Simultaneous multiple congenital anomalies in a young male

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Congenital malformations associated with disorders of neuronal migration leading to seizure disorder in a young population are rarely seen. Here, we report, six congenital malformations simultaneously present in brain imaging in a young patient. A 35-year-old male presented with a history of seizure disorder since childhood presented in the clinic with on and off episodes of seizures. The seizures were right focal with secondary generalized tonic-clonic or generalized tonic-clonic type but he was not taking medications regularly. His peri-natal history was asymptomatic. There was no developmental delay in milestones. Furthermore, there was no significant family history. General, systemic, and neurological examinations were within normal limits. On investigations, his magnetic resonance imaging brain showed open lip schizencephaly, polymicrogyria, hetero-trophic grey matter in the left frontoparietal region, focal cortical dysplasia, and corpus callosum agenesis (Fig. 1a-e). He also had evidence of cerebrospinal fluid signal intensity cystic spaces in the posterior fossa directly communicating with the fourth ventricle, compressing the cerebellar hemispheres, and absence of vermis suggestive of Dandy-walker malformation (Fig. 1f). Genetic analysis was not done in this patient. The patient earlier tried various antiepileptics, but his seizures finally got well controlled on tablet zonisamide 150 mg twice daily and tablet clonazepam 0.25 mg thrice daily. At present, the patient is working in the education department and doing well despite all these malformations.

Various congenital malformations associated with seizure disorders are described [1-3]. Most of them are due to a direct result of faulty neuroblast migration [4]. During early gestation (8–15 weeks), most severe migratory defects occur, affecting events in the gross formation of the neural tube and cerebral vesicles. Later defects of neuronal migration can present as disorders of cortical lamination or gyration such as lissencephaly, pachygyria, and cerebellar dysplasias [5]. In our patient, six different congenital malformations were present with the main presentation of generalized tonic-clonic seizures, and seizures were adequately controlled with anti-epileptic drugs. Therefore, congenital malformations, especially disorders of neuronal

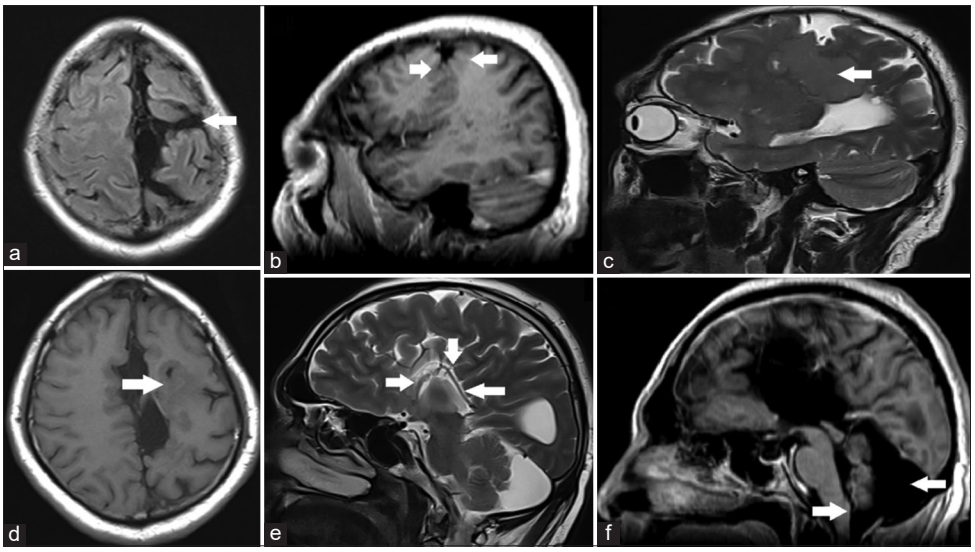

Figure 1: Multiple congenital abnormalities seen in MRI marked with white arrows. (a) Open lip schizencephaly, (b) polymicrogyria, (c) hetero-trophic grey matter in left fronto-parietal region, (d) focal cortical dysplasia, (e) corpus callosum agenesis, and (f) Dandy-walker malformation

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migration need to be identified as an important cause of seizure disorder. Medical management is effective despite multiple malformations.

CONTRIBUTORS DETAILS

All authors made equal contribution in study design, data analysis, patient care, patient management, data acquisition, and manuscript writing.

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