Doose Syndrome

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# Doose Syndrome

Doose syndrome also known as myoclonic-astatic epilepsy is a rare seizure disorder that occurs during early childhood. In 1970, It was first described by Dr. Hermann Doose. Doose syndrome is a type of generalized epilepsy and seizures that is not managed easily with medication. When children reach adulthood, the symptoms may improve and treatment for this might not be needed anymore. Since 1970, the genetic background and knowledge of Doose syndrome have continued to grow. Much research has been conducted in which the efficacy of different treatment for Doose syndrome is seen. Doose syndrome is a most common disease condition with an incidence of 1 in 10000 children and constitutes about 2.1% childhood-onset epilepsy. Doose syndrome is more common in males1. In almost 95% of the cases, disease onset occurs within the first five years of age but 25% of children suffer first seizure in their first year of life. In some children, a further seizure cannot occur for short period that can delay the myoclonic-astatic epilepsy diagnosis. Doose syndrome is connected with different types of seizures. Myoclonic seizures are associated with quick jerking movement which occurs axially or truncally2.

In Doose syndrome, genetics plays a very important role and becomes an additional technique to differentiate it from other syndromes. The first person to draw attention to the increase incidence of seizures was Doose. Findings of ECG among the affected individual family members also shows the same effect. It is reported that the abnormal findings of ECG prevalence was approximately 68.1% among the immediate family members and 81% among the distant relatives. The previous finding has reported that seizures occur in 39% of the relatives of the individuals with this syndrome2. The main findings of ECG in family members are unusual ‘theta background rhythm’ and ‘photosensitivity’. The multifactorial inheritance is more common in this disorder. This is shown by the detail that Doose syndrome has many different kinds of seizure manifestation. While many of the children do not have recognized gene mutation, in some cases mutation in several genes including *SCN2A*, *SLC2A, SCN1A, SCN1B, KIAA2022, and SYNGAP1* have been determined in many cases.Individuals with this disorder were diagnosed first with the mutation in sodium channel neuronal type 1 alpha subunit with febrile seizure and general epilepsy disorder. Mutation in gamma-aminobutyric acid and sodium channel subunit 1 have also been found in individuals with Doose syndrome.

 It is reported that Doose syndrome is difficult to treat. Less traditional therapies and anticonvulsant medications have been reported in different studies for Doose syndrome treatment. One of the initial therapies for Doose syndrome was corticosteroid and a very high dose of dexamethasone. It was first stated by Doose in his study, the use of high doses of 80 IU adrenocorticotrophic hormone and 1mg/kg dexamethasone for the treatment of seizures3. The main adverse effect of the use of steroids is a recurrence of seizures after discontinuation of treatment2. Ethosuximide is found to be the most effective anticonvulsant for the treatment of absence seizures. Lamotrigine and Valproic acid have also found beneficial in Doose syndrome treatment. However, lamotrigine should not be used in patients with myoclonic seizures. Zonisamide and Levetiracetam have been used anecdotally for Doose syndrome. Evidence on the use of anticonvulsants, such as lacosamide, ralfinamide and clobazam in Doose syndrome is not readily available in the literature3. It is determined that phenytoin, carbamazepine, and vigabatrin can exacerbate seizures in Doose syndrome. Most of the authors have stated that anticonvulsants should be used with caution in patients with Doose syndrome. The ketogenic diet has also been reported as most efficacious therapy for the management of Doose syndrome

**Reference List/Endnotes**

1. Kelley SA, Kossoff EH. Doose syndrome (myoclonic–astatic epilepsy): 40 years of progress. *Dev Med Child Neurol*. 2010;52(11):988-993.

2. Pittau F, Korff CM, Nordli Jr DR. Epileptic spasms in epilepsy with myoclonic‐atonic seizures (Doose syndrome). *Epileptic Disord*. 2016;18(3):289-296.

3. Dragoumi P, Chivers F, Brady M, et al. Epilepsy with myoclonic–atonic seizures (Doose syndrome): When video-EEG polygraphy holds the key to syndrome diagnosis. *Epilepsy Behav Case Rep*. 2016;5:31-33.