rare epilepsy landscape analysis

July – December 2019

Ilene Penn Miller

December 1, 2019

APPENDIX OVERVIEW

The following APPENDIX supplements the Rare Epilepsy Landscape Analysis (RELA). It includes more detailed inputs from 43 out of 44 RELA respondents. 1 respondents participated in the survey but opted out of the Appendix.

APPENDIX A. RELA ANNOUNCEMENT

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ANNOUNCING THE RARE EPILEPSY LANDSCAPE ANALYSIS (RELA)

Epilepsy Foundation is pleased to announce Ilene Penn Miller was recently hired as an independent contractor to conduct a **Rare Epilepsy Landscape Analysis** (RELA).

RELA Genesis
In the past decade, our understanding of epilepsy has evolved from a single disease to a complex of syndromes. A proliferation of nonprofit rare epilepsy syndrome focused 501c3 organizations and groups (hereinafter "rares") have emerged. Through the years, multiple collaborative initiatives including Institute of Medicine (IOM), Epilepsy Leadership Council (ELC), Rare Epilepsy Network (REN), Epilepsy Learning Health System (ELHS), Epilepsy Genetics Initiative (EGI), etc have formed to drive common objectives; however, a thorough scan of the rare epilepsy landscape to better understand the individual and collective goals, strengths, and needs of the disease focused epilepsy organizations has not been undertaken. Ilene Miller proposed undertaking a Rare Epilepsy Landscape Analysis to EF's Brandy Fureman and subsequently to Phil Gattone in early 2019. Epilepsy Foundation (EF) awarded a contract and the project will commence July 1 through December 31, 2019 with prep beginning immediately.

Opportunity
Staff, time, and financial resources are being consumed as each rare organization sets up independent operations, as well as develops information, education, support services, and research programs. In some instances, these resources are being expended for rare epilepsies with very small populations. There is collective interest in better understanding where goals align; what strengths exist within the rare epilepsy community; which resources are missing; and what priorities could be undertaken as collaborations. Some rares have expressed concerns regarding EF's alignment in mission and goals with rare organizations and potential ulterior motives for the RELA undertaking. EF is supporting this independent consultancy to gather insights to inform future strategic planning for the EF & REN, the rare epilepsy community, and the broader epilepsy community at large. This initiative prioritizes listening versus telling; transparency in process and communication; patient/caregiver centric outcomes, and identifying rare driven priorities and opportunities.

Process
The RELA will combine interviews, teleconferences and meetings, and a survey developed by and for the Rares to determine if and where there are opportunities for new more efficient, less duplicative, larger number collaborations and infrastructures in support of research strategizing and grant making; public and professional education and information development and dissemination; support and community building; operations, administration and compliance; and fundraising. The workplan begins with an internal audit of Epilepsy Foundation's (EF) leadership to assess existing mechanisms and opportunities to support rare patients and caregivers as well as partnerships with rare epilepsy organizations. Next, Rare epilepsy organization leaders will be engaged to develop a survey to determine strengths and needs across key program areas. Survey findings will be presented to EF leaders and the REN and Rares community in the timeframe of AES. The survey and analysis are intended to be a first step towards accruing baseline information and identifying new opportunities for collaboration.

Timeline
The project is broken into 4 milestones summarized below:

(1) **RELA PREP (5/15 to 6/15)** Interview EF leaders; Identify RELA partners; Courtesy calls to some rare org leaders and epilepsy influencers.
(2) **RELA LAUNCH & INFO GATHERING (6/1 to 7/15).** Publicly announce RELA. Build Advisory Committee including (small/large dz interests, genetic/other causes, newly organized to well established orgs) to draft/test survey. **RELA kick off call on 6/24 @ 3 PM.**
(3) **SURVEY DEVELOPMENT & IMPLEMENTATION (7/15-10/15)** Develop, build, test, revise and implement survey with Advisory Committee and EF staff designees.
(4) **ANALYSIS & REPORT OUT (10/15-12/15)** Analyze surveys and interviews. Report out to EF leadership and REN/Rares in Dec. 2019/AES timeframe. Coordinate strategic planning discussion re: next steps.

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KPIs
While the ultimate outcome from the survey and analysis is unknown and will be driven by the findings and analysis, several KPI's will include:

- Raising the Rares profile with EF leadership during interviews with 20+ EF leaders;
- Ensuring the most severe epilepsy concerns are included with intention in strategic planning throughout EF;
- Informing the broad epilepsy community about the strengths, gaps, and needs of the rares;
- Identifying strengths, priorities and gaps within Rare organizations; and
- Pinpointing priority areas for collaboration

About Ilene Penn Miller, JD, LLM
Ilene Penn Miller is as results driven leader with 20+ years' experience in nonprofit management, strategic planning, coalition building, marketing, fundraising and program development and oversight. She is a passionate advocate with proven ability to develop a vision, achieve consensus and deliver results in both for and nonprofit settings. She has been contracted as an independent contractor for this engagement and this engagement does not reflect the goals or objectives of Hope for Hypothalamic Hamartomas. Ilene co-founded and serves as President of Hope for Hypothalamic Hamartomas (hopeforhh.org). She serves as the Advocacy Co-Chair on the 2020 NINDS Curing the Epilepsies Conference, and is an active member on the Epilepsy Leadership Council (ELC), the Rare Epilepsy Network (REN), the Epilepsy Learning Health System (ELHS) and a former Advisor on the NIH National Institute for Neurological Disorders and Stroke (NINDS) Advisory Council (2013-2017). Previously, Ilene served as Executive Director of the Cure for Lymphoma Foundation and as a Senior Associate at Podesta Associates where she counseled a coalition of major national cancer advocacy organizations and implemented legislative, executive branch, grassroots, and media strategies to increase federal cancer appropriations. Ilene earned a B.S. in communications from Boston University; a J.D. from the Columbus School of Law at Catholic University of America (Washington, D.C); an LL.M. in advocacy from Georgetown University Law Center (Washington, D.C) and a Nonprofit Management Certificate from Georgetown University (Washington, DC).

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APPENDIX B. RARES IN THE SAME SPACE

Batten Disease	CDKL5	GNAO1	GRIN
Batten Disease Family Association and Norwegian Speilmeyer-Vogt Association Batten Disease Support & Research Association	CDKL5 Alliance CDKL5 Brazil CDKL5 India CDKL5 Research Collaborative Hope4Harper International Foundation for CDKL5 Research Loulou Foundation	Italy Famiglie GNAO1 Netherlands Stichting GNAO1 NL The Bow Foundation UK Mondo UK	Austin's Purpose Cure Grin GRIN2A Support Group GRIN2B Europe GRIN2B Foundation
ISAN	**LaFora**	**Neurodegeneration with Brain Iron Accumulation Disorders (NBIA)**	**NORSE**
Mickie's Miracles	Association France-Lafora Associazione Italiana Lafora (AILA) Chelsea's Hope	BPAN Warriors NBIA Disorders Association (US)	Association Paratonnerre in Paris NORSE Institute
Phelan McDermid Syndrome	**RASopathies**	**Ring 20**	**SCN8A**
CureSHANK PMSF	CFC International Children's Tumor Foundation Costello Syndrome Family Network French Costello/CFC French Association of Costello Syndrome and Cardio-Facio-Cutaneous International Costello Syndrome Support Group Italian Costello/CFC/RASopathies Association NF Network Noonan Syndrome Association - UK Noonan Syndrome Foundation Noonan Syndrome UK	Ring 20 Research & Support UK Ring Chromosome 20 Alliance	Ajude o Rafa Shay Emma Hammer Research Fnd The Cute Syndrome Foundation Wishes for Elliott www.scn8a.net
SYNGAP	**Tuberous Sclerosis**	**Copy Number Variants (CNVs)**	
Bridge the Gap SynGAP Leon & Friends (Austria) SYNGAP Elternhilfe (Germany) OverCome Syngap1 (Canada/France) SynGAP Research Fund Syngap1 Spain Syngap1 Switzerland	Tuberous Sclerosis Alliance Tuberous Sclerosis Association (United Kingdom) Tuberous Sclerosis International	8p Dup15Q	

APPENDIX C. FOUNDING STORIES

Organizations were invited to share how and why their organization was founded and encouraged to include the need as well as the founding story. Where applicable, this section also includes citations to key resources denoted by each organization. Clinical guidelines for treatment, management and diagnosis are also referenced here as well.

Batten Disease Support and Research Association, www.bdsra.org (1998) 501c3 OH
The Batten Disease Support and Research Association (BDSRA) is dedicated to funding research for treatments and cures, providing family support services, advancing education, raising awareness, and advocating for legislative action.

Founded in 1987, by parents seeking to build a network for those diagnosed with Batten disease, BDSRA is now the largest support and research organization dedicated to Batten disease in North America. BDSRA believes that to effectively unravel the mysteries of Batten disease, the worlds of medical science, research, and families must work together toward a common goal: discover treatments and cures while assuring a better quality of life for those living with the disease.

>>RESOURCE:
The Neuronal Ceroid Lipofuscinoses (Batten Disease) (Contemporary Neurology Series) 2nd Edition by Sara Mole (Author), Ruth Williams (Author), Hans Goebel (Author)

BPAN Warriors, https://www.bpanwarriors.org/ (2018) 501c3 FL

Sarah Chisholm, founded BPAN Warriors in 2018, ten months after her youngest daughter (then four) was diagnosed with BPAN. Similar to many rare disease parents, she never envisioned leading the development of a research network prior to her daughter's diagnosis. After many frustrating months of seeking medical/clinical care support and guidance, she recognized that there was a copious amount of unmet needs in the BPAN community and no BPAN specific organization existed in the world. Out of necessity and love, and determination, she redirected her early frustrations and launched the BPANWarriors.org website to provide a voice for the patient community along with much needed resources.

Using her 20+years of experience in sales and new business development/marketing, she has since turned her focus towards building a patient-centered research plan. Much of this last year has been invested in developing an infrastructure to support the fledgling non-profit, learning more about BPAN (the disease mechanism, the science and the existing research), identifying gaps in the overall research structure, and then diligently working to flush out a strategic plan to meet the needs of the growing BPAN community. The focus has been on how to capitalize on the existing research within the Neurodegenerative space and what technologies or collaborations would help bypass existing inefficiencies or might serve to codify a strategy towards uncovering a therapeutic path.

Although the science may be complex, BPAN Warriors remains undaunted. By leveraging new technologies, patient-centered data collection systems and ongoing data analytics and systems biology, we remain confident that we will be able to identify biomarkers, test new and existing drugs and potentially apply existing gene therapies to cure a relatively "simple", monogenic disease.

Since its incorporation in 2018, BPAN Warriors has been simultaneously focused on putting its infrastructure in place while also trying to bridge the gap between scientific discoveries and meaningful progress toward treatments and a cure for BPAN. Early on, we recognized the need for a collaborative network of scientists, researchers, patients and other stakeholders (those in the pharmaceutical and biotech industries). A research network will be essential in helping us understand and prioritize the most relevant and promising research approaches for this disease. Currently there are no FDA approved treatments for BPAN, no clinical trials underway, and no labs investigating cutting edge approaches, such as gene therapy, for BPAN. As an organization whose aim is to fund the most promising research, it is essential that we have consensus on what research questions are most important to answer in order to have the greatest impact for patients.

>>RESOURCE:
https://ghr.nlm.nih.gov/condition/beta-propeller-protein-associated-neurodegeneration#diagnosis
https://www.ncbi.nlm.nih.gov/books/NBK424403/
https://mastermind.genomenon.com/search?gene=wdr45

>>GUIDELINES:
https://www.ncbi.nlm.nih.gov/books/NBK424403/ There are "treatment" guidelines, however they are for the treatment of some of the symptoms, rather than the disease. As for diagnosis and evaluation guidelines, they exist, however we cannot really say whether or not clinicians actually use them.

Bridge the Gap - SYNGAP Education and Research Foundation, https://bridgesyngap.org (2014) 501c3 TX

In 2014, Monica Weldon became the Founder, President and Chief Executive Officer of Bridge the Gap - SYNGAP Education and Research Foundation. The foundation was established soon after Monica's son Beckett was diagnosed with an SYNGAP1 mutation in 2012. He was the first child identified at Texas Children's Hospital Genetics Clinic and was one of 6 individuals in North America identified at the time. Since its inception in September of 2014, the organization has grown rapidly because of the tireless efforts of the volunteer board of trustees and parents. In May of 2015, the foundation and scientific advisory board published the first combined descriptive summary of SYNGAP1 mutations published by the National Organization of Rare Disease.

Bridge the Gap - SYNGAP ERF is also partnering with several on-going research studies across the globe that are aimed at understanding epilepsies and autism spectrum disorders. We have two current studies focused specifically on epilepsies. The first being The Rare Epilepsy Network (REN) which is a collaboration between

the Epilepsy Foundation, RTI International, Columbia University and many other different organizations that represent patients with a rare syndrome or disorder that is associated with epilepsy or seizures. The REN will establish a registry of these patients which includes patient or caregiver-reported data in order to conduct patient-centered research. The second is a study being conducted by the University of Melbourne in Australia directed by Dr. Ingrid Sheffield. This research will assist us to understand the nature of SYNGAP1 disorders and epilepsy and other disorders associated with them. The hope is that the information collected will help families and their doctors to diagnose this condition, identify the seizure types and associated disorders, and select appropriate medication.

Our patient group is participating in an autism study with Simons VIP Connect. The research study is aimed to better understand the medical, learning and behavioral features of individuals with genetic changes associated with features of autism, developmental delays, and other neurodevelopmental concerns (like seizures). They identify the needs of the families while providing support through education, access to experts, and by connecting with other families.

In April 2016, the foundation was awarded by the National Organization of Rare Disease and the US Food and Drug Administration, the first and largest Natural History Study and Registry for SYNGAP1 (MRD5). This is a five-year project that will produce specific data about SYNGAP1 mutations and shared with researchers who study SYNGAP1 to find better treatments. Monica Weldon is the primary investigator on the project that includes 7 physicians and 3 Bridge the Gap - SYNGAP ERF charter members.

The organization has consistently and specifically worked to further education and research efforts to battle the effects of the Syngap1 gene mutation.

Bridge the Gap -SYNGAP Education and Research Foundation (501(c) 3) is a non-profit organization whose mission is to serve, educate and fund research for families coping with the effects of SYNGAP1 mutations. The leading organization advocating and raising funds for research and treatments for SYNGAP1. Our international outreach for SYNGAP1 children gathers critical information, which is needed to drive research towards more immediate therapeutic solutions. Bridge the Gap-SYNGAP mission is to improve the quality of life for people affected by SYNGAP1, provide family support, accelerating research and raising awareness so that every family and every child with SYNGAP1 can provide information that can guide us to a cure.

>>RESOURCE:
https://www.ncbi.nlm.nih.gov/pubmed/31454529
https://www.ncbi.nlm.nih.gov/pubmed/31395010;
https://www.ncbi.nlm.nih.gov/pubmed/30789692

CFC International, www.cfcsyndrome.org (1999) 501c3 NY

CFC International was founded in 1999 as the CFC Family Network by mom, Brenda Conger. After an exhaustive search for answers for her son, born in 1993, she found a group of 20 parents who interacted with one another over a listserv, and found one another through an ad in a magazine.

When the Conger's founded the organization, their goal was to create brochures, a website so new families could easily find answers, clinical guidelines, a Medical Advisory Board, and a family medical conference. By 2004, the organization had achieved all of its goals and set out to expand its reach by adding a community of parents to its board of directors.

With more than 100 diagnosed families, the organization decided in 2004 to change its name to CFC International to be better recognized as a global support organization. They also decided to boldly move forward to bank their own DNA through the jointly owned Genetic Alliance BioBank Program. This venture captured the attention of researchers from around the globe. CFC International partnered with a team of researchers led by Dr. Katherine Rauen at the UCSF Comprehensive Cancer Center. Two years' worth of clinical data and DNA storage paid off as the collaborative team efforts quickly located three of the genes responsible for CFC syndrome: BRAF, Map2k1 and Map2k2. Publication of the gene discovery occurred in one of the most highly visible formats in all of science and medicine in March 2006. Additional publications followed quickly, along with an NIH grant to host the first International Symposium on CFC and Noonan syndromes in November 2006.

By the end of the decade CFC International had hosted 5 family medical conferences and led the discovery of a 4th gene that causes CFC Syndrome, KRAS. The development of mouse models and new research led to the discovery of better treatment for cardiac complications, making children with the diagnosis more stable.

Brenda Conger continued to lead CFC International as the Executive Director until 2017 when she decided to retire. In 2018, Tuesdi Dyer took the helm as the first full-time executive director to lead the organization into greater strategic growth. With more than 500 registered families, CFC International is now expanding its conference across the globe, raising 50% more in revenue to expand its reach, and embarking on its first fully funded research on seizures at the University of Minnesota. Celebrating its 20th Anniversary, CFC International is working hard to meet the needs of families as we prepare to welcome an expected 2000 diagnosed individuals by 2030.

>>GUIDELINES:
https://rasopathiesnet.org/wp-content/uploads/2014/01/CFC-Parent-Guide_R4.pdf

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Chelsea's Hope chelseashope.org (2009) 501c3 CA

Chelsea's Hope was founded in 2007 by Linda Gerber, mother of Chelsea Gerber, along with 6 members of her synagogue. It's mission was to raise money for research and to support families. This was in the early days of internet connectivity and social media had not yet become mainstream. Chelsea's Hope enabled approximately 30 families to connect and support one another through an online "chat room".

