



Living with Dravet Spectrum Disorder

A guide for Newly Diagnosed Families

Helping the Child While Finding the Cure

Introduction

As the parents of a child newly diagnosed with a Dravet Spectrum Disorder, one of the first things you probably wondered is: “how are we going to manage all of this?” This guide is designed to help you do just that. You’ll get the answers to questions that you’ve no doubt already started asking -- and to the questions you haven’t even thought of yet. You’ll get:

- what Dravet Spectrum Disorders are -- and aren’t
- answers to the questions you are asking right now
- tips for getting plugged into the network of available resources and educating yourself
- suggestions for how to talk to your doctors, caregivers, and teachers about Dravet Syndrome Disorders
- tips and techniques for taking care of yourself so that you're able to care for your child better

**Epilepsy is not a
mental disorder**

This guide was written by parents who have been right where you are now. Over the years, they’ve heard every new parent ask the same questions that they did, too. So they asked themselves: “*What do I wish that someone had told me when my child was first diagnosed?*” and pooled their best advice just for you. They wrote this guide because the more informed you are, the better decisions you can make for your child, and the happier and healthier his or her life will be because of you.

And because you and your child are not alone!

What are Dravet Spectrum Disorders— and What Aren't They?

First, it's important to emphasize that *you didn't do anything to cause this*. There is nothing you could have done differently that would have prevented your child from having a Dravet Spectrum Disorder. It's not a punishment for anything you have or haven't done. So do yourself a favor right now and let yourself off the hook!

Dravet Spectrum Disorders are a group of related epilepsies having a similar genetic cause -- most commonly, a mutation in the SCN1A gene that helps control the flow of sodium ions to neurons in the brain and to muscle and endocrine cells throughout the body. Essentially what happens is that the mutated SCN1A gene allows too many or too few sodium ions to reach neurons and cells, triggering an "electrical storm" in the brain that manifests as seizures. Researchers still don't fully understand how these genetic mutations directly cause epilepsy. The disorders in the Dravet spectrum, listed from least to most severe, are:

Dravet Spectrum Disorders are so rare ..they often take a long time to diagnose.

- Familial Febrile Seizures (FS)
- Generalized Epilepsy with Febrile Seizures Plus (GEFS+)
- Intractable Childhood Epilepsy with Generalized Tonic Clonic Seizures (ICE-GTC)
- Severe Infantile Multifocal Epilepsy (SIMFE)
- Severe Myoclonic Epilepsy Borderline (SMEB)
- Severe Myoclonic Epilepsy of Infancy (SMEI); also called Dravet Syndrome

You can learn more about each of these on the Dravet.org website (www.dravet.org).

Dravet Spectrum Disorders are distinct from other types of epilepsy that are caused by physical defects or lesions in the brain, head injuries, strokes, or brain tumors. In Dravet Spectrum Disorders -- the most severe of which is called Dravet Syndrome -- the first seizures usually occur before one year of age, with no known cause other than fever or illness. Seizure severity and control varies depending on the disorder. Developmental delay, cognitive impairment, and physical handicaps also increase with the severity of the specific disorder within the spectrum. **Understanding** the causes of these epilepsies presents an ongoing challenge to researchers and doctors. They occur equally in both genders, and have no geographic or ethnic boundaries. As more is learned about these disorders and awareness of the clinical spectrum broadens, the number of people diagnosed with Dravet Spectrum Disorders is increasing.

Epilepsy is not a mental disorder. However, many children with Dravet Spectrum Disorders experience developmental issues that may be minimized with effective and early treatment. The fact that you are reading this right now means that you have already taken steps to get treatment for your child, which will greatly improve your child's prospects for living a healthier, happier life.

First Questions

It has probably been a long, hard road to get to your child's diagnosis. Because Dravet Spectrum Disorders are so rare and not as well-known as other forms of epilepsy, they often take a long time to diagnose. Just a few years ago, many parents just like you were trying for four, six, eight, and sometimes even ten years or more to get an accurate diagnosis. If your child is SCN1A-negative, it probably complicated the diagnosis. It may have taken you a lot of effort to get the hospital to agree to do SCN1A testing in the first place. Thanks to better education -- and the efforts of many parents who have come before you -- many hospitals are changing their testing philosophy. Today, many more children are being diagnosed in infancy.

