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Case Study

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DRAVET SYNDROME: A PAEDIATRIC CASE SCENARIO IN A TERTIARY CARE HOSPITAL IN SOUTHERN INDIA

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ABSTRACT

Dravet syndrome (DS) is an epileptic encephalopathy that begins with prolonged seizures in infancy, often accompanied by a fever, and is initially classified as a febrile seizure. In 80 to 90 percent of cases, DS is linked to a mutation in the SCN1A gene. Early identification of Dravet syndrome in a child who presents with febrile seizures is critical, as it allows appropriate care to be instituted. This case study presents Dravet Syndrome of a 7 year old female child who is presented with the chief complaints of fever and cough for the past two days, abdominal distension, and multiple episodes of abnormal body movements for one week. Based on the subjective evidence and patient's history,she had dravet syndrome with acute respiratory tract infection. The treatment given to the child include ampicillin, paracetamol, valproic acid, clobazam, phenobarbitone and salbutamol. The child was discharged after complete resolution of symptoms. The patient's condition had improved and there were no new concerns when the review was done two weeks later. This case report emphasizes early diagnosis and treatment of DS and the importance of giving proper treatment in improving the clinical outcome and in preventing serious complications.

KEYWORDS: Dravet syndrome, Pediatrics, Epilepsy, SCN1A gene, Febrile convulsion.

INTRODUCTION

Dravets Syndrome(DS), initially known as severe myoclonic epilepsy of infants, is a rare and severe form of epilepsy with frequent prolonged seizures that is triggered by an increase in temperature,delay in development, impairment in speech, sleep disturbances and some other health problems.In most cases of DS, there is a mutation within the SCN1A gene.[1,2]

International League Against Epilepsy (ILAE) has classified DS as an epileptic syndrome.[3] It is characterised by hemiclonic or generalized clonic seizures during the first year of life that is often triggered by fever in the child.It then progress to other forms like myoclonic,absence, focal and generalised tonic-clonic seizures within the second year. Even Though development is normal during the first years of life, these children suffer from intellectual impairment. Also there is an increase in mortality rate.[4] However, early identification and treatment of these individuals can result in improvement in the cognitive, behavioral and motor impairments.

This case report emphasizes on the early diagnosis and treatment of DS using suitable antiepileptics to reduce the seizure burden thereby improving the quality of life of patients.

CASE REPORT

A 7 year old female child was admitted in the hospital with chief complaints of fever and cough for the past two days, abdominal distension, and multiple episodes of abnormal body movements for one week. The child was a known case of seizure disorder and was on treatment with multiple anticonvulsants. First episodes of seizure occurred at the age of 5 months following DPT second dose immunization. At seventh month, the child had recurrent episodes of seizure and was admitted in ICU for 5 days. The child was also having a history of recurrent lower respiratory tract infection and was on nebulization. Regarding the present condition, the child was a known case of seizure disorder on treatment and is presented with multiple episodes of abnormal body movement. The movements occured are tonic clonic in nature, lasting more than 20 minutes and was associated with the loss of consciousness, involuntary micturition and post ictal phase. There has been an increased frequency for the past one week. The child was also presented with abdominal distension and had fever with cough for the past 2 days with no chills and rigor.

The child is 115 cm in height and weighs 22 kilogram. On examining the vitals it was found that the child was active and alert with the pulse rate of 112/min, respiratory rate of 22/min, blood pressure of 110/60 mmHg and afebrile temperature. The child was having mild parlour with no icterus, clubbing and cyanosis. From head to foot examination it was found that head, hair and hairline, face, eyes, ears, oral cavity and chest were found to be normal. From the systemic examination, S1 and S2 were normal and no murmurs were present regarding the cardiovascular system. Regarding CNS, the child was conscious and oriented. Regarding GIT, the abdomen was soft with nontender and positive vowel sounds were found. Regarding the respiratory system, bilaterally equal air entry was found.