>>RESOURCE:

https://www.valerion.com/advances-in-neurodegenerative-disease-research-and-therapy/
https://www.valerion.com/technology/lafora-disease/
https://www.cell.com/cell-metabolism/fulltext/S1550-4131(19)303754?_returnURL=https%3A%2F%2Flinkinghub.elsevier.com%2Fretrieve%2Fpii%2FS1550413119303754%3Fshowall%3Dtrue
https://www.aesnet.org/meetings_events/annual_meeting_abstracts/view/546387
http://www.jbc.org/content/293/19/7117
https://indiana.pure.elsevier.com/en/projects/lafora-epilepsy-basic-mechanisms-to-therapy
https://www.nature.com/articles/s41582-018-0057-0
https://www.jle.com/fr/revues/epd/e-docs/lafora_disease_308085/article.phtml?tab=texte
https://www.ionispharma.com/indications/lafora-disease/

Christianson Syndrome Association, www.csa-cares.org (2011) 501c3 TX

The Christianson Syndrome Association is a non-profit organization founded by Debbie Nash and her family in August 2011 in honor of her son, Andrew. Andrew was diagnosed with Christianson Syndrome in August of 2010. Soon after receiving the diagnosis, the Nash family realized how very little was known about this newly found genetic disorder. From that point forward, it has been the goal of Debbie and her family to support the research into the cause and treatment for this disorder as well as raise awareness both nationally and internationally. Once diagnosed families need to be able to talk and network with other families to know that they are not alone. CSA was born out of Debbie's desire to reach other families whose children are affected by CS and to unite them together as one family with the goal of raising awareness and to ultimately find a treatment to help our boys. CSA has organized 4 conferences since its inception, bringing families and doctors together to address the many issues of Christianson Syndrome.

2013- Brown University RI Inaugural CSA family meeting with researchers at Brown
2015- Houston Tx - 1st International Basic and clinical science conference on Christianson Syndrome
2017 -McGill University- 2nd International Basic and clinical conference on CS
2019- CS Europe conference

Chromosome 9pMinus Network, 9pminus.org 501c3 UT

The Chromosome 9p Minus Network is a continually growing nonprofit, parent-based support group with a mission to improve the lives of families affected by 9p Deletion Syndrome by connecting families, offering knowledge, and improving access to information about this rare genetic disorder.

CureGRIN Foundation www.curegrin.org (2019) 501c3 CO

CureGRIN was founded the same way many rare disease organizations are founded; by parent advocates searching for a cure. Many times, parents have to be the ones to lead the way, and that is exactly how CureGRIN came to be. Keith McArthur and Denise Rehner first met at a parent meetup in Colorado shortly after their children were diagnosed with GRIN Disorders in 2016. In 2018, they began working with other parents to put together an organization that would help drive GRIN Disorder research. We were blessed to have a group of doctors and researchers already studying GRIN Disorders, but there was not a US based Foundation that was focused on GRIN genes as a whole (as opposed to JUST focusing on GRIN1, GRIN2A, GRIN2B, GRIN2D)...Thus, CureGRIN was formed. We recognize that a cure for one GRIN gene will help the entire GRIN family.

CureSHANK CureSHANK.org (2019) 501c3 pending CA

It is the mission of CureSHANK to accelerate the development of cures and treatments for Shankopathies through research. The founders of CureSHANK are three parents of children affected by Phelan-McDermid Syndrome [a shankopathy], who believe that a better life for our children is within reach. CureSHANK works towards this goal through funding targeted research and holding scientific meetings to connect diverse stakeholders.

>>GUIDELINES:
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4362650/ This paper includes guidelines for monitoring. There are no approved treatments, though the practice parameters paper has some general recommendations about things like referral to speech, OT, ABA, etc.
Diagnosis: https://www.ncbi.nlm.nih.gov/books/NBK1198/

DDX3X Foundation www.ddx3x.org (2018) 501c3 DE

In 2014, DDX3X Syndrome was identified as a cause of intellectual disability when several gene sequencing labs around the world began to detect a mutation in the DDX3X gene in children exhibiting a similar phenotype. Shortly after, the two founders of the DDX3X Foundation, Beth Buccini, and Liz Berger were connected through their daughters' doctors, Elliot Sherr at UCSF and Kevin Strauss at the Clinic for Special Children. They convened the first gathering of DDX3X families in 2015.

Today there are approximately 500 known cases of DDX3X in the world and the rate of growth of identified cases is about 100% per year. It is expected that 1-3% of all intellectual disability in girls is explained by mutations in the DDX3X gene. This equates to an expected population of 20,000-40,000 individuals in the U.S. The DDX3X Foundation was established to connect families, researchers and medical professionals. Our mission is to facilitate continual research regarding DDX3X syndrome, outline best practices for treatment and management of this disease, and to improve the overall quality of life this impacted by this syndrome.
>>RESOURCE:

Snijders Blok, et al. "Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling," American Journal of Human Genetics. July 30, 2015. DOI: https://doi.org/10.1016/j.ajhg.2015.07.004

DNM1 dynamos - Connecting DNM1 Families https://www.facebook.com/groups/1065172710162616/ (2015) Non incorporated support group

This organization is a private Facebook Group founded August 2015. It was founded in order to connect families around the world whose children share a very specific diagnosis (a Mutation in Gene DNM1). DNM1 gene mutations renders children with severe refractory Epilepsy (Early Infantile Epileptic Encephalopathy Type 31 - EIEE31) and severe to profound global developmental delays.

Since its inception - the group has grown to include more than 60 families with children who have mutation in DNM1 gene. This growth has been through the DNM1 children's clinicians and researchers familiar with or working on DNM1 related topics.

The group acts as a support group to families and help share information with each other related to managing their children's epilepsy and everyday therapeutic needs, share information on the associated challenges and how to tackle them.

Additionally - the purpose of the organization is to promote research so that one day there will be a cure for DNM1 related Epileptic and Developmental Encephalopathy.

Doose Syndrome Epilepsy Alliance www.doosesyndrome.org (2002) 501c3 CO

Informal 2002 formed on yahoo group. In 2011 Doose families had their first gathering at the EFOA walk. There were 80 or so families. We had the largest team that year and raised the second-largest amount of money. This was very life changing for so many families as the vast majority did not have an opportunity to meet another family with their child's diagnosis. We had veteran families whose children were still seizing, but some who had gone into remission and live relatively "normal" childhoods and young adulthoods. We had one family that was diagnosed the week before who lived in the area. We decided we would like to formalize the organization with three core values in mind. 1. further research into the genetic marker of Doose. 2. fulfill grants for families needing things their insurance company would not cover. 3. Build a community

We were co-founded by two mothers (I am one of them). Shortly after the founding one of the mothers went through a family issue and stopped helping. Shortly after that the other founder started another nonprofit and DSEA was back burnered (stopped fundraising) but continued to provide an online gathering community for families to receive ongoing support. I have not been able to previously find the time to pick DSEA back up but leave it as a nonprofit for when we can, or there is interest in the group from a parent or two to take the reins.

All in all we are a very small organization, not well funded, and somewhat dormant.

Successes: we have a wonderful online community that is extremely supportive with very little discourse, I can think of a handful of times since 2009 when first forming our yahoo group (now we are on FB). We did get two genetic studies off the ground, one with Manton Center for Orphan Disease Research. One with Kings College in London. We also have fulfilled all grant requests that have come in through the years, even without fundraising, I have made those personally.

Challenges: Parents aren't able to do much more than they are doing to take care of their kids, or do not have the skillset or the desire. We stopped fundraising. distance. Not meeting regularly.

Dravet Syndrome Foundation https://www.dravetfoundation.org (2009) 501c3 CT

Our organization was founded by 4 parents of children with Dravet syndrome who were frustrated by the lack of research in the field and lack of understanding of the syndrome. When they looked at epilepsy research funding in general, they found that it was generally underfunded and by the time you funneled down to a rare epilepsy like Dravet syndrome it was non-existent. It was at that point that we recognized if things were going to change, it needed to be driven by affected families.

>>GUIDELINES:
https://www.pedneur.com/article/S0887-8994(16)31037-2/fulltext

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Dup15q Alliance www.dup15q.org (2004) 501c3 OR

Dup15q Alliance is a nonprofit 501(c)(3) corporation. Originally founded under the name of IsoDicentric 15 Exchange, Advocacy and Support in 1994 by Donna Bennett, mother to Joshua (a young man with idic(15)) and Brenda Finucane, MS, LGC, who was then the Director of Genetic Services at Elwyn Inc., as a list of 13 families raising children with chromosome 15qduplications, the support group grew and in 2004 it was officially incorporated into a nonprofit organization. The organization's name was changed in 2011 to Dup15q Alliance to encompass both interstitial and isodicentric variations of the disorder known as chromosome 15q11.2-13.1 duplication (dup15q) syndrome. Today, there are almost 1,800 families

from around the world affiliated with Dup15q Alliance. The organization is a mostly volunteer run organization supported by grants, donations and many hours of volunteer effort.

FamilieSCN2A Foundation www.scn2a.org (2014) 501c3 MA

Ben was born healthy and gorgeous with a full head of hair. His first year was blissful. Being my first child, I enjoyed every second of being a Mom. Ben made it easy too. He was so happy, sweet and easy going.

On November 7, 2011, he had his first seizure, followed by hundreds more, all on the same day. The next day, he was diagnosed with Infantile Spasms, a catastrophic form of epilepsy. Our world stopped. Ben became very ill very quickly. He went from a walking, babbling toddler to a newborn baby again. He lost every milestone he had gained in his first year. After failing 7 anti-epileptic drugs and the ketogenic diet, the doctor told us that there were few other options for us, and that Ben would always have seizures and would need a lifetime of care. So, we did what any other parents would do, we got another doctor.

Ben's new doctor told us there would always be another option and showed me how to be an active participant in his care. Not only were we able to stop the seizures, we also learned the cause was a genetic change to the SCN2A gene.

S - C - N - 2 - A - what does that mean?? Nothing was known at the time, no research, no treatments, no other patients to connect with. We were told he is the only one. This knocked us back down to the ground, but hey, we'd been down before and we now knew how to rise, so rise we did.

We started the first SCN2A online support group in 2013 to try to find another family, and we did, then another, and another. We grew to 50 families in under a year and each one that joined the group said the same thing, "we thought we were the only ones." Carla Forbes, the 6th family to join the online community, and I took the initiative to start the FamilieSCN2A Foundation to change the wish to a plan. We received our 501(c)3 in 2014 and we have been successful at getting SCN2A on the map. We support the Global research community in cutting edge transnational research that is leading towards new treatments for SCN2A Disorders.

We do not know what a cure will look like for our children, however we know we will see one soon. If we can watch just one child, one baby, saved from this disease, we will know we made an impact. Carla and I are not trying to change the world, but we are trying to change our world.

Glut1 Deficiency Foundation https://www.g1dfoundation.org (2011) 501c3 IND

The Glut1 Deficiency Foundation was formed originally in 2009 - born of a desire for Glut1 families to get together to meet, share, and learn from one another. Glut1 Deficiency is such a rare diagnosis that many families, and especially our Glut1 children, have never had the experience of meeting others who share this journey.

The Yahoo Health Group for Glut1 DS was our original meeting place, and as our connections with and dependence upon each other deepened and strengthened, we started talking about how good and helpful it would be if we could meet in person. A group of Glut1 Deficiency families in Australia had a gathering a few years earlier, so this was the inspiration for us to do the same. A German Glut1 parent group (www.glut1.de) also has regular family get-togethers, and we very much wanted to have a similar experience.

Jen Lazar, mom to Sam, had the courage to take on the task of organizing our first family meeting in Chicago, which quickly grew into a full-fledged conference as world-renowned Glut1 Deficiency researchers, doctors, and dietitians were eager to join us. The knowledge, experiences, and emotions shared there certainly impressed upon us the need to continue having conferences to be able to have time with other families and to continue to learn about the work being done by the specialists on behalf of our children. As relationships developed with these Glut1 experts, we soon learned that there were very few sets of eyes and very few dollars focused on researching Glut1 Deficiency and we wanted to help change that.

As our second conference rolled around in 2010, hosted by the Steele family (Macie), we were well on our way to building a formal organization (originally naming ourselves glut1ds.org) and trying to make a difference for our children. The Meyers family (Katie) spearheaded a $25,000 fundraising effort that was presented at the conference for a clinical trial at UT Southwestern for the use of C7 oil (triheptanoin) as an alternative treatment for Glut1 Deficiency. The Louisville conference also gave us an opportunity to meet with experts and other interested families to begin to plan the process for becoming a formal non-profit group. A leadership board began to emerge in early 2011, and the groundwork began to be laid for what became called the Glut1 Deficiency Foundation. We received our 501 (c)(3) designation from the IRS in July of 2011.

Our third conference was held in New Orleans in 2011 and hosted by the Meyers family. Board members had the opportunity there to meet with our esteemed Glut1 Deficiency experts and work on setting goals and developing plans for future projects. We have continued to grow our service programs and our impact, have expanded our Board of Directors, and have added both a Medical Advisory Board and Scientific Advisory Board to help guide our endeavors.

With so little government funding available for rare diseases, we know that the future of research, advancements, diagnosis, and improvements in treatments and quality of life for Glut1 Deficiency patients rests largely in the hands of families. We know that we can do so much more together than any of us can do alone, and there are many projects, both large and small, where we can help move things forward. We are already hard at work trying to help all people, present and future, with Glut1 Deficiency, and we hope to one day be able to help bring about the ultimate treatment - a cure.

>> RESOURCE:
Glut1 Deficiency Foundation, Review: https://www.ncbi.nlm.nih.gov/books/NBK1430/
>>GUIDELINES:
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5983110/

GRIN2B Foundation www.grin2b.com (2017) 501c3 ILL

When our daughter, Lucy, was diagnosed with GRIN2B-Related Neurodevelopmental Disorder in November of 2014, there was no organization, support system or network of families anywhere in the world. At the time of Lucy's diagnosis, the medical community knew of only 10 cases of GRIN2B mutations worldwide.

Our family was devastated and felt alone without any sort of family support system or structure. Slowly, through social media, we discovered other families around the United States and around the world. We were the second family to join the GRIN2B Family Support Group on Facebook. Today this group contains several hundred members from all around the world. In 2016, my wife, Liz Marfia-Ash began writing www.grin2b.com, because she was determined to create a place for newly diagnosed families on the Internet. The success of the website led Liz to found GRIN2BFoundation in 2017.

GRIN2B Foundation is a parent-run organization dedicated to furthering research on the GRIN2B gene and providing support and education to the small, but growing community of individuals and families impacted by a GRIN2B diagnosis.

Hope4Harper www.hope4harper.com (2012) 501c3 TX

Hope4Harper was founded after our youngest daughter, Harper Howard, who was born with CDKL5 Deficiency Disorder (CDD). CDKL5 is a rare, life-threatening, genetic disorder that carries with it a multitude of debilitating symptoms including uncontrollable seizures. We were desperate to find a way to improve Harper's quality of life with CDKL5 by stopping symptoms such as seizures associated with the disorder.

In 2011, my mother-in-law saw Dr. Francis Jensen, former 2012 president on 60 minutes discussing seizures in infants and suggested I reach out. At that time Dr. Jensen was still seeing patients as well as overseeing research. I was put on a waiting list and a spot opened in two weeks. So I flew from Texas with a binder full of medical records and 4 month old Harper for a visit. After a multitude of tests and an extensive evaluation, we were sent home to wait. Four months later, the results came in.....Harper had CDKL5 disorder and nothing could be done! At that point we asked Dr. Jensen if her lab could do research and find a way to stop Harper's seizures, thus the birth of the organization, Hope4Harper began and our connection to AES was made.

>>RESOURCES:
https://www.jneurosci.org/content/early/2019/04/05/JNEUROSCI.2041-18.2019/tab-article-info?versioned=true
https://www.louloufoundation.org/announcements.html

International Foundation for CDKL5 Research https://www.cdkl5.com (2009) 501c3 OH

In 2004 CDKL5 was discovered as disease-causing by Dr. John Christodoulou and Dr. Vera Kalscheuer. In separate case-studies, they each link CDKL5 genetic mutations to a phenotype that overlapped with Rett Syndrome. The International Foundation for CDKL5 Research began in late 2008 as a group of seven parents whose children had CDKL5 Deficiency Disorder. A small Yahoo group brought us together as we dared to dream of a new future for our children. We found some initial research available in the Rett Syndrome community and Rettsyndrome.org (IRSF at the time) graciously mentored us in our founding years. With education and research, we believe a life-changing cure will be found. In 2009, we incorporated the International Foundation for CDKL5 Research as a non-profit organization. Since then, we have gone on to fund ground-breaking research and establish CDKL5 Centers of Excellence across the United States.

>>RESOURCE:
https://rareomics.healx.io/disease/cdkl5-disorder

Jeavons Syndrome Facebook Group https://www.facebook.com/Jeavons-Syndrome-260411829399/ Founded 2008

I started the Jeavons Facebook site 11 years ago. The reason I started it was due to the Mayo Clinic diagnosis that my then 13 year old daughter received. It was very distressing to hear that there was no cure for this life-long disease. Even more frustrating was the fact that there was very little information available on the web. Lastly, there was no one I could find that had the disease. I felt alone! The Facebook site was born out of this situation.

Since its inception, the site has reached people around the world. It has become a place to share information, experiences and provide support. It has been a comfort to many of the people that had felt just like me. They come to the site and gain the knowledge and confidence of how to deal with Jeavons but most importantly know that they are not alone.

KCNMA1 Channelopathy International Advocacy Foundation (KCIAF) kciaf.org (2019)501c3 NY

KCNMA1-linked channelopathy is only defined by mutations in the KCNMA1 gene that are associated with seizure and/or movement disorder. A neurologist treating a patient (Dr. Sotirios Keros) met a basic scientist/researcher (Dr. Andrea Meredith) as a result of the patient being profiled on Netflix. Knowing that the debut of the episode would identify more patients, we co-founded the advocacy group to serve as a connection platform and repository of information while the defining attributes of the disorder are being established. We are struggling to get parents of patients involved, as most don't have the time and resources to help us with KCIAF organizational endeavors.

