



NAVIGATING LIFE WITH DRAVET SYNDROME

Information and support for parents and caregivers

NAVIGATING LIFE WITH DRAVET SYNDROME

Information and support for parents and caregivers

Contributors:

Dr. Martina E. Bebin

Dr. Robert Flamini

Dr. Kelly Knupp

Dr. Linda Laux

Dr. Scott Perry

Dr. Joseph Sullivan

Dr. James Wheless

Dr. Elaine Wirrell

Dravet Syndrome Foundation
Executive Team

TABLE OF CONTENTS

INTRODUCTION	6
---------------------	----------

QUESTIONS	8
------------------	----------

Overview of Dravet syndrome	8
------------------------------------	----------

1. What is Dravet syndrome?	8
2. What is the cause of Dravet syndrome?	10
3. How is Dravet syndrome diagnosed?	10
4. Is Dravet syndrome inherited? Are my other children at risk?	11
5. What type of doctors and specialists might my child need to see regularly?	13
6. Can childhood vaccines cause Dravet syndrome? Should I vaccinate my child?	14

Seizures and treatments	15
--------------------------------	-----------

7. What types of seizures are usually seen in Dravet syndrome?	15
8. What is SUDEP?	16
9. Besides seizures, what are the other medical issues related to Dravet syndrome?	18
10. What medications are used to treat seizures in Dravet syndrome?	20
11. What are other medication considerations?	21
12. Are there credible non-medication treatments for controlling seizures?	22
13. In addition to medication, how can I help reduce the risk of seizures in my child?	23

Caring for people with Dravet syndrome	24
---	-----------

14. What should I do when my child has a seizure?	24
15. What are trigger factors for seizures?	25
16. What daily life difficulties might my child face?	26

Family life	27
--------------------	-----------

17. How will our family's daily life change?	27
18. How do I explain Dravet syndrome to my other children?	28
19. How do I explain Dravet syndrome to my family and friends?	29

Dravet syndrome in childhood	31
-------------------------------------	-----------

20. What is the course of Dravet syndrome in childhood?	31
21. Can my child attend school?	33

Dravet syndrome in puberty and adulthood	35
---	-----------

22. What services might my child need as a teen?	35
23. What services might my child need as an adult?	37

GLOSSARY	38
-----------------	-----------

RESOURCES	42
------------------	-----------

NOTES	43
--------------	-----------

INTRODUCTION

Biocodex, in partnership with leading Dravet syndrome doctors and the Dravet Syndrome Foundation, created this booklet to be a useful guide for families, relatives, and the communities surrounding patients with Dravet syndrome. This guide provides an overview of Dravet syndrome, capturing the daily challenges that may arise at different stages of a patient's life.

We realize that it is very easy to get lost in all of the information shared across different platforms these days, and we hope that this guide provides clear and concise information you may need throughout your journey in understanding Dravet syndrome.

This booklet is designed to answer questions that you may have after your child's diagnosis and later down the road. You will also find useful information that you can pass on to your family members and/or community.

We know that this booklet will not answer all of your questions, but we hope that you find it to be a good starting point. For additional questions, your child's doctor remains your best contact. We have included a section for notes in the back of the book, where you can write any additional questions you may have and then bring them to your doctor. We have also included a glossary of medical terms you will encounter as you research Dravet syndrome in this guide and elsewhere.

The information contained in this booklet is not a substitute for professional medical advice, nor is it intended to be used for medical diagnosis or treatment. Talk with your healthcare provider about any questions or concerns you may have.

OVERVIEW OF DRAVET SYNDROME

1. What is Dravet syndrome?

Dravet syndrome is a rare type of epilepsy that is most often caused by a **pathogenic variant** or pathogenic mutation (you can think of it as a genetic typo) most commonly seen in the *SCN1A* gene.

