

DRAVET SYNDROME FOUNDATION

# 5 YEAR STRATEGIC PLAN

2018-2022



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# WELCOME

The Dravet Syndrome Foundation (DSF) was founded in 2009 when a few parents came together who were anxious to see research for Dravet syndrome (DS) that would improve the outcomes for their own children. When this group learned that the governmental funding available for epilepsy research in general was limited, they knew for a rare disease like DS the efforts would need to be parent-driven.

I have had the honor of being one of the founding members of DSF, and over the years I have had the privilege to see first-hand the many accomplishments that our community has achieved in a relatively short time. By offering research grants for Dravet-specific research with a novel approach, we can help move researchers and the medical community forward to find better treatments and a cure for Dravet syndrome and related epilepsies. Through advocacy, education, and support, we can assure caregivers and medical professionals understand this complex disorder and that patients are receiving the highest quality of care.

Whether you attend or sponsor an event, complete a survey, participate in our registry, volunteer your talents, host a fundraiser, or offer advice or support to another parent who is struggling – those are all things that make our community stronger and help propel our mission forward. Our community members, supporters, clinicians, researchers, other nonprofit organizations, and industry partners are all essential collaborators in achieving our vision and mission. DSF exists and continues to gain momentum in the fight against DS because of each of you and the important role you each play.

Rather than feeling hopeless, I want you to better understand the work DSF is doing, our plans for the future, and how you can play a role and feel empowered. We truly are a powerful group when we work together and we have the ability to make great change. Thank you for being a part of that change and working with DSF towards a better future for all of those with DS.

*Mary Anne Meskis*

Executive Director

# LOOKING AHEAD

Much has changed in the field of Dravet syndrome (DS) since the inception of the Dravet Syndrome Foundation (DSF) in 2009. The purpose of this strategic plan is to articulate where we are today and the long-range direction and priorities for DSF. The content for our strategic plan was derived by examining the DS landscape. We listened to a broad range of input from key stakeholders within our community regarding current community priorities, emerging needs, and organizational strengths and vulnerabilities.

The objectives and strategies described in this plan have been reviewed and approved by our Board of Directors, Scientific Advisory Board, and Medical Advisory Board. These strategies will guide DSF's efforts and investments over the next five years and will balance our current and future needs.

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## OUR MISSION

The mission of Dravet Syndrome Foundation (DSF) is to aggressively raise research funds for Dravet syndrome and related epilepsies; to increase awareness of these catastrophic conditions; and to provide support to affected individuals and families.

## WHAT IS DRAVET SYNDROME?

Dravet syndrome is a rare, catastrophic, lifelong form of epilepsy that begins in the first year of life, with frequent and/or prolonged seizures. It affects 1:15,700 individuals, 80% of whom have a mutation in their SCN1A gene.

While seizures persist, other comorbidities such as developmental delay and abnormal EEGs are often not evident until the 2nd or 3rd year of life. Common issues associated with DS include:

- Prolonged and/or frequent seizures
- Behavioral & developmental delays
- Movement & balance issues
- Orthopedic conditions
- Delayed language & speech issues
- Growth & nutrition issues
- Sleeping difficulties
- Chronic infections



Patients with DS face a 15-20% mortality rate due to SUDEP (Sudden Unexplained Death in Epilepsy), prolonged seizures, seizure-related accidents (such as drowning), and infections.

Current treatment options are limited, and the constant care required for someone suffering from Dravet syndrome can severely impact the patient's and the family's quality of life. Receiving a diagnosis of DS can be overwhelming and leaves families with many unanswered questions while they adjust to the many needs of their loved one. Because DS is a rare disease, many medical professionals are not as familiar with the disease, leaving families to learn all they can to assure their child is receiving the best care.

Life with Dravet syndrome is difficult. Research for a cure offers patients and families hope for a better quality of life for their loved ones.

## ACHIEVEMENTS

Since our inception, Dravet Syndrome Foundation has made significant strides in uniting the Dravet community and funding research. From 2010-2017, DSF granted awards totaling more than \$3.6M for Dravet-specific research grants and postdoctoral fellowship awards. By uniting our patient community and supporting initial stage research, we have brought Dravet syndrome to the forefront - increasing interest among researchers, expanding funding mechanisms, and broadening the research community.