>> RESOURCE:
http://jgp.rupress.org/content/jgp/151/10/1173.full.pdf?with-ds=yes

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KIF1A.ORG kif1a.org (2017) 501c3 NY

In August 2016, our co- founders Luke Rosen and Sally Jackson's two-year-old daughter, Susannah, was diagnosed with a rare, neurodegenerative disease called KIF1A Associated Neurological Disorder (KAND). There is no cure or treatment for this condition, and the lack of information and resources available to KAND families left them desperate to find treatment for Susannah. KIF1A.ORG was created to help other families affected by KAND and fund research for a cure. We received our 501(c)(3) nonprofit status in March 2017.

FIRST FAMILY MEETING
Initially, all communication amongst families affected by KAND were powered through our private virtual support group. We still use the group to stay up to date, ask questions, and share stories and advice, but we took it a step further. With help from the team at Columbia University Medical Center, 10 families came together to share information and create an urgent plan to find treatment for our children. We had our first family meeting in April 2017. Families were able to meet and connect with researchers studying the disorder in person to learn more about KIF1A, ask questions and share their experiences. Through the family meeting, we developed our vision: care until the cure. KIF1A.ORG's priorities are: Expanding a global community of families, and engaging the scientific community to inspire new research Growing the KAND Natural History Study a systematic collection and analysis of comprehensive, longitudinal data collected from families diagnosed with KAND Securing mouse models and cell lines to test potential therapeutics Developing tools and a treatment development process to efficiently partner with pharmaceutical and biotech companies Raising funds to accelerate research for treatment and a cure.

RESEARCH PARTNERS Wendy Chung, MD, PhD and her team from Chung Lab at Columbia University have been our primary research partners from the very beginning. Dr. Chung is a leader in rare disease research, and families with nowhere else to turn are often referred to her seeking care and an end to the diagnostic odyssey. Such was the case with Susannah Rosen. After meeting the Rosen family and understanding the progressive nature of this disease, Chung and her team urgently set out to find treatment and a cure for KAND. Other vital members of our core research team include The Jackson Laboratory and Berger Lab at University of Vermont. Today, KIF1A.ORG fosters patient-driven collaboration with a growing network of researchers and innovators around the world dedicated to KIF1A and related disease areas.

LGS Foundation https://www.lgsfoundation.org (2008) 501c3 NY

The LGS Foundation is a non-profit organization dedicated to providing information about Lennox-Gastaut Syndrome while raising funds for research, services and support for individuals living with LGS and their families.

The LGS Foundation is based in New York and provides services and information to thousands of members across the world.

Lissencephaly Foundation Inc http://lissfoundation.org/ (2019) 501c3 Pending CA

Empowering Families & Educating Communities by hosting awareness events & Regional gatherings. Spreading awareness for Lissencephaly families to create a more accepting world around us.

>>RESOURCE:
https://www.ninds.nih.gov/Disorders/All-Disorders/Lissencephaly-Information-Page

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Mickie's Miracles www.mickiesmiracles.org (2016**)** 501c3 CA
Thank you for the opportunity to share about our family's journey with pediatric epilepsy and the organization I launched to support warrior families still in the fight to save their baby's life. Mickie's Miracles allowed me to take the life experience that almost broke us and turn it into an organization committed to helping families obtain a proper diagnosis and effective treatment. Ultimately, early intervention preserves the baby's brain and gives the child the best chance at their highest quality of life.

The journey to save our newborn daughter's life from pediatric epilepsy was the scariest and loneliest experience of my life. When she was three months old, I will never forget watching her eyes roll back into her head and her body becoming rigid and trembling as her first seizure took hold. I did not know the battle we would be facing in the months and years to come. I watched in terror as Mickie failed eight anti-seizure medications in eight months.
By the grace of God, a family friend connected us to CHOC Children's Hospital in Orange County, CA where Mickie would be evaluated for brain surgery. The day before Mickie's first birthday, a surgeon removed 25% of her brain to stop her seizures. We were told Mickie would never walk or talk, that she may live in a vegetative state or die from her disease. She turns eight this fall and is thriving in swim lessons, dance class and is mainstreamed with an aide in her second-grade class. She is a tenacious, talented, driven, lady and the definition of a miracle.

Our experience inspired me to start Mickie's Miracles over three years ago. Our mission is to create global pediatric awareness, education and advocacy. We have helped more than 500 children around the world access critical epileptic care. Most families have no idea how devastating epilepsy is to the developing brain of a child and that it is more vulnerable than a fully developed brain. This drives our urgency to educate families and help them on their journey to get the proper diagnosis and treatment of their specific seizure disorder. Swift action determines a child's quality of life. Time is neurons!

As the founder of Mickie's Miracles, I dream of a day where there are no barriers to care for babies and children suffering from Pediatric Epilepsy. Our vision was seeded in the early days by our own battle to save Mickie's life. Now every day is a battle to raise the global consciousness about epilepsy--to bring it out of the darkness and into the light where we can change attitude, shape policy and continue our mission.

>>GUIDELINES:
https://www.uptodate.com/contents/clinical-features-and-diagnosis-of-infantile-spasms

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NORSE Institute http://www.norseinstitute.org **(2015) Non incorporated Support or other Group**

When my son died on December 3, 2013, there was no explanation for his death. One doctor handed me an article on NORSE without giving any further explanation shortly before she left his medical team. Another doctor would not confirm this diagnosis. The head of Neurology in my local NJ hospital, Harvard Medical School, Mass General, had never heard of the term NORSE but said "the entity is one we all see from time to time." Not until several months after his death did one of his physicians indirectly confirm this diagnosis by referring me to two doctors Drs Larry Hirsch and Nicolas Gaspard who were doing research on patients like my son, a study that included NORSE.

I met with Drs Hirsch and Gaspard, learned they were proposing a larger study of refractory status epilepticus which included NORSE patients. I told them I wanted to learn more about and support NORSE research in particular. My husband and I are supporting the only multi-center, prospective, observational study of NORSE patients collecting clinical data and bio specimens. Hirsch, Gaspard and I kept up conversation. We asked other epilepsy and critical care doctors to join in our conference calls. They became my medical advisory board with Hirsch and Gaspard as my co-chairs. I created the NORSE Institute in 2015 as the platform to integrate the medical and patient/family perspective.

We are biased towards the medical perspective because I have the most sustained interaction with doctors and researchers. Because of the sudden, catastrophic nature of the syndrome, families are often left in shock, bereaved or overwhelmed with caregiving responsibilities. There is little time, energy or money to get involved with our organization. Most importantly, the term NORSE is new. Not all physicians are aware of the term so they do not inform their patients/families they have had it. Consequently, families don't know to look for the NORSE Institute.

>> RESOURCE:
Most extensive explanation (and largest series study of NORSE https://www.ncbi.nlm.nih.gov/pubmed/26296517
Recent review of integrated NORSE and FIRES literature https://www.ncbi.nlm.nih.gov/pubmed/30482654
Proposed consensus definitions https://www.ncbi.nlm.nih.gov/pubmed/29399791
State of the art: https://www.ncbi.nlm.nih.gov/pubmed/29476535

Phelan-McDermid Syndrome Foundation https://www.pmsf.org/ (2013) 501c3 FL

Established in 2002, the Phelan-McDermid Syndrome Foundation is the leading source worldwide for information about the rare genetic condition. But it was events more than 10 years before that which set us on the course, we're on today. We offer support to families around the world, advocate for our cause and encourage research that one day may lead to therapies or effective drugs to treat those with PMS. A look back at how we got to where we are today: https://www.pmsf.org/pmsf-history/

>>RESOURCE:
Gene Cards: https://www.genecards.org/cgi-bin/carddisp.pl?gene=SHANK3
GeneReviews - https://www.ncbi.nlm.nih.gov/books/NBK1198/

Project 8p www.project8p.org (2019) 501c3 DE

I founded the organization because of my daughter. Here is my story:
My daughter beams with light and gives hugs to everyone around her, from a stranger to her brother, so long as she senses positivity. Even all of her specialists (over a dozen!) receive a huge embrace with a cheek to-cheek-smile from her. She is smart, and she captivates those around her.

She was born with what appeared to be healthy, except for a rare genetic mutation in the 8th chromosome. Some of her genes are missing, some are duplicated, and some are flipped around. And nobody has a clue as to how this happened.

When we first received the Chromosome 8p diagnosis, we were told, -Good luck, she's not going to Harvard. You should seek parent counseling.,- As you can imagine, there were endless tears that emptied the tissue box on the car ride home. And after. The 1st brochure we received from a genetics counselor came with the caveat that it was 30 years old. This diagnosis was so rare that they couldn't tell me what her symptoms would be, all they could do was offer a range. The range was so broad, it spanned from minor global delays to wheelchair bound and questionable health. I was utterly scared and confused. All I kept thinking was- That's all you can tell me?

That's when I learned there was no central database for Chromosome 8p disorders or genetic conditions in this country, never mind the world. Months went by after the diagnosis with no info. Then I started scouring the internet. I found a couple of organizations and some basic databases. I started contacting any families I could find that were impacted by Chromosome 8.

Along the way, we decided to see our daughter for who she is and not what a brochure told us she could be. I have learned to forget about milestones and value the inch stones at her own pace, not mine.

When I hear my child's voice with a faint "hhhiii," a dozen things go through my mind: my heart melts, I wonder how hard that must have been, I imagine neurons in her brain trying to send a message to the facial muscles, misfiring, and eventually her tongue moving and her breath producing that, "hhhiii,". I am aware that it took 2 minutes for that motor plan, I am upset that it took that long, I am proud of her, I have tears of happiness and sadness. I can't imagine how frustrating it must be when she has so many ideas to express and cannot, she wants to run alongside friends in the playground and cannot, and she wants to shout that she's hungry and exhausted after a full day and cannot.

But she can say hi, even if it took around 5 years and approximately 8,000 hours of all kinds of therapies. Could our journey to ,"hhhiii" help other families? Could all of our collective experiences help each other if we only had a central repository of knowledge?

I am motivated to make sure my daughter can do everything she wants to do. That means coming together to find treatment. To help prevent others from being born with a genetic condition like this. To provide resources and a hand to hold for new families so that nobody needs to start from scratch and navigate the unknown like we did. To find a way to help the 8p heroes continue persevering with their beautiful smiles.

With Project 8p, I want to represent my daughter's voice and its truth. I am her spokesperson, and one day, she will be the spokesperson for this foundation. I carefully listen to her communication cues, often it's a wide eyed stare that sees right through my soul. I have full conviction that she is my teacher and has given me a purpose that is beyond our little family. As I say this out loud to her, she claps and reaches her arms out to embrace me.

So you see, I almost need her more than she will ever need me. I hope when you read this, you can relate somehow. She is my inspiration to find a better, smarter way to respectfully fulfill her dreams. And there are infinite possibilities.

PVNH Support & Awareness www.PVNHSupport.com (2009) Non Incorporated Support or Other

PVNH Support & Awareness was created out of necessity, after Yolaine Dupont's daughter Ella passed away while still undiagnosed in 2009. She was 7 month and 20 days. Back then, even though PVNH was already identified on an MRI, there were no information available, except a few academic papers published, and because her symptoms did not match what the medical community knew of PVNH (periventricular nodular heterotopia) at the time, which was very little, PVNH was not believed to be the cause of her deadly lung disease. It was only because Yolaine pushed for answers that an autopsy was performed which then pointed to X-linked PVNH as a possible culprit, despite the fact that some doctors believed it was because of something else entirely. Once the report was received, Yolaine pushed for genetic testing, to have closure but also because she wanted to give a brother or sister to Ella and wanted to avoid having other families face a similar fate. After two refusals by her provincial health services, Yolaine was finally told by her geneticist who had pursued the quest of testing with her, that payment for testing was approved. On 1st October 2009, Yolaine was told was formally given a DNA diagnosis of X-Linked PVNH. She did not stop there and kept digging in her own medical history. Six days later, she discovered her mom had a PVNH diagnosis that had been made 13 years prior to Ella being born, but no one had explained to her mom. Further testing confirmed what Yolaine already knew, that the disorder that took Ella's life was also what had afflicted her health all throughout her life. In was in the Fall of 2019 that PVNH Support & Awareness was created: to provide support to families with a PVNH diagnosis, and to educate the medical professionals and other groups of interest. Yolaine, to this day, is the sole volunteer and champion of everything PVNH. She has also added SBH (Subcortical band heterotopia) to the families she represents and supports. PVNH Support & Awareness now supports more than 5000 families in 33 countries and also fundraises for research, hosts World PVNH Day and World PVNH Month, conferences, and participates in research. PVNH is a complex disorder which can affect multiple organs in addition to causing epilepsy in most patients and can be mild to very severe in presentation. Yolaine has been active in social media and on forums and families find her and the organization weekly.

>>RESOURCE:
https://www.ncbi.nlm.nih.gov/books/NBK1213/

RASopathiesNet
Connect ~ Collaborate ~ Cure

RASopathies Network Https://RASopathiesNet.org (2010)501c3 CA

As the parent of a child with Costello syndrome whose son died of cancer related to the syndrome, I had accumulated a lot of information and learned I enjoyed sharing what I learned either from the families with the clinicians/researchers and what I learned from the clinicians/researchers with the families. I became president of CSFN (Costello Syndrome Family Network) for two terms 2003-2007, and then Board Secretary until 2017. In 2006, with the first cardiofaciocutaneous (CFC) syndrome mutations identified as the 'third' datapoint (PTPN11, identified in 2001 which ~50% of individuals w/Noonan syndrome have; HRAS, identified in 2005 preceding the CFC syndrome mutations find), Dr. Katherine A. Rauen connected them molecularly by identifying their relationship: all are mutations along the RASopathies mitogen activated protein kinase (MAPK) pathway. It was she who coined the term, RASopathies, to encompass CS, CFC, Noonan syndrome (NS) and neurofibromatosis type 1 (NF1) syndromes - in large part to define them molecularly, as well as frame a strategy for therapeutic intervention, given that 1/3 of all malignant tumors also have mutations along the same pathway - though within the tumor (somatically) and not germline, and all the syndromes are affected by their mutations in a gain-of-function way, so the possibility of using targeted chemo (at reduced doses) could slow the signaling instead of silencing the signaling to kill the malignancy). The name was formalized by a vote - the first order of business prior to setting up the RASopathies Network, in 2009.

After attending numerous workshops hosted by NORD and the Genetic Alliance on encouraging researchers to hold NIH R13-funded scientific meetings to jumpstart rare disease research, I collaborated with Dr. Rauen on the First International Costello syndrome symposium in 2007 (her idea), in conjunction with the 2007 biennial CS Family Conference.

For 2009, Dr. Rauen and I continued pairing the research meeting with and Costello Syndrome Family Network conference, and expanding the concurrent events to include the CFC International and The Noonan Syndrome Family Support Group family conferences - and invited NF Network and the Children's Tumor Foundation as well. After that meeting, the leadership from the family organizations of CSFN, CFC International and The Noonan Syndrome Support Group suggested that I (with my

Masters in Fine Arts degree), as the research maven, start a parent-led research network; and when the researchers agreed that they needed an organization like the Children's Tumor Foundation to keep them focused, I stepped to the plate to set up and run a non-profit RASopathies networking organization, which was formalized in October, 2010.

>>RESOURCE:
* Giannoulatou et al. https://www.pnas.org/content/110/50/20152 2013

Ring Chromosome 20 Alliance https://www.facebook.com/pg/RC20Alliance/about/?ref=page_internal (2015) 501c3 PA

We began as 2 mothers who joined together out of desperation to support each other and better understand Ring Chromosome 20 mutation. Out of our collaboration and communication with other RC20 families in the US and internationally, we saw the need to form a non-profit group to advance awareness of RC20 among professionals and the epilepsy community, support RC20 families, promote research and seek treatment of Ring20.

>> RESOURCE:
Rinaldi et al. Orphanet Journal of Rare Diseases (2017) 12:69 DOI 10.1186/s13023-017-0606-4

Ring14 USA www.ring14usa.org (2011) 501c3 CA

Ring14 USA, Yssa DeWoody's Perspective. Ring14 USA was co-founded by Terri Granard and Yssa DeWoody in October of 2011. We found each other through the internet. Terri's husband had built a beautiful website devoted to his son Eli and I happened to stumble upon it in yet another midnight search for answers. I reached out to Terri and we soon discovered that both of us were pursuing the idea of founding a non-profit to support families living with the ultra-rare disorder, Ring14 Syndrome.

I had already acquired first-hand knowledge of how powerful a patient advocacy organization could be. In fact, my daughter Marie, who was diagnosed with Ring14 Syndrome at 3 months shortly after her seizures began, was just 18 months old in October of 2006 when Ring14 Italy (then called the Ring14 Association) held its first Ring14 International conference for both scientists and families. We were quite fortunate to have found this Italian organization, which was founded by Stefania Azzali in 2002, and even more fortunate to have been able to attend their first international conference. The connection to this tiny group of families was immediate and profound despite the language barriers. If I recall right, there were only about 20 - 30 families from around the world that were able to attend (~10 Italian families, ~5 from USA , ~3 from France, ~3 from UK, and ~5 from several other countries). This was our first experience with the rare disease community.

My family continued to be involved with the Italian organization by donating funds, going to conferences, and attending family camps, but something shifted in 2011. After attending their first research workshop, after hearing about the science that needed to be funded in order to begin to find effective therapies, we knew we must do more than just be participants. My husband, Andrew, joined their Scientific Advisory Board and I began to plan founding Ring14 USA. Not only did I wish to help raise the money to fund the research, but I also wanted to help bring the support programs of Italy to the United States. The Italian association had proven that a small group of moms really could do big things, promote research and change lives - it was time to replicate this in the United States.

It was right after coming back from this workshop that I found Terri. We were well aligned with the common desire to fund research, raise awareness, and provide community to the families of North American. It only made sense to join forces! We quickly found 4 other mothers who shared our vision and our first board of 6 moms was born - we lived in CA, IN, OK, PA, FL and MD. It would actually be 3 years before we all met face to face.