There are many different types of epilepsy. They are often diagnosed by what the seizures look like. Dravet syndrome usually appears in the first year of life, with long seizures often triggered by changes in temperature. Some examples of temperature changes that can trigger seizures are fever (related to illness or immunizations), getting in and out of the bath, being outside on a warm afternoon, or sleeping in a hot room. The seizures in the first year of life may often (but not always) be **hemiclonic**, which is a type of seizure where one side of the body shakes while the other side does not. These seizures tend to be very long and may require treatment in an emergency department or intensive care unit.

Seizures tend to change over the patient's lifespan. In the second year of life, seizures will often change from long seizures into several different types (see Question 7). These seizures do not always respond well to treatment.

During school-aged years, seizures begin to occur more frequently during sleep than while awake. These seizures are usually much shorter in duration but may happen in clusters.

During adolescence and adulthood, while seizures may decrease in number, they still often occur in clusters. With age, triggers tend to provoke seizures less.

There are other symptoms that are often seen in Dravet syndrome. Caregivers will often notice a slowing of development in their child, usually starting at preschool age. At this age, children often have difficulty focusing on one task for long periods of time. Their pain tolerance is usually very high, which can lead to injuries and risky situations.

Starting at 1 year old, children may have problems with balance. Motor control issues and problems walking are common after age 5. As children enter into adolescence, they may develop a **crouched gait**, meaning that there is too much bend in the hips and knees.

2. What is the cause of Dravet syndrome?

Dravet syndrome is most often caused by a **pathogenic variant** in a gene called *SCN1A*. However, further genetic testing may show that other genes can be associated with Dravet syndrome. Genes provide the code that tells our cells and body how to work and grow. The *SCN1A* gene codes for a sodium channel called NaV1.1, which is thought to be involved in telling our cells when to “quiet down.” When cells are not able to be calmed down, it can lead to overexcitement of the brain, which presents as a seizure.

This sodium channel is present in the brain as well as other parts of the body, which may be the cause of some of the other symptoms that are seen with this syndrome, like gait problems and constipation. Scientists are still working to fully understand how the *SCN1A* gene works.

3. How is Dravet syndrome diagnosed?

A healthcare provider (usually a neurologist or geneticist) may recommend running a gene test, which requires a blood test or mouth swab. They will look at the child’s history and any genetic test results to determine if they have Dravet syndrome.

Diagnosing Dravet syndrome can be complicated, because some people will have a mutation in the gene and not have seizures, and other people may have seizures that look like Dravet syndrome but not have a mutation in the *SCN1A* gene. In early genetic testing, it may be difficult to know if a child will have Dravet syndrome until other seizure types emerge. At this point, some healthcare providers may call this “presumptive Dravet syndrome.”

4. Is Dravet syndrome inherited? Are my other children at risk?

First, you should not feel guilty or responsible for your child’s disease. No one is to blame for it. In most cases, variants of the *SCN1A* gene occur “**de novo**,” meaning that they are “new” mutations—neither parent carries the mutated gene. These mutations occur spontaneously before conception or just after the time of conception.

Boys and girls are equally affected. If you have a child with Dravet syndrome, it doesn’t mean your other children will also have this disease. Your older children run no risk of developing Dravet syndrome.

Although most cases of Dravet syndrome are the result of **de novo** (new) mutations, around 5%-10% of cases are inherited by children from their parents. In such cases, the disease follows an **autosomal dominant inheritance pattern**. That means that one of the parents will test positive for the same *SCN1A* gene mutation as their child.

Based on medical science's current understanding, if your child carries a **de novo mutation** in the *SCN1A* gene, your risk of having another child born with Dravet syndrome may be slightly greater than that of the general population (ie, 1 in 15,700 births). This small increase in risk may be caused by a parent carrying the *SCN1A* gene mutation. Sometimes, people can carry the gene mutation even if they test negative for it, either because they have a very small number of affected cells, or because the mutation is in non-blood cells. There are new tests being developed that may more accurately determine your risk of having another child with Dravet syndrome.

Having an *SCN1A* mutation does not always mean that someone will have Dravet syndrome. *SCN1A* mutations can also be found in patients with other types of epilepsy, such as genetic epilepsy with febrile seizures plus (GEFS+), a condition that causes varying types of seizures and syndromes across family members.

