Journal of Bangladesh College of Physicians and Surgeons
Vol. 44, No. 1, January 2026

Dravet Syndrome: A Case Report

A AKTER[a], ST ALAM[b], S AHMED[c]

Abstract

Dravet syndrome is a rare genetic epileptic encephalopathy characterized by drug resistant epilepsy. Patients present with prolong febrile seizure with normal developmental status in infancy. However patient may experience a halt or decline of development usually after one year of age. This type of presentation may mimic other neurological disorder like simple febrile seizure, mitochondrial disorder and other epileptic encephalopathies. Though clinical presentations almost similar, confirmation require genetic evaluation for counseling and management purpose. We recently observed a two-year-old boy with Dravet syndrome confirmed by identifying mutation of SCN1A gene. MRI of brain revealed moderate cerebral atrophy and enlarged brain ventricles.

Keywords: Dravet syndrome, Epileptic encephalopathy, SCN1A Gene

(J Bangladesh Coll Phys Surg 2026; 44: 64-67)
DOI: https://doi.org/10.3329/jbcps.v44i1.87291

Introduction:

Dravet syndrome is a rare and severe form of epileptic encephalopathy previously known as severe myoclonic epilepsy of infancy (SMEI), typically presents initially as complex febrile seizures during infancy. This condition was first identified by Charolette Dravet in 1978.[1] Dravet syndrome is characterized by repeated febrile , afebrile seizure including epileptia pertialis continua that are resistant to medication, along with various associated conditions such as cognitive impairments, behavioral issues, sleep disturbances, walking abnormalities, and an increased risk of sudden unexpected death in epilepsy (SUDEP).

The prevalence of Dravet syndrome (DS) is estimated to be approximately 1 in every 15,700 births.[2,3] Children with Dravet syndrome usually show normal development until the onset of seizures. Seizures typically begin within the first year of life, followed by developmental delays. The initial seizures often manifest as prolong focal seizures, commonly resembling febrile seizures, which can make them challenging to distinguish from benign febrile seizures. Triggers for Dravet syndrome episodes may include infections, vaccinations, hot weather, warm baths, fatigue, sun exposure, or physical exertion.

Approximately 80% of individuals affected by Dravet syndrome have pathogenic mutations in the SCN1A gene, which codes for the voltage-gated sodium channel alpha I.[4,5] Other genes such as PCDH19, SCN2A, SCN8A, SCN1B, GABRA1, GABRB3, GABRG2, KCNA2, CHD2, STXBP1, and others may also contribute to Dravet syndrome or symptoms resembling those of Dravet syndrome.[6] Apart from Dravet syndrome, SCN1A Mutation also causes other epileptic encephalopathies, like genetic epilepsy with febrile seizure plus (GEFS+), West syndrome, Lennox Gestaut syndrome (LGS), Epilepsy of infancy with migrating focal seizure (EIMFS),Rett Syndrome.[7]

Early seizures in Dravet syndrome may resemble other neurological disorders. These include simple febrile seizure, mitochondrial disorder and other epileptic encephalopathies such as West syndrome, LGS, and EIMFS. Dravet syndrome can be distinguished from other conditions through a thorough analysis of medical history and clinical examination. In simple febrile seizures, the developmental status remains unaffected. Other epileptic encephalopathies can be distinguished based on seizure semiology. In situation involving mitochondrial disorder, a suggestive family history and multisystem involvement are key factors in differentiation.

