Article published online: 2025-02-20

Neurofibromatosis type 1 and Hemimegalencephaly: A Rare Association

Bramhini Bhargavi Koneti[1] Singanamalla Bhanudeep[2]

[1] Department of Dermatology, Kaya Clinic, Jubilee Hills, Above Krishna Pearl, Hyderabad, Telangana, India
[2] Department of Pediatrics, Paramitha Group of Hospitals, Hyderabad, Telangana, India

Address for correspondence Singanamalla Bhanudeep, DM, Consultant Pediatric Neurology, Paramitha Group of Hospitals, Hyderabad, Telangana, India (e-mail: singanamalabhanudeep0@gmail.com).

Int J Ep

A 2-month-old boy, second-born to healthy parents, was brought with left-sided clonic jerks and asymmetric spasms in clusters for the past 10 days. His birth history was uneventful. He cannot recognize his parents, does not follow objects, and does not turn his head to sounds. On examination, his head circumference was 39 cm (Z-score of -0.26). He has multiple café au lait macules over the trunk and back (►**Fig. 1A, B**), normal tone, and just elicitable deep tendon reflexes with no paucity of movements. Electroencephalography revealed a hemi-hypsarrhythmia pattern (►**Fig. 1C**). Neuroimaging was suggestive of right hemimegalencephaly (►**Fig. 2**). He was started on vigabatrin and oral steroids (4 mg/kg/d). Whole exome sequencing showed an autosomal dominant heterozygous pathogenic missense variant (c.3104T > G) in exon 23 of the *NF1* gene. On follow-up till 7 months of age, the spasms reduced to two per day. He had partial neck control and cannot roll over, but can recognize his parents. The parents were advised for hemispherotomy and the surgery was delayed due to the financial constraints.

Hemimegalencephaly is a hamartomatous overgrowth involving one cerebral hemisphere that occurs due to congenital malformation of cortical development secondary to abnormal cell proliferation and apoptosis.[1] It can occur in isolation or often associated with various neurocutaneous syndromes such as organoid nevus syndrome, Proteus' syndrome, tuberous sclerosis, hypomelanosis of Ito, McCune–Albright syndrome, CLOVES syndrome, Klippel–Trenaunay syndrome, and rarely neurofibromatosis type 1.[1–3] The infants usually present with drug-resistant seizures and severe global development delay. The recommended treatment is early hemispherotomy.[1]

Authors' Contributions

B.S. and B.B. reviewed the literature, prepared the initial draft of the manuscript, and share the first authorship.
B.B. provided the concept, design, intellectual content, and finalized the manuscript.
All the authors approved the final version of the manuscript.

Funding

None.

Conflict of interest

None declared.

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DOI https://doi.org/10.1055/s-0045-1802981.
ISSN 2213-6320.

Thieme Medical and Scientific Publishers Pvt. Ltd., A-12, 2nd Floor, Sector 2, Noida-201301 UP, India

