Brief Communication

Journal of Child Neurology
Volume 23 Number 6
June 2008 690-694

10.1177/0883073808314162
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Coping With a Child With Dravet Syndrome: Insights From Families

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Dravet syndrome is a truly catastrophic childhood-onset epilepsy. Families are faced with repeated episodes of status epilepticus, intractable seizures, mental handicap, behavior disorders, and a life of dependency; understandably, coping with Dravet syndrome is very difficult. Twenty-four families with a child with Dravet syndrome were interviewed and identified some practical suggestions to improve their daily life. These included inserting an indwelling venous access device, creating a portable microenvironment, writing an emergency department protocol, establishing emergency routines for the family, assigning a parent on call to lessen the effect on siblings, creating personal time to decrease parental stress, finding respite care, and contacting an Internet support group. Unresolved and common issues included transition to adult care, the utility of early diagnosis, and social isolation. These solutions and issues may be helpful to many families with a child with Dravet syndrome and possibly other severe childhood-onset epilepsies.

Keywords: Dravet syndrome; management; social problems; Internet support

Dravet syndrome is an intractable epilepsy syndrome that evolves through 3 distinct stages. Stage 1 begins in the first year of life and consists of prolonged, frightening, convulsive tonic–clonic or clonic seizures and frequent episodes of status epilepticus, typically associated with fever.[1] Stage 2, which usually begins at about 1 year of age, is distinguished by additional seizure types and developmental plateau or regression.[1,2] Individuals who reach late childhood or early adolescence may enter stage 3, with a decrease in seizure type and frequency.[1] At all stages, however, seizures are notoriously unresponsive to medication, and a seizure-free state is seldom attained.[3]

We have previously described the specific challenges faced by families at each of the 3 stages.[4] During our interviews with 24 families, parents anecdotally reported interventions and coping strategies that they found particularly helpful for their child or their family. Many of these strategies were novel and interesting, and could be helpful for children and families affected by other types of intractable epilepsy. Here we present a number of these special strategies that we considered particularly helpful.

Venous Access

This child arrived at the emergency department of her local hospital during her 15th seizure in less than 4 months. Because blood to measure antiepileptic drug levels had been drawn twice each week, many of her veins were inaccessible owing to scarring and collapse. On this occasion, 2 physicians spent 40 minutes with unsuccessful attempts to insert an intravenous line. As a consequence, the status epilepticus continued for 50 minutes before a rectally administered medication stopped the seizure. After this experience, a nurse at a nearby tertiary care hospital suggested the insertion of a permanent intravenous catheter with percutaneous access, a suggestion that was promptly acted upon. The child's family has viewed this venous access device as lifesaving. In addition, stress for the child, family, and emergency personnel has decreased because they no longer worry about unreliable venous access.

An important concern for children with intractable epilepsy is the prompt availability of venous access for the treatment of status epilepticus. Venous access in children can be difficult at the best of times, and during a seizure, the often-febrile child presents an additional challenge. For this reason, many children in our study had a permanent indwelling intravenous catheter, such as a percutaneous indwelling venous access system (Port-A-Cath, Smiths Medical MD, Inc, St Paul, Minnesota). Parents of children who had received such a device were pleased with the results of quick access for treatment, which led to a feeling of additional security for the family.

Portable Microenvironment

Severe hyperactivity developed in this child with Dravet syndrome at about 5 years of age. Her mother was hesitant to

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J Child Neurol. 2008;23:690-694.

take her outside their home, fearing that she would run off. She decided to place her daughter in a stroller for outings. In addition to restraint, the stroller served as a portable environment, providing an excellent location for storage of emergency equipment and medications. These included the child's emergency protocol, oxygen, a suction machine, a rectal diazepam kit, and a cellular telephone. Her hyperactivity later subsided, but mobility problems developed, for which the stroller continues to remain helpful.

In our study, 88% of the children had a history of behavioral problems, most commonly hyperactivity. Ataxia was a problem with the older children, and 3 of 10 children aged older than 12 years were unable to walk. Every child in our study required emergency medication and other equipment to be nearby at all times. Many parents coped with all of these problems with the help of a transportable environment. In most cases, this environment consisted of a stroller in which the child was placed for mobility, accessibility of needed equipment, and also for restraint. The stroller can be outfitted with important medications and other equipment, such as suction and a cellular telephone, ensuring that needed items for use in the case of emergency are always close at hand. A written status epilepticus protocol, as described in the next section, is part of the equipment. Other items that parents found helpful to carry with them at all times included a change of clothes, diapers, cleansing wipes, and medication for the day. One parent advised us of the importance of having a lock for the stroller, because most are too big to fit in emergency transport vehicles should the child require urgent transport to the hospital.