We present a case of a 2-year-3-month-old male child with clinical suspicions of Dravet syndrome, which was

a. Dr. Aireen Akter, Department of Pediatric Neurology, Institute For Paediatric Neurodisorder And Autism in BMU, Dhaka, Bangladesh

b. Prof. Dr. Syeda Tabassum Alam, Professor and Director, Department of Pediatric Neurology, Institute For Paediatric Neurodisorder And Autism in BMU, Dhaka, Bangladesh

c. Dr. Sanjida Ahmed, Assistant Professor, Department of Pediatric Neurology, Institute For Paediatric Neurodisorder And Autism in BMU, Dhaka, Bangladesh

Address of Correspondence: Dr. Aireen Akter, Department of Pediatric Neurology, Institute For Paediatric Neurodisorder And Autism in BMU, Dhaka, Bangladesh, E-mail: aireenakter785@gmail.com, Mobile: 01926300970

Received: 07 Dec., 2025 **Accepted:** 15 April 2025

confirmed through genetic testing revealing a mutation in the SCN1A gene, responsible for encoding the voltage-gated sodium channel.

Case presentation: A 2 year 3 month old boy, immunized, 1st issue of his non consanguineous parents got admitted into inpatient department of pediatric neurology, IPNA, Bangladesh Medical University, Dhaka, Bangladesh with the complaints of repeated seizure since 3 months of age. According to informant mother, he was previously healthy until 3 months of age, after which he experienced first seizure associated with fever following vaccination. Since then he developed febrile seizure for several episodes at 10 to 15 days interval. Initially, the seizures manifested as focal clonic episodes of prolonged duration, which subsequently evolved into generalized tonic-clonic seizures, each lasting approximately 10 to 15 minutes. Over the past year, he required admission in intensive care unit (ICU) on three occasions due to prolonged febrile seizures following vaccination.

The patient was initially managed with intermittent prophylaxis for febrile seizure using oral diazepam and acetaminophen during febrile illnesses. Despite these measures, the frequency and duration of his seizures progressively increased. Then he was treated by registered physician with oral oxcarbazepine, considering the presence of focal seizures. However, seizure control was not achieved; instead, his seizure burden worsened. Consequently, he was admitted to the Department of Pediatric Neurology at **BMU** for further evaluation and management.There, he was treated with sodium valproate, followed by clobazam , after discontinuing oxcarbazepine, leading to some improvement of seizure.

The mother also noticed a decline in motor skills with difficulty in ambulation along with speech regression over the past 3 months, accompanied by abnormal posturing . Despite treatment with multiple antiepileptic medications, there has been no observed significant improvement in his developmental status.His birth history was unremarkable. Developmentally, he achieved age-appropriate milestones up to 2 years of age.

During examination, he presented as ill-looking but conscious, alert, and responsive to his environment, having a dystonic posture. He was afebrile, anicteric, hemodynamically stable, and thriving well anthropometrically. Skin examination was unremarkable with a visible BCG scar. Developmentally, he previously met appropriate milestones but currently exhibits a loss of neck control, social smile, and speech.

Neurological assessment revealed normal muscle bulk, increased tone in all four limbs, 3/5 muscle power in all extremities, bilaterally exaggerated reflexes, and extensor plantar responses. Cranial nerve and sensory examination findings were within normal limits. Systemic examinations did not reveal any additional abnormalities.

The initial EEG exhibited normal findings upto 1 year, but subsequent assessments indicated generalized slowing and focal epileptiform discharge. MRI results of the brain revealed moderate generalized cortical atrophy and ventricular dilatation. Whole exome sequencing till now not done in our country so we did it abroad which identified a mutation in the SCN1A gene.