5. What type of doctors and specialists might my child need to see regularly?

It is always helpful to have a strong care team to help support your child with their medical and developmental needs. Your primary source of care and information will likely be your child's neurologist.

In addition to your neurologist, a geneticist may also be part of the team that can help you understand Dravet syndrome. The geneticist can explain more about the test results and what they might mean for your child. If you still have questions about future children, do not hesitate to ask your geneticist questions regarding future pregnancies and risk factors for other members of your family.

Depending on any other disorders your child may have, they may benefit from seeing a:

- psychiatrist
- orthopedist
- endocrinologist
- nutritionist
- psychologist/psychiatrist
- physical, occupational, and/or speech therapist

6. Can childhood vaccines cause Dravet syndrome? Should I vaccinate my child?

Vaccinations do not cause Dravet syndrome. As previously discussed, the usual cause of Dravet syndrome is a **de novo mutation** of the *SCN1A* gene.

In the past, it was believed that vaccination was linked to Dravet syndrome since the first seizure often happened around the time of the first vaccine. However, while vaccination may trigger a seizure, it is not a cause of the underlying condition. The exact mechanisms that link vaccination to seizures are not fully understood.

Vaccination-induced seizures do not change the course of Dravet syndrome at all, either in terms of seizure severity, developmental concerns, or intellectual ability.

Before your child receives a vaccination, discuss ways to manage the risk of seizures with your child's doctor.

SEIZURES AND TREATMENTS

7. What types of seizures are usually seen in Dravet syndrome?

Infants with Dravet syndrome most commonly present with seizures that involve rhythmic shaking of one side (**hemiclonic seizure**) or both sides of their body (**generalized tonic-clonic seizure**). **Hemiclonic seizures** typically switch sides from one to the next, affecting the right side with one seizure and the left side with the next. In infancy, seizures tend to be prolonged (often longer than 10 minutes) and are often triggered by fever, illness, or immunization.

Later, between 1.5 and 5 years of age, additional types of seizures may occur. These include:

- **Myoclonic seizures:** brief, isolated jerks of one or more extremities, lasting 1 or 2 jerks
- **Focal impaired awareness seizures:** characterized by unresponsive staring, at times with fiddling with

the hands or chewing movements of the mouth. During the event, the child is poorly responsive, and after the event, they will typically sleep

- **Atypical absence seizures:** characterized by brief periods of staring and decreased responsiveness without sleepiness

Sometimes, children can have clusters of myoclonic, focal impaired awareness, or atypical absence seizures that occur so frequently that the child cannot recover between them and may appear foggy or confused. If this state lasts longer than 30 minutes, it is called **non-convulsive status epilepticus** or **obtundation status**.

Tonic seizures are seizures where the whole body becomes stiff. **Tonic-clonic seizures** have a tonic phase followed by whole-body rhythmic shaking activity (clonic). They occur mainly in clusters during sleep as children get older, around 4-5 years old. In adulthood, these seizures often become more evident.

8. What is SUDEP?

Sudden unexpected death in epilepsy, or SUDEP, refers to the sudden and unexpected death of someone with epilepsy who was previously healthy. In cases of **SUDEP**, which may occur in up to 1% of

people with Dravet syndrome each year, the patient is often found face down and unresponsive in bed. There may or may not be evidence that the patient suffered a recent seizure—no specific cause of death is found when an autopsy is done. The exact cause of **SUDEP** is not known, but it has been proposed that seizures may induce an irregular heart rhythm or breathing difficulties.

SUDEP is most common in people with poorly controlled seizures, especially when **generalized tonic-clonic seizures** are occurring more than once a month. This is, unfortunately, the case for many people with Dravet syndrome. Although it is not possible to prevent **SUDEP**, the risk can be reduced by improving seizure control and decreasing the frequency of **generalized tonic-clonic seizures**, particularly at night. This is one of the most important treatment goals for you and your child's healthcare team to work toward.

Several seizure alarms are now available on the market to alert families and caregivers if the person is having a seizure. These alarms are not foolproof as they can miss some seizures and there can be false alarms. You can find more information on these alerts at Epilepsy.com.

