



HEMIMEGALENCEPHALY PRESENTING WITH EARLY-ONSET EPILEPTIC ENCEPHALOPATHY AND DEVELOPMENTAL DELAY: A CASE REPORT

Dr. Nivedha M*	Junior Resident, Department of Paediatrics, Sree Balaji Medical College and Hospital, Chromepet, Chennai. *Corresponding Author
Dr. Sree Divya M	Assistant Professor, Department of Paediatrics, Sree Balaji Medical College and Hospital.
Dr. Sundari S	Head of Department, Department of Paediatrics, Sree Balaji Medical College and Hospital.

ABSTRACT Hemimegalencephaly is a rare congenital malformation of cortical development characterized by enlargement and dysplasia of one cerebral hemisphere and is commonly associated with early-onset epilepsy and developmental delay. We report a two-year-old male child with recurrent seizures since the third day of life and global developmental delay. The seizures were characterized by sudden stiffening of the limbs, up-rolling of the eyes, and jerky movements predominantly involving the left upper limb. Neurological examination revealed contralateral spastic hypertonia. Magnetic resonance imaging showed enlargement of the right cerebral hemisphere involving the frontal and temporal lobes, with cortical dysplasia and abnormal gyral patterns, consistent with right hemimegalencephaly. Video electroencephalography demonstrated continuous periodic lateralized epileptiform discharges with suppressed background activity, suggestive of severe epileptic encephalopathy. The child was treated with multiple antiseizure medications and pulse methylprednisolone therapy and was advised regular neurological follow-up and developmental rehabilitation. This case emphasizes the importance of early neuroimaging and electrophysiological assessment in children with early-onset seizures and developmental delay.

KEYWORDS : Cortical Dysplasia; Epileptic Encephalopathy; Magnetic Resonance Imaging; PI3K-AKT-mTOR Signaling Pathway

INTRODUCTION

Malformations of cortical development are an important cause of epilepsy and developmental delay in children. These abnormalities arise from disturbances in neuronal proliferation, migration, or cortical organization during fetal brain development (Guerrini & Filippi, 2005; Leventer et al., 2008). Hemimegalencephaly is a rare congenital disorder characterized by enlargement and dysplastic development of one cerebral hemisphere and is frequently associated with severe epilepsy, developmental delay, and neurological deficits (Flores-Sarnat, 2002). Magnetic resonance imaging plays a central role in establishing the diagnosis.

Case Report

A two-year-old male child presented with recurrent seizures and delayed developmental milestones. The child had a history of seizures since the third day of life. The episodes were described as sudden stiffening of the limbs associated with up-rolling of the eyes and jerky movements predominantly involving the left upper limb, occurring approximately once per month.

The child was born at term to non-consanguineous parents by normal vaginal delivery with a birth weight of approximately 2.9 kg. The neonatal period was complicated by seizures occurring on the third day of life, requiring neonatal intensive care admission. Developmental assessment revealed global developmental delay.

On examination, the child was conscious and responsive. Vital signs were stable. Neurological examination revealed global developmental delay with contralateral spastic hypertonia. No dysmorphic features or neurocutaneous markers were noted.



Figure 1. Clinical Photograph of the Child; the Eyes are Masked to Protect Identity.

Magnetic resonance imaging of the brain revealed enlargement of the right cerebral hemisphere involving the frontal and temporal lobes, with cortical dysplasia and abnormal gyral patterns consistent with right hemimegalencephaly.

Video electroencephalography demonstrated continuous periodic lateralized epileptiform discharges with suppressed background activity, suggestive of severe epileptic encephalopathy.

The child was treated with multiple antiseizure medications, including levetiracetam, oxcarbazepine, and clonazepam. Pulse methylprednisolone therapy was also administered. Clinical improvement was noted, and the child was discharged with advice for regular neurological follow-up and developmental rehabilitation.

DISCUSSION

Hemimegalencephaly is a rare malformation of cortical development characterized by unilateral hemispheric enlargement and dysplasia resulting from disturbed neuronal proliferation and cortical development (Flores-Sarnat, 2002; Leventer et al., 2008). In the present child, the very early onset of seizures and global developmental delay were consistent with the recognized clinical spectrum of this disorder.

Molecular studies have linked hemimegalencephaly to abnormalities in growth-regulating pathways, particularly the PI3K-AKT-mTOR/GATOR1 signaling network. Germline or mosaic variants affecting these pathways can lead to abnormal neuronal growth, cortical organization, and hemispheric enlargement (Edmonds et al., 2024; Vasquez et al., 2024).