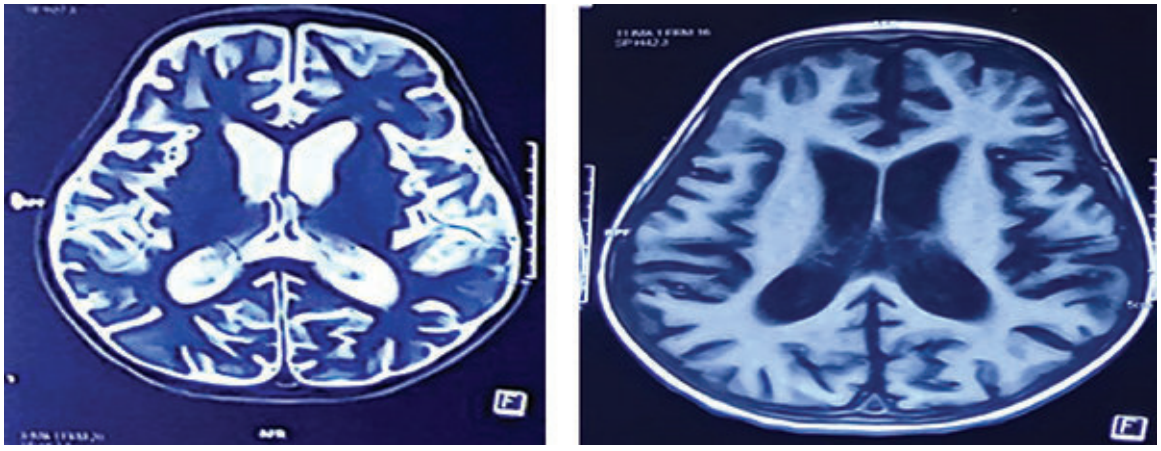

Figure-1: *Axial brain MRI showing cortical atrophy and ventricular dilatation.*

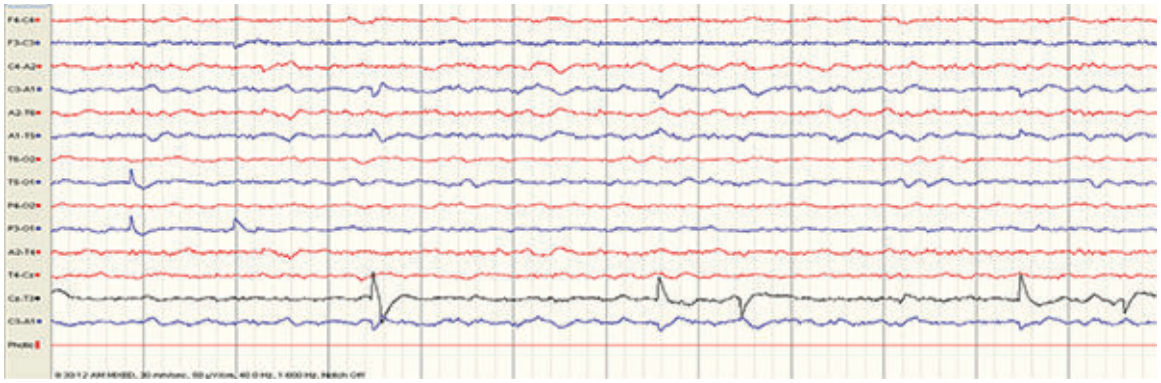

Figure-2: *Electroencephalography shows slowing with no epileptiform discharge at 1 year of age.*

Table-I

Result of Whole Exome Sequencing Report

Case		Variance	Gene	Zygosity	Inheritance	Disease	Significance
Piyarul 2year 3 months	Repeated prolonged febrile seizure, Neuroregression	c.3078del p.Arg1028GlufsTer18 [Depth:55x]	SCN1A (NM_001165963.4)	Heterozygous	Autosomal Dominant	SCN1A related disorder	Pathogenic

Discussion:

Dravet syndrome, a rare disorder typically seen in infancy and childhood, poses a challenge due to frequent misdiagnosis and limited awareness. Identifying the condition accurately and starting early treatment in affected children can be notably difficult. The primary seizure type in Dravet syndrome within the first year of life is febrile focal seizure, with some patients also experiencing myoclonic and other types of seizures.[8,9] Our patient exhibited focal prolonged febrile seizures. These seizures are commonly triggered by fever, illness, immunization, and exposure to hot water in the initial year of life. In our case, the patient required ICU admission three times, each episode being triggered by vaccination.[10]

The first episode of prolonged febrile focal convulsion in an otherwise normal infant has many potential etiologies. Children with DS are frequently initially diagnosed with febrile seizure or febrile status epilepticus. Focal epilepsy due to an occult structural lesion should be excluded by neuroimaging. Other differential diagnoses include myoclonic epilepsy of infancy , epilepsy with myoclonic atonic seizure, progressive myoclonus epilepsy (PME), Lennox-Gastaut Syndrome (LGS), mitochondrial disorder[11]. These alternatives were excluded through a thorough review of history, clinical symptoms, examination results, and EEG.

