

Pictorial CME

MR Imaging in Neurofibromatosis Type I

K Mugundhan¹, M Sathishkumar, P R Sowmini², K.Sakthivelayutham²,
K Malcolm Jayaraj², R Viveka Saravanan¹

A 12 year old boy presented with flat, dark brown macules of varying size and shape present over anterior aspect of left side of chest above nipple, anterior abdominal wall, lumbosacral and left calf region since birth. He presented with seizures at the age of twelve years. He had no history of headache, vomiting, fever or any other focal neurological deficit. His Intelligence Quotient was normal.

On dermatological examination, the child showed café au lait spots of varying size and shape over anterior aspect of left side of chest above nipple and anterior abdominal wall (Fig 01). He also showed multiple café au lait spots over lumbosacral region (Fig 02). A large triangular shaped café au lait spot over left calf region (Fig 03). Based on the above clinical findings, it was diagnosed as neurofibromatosis type 1 (NF1). His EEG showed bilateral epileptiform activity. MRI Brain (T2W & FLAIR) showed focal hyperintense lesions at right basal ganglia, left periventricular whitematter and right medial cerebral peduncle suggestive of non neoplastic hamartomas (Fig 04, 05 and 06). X-ray long bones, skull PNS view and Ultrasonogram abdomen were normal.

Neurofibromatosis type 1 is associated with neurological manifestations like seizures, tumors of brain, spinal cord and optic nerve. 60-78% patients with NF1 have hyperintense lesions within the basal ganglia, thalamus, brain stem and cerebellum on MRI brain (T2w & FLAIR)¹. Hyperintense lesions are highly prevalent and characteristic in patients with NF1. MR imaging contributes to a definitive diagnosis of NF1².

This case highlights the characteristic MRI findings of neurofibromatosis type1.



Fig 01 - café au lait spots of varying Size & shape over anterior aspect of leftside of chest above nipple and anterior abdominal wall.



Fig 02 - Multiple café au lait spots over lumbo sacral region

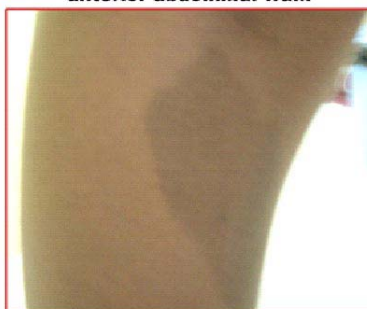


Fig 03 - A large triangular shaped café au lait spot over left calf region



Fig 04 - MRI (T2W) showing hyperintense lesion over right basal ganglia



Fig 05 - MRI (FLAIR) showing hyperintense lesion over left peri ventricular white matter region.



Fig 06 - MRI (FLAIR) showing hyperintense lesion over right medial cerebral peduncle.

REFERENCES

- 1 Walter G Bradley — Neurology in clinical practice 2008;2: 1874-77
- 2 Menor, F., Marti-Bonmati, L., Arana, E., Poyatos, C., et al — 1998, "Neurofibromatosis type1 in children: MR imaging and followup studies of central nervous system findings" Eur J Radiol, Vol.26, PP, 121-131

¹Associate Professor, Department of Neurology, Govt. Stanley Medical College, Chennai 600001

²Assistant Professor, Department of Neurology, Government Stanley Medical College, Chennai 600001