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NEUROCUTANEOUS MELANOSIS : RADIOLOGICAL PERSPECTIVE

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Introduction

Neurocutaneous melanosis (NCM) is a rare, non-familial sporadic congenital neurocutaneous syndrome (3). Usually, a disease entity, diagnosed in early infancy with a spectrum of severity comprising both benign and malignant transformation. Most present with giant cutaneous melanocytic naevi (GCMN) or multiple cutaneous melanocytic naevi (MCMN) albeit presentation due nervous system complication not uncommon. Radiological evaluation plays a vital role in diagnosis as well as follow up nevertheless archetypal imaging findings are yet to be described. We present a case of NCM in perspective of presentation and diagnosis albeit proper diagnostic criteria, management guideline and poor prognostic factors are scarcely described as only handful of literature available.

Case presentation

A 5-month-old male infant presented with a few episodes of myoclonic convulsions since early neonatal period with the same semiology. He also had mild to moderate size (1-3cm in diameter) congenital melanocytic naevi distributed in the scalp, back of the chest, buttock, and lower limbs (largest) with a slight increase in size but the number and color with time. He also had several episodes of culture-positive urinary tract infection (UTI) due to partial posterior urethral valve without vesicoureteric reflux or nephropathy.

Antenatal history and perinatal history is unremarkable and no family history of any neurocutaneous diseases.

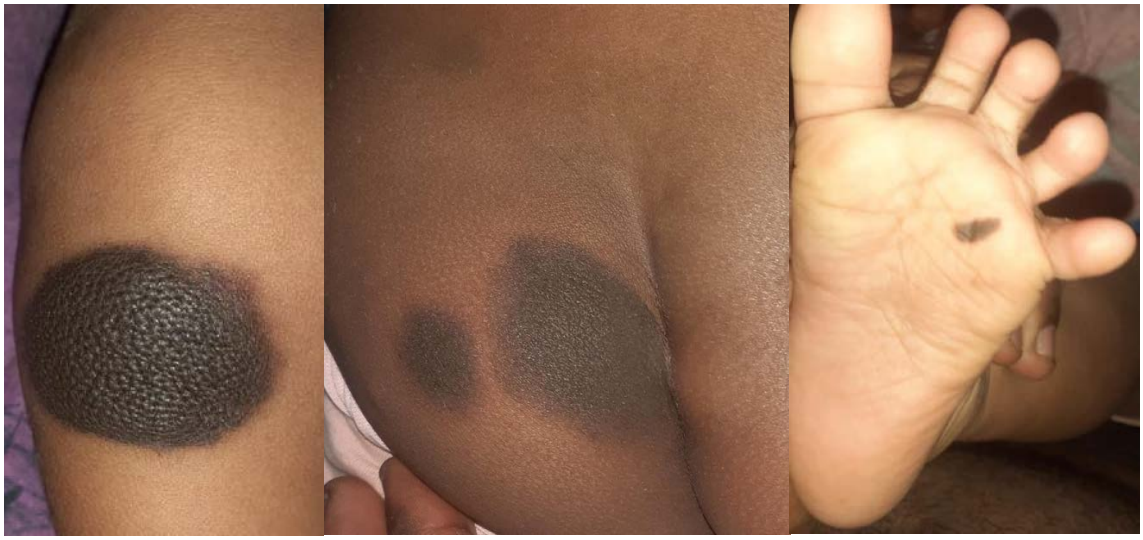

Figure 01. Cutaneous congenital melanocytic naevi in right thigh, buttock & sole of left foot

The electroencephalogram (EEG) was normal. The non-contrast computed tomography (NCCT) of the brain showed heterogenous hyperdensity in bilateral mesial temporal lobes (Right more than left), pons and bilateral cerebellar hemispheres. No evidence of hydrocephalus (Figure 02).