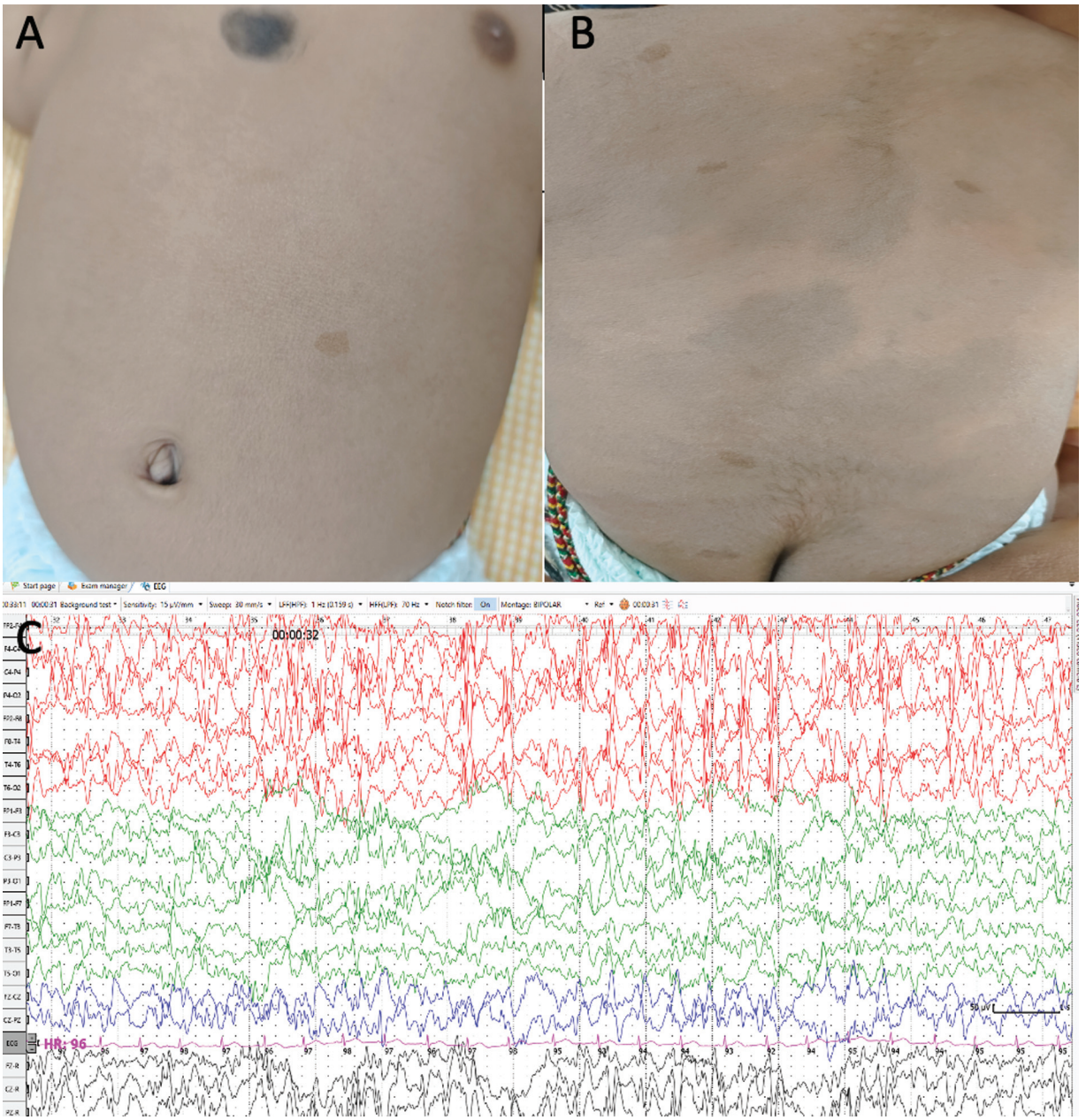

Fig. 1 (**A**, **B**) Multiple café au lait macules over the trunk and back with extensive Mongolian spots. **(C)** Electroencephalography (sensitivity: 15 mcv/mm; sweep: 30 mm/s) showing high-voltage multifocal independent spike discharges over the right hemisphere.

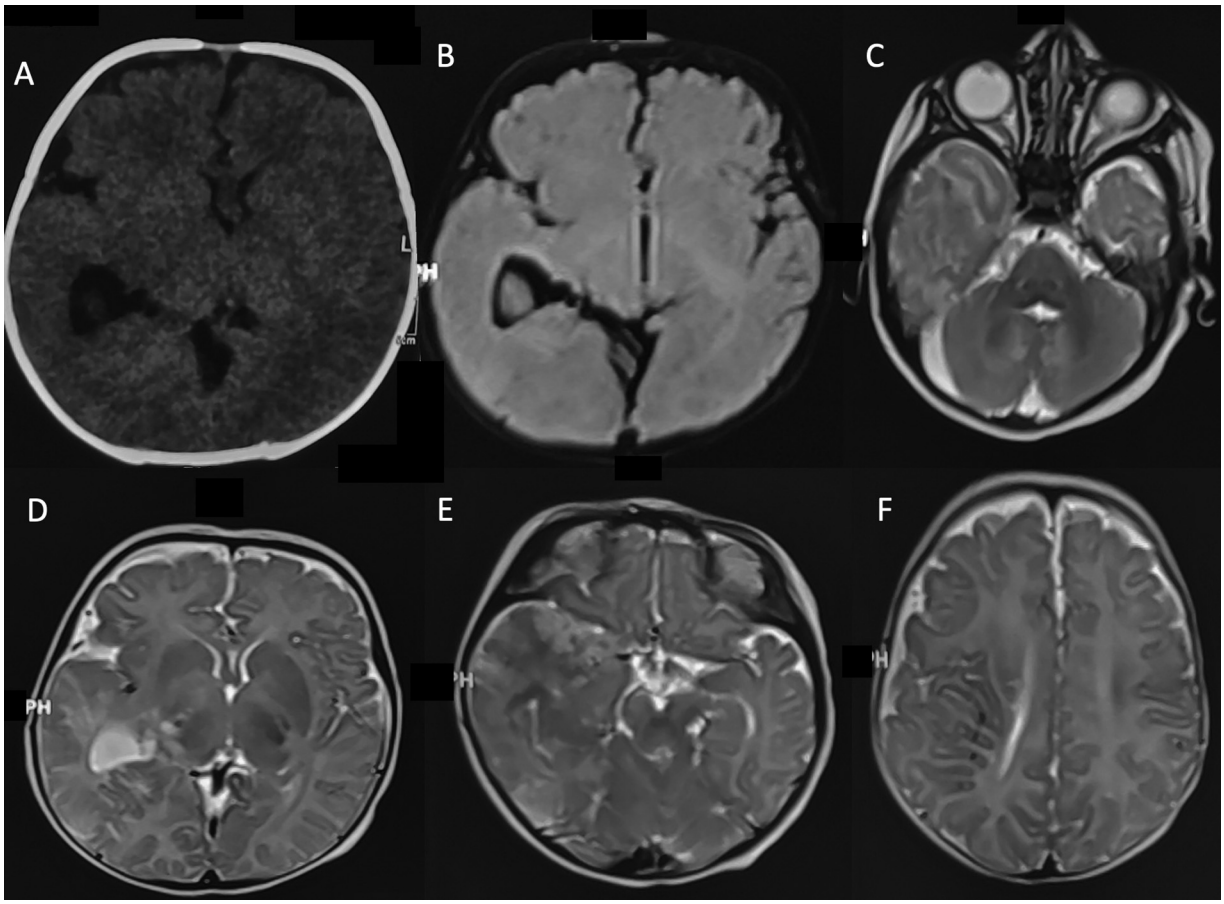

Fig. 2 (A) Noncontrast computed tomography of the brain showing large right temporal and occipital lobes with enlarged temporal horn, (B) fluid attenuated inversion recovery weighted image showing right hemimegalencephaly in the right temporal lobe, and (**C–F**) T2-weighted images showing right hemimegalencephaly with associated white and gray matter hyperintensities, heterogeneous right thalamoganglionic region, and corona radiata.