Research on Dravet syndrome in Asian countries is scarce, with only a few case series and reports published. An Indian case series detailed nine cases of Dravet syndrome, all showing consistent history and clinical course. Seizures in Dravet syndrome are often provoked by fever, illness, immunization, and exposure to hot water in the first year of life.[12] Our patient's symptoms appeared earlier than the typical age of onset. Initially, affected infants usually exhibit normal physical and psychomotor development before seizure onset. However, developmental stagnation and regression typically begin around the second year of life. Our patient showed normal development until 2 years of age, after which cognitive and motor regression, along with abnormal involuntary movements like dystonia, became evident. These observations are consistent with findings from other studies.

EEG features in Dravet syndrome are usually normal initially but become abnormal as seizure frequency rises, typically after the first year of life. [13] In our case, similar

pattern was found during EEG evaluation. MRI of brain shows focal brain atrophy, cortical dysplasia, and hippocampal sclerosis, are found in a minority of patients with Dravet syndrome.[14] Whilst generalized cortical atrophy and ventricular dilatation, were observed in our patient.

Managing Dravet syndrome is complex and requires a multidisciplinary approach to enhance patient care and quality of life. Early diagnosis and genetic testing are crucial to avoid treatments that may worsen seizures. Treatments such as valproate and clobazam are often inadequate to control seizures.Precision medicine like stiripentol, cannabidiol, fenfuramine which is not available in our country and ketogenic diet have been proposed as potential options.[15] Despite treatment challenges, immunization should not be withheld in patients with Dravet syndrome, emphasizing the need for innovative treatments to address the unmet needs of this condition.

Conclusion

Dravet syndrome is a challenging rare, genetic epileptic encephalopathy, presents with focal prolong febrile seizure in infancy. Despite multiple Antiepileptic drugs, the seizures are intractable and causing neuro-developmental impairment. Early recognition and genetic testing for SCN1A mutations are important for optimizing outcomes.

Acknowledgements

We would like to acknowledge and thank to the all residents, doctors and faculties of Department of Paediatrics Neurology, IPNA, BMU.

Conflicts of interest: The authors have no conflict of interest to declare.

Ethical Approval: Informed consent was taken from the parents of the patient.

