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SCN1-1 RELEATED DISORDERS: DRAVET SYNDROME

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Annotation

SCN1A-Related Disorders - the most common epilepsies exacerbated by fever are caused by pathogenic variants in the SCN1A gene, which lead to various syndromes, including Dravet syndrome (DS) and genetic epilepsy with febrile seizures plus (GEFS+). DS in particular is highly associated with loss-of-function SCN1A mutations; GEFS+ is more heterogeneous and is associated with multiple other genetic variants as well as some SCN1A gain-of-function mutations.

Key words:

Dravet Syndrome: DS is suspected in children with complex febrile seizures evolving into difficult-to-control epilepsies, exacerbated by antiseizure medications with sodium blocking mechanism of action (carbamazepine, oxcarbazepine, phenytoin, or lamotrigine). The first seizures are frequently febrile and prolonged, and can be generalized or focal clonic in type. Febrile and nonfebrile seizures recur, sometimes as status epilepticus. Generalized myoclonic or atonic seizures appear after 1 year of age. Complex partial seizures with secondary generalization may also occur. Coincident with the onset of myoclonic/atonic seizures are the slowing of development and the gradual appearance of ataxia and hyperreflexia.

Diagnosis. The diagnosis is based on the phenotype, but genetic testing is increasingly important. Up to 80% of patients have a mutation on SCN1A with a negative effect on sodium channel function. Genetic testing (positive in 80% of cases) is helpful and may prevent further unnecessary EEGs or imaging studies.

Management. Seizures in Dravet syndrome are difficult to control but can be reduced by anticonvulsant drugs. Some anticonvulsant medications (such as oxcarbazepine, carbamazepine, phenytoin, and lamotrigine) should not be used on a daily basis as they may make seizures worse. A ketogenic diet, high in fats and low in carbohydrates, also may be helpful. Avoid sodium channel drugs: medications such as levetiracetam, 37 divalproex sodium, topiramate, zonisamide, rufinamide, and

management with a ketogenic diet are good options. Epidiolex (cannabinoid) is a US Food and Drug Administration (FDA)– approved drug for the treatment of DS. Fintepla (fenfluramine) was recently approved for use, and clinical trials show such significant efficacy in DS that we recommend clinicians consider initiating it as soon as the diagnosis is confirmed. Patients should have a soft helmet to prevent head injury during seizure-related falls. Encourage parents to take a class in cardiopulmonary resuscitation (CPR). A written emergency room protocol that families carry with them is often helpful. Prenatal counseling and genetic testing of the patient's siblings should be considered.

In conclusion, Dravet Syndrome is an epilepsy syndrome that begins in infancy or early childhood and can include a spectrum of symptoms ranging from mild to severe. Children with the disorder typically have normal development in the first few years of life. As seizures increase, the pace of acquiring skills slows and children start to lag in development behind their peers. Other symptoms can begin throughout childhood with changes in eating, appetite, balance, and a crouched gait (walking).

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