9. Besides seizures, what are the other medical issues related to Dravet syndrome?

Seizures are just one part of Dravet syndrome. People with Dravet syndrome have other symptoms that can have an impact on their quality of life. These symptoms include:

Intellectual disabilities and learning problems

When seizures first start happening, children usually have normal development. However, their developmental progression slows over time, which may be noticeable from 1 to 5 years old. Rarely, a child can lose skills after a prolonged episode of **status epilepticus**. Language development is often delayed. Most children develop intellectual disabilities that range from mild to severe. It is recommended that children with Dravet syndrome have a formal assessment with a neuropsychologist or developmental pediatrician before starting school, to ensure that the support they will need will be in place.

Behavior and attention disorders

Many patients develop behavior disorders, and some have attention issues and hyperactivity. Medications for behavior and attention problems may be helpful in certain cases and generally do not worsen seizures.

Walking and coordination difficulties

Over time, most people with Dravet syndrome develop a **crouched gait**, typically by late childhood to adolescence. Poor coordination is also a concern. Assessment by physical and occupational therapy may be helpful.

Sleep disorders

Many people with Dravet syndrome have sleep problems, which can include difficulties falling asleep and staying asleep. This can also lead to excessive fatigue during the day.

It is important to encourage regular sleep habits. Have a regular bedtime routine and avoid exciting activity around bedtime. While some children may need short naps during the day, it is important to avoid frequent long naps, as this may interfere with nighttime sleep.

Growth and appetite

Many children with Dravet syndrome have poor appetites compared to their siblings. Some of this may be medication-related, so it is important to discuss these concerns with your healthcare provider. Studies have shown that children with Dravet syndrome have slower growth.

Autonomic dysfunction

Many people with Dravet syndrome will sweat abnormally, or have purplish or bluish discoloration of their hands or feet when in cooler environments. Generally, no treatment is needed for such symptoms.

10. What medications are used to treat seizures in Dravet syndrome?

There is currently no cure for Dravet syndrome. Treatments for the seizures associated with Dravet syndrome are called anti-seizure drugs, or ASDs (also called antiepileptic drugs, or AEDs). Your child may be prescribed a combination of ASDs. It often takes time to find the best combination, and it may change over time as your child's seizure types change. Any treatment changes should be made by a healthcare provider.

Some anti-seizure medications, called sodium channel blockers, may make seizures worse for patients with Dravet syndrome. Some examples of these medications are lamotrigine, carbamazepine, and oxcarbazepine.

A seizure response plan may include an on-hand "rescue" therapy. Rescue medicines are fast-acting

anti-seizure drugs that help to stop a seizure or seizure cluster quickly before it progresses to a medical emergency, such as **status epilepticus**. Nasal formulations have recently become available, in addition to a rectal gel formulation already on the market. When on-hand rescue therapy does not control a seizure, caregivers should contact local emergency services for assistance by calling 911.

11. What are other medication considerations?

Some types of anti-seizure therapy have been shown to worsen seizures in Dravet syndrome. These include Dilantin® (phenytoin), Cerebyx® (fosphenytoin), Tegretol® (carbamazepine), Trileptal® (oxcarbazepine), Lamictal® (lamotrigine), Banzel® (rufinamide), Sabril® (vigabatrin), and generic sodium channel blockers. Talk with your healthcare provider for more information on potential treatment interactions.

12. Are there credible non-medication treatments for controlling seizures?

A **ketogenic diet** is often a good alternative or complement to medication for seizure management in

children with Dravet syndrome. This diet is very low in carbohydrates and high in fat, which helps put the body into a metabolic state called ketosis. When a person is in ketosis, the body burns fat for energy, instead of burning carbohydrates. A **ketogenic diet** for seizure control should only be followed under medical supervision.

