight that repeated lumbar punctures may be required to establish a diagnosis of carcinomatous meningitis.[3] A high degree of suspicion, careful interpretation of the smears, and appropriate Immunohistochemistry (IHC) stains will aid in an early diagnosis. Carcinomatous meningitis results in a typical leptomeningeal enhancement over the cerebellum and other neural structures on MRI, termed zuckerguss (sugar frosting in German). T1 sequences will demonstrate that these lesions are hyperintense and characteristic of this condition. One must also remember that substances other than melanin such as fat, methemoglobin, protein-rich exudates, and manganese can also appear hyperintense in T1.

Diffuse meningeal melanomatosis is a highly aggressive and rapidly progressive disease. It can lead to various neurological symptoms such as headaches, seizures, changes in vision, motor deficits, and cognitive decline. Due to its rarity and the similarity of symptoms with other neurological conditions, the diagnosis of diffuse meningeal melanomatosis can be challenging and often requires a combination of imaging studies, cerebrospinal fluid analysis, and biopsy. Treatment options for diffuse meningeal melanomatosis are limited, and the prognosis is generally poor.[4] The goal of treatment is to relieve symptoms and improve the quality of life for the patient.

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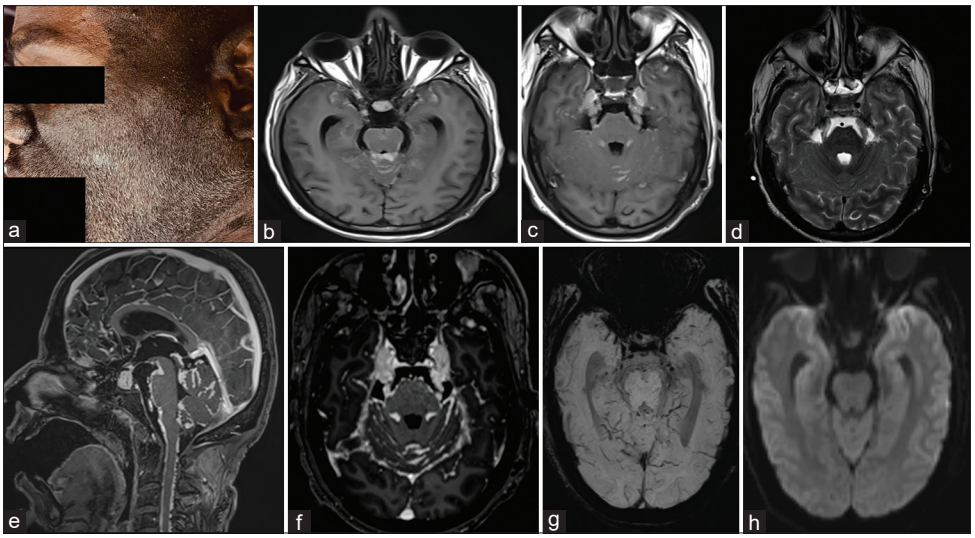


**Figure 1:** (a). A large nevus with distinct boundaries and uniform pigmentation over the left half of the face. (b and c). T1 (non-contrast) sequence showed hyperintensities over the cerebellar folia and the anterior temporal lobes. Note the intense thickening and T1 hyperintensities over the trigeminal nerve. (d). T2 axial demonstrating thickened trigeminal nerves. The rest of the brain parenchyma was normal. (e and f). T1-Gd+ images showing intense leptomeningeal enhancement over the cranial nerves, brainstem, cerebellum, and spinal cord ("*zuckerguss*"). (g and h). SWI and DWI sequences did not show areas of abnormal susceptibility or restriction

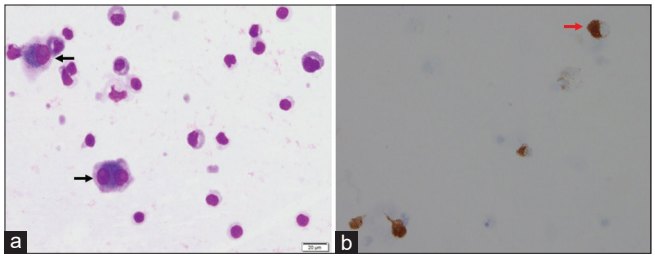


**Figure 2:** (a). Large atypical cells in CSF smear with intracytoplasmic pigment (black arrows). Some of these cells have a bilobed nucleus. (b). Melanoma cells confirmed by HMB45 immunostaining

## Declaration of patient consent

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

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Nil.

## Conflicts of interest

There are no conflicts of interest.

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