**An accurate diagnosis
is critical for best
treatment results.**

And now, you're probably beginning to realize that the long road to getting to a diagnosis was just the first couple of miles along an even longer road that you're beginning to see stretching out ahead of you. Here are some factual answers to the most urgent questions you are probably asking right now:

Is there a cure for Dravet Spectrum Disorders?

No, there is no cure right now. There are treatments that will help reduce the severity and frequency of seizures (see below). Although treatment does not cure the disease, seizure control helps improve quality of life for both patients and their families. Optimal control helps children learn and grow to their full potential.

What is my child's prognosis? Is Dravet Syndrome fatal?

Prognosis for children with Dravet Syndrome used to be poor, but with so many more children being diagnosed in recent years, the prognosis may improve. If left untreated, seizures can be fatal. According to the National Institutes of Health (NIH) Office of Rare Diseases Research, the long-term prognosis and life expectancy for children with Dravet Syndrome is not well understood. Current research suggests that intellectual impairment is

correlated with the frequency of seizures, and that the decline in cognitive function tends to stabilize after the child is 4 years old. According to NIH, "[a]n individual with Dravet Syndrome has an 85 percent chance of surviving into adulthood." Many adult Dravet Syndrome patients are dependent on caregivers, however.

Will my child regress?

When a child with a Dravet Spectrum Disorder is on the proper medications, they won't necessarily regress. In fact, children with febrile seizures and GEFS+ generally don't experience regression. However, it's not uncommon for children to "plateau" for extended periods. Although with the correct treatment regressions and plateaus can often be effectively managed (see below), unfortunately many times, despite a parent's best efforts to control seizures and to optimize treatments and therapies, their child may still regress.

At present, there's no way to be certain what your child's outcome will be, but you can take heart in knowing there are adults with Dravet Spectrum Disorders who are married and have children. While this is not common for people whose disorders are in the severe end of the spectrum, as more physicians recognize and test for Dravet Spectrum Disorders we will likely see more children with less severe disabilities.

What treatment options are available? What medicines work best in treating Dravet Spectrum Disorders?

Treatments will differ depending on the severity of the disorder. They include the use of antiepileptic drugs, rescue medicines for prolonged seizures, and alternative treatments such as ketogenic diet (KD) and vagus nerve stimulation (VNS). We'll cover each one briefly.

The treatment of seizures and related conditions is unique to each person diagnosed with a Dravet Spectrum Disorder. An accurate diagnosis is critical for best treatment results. Because these are rare diseases, many families find it helpful to consult with an epileptologist (a neurologist who specializes in treating epilepsy). Remember that your child's reaction to treatment will be unique, and that even with the best medication regimen, seizures may persist.

A recent study has found that children with SCN1A mutations may also have

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among seizure control,
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and quality of life**

mitochondrial disease. Because the treatment of seizures for these two diseases are different -- and in some cases can actually cause harm -- you may want to consider obtaining a complete diagnostic evaluation of your child to ensure that he or she doesn't also have mitochondrial disease. If your child tests positive for mitochondrial disease, speak with your physician about suitable treatment options, such as supplements.

1. Antiepileptic Drugs

Seizure-preventing medicines, also called antiepileptic drugs (AEDs) or anticonvulsant drugs, are prescribed for the various Dravet Spectrum Disorders. Doctors prescribe medicines with the goal of seizure-freedom with few or no side effects from therapy. Often, it's not possible to achieve complete seizure control; instead, you may have to seek a balance among seizure control, medication effects, and quality of life that is right for your child and you. The drug regimen will vary depending on the severity of the disease; children with inherited febrile seizures, for example, may only need one anti-epileptic drug -- or none at all -- whereas children with Dravet Syndrome are often on polytherapy (three drugs) and also on the ketogenic diet (KD) or have a vagus nerve stimulation (VNS) implant. (KD and VNS are discussed in more detail below.)