Some common healthy foods included in a **ketogenic diet** are:

- seafood
- poultry
- low-carb vegetables like cauliflower, cabbage, broccoli, or zucchini
- avocados
- dairy products like cheese, plain Greek yogurt, or cottage cheese
- eggs
- nuts and seeds
- coconut or olive oil

Please consult with your doctor before incorporating a **ketogenic diet** into a patient's treatment plan.

13. In addition to medication, how can I help reduce the risk of seizures in my child?

Creating a good sleep routine for your child, making sure they take all their medicines at the right time, and controlling shifts in temperature are all things you can do that may reduce their risk of seizures.

Because of this condition's sensitivity to heat, temperature control and proper fever management are very important.

CARING FOR PEOPLE WITH DRAVET SYNDROME

14. What should I do when my child has a seizure?

It's very important not to panic when your child has a seizure. Instead, try to stay calm and observe your child.

To keep your child safe and prevent accidental injury during a seizure:

- Prevent choking by laying them on their side or stomach
- Ensure that they are breathing adequately. Your healthcare provider can help show you how to check your child's breathing
- Do not place anything in the child's mouth
- Gently clean saliva and debris from their mouth with a soft cloth
- Note the time that the seizure begins and ends, what your child was doing when the seizure occurred (eg, using stairs, swimming, walking), and if the seizure occurred on only one side of the body
- Administer any treatment that your doctor has prescribed to stop the seizure

- Call 911 if the seizure does not stop within 3-5 minutes or your child does not fully regain consciousness
- If the seizure was caused by a fever, consult your doctor about giving your child a medication to reduce the fever (eg, acetaminophen, ibuprofen) when they are fully awake

A prolonged seizure, or **status epilepticus**, lasting more than 5 minutes requires the use of a rescue treatment. If this seizure does not stop in spite of this procedure, your child must be taken to an emergency room. We recommend having a written family emergency plan prepared and reviewed by your child's doctor.

15. What are trigger factors for seizures?

Triggers may be different from one person to the next. Possible triggers for a seizure include:

- fever and/or illness
- stress
- sleep deprivation
- emotional upset
- intense exercise
- flashing lights
- loud music

These triggers usually immediately precede the seizure.

16. What daily life difficulties might my child face?

Each child will have a different experience with Dravet syndrome, its associated seizures, and the impact they have on their life. It is important to consider factors beyond just seizure control when caring for your child.

It is recommended that you consider screening for learning difficulties, as well as for behavioral and psychiatric problems. These issues could include:

- attention deficit hyperactivity disorder
- autism spectrum disorder
- developmental coordination disorder
- depression, anxiety, or aggression

FAMILY LIFE

17. How will our family's daily life change?

Every patient is different, so the healthcare needs and seizure triggers will be unique to each child and will affect each family's everyday life in different ways.

Patients typically require around-the-clock monitoring and care. There will be frequent healthcare appointments to properly manage seizures and other associated health issues, as well as therapy sessions to assure the best outcome for your child's development.

Daily life will change. Many activities you took for granted will now require planning, and last-minute changes to plans and cancellations will be common. Families often avoid situations that could trigger a seizure in their child. Common seizure triggers for patients with Dravet syndrome include overexcitement, overheating, pattern/light sensitivity, and illness. Often, a parent may leave their job to become the primary caregiver for their child. These changes will be a difficult adjustment for everyone in the family, but you will learn to adapt. If there are siblings in the home, it will be important to talk to them about their concerns and fears.

Parenting may not turn out to be what you imagined, but you will find your family's new normal. Over time, you will become your child's best advocate and will learn how to most effectively manage this complicated care. Throughout this journey, some days will be harder than others, and you will experience a roller coaster of emotions such as fear, anger, and grief. Know that these emotions are normal, and you will get through it. The Dravet Syndrome Foundation (DSF) offers a private parent support group that offers information and strength from others who are going through the same struggles as you and your family.

18. How do I explain Dravet syndrome to my other children?

Living with a brother or sister, including one with a disability, can be rewarding, confusing, and stressful. Siblings will want to understand why there are some things their affected sibling cannot do. It is important to be open and honest and answer any questions in an age-appropriate way, as well as to give them a chance to help out. This will allow them to feel valued and secure about their place in the family and will allow them to bond with their sibling.