References

1. Hurst DL. Epidemiology of severe myoclonic epilepsy of infancy. Epilepsia. 1990 Jul-Aug;31(4):397-400. doi: 10.1111/j.1528-1157.1990.tb05494.x. PMID: 1695145.
2. Rosander C, Hallböök T. Dravet syndrome in Sweden: a population-based study. Dev Med Child Neurol. 2015 Jul;57(7):628-633. doi: 10.1111/dmcn.12709. Epub 2015 Mar 13. PMID: 25772213.
3. Wu YW, Sullivan J, McDaniel SS, Meisler MH, Walsh EM, Li SX, Kuzniewicz MW. Incidence of Dravet Syndrome in a US Population. Pediatrics. 2015 Nov;136(5):e1310-5. doi: 10.1542/peds.2015-1807. Epub 2015 Oct 5. PMID: 26438699; PMCID: PMC4621800.
4. Nakayama T, Ogiwara I, Ito K, Kaneda M, Mazaki E, Osaka H, Ohtani H, Inoue Y, Fujiwara T, Uematsu M, Haginoya K, Tsuchiya S, Yamakawa K. Deletions of SCN1A 5' genomic region with promoter activity in Dravet syndrome. Hum Mutat. 2010 Jul;31(7):820-9. doi: 10.1002/humu.21275. PMID: 20506560.
5. Mulley JC, Nelson P, Guerrero S, Dibbens L, Iona X, McMahon JM, Harkin L, Schouten J, Yu S, Berkovic SF, Scheffer IE. A new molecular mechanism for severe myoclonic epilepsy of infancy: exonic deletions in SCN1A. Neurology. 2006 Sep 26;67(6):1094-5. doi: 10.1212/01.wnl.0000237322.04338.2b. PMID: 17000989.
6. Wirrell EC, Hood V, Knupp KG, Meskis MA, Nabbout R, Scheffer IE, Wilmshurst J, Sullivan J. International consensus on diagnosis and management of Dravet syndrome. Epilepsia. 2022 Jul;63(7):1761-1777. doi: 10.1111/epi.17274. Epub 2022 May 12. PMID: 35490361; PMCID: PMC9543220.
7. Ding J, Li X, Tian H, Wang L, Guo B, Wang Y, Li W, Wang F, Sun T. *SCN1A* Mutation-Beyond Dravet Syndrome: A Systematic Review and Narrative Synthesis. Front Neurol. 2021 Dec 24;12:743726. doi: 10.3389/fneur.2021.743726. PMID: 35002916; PMCID: PMC8739186.
8. Dravet C. The core Dravet syndrome phenotype. Epilepsia. 2011 Apr;52 Suppl 2:3-9. doi: 10.1111/j.1528-1167.2011.02994.x. PMID: 21463272.
9. Wirrell EC, Laux L, Donner E, Jette N, Knupp K, Meskis MA, Miller I, Sullivan J, Welborn M, Berg AT. Optimizing the Diagnosis and Management of Dravet Syndrome: Recommendations From a North American Consensus Panel. Pediatr Neurol. 2017 Mar;68:18-34.e3. doi: 10.1016/j.pediatrneurol.2017.01.025. Epub 2017 Feb 4. PMID: 28284397.
10. Brunklaus A, Ellis R, Reavey E, Forbes GH, Zuberi SM. Prognostic, clinical and demographic features in SCN1A mutation-positive Dravet syndrome. Brain. 2012 Aug;135(Pt 8):2329-36. doi: 10.1093/brain/aws151. Epub 2012 Jun 19. PMID: 22719002.
11. Incorpora, G. Dravet syndrome. *Ital J Pediatr* 35, 27 (2009). https://doi.org/10.1186/1824-7288-35-27
12. Kapoor D, Anand A, Sharma S, Mukherjee SB, Marini C, Mei D, Chopra SS, Chettali AM. Dravet Syndrome: A Case Series. Indian J Pediatr. 2021 Jan;88(1):82. doi: 10.1007/s12098-020-03383-z. Epub 2020 Jun 26. PMID: 32591999.
13. Bureau M, Dalla Bernardina B. Electroencephalographic characteristics of Dravet syndrome. Epilepsia. 2011 Apr;52 Suppl 2:13-23. doi: 10.1111/j.1528-1167.2011.02996.x. PMID: 21463274
14. Guerrini R, Striano P, Catarino C, Sisodiya SM. Neuroimaging and neuropathology of Dravet syndrome. Epilepsia. 2011 Apr;52 Suppl 2:30-4. doi: 10.1111/j.1528-1167.2011.02998.x. PMID: 21463276.
15. Mahesan A, Kamila G, Gulati S. Advancements in Dravet Syndrome Therapeutics: A Comprehensive Look at Present and Future Treatment Horizons: A Focused Review. Ann Indian Acad Neurol. 2024 Jul 1;27(4):352-357. doi: 10.4103/aian.aian_49_24. Epub 2024 Aug 16. PMID: 39196806; PMCID: PMC11418756.