>>RESOURCE:
Bibliography: http://ring14usa.com/index.php/research/bibliography/
>>GUIDELINES:
Article published in 2017 here: https://ojrd.biomedcentral.com/articles/10.1186/s13023-017-0606-4

Shay Emma Hammer Research Foundation shaysgift.org 501c3 AZ

The Shay Emma Hammer Research Foundation is committed to improving the quality of life of children's suffering from epilepsy and other brain disorders by supporting scientific research to improve our understanding of the fundamental processes that regulate brain function and to discover new treatments.

>>RESOURCE: scn8a.net

SLC6A1 Connect. https://slc6a1connect.org (2018) 501c3 CO

My 15 month-old son was diagnosed with the SLC6A1 mutation. I left my career in equity analysis to find a path forward to develop a treatment for this disorder.

SYNGAP RESEARCH FUND
Collaboration. Transparency. Urgency.

SynGAP Research Fund https://www.syngapresearchfund.com (2018) 501c3 CA
Letter from Mike and Ashley Graglia - Founders of Syngap Research Fund:

Tony was four when we received the diagnosis that the cause of his persistent and otherwise puzzling global developmental delays and epilepsy was genetic, a de novo mutation on one of his 6,000,000,000 base pairs, which causes his brain to produce only half of the required syngap, a crucial protein for learning and memory. Tony's SynGAP-1 diagnosis was crushing, of course , we knew then that there was not a cure for his condition and that he would not outgrow it; in fact, as he continued to build his brain on a faulty foundation, the gap between him and his peers would only grow.

Our SynGAP-1 diagnosis, though, gave us direction and a way to hope. By joining a community of parents, we found that there are over 400 other families whose children suffered in a similar way. By engaging with leading neuroscientists and researchers, we found that the SynGAP-1 gene is well characterized, that there have been treatments for haplo-insufficiencies like SynGAP-1, and that there are other genetic epilepsies with potential treatments in clinical trials.

My husband and I founded the SynGAP Research Fund (SRF), then, because we believed it would help us to fulfill our promise to Tony. Our mission is, through research, to identify the treatments and the therapies that will enable patients with SynGAP-1 to live their best lives. We are seeking to raise $3-5m in funds to support research focused on gene therapies for our patients.

The values of the SynGAP Research Fund are collaboration, transparency, and urgency. We ask the researchers we fund and our volunteer team who work with us to live these values as we together seek a cure.

Collaboration: The over 400 diagnosed patients in the SynGAP community share a painful bond, and we will help none of them if we do not work together. The SynGAP Research Fund is a member of the SynGAP Global Network, whose organizations in Australia, England, Germany, Spain, France, and Canada knit together our community and the researchers supporting it. The rare disease community includes 30m patients in the US alone. 80% of rare disease is due to faulty genes and as such may benefit from this research. As such, we have also actively sought out the insight and experience of other patient communities such as the Dravet Syndrome Foundation, EBRP, the Grace Science Foundation, as well as organizations such as Global Genes.

Transparency: We are a non-profit, incorporated in the State of California, and we have made our financials, our by-laws, and our scientific advisors, as well as all of the projects we fund, public on our website. We have asked our researchers to provide regular updates on their progress, which we will also share.

Urgency: Our children's brains are being built. There is no time for delay. We are all volunteers and we are interested only in discovering, as quickly as possible, what can be done to help our children, and then doing it.

Tony, and all children with SynGAP-1, wake up every morning and find their way through the day, with insufficient amounts of a critical protein, with a steady increase in suffering, and without any real understanding of why.

We are determined to be, not only witnesses of our child's journey, but also his guides. We seek a path for him that is as fulfilling as possible. We founded the SynGAP Research Fund because it is our belief and our hope that, with our help, his path may include a cure for his disease.

>>RESOURCE:
http://epilepsygenetics.net/syngap1-this-is-what-you-need-to-know/

TBC1D24 Spectrum Family Network

My daughter, Amy was diagnosed in 2015 after a 38 year search for a definitive diagnosis one of the diagnostic doctors asked me to start a website for the families of children with a TBC1D24 diagnosis. I happily complied with his wish. We are not a formal Foundation and are not Non-Profit as yet.

TESS RESEARCH FOUNDATION
Treatments for Epilepsy & Symptoms of SLC13A5

TESS Research Foundation tessresearch.org (2015) 501c3 CA

Kim and Zach Nye live in California with their four young children. Two of their children, Tessa and Colton, recently received SLC13A5 Deficiency diagnoses. Despite uneventful pregnancies and deliveries, Tessa and Colton both began having seizures the day they were born. For a decade, the underlying cause of Tessa's seizures and developmental problems remained unknown, despite consulting dozens of specialists across the country and trying dozens of medications. Tessa continues to have hundreds of seizures daily. When Colton was born in 2013 and also began having seizures, our team of experts realized the problem was genetic and were able to isolate a genetic marker for the disease: SLC13A5. Finding a genetic marker is a huge step in finding a cure.

The Nye family started TESS Research Foundation in order to improve the lives of those living with the genetic neurological disorder SLC13A5. TESS Research Foundation aims to raise awareness, unite families affected by the disease, and fund research in order to better understand this disease.

>>RESOURCE:
https://www.cell.com/ajhg/fulltext/S0002-9297(14)00268-7

The Bow Foundation

The Bow Foundation https://gnao1.org/ (2017) 501c3 VA

The Bow Foundation is dedicated to supporting GNAO1 families, research and awareness.

The Foundation was launched in 2017 by parents of children with GNAO1 disorders. Headquartered in the United States, the Foundation is recognized by the IRS as a nonprofit charitable organization.

Our vision is to build a better tomorrow for GNAO1 patients and their families by fundraising to support medical research that leads to a more informed GNAO1 body of knowledge, better patient treatment options and an eventual cure.

The Brain Recovery Project: Childhood Epilepsy Surgery Foundation
https://www.brainrecoveryproject.org (2011) 501c3 CA

The Brain Recovery Project was formed in May of 2011 by Brad and Monika Jones. Brad and Monika's son Henry has undergone three epilepsy surgeries , modified lateral hemispherectomy, revision, and then anatomical hemispherectomy , for intractable epilepsy, including infantile spasms, caused by total syndromic hemimegaloencephaly.

After his hemispherectomy at the age of three months old, Brad and Monika grew increasingly frustrated by the lack of information in the medical and therapy community about how to best rehabilitate their child. Would he learn to walk? Would he talk? How would they get him to reach his full potential? When no clear answers were provided, they decided to form this non-profit organization.

Since our inception in 2011, our primary focus has been funding research to better understand functional outcomes after hemispherectomy and how to improve those outcomes. In November of 2016, our board of directors approved our new initiative: to expand our research to better understand functional outcomes after pediatric epilepsy surgery , and how to improve them ,ì especially large resective procedures like hemispherectomy, hemispherectomy, lobectomy, temporoparietaloccipital disconnection, and corpus callosotomy, and to provide resources to families and children affected by these procedures. Because functional outcomes are tied to timely referral to surgical evaluation, we now help parents understand when they should be referred, the various surgeries offered, and their functional implications. We also offer programs post-operatively for the children.

The Brain Recovery Project received its non-profit status from the Internal Revenue Service in August of 2011. We are a 501(c)(3) not-for-profit corporation.

>>GUIDELINES:
https://onlinelibrary.wiley.com/doi/full/10.1111/j.1528-1167.2006.00569.x

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The Cute Syndrome www.thecutesyndrome.com (2014) 501c3 MD

TCSF was founding in 2013 when our founder, Hillary Savoie, was looking for answers and raising money for her daughters then diagnosis PCDH19. One year later Esme was diagnosed with SCN8A and Hillary connected with another parent Juliann Bradish who had a Facebook Support Group for parents with children with SCN8A EE. Esme's phenotype was more like the SCN8A children and Hillary quickly found a home amongst the SCN8A community. Despite Esme being diagnosed with more genetic mutations Hillary and TCSF remain dedicated to the SCN8A community. The lack of SCN8A awareness and research still drives us to fund research and empower families with knowledge about the devastating effects of living with SCN8A EE.

>>RESOURCE:
Influence of age at seizure onset on the acquisition of neurodevelopmental skills in an SCN8A cohort Alejandra C. Encinas1 | Ida (Ki) M. Moore2 | Joseph C. Watkins1,3 | Michael F. Hammer1,4
The spectrum of intermediate SCN8A-related epilepsy Katrine M. Johannesen1,2 et al
The phenotypic spectrum of SCN8A, Jan Larsen, MSc*‡ Gemma L. Carvill, PhD*et al
>>GUIDELINES:
https://www.thecutesyndrome.com/reference-guide.html

The TBCK Foundation www.tbckfoundation.com (2019) 501c3 ILL

Our organization was founded by our family after a diagnosis of TBCK Syndrome and a visit to our son's researchers that determined that funding was a barrier to research beginning. Our founding team was a mother, father, and friend. Both of the later who are lawyers. We chose intentionally to keep the founding team small for simplicity and communication.

Tuberous Sclerosis Alliance https://www.tsalliance.org (1975) 501c3 CA
The TS Alliance is dedicated to finding a cure for tuberous sclerosis complex, while improving the lives of those affected.

Our story is one of determination and devotion, unyielding determination to help bring about better treatments and a cure for tuberous sclerosis complex (TSC) and an intense devotion to all those who share this battle with us. Individuals and families. Health care professionals. Medical researchers. Friends and family. Everyone who has a stake in this fight can multiply his or her impact by linking arms and working together to bring this disease to the brink of a breakthrough.

The Tuberous Sclerosis Alliance (TS Alliance) was founded on the core belief that community is a sustaining strength in the face of difficult challenges. In 1974 four mothers who shared the common bond of tuberous sclerosis complex came together to provide fellowship, generate awareness, pursue more knowledge and offer hope to each other. That was the birth of the Tuberous Sclerosis Alliance, and the spirit and intention of those founders permeate everything we do to this day.

>>RESOURCE:
Tuberous Sclerosis Complex: Genes, Clinical Features, and Therapeutics

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Wishes for Elliott: Advancing SCN8A Research www.wishesforelliott.org (2015) 501c3 DC

No answers came with diagnosis, at 15 months, which was devastating. No prognosis. All drugs were a crap shoot with our Dr, the head of Epileptology saying: "well you could try x AED next - it might help but it could also kill him. " It was an intolerable position to be in - and while we never really presumed we could see results yield in time to help Elliott, we committed to focusing on advancing research - so no parent or medical professional would be in the unbearable position we were in.

APPENDIX D. TOP 3 STRATEGIC PRIORITES

Organization	Describe your TOP 3 Strategic Priorities
Batten Disease Support and Research Association	Support Batten families regardless of where they are in their journey with the disease. - Host annual conference - host and support online caregiver page - support families one on one over phone, email, and visits - etc. Fund and facilitate research in all forms of Batten - Fund grants in as many forms of the disease as funds allow - connect families with research opportunities - write letters of support for researchers - Engage with industry Advocate for change - sign on to relevant state and federal letters in support of bills - Teach families how to be advocates - Provide relevant policy updates to community
BPAN Warriors	Creation of a BPAN collaborative research network: identification of researchers within BPAN, DEE, autophagy, iron storage disorders, ALZ, Parkinson's and related Neurodegenerative diseases; included platform, logistics and guideline development Host initial BPAN Research Workshop, which will serve as a first in a series of discussions which will serve as a high-level forum for sharing views, information, and analyses related to harnessing science and technology for catalyzing research, supporting collaboration and implementing, follow-on advisory work Identification of key therapeutic path(s) and validation of funded research required to bring to market
Bridge the Gap - SYNGAP Education and Research Foundation	SYNGAP1 (MRD5) Natural History Study Registry The primary aim of the SYNGAP1 (MRD5) Natural History Study Registry is to conduct a prospectively-planned and efficient natural history study that will result in the most comprehensive understanding of the disease and its course and pace over time. A critical amount of clinical information is needed to understand what is causing any disease and the purpose of registries are to capture information from each patient that may shed light on understanding how to pursue treatments. Since SYNGAP is rare, there are so few patients, it is critical that every patient is registered. This information will bring together information in order to understand the diversity of the disease. This program is made possible with the help and partnership of the National Organization of Rare Disease (NORD) and the US Federal Drug Administration (FDA). SYNGAP1 Family Meetups Our primary objective is to empower the patient and stress the importance of their involvement in driving research. With this goal in mind, we believe that having all the stakeholders in conversation will communicate the importance of patient advocacy and their role in accelerating ongoing research and get to treatments faster. Our goals are to provide support, education, and resources. Foundation representatives will provide up to date research information and present current findings on data collected from the SYNGAP1 Registry and Natural History Study. This will also afford better communication and patient engagement in the ongoing research driven by the foundation registry charter. Clinicians and scientists will be presenting information on current research and be available to answer questions in an open forum. This will also give families an opportunity to ask questions and get clarification from the experts. The education the families will take away from the meeting is intended to reach treating clinicians of SYNGAP1 patients and give families and caregivers a secure knowledge base of the disease and how to best approach available treatments. Build SYNGAP1 Centers of Excellence Our participating Institutes offer a comprehensive set of services: Not just core services, but advanced services and support services specifically geared to treating patients with SYNGAP1 Syndrome. The range of services often spans the entire continuum of care, not merely the acute care procedure. We currently are working with Texas Children's Hospital and Johns Hopkins to be our first SYNGAP1

	Centers of Excellence. We will work in the next five years to obtain two additional Centers of Excellence, one on the West Coast of the United States and another in Europe to serve and support our International families abroad.
CFC International	1. Expand our global reach to support patients around the world by adding family medical conferences in Europe. 2. Expand access to information to geographic regions for newly diagnosed families. 3.Provide medical conference access to families at no cost, by eliminating registration fees for all, and providing travel scholarships to those that income qualify.
Chelsea's Hope	1. Keeping researchers' "eyes on the prize". The leadership of the Lafora Epilepsy Cure Initiative (LECI) has impressed upon us how patient advocacy directly impacts the rate of scientific progress. Since 2014, there have been 5 International Lafora Workshops. This year it was held in Acala, Spain. Frank Harris, President of Chelsea's Hope, gave a stirring address and, as in all of the workshops, we prepared a video tribute to highlight a patient with Lafora Disease. In past years these have introduced scientists, most of whom have never seen a patient with LD, to the ravages of the disease as it inevitably progresses toward death within a decade of diagnosis. This year, we chose to highlight a newly diagnosed young man whose life is still filled with promise, who stands to directly benefit from the cure that is close at hand. As in prior years, the feedback we've received is that putting a human face on LD is a powerful motivator, causing researchers to put aside their competitiveness and individual priorities, and to work collaboratively toward their common goal of delivering to the bedside the first curative precision therapy for epilepsy. 2. Reaching every patient diagnosed with LD in North America, keeping them informed about the progress of the LECI, and impressing upon them the importance of enrolling in the Natural History Study that is currently underway and the critical importance of genetic testing of younger asymptomatic siblings. At the most recent workshop, the drug companies asked us to educate patients enrolled in the natural history study about the importance of consenting to blood draws and spinal punctures to obtain CSF for biomarkers. The protocol makes these optional, but the drug companies are telling us that they will greatly strengthen application for FDA approval for clinical trials. 3. At the 2018 Workshop, Chelsea's Hope was asked to begin to develop and house a patient registry in preparation for clinical trials. We were beginning to explore the best way to do that, contacting other members of REN and exploring platforms available through Global Genes and NORD. At the recent 2019 Workshop this request was withdrawn. Although researchers had previously stated their preference for the registry to be housed by an impartial third party where it would be sequestered from political considerations, apparently a platform has been devised and will be housed by one of the three Spanish academic research facilities. Thus this strategic priority has been tabled.
Chromosome 9pMinus Network	Yearbook for families to tell their story 9p minus Family Reunion in Iowa in August 2020. The reunion occurs every 3 years in a different state. past reunions were in Cape Cod, St Louis, Montpelier, VT and Lake Charles, LA Research - funding for new research with our team of doctors at Washington University, St Louis MO
CureSHANK	We want to de-risk preclinical research through the following approaches: Incentivizing the replication and validation of key findings in our field Developing assay standards for cells and model systems Developing and validating clinically translatable predictive biomarkers
DDX3X Foundation	1. Identifying the population affected by DDX3X syndrome through expanded awareness and education about the disorder and growing our community. 2. Expanding our patient and contact registry. 3. Promoting research of DDX3X syndrome and expand our knowledge into basic science of DDX3X as well as the clinical understanding of the disorder.
DNM1 dynamos - Connecting DNM1 Families	1. Promote awareness of EE caused by DNM1 mutation among researchers and clinicians in order to expedite research into treatment options. This is to be achieved by partnering and interacting with organizations such as Epilepsy foundation to increase DNM1 specific awareness. Also spread the word through clinicians and contact researchers via emails and networking platforms 2. Provide support to constituent families to help traverse the challenges faced everyday
Doose Syndrome Epilepsy Alliance	We are not currently active. I am trying to get in touch with the parents who built the website and gain access again to the email account. Sadly we have not kept up with these things. 1. update the website. 2. gain access to email and recruit parents to monitor.
Dravet Syndrome Foundation	1. Development of donor development program and additional time spent on grant opportunities to further expand diversity in our revenue streams. 2. Enhancement of our communications calendar and social media presence to increase awareness and education of Dravet syndrome.