RESEARCH	AWARENESS & SUPPORT
Over \$3.6M in Research Grants	3 Biennial Family & Professional Conferences
8 Research Roundtables	\$140K in Patient Assistance Grants
North American Care Consensus	DSF Family Network
Clinical Research Award Established	Day of Dravet Regional Workshops

## RESEARCH OVERVIEW

In the US, most biomedical research is funded through the National Institutes of Health (NIH), which awards large grants of \$100K-\$400K per project per year to highly qualified researchers. Epilepsy has traditionally been underfunded, receiving just one-quarter of the research dollars awarded to breast cancer, despite a similar incidence. As a rare form of epilepsy, DS projects received just \$6.3M from the NIH between 2002 and 2009 before DSF was founded, and only about 30 studies were published.

When DSF began awarding research grants of \$50K-\$150K in 2010 in the hopes of drawing researchers into DS and establishing dedicated laboratories throughout the US, interest in DS grew exponentially. From 2010-2017, the NIH spent \$44.6M on DS research, an increase of 600%, and over 300 studies were published on DS. Six of the nineteen researchers in whom DSF invested in between 2010 and 2015 later applied for and received large NIH grants, representing a 32% rate of return.

# RESEARCH OVERVIEW CONTINUED

DSF’s strategic \$3.6M investment has broadened the research community, increased DS’s importance under other funding mechanisms, and has had lasting impact that sustains research growth.

DSF has hosted 8 annual Research Roundtables, which bring together researchers, clinicians, geneticists and other professionals with a strong interest in DS and epilepsy for an evening of presentations, group discussion, and brainstorming about where research needs to go to advance knowledge toward a cure. What started as a meeting of approximately 30 researchers has grown to almost 150 attendees in 2017.

These Roundtables, led by DSF’s highly esteemed Scientific Advisory Board, have been extremely successful in drawing top scientists and clinicians into the fold, allowing DS to be at the forefront of conversations about epilepsy research throughout the year. They have also allowed DSF the opportunity to establish a Research Roadmap to guide us in funding research projects that address the critical challenges of the syndrome and which will offer the most promising breakthroughs at the fastest pace.

RESEARCH AREA	\$ USD
SUDEP (Sudden Unexplained Death in Epilepsy)	\$300,000
Gene Therapy	\$703,000
Drug Discovery, Screening or Treatment	\$1,063,000
Genetics	\$714,000
Epidemiology	\$273,000
Neuronal Network	\$345,000
Other	\$300,000
<b>Total</b>	<b>\$3,698,000</b>

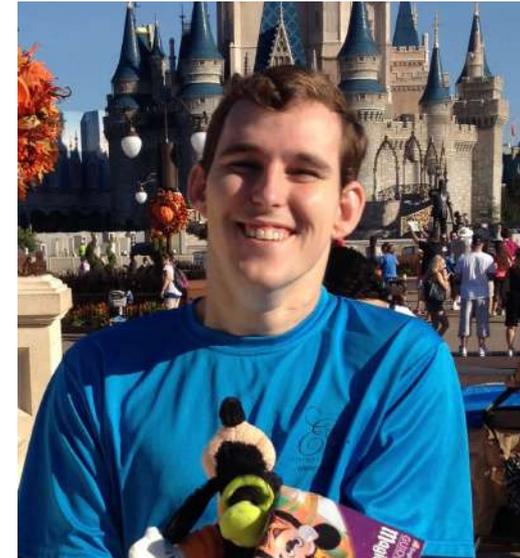


## AWARENESS & EDUCATION

In 2012 DSF expanded our mission statement by adding “to increase awareness of these catastrophic conditions” with the hopes of providing all professionals with the information needed for a time-sensitive diagnosis amongst children, adults, and populations not served by large centers with expertise in Dravet syndrome. Awareness began with the first online Facebook Support Group, which has grown from about 100 members in 2009 to over 2,000 members in 2018.