Emergency Protocol

The child and her family were very familiar with the local hospital because she had presented there several times in status epilepticus. The staff had a clear understanding of the most suitable interventions for her. During one prolonged seizure, however, her ambulance was rerouted to another hospital. The staff at this hospital was not familiar with the child and, particularly, with the established inefficiency of certain emergency antiepileptic medications for the management of her status epilepticus. Despite her parents' protests, a variety of these medications were given and were ineffective. The result was a very prolonged seizure leaving her obtunded and necessitating admission to the intensive care unit. After this episode of status epilepticus, her family decided to create and establish a clear protocol for the emergency management of the child's prolonged seizures. The protocol was drafted with input from the local emergency department staff and her consultant neurologist. Now the protocol is posted in their local emergency department. A copy is carried at all times with the child; in addition, her mother also carries a copy at all times. The security of carrying a clearly defined emergency protocol has allowed her family to travel further from the area covered by her home hospital with less stress. In addition, they have found that emergency care at her home hospital has also improved by rapidity of triage and evaluation and accurate delivery of emergency rescue medications. New emergency department staff has accepted the written protocol without question.

Ten of 24 parents discussed the benefit of having a clearly established emergency protocol for their child. Such a protocol is kept in any setting that the child frequents: with the parents, with the child, in local responding ambulances, and most important, in the hospital where care is usually given at the time of an emergency. These protocols may include patient demographics, a brief history, diagnosis and allergies, caretaker information, physician to notify of arrival at the emergency department, daily medications and specific medications to be administered en route to the emergency department, and specific steps to be taken by emergency department personnel for quick and appropriate management.

Parents found the emergency protocol helpful for a variety of reasons. First, the establishment of an emergency protocol gave parents a sense of control in their child's care. Second, 15 of 24 of study participants described frustration with health care professionals who did not listen to their own experience and expertise or follow the recommendations of physicians involved in the long-term management of their child. In most cases, those who seem to benefit most from the easily accessible protocol are the emergency room personnel or first responders who encounter the child for the first time. An emergency protocol helps to avoid situations that result from a lack of familiarity with a rare condition like Dravet syndrome. The parent and trusted health care providers can work carefully together to construct an outline to ensure prompt, effective emergency treatment with specific interventions that are known to benefit the child.

Planning for Emergencies

This child lived in a remote community, far from the closest small hospital. His family had a well-established routine for managing his frequent seizures. When his seizure did not end spontaneously, his mother attempted to treat him at home with rectal diazepam. If this was ineffective, she ran next door to the home of the local paramedic. The paramedic attempted to treat the child, and if unsuccessful, the local hospital was alerted. The child was placed in the ambulance

and they began the long trip to the hospital, mindful of the timing because the trip entailed 2 ferryboat crossings.

In addition to an emergency protocol for use by health care professionals, many families discussed the importance of clear planning for the management of seizures and other emergencies at home. Most families had established guidelines regarding placement of the child in a safe location, administration of rectal, oral, or nasal benzodiazepines, contacting emergency personnel, and transportation to the local emergency department. Families found that having a clear routine decreased the stress of a prolonged seizure or other emergency event.

Effect on Siblings

A family found that their frequent trips to the hospital for their young son with Dravet syndrome were negatively affecting their older daughter; for example, she would often be removed from her after-school activities to accompany her family to the emergency department. The family was concerned that their daughter would become resentful of her younger brother. For this reason, they implemented a plan whereby at any given time, 1 parent was responsible for 1 child. Should their son have a prolonged seizure, the parent on duty on that day would accompany him to the hospital. The other parent would remain with their daughter and continue her regular activities.

Children with Dravet syndrome may have literally hundreds of emergency department visits, each lasting many hours. So the concern for siblings is very real. Parents had mixed reactions about the effect of a child with Dravet syndrome on siblings. Of 21 families with multiple children, 16 believed that the siblings were more caring, empathetic, and compassionate as a result of living with a child with a disability. However, 16 parents believed that the siblings were at times resentful of the increased attention paid to their brother or sister. Parents certainly struggled with this reality, and few felt that they had found a satisfactory solution. The presented example of having a parent on call provided a sustainable solution for 1 family. Other solutions included keeping the sibling well informed about Dravet syndrome and involving him or her in emergency planning. Families discussed the importance of giving the siblings a clearly established role in the case of a prolonged seizure; for example, the sibling might be charged with retrieving certain medications or calling emergency response personnel. Many parents believed that giving siblings specific roles decreased their anxiety during emergency situations that could otherwise be frightening for children. For single-parent families with several children, the strategy of a parent on duty clearly does not work. In some cases, understanding neighbors were described as a "godsend," especially when status epilepticus began at night.

Coping Mechanisms

This girl's mother has found that taking a small amount of time each day for herself decreases her stress levels and helps her better care for her daughter. While her daughter is at school or at home with another caregiver, she goes for daily walks; however, she knows that she must always be available should an emergency arise. For this reason, the mother walks in small neighborhood loops so that she is never more than 10 minutes from home. She carries a cellular telephone for emergency contact. The short time she has to herself each day is essential for her own well-being and for that of her child.