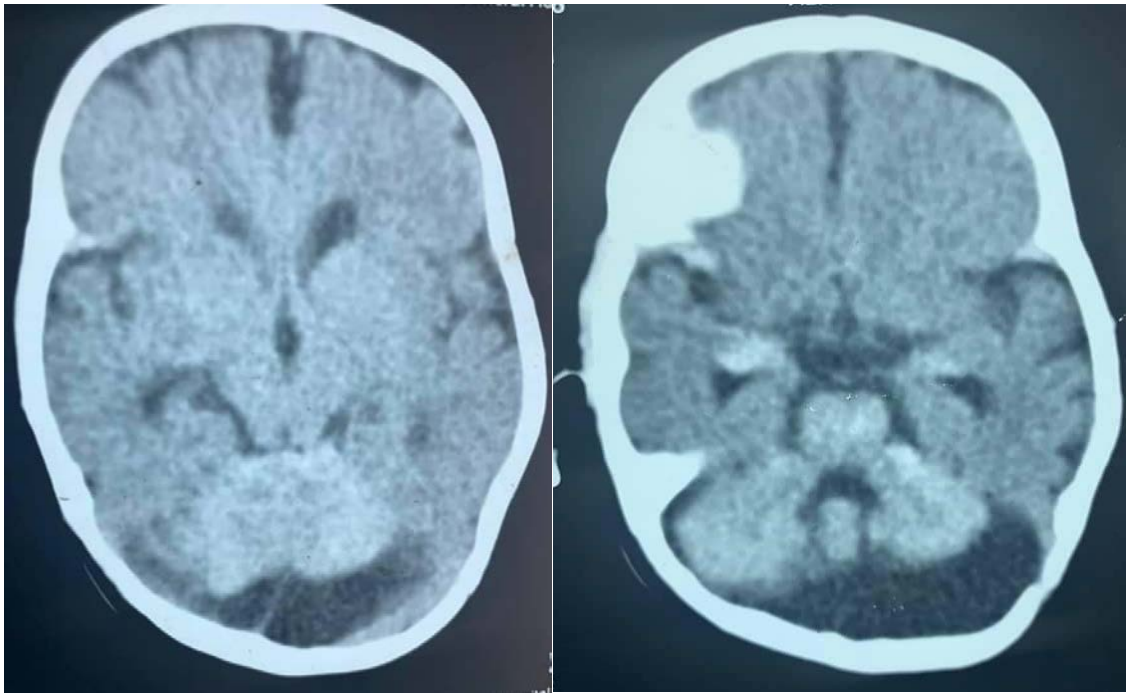

Figure 02. Non contrast brain, axial images at the level of cerebellum.

The contrast enhanced magnetic resonance imaging done with T1W/T2W volume acquisition images, FlAIR axial/coronal, SWI, DWI/ADC and post gadolinium volume acquisition. It showed high signal intensity focal areas in T1 weighted (T1W)images and iso to low signal intensity in T2 weighted(T2W) images distributed in bilateral amygdalae, ventral pons and cerebellum corresponding to hyper dense lesion in the NCCT (Figure 03). No fat suppression, blooming, diffusion restriction or contrast enhancement. Similar signal characteristics are also seen in cerebellar folia and leptomeninges of cerebellum indicating leptomeningeal involvement. No hydrocephalus was seen at the time of study.
There is a retro cerebellar arachnoid cyst measuring 3x6 cm In size with mild compression on left cerebellar hemisphere.

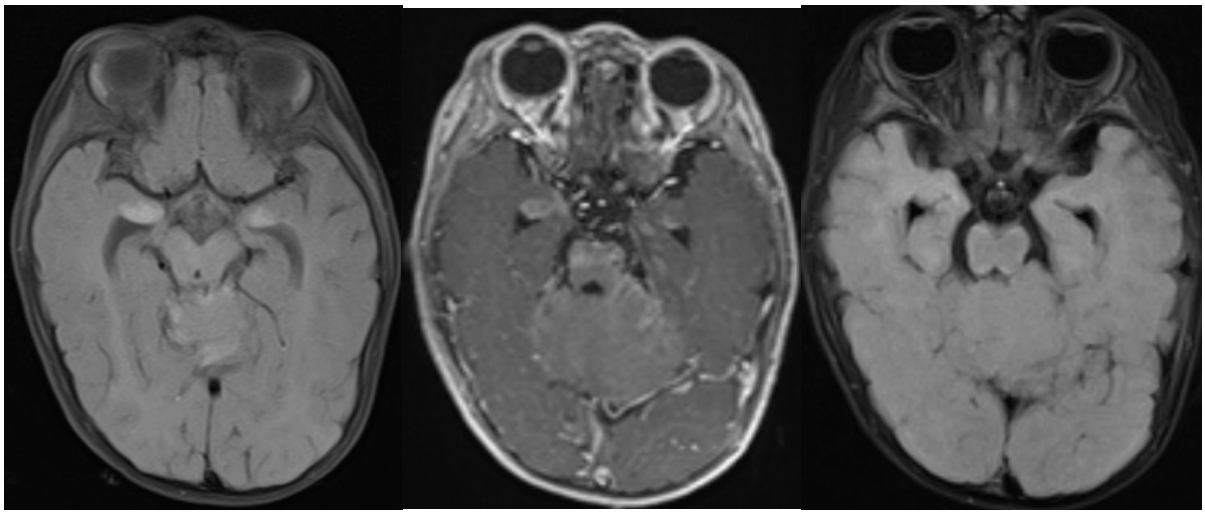

Figure 03. T1 weighted axial, T1 post contrast images, T2 FLAIR images showing melanin deposition in bilateral amygdalae, ventral pons and cerebellum.