	3. Increase the number of qualified applications that are submitted for our research grant program, as well as expand and adapt our grant program to meet the unmet needs of our professional community.
Dup15q Alliance	Identify Board Member Skills needed for Dup15q Alliance Recruit New Board Members Committee Restructuring and Development 100days document split into links Structured Communication with all incorporated International groups
FamilieSCN2A Foundation	1. Community / patient / family support through education and empowerment - conferences - webinars - family meet ups 2. Improve clinical treatment protocols for children with SCN2A Disorders - centers of excellence - conferences - webinars - data collection for NHS 3. Research that is directly related to finding new treatments for SCN2A Disorders - Action Potential Grants for post docs - bio marker studies
Glut1 Deficiency Foundation	1. 9th Biennial Conference - families and professionals. We held this meeting in Washington, DC in July. 2. Scientific Research Grants - $187,000 awarded in April 2019 3. Educational exhibits at major medical meetings to raise awareness and education professionals.
GRIN2B Foundation	Our 2019 priorities have been fundraising in order to create our first grant research cycle. This cycle was completed last month and we are excited to announce the initiative later this month. Our fundraising efforts have brought in tens of thousands of dollars for this grant cycle. In March, we conducted our third annual GRIN2B Awareness Month, where we used social media to spread awareness of GRIN2B-Related Neurodevelopmental Disorder. This month also helped our overall fundraising efforts to both launch our grant cycle and to help with overall family support and education. In September of this year, we attended a GRIN Disorder conference conducted by Emory University in Atlanta, GA. This conference was attended by the leading researchers and families for all GRIN-Related disorders (there are a total of 7 different GRIN genes). This conference enabled researchers to share data face-to-face to aide in the groundwork for future patient registries.
Hope4Harper	The first 3 years of Harper's life were extremely difficult. With each passing day she became more and more medically fragile. Every morning we wondered when we went to her room if she would be alive. With the misunderstanding among other organizations as to our desire to actually contribute rather than compete with them, it was a struggle to find the desire to keep the organization in existence while living the daily struggle of caring for Harper. But every time I thought we should quit I would get an email or a message from a parent and realize that what I was contributing mattered. We then began to think "what if Harper dies? How can we keep making a difference?" And the topic of organ donation for scientific research surfaced. This topic is so under discussed that it took us two years to figure out how to donate Harper when she passed away. Thankfully, we figured it out with the help of Dr Jensen and other's willing to discuss the topic 30 days prior to Harper's passing we knew the exact steps to take. In January 2016, Harper became the first CDKL5 brain donor for scientific research. This changed everything and expedited research in phenomenal ways. In April 2019, with the use of Harper's brain donation Dr Jensen was able to publish our CDKL5 research in the Journal of Neuroscience. With Harper's passing we were able to really asses the existence of our organization and its purpose. We feel we contribute strongly in 3 main areas: 1) Research using Harper's donated organs 2) Organ Donation Advocacy for scientific research 3) CBD Advocacy - In 2013, Harper began using CBD and her success changed the path of our funded research. In addition it changed CBD laws in 3 countries over a 2 year period of time.
International Foundation for CDKL5 Research	1. Grow patient/caretaker outreach and tools 2. Develop our clinical research network infrastructure with a centralized smart IRB and repository that works well with red cap 3. Develop an ambassador program.

Jeavons Syndrome Facebook Group	- Patient / Caregiver support - This is the most critical mission of the Jeavons site. This is the primary purpose to let people know that no matter what country they live in and what type of access they have to Neurologist's and epileptologists, they are not alone and they are not without hope! - Patient Information- It is important to provide factual information and to make clear that the experience that one patient has is just that. It is there individual experience. We do not go condone certain treatments or medications but rather it is a place that people can find out options whether it is info about polarized sunglasses, vitamin supplements or traditional medicine. The other areas are not critical to our mission.
KCNMA1 Channelopathy International Advocacy Foundation (KCIAF)	Educate patients, families, the public and the scientific community about KCNMA1-associated conditions Encourage and promote efforts to find effective treatments for the wide range of symptoms which can be caused by KCNMA1 mutations Provide opportunities for patients and families to meet, whether in-person or otherwise, and share their experiences
KIF1A.ORG	*Expanding the number of diagnosed patients and engaging them in our community *Funding research *Developing therapeutic strategies that will benefit this generation of patients
LGS Foundation	PFDD Meeting Family Conference Build local meet-ups
Lissencephaly Foundation Inc	To execute and release our packet programs; we are preparing packets/pdf files to educate & give hope to newly diagnosed families. This packet will be distributed to new families after diagnosis, we hope for it to become widely available and be a resource for families who are just starting their journey. Bring on at least 2 additional regional directors to help establish connections within the United States. After establishing that we will move onto Canada, United Kingdom, & Australia. Start a membership plan which will provide us with monthly/yearly income from individuals. This membership will be something we will be speaking about in 2020, I do not have details at this time.
Mickie's Miracles	Education, advocacy, and awareness. Education: Mickie's Miracles strives to educate not only the general public, but law makers, politicians, healthcare professionals, and care givers. The more education that is available about Infantile Spasms, the more babies' lives will be saved. Advocacy: If a child fails the first form of epileptic treatment, they must be seen by a Pediatric Epileptologist. These doctors have a higher level of understanding epilepsy than a neurologist or pediatrician. Oftentimes, parents are not told that a Pediatric Epileptologist even exists, and if they are aware of them, then they face barriers from all sides such as insurance coverage, or doctors refusing to provide a proper referral. We advocate for our families to be able to urgently receive this higher level of care. We also advocate for all people with epilepsy by helping to end the stigma around epilepsy. Awareness: Early Diagnosis is critical. Pediatric Epilepsy is catastrophic to the developing brain. Epilepsy is a progressive disease. Early Intervention is essential and a matter of life and death. Urgency is imperative. Parents often miss signs of catastrophic seizures because "babies do weird things." And oftentimes when parents do notice the spasms, doctors will misdiagnose it with things like reflux, movement disorders, etc. It is our job to raise awareness of how to recognize the signs, as well as raising awareness that Infantile Spasms IS a medical emergency.
NORSE Institute	1) Increase awareness of NORSE in doctors which includes the awareness that time to treatment is vital so they can provide more timely treatment. Also to increase awareness of NORSE in patients/families so they can find a community, and to provide basis for future research and funding. 2) Develop consensus proposed diagnostic workup and treatment management protocol based on integrated perspectives from the previously separate fields of pediatric FIRES and adult NORSE. 3) Bring together clinicians and basic scientists to identify elements to build preliminary NORSE model in laboratories and develop consensus on ways to collect bio specimens.
Phelan-McDermid Syndrome Foundation	1. Develop the board and operations infrastructure to handle the expected growth of the organization. This includes establishing scientific and medical advisory committees, board development and staff expertise. 2. Establish clinical care guidelines and identify and build centers of excellence for patient-centered care and research. 3. Increase awareness and funding through the continuous outreach, development, and nurture of strategic relationships in science, medical, business and patient communities.
Project 8p	Patient-led funding research to understand the molecular biology of the disorder Funding research for novel technologies and bioinformatic tools to support downstream strategies Determining meaningful outcomes and a standard of care Cultivating clinical experts in 8p internationally

PVNH Support & Awareness	Better definition and education of the cortical malformation that is PVNH. We are participating in a European brain consortium which is redefining cortical malformations, including PVNH, as well as preparing recommendations for diagnostic work-up. Next step will be to prepare a natural history study questionnaire that will look at how PVNH patients' daily lives are affected and identify what should be investigated to make life easier for patients. Form alliances with groups of interests subject to see or have PVNH patients in their own groups: connective tissue disorders, epilepsy groups, aortic disorders, etc. so that we can provide better support PVNH families with our allies. Continue to fundraise for research and conferences. We have now a small research fund and an even smaller budget for a conference to happen in 2020. This in turn will enable to get more patients diagnosed.
RASopathies Network	NOTE: to Patient/Caregiver Support & Community, we rely on the CS, CFC, Noonan and NF1 family organizations to provide direct support to families, and work to connect families to their appropriate family organization if they come to us first. For those who have an identified RASoapthy gene mutation but have no family organization to go to (because there doesn't exist one), we mean to provide patient/caregiver support and to bring them together and will do what we can to support them building their own communities. 1. Convene biennial scientific research symposia for researchers, clinicians and families 2. Review our outreach strategies for updating - for families as well as for professionals (researchers, clinicians, therapists) 3. Formalize a family support position (a parent)
Ring Chromosome 20 Alliance	1. Secure start-up funding from corporate donors 2. Recruit board members 3. Recruit volunteers
Ring14 USA	1. Recruit and engage more scientists or physicians to consider Ring14 Syndrome. 2. Define new research initiatives to guide research going further. Both 1) and 2) have been pursued by hosting our 3rd International Scientific Workshop in last October of 2019. This is an invitation only conference and much thought goes into who is invited with the vision to engage these participants in our community. 3. Strengthen collaborative efforts between Ring14 USA and other epilepsy associations. We are at the point in our organization where we truly need to learn from others how to expand beyond the very narrow objective we have had in the past.
Shay Emma Hammer Research Foundation	Raise funds to support the Registry. Raise funds to support research Increase awareness of this rare disease.
SLC6A1 Connect	SLC6A1 only funds research that will result in a curative approach.
TBC1D24 Spectrum Family Network	1. Family Support 2. Communication with specialists in our field of need 3. Becoming a not for profit organization
TESS Research Foundation	1. Clinical Trial Readiness We are trying to figure out the best metrics for natural history studies and clinical trial outcome measures, including non-seizure endpoints. We are aiming for disease-modifying treatments. We are hoping to treat patients without having a placebo arm. 2. Gene Therapy continued research We have developed a gene therapy for SLC13A5 Deficiency that continues to be studied for toxicity and efficacy. 3. Basic Research We continue to develop model systems, including iPSCs, organoids, and animal models with the aim of using them for drug screens, etc.
The Brain Recovery Project: Childhood Epilepsy Surgery Foundation	1. Launch of Global Pediatric Epilepsy Surgery Registry (completed); 2. Research meeting re functional outcomes after large resective/disconnective procedures (completed) 3. Assembling research consortium
The Cute Syndrome	Professional Education and Research - Through collaboration, communication, and financial support with our research community, we can improve the timeline for better treatments for SCN8A and comorbidities. Increase knowledge throughout the healthcare system, we can increase diagnosis and assure the best quality of care. Family Education & Advocacy- We will continue to support patients and families with disease education and advocacy needs through expansion of patient resources, as well as family support. Increase Funding - 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.

The TBCK Foundation	1. Advancing research: this is our focus and also the most challenging as we learn how to best navigate this. We hope to send funding to CHOP where our researchers are located to begin a natural history and patient registry. 2. Advocacy/ education Since our disease is so rare (less than 60 cases) advocacy is directed at sharing the story of our rare disease; but also extends more broadly about advocacy for inclusion, accessibility, and issues faced by the disability community. 3. Family support. To date, we don't have the funding to support families how they often need it most: financially, but our goal is through the existence of our foundation we are bringing families together with our Facebook group and our foundation as a whole.
Tuberous Sclerosis Alliance	1. Accelerate Research • Collaborate and extend partnerships with other nonprofits, academic institutes • Advocate for Federal research funding • Increase Preclinical Consortium productivity by engaging researchers, improving models, adapting to priorities of members • Expand capabilities of centers in the Clinical Research Consortium and fund pilot or add-on studies with pathways to outcomes. • Increase rate of biosample collection by enrolling individuals anywhere in US • Invest research grant funding in postdoctoral trainees and highly novel research ideas • Convene targeted workshops on urgent research problems and follow-up with funds. 2. Improve Access and Quality of Care • Advocate for state funding for research and/or Centers of Excellence • Develop a support hub to navigate access to FDA approved therapies • Build evidence for what constitutes high quality care for TSC • Expand knowledge of evidence-based, best practices of TSC clinical care among TSC Clinic healthcare professionals and other medical providers involved in the treatment of TSC • Expand access to care for people in regions with no TSC Clinic or TSC- knowledgeable healthcare professionals. • Improve access and delivery of evidence- based, high quality clinical care for patients with TSC, including comprehensive adult care for inpatient and outpatient services. 3. Support and Empower Constituents • Create a premiere support services volunteer corps through Clinic Ambassadors, Junior Leaders, Dependent Adult Transition Resource Coordinators, Adult Regional Coordinators, Spanish Support Network, Education Parent Mentors and Community Alliance leadership • Expand skills of parents/caregivers and young adults to advocate for and access appropriate educational services to improve quality of life • Grow collaborations with other nonprofit organizations internationally, nationally and locally to ensure access to resources, support services, transition tools and information • Expand reach of education and support through use of technology • Increase scientific knowledge in the TSC community • Grow and advance the Global Alliance Program
Wishes for Elliott: Advancing SCN8A Research	1.supporting the highest quality scientific, peer-reviewed research to accelerate answers and hope for children struggling with SCN8A epilepsy and related disorders 2. Convening experts/facilitating collaboration of scientists researching SCN8A and related disorders 3. Integrating specially targeted information and support for families with the children most severely affected by Developmental Epileptic Encephalopathies

APPENDIX E. OVERARCHING CHALLENGES

Organization	**Describe the TOP 3 challenges.**
Batten Disease Support and Research Association	1. Fundraising- each year we support more and more families but we never stop supporting those who are already a part of our community 2. Changes in health care - it is becoming increasingly difficult for our families to have access to the necessary medical and social supports 3. limited staff - our small staff can only do so much but there are many more ways we could be helping families
BPAN Warriors	Lack of cooperation from established research community in the US, which is very protective of their long established "expert" status in the US. Working to establish a collaborative without rocking patient sense of security and alienating the establishment has made funding and real communication and collaboration difficult. Most of our BPAN patients have been led to believe that research is limited to one specific academic institution in the US, whereas the research in autophagy, iron storage disorders, DEE and neurodegenerative diseases as a whole is expansive. Few have ventured beyond the diagnosis and even fewer are able to understand the science (see below). Digestible, distilled science that the patient community understands: This is currently the biggest hurdle that applies to patient engagement, fundraising, and overall effectiveness of all future community participation. With the rarity of the disease and limited clinician awareness of BPAN, we cannot expect the patient community to necessarily take on the challenge to become the researchers

	(although many of us have done just that). In a disease that has so many symptoms and affects so systems, having simple, patient consumable copy is critical. This is an urgent need as each rare disease community is trying to desperately to piece together content to package and communicate to their disease specific community, attempting to piece together patient friendly copy from very complex medical and scientific research is daunting. Attempting to communicate about the research landscape and discussing the merits of the research on the horizon is difficult without the proper language and tools to communicate. As an organization, we are working towards rapid growth, however we are limited in skilled support. Currently, BPAN Warriors has no paid staff. Sarah leads the day to day operations and is supported by a working board of directors and volunteers are supporting the operations of the organization in the areas of organizational development, creation of a Scientific and Medical Advisory Board, fundraising, awareness and outreach. The organization utilizes the services of consultants (as needed) with areas of expertise in nonprofit organizational development, fundraising and marketing. Having no paid staff, or office space, allows us to keep our operating costs low and allocate the majority of our funding to direct programming and research initiatives. To really expand and grow, the organization must hire or onboard 1-2 full time/part time personnel to help with administration, coordination of key planning items and facilitate with follow-up and follow through. This will require additional funding and structuring to allow for learning curve.
Bridge the Gap - SYNGAP Education and Research Foundation	Newly organized groups with similar missions in the same space has led to confusion and undermined progress in building community, raising funds and collaborating with researchers.
CFC International	1. Exhaustion of our donor base, which is mostly our patient families. 2. Expanded into new forms of communication to our families that aren't email or Facebook. 3. Reaching families as we see an increase of diagnosis from 1 per month, to one per week.
Chelsea's Hope	We are too small (25 known patients in the US) and we do not have the funding to hire professionals. In a perfect world, we would hire or outsource professionals in the following areas: 1. Social media, website development and management. We have a parent with a background in website design who has recently joined the Chelsea's Hope Board. We hope that this will enable us to transform our website into a polished and fully functional center of communication between Lafora families, LECI researchers, drug companies, and donors. 2. Grant writing. We also have a new board member with experience in grant writing and we hope this will allow us to more fully take advantage of funding opportunities we know are out there, but we've not had the expertise to tap into thus far. 3. Fundraising
Chromosome 9pMinus Network	More families are asking their question on Facebook....only using the Network at a resource. Decrease in donations smaller group of families stay active in Network.
CureGRIN Foundation	1. Fundraising: Cures/Therapies are expensive. We are just starting out and we know we have a long way to go. But we have lofty goals for the next two years and we intend to meet them by heavily involving patient families. Our first goal will be to recruit 100 "GRIN Champions" to each raise $2,500. 2. Awareness: GRIN disorders are still very much unknown. Not only amongst the general population, but also among clinicians. We need more doctors/researchers to become aware of GRIN disorders and how they affect the brain. While we are lucky to have dedicated doctors/researchers already studying GRIN, we know the importance of raising recognition. While GRIN disorders are "ultra-rare" their awareness does not have to be.
CureSHANK	1. So far, there has been a lack of a cohesive scientific story, with multiple research teams pursuing a lot of different types of projects with a lot of variability in their approaches. 2. There has not been a clear path or roadmap for progress in our disease. 3. There has been a failure to move some interesting preclinical findings into a commercial pipeline.
DDX3X Foundation	Out three largest challenges: 1) Financial resources are insufficient to hire full-time staff and fund the research objectives that we would like to fund. 2) The number of identified families needs to expand in order to make us more attractive to researchers and those developing therapies. 3) Volunteers are difficult to engage due to the extensive care our children require.
DNM1 dynamos - Connecting DNM1 Families	1. Challenge to have research organizations make DNM1 research as their primary focus. The reasons for this are: - Approx. 100 children known to have impacted to DNM1 mutations. Not enough business incentive for drug companies to invest in DNM1 specific research - Not just an epileptic encephalopathy but a developmental encephalopathy as well. Developmental delays likely unrelated to Epilepsy and therefore controlling just epilepsy via medicines is unlikely to help developmental progress