Although there are many seizure-preventing medicines in use worldwide, their availability can vary widely, even between the United States and the European Union. Most antiepileptic drugs are taken by mouth. Some people experience side effects, others will not. Side effects generally are related to the dose; the greater the dose, the greater the likelihood of a side effect. Ask your child's doctor about possible side effects. Also, ask about drug interactions with other medicines or foods.

Antiepileptic medicines that **may help** patients with Dravet Spectrum Disorders:

- clobazam (Frisium, Urbanyl, Onfi)
- clonazepam (Klonopin, Rivotril)
- clorazepate (Tranxene, Novo-Clopate)
- divalproex sodium and derivatives (Depakote, Depakene, Epilim, Epival, Micropakin)
- leviteracetam (Keppra)
- stiripentol (Diacomit, which is available only in the European Union)
- tiagabine (Gabitril)
- topiramate (Topomax)

Antiepileptic medicines that **may make seizures worse** in patients with Dravet Spectrum Disorders:

carbamazepine (Tegretol, Calepsin, Cargagen, Barbatrol, Epitol Finlepsin, Sirtal, Stazepine)
fosphenytoin (Cerebyx, Prodilantin)
lamotrigine (Lamictal)
oxcarbazepine (Trileptal)
phenytoin (Dilantin, Epanutin)
vigabatrin (Sabril, Sabrilan, Sabrilex)

Caring for your child
will be a team effort.

Febrile seizures are not always treated with antiepileptic drugs. They may only require intermittent treatment during the febrile episodes or illness. For the less severe disorders like FS or GEFS+, monotherapy (the use of a single anti-epileptic drug) is often enough to control seizures in treatment, while for Dravet Syndrome the use of three or more drugs is common.

2. Rescue Medicines

Medicines that stop seizures in progress, also called rescue medicines, are prescribed for those with a history of prolonged seizures (also called *status epilepticus*). These drugs are usually in the benzodiazepine class such as clonazepam (Klonopin), diazepam (Diastat, Valium), lorazepam (Ativan), or midazolam (Versed). Your doctor will provide you with specific instructions for their use according to the specific needs of your child.

3. Non-Pharmaceutical Treatment Options

The **Ketogenic diet** (KD) is a very strict diet that is high in fats and low in carbohydrates. Because there are potentially very serious risks associated with KD, it is prescribed under the careful supervision of a doctor who is familiar with its use; a team of doctors, dietitians, and nurses work together to manage your child's KD and you should not attempt it on your own. Other dietary therapies such as the low glycemic index diet or other modified low carbohydrate diets can be useful with some patients. These diets also require the advice of an experienced team to get the most benefit while also reducing the risks. If you decide to start your child on a Ketogenic diet, sooner is better. If they are allowed to develop tastes for non-KD foods, it will be harder to get them to make the switch to the kinds of food that are allowed in KD. KD is used as a treatment for children whose epilepsy is at the more severe end of the spectrum; it's not likely to be recommended for children with febrile seizures or GEFS+.

Vagus nerve stimulation (VNS) therapy involves sending small pulses of electricity to the brain through the vagus nerve, which is a large nerve in the neck. A surgeon implants the VNS generator device under the skin on your child's chest and attaches wires to the vagus nerve. Like KD, VNS is typically recommended for children whose epilepsy is at the severe end of the Dravet spectrum.

Getting Connected and Getting Educated

Although you may be feeling alone and isolated right now, there are many people, resources, and tools available to you when you need them. Most of them are just a phone call or a mouse click away. The more connected and the better educated you are, the more help you will be to your special needs child.

Meet the Parents. Early on, it's important to start getting in touch with other parents and to begin seeking out accurate, reliable information from websites that are focused specifically on forms of intractable childhood epilepsy such as Dravet.org. A lot of the information you'll find on the Web is outdated or just plain inaccurate, but thanks to the efforts of the IDEA League (the predecessor of Dravet.org), in recent years much more research and accurate information has been published about Dravet Spectrum Disorders. Hearing other parents tell their stories will help make your situation seem not quite so catastrophic, and they'll be able to point you in the right direction for finding resources.