Six parents emphasized the importance of having an activity outside the home or taking some time for themselves each day. Parents also described many other interesting interventions and coping mechanisms, including access to multidisciplinary health care (such as occupational therapy, speech language pathology, social work, and others), faith, and personal research about Dravet syndrome. Some parents found summer camps for children with disabilities to be an excellent source of respite care, whereas others found the few hours a week of respite provided by an early childhood intervention program for children with disabilities to be helpful both for their children and for themselves.

Respite Care

One family found that it was nearly impossible to obtain a baby sitter for respite care for their daughter during the first and second stages of Dravet syndrome. Local resources were scarce, and traditional baby sitters felt appropriately ill equipped to manage the child's frequent seizures. Finding a person that they trusted was impossible. During a prolonged hospital admission, the family came to know quite closely several nurses and nursing assistants. When their child was discharged, one of these qualified nursing assistants offered to provide respite care as needed for the family. They have been incredibly grateful.

Respite care, even routine baby sitting, is critical for parents and families but incredibly difficult to arrange for children with Dravet syndrome, as noted by 17 families in our study. Those who had access to government-supported respite care often had to wait years before they received these services. In general, extended family members provided little respite support; however, 15 families received some respite care by other family members. Most grandparents and relatives were, understandably, frightened of the seizures. Health care professionals were used for respite care in 15 families. The best source of these helpful individuals is probably a children's hospital.

Support Groups

This child and her mother live in a small city in a remote area of Canada. Few other children with intractable epilepsy live in the area and likely none with Dravet syndrome. Her mother is a single parent with few family supports. She has many friends but finds that they are unable to understand the difficulties of caring for a child with Dravet syndrome. She has found that the most important coping mechanism in her life is access to an Internet-based support group for parents of children with Dravet syndrome.

Twelve of the families in our study were members of an Internet-based support group, and most of these parents found that the chance to communicate with other families in similar situations was very helpful. Several parents who attended general epilepsy support groups found them to be less helpful. Their experiences differed greatly from those parents of children with less severe epilepsy syndromes. In addition, many parents thought their presence at general epilepsy meetings was detrimental to other parents because their experiences were so severe that the mood of the group was negatively affected. For these reasons, specific support groups for families of children with Dravet syndrome or other types of intractable epilepsy tended to be more helpful for parents. Many parents of children in our study subscribed to a support group available through the International Dravet Syndrome Epilepsy Action League (www.idea-league.org).

Unresolved Issues

Although we found that the parents in our study provided a wealth of information and helpful tips, there were some issues for which no obvious solutions or helpful interventions were available.

Transition to Adult Care

Dravet syndrome has only recently come into focus for child neurologists, although it was first described by Dravet in 1978.[5] Because children with Dravet syndrome may live a long life with persistent seizures, transition to adult epilepsy care is an important step. The oldest participant in our study was 24 years old. Although 2 participants in the study were older than 18 years, which is the traditional age of transfer to adult care, neither had made the transition successfully. These patients were still visiting a pediatric neurologist for their health care needs. Issues such as the inadvisability of lamotrigine and the ongoing need for status epilepticus treatment may not be widely appreciated in the adult neurology community.

Value of Early Diagnosis

Reactions from parents in our study were mixed regarding the value of an early diagnosis. Many believed that an earlier diagnosis would have increased the options available to their child and might have led to a better outcome. Some parents, however, believed that an earlier diagnosis would have been overwhelming and disheartening. It remains unclear if an early diagnosis of Dravet syndrome improves the long-term seizure or intellectual outcome; however, issues such as those we have outlined suggest that there should be value in early diagnosis. The spectacular failure to make the diagnosis of Dravet syndrome in the child described in the book *The Spirit Catches You and You Fall Down*[6] led to many misunderstandings and inappropriate foster care of the child. This book, of course, is mainly about cultural misunderstandings. The advent of testing for mutations of the voltage-gated sodium channel alpha subunit type 1 (*SCN1A*) gene can assist earlier diagnosis.[7]

Social Isolation

Parents described increasing social isolation from family, friends, and their spouse throughout their child's life. This isolation was often attributed to decreased time available to spend with friends and family as they devoted increasing amounts of time to the care of their child. Parents also thought their friends and family had difficulty relating to their situation, and they often felt awkward discussing their child. Relationships with spouses were almost universally negatively affected by the stress of caring for a child with Dravet syndrome. It is possible that early social intervention could decrease stress and improve relationships with others, thereby decreasing the social isolation experienced by many of these parents. This may be an argument in favor of early diagnosis with the opportunity for contact with other families.

Conclusion

At times, physicians may learn more from their patients than vice versa, and our experiences with the families of children with Dravet syndrome illustrate this phenomenon. Many of the strategies and interventions used by these families were novel and interesting to us. We hope that what we have learned can help to provide new strategies for families of children with Dravet syndrome and possibly other childhood-onset intractable epilepsy syndromes.

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