There are books available geared toward children about epilepsy and disabilities that may help younger children begin to understand their sibling's special needs. Check with your hospital to see if there are any programs offered for siblings (Sibshops is one available resource). Also, many siblings see a therapist to help them process their emotions and fears.

19. How do I explain Dravet syndrome to my family and friends?

Dravet syndrome is a rare and treatment-resistant form of epilepsy that begins in the first year of life. It is a chronic and lifelong condition with many associated health issues. There is currently no cure. Today's treatment options are limited and work differently for every patient.

Patients take multiple daily medications and also require emergency interventions for prolonged seizures. Medication side effects can include hyperactivity, lethargy, and anorexia, which in turn also need to be managed. People with Dravet syndrome have an increased mortality rate.

Management of Dravet syndrome impacts every aspect of daily life not only for the patient but the entire family. Caregivers face constant worry about keeping their child healthy and safe. They must help the child avoid seizure triggers, manage other conditions they may have, arrange for care management, and much more. Dravet syndrome can have a considerable financial impact on the family. Most people with Dravet syndrome are not able to live independently.

DRAVET SYNDROME IN CHILDHOOD

20. What is the course of Dravet syndrome in childhood?

Dravet syndrome typically starts in the first year of life, with most children experiencing their first seizure at 5-8 months of age. Seizures at this time are often triggered by fever, illness, or vaccinations, but can also occur when the child is otherwise healthy.

Seizures may be prolonged (from 5 to over 30 minutes) and present in multiple forms—most typically, **generalized tonic-clonic** and **hemiclonic** type seizures (see Question 7 and Glossary). Development is normal during this initial stage of Dravet syndrome, and tests like the **electroencephalogram (EEG)** and **magnetic resonance imaging (MRI)** also often show normal results.

Between the ages of 1 and 5, multiple seizure types begin to appear such as **tonic-clonic**, **myoclonic**, **atypical absence**, and **focal impaired awareness**

seizures (see Question 7). These can occur both with and without fever. For many, seizures reach their highest frequency during these years. It is during this time that developmental progression may slow after prolonged or frequent seizures. Other problems such as inattention, hyperactive behavior, and sleep difficulties may arise.

Patterns of seizures and development may continue to change from age 5 to adulthood. Seizures often stabilize in frequency as the child ages, but the seizures never completely go away. The duration of seizures often shortens with age, and hospitalizations for prolonged seizures are, in turn, less common. While delays in learning and development will persist, children with Dravet syndrome can still gain new skills over time. The severity of cognitive delay is different for every child with Dravet syndrome and is fairly evenly split between mild, moderate, and severe. Around the age of 6, some children will develop an unsteady gait, which may limit their ability to walk long distances. As people with Dravet syndrome reach adulthood, they continue to have cognitive delays and limitations in their ability to live independently.

21. Can my child attend school?

Children with Dravet syndrome can attend preschool and grade school, just like any other child. As with any child, there is a higher likelihood of exposure to illness when in school settings, but this should not keep a child with Dravet syndrome from school. Practicing proper handwashing and avoiding contact with people who are sick can help your child avoid illness.

School personnel should be familiar with the child's diagnosis and know basic seizure first aid. They should also be familiar with seizure triggers (like overheating and fever) and be aware of how to limit exposure. All children should have a seizure action plan available at the school, along with rescue medication if prescribed by their doctor.

Children with Dravet syndrome will often have an individualized education program (IEP) in place with the school, which details the learning goals for the child and the accommodations expected from the school to help them achieve those goals.

Many children will also receive specialized therapies such as speech, physical, and occupational therapy

as part of their schooling. For many, school is an important component in the child's development, both cognitively and socially. In addition to the child's education, school can provide an important source of respite for caregivers, allowing them to tend to their own needs as well.

DRAVET SYNDROME IN PUBERTY AND ADULTHOOD

22. What services might my child need as a teen?

Dravet syndrome is a spectrum disorder, meaning that each individual's experience is different. The severity of symptoms varies, and some children may not experience all of the symptoms discussed in this booklet.