	- No specific source of funding identified. Crowd source funding likely not an option.
Doose Syndrome Epilepsy Alliance	1. not active 2. Haven't found a family that would like to take over management of the NP
Dravet Syndrome Foundation	Our community and foundation continue to be fortunate in having unexpected opportunities that arise that sometimes shift our timelines on other projects. Our rapid growth is exciting, but we often find ourselves short-staffed in doing all that we want to do and sometimes have to bump certain objectives down on our list of priorities.
Dup15q Alliance	Not enough staff to complete all tasks
FamilieSCN2A Foundation	Too few patients, most in crisis mode so unable to participate. Natural History Study - too few patients and too new of disease. For profit Industry collaboration.
Glut1 Deficiency Foundation	1. Money is an issue due to the example given above, and also that some families seem happy to let others do the work and remain unengaged. 2. Time is an issue. There never seems to be enough. I am the only paid staff member and volunteer efforts have waned recently. 3. Prioritization - related to time and money. With limited resources and limited time, prioritizing where to spend both is difficult as it is hard to decide what programs and projects are most beneficial and give the most return on our investment.
GRIN2B Foundation	Our first challenge was encouraging families to mobilize and fundraise for our foundation. Many families did so, but many more did not. Our fear is that this will lead to over extension of some families. Our second challenge was that we discovered that GRIN2B is one GRIN gene that exists within a family of GRIN genes. This makes us very different from other rare diseases and has forced us to re-evaluate our non-profit model. This year Cure GRIN Foundation was created to raise monies to research and hopefully cure all GRIN-Related disorders. Though we have overlap, our missions do vary slightly, and we have had to work hard to figure out how to work with this organization and where there reach ends and ours begins. This collaboration has caused us to vary slightly from the original path we thought we were on. Our other challenge is coordinating with researchers to develop an international patient registry for GRIN2B patients. Too many researchers have their own ideas regarding how to do this and it is hard to streamline this registry process so that we can fund its efforts.
Hope4Harper	Hope4Harper is a small nonprofit with a large international presence thanks to technology and our willingness to be transparent in with our lives. We have impacted millions through our CBD advocacy and our willingness to donate our daughter to science and talk about it. The research we are a part of has connected our rare childhood disorder of CDKL5 to the major adult disorder of Alzheimer's. Despite our impact we face 3 major challenges in achieving our goal of treatment for CDKL5. 1) Finances - research costs money and our money comes from grass roots fundraising efforts. Our story is powerful but for some reason we struggle in getting it to the masses. I have traveled the world publicly speaking to thousands but our following and contribution levels remain the same. I have even been on Fox 4 Dr Manny in New York that reruns internationally on any Fox station in the world. 2)Awareness - Again our story is powerful but for some reason we struggle in getting it to the masses and struggling to help them understand unless explained one on one how impactful Harper has been to millions. Our social media is lacking because Facebook laws have banned our ability to promote our efforts because our research that is done at a public university is in part CBD related. 3)Volunteers - We do have an end goal that is attainable and that is a treatment for CDKL5. This will be completed in my lifetime. The Orphan Disease center in Pennsylvania believes strongly enough in the ability to find a treatment that of the 7000 options they are contributing funding to CDKL5 now that Harper has donated her organs to the research of it. I expect Harper's story to be around forever but I expect our organizations purpose of finding treatment for CDKL5 to come to an end. But for that to happen I need help.
International Foundation for CDKL5 Research	1. Lack of budget for paid executive staff - we were able to add a paid science advisory position in 2018 and hope to add some additional paid staff in the next 5 years 2. Slow development of community financial support and participation in research with extreme caretaking burden and high medical bills. 3. High front end costs to collect data and clinical research in a manner that can be validated and used
Jeavons Syndrome Facebook Group	Because of the nature of the site, we do not need funding. That said it would be truly wonderful if there was money to have Jeavons studies conducted because so little is really known about it. Every single neurologist that I have met either knew very little about Jeavons or knew nothing at all! Some neurologists don't even acknowledge that Jeavons is a disease! I have had to educate these doctors (and I am not the one with a medical degree!). This is my biggest challenge.
KCNMA1 Channelopathy International Advocacy Foundation (KCIAF)	Lack of patient/parental involvement in organization mission or operation Lack of neurologist/physician involvement in information sharing Lack of volunteers to assist with anything

KIF1A.ORG	'-Time (we are in a race against time with a degenerative disease that has no treatment) -Money (to shorten the time it takes to get to treatment, we must scale up the resources going into research and discovery)
LGS Foundation	Time. Money. Trained staff.
Lissencephaly Foundation Inc	Reaching these newly diagnosed families, our only way to reach them at the moment is via our website, email, & social media. We need to establish a way to reach them while they are in the hospital or early on in diagnosis. Finding adequate individuals to commit their time to being on the board. We currently don't have anything to offer members since we are such a new foundation, our biggest struggle will be finding ways to "compensate" or give members benefits for being a continuous donor to our foundation.
Mickie's Miracles	Financial Finances are unfortunately an integral part of a nonprofit organization. Money is absolutely necessary to be able to fund our mission to urgently connect families all over the world to level 4 epilepsy centers throughout the country. Families go into financial destitute trying to get proper treatment and we strive to be able to support these families through their diagnosis with Infantile Spasms. Geographical Our families come from all over the world. For example, we recently helped a baby in Canada as well as a baby in India. Both families were desperate to get their hands on the front line drug for Infantile Spasms to help stop the catastrophic seizures happening in their child's brains. We worked diligently to overcome these geographical issues, as taking a currently seizing child on an airplane is just not medically safe. We were successful in transporting medication to these babies. Resources/Barriers to care Pediatric Epileptologists are epilepsy specialists who work specifically on the brains of infants and children. Tragically, the amount of children needing this kind of doctor far beyond exceeds the amount of practicing epileptologists. And the existing pediatric epileptologists are generally booked out for months seeing current and new patients.
NORSE Institute	1) NORSE terminology is new, there are no identified biomarkers, and it is a syndrome that probably encompasses several etiologies. Dissemination of this complex information characterized by uncertainty is slow. 2) Consequences of NORSE are up to 30% mortality (depending on study) and significant proportion of survivors have devastating brain damage, almost all survivors develop epilepsy. Families with survivors spend most of their time, energy and money on their loved ones. Bereaved families are often in shock. Many families are not even aware their loved ones had something called NORSE. These facts explain the family low involvement and small funding base for our organization. 3) Poor communication between doctors: among medical teams and between medical teams within hospitals, between individual doctors studying NORSE, between clinicians and basic scientist. Information is stuck in silos.
Phelan-McDermid Syndrome Foundation	1. Our syndrome is greatly underdiagnosed, so getting the word out about the syndrome and the importance of genetic testing to patients who may be undiagnosed is critical but challenging due to the expense of global communications outreach. 2. Our fundraising is currently primarily based on donations from family and friends, who are already financially overextended and oversaturated with the ask and overwhelmed with the care of their loved ones. We are faced with the challenge to engage and cultivate partners outside of that circle through the use of peer-to-peer, monthly giving, legacy, and other donor development tactics. 3. We need consensus in our science and medical communities so we can better understand opportunities to make progress in research and medical support to our members.
Project 8p	Competitive landscape and lack of transparent sharing with PAGs More patients that scientists - lack of funding. Incentive model for researchers and biopharma to be interested is not beneficial to eradicating disease - it plays to self-preservation and ego. Nothing to do with actual healthcare.
PVNH Support & Awareness	Getting more families involved in the actual work that goes behind the scene. As a sole volunteer, I am now faced with too much work and must surround myself with the proper talent to continue to proper us forward. We need to pursue a nonprofit status in 2020 in order to grow and achieve our goals, which will also mean being able to grow our fundraising by potentially securing grants. The diagnosis and information piece in the medical community remains a challenge. PVNH is seen by many as a brain disorder which does not cause much damage, when it fact it is multisystem from mild to extremely severe. As heterotopias are found in asymptomatic individuals as well as with those with symptoms, we need to continuously push for better depictions and accurate information sharing.
RASopathies Network	1. As a working volunteer Board, each member with their own "salaried" full-time jobs, and parents of a child with a RASopathy (except my husband and me; our son died of cancer related to Costello syndrome), it is a challenge for Board members to carve time to run the organization. 2. With increasing dawning from the cancer research community - including the maturation of the Ras Initiative at the National Cancer Institute (about 4 years old now), come an exponentially growing bibliography of peer-reviewed publications that could potentially be of value to shed light on the RASopathies. We've gone from Needing More Research to Not Being Able to Keep Up.

	3. And as always, the amount of energy to raise funds is limited, and takes away from programming. We have succeeded in earning NIH R13 scientific meeting funding, and now of course, I need to carve time to maintain our SAM and Grants.gov accounts - and submit the paperwork to receive and report on the usage of the funds. 4. To this, we continue to seek someone with financial and business acumen who isn't afraid of the human genetics science - and clinical trials acumen "needed" to understand, promote and protect families affected by the RASopathies, to join the organization.
Ring Chromosome 20 Alliance	Goals for 2019 were sidetracked by personal/family demands.
Ring14 USA	One of the major challenges with regards to engaging researchers in Ring14 or even in the study of large deletions of the 14 chromosome is a lack of tools to deal with large structural problems at the chromosomal level. We have to develop better techniques to go from large variants to identifying the relevant genes and pathways affected. There is a huge focus on monogenic syndromes, and rightly so - this is the place to start, but we need someone today developing the techniques that will prove useful in the future as more complex syndrome are explored. Money is usually an issue with funding rare syndrome programs, and this is certainly exaggerated within the ultra-rare space. Not only is hard to engage the very few families to give or to fund raise, but it is also hard to engage pharma when they already have established relationship with other rare epilepsy organization who have a larger patient population (one that will support clinical trials. Time is also a precious commodity within volunteer organizations, Families can be vocal about services they need, but it's hard to make that happen without manpower. But the majority of our volunteers are the very families who need help - these families are just trying to survive.
Shay Emma Hammer Research Foundation	Rare disease has few advocates, leading to difficulty in raising awareness and funding.
SLC6A1 Connect	The largest challenge is finding appropriate scientific efforts followed by fundraising.
TBC1D24 Spectrum Family Network	Patients and their families are from all over the world. Our disorder is extremely rare yet children are being diagnosed more often as DNA testing becomes more affordable and accessible. We don't have anyone who is willing to be the Treasurer, and one needs a Treasurer to become a non-profit
TESS Research Foundation	1. funding 2. low incidence and prevalence 3. lack of understanding of gene therapy and disease modifying treatments
The Brain Recovery Project: Childhood Epilepsy Surgery Foundation	1. Patient donation pool is small, although we have tremendous support 2. Current staff is at maximum bandwidth. Funding limits our ability to hire more; 3. We are a research-based organization and cover a wide range of topics on our website. These topics require very deep dives in the science - it can take months to reach conclusion, update website, etc. on one small topic.
The Cute Syndrome	Volunteer retention - With just two full time volunteers the workload is too much and a financial strain on their families. Too few diagnosis - need more diagnosed to have adequate information to provide to research. Also with a small population not enough are giving financially to the cause. Families are overburdened with medical care and cannot extend to volunteering.
The TBCK Foundation	1. Funding. It seems there is a lot of confusion and just lack of understanding of what our foundation and what foundations in general do. We have done a few online fundraisers and have had minimal participation from families. I think a lot of that is them being so overwhelmed by their medical needs and also because a lot of families depend on asking for money to support their families and complex kids. 2. Depth of understanding of the foundation, our mission and what we do. Building off number one, it seems there is a disconnect. I don't get the impression people always see the mission as one that applies to them. I think they want research, but don't know how to engage. 3. Confusion on Engagement. I think people want to help, but just don't know how to engage their audience and their network. Our family included! I think they want to host an event or fundraiser, but don't feel equipped.
Tuberous Sclerosis Alliance	1) Making easier for TSC community to donate biosamples to the TS Alliance Biorepository Project. 2) Aligning clinic standards with consensus guidelines and patient/family feedback. 3) Providing resources and information to the TSC community to engage them using digital technology in ways they communicate today.
Wishes for Elliott: Advancing SCN8A Research	1. Fractions, inefficiency, within community . 2 .Challenges connecting with families 3. Time, energy given intense family need

APPENDIX F. SIGNATURE PROGRAMS

Organization	List and describe any signature content or program(s) that your organization has developed that it would be willing to share with other Rares. If you haven't developed your own programming or content, feel free to mention programming or content you "covet" and have borrowed or adopted from another organization.
Batten Disease Support and Research Association	Annual Family Conference: 3.5 days of programming for families including a separate sibling camp, nursing care for affected loved ones, and daycare for children 2-5. Programming includes research updates, educational sessions, and a memorial ceremony. Closed Facebook page for parents and caregivers for peer-to-peer support. Page is monitored by staff to keep it a safe space. Ask-An-Expert chats hosted on our closed caregiver page where a topic expert is brought in to answer questions from caregivers. Topics cover the lifetime of the disease including grief and bereavement.
BPAN Warriors	BPAN Warriors Slide Deck: Our Story, Our Disease, Our Mission, Our Goal We hope to adopt a one page clinical care guideline similar to the one on the Phelan McDermid website, more as an easy patient starting point to engage clinicians and specialists. For more complete patient care guidelines, we hope to use the CDKL5 community's as our template as their symptoms and care provisions are nearly a complete overlap with our community.
Bridge the Gap - SYNGAP Education and Research Foundation	Family Meetup Program
CFC International	1. Family Liaison Program - a program dedicated to ambassadorship for newly diagnosed families. 2. Self-Care workshops (monthly via computer or call-in) 3. Emergency Medical Funding for families in need. 4. Seizure Natural History Study with the University of Minnesota 5. Patient Registry through Invitae
Dravet Syndrome Foundation	DSF Research Grant Program - our grant program is made up of four arms of seed funding to allow researchers to collect the data that is helpful in proving their hypothesis and allows them the data needed to apply for funding from larger grant mechanisms. DSF Research Roundtable - this annual 4 hour meeting brings together the top researchers and clinicians, along with industry members, to discuss the latest advancements in Dravet syndrome and future collaborations and next steps. DSF Family Network - this is a structured program that allows us to gather information on our patient families and includes a variety of initiatives, including annual Day of Dravet regional workshops; regional Family Ambassadors; our online support groups; and Dravet Dialogues (live virtual events that offer education and interaction for our community members). Day of Dravet Regional Workshops - mentioned above, these 1-day meetings are more casual gatherings for Dravet families, allowing them to learn about the latest in Dravet syndrome while connecting with families in their region. We take the opportunity to educate them on the need of the patient voice in research and how they can participate, as well as assure that a large portion of the meeting is reserved for guided discussions to help us identify the needs of our community and gap of care, to help guide our future priorities. It also includes a Sibs Camp to give a chance for siblings to bond. DSF Biennial Family & Professional Conference - this 3-day event is held in even numbered years, our conference offers a CME track, as well as sessions on patient and family needs. Sessions are recorded and archived for those unable to attend. There is also a sibling camp and activities for Dravet children throughout the event. Each evening, we hold a family gathering dinner and activities to allow families a chance to connect. DSF Patient Assistance Grant Program - offers grants for durable medical goods and educational needs for our patient community. Caregiver Connect - our newest program is made up of several initiatives, including DSF Birthday Buddies (where the Dravet child receives a special gift on their birthday); Connect Grants (to encourage regional gatherings for families); and a series of Caregiver Burden videos and a workbook (which is currently under development).
FamilieSCN2A Foundation	NPO org chart and flow
Glut1 Deficiency Foundation	Find Some1 with Glut1 - our professional guide we share at medical conferences through our exhibit tables Family and caregiver brochure Video series - interviews with professionals, other families who share their journeys
Hope4Harper	This is the resource I am most proud of because it was the most challenging to develop being we had to live through it in order to create it..... https://dev.hope4harper.com/organ-donation/ The other resource I have created is #LifeAfterCDKL5 Facebook support group and in person support section in the CDKL5 Family conference hosted by IFCR. Though there are some in the support groups that see me as a "lack of hope" I strongly disagree. I am living

	proof of HOPE. Hope for not only treatment for CDKL5 from the donation of Harper to science but also HOPE that one can survive life after CDKL5. I am sensitive topics that need to be heard.
International Foundation for CDKL5 Research	We have developed CDKL5 guides that can easily be modified for other disorders. We recently worked with Dup15Q to access our Medical Advocacy guide - find all the guides here: https://www.cdkl5.com/cdkl5-resource-library/ We have borrowed Dr. Tim Benke's multidisciplinary model and took it a few steps further with the development of the Clinical Research Network mentioned earlier. Any complex disorder will benefit from a multidisciplinary clinical model - this is scalable.
KCNMA1 Channelopathy International Advocacy Foundation (KCIAF)	We are a new organization and have no programs yet.
KIF1A.ORG	Tools for Development: These tools support every function of research and development — from pre-clinical discovery through clinical trial readiness and a prepared, proven regulatory pathway. KIF1A resources are powered to support any therapeutic program working to bring treatment to children diagnosed with KIF1A Associated Neurological Disorder: //www.kif1a.org/research/tools-development/ Family & Scientific Engagement Conference: https://www.kif1a.org/2019conference/ Research Simplified: We work with KIF1A researchers to summarize their scientific research in plain language for our families and follow up with interviews to discuss questions gathered from our community: https://www.kif1a.org/category/research-simplified/
LGS Foundation	We borrow extensively from TS and DSF. Our family conference is pretty amazing and is always a huge success. **From interview with Tracy, Dravet has SUDEP Risk; Phelan has End of life Toolkit; LGS has PFDD/FDA; TS has clinic model and ambassadors; IFCR has centers of excellence; TESS has SUDEP; Bridge SYNGAP has education.*
NORSE Institute	Our website is modeled after the CDC travel inoculations website. Professionals and laypeople come to same site. Each side varies in focus and depth of information, but users can choose to read either or both sides. Simple layout, no flash. For families, we have basic glossary of terms and discussion of uncertainty inherent in NORSE. For professionals, there is reference list: a curated reading list from Dr Nicolas Gaspard, annotated with his notes. I want to develop ways to prepare families for bad outcomes, make them aware palliative care from hospital can accompany aggressive treatment. And develop wording to bring choices of autopsy and donation of brain specimens to science.
Phelan-McDermid Syndrome Foundation	Our organization has developed a patient registry that includes patient-reported data, genetic reports, and electronic health records. The genetic reports are curated by a genetic counselor and reviewed by genetic advisors to ensure high-quality data. We have developed expertise in creating a registry. We have a data acquisition review process to review all requests for registry data for scientific rigor and patient interest/merit. We have toolkits for fundraising and advocacy. We are developing our peer mentor and new family welcome capacity and would be willing to share handbooks and training videos. We have experience producing international family and science conferences for over 20 years.
Project 8p	A lot of ideas brewing to kick off programs. Our conference was quite successful as a first and I am happy to share how we managed all volunteers, tight budget, and 2.5 days of agenda with clinicians and everyone meeting for the 1st 8p conference. I try not to reinvent the wheel - comradery is critical to advance all of our disorders. Working on a Commission to drive novel technologies for neurodevelopmental CNVs in its infancy stage with Dup15q.
RASopathies Network	* developed at UCLA Mattel Children's Hospital Hem/Onc Department for neutropenic children: wallet-sized cards that explain that the child (name) is neutropenic so please place the child in an isolated room if they come to your Emergency Room. * [what I wanted to do when I worked at our State Title V Program for CYSHCN (children and youth with special health care needs) but didn't get to implement before leaving: to go into in-hospital insurance authorizations: cardboard "table tents" that say, "Thank you for washing your hands before treating [name]!"] * developed business card-sized cards that explained what Costello syndrome is, for families to hand out if people gave them "funny looks" - with website that has info about Costello syndrome printed on it * paid for a "PSA" animation to explain the RASopathies in 5 minutes - useful for families and researchers

