NEUROCUTANEOUS MELANOSIS: CLINICAL AND IMAGING CHARACTERISTICS IN 7 PATIENTS

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INTRODUCTION

Neurocutaneous Melanosis (NCM) is a rare genetic syndrome characterized by the presence of large and multiple congenital melanocytic nevi associated with leptomeningeal melanosis and/or melanomas of the central nervous system (CNS) as a result of an error in morphogenesis of neural ectoderm in the developing embryo.

Neurologic manifestations generally occur before the age of 2 years in the majority of patients. Clinically it presents with intracranial hypertension symptoms, focal deficits, epilepsy and cognitive impairment.

Magnetic Resonance Imaging (MRI) shows hidrocephalus in at least two thirds of the patients as well as typical lesions due to melanin deposits in parenchyma and meninges.

Keywords: Neurocutaneous melanosis, melanocytic nevi, MRI, leptomeningeal enhancement.

OBJECTIVE

Describe the clinical presentation of 7 patients with diagnosis of Neurocutaneous Melanosis.

MATERIALS AND METHODS

Retrospective and descriptive study of the clinical and imaging characteristics of patients with NCM treated at our hospital service from 2009 to 2021.

RESULTS

Seven patients were diagnosed with NCM, 5 boys and 2 girls who were twins. Diagnosis was established within the first week of life in 5 of the patients. The other two patients were diagnosed before the first year of life in one case at the age of 10 in the other. The most frequent symptom was the presence of large melanocytic nevi (n:6), most often scattered or localized in the thoracoabdominal region (n:3), and less often limited to the lower limbs (n:2). All the patients had characteristic skin lesions.

Two patients presented focal epilepsy consistent with electroencephalogram findings. With regards to the neurodevelopment, a general developmental delay or language impairment was observed in four of the patients.

In the MRI, five patients presented parenchymal involvement and six leptomeningeal enhancement and spinal cord lesions. Four patients required ventriculoperitoneal shunt to control hydrocephalus. In two of them the eye fundus examination was altered.

CONCLUSIONS

Although NCM is rare and has wide clinical variability, awareness of the disease is essential. An interdisciplinary approach is the cornerstone to improve outcomes in this pathology.

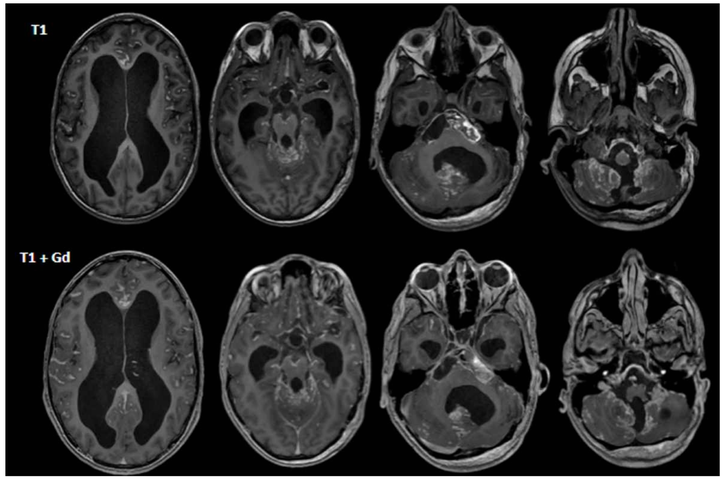

Figure 1. Brain MRI, axial cuts, **T1-weighted without contrast (top row) and with contrast (bottom row)**. In the top row, we can observe deposits lesions spontaneously hyperintense in leptomeningeal topography in brain gyri on the internal face of the frontal lobes, Sylvian fissure and cerebellar interfoliar spaces. A dominant lesion in left paramedian prepontine cistern produces mass effect on the homolateral middle cerebellar peduncle. In the bottom row, diffuse leptomeningeal enhancement is also observed which is more evident in anterior interhemispheric gyri and Sylvian fissure. Note the dysmorphic dilatation of the supra and infratentorial ventricular system.

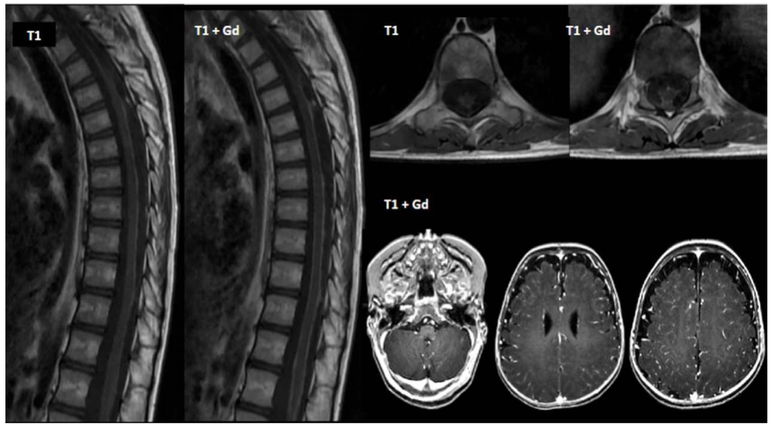

Figure 2. Dorsal spine MRI in sagittal and axial planes T1-weighted with and without gadolinium. In the records without contrast, deposits spontaneously hypertense are observed on leptomeninges, more evident in the posterior medullar contour. After the administration of gadolinium, the rest of the leptomeninges shows diffuse linear enhancement. **Brain MRI, axial cuts T1-weighted with contrast** which shows subtle leptomeningeal enhancement more notorious in gyri of the convexity.

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