Dravet syndrome involves more than just seizures—the majority of children with this condition do have some degree of intellectual disability, behavioral issues, sleep difficulties, and motor impairments. For these reasons, finding an appropriate, supportive learning environment will be very important. You should work closely with your school district office of special education and ask for an individualized education program (IEP). The IEP sets specific academic and cognitive goals designed specifically for your child, but may also include interventions targeting behaviors and motor impairments. This plan could involve a behavioral therapist along with occupational, physical, and speech therapy.

Mental health is also important. It is often difficult to assess mental health in teens with intellectual disabilities, but this does not mean that it should be ignored. Teens with epilepsy may be aware that they are different, and this can result in anxiety and/or depression. Having access to a mental health provider or therapist that is skilled in working with patients with intellectual disabilities can be very helpful.

Transitioning to a new medical provider can also cause a lot of anxiety. You and your child will have likely developed a close relationship with your pediatric neurologist or epileptologist. Having a discussion with your doctor in the early teenage years about a transition plan is extremely important so this transition of care can occur more smoothly over an extended period of time.

23. What services might my child need as an adult?

While there are many possible outcomes for teens transitioning into adulthood, the majority of adults with Dravet syndrome will not be able to live independently for many of the reasons discussed here. This brings up additional issues that need to be addressed, such as conservatorship, making caretaking plans, and finding options for where they may live.

The burden of seizures may actually decrease in adulthood, but motor impairments may continue to worsen, affecting posture and gait. Some patients may also develop a movement disorder that is similar to Parkinson's disease, where overall movements are slowed. These motor impairments bring additional challenges to mobility at home and in the community.

GLOSSARY

Autosomal dominant inheritance pattern: when a trait is dominant, a single copy of a disease-associated gene mutation is enough to cause the disease. This means that if one parent has a mutated *SCN1A* gene and they pass it on to their child, the child has a higher risk of developing Dravet syndrome. “Autosomal” means “not on a sex chromosome,” so these patterns affect boys and girls equally.

Attention deficit hyperactivity disorder (ADHD): a medical condition that often begins during childhood and includes symptoms such as poor concentration, hyperactivity, and impulsivity.

Crouched gait: a walking abnormality characterized by an increased flexion of the hips, knees, and ankles that makes it difficult to walk long distances. The younger the gait disturbance starts, the worse it will be when reaching adulthood. Nevertheless, most patients can walk at home, school, or work. A wheelchair may be necessary for some.

De novo mutation: a “new” mutation that occurs spontaneously before conception or at the time of conception—neither parent carries the gene mutation.

Dravet syndrome: Dravet syndrome is a rare, hard-to-treat, drug-resistant form of epilepsy that typically begins in the first year of life in an otherwise healthy infant and lasts throughout the person’s life.

Electroencephalogram (EEG): a test used to evaluate the electrical activity in the brain. Brain cells communicate with each other through electrical impulses. An EEG can be used to help detect abnormal activity that suggests a higher risk of seizures.

Hypertonia: increased muscle tightness and reduced flexibility due to dysfunction in the brain or nervous system.

Ketogenic diet: a diet that is very low in carbohydrates and high in fat that may reduce seizures.

Magnetic resonance imaging (MRI): a medical imaging technique using strong magnetic fields and radio waves to create images of the body.

Pathogenic variant: a genetic alteration that increases an individual’s susceptibility or predisposition to a certain disease or disorder. When such a variant (or mutation) is inherited, development of symptoms is more likely, but not certain. Also called a deleterious mutation, disease-causing mutation, predisposing mutation, and susceptibility gene mutation.

Seizures, types of:

Atypical absences: when experiencing atypical absences, your child looks like they are lost in their own world. These absences may last from a few seconds up to several minutes. This type of seizure may persist up to around age 12. These have a less clear beginning and ending than typical absences.

Clonic seizures (convulsions): rapid rhythmic jerking movements that may affect one limb, one side of the body, or the whole body.

