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UNMASKING NEUROCUTANEOUS MELANOSIS IN A PATIENT WITH CONGENITAL GIANT MELANOCYTIC NEVUS: A CASE REPORT

Radio-Diagnosis

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ABSTRACT

Congenital melanocytic nevi (CMN) can present with neurological manifestations or complications like Neurocutaneous melanosis (NCM) with Melanocytosis/Melanomatosis. This occurs when the melanocytes invade the CNS. Investigations, specifically MRI, play a crucial role in diagnosing and characterising these abnormalities. Symptomatic NCM has a poor prognosis, and patients can present with symptoms like seizures, lethargy, and cranial nerve dysfunction. Therefore, sensitization of concerned specialities is necessary for early diagnosis and better management of such cases.

KEYWORDS

Case Report, Congenital Giant Melanocytic Nevus, Neurocutaneous Melanosis, Phakomatosis, Melanocytosis, Melanomatosis.

INTRODUCTION

A Congenital Giant Melanocytic Nevus is a pigmented/melanocytic lesion that is present at birth and reaches up to a size of >20 cm in adulthood. Despite being a rare entity, early diagnosis is crucial as it is associated with various complications like malignant transformation of the lesion, neurocutaneous melanosis, which affects the nervous system and has a severe psycho-social impact on the patient and family members. The lesions are generally brownish, with a flat and mammillated surface, are well-defined, and show hypertrichosis. The diagnosis is primarily made on clinical examination.

These lesions can affect any region of the body; the most common location is the trunk and back, followed by the limbs and head.

Neurocutaneous Melanomatosis, also known as Neurocutaneous Melanosis, is a rare but serious complication of a giant congenital melanocytic nevus. It is a sporadic phacomatosis that is characterised by multiple congenital cutaneous nevi and meningeal melanocytosis/ melanomatosis. Diagnosis is often made when the patient presents with features of raised intracranial pressure/hydrocephalus due to meningeal involvement.

It is proposed that neurocutaneous melanosis occurs as a result of congenital dysplasia of melanocyte precursors (melanoblasts), which are of neural crest cell origin and are located in the meninges, inner ear, skin, etc.

Case Report

An 18-year-old male presented with chief complaints of fever, generalised weakness, shortness of breath, left ear tinnitus and vomiting for 5 to 7 days.

On clinical examination, the patient showed multiple congenital melanocytic nevi with a giant melanocytic (bathing trunk) nevus and a few nodular lesions, which were clinically suspicious of neurofibromas. Apart from these lesions, multiple black macules were present over the trunk, back, axillary region, inguinal region, thigh and on the face.

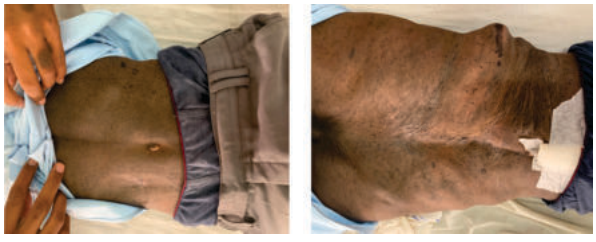

Figure 1. Clinical images of a giant melanocytic nevus covering the entire trunk and back "bathing trunk" with hypertrichosis, irregular surface and nodular areas.

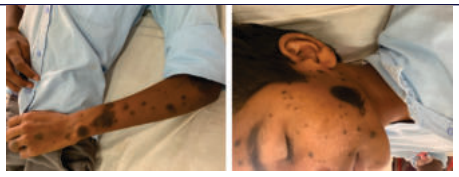

Figure 2. Multiple satellite lesions (with hypertrichosis) associated with the giant nevus are also noted on the arms and face.

The patient was referred to the ENT and ophthalmology department for further evaluation.

