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Dravet Syndrome: A Case Report

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Abstract **Case Report**

Dravet syndrome is an epileptic encephalopathy caused by mutations and deletions in the SCN1a gene in chromosome 2q. Child usually infants present with recurrent febrile seizures later may be associated with afebrile seizures. Initially developmental history normal but history of regression of milestones is present. Early recognition and diagnosis by genetic work up and timely treatment with appropriate anti convulsant therapy may improve neurodevelopmental outcome and reduces seizure frequency.

Keywords: Dravet Syndrome, convulsant therapy, neurodevelopmental outcome.

INTRODUCTION

Dravet Syndrome (DS) was first described as severe myoclonic epilepsy of infancy (SMEI) by Charlotte Dravet in 1978 and was later renamed Dravet Syndrome in 1989. It is rare form of early onset genetic epilepsy syndrome that manifests as intractable epilepsy and neurodevelopmental delays. It affects an estimated of 1 in 15700- 40000 live births, accounted for 3% of cases of epilepsy among children presenting with a seizure within first year of life [1].

CASE PRESENTATION

8 month old girl child presented to paediatrics emergency at Rajindra Hospital Patiala with history of abnormal body movements in form of tonic clonic movements of both upper limbs and lower limbs associated with uprolling of eyeballs with clenching of teeth. There was no significant family history. Antiepileptic phenytoin loading dose given and put on maintenance dose, child again had repeated seizure episodes another anti epileptic's valparin and levitiracetam added accordingly. All routines investigations including EEG, MRI Brain, lumbar puncture and metabolic work up done which shows normal results. Developmental milestones were achieved normally upto 1 year and delayed after that.

Then child had episodes of high grade fever associated with tonic clonic seizures multiple times. Antiepiletics topiramate and clobazam added. As child didn't respond to above said treatment modalities keeping in mind possibility of multi drug resistant genetic disorder we have gone for gene analysis, in which SCN1a came out to be positive.

RESULTS

PATHOGENIC VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS DETECTED

Gene# (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification
SCN1A (-) (ENST00000674923.1)	Exon 14	c.1930del **(p.Thr644LeufsTer28)**	Heterozygous	Generalized epilepsy with febrile seizures plus, type 2 or familial febrile seizures-3A, Dravet syndrome	Autosomal dominant	Pathogenic

DISCUSSION

It is an epileptic encephalopathy, which is clinically characterized by pleomorphic seizures starting as early as infancy, and exhibit neurodevelopmental delay, cognitive and motor impairment [1]. The aetiology of Dravet syndrome is genetic. Mutations and deletions in the SCN1A gene on chromosome 2q are found in 70% to 80% of patients with Dravet syndrome. Of the gene abnormalities, 85% are de novo mutations. 7-9 Familial SCN1A mutations occur in 5% to 10% of patients, and a family history of febrile seizures and epilepsy has been reported to be between 25% and 75% in some studies [3].

Like other epileptiform encephalopathies, DS patients are also multi drug resistant. The first line of drug is valproate, topiramate and stiripentol. Other efficacious therapies are cannabinoids and a ketogenic diet. In patients refractory to Antiepileptics and ketogenic diet, surgical therapies of deep brain stimulation or vagal nerve stimulation have been used. Overall, DS has a poor prognosis and need multidisciplinary healthcare management [1].

CONCLUSION

DS is a severe epileptic encephalopathy that is difficult to recognize at time of onset. Early recognition and diagnosis of DS and management with appropriate anticonvulsants and treatment plan may reduce seizure burden and improve long term developmental outcome.

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