Create an emergency room plan that includes step-by-step treatment in case of seizure.

Consider Seeking Out Reputable Online Resources. Many families connect with each other online via the Dravet.org group on Facebook, through the forum on the Dravet.org website, and via blogs. These resources might be a good way for you to connect with other parents all over the world -- some of whom who are probably not too far away from you -- who are going through the same thing that you are. You can also meet family members and learn more about the latest research and treatments at Dravet.org's biennial conference (visit www.dravet.org for more information).

Educate Yourself about SUDEP. Too many parents are unaware that their Dravet child is vulnerable to something called SUDEP, which stands for Sudden Unexpected Death in Epilepsy. As its name implies, the causes of SUDEP are currently unknown. Because most instances of SUDEP occur at night, you can cut down on the risk by speaking with your physician about preventive measures that you can take, such as a video monitor, Emfit® Movement Monitor, or pulse oximeter.

Know that Your Child's Drugs Will Change over Time. Your doctor will help you find the appropriate drug or drug combination for treating your child's symptoms. Don't try them all right away; go slowly. Otherwise, you won't have time to really identify which drug is working. Also, be prepared to try other drugs in the future. As your child grows, their metabolism will

naturally slow, affecting dosage levels and drug effectiveness; over time, you will need to try different drugs (alone and in combination) to be able to continue managing their symptoms.

Track Your Child's Health. Using a 3-ring binder, a notebook, your computer, or your smart phone, create a portable patient notebook for your child that you can take with you to the hospital and on trips. The notebook should document essential medical data such as your child's care plan, seizure logs, a list of medications, doctor's visits, and contacts. You can also create a master notebook that stays at home and a smaller travel version, if that works better for you. Another handy resource is Seizure Tracker (www.seizuretracker.com), a free online service that lets you log and track seizure activity, doctors' appointments, and medication schedules, print seizure logs, and generate customized reports through your computer and smartphone.

Caring for a child with chronic medical issues is not easy, and you're going to need some help along the way.

Talking with Doctors, Caregivers, and Teachers

Caring for your child will be a team effort. Chances are, your child's pediatrician, neurologist, nurses, caregivers, and teachers will never have met a child with a Dravet Spectrum Disorder. You will probably end up educating them about your child more than they will be educating you. Don't worry about that -- the more you learn about your child's diagnosis (DSD here seems redundant), the more of an expert you will become about your child's condition. And throughout it all, **trust your instincts!**

You may need to help your child's physician to recognize that many Dravet Spectrum Disorders (particularly ICE-GTC, SIMFE, SMEB, and Dravet Syndrome) are considered a severe disability. While many children with Dravet Spectrum Disorders look and act normally most of the time, this does not mean that they do not have a serious medical condition. Just because they may not "act sick" when they are visiting a physician, doesn't mean that they don't require constant supervision and medication to prevent a sudden seizure. They have a very complicated condition that can -- and will -- change often. Therefore, when introducing your child's condition to a new physician, it might help if you use the term "catastrophic epilepsy with associated developmental delays", or "intractable (drug resistant) epilepsy" instead of "Dravet Syndrome" or "Dravet Spectrum Disorder." They will understand that language and respond accordingly. And if you have any doubts, **don't be afraid to get a second opinion.** You are the health care consumer; it is your choice and your right.

Work with your child's physician to create an emergency room (ER) plan that includes step-by-step

treatment in case of seizure. Include instructions for paramedics and school nurses in the plan. Contact your local 911 squad and inform them about your child's needs. Be sure to let them know that your child's condition is **high priority** because the seizures can be life threatening. Otherwise, they might mistake an emergency call from you for a common form of epileptic seizure and designate the response as a low-priority.