Education, a key component for a well-organized patient community that is actively engaged in collaboration with clinicians, has been an underlying theme of many of DSF’s programs. Our biennial conferences focus on families and professionals. We offer education for professionals through Continuing Medical Education (CME) sessions, ensuring accurate and timely diagnosis, standard of care, and new treatment options. For families, we offer a series of caregiver-relevant presentations covering topics such as educational plans, clinical trial participation, avoidance of seizure triggers, and understanding the pathophysiology of DS.

Our conferences take place in even-numbered years, with our 1st conference in Illinois in 2014; the 2nd conference in Florida in 2016; and our 3rd conference in Colorado in 2018.



## SUPPORT & ADVOCACY

DSF provides support and advocacy to families through several programs. Our Patient Assistance Grant (PAG) program has awarded over \$140K to families for medical and educational equipment not covered by traditional insurance plans, including iPads for communication; adaptive strollers/wheelchairs for ambulation; cooling vests to help with temperature regulation; and specialized glasses to help with photosensitivity. In 2017, thanks to a generous independent sponsorship, DSF was able to establish a separate fund to assist families recovering from natural disasters such as hurricanes, fires, and flooding.

In 2016, DSF implemented the DSF Family Network, which allows families to come together for mutual support and information-sharing. This family-centered and family-driven network is divided into 5 regional groups with Family Ambassadors assigned to each group to facilitate networking, support, and awareness to newly diagnosed and veteran members seeking information about particular topics.



In 2017, DSF hosted its first series of annual *Day of Dravet* workshops in each of the 5 DSF Family Network regions across the US. These events brought education about Dravet syndrome, comorbidities, patient-centered outcomes research, and caregiver stress to families unable to travel to the biennial conferences.

## STRATEGIC OBJECTIVES

Dravet Syndrome Foundation has developed four strategies with associated objectives. Our strategic plan includes resource plans, timelines, and measurable outcomes. Our strategic drivers are the guiding force behind our five year plan and each category is critical in shaping the future and direction of DSF and advancing our mission.



## RESEARCH AND PROFESSIONAL EDUCATION

Through collaboration and communication with our research community, we can improve the timeline for better treatments and a cure. By supporting opportunities that allow for greater education on Dravet syndrome throughout the healthcare system, we can increase diagnosis and assure the best quality of care.

GOALS	STRATEGIES
Announce one or more funding opportunities per year that help build the field of DS research	<ul style="list-style-type: none"> <li>• Increase funding and support of basic science and clinical research, through a coordinated funding strategy</li> <li>• Increase number of qualified research grant and postdoctoral fellowship applications</li> </ul>
Continued refinement of DSF's Research Roadmap	<ul style="list-style-type: none"> <li>• Identify high priority areas for focused research</li> <li>• Outline advances in the field of DS</li> <li>• Track non-DSF funded research to identify funding gaps</li> </ul>
Improve communication and collaboration between researchers in the field of DS research	<ul style="list-style-type: none"> <li>• Host our annual Research Roundtable meeting</li> <li>• Host a biennial Research Meeting specific to DS and associated sodium channelopathies</li> <li>• Partner with institutions to create alliances to accelerate research, increase diagnosis and improve treatment options</li> <li>• Influence DS research and stimulate new ideas and projects through ongoing communications with our scientific community</li> </ul>
Assure ready availability of professional educational resources	<ul style="list-style-type: none"> <li>• House recorded presentations from our biennial conferences on our website</li> <li>• Develop webinars on topics such as diagnosis, management, and adult transition of care</li> <li>• Exhibit at one or more professional conferences per year (such as the American Epilepsy Society Meeting)</li> <li>• Increase knowledge of clinical care best practices through distribution of the North American Treatment Consensus information</li> </ul>
DS Natural History Study	<ul style="list-style-type: none"> <li>• Collaborate with professionals in the development of a Natural History Study</li> <li>• Encourage and support patient enrollment</li> </ul>

# INCREASE REVENUE TO SUPPORT OUR PROGRAMS

Funding growth is challenging, particularly for a rare disease nonprofit. By increasing and diversifying our revenue streams we can ensure we have a sustainable operating model that meets the demands of our continuously growing community, and guarantee our success and longevity.