	* invite clinicians and professionals to talk about their subject matter related to the RASopathies - record and share link on website * share (vetted) strategies learned from families on caring for the RASopathy-related feature (e.g. g-tubes) as well as resources from communities that coalesce around the specific feature, e.g. the Oley Foundation https://oley.org/ for enteral and i.v. feeds, etc.
Ring14 USA	We have not really developed any resources to share. Although we do have a patient registry in Italy which is maintained through physician entered data, we would like to develop a patient registry here in the States that is patient/caregiver entered. This initial survey could certainly be borrowed from another organization. Our child what multiple seizure types which are often hard to identify. We are interested in shared repository of seizure identification videos. We are most definitely interested in a rare epilepsy center of excellence!
Shay Emma Hammer Research Foundation	a registry questionnaire
SynGAP Research Fund	1. Warrior Wednesday. We release a patient story with pictures every Wednesday 2. Medium Channel: By parents for parents. Provide useful content for families We are in the process of developing other programs. Here are some areas of focus a. Resource section of the website by state (access to services) b. Info packet for new families c. Info packet for neurologists d. Awareness Campaign - Promote genetic testing and how important it is to get the right diagnosis d. Peer to Peer webinars: Facilitate discussions between families on key topics affecting day to day life e. Meet the scientists - presentation from scientists working on Syngap
The Brain Recovery Project: Childhood Epilepsy Surgery Foundation	IEP training; school training; IEP webinars educational guides functional outcomes guides
Tuberous Sclerosis Alliance	The TS Alliance has developed many programs to provide support for the TSC community including: https://www.tsalliance.org/individuals-families Contact Dena Hook, VP Support Services for more information at dhook@tsalliance.org
Wishes for Elliott: Advancing SCN8A Research	Partnership with AES to sponsor early investigators benefiting both from comprehensive outreach, rigorous peer review, and shared funding of highest scoring proposals Hosting of early scientific meetings to develop synthesis on state of knowledge of SCN8A resulting in consensus publication, new collaborations, and 1st article in Gene Review

APPENDIX G. RESEARCH PROGRAM DEVELOPMENT

Organization	**Describe how you developed your research program and something you knew or understood that you would do differently now.**
Batten Disease Support and Research Association	Our research program has been around for the last 30 years. It started with small grants to fund postdocs or small projects. The organizations has funded over 7 million dollars in that time. Now we are working on making sure we are not reinventing the wheel. If we know that a project has been done already we will not fund a similar project unless we know that something significant has changed. This involved making sure the community is aware of all the research and results of said research in the field.

BPAN Warriors	We are in the early stages of establishing our research program. We were very strategic in our approach as to avoid duplication of any efforts and ensure that funding was well used for projects that would yield best possible outcomes for therapeutic advances. Given that our organization was established in 2018 as a family support organization and only in the last six months focused resources towards research, we believe we are making great progress. We always would like to move more quickly; however we have learned to use the lessons learned from other rare disease communities to support our strategic plan in an efficient and time-sensitive manner. 1) Comprehensive review of existing research landscape 2) Assess the existing research - luckily we have a great corporate partner, Genomenon who provided us access to their web-based research tool, Mastermind, which allowed us to collect all the existing research on BPAN and any other related gene and/or disease in one platform 3) Before we could develop any type of research program we had to understand the disease state, identify what was known and what was missing in existing research • Cellular/Molecular Process/Pathways • Animal Models • Assays • Natural History/Patient Registries/Bio-Repository 4) Review Identify researchers who have studied BPAN; develop database on existing researchers 5) Currently we are looking at parallel tracks of research in both therapeutic repurposing of existing drugs to slow the progression of the disease and reviewing BPAN isoforms for potential targets for gene therapy. Given the silos of information and limited dissemination of research and clinical care guidelines, we are actually pleased with the progress that we have made in assessing the science.
Bridge the Gap - SYNGAP Education and Research Foundation	Found researcher working on the gene and found others that were interested.
CFC International	We began our natural history study of seizures because we began to review anecdotal data from parents of our children and found that mortality was uniquely linked to seizures, even though published data was unclear. We had a very organized group of funders ready to take on this study, but we should have spent more time really reviewing the procedures for funding. We had limited time, and we just jumped in.
Chelsea's Hope	Since its establishment in 2007, Chelsea's Hope has raised about $1 million for research. At first all money raised (through activities such as live and online auctions and a few small grants) was directly donated to the labs of Dr Delgado-Escueta (UCLA) and Dr Berge Minassian (formerly at Sick Children's Hospital in Toronto, now at UT - Southwestern). In 2014, the first International Lafora Workshop was conceived and organized by Chelsea's Hope with a considerable boost from Jack Dixon, PhD, a distinguished biochemist at UCSD whose lab was one of the few in the world working on various aspects of LD. $40,000 was raised by Chelsea's Hope to fund the conference. Jack Dixon's lab identified and invited participants from North America and Europe and planned the scientific agenda. Chelsea's Hope planned and organized meals, lodging, and made travel arrangements for participants. Researchers who had previously been in competition with one another recognized potential synergies that had not been previously recognized and they were inspired at the workshop to collectively apply for a NIH "U54" grant. This initial effort was unsuccessful, but they persevered, forming the Lafora Epilepsy Cure Initiative (LECI) and in 2016 were awarded a $9 million 5 year NIH Program Project grant. This provided a tremendous boost to research on LD and progress has been accelerating ever since. International workshops are now held annually. The 6th will take place in San Diego in September 2020. Currently researchers are collaborating in a natural history study and preparing for clinical trials. Additionally there are now 3 biotech companies involved, two of which have established proof-of-concept in animal models and have drugs in the pre-clinical pipeline.
Chromosome 9pMinus Network	A family has committed to fund the initial study to use advanced gene code deciphering and computer based analysis strategies to help understand what genes are most critical in defining the characteristics that people with 9p- syndrome have.... The researchers are very familiar with the 9p- minus Network and have worked with us before. They are personal doctors to several of our Network's children. We are very lucky to have a family that can fund this project. We hope to have this research and sample gathering beginning in the 1Q of 2020.
CureSHANK	CureSHANK has a singular purpose: to accelerate the development of treatments for SHANK-related disorders. We launched CureSHANK with the intention of overcoming critical barriers to successful drug development in our disease area.
DDX3X Foundation	Our research is driven by the interest of researchers and has developed organically. Our first significant study was conducted by Sherr Lab at UCSF. Elliot Sherr was one of the first doctors to diagnose a child with DDX3X syndrome. After seeing several patients with DDX3X, Dr. Sherr began a research study in 2016. At the same time, other research was being conducted by scientists in Europe, yet not in collaboration with the Sherr Lab. In 2018, the Seaver Center for Autism also decided to undertake research in regard to DDX3X. That same year we brought these researchers, as well as other scientists, together to discuss progress and challenges related to this syndrome. Our organization's main goal is to serve as advocates for those impacted by DDX3X syndrome. We play a critical role in the development of research by identifying clinical needs and defining the path to a cure. We intend to take a more active role in setting research priorities and directing patients to appropriate studies. We strive to collect and maintain critical care resources such as important contacts and knowledge related to disease progression to better serve the needs of this small but important population.
Doose Syndrome Epilepsy Alliance	This was started about 10 years ago and has gone dormant. I wish I would have collaborated with other rare disorders to get more "bang for our buck" with similar diagnoses. I would have engaged clinicians that could carry it forward, not just the research institution. Over the years we could have had a lot of genetic samples given.

Dravet Syndrome Foundation	We developed our program under the advice and oversight we received from Dr. Jack Parent at the start of our organization. As the program and our resources have grown, we have added other arms to it to meet the needs of our entire professional community, including our clinicians. The development of these arms has been done with input and oversight from our Medical and Scientific Advisory Boards.
FamilieSCN2A Foundation	We developed our research program through close collaboration with scientists involved in the space and our SAB. The goal was and still is to be stay true to our mission of finding curative treatments.
Glut1 Deficiency Foundation	We have typically used our excess funds to support our research grant program. There is never any guarantee of how much money we will have to give each year, and we are always mindful of finding the balance between being conservative and holding some reserves over vs. putting the money to work right away. We struggle with knowing if it is better to give small amounts as we are able, or save it for a larger pool of funds, which may be more impactful. We have also relied on researchers to come to us with the ideas of how to spend our money, and I wish we could be more strategic in deciding what our community thinks our research priorities should be, fundraising specifically for those projects, and then helping make them happen with the researchers willing to help us with those priorities (rather than their own). We are looking into models for a collaborative research network that use this approach.
GRIN2B Foundation	We developed our grant application in conjunction with medical researchers on our Foundation's Medical Advisory Board. Through this collaboration we developed a grant application. The application process yielded us with 7 applicants from around the world, all competing for $45,000 in grant monies. To help our Foundation vet these applicants, we offered a small stipend and created a medical research liaison. This individual reviewed the applications and organized them for us to review. Our Medical Advisory Board also offered their recommendations In the end, we awarded a grant in the amount of $56,000 to an individual tasked with discovering GRIN2B-Related Neurodevelopmental Disease Biomarkers through patient EEGs.
Hope4Harper	I fell into our research. As described earlier my mother in law saw Dr Frances Jensen on 60 minutes discussing seizures in infants and mentioned to me that I should reach out to her. I ended up on a waiting list and then with an appointment 2 weeks later when Harper was about 4 months old. After a multitude of tests and an in depth evaluation we were sent home to wait on results. When Harper was 9 months we received a phone call from Dr Jensen with the results. Harper had CDKL5 and there was nothing we could do to help her. I plugged myself in to the only online group there was because at the time in 2011 there were only 600 known cases worldwide. After plugging myself in to the group and seeing patterns in Harper and others with CDKL5 I reached back out to Dr Jensen 6 months later asking if her research lab would be interested in helping me find a treatment for what I saw as Harper's biggest symptom, seizures. It wasn't until many months later when we were at Texas Children's in Houston and I asked their Genetics department if they would be willing to research CDKL5 that I understood how obscure it was that a mom could ask a research scientist to do a research project and it be taken seriously. I am glad I did not understand this because then I may not have asked and CDKL5 might still not have a brain to use in research. So you have more of an understanding on the uniqueness of this, 8 children passed away the year before Harper did. That is 8 opportunities to advance science that passed because nobody would talk about the importance of organ donation. Nowhere in any CDKL5 organization or group was this information disclosed. Nowhere was it acceptable to ask about life expectancy and as a matter of fact when that question was brought up it was removed by the admin of the group.
International Foundation for CDKL5 Research	We were mentored in the early days by Rettsyndrome.org - they told us what they would have done differently and that led us to quickly find a partner for a caretaker driven natural history study. We also had a roadmap to a cure, much like everyone else, that we used to educate our community about the amount of work we need to accomplish. A CDKL5 patient was invited to the Denver Rett Clinic and shared their follow-up report/experience with our organization. That is where our Clinical Centers of Excellence model was born. In hindsight, the infrastructure to support the collection of data should have been thought out before data collection started. We are moving on to 2.0 and satisfied with work accomplished under multiple data collection sites despite no centralized collection point. We now are focused on correcting that so our clinical research can move forward efficiently. We would also require a lay summary of all research funded in our contractual terms.
LGS Foundation	Started with small seed grants to young and established investigators. Also open to PhDs and MDs. We got great advice from others who already had research programs (like TS Alliance and others) so we feel like we did it right the first time. It's working.
Lissencephaly Foundation Inc	We have not launched our program yet; we are aiming to start our research program in 2022
NORSE Institute	From the very start, my emphasis has been on collaboration among clinicians. I have only recently expanded that to the collaboration among clinicians, basic scientists and patient/families. Clinicians want translational research to identify effective treatment. Basic scientists need help to identify what to test in animal models and they need human bio samples to test genetics and other human processes. Clinicians and scientists must have a dialogue and include families because they are the ones who will donate specimens and money.
Phelan-McDermid Syndrome Foundation	We are in the process of redeveloping our new Strategic Plan for Science. We accomplished most of what we set out to accomplish in our first 5 year plan.

Project 8p	We don't have a formal research program. We are just getting our feet wet and it is basically me. I have learned a lot in less than 1 year and it is that many brilliant researchers or clinicians do not have experience in driving research to the clinic - and that could waste years in the lab and years meeting patients without actually improving their lives. Truly vetting out whether researchers can understand what is meaningful strategy to get to clinic - whether it is an n=1 trial OR knowing what is a "Waste of time in the science" and taking advantage of innovative methods today doesn't really exist. Everyone works in their own silo and the PAG leader in my case is responsible for hand holding a true collaborative effort. I learned that I cannot rely on the scientists yet - we are NOT the low hanging fruit and I need to make our patients relevant for them. I need to have them see and experience what the disorder means and connect them to real patients. There is a steep learning curve here if we want to have treatment options on the table in our lifetime.
PVNH Support & Awareness	Our research program is in its infancy, still growing funds. We approached researchers with the goals of having them working together with our community. Many are all in, but a some were not good players and are not being easily opened to collaborations with other researchers and therefore we have had to make choices. In the end, those that stick with us and are open to collaborations are those that are moving the needle forward, ever so slowly though.
RASopathies Network	[Other parents and I follow (dog) the researchers/clinicians who show interest in the syndromes
Ring14 USA	Our research program was developed by hosting international research workshops where we handpick and invite specialist in fields that we think would be fruitful to explore. These experts had no knowledge of Ring14 Syndrome, but we enticed them to share their expertise and consider ways that it might be applied to Ring14 research by providing a trip to Italy, where Ring14 International is housed. Honestly, this has been a very effective means of engaging scientist who had no interest in exploring Ring14 as a new research interest. The conference is typically held at a secluded location, which gives us the opportunity to develop personal connections with the scientists. We then invite these participants to apply for our seed grants. About 30 percent of the participants follow through. We have also expanded our Scientific advisory board through participation in these workshops. This might seem extravagant, but with at least half the participants being European, it really is not. This strategy might only work for an international program, but point is that you have to actively go after the best and it help to offer them a unique experience. This has been great to encourage new scientists to take an initial look at our syndrome - the problem has been getting funding beyond our seed grants. I wish I had written more about the outcome of these workshop in lay-term so that the value of these workshop was appreciated by our patient population.
The Brain Recovery Project: Childhood Epilepsy Surgery Foundation	Our research program is by invitation only based on what the CEO believes should be researched. We now have a scientific advisory board and will develop a formal grant program for identified research goals.
The Cute Syndrome	Our research program began with small seed grants and we still continue with seed grants and see the importance of them. We developed into having a gathering with clinicians, researchers, and families in conjunction with AES. This gathering has developed over the past 4 years to a place for SCN8A professionals to share new developments in their research. This summer we had our first researcher roundtable where we had a small group of invited researchers and clinicians present and share their findings and have detailed SCN8A discussion on where new developments are needed. This area is one where we, TCSF, excel. Constant communications and developing relationships between the SCN8A professionals, volunteers, and families has been integral. I wish that we had researched and created our own, TCSF, registry from the beginning.
Tuberous Sclerosis Alliance	The first research grant was funded in 1984 to a researcher at the University of California, Irvine. In 1992 the organization testified before Congress about increased research funding for TSC and in 1998 introduces formal research program and funds $886,741 in research grant awards. In 2001, the TS Alliance obtains support from Congress to start the TSC Program in the Department of Defense Congressionally Directed Medical Research Program (TSC CDMRP). In 2002, partners with NIH to hold a scientific conference to produce a comprehensive research plan for TSC. In 2017, the TSC CDMRP receives another $6 million appropriation, bringing the cumulative funding to $71 million since 2002, thanks to its successful grassroots efforts. The TS Alliance also cultivates relationships with private donors who pledge major dollars toward research.

