Dravet Syndrome

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Dravet Syndrome Information Page.

Dravet syndrome, previously called severe myoclonic epilepsy of infancy (SMEI), 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 Dravet syndrome initially show focal (confined to one area) or generalized (throughout the brain) convulsive seizures that start before 15 months of age (often before age one). These initial seizures are often prolonged and involve half of the body, with subsequent seizures that may switch to the other side of the body. These initial seizures are frequently provoked by seizures or exposure to increased temperatures or temperature changes, such as getting out of a bath. Other seizure types emerge after 12 months of age and can be quite varied. Status epilepticus – a state of continuous seizure requiring emergency medical care – may occur frequently in these children, particularly in the first five years of life.

Children with Dravet syndrome typically have normal development in the first fews 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, appetitie, balance, and a crouched gait (walking).

In at least 80 percent of cases, Dravet syndrome is caused by defects in a gene required for the proper function of brain cells. Mutations in the SCN1A gene (a gene that encodes as a sodium channel, a part of the cell membrane involved in nervous system function) are the primary causes of Dravet syndrome. Borderline SMEI (SMEB) and another type of infant-onset epilepsy called generalized epilepsy with febrile seizures plus (GEFS+) but which is much less severe, are caused by defects in the same gene. Dravet syndrome is a lifelong condition.