Caregivers should understand that children with Dravet Spectrum Disorders require **constant supervision**. They should never be left alone. Furthermore:

- They may not understand danger.
- They can have a seizure at any time.
- They have a very high pain threshold.

That means that by the time anyone notices that something is wrong, it could be well after the injury or illness that caused it. So constant monitoring is essential.

There are a number of low- or no-cost health care options that your child **may** qualify for because of their condition. They include:

- Early Intervention: a range of services provided to infants and toddlers with disabilities
- Special Education: school services that provide for your child's education in a way that accommodates their condition
- Home and Community Based Waivers: also called "Katie Beckett Waivers," these are waivers that allow your child to continue receiving Medicaid coverage while being treated at home
- Medicaid Waivers: money that pays for services in addition to doctor appointments, hospital expenses, medicine, therapy, and some adaptive equipment that Medicaid already pays for
- Consumer Directed Option (CDO): a type of Medicaid waiver that lets you hire your own caregiver
- Social Security Benefits
- Medicaid

You can find much more information about all these options on the Dravet.org website. Just click on "Help for Patients and Families" at the top of any page on the website.

Adjusting Your Family and Personal Life

The panic that hit you when your child got his or her diagnosis will subside. But the grieving process that you have also begun may never really end. It's important to deal with your grief so that you can continue to be good parents. It's normal, it's healthy, and it's important. Don't deny that of yourself, your loved ones, or your friends.

Consider seeking the assistance of a professional counselor to help you and your family find a way to navigate through this challenge -- there is no shame or failure in seeking this important kind of help. Caring for a child with chronic medical issues is not easy, and you're going to need at least some help along the way. Little things like going to the mall or playing at the park may be taken away for a time, but those memories can be replaced with other activities that are centered around your special needs child.

It affects your lifestyle. A diagnosis of a Dravet Spectrum Disorder -- especially of a disorder at the more severe end of the spectrum -- can be a complete lifestyle change that affects your entire extended family. You are pioneers setting out on a new and unexplored frontier called "life with a special needs child." For a while, everything you do is going to be a first.

Give yourself patience to learn, and to make mistakes. But also remember that you *will* get better at this. It takes time to adjust to daily life with a Dravet Spectrum Disorder, but with a good support system and the appropriate tools, you will eventually adjust to your new normal.

It affects your relationships. Having a child with special needs may have an impact on personal and family relationships. Some of your current friendships, frankly, will not survive the diagnosis. On the other hand, many friends will want to help and support you -- and those are the friends you'll want to have along on your journey.

Don't be afraid to ask others for help. One of the hardest adjustments for parents to make is the need to ask others for help when they're tired. Caring for a child with Dravet every day -- and every night -- can be exhausting. Don't be afraid to ask for help. You can start with the suggestions that are mentioned in this Guide, and also visit the Dravet.org website for even more ideas.

Start seeking out respite services now. Take the time to train someone else to take care of your child, in case of an emergency -- or just when you need a break. The more rested you are, the better you can take care of your child.

It requires caring for yourself, too. The better able you are to care for yourself, the better care you'll be able to provide your child. Set aside time for yourself, time to relax, read a book, meet a friend for lunch, get a massage, or even seek professional help if need be. Some parents seek out a therapist or psychologist in advance, someone they can turn to if the going gets tough.

Some Final Thoughts . . .

Focus on the day-by-day, and try not to get caught up in worrying about how things might be a year, five years, or 10 years down the road. Instead, you should try to focus on and celebrate your successes -- even the small ones. That sounds hard now, but it will get easier, especially if you celebrate them with friends and family who know what you are going through.

Find other parents to share your experiences with. The network of Dravet parents is very strong and active. They will cheer along with you and cry along with you too. A diagnosis of a Dravet Spectrum Disorder may not be easy, but if you have the support of friends, family, and professionals, they'll catch you when you fall and they will help you get back on your feet again.



The leading global patient advocacy organization for promotion and funding education, family support, and medical research to find cures for Dravet Spectrum Disorders.

For additional resources and to support our work, visit www.dravet.org.

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