APPENDIX H. ADVANCING RESEARCH IN YOUR DISAESE

Organization	**If you were advising a new rare epilepsy organization, list and describe the first, second, and third initiatives you would undertake to advance research your disease.**
Batten Disease Support and Research Association	1. A natural history study that is owned by patients and that collects a broad spectrum of data so it can be usual for many studies and for many years. 2. Understanding the mechanisms of the disease 3. Biomarkers

Bridge the Gap - SYNGAP Education and Research Foundation	#1Stakeholder Conference #2Natural History Study #3Centers of Excellence
CFC International	Constantly ask yourself the question, "what would I do if $1 million for research dropped in my lap tomorrow?" Even if it seems like a dream, you need to be ready. When the funding comes, you have to know what you're going to do with it immediately.
Chelsea's Hope	Identify all of the researchers currently working on some aspect of the disease and find a way to bring them together. In the field of LD research, the synergies discovered at the first workshop were astonishing and it was a profound catalyst to research progress.
Chromosome 9pMinus Network	1) find the correct doctors 2) Funding 3) set reasonable objectives
CureSHANK	1. Learn as much as possible about the science related to your disease (and related diseases). 2. Ask a lot of questions about drug development in your disease and identify what the critical bottlenecks are. 3. Invest in projects that address the critical bottlenecks.
DDX3X Foundation	1) Understand the experiences of other organizations. We have much to learn from other's successes and mistakes. Incorporating this experience is enormously helpful and saves time and money. 2) Know who your patients are and build a tight-knit community. Our community is our biggest asset. Not only is it the source of our fundraising, but our parents volunteer considerable time and resources, spread awareness and are motivated to travel to participate in studies. 3) Form a scientific advisory committee with clear deliverables and meeting times.
DNM1 dynamos - Connecting DNM1 Families	Find the associated researchers. Contact them tell your story- make their work more meaningful
Doose Syndrome Epilepsy Alliance	1. Discover the interest of patients and clinicians 2. Find the right research institution 3. Disseminate results for improved outcomes
Dravet Syndrome Foundation	1. Fundraise and make seed funding available for researchers. 2. Have a presence at meetings to make sure that your disease is being represented. 3. Recognize that it is not uncommon, even in rare diseases, for there to be multiple organizations that are supporting a single rare disease. Egos need to be pushed to the side and the patient leaders need to work with one another collaboratively if they hope to make headway. There is no benefit to divisive or duplicate efforts when there are limited resources - both financial and patient families.
FamilieSCN2A Foundation	1) registry that can evolve into a NHS and you have control over 2) create a strong SAB of professionals with different backgrounds 3) do not throw $ at any research - be strategic and have a thorough review process of each application
Glut1 Deficiency Foundation	1. establish a registry 2. determine natural history 3. engage community to determine research priorities
GRIN2B Foundation	1) I would first advise the new organization to develop clear goals for their research. What does their organization want to get out of this research and how will it, ultimately, help patient and families? 2) I would encourage the organization to create a medical advisory board of interested physicians that can act as council for the organization for research. These individuals can be persons who research the disease or simply trusted physicians with experience in analyzing medical data and correspondence. 3) 3) I would create a fundraising goal for the organization so their research proposals can be properly funded.
Hope4Harper	1) gather parents - organize them but do not try to control them. Allow for open discussion and kindness. The only thing you should expect from them is respect for one another not agreement with one another. Become a resource of complete and total knowledge. What to expect, what resources you will need, specialists you should be seeking out, etc... 2) find researchers that are researching a similar disorder. You do not have to have a website or nonprofit status to start. We began by connecting with Dr Jensen who then connected us to the hospital that helped us set up a donation page on their hospital site and routed money through them to Dr Jensen's lab while we began the process of becoming a nonprofit. 3) Try to encourage parents to form organizations by regions/countries. Example: CDKL5 UK, CKDL5 Brazil etc....
International Foundation for CDKL5 Research	1. Natural history/contact registry 2. animal models 3. Find research clinicians - they are rare gems
LGS Foundation	1. Bring the community together and try to get some consensus on a research plan/vision. 2. Fund small seed grants to address that plan/vision. 3. Grow your funding dollars, consider direct funding to solve a problem, and convene the community again every 2-3 years.

Phelan-McDermid Syndrome Foundation	1. Locate researchers in your disease and related disease. Let them know you have organized the patient community and that you will help them in collecting data and recruitment. 2. Develop a 3 years research plan based on the recommendations of your Scientific Advisors. 3. Develop a patient registry (at a minimum start with contacts)
RASopathies Network	do what you're doing: come together, share stories, identify resources, seduce clinicians and researchers...
Ring Chromosome 20 Alliance	1. Review all the existing research & literature from beginning to most recent, looking for what has already been proven in research and how next research questions can be built on what is known scientifically already. 2. Compile a master library of the available literature and research (no patient identification) on the rare epilepsy and distribute/give access to medical & genetics students, interns, residents, neurologists, geneticists. 3. Create a patient registry with patient permission to de-identified data.
Ring14 USA	1) Engage a clinician first to start defining the syndrome through a natural history study. 2) Develop some kind of a repository for researchers to access, either a biobank of sample or a clinical database, to make their job easier by letting them focus on science nor recruitment. 3) Find someone passionate about your syndrome who speaks science. You have to have a bridge between the families and the science; someone who can speak to both groups.
The Cute Syndrome	1. Fundraising 2. Begin Natural History study 3. Patient Registry
Tuberous Sclerosis Alliance	1) convene researchers and patients via conferences and workshops, 2) build a patient registry with ability to re-contact participants, 3) do more of #1 and include program officers from NIH.
Wishes for Elliott: Advancing SCN8A Research	1. Convene scientists for a dedicated scientific exchange; promote new collaborations across specialties and related disease groups 2. Support (facilitate/fund) publication by gathered scientists on a consensus paper on the " state of the science" in your disease area including critical gaps and emerging priorities 3. Commit to funding early investigators annually; not only build knowledge but nurtures the development of a growing pipeline of new investigators dedicated to research related to your disease Mostly recognize you can't do everything. Look for critical gaps or targets of opportunity where you can maximize your impact despite what are always limited resources and capacity. Set clear priorities. Invest in collaborative efforts to make progress on larger challenges where objectives/interests are shared and more can be achieved by working together.

APPENDIX I. ANIMAL MODEL IMPACT ON DISEASE UNDERSTANDING & THERAPY

Organization	**Describe how the animal model has been effective at advancing understanding of your disease and/or advancing the development of therapies? Describe any limitations.**
Batten Disease Support and Research Association	Discovering a naturally occurring dog model for one of our forms has allowed many preclinical studies to be done that eventually led to the current treatment. The challenges were making sure they were available to other researchers (outside the home institution).
BPAN Warriors	Fortunately, we have several mouse models, however the two that are most similar in phenotype to BPAN patients are documented in research from China. Although we have explored the option of accessing these mouse models in China, our consultants at Jackson labs have indicated that transfer of such mice might be a difficult process. Fortunately Jackson labs also houses a KO mouse in house and believes that we can age out the mouse and run testing to see how closely they resemble BPAN symptoms. There is a forth KO mouse that our patient community funded through the NBIA Disorders Association in 2015, however, to date we cannot get any information about the mouse and not papers have been written as to how the mouse presents symptomatically. This is area of great interest as we are currently in the process of identifying potential isoform(s) for targeting in creation of a vector for gene therapy. We are at the same time ensuring that the mouse model is available for testing should gene therapy be a possible therapeutic path.
Bridge the Gap - SYNGAP Education and Research Foundation	Yes, finding mechanisms and translational science https://www.ncbi.nlm.nih.gov/pubmed/31395010 https://www.ncbi.nlm.nih.gov/pubmed/30455457
CFC International	The CFC Syndrome mouse model has been effective in researching Mek Inhibitor use to combat cardiac manifestations in the disorder. This could lead to clinical trials in humans with CFC Syndrome and has already led to trials in other Rasopathy disorders.
Chelsea's Hope	Laforin -/- and Malin -/- knock-out mice have been developed and are made available to LECI researchers at multiple sites. Double knock-out mice, lacking both laforin or malin PLUS an enzyme necessary for glycogen synthesis in vivo, are normal. Since Lafora disease is caused by abnormal glycogen accumulations in brain cells, blocking glycogen synthesis rescues the disease in mice. This has opened up numerous lines of research including Anti-sense oligonucleotides (ASOs) and small molecules designed to down-regulate glycogen metabolism.
CureSHANK	The animal models have been useful for understanding the disease mechanisms. Behavioral assays in mouse models may have limited value for understanding human disease phenotypes. Clinically translatable biomarkers might be a useful alternative to mouse behavior. The animal models have also been useful for identifying a handful of lead compounds that might be effective, including a few that have already moved into clinical trials.

	We have 15 mouse models of our disease. The genetic constructs are different, the mouse backgrounds are different, and the assays are different. We don't know if variability in key findings can be attributed to real differences resulting from specific genetic constructs or if (or how much) the variability in findings is related to all the other sources of experimental variability.
DDX3X Foundation	The mouse model was developed by the Seaver Center for Autism. Preliminary evidence suggests that there is a consistent phenotype. However, it is too early to test therapies on the mouse model. A second mouse model is being used at the Silver Lab at Duke to understand prenatal brain development. Again, results are not yet available.
DNM1 dynamos - Connecting DNM1 Families	One of the ways animal model has been informative is that the DNM1 encephalopathy leads to behavioral comorbidities and developmental delays independent of epilepsy.
Dravet Syndrome Foundation	We have multiple animal models, all of which have been helpful in understanding our disease. The zebrafish and drosophila models have been beneficial as they are far less expensive to maintain than the rodent models.
Glut1 Deficiency Foundation	The mice models have given a great deal of insights into the disease mechanisms. They've been used to test hypotheses before moving to human trials for treatments. They've also been used for functional studies for specific mutations. Our researchers always remind us that mice are not men. A couple of studies have not shown the same results in humans that we see in mice.
Hope4Harper	The animal model was not initiated by our organization. The animal model is challenging for our purpose because we desire to treat symptoms associated with the disorder beginning first with the seizures but the mice model does not seize.
International Foundation for CDKL5 Research	the R59X has been used around the world
KCNMA1 Channelopathy International Advocacy Foundation (KCIAF)	Because the majority of the gene mutations are de novo, we expect to use the animal models to first determine the causal relationship between KCNMA1 mutations and disease symptoms.
Phelan-McDermid Syndrome Foundation	(This is from a 2016 PMS Research Landscape Report) Since the publication of the first Shank3 mouse in 2003, there has been a strong commitment of the PMS field to continued animal model development and involving a range of different species. The success of model development in the PMS field, particularly efforts that have emerged around Shank3, has been a real strength of the PMS community. It has been a game-changer in terms of influencing the pace of research and enabling translational work to take hold. For example, the treatment hypothesis surrounding IGF-1 was evaluated in murine models of Shank3 before advancing into clinical trials for PMS (oxytocin, which is currently being evaluated in PMS subjects, was also investigated in a rat Shank3 model, but published after the trial had begun). When evaluating the PMS research space to assess the current state of animal model development related to PMS, there is good news to share. Like many rare neurodevelopmental disorders with known, or suspected, genetic etiology, the field of research around PMS has benefited by having genetics to lean on when it comes to animal model development. Much of this work has focused on SHANK3 in the neurological components of PMS. The PMS-research community has developed a wide range of animal models of PMS based on the genetic risk factor SHANK3. Across most of these models, there is good face validity to the diverse neuroanatomical, physiological and behavioral phenotypes for the clinical presentation of PMS In recent years there have been primates models developed as well.
Project 8p	Difficult for animal model for this disorder. hence looking into neurons/organoids first.
Ring14 USA	So far it hasn't been very effective. We tried to achieve a somewhat comparable mouse model, but it was not viable - plus it was poor comparison since the genetic material on the 14th human chromosome maps to parts of mouses 14th and 12th chromosome. So ring 14 mouse would not the analogous genetic material as a ring14 human. Dr Spinner has endeavored to turn a ring14 IPSC line into an organoid in her NIH grant but we have heard it was not successful. We actually have several stable IPSCs lines, but they have yet to use in a study that has been published.

APPENDIX J. BIOSAMPLE REPOSITORIES

Genes	Please specify the name of the repository (ies).
15q11.2-13.1	• Coriell and Rutgers • UConn
22q13 Deletion Ring 22, SHANK3 mutations	• Brain tissue at Autism Brain Net • iPSC resources at NIMH Stem Cell Repository • NIMH Rutgers Collection - https://www.rucdr.org/ • CIRM - https://www.cirm.ca.gov/
Brain Recovery	• We do not serve a specific disease; however, UCLA houses a biosample collection for Rasmussen's encephalitis which requires hemispherectomy.
chromosome 8p	• we are in talks with Corielle, RUDCR, and UConn Core
CLN1, CLN2, CLN3, CLN4, CLN5, CLN6, CLN7, CLN8,	• Massachusetts General houses our biosamples

CLN10, CLN11, CLN12, CLN13, CLN14	
DDX3X mutation	• Seaver Center for Autism and Sherr Lab at UCSF. There is no centralized biorepository.
DNM1	• There is no repository of biosamples. However biosmaples from few patients are collected by TGEN for research purposes - to develop Zebrafish models and STEM cells (IPSCs)
Doose	• Manton Center for Orphan Disease Research and Kings College in London have samples.
LGS	• There are many repositories for etiologies but again nothing SSW or GPFA specific. We are glad these exist but mostly leave them to the gene and cause-specific groups to run. The only real repository is from Epi4K and the exomes of the LGS patients in there. They may also have some blood stored, but not much.
NORSE	• The NORSE prospective study done through the consortium (CCEMRC) houses the bio samples at Yale
ring chromosome 14	• Telethon Network of Genetic Biobanks (TNGB, http://biobanknetwork.telethon.it) and, in particular, the Biobank located at Galliera Hospital in Genoa http://ggb.galliera.it/ • Anthony Wynshaw-Boris at Case Western has stable IPSC cell line available
ring chromosome 20	• Children's Hospital of Philadelphia
SCN1A	• The two repositories are only collecting brain tissue, after death - NASR (North American SUDEP Registry) and STOP SUDEP Registry.
SCN2A	• Simons has a biobank of SCN2A patient's iPSCs, blood, saliva
SCN8A	• CNMC Dr. John Schreiber • University of MI
SLC13A5	• Coriell
SLC2A1 gene	• Coriell
slc9a6	• Dr Eric Morrow - The Warren Alpert Medical School of Brown University
STK9, CDKL5	• Harvard Brain Bank • Dr Allyson Moutri's stem cell research lab houses the IPS Cells and is beginning the creation of the organoids.
SYNGAP1	• Texas Children's Hospital
TSC1, TSC2	• Van Andel Research Institute
WDR45 / WIPI4	• Friedrich-Baur-Institut and Oregon Health and Science University - we have been told that the biobank will be used to advance NBIA research, enable faster access to worldwide clinical trials and draw greater interest from potential funding sources to bring a drug to trial, however neither institution has draws samples from the other and the IRB process is prohibitive. • We are now establishing a centralized bio-repository including iPSC cell lines at the Coriell Institute so that the biosamples are more readily accessible. • There are also brain bio-samples that are stored at both OHSU and other institutions. Also, we fully concede that there may be many other patient samples that are yet stored in other institutions and academic settings that we have no knowledge of yet. This will be part of the learning curve of establishing a centralized biorepository.

APPENDIX K. KEY MESSAGES TO PROFESSIONALS

Organization	What is the most important message your target groups need to know/learn about your disease?
Batten Disease Support and Research Association	early diagnosis is very important for quality of life and treatment options
BPAN Warriors	Symptoms should not simply be dismissed because of BPAN diagnosis. Although treatments/therapies are not available currently, this does not mean that underlying issues such as hormone imbalances, vitamin deficiencies, endocrine issues, metabolic dysfunction, GI issues, irregular sleep, etc. cannot be addressed. We are at such an early stage in understanding BPAN and it is critical that additional testing and evaluation be performed to get to the root cause of many of the patient symptoms. Listen to your patients/caregivers and recognize that they have insights to the disease and symptoms. They may not know the diagnostic path to address some of the seemingly "odd" or unrelated issues, however it is your duty to provide critical guidance to families who are just doing their best to survive. Please do not dismiss, doubt, shame, or second guess, patient caregivers when so little is known about BPAN. We are learning as we go and need support from professionals not push back.
Bridge the Gap - SYNGAP Education and Research Foundation	Get Patients to Register for the Natural History Study
CFC International	We struggle to get providers to understand that all malformations are attributed to CFC Syndrome. Because of our name, providers often assume that the only complications are heart, facial, and skin related. They often see things like Cerebral Palsy and Seizures and uncommon and unrelated.
Chelsea's Hope	LD is usually initially misdiagnosed as the much more common Progressive Myoclonic Epilepsy. Up until now, delayed diagnosis didn't cause a great deal of harm since there was effective therapy. However, now that clinical trials will soon be underway, early diagnosis will be of critical importance. All pediatric epileptologists must be made aware that genetic testing should now be done immediately for all patients with adolescent onset seizures.

Christianson Syndrome Association	How differently it can affect each person and to really listen to what is being told to them. These boys have a mold of their own and the doctors really need to think outside the box when treating them.
Chromosome 9pMinus Network	Also called Alfi's Syndrome. Every child with the 9p- chromosome is different but most share common abnormalities.
CureGRIN Foundation	GRIN Disorders affect NMDA receptors. NMDA receptors affect other Neuro-Developmental Disorders like Alzheimer's, schizophrenia etc...so maybe getting them to understand that piece could help bring more familiarity with GRIN.
DDX3X Foundation	DDX3X is believed to be the most common cause of intellectual disability in girls yet there are less than 500 cases that have been identified in the world. Our children present very differently, so it is critical that genetic testing is undertaken to diagnose patients.
DNM1 dynamos - Connecting DNM1 Families	Do Literature Survey, follow ongoing research, engage and help advance it
Doose Syndrome Epilepsy Alliance	Early diagnosis and treatment leading to better outcomes.
Dravet Syndrome Foundation	While early diagnosis is ideal, diagnosis at any age is vital to assure proper treatment and quality of life.
Dup15q Alliance	That networking with other clinicians helps to create confident care providers, it is not a sign of weakness
FamilieSCN2A Foundation	A very clear phenotype / genotype correlation has been validated and pharmacological response can be predicted. Before recommending a treatment course, this should be taken into consideration.
Glut1 Deficiency Foundation	Any patient with suggestive symptoms across a broad range of severity should be tested - there is a very effective treatment in the ketogenic diet.
GRIN2B Foundation	GRIN2B-Related Neurodevelopmental Disorder may be rare, but it's most likely not as rare as many think. GRIN genes affect how we think and interact with the world and may be the key to unlocking the mysteries behind other more common diseases such as epilepsy and autism spectrum disorder. Our goal is to educate everyone about the complexities of this disease so that someday researchers and doctors will take the science behind GRIN genes and NMDA receptors further toