

Transmantle Heterotopia or Closed Lip Schizencephaly: A Diagnostic Dilemma

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CASE HISTORY

A 41-year-old adult male presented with two episodes of generalized tonic clonic seizures. He had past history of seizures from childhood with seizure-free intervals, and the last episode was about 2 years back. The patient was taking phenytoin off and on for last 10 years which was prescribed by a local practitioner. On physical examination, the patient was afebrile, disoriented, and showing gum hypertrophy. The neurological examination suggested dysarthria, nystagmus, and positive cerebellar signs. The patient had not been evaluated with any previous imaging for the epilepsy. Magnetic resonance imaging (MRI) was done with 1.5 Tesla GE machine. On MRI, T1- and T2-weighted sagittal images showed continuous altered signal intensity areas at two sites in the right frontal lobe extending from the ventricular wall to the cortex [Figure 1a & 1b]. The altered signal intensity areas were iso-intense to gray matter on all pulse sequences without obvious intervening cerebrospinal fluid (CSF) cleft. They were seen continuous with overlying gray matter of the cortex and causing bulge on the lateral ventricular wall. The left cerebral hemisphere showed multiple foci of similar signal intensity in the subcortical region of the frontal lobe on T1- and T2-weighted images [Figure 1c and 1d]. In addition to the above imaging findings, there was diffuse bilateral cerebellar and vermian atrophy with prominent posterior fossa extra-axial spaces [Figure 1e & 1f]. The bilateral cerebellar atrophy was developed as consequences of long-term phenytoin use explaining positive neurological examination.

DISCUSSION

The continuous layer of gray matter extending from the ventricle to cortex is seen in transmantle heterotopia and closed lip schizencephaly, both of which are difficult to differentiate on imaging. Transmantle heterotopia is a rare entity in which gray matter is seen extending from the ventricular wall to overlying cortex without intervening white matter or CSF cleft.^[1-5] Schizencephaly refers to gray matter lined cleft extending from the ventricular wall to cortex and can be open or closed lip type. In open lip, the cleft walls are well separated and filled with CSF, whereas they are in apposition in closed type.^[1,4] The differentiating feature between closed lip schizencephaly and transmantle heterotopia is the CSF cleft which is sometimes difficult to discern.^[5] In our opinion, the best differentiating feature between these two conditions is “dimple sign” which

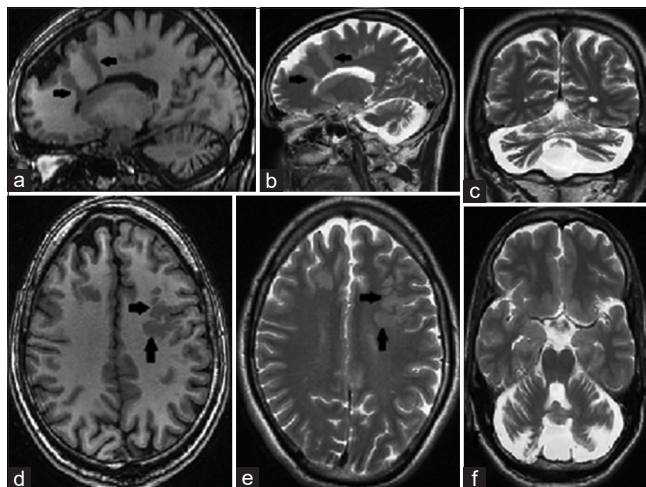


Figure 1: Magnetic resonance imaging (MRI) T1- (a) and T2- (b) weighted sagittal images showing continuous layer of gray matter in the right frontal lobe (arrows) at two sites extending from ventricular wall to cortex and causing bulge on the ventricular wall without intervening CSF cleft suggesting transmantle heterotopia (typical imaging features to differentiate from closed lip schizencephaly). T1- (c) and T2- (d) weighted axial MRI images showing multiple foci of gray matter in the left frontal lobe (arrows) suggesting subcortical heterotopia. T2-weighted coronal (e) and axial (f) MRI images showing bilateral cerebellar and vermian atrophy

is always seen in closed lip schizencephaly. In closed lip schizencephaly, a dimple is seen on the ventricular outline where gray matter meets the ventricular wall while heterotopic gray matter bulges into the wall. In our case, the absence of CSF cleft and dimple sign with gray matter bulging on the ventricular wall suggested transmantle heterotopia in the right cerebral hemisphere with focal subcortical heterotopia in the contralateral hemisphere.

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Submitted: 19-Jan-2021 **Revised:** 12-Feb-2021 **Accepted:** 26-Feb-2021

Published: 21-May-2021

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DOI: 10.4103/aian.AIAN_33_21

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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