Clinically, hemimegalencephaly commonly presents with seizures beginning in infancy or the neonatal period, developmental delay, hemiparesis, and variable cognitive impairment. Epilepsy is frequently drug resistant (Günbey et al., 2025; Flores-Sarnat, 2002). The neonatal onset of seizures and contralateral spastic hypertonia in this child are therefore characteristic of the condition.

Magnetic resonance imaging is the investigation of choice. Typical abnormalities include enlargement of the affected hemisphere, cortical thickening or dysplasia, abnormal gyral patterns, and ventricular asymmetry (Günbey et al., 2025; Sepulveda et al., 2024). In this case, MRI demonstrated enlargement of the right cerebral hemisphere with cortical dysplasia and abnormal gyral patterns.

Electroencephalography is useful for defining the epileptiform burden and assessing the severity of the epileptic encephalopathy. The present

child showed continuous periodic lateralized epileptiform discharges with suppressed background activity, indicating marked hemispheric epileptiform dysfunction.

Management is directed primarily toward seizure control and developmental support. Antiseizure medications are often insufficient in drug-resistant disease, and functional hemispherectomy or hemispherotomy may be considered in appropriately selected children (Edmonds et al., 2024; Pepi et al., 2023). Early multidisciplinary evaluation can facilitate timely treatment planning, surgical assessment when indicated, and rehabilitation.

CONCLUSION

Hemimegalencephaly should be considered in children with seizures beginning in the neonatal period or early infancy, particularly when accompanied by developmental delay and focal neurological abnormalities. MRI and EEG are central to diagnosis and characterization. Long-term management requires seizure control, developmental rehabilitation, regular neurological follow-up, and consideration of epilepsy surgery in drug-resistant cases.

REFERENCES

- a. Günbey, C., Oğuz, K. K., Bilginer, B., Öztoprak, Ü., Ilgaz, F., Akalan, N., Topçu, M., Turanlı, G., & Yılmazoğlu, D. (2025). Clinical and radiological evaluation of children with hemimegalencephaly and epilepsy: A single-center study. *Seizure*, 132, 186–191. <https://doi.org/10.1016/j.seizure.2025.09.011>
2. Edmonds, B., Ngo, J. P., Groves, A., Reyes, B., Gott, R. A., Chia, D. J., Mirbaha, H., Magaki, S., Khanlou, N., Pineles, S. L., Salamon, N., Thompson, R. M., Newman, M., Rajaraman, R. R., Hussain, S. A., Fallah, A., Russell, B., & Nariai, H. (2024). Multidisciplinary team approach for pediatric hemimegalencephaly: Insights from a single institutional case series. *Epilepsia Open*, 9(6), 2510–2517. <https://doi.org/10.1002/epi4.13079>
3. Sepulveda, W., Sepulveda, F., Schonstedt, V., Stern, J., & Diaz-Serani, R. (2024). Neuroimaging findings in fetal hemimegalencephaly: Case study and review. *Fetal Diagnosis and Therapy*, 51(2), 133–144. <https://doi.org/10.1159/000535406>
4. Vasquez, A., Miller, K. J., Youssef, P. E., Selcen, D., Patterson, M. C., & Starnes, K. (2024). A case report of hemimegalencephaly with super-refractory status epilepticus and brain atrophy associated with NPRL3 gene mutation. *Seizure*, 116, 156–158. <https://doi.org/10.1016/j.seizure.2023.02.010>
5. Pepi, C., De Benedictis, A., Rossi-Espagnet, M. C., Cappelletti, S., Da Rold, M., Falcicchio, G., Vigeveno, F., Marras, C. E., Specchio, N., & De Palma, L. (2023). Hemispherotomy in infants with hemimegalencephaly: Long-term seizure and developmental outcome in early treated patients. *Brain Sciences*, 13(1), 73. <https://doi.org/10.3390/brainsci13010073>
6. Guerrini, R., & Filippi, T. (2005). Neuronal migration disorders, genetics, and epileptogenesis. *Journal of Child Neurology*, 20(4), 287–299. <https://doi.org/10.1177/08830738050200040401>
7. Flores-Sarnat, L. (2002). Hemimegalencephaly: Part 1. Genetic, clinical, and imaging aspects. *Journal of Child Neurology*, 17(5), 373–384. <https://doi.org/10.1177/088307380201700512>
8. Leventer, R. J., Guerrini, R., & Dobyns, W. B. (2008). Malformations of cortical development and epilepsy. *Dialogues in Clinical Neuroscience*, 10(1), 47–62. <https://doi.org/10.31887/DCNS.2008.10.1/rjleventer>
9. Kliegman, R. M., St. Geme, J. W., Blum, N. J., Shah, S. S., Tasker, R. C., & Wilson, K. M. (Eds.). (2023). *Nelson textbook of pediatrics* (22nd ed.). Elsevier.