Lab investigations

Lab parameters	Normal value	Observed value
RBC	3.38-5.08 million/mm^3	4.1
Hemoglobin	11-16 gm%	13.2
PLT	1.5-3.8 L/mm^3	3.2
WBC	4.5-11 cells/mm^3	8.7
Neutrophils	50-70 %	62
Eoisinophills	0.8-4 %	**6**
Lymphocytes	20-44 %	28
Basophills	0.3-1.8 %	1.2
Monocytes	4.1-9 %	**10.2**
ESR	0-10 mm/hr	4
HCT	34.5-46.3 %	41.2
MCV	18.4-95.9 fL	72
MCH	27.2-33.5 pg	32.4
MCHC	32.5-35.2 g/dl	34

LFT and RFT

Lab parameters	Normal value	Observed value
Total bilirubin	0.2-1mg%	0.6
Direct bilirubin	0-0.25 mg%	0.22
Serum cholesterol	<170 mg/dl	**210**
SGOT	5-42 IU /L	88
SGPT	5-38 IU/L	22
Serum albumin	3-5 gm%	4.1
ALP	20-112 IU/L	**220**
Serum globulin	2.5-3.5 gm%	
Serum creatinine	0.6-1.4 IU/L	0.9
Blood urea	7-20 mg/dl	12

From the urine routine examination it was found that albumin and sugar was absent. Microscope shows the pus cells (0-1/hpf), epithelial cells (0-1/hpf), and RBCs, Casts, crystals and bacteria were absent.

Diagnosis

Based on the subjective evidence of abnormal body movements and from the history of the patient, the patient is having dravet syndrome with acute respiratory tract infection.

Treatment

Here the child is presented with seizure disorder associated with dravet syndrome. On the day of admission, Inj Ampicillin 500mg I IV was given Q6H, Syp Calpol 250 was given 6ml for Q8H, Syp Encorate (7.5ml 1-0-1), Tab Frisium 5 (1-1-1), Syp Gardenal (2.5ml BD) and Syp Asthalin (7.5ml 1-1-1) were given, as the child is having cough. On the second day of admission, all the medicines were continued and Asthalin was nebulised (6ml 1-1-1). On the third day of admission all medicines were continued. On the fourth day of admission, in addition to the ongoing drugs Syp Mox (5ml 1-1-1) was also given. The child was discharged with the following medications, Syp Moxclav (5ml 1-1-1) for 5 days, Syp Encorate (7.5ml 1-0-1), Tab Frisium 5mg (0-1-0), Syp Gardenal 2.5ml (1-0-1) for 2 weeks and Syp Asthalin 5ml to be continued. The patient was discharged in a stable condition. The review was made after two weeks and the condition of the patient got improved and had no fresh complaints.

DISCUSSION

The incidence of DS is about 1 in every 40000 infants in the US. The typical clinical presentation for DS includes seizures, which is often triggered by hyperthermia that lasts more than 10 minutes, developmental delay, ataxia, speech impairment and sleep disturbances. In this case, the child was presented with these symptoms like seizure that is tonic-clonic in nature, lasting more than 20 minutes with loss of consciousness. The seizure was triggered by fever. The child was also presented with developmental delay, involuntary micturition and had a postictal phase.

The first line agents used for treating DS are valproic acid and clobazam and their use is supported by expert opinion. Other drugs used include topiramate, levetiracetam, zonisamide, bromides, and ethosuximide. Ketogenic diet, and vagus nerve stimulation are some non pharmacological measures that can be adopted.However,there are some newer drugs like stiripentol, fenfluramine and cannabidiol that are found effective in DS.

It is of equal importance that sodium channel blocking agents like carbamazepine, oxcarbazepine, and phenytoin along with lamotrigine has to be avoided as it has the potential to exacerbate and worsen the condition of the patient with DS. Early detection of DS and its management with suitable antiepileptic medications can help patient in preventing seizure episodes and to improve the quality of life.

CONCLUSION

DS is a rare kind of genetic neurological disorder that has no permanent cure. This case report mainly focussed on early detection and diagnosis of DS along with treatment using suitable antiepileptic agents. The child's parent was given counselling about the importance of administering the drugs, especially the antiepileptics

being given. Parents were also instructed to keep the child safe from seizure triggers like hyperthermia. However the patient was reviewed after 2 weeks with an improvement in his condition.

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CONFLICT OF INTEREST
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ABBREVIATIONS
SCN1A - Sodium Voltage-Gated Channel Alpha Subunit 1.
DPT - Diphtheria Pertussis Tetanus.
WBC - White Blood Cell.
RBC - Red Blood Cell.
PLT – Platelet.
ESR - Erythrocyte Sedimentation Rate.
MCV - Mean Corpuscular Volume.
MCH - Mean Corpuscular Haemoglobin.
MCHC - Mean Corpuscular Haemoglobin Concentration.
HCT – Hematocrit.
SGOT - Serum Glutamic-Oxaloacetic Transaminase.
SGPT - Serum Glutamic-Pyruvic Transaminase.
ALP - Alkaline Phosphatase.

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