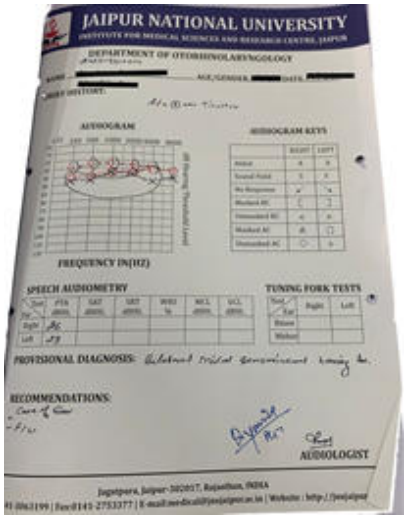

Figure 3. Audiometry was done, and it revealed bilateral mild sensorineural hearing loss.

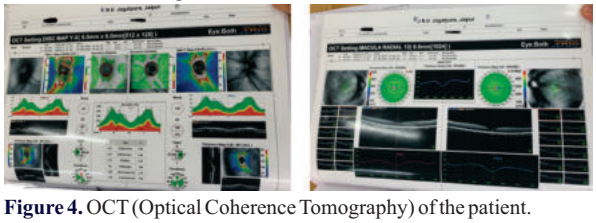

Figure 4. OCT (Optical Coherence Tomography) of the patient.

The patient was referred to the radiology department for an MRI of the Brain with Orbit and whole spine (contrast scan), which revealed the following -

MRI Brain With Contrast -

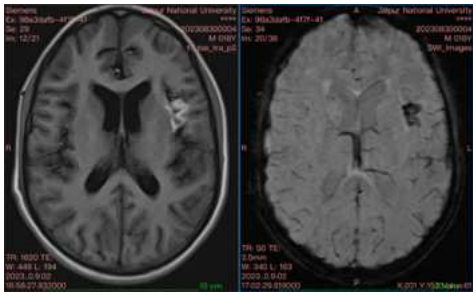

Figure 5. Nodular as well as gyri-form lesions are seen in the left insula, showing intrinsic hyperintensity on T1 images and blooming on SWI images.

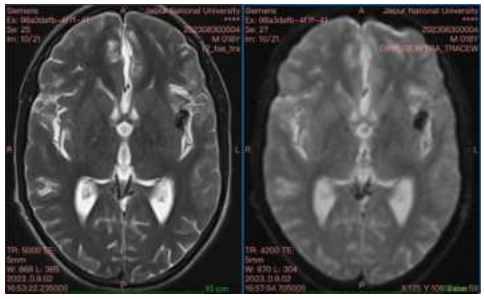

Figure 6. The same lesion appears hypointense on T2 and diffusion images.

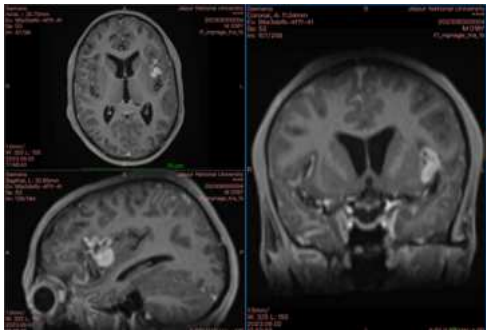

Figure 7. Enhancement of the lesions is noted on T1 post-contrast images.

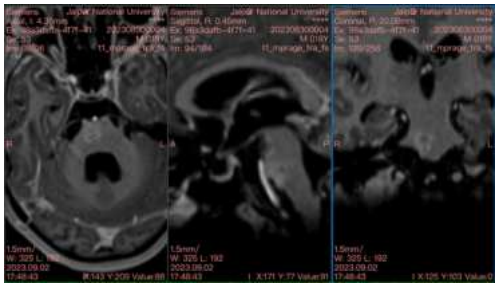

Figure 8. Subtle similar lesions are also noted in the lower pons and medulla.

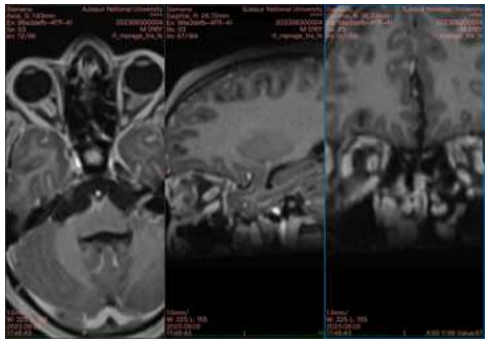

Figure 9. Enhancement is also noted in the right optic nerve sheath.