GOALS	STRATEGIES
Maintain, manage, and increase funds raised through our existing fundraising programs	<ul style="list-style-type: none"><li>• Continue engagement through peer-to-peer fundraising opportunities</li><li>• Increase funds donated by major investors</li><li>• Increase industry and corporate support</li></ul>
Increase fundraising programs and opportunities while engaging our community stakeholders and their extended networks	<ul style="list-style-type: none"><li>• Establish an Endowment Fund that will generate an ongoing stream of revenue</li><li>• Develop a Planned Giving program</li><li>• Develop new fundraising programs and increase events to make fundraising easier for our constituents</li></ul>



## EXPAND FAMILY EDUCATION & ADVOCACY

We will continue to support patients and families with disease education and advocacy needs through maintenance and expansion of patient resources, as well as family-to-family support.

GOALS	STRATEGIES
Provide readily available tools and resources to improve patient access of care	<ul style="list-style-type: none"> <li>• Supply educational downloadable pdfs on our website</li> <li>• Make printed brochures available on request</li> <li>• Increase the number of physicians listed on our website on the <i>Find a Doctor</i> page</li> </ul>
Empower our community through the development of new tools and resources	<ul style="list-style-type: none"> <li>• Develop and distribute surveys to respond to evolving issues and emerging needs</li> <li>• Identify common patient and family needs to provide direction for our program priorities</li> <li>• Strengthen our relationship with the growing Dravet adult population and their families to include their voice in our program evolution</li> </ul>
Engage and advocate, when appropriate, on behalf of legislative solutions that directly benefit or impact our patient population	<ul style="list-style-type: none"> <li>• Inform the community about legislative changes that directly affect them (such as social service benefits)</li> <li>• Partner with other advocacy organizations by representing the unique voice of the DS community regarding the impacts of national and state legislation</li> </ul>
Increase our in-person initiatives to allow families to connect with and support one another	<ul style="list-style-type: none"> <li>• Continue to host our biennial conferences</li> <li>• Increase <i>Day of Dravet</i> regional workshops and family gatherings</li> <li>• Increase membership of our private online support groups</li> </ul>
Expand our DSF Family Network and Family Ambassador Program	<ul style="list-style-type: none"> <li>• Provide annual training for family ambassadors that will give them the skills and tools to build relationships with families and potential supporters</li> <li>• Continue community outreach to connect families regionally</li> </ul>

## ENHANCE COMMUNITY DEVELOPMENT & COMMUNICATIONS

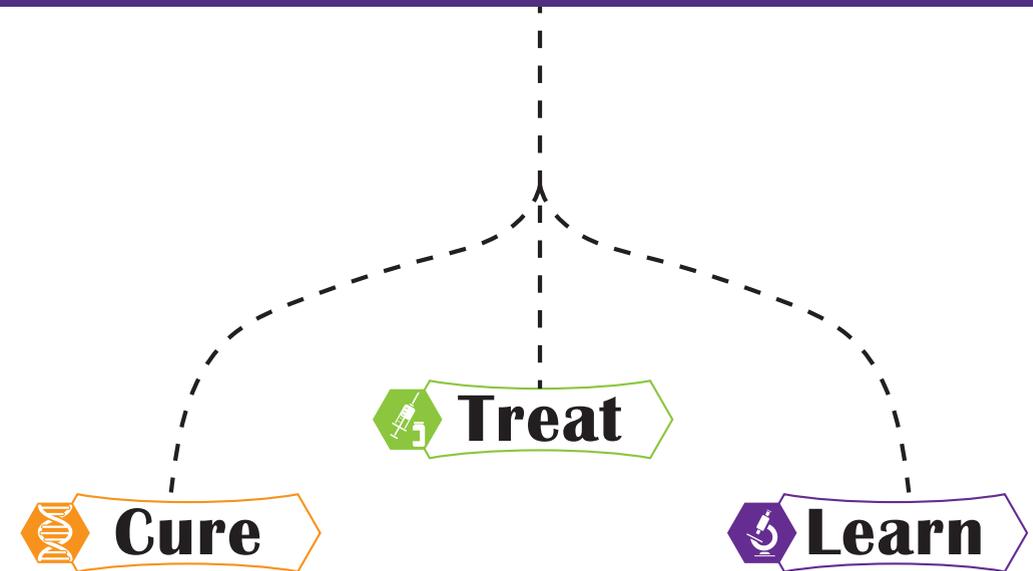
By increasing the leadership roles of our staff, board, volunteers, and donors we can build on current strengths through internal capabilities and resources, while strengthening our community connections. We recognize the importance of an ongoing assessment of our role within the community and the most effective use of our resources, as well as the need to continually communicate our priorities, goals, strategies and accomplishments to inform and engage our stakeholders.