Convulsive seizures: seizures involving more violent motor phenomena (hypertonia, convulsions).

Febrile seizures: epileptic seizures brought on by a fever in an immature brain. These commonly occur in healthy infants or small children and are usually outgrown by age 5. In children with Dravet syndrome, fevers or hot environments can commonly trigger seizures.

Seizures, types of (cont'd):

Focal impaired awareness seizures: seizures that start in one area or side of the brain, causing the person to lose awareness of their surroundings. These seizures typically last 1-2 minutes and are often followed by fatigue or confusion.

Focal seizures (partial seizures): seizures that start in a small area of the brain and can be detected by EEG recordings. They have a range of manifestations, including muscle stiffening, irregular breathing, and sweating. Overall, these seizures are usually brief, lasting less than 2 minutes.

Generalized seizures: seizures affecting both sides of the body.

Generalized tonic-clonic seizures: seizures that involve the entire body and consist of generalized stiffening, followed by rhythmic jerking. They can also be called grand mal seizures.

Hemiclonic seizures: seizures with rhythmic jerking that only affect one side of the body.

Myoclonic seizures: sudden, very brief twitching movements, sometimes repeated, similar to how someone might jerk when they are startled. They are generally of moderate intensity. Fever may increase their intensity and frequency.

Non-convulsive status epilepticus: a prolonged seizure that manifests primarily as altered mental status as opposed to the dramatic convulsions seen in generalized tonic-clonic status epilepticus.

Obtundation status: a special type of seizure in Dravet syndrome consisting of fluctuating alteration of consciousness with reduced postural tone and myoclonic jerks.

Status epilepticus (prolonged seizures): a medical emergency defined as a continuous seizure lasting more than 30 minutes, or 2 or more seizures without full recovery of consciousness between them. It is associated with significant morbidity and mortality risk.

Tonic seizures: seizures where the body, arms, or legs suddenly become stiff or tense.

Tonic-clonic seizures: seizures that have 2 stages:

- Tonic phase: the muscles stiffen; air being forced past the vocal cords causes a cry or groan; the person loses consciousness and falls to the floor. The tongue or cheek may be bitten, so the patient may drool saliva mixed with blood, and their face may turn blue
- Clonic phase: the arms and legs jerk rapidly and rhythmically, bending and relaxing at the elbows, hips, and knees. After a few minutes, the jerking slows and stops. Bladder or bowel control is sometimes lost when the body relaxes. Consciousness returns slowly, and the person may be drowsy, confused, agitated, or depressed. These seizures generally last 1-3 minutes.

A tonic-clonic seizure that lasts longer than 5 minutes requires medical help. Caregivers should contact local emergency services for assistance by calling 911.

Sudden unexpected death in epilepsy (SUDEP): the sudden and unexpected death of a person with epilepsy who was previously healthy.

Typical absences: brief losses of consciousness without convulsion. The child stops what they were doing, gets a vacant stare, and becomes unresponsive. These seizures generally last a few seconds or longer. They have a clear beginning and ending. They may pass unnoticed.

RESOURCES: ADDITIONAL INFORMATION

FAMILY SUPPORT WEBSITES AND ASSOCIATIONS

- Dravet Syndrome Foundation:
<https://www.dravetfoundation.org>
- Epilepsy Foundation:
<https://www.epilepsy.com/learn/types-epilepsy-syndromes/dravet-syndrome>
- National Organization for Rare Disorders:
<https://rarediseases.org/rare-diseases/dravet-syndrome-spectrum/>

You can also find local support groups through your health system or on the Dravet Syndrome Foundation website at <https://www.dravetfoundation.org/dsf-family-network/>.

CONTACT INFORMATION



BIOCODEX is a family-owned multinational pharmaceutical company founded in France in 1953, with proven expertise in treatments for the central nervous system. We are dedicated to providing education and support in the field of Dravet syndrome to healthcare providers, affected individuals, and their families.

All trademarks are the property of their respective owners.



© 2020 BIOCODEX, Inc. All rights reserved. Printed in USA.
BBD-00022-062020