MRI Spine With Contrast -

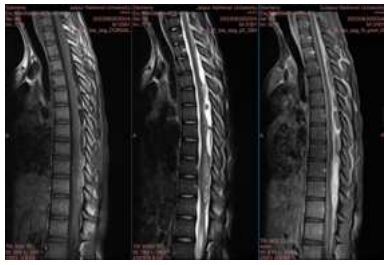

Figure 10. Multiple nodular as well as plaque-like lesions are seen in the thecal sac of the dorsal spine in the intradural extramedullary space, causing mild cord compression. These lesions show intrinsic T1 hyperintensity, appear T2 hypointense and show avid enhancement.

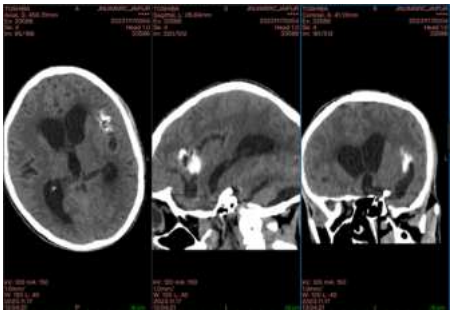

Figure 11. The patient came after 2 months for a follow-up, and the NCCT-Brain scan revealed communicating moderate hydrocephalus with multiple hyperdense parenchymal and meningeal melanocytic deposits.

DISCUSSION

Neurocutaneous melanosis is a rare neurocutaneous syndrome that is characterised by the presence of congenital giant and/or multiple melanocytic nevi of the skin with meningeal involvement. Though the exact cause is not known, it is proposed to be an aberration of the neural crest cells during the early embryonic period. It can also occur in association with Sturge-Weber syndrome and Dandy-Walker malformation.

The majority of the patients present with a large/giant nevus covering the back, known as a "bathing trunk" congenital nevus. Another rarer form exists, termed "forme fruste" or incomplete form, where the patient presents with isolated meningeal involvement without any skin lesions.

In cases with neurocutaneous melanosis/melanocytosis, the lesions can occur anywhere in the brain, predominantly involving the pons, amygdala, cerebrum and spinal cord.

Patients can present with features of raised intracranial pressure (hydrocephalus) due to meningeal melanomatosis.

Diagnosis is primarily made on clinical features combined with radiological investigations, specifically MRI. The lesions show intrinsic hyperintensity on T1-weighted images due to the presence of melanin, which has paramagnetic properties, specifically its ability to interact with the magnetic field in such a way that it shortens the T1-relaxation time.

It is important to differentiate melanin from other substances that appear bright/hyperintense on T1-weighted images, namely lipid, protein, methemoglobin, calcium, copper and manganese. Careful comparison of other sequences, like T2-weighted images (melanin shows low signal intensity), SWI and T1 post-contrast images, is crucial for accurate diagnosis.

It is important to determine the benign nature of the skin lesions, which, if not, can result in metastasis. Leptomeningeal melanomatosis is different from neurocutaneous melanosis, as in neurocutaneous melanosis dura is usually spared while the cerebral parenchyma, ependyma and choroid plexus are affected.

Prognosis is poor in symptomatic cases, especially when associated with other conditions like Dandy-Walker malformation.

No specific treatment is available; most of the therapies are aimed at reducing/controlling symptoms due to complications.

CONCLUSION

A multidisciplinary approach is needed for cases with congenital melanocytic nevi, as the condition is not only limited to the skin, but as it progresses, the patient can develop multi-system involvement. Though rare, neurocutaneous melanosis is a serious complication, especially if associated with other conditions like Dandy-Walker malformation. The diagnosis is made based on clinical features combined with MRI findings, which show lesions with intrinsic T1 hyperintensity due to the presence of melanin. A regular dermatological and neurological follow-up, along with imaging, should be advised due to the risk of malignant transformation and rapid progression of the disease.

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