GOALS	STRATEGIES
<p>Maintain a solid infrastructure to support a high functioning organization</p>	<ul style="list-style-type: none"> <li>• Maintain transparent and effective financial policies, procedures, budgets, and plans to continually expand the organization’s financial capacity to achieve its vision, mission, and strategies</li> <li>• Continue to develop and maintain operating policies, procedures, and records to ensure responsibilities are clearly communicated throughout the organization and tied to the goals and strategies of the foundation</li> </ul>
<p>Develop and implement a comprehensive communications plan to better address the needs and concerns of the Dravet community</p>	<ul style="list-style-type: none"> <li>• Increase social media presence through a coordinated plan to inspire and activate the Dravet community</li> <li>• Develop a community messaging plan, including the use of emerging technologies, that promotes the accomplishments and goals of the foundation</li> <li>• Ensure timely and accurate dissemination of information of interest to the Dravet community</li> <li>• Participate in rare epilepsy coalition groups and meetings to assure the voice of the Dravet community is represented</li> <li>• Identify common patient and family needs to provide direction for our program priorities</li> </ul>
<p>Acquire and retain committed board members, staff and volunteers</p>	<ul style="list-style-type: none"> <li>• Actively recruit, develop and evaluate staff and volunteers who embody the foundation’s vision and mission</li> <li>• Further expand and develop our volunteer committees to allow volunteers to become more involved and serve in a more strategic capacity</li> <li>• Develop an organizational structure that reflects clear and effective leadership, advisory, staff, and volunteer reporting</li> <li>• Establish a nominating committee for recruiting and interviewing future board members</li> </ul>

## RESEARCH PLAN

Dravet Syndrome Foundation (DSF), with the input of our community stakeholders, has developed a strategic research plan for Dravet syndrome (DS) to accelerate the research and development of new therapies.

Our 5-year strategic plan addresses the three highest priorities for research in our community. 1) Cure 2) Treat and 3) Learn. By distributing resources among these arms, DSF will continue to balance the need for progress toward a cure with the immediate need for better treatments and new pathways toward that cure.



## PROGRAM 1: CURE

As a disease with a distinct genetic cause, Dravet syndrome seems well-positioned for a cure based upon gene therapy or related treatments that modulate gene expression. Caregivers recognize that a cure will target the underlying problem rather than simply treat seizures, and progress of gene therapy in other disorders is exciting. However, this will be a long-term investment for Dravet syndrome. Research has shown that treating the genetic mutation - which occurs in every cell of the patient's body but causes the most dysfunction in the inhibitory interneurons nestled in the brain, away from the bloodstream - is more difficult in Dravet syndrome than in other genetic disorders. DSF is committed to overcoming these barriers from the ground up, beginning with

investigating whether increased *SCN1A* reverses the course of DS or alleviates symptoms. Potential cures or curative treatments for DS may involve:

- One-time gene therapy treatments, in which the mutated gene is corrected in the proper neurons
- Upregulation of the healthy copy of *SCN1A* or its transcripts to overcome haploinsufficiency caused by the mutated copy
- Altered regulation of other genes that compensate for the defective gene, such as other sodium channel isoforms

## PROGRAM 2: TREAT

Patients need better treatments for their condition now, long before a cure is developed. Traditional anti-seizure medications and treatments have proven unsuccessful in controlling all seizures and reducing comorbidities in DS. DSF is committed to investing in drug discovery, non-pharmaceutical company-sponsored clinical trial support, and research on treating the comorbidities associated with DS.

DSF will continue to help advance research to develop new treatments and improve the health and overall well-being of those with DS. With support from our community, we have played an instrumental role in advancing the field of DS by directly supporting research while encouraging and facilitating collaboration; collaborating with pharmaceutical companies; and developing a research-ready community.



