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Perspective

Clinical Findings and Epidemiology of Dravet Syndrome in Children

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DESCRIPTION

A rare form of epilepsy that manifests in infancy is known as Dravet syndrome, often referred to as Severe Myoclonic Epilepsy of Infancy (SMEI). It is a chronic, crippling condition that can significantly lower the patient's quality of life. Patients have delayed growth, frequent seizures, and poor seizure management. When your child has a fever, initial seizures are frequently extended events. Later on, in the second year of life, different seizure types (frequently without a temperature) start to appear. Dravet syndrome develops at random; while being a genetic condition, the mutations are frequently fresh alterations that only impact the affected child. In other words, no additional family members have the gene mutation. Up to 80% of people with Dravet syndrome will also test positively for a SCN1A gene mutation, aiding in the confirmation of the clinical diagnosis. It's crucial to understand that a diagnosis is not automatically ruled out in the absence of a SCN1A mutation. Most nations now provide commercial genetic testing. If a genetic test is successful, the parents may also be tested to determine inheritance. SCN1A mutations, specifically loss-of-function mutations in one copy of the SCN1A gene, are the most common cause of Dravet syndrome in those who have been given the diagnosis. These people produce fewer sodium channels, which are frequently seen on Gamma-Amino Butyric Acid-GABAergic neurons in the brain, as a result of having only one functional copy of the SCN1A gene. This results in a reduction in inhibitory action. Seizures and other Dravet syndrome symptoms occur from the breakdown of the excitatory-inhibitory balance in the brain.

The incidence in the general population is 1/40000, 1/20000, or 30000. Males are harmed more commonly than females (2:1). The incidence of epilepsy among patients ranges from 3 to 5 percent in the first year of life and reaches 7 percent by the age of three. In a child who is otherwise healthy, tonic-clonic seizures almost invariably start in the first year of life, or more frequently in the first six months. Because of this, the syndrome's beginning may be difficult to tell apart from febrile seizures.

Children with Dravet syndrome always have cognitive impairment. Patients are fully normal throughout the first year of birth, but by the second year, it is clear that psychomotor development has slowed or stopped. During this time, behavioral abnormalities that more frequently show up as hyperactivity and less frequently as autistic symptoms were also observed. Nearly all children begin to show severe mental impairment around the fifth year, which affects every functional area. Sometimes, cognitive function increases noticeably at the same time that the frequency of epileptic seizures declines. The motor and visuomotor functions are better preserved in the early stages of the illness than the progressively deteriorating language abilities.

An Electroencephalogram (EEG) is typically normal at the start of the symptoms. When febrile and afebrile seizures reoccur, abnormalities such as generalized, localized, or multifocal anomalies-such as multifocal spikes, spike and waves, polyspike and waves discharges, and a slowdown of background activity become apparent. Even before a year, the intermittent light stimulation can be beneficial. There is a photosensitivity spectrum in SMEI patients because this response can remain and in some cases can be exploited as self-stimulation. Photosensitivity is not a permanent trait throughout the course of the disease. The clinical diagnosis of Dravet syndrome is still mostly dependent on the patient's seizure history, clinical features, neurologic examination, EEG pattern, and extended observation. Therefore, DNA analysis combined with an assessment of the SCN1A gene can typically confirm the diagnosis. Some medications, including lamotrigin, carbamazepine, phenobarbital, and phenytoin, may make seizures worse. The three medications with some effect that are most usually used are valproate, clobazam, and topiramate. The drug stiripentol (unapproved in the US) is seen as helpful. Although other treatments like the ketogenic diet have been explored, the results remain debatable.

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