Lumbar puncture was unremarkable and skin biopsy was not performed.

Patient is managed with antiepileptic and annual surveillance of the cerebral lesions with MRI brain and spine was planned.

Discussion

NCM is a rare, sporadic neurocutaneous disease ,but with an incidence of 23-30% in patients with neurocutaneous melanocytic naevi who are been imaged with MRI brain to assess CNS disease(3). Most of the reported cases are non-familial, but contrary some articles describe incidence of almost 25%, still no reported increased risk among monozygotic twins(1, 3). There is no gender predilection and most of the symptomatic cases present before 2nd year of the life, if so it carries a bad prognosis (5).

The definite pathogenesis and precursors are still uncertain, but abnormal neural crest differentiation having common origin to both cutaneous and leptomeningeal melanocytes is thought to be the culprit (2, 3). So the lesions distribute in skin as well as in leptomeninges and brain parenchyma around vascular beds where melanocytes are usually present.

Patients usually present with a congenital melanocytic naevi, which can be giant melanocytic naevi/GMN (2/3 of the cases) or smaller but multiple in number. The GMN usually distribute in lower back giving “bathing suit” appearance and scalp. The presence of satellite lesions are more favor in the disgnosis. In our case, multiple small congenital naevi were seen distributed in scalp, back as well as in extremities (largest lesion), but no giant naevi or satellite lesions.

They also can present with CNS manifestation, either due to symptoms and sign of increased intra cranial pressure following hydrocephalus or seizure episodes. Our patient had episodes of myoclonic seizures from the neonatal period which responded to anti-epileptic. There are reported cases of associated renal anomalies mainly upper urinary tract anomalies, Meckel’s diverticulum and cerebral posterior fossa anomalies. Our patient had a partial posterior urethral valve causing recurrent urinary tract infection, but no upper tract anonalies. (1) Diagnosis is usually made either CNS screening for suspicious cutaneous naevi or imaging of the brain and spine following CNS manifestations. The most recent diagnostic criteria contains clinical, imaging and histological integration, and it comprises 1. giant melanocytic naevi (diameter more than 20cm in adults, 9-6cm in children) or multiple in number (more than 3) with central nervous system(CNS) lesions 2. CNS involvement without cutaneous melanoma except biopsy-proven CNS disease 3. CNS disease without melanoma except in biopsy proven cutaneous benign disease (2). At least on is essential for the diagnosis (Table o1).

Table 01. Diagnostic criteria of NCM

01	Giant melanocytic naevi (diameter more than 20cm in adults, 9-6cm in children) or multiple in number (more than 3) with central nervous system(CNS) lesions
02	CNS involvement without cutaneous melanoma except biopsy proven CNS disease
03	CNS disease without melanoma except in biopsy proven cutaneous benign disease

EEG will sometimes show the epileptic focus, but not in our case as it was normal. Cerebrospinal fluid analysis occasionally gives a clue, showing elevated opening pressure, melanotic granules, leukocytes and xanthochromia due to melanine and hemorrhages (3).

Ultrasound of the brain is often normal unless the patient developed hydrocephalus due to leptomeningeal involvement.

Usually non contrast CT of the brain will show hyperdense lesions in typical sites such as, bilateral mesial temporal lobe (amygdalae), pons mainly ventrally, bilateral cerebral parenchyma or foliae and contrast enhancing leptomeningeal lesions following intra venous contrast. The MRI is the imaging choice for screening of the brain using T1W, T2W volumetric acquisition with contrast images additional to routine epilepsy protocol as well as the spine (2). The lesions characteristically show of high signal intensity in T1W, iso/low signal intensity in T2W images, without mass effect or contrast enhancement. It can also detect leptomeningeal enhancing lesions in brain and spine with or without hydrocephalus. Imaging features also can be helpful to

assess lesions for malignant transformation, with favoring features of contrast enhancement, necrosis, hemorrhages and mass effect(3,) and associated posterior fossa anomalies such as Dany walker malformation. Our patient has lesions with typical characterization and distribution and retro cerebellar arachnoid cyst without features of hydrocephalus or melanoma.
The management is usually symptomatic such as antiepileptic for seizure, ventricular shunts for hydrocephalus, and surgical excision of cutaneous lesions for cosmetic reasons. Prognosis is usually poor mainly for symptomatic patients specially due to complications of leptomeningeal involvement. Our patient's convulsions are controlled with anti epilileptic and follow up imaging recommended for screening of hydrocephalus or malignant transformation.

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