## PROGRAM 3: LEARN

Our current understanding of Dravet syndrome is light years ahead of where it was when *SCN1A* was first associated with DS in 2001. With every expansion of knowledge comes the potential for new pathways for treatment and a cure, and it is this potential that strengthens DSF's commitment to supporting Dravet syndrome basic science. Recent examples of the role basic science plays include the discovery of the effects of 5-HT receptors on DS (which have led to two Orphan Drugs, one of which has been designated as a Breakthrough Treatment), as well as the connection between cardiorespiratory failure and SUDEP (Sudden Unexplained Death in Epilepsy). Basic science has the power to arm researchers with preliminary data and preclinical evidence needed for pursuing larger grants from other agencies. This learning can be accomplished through several avenues including the use of:

- Traditional mouse models
- Alternative animal models such as zebrafish and drosophila (fruit fly) models
- Induced pluripotent stem cell (iPSC) models
- In vitro organoids

## SHORT TERM GOALS (1-2 YEARS)

Maximize opportunities to address immediate needs

CURE	TREAT	LEARN
<ul style="list-style-type: none"> <li>• Discover the effects of therapeutically increasing <i>SCN1A</i> expression on symptoms and disease course</li> <li>• Determine whether targeting specific interneurons is necessary</li> <li>• If necessary, investigate mechanisms for specifically targeting interneurons for increasing <i>SCN1A</i> expression</li> <li>• Investigate viruses with interneuron-specific promoters</li> </ul>	<ul style="list-style-type: none"> <li>• Establish a Clinical Research Award</li> <li>• Work towards establishing an ICD-10 code specific to DS</li> <li>• Prepare the patient community for involvement in research</li> <li>• Identify and describe the natural history of DS against which new treatments can be measured</li> </ul>	<ul style="list-style-type: none"> <li>• Support organoid and other models of DS</li> <li>• Explore mechanisms related to SUDEP including breathing</li> <li>• Identify and describe the natural history of DS</li> <li>• Continue research on the mechanisms of DS and identification of potential pathways for treatment</li> </ul>

## LONG TERM GOALS (3-5 YEARS)

Ready for expansion & growth

CURE	TREAT	LEARN
<ul style="list-style-type: none"> <li>• Target <i>SCN1A</i> upregulation to interneurons</li> <li>• Explore other mechanisms of increasing expression</li> <li>• Investigate gene therapy techniques that may prove useful in future DS studies</li> </ul>	<ul style="list-style-type: none"> <li>• Identify the best interventions for seizures and comorbidities</li> <li>• Support clinical studies through research grants and patient engagement</li> </ul>	<ul style="list-style-type: none"> <li>• Support researchers as they explore new models</li> <li>• Examine new imaging techniques to assess network dysfunction at the cellular level</li> </ul>

## NEXT STEPS



When families receive the news that a loved one has been diagnosed with Dravet syndrome, they are often overwhelmed, scared, and confused. DSF is taking an active role in involving our community in finding a cure, while supporting patient families impacted by Dravet syndrome.

This 5 year strategic plan report represents an evolving and changing, multiyear effort that we hope will allow us the flexibility to meet foreseen and unforeseen changes ahead of us. In order to fulfill our strategic plan, we will require support from our entire community – funding from donors, engagement with caregivers and volunteers, and disease awareness generated by those involved in the Dravet community.

**There is no time to lose.** As we move forward and execute the strategies outlined in this strategic plan, we urge you to give today. Whether that means you are giving of your time, talents, or money, we need YOU. Our success is due to our community support. Visit [www.dravetfoundation.org](http://www.dravetfoundation.org) to make a donation, learn about upcoming events, or join our volunteer network.

Through all of our efforts, we remain committed to achieving DSF's mission as quickly as possible and to the benefit of the Dravet community. If you have any questions, would like to offer comments, or just want to become more involved, please contact Mary Anne Meskis, DSF Executive Director, at [maryanne@dravetfoundation.org](mailto:maryanne@dravetfoundation.org).



[www.dravetfoundation.org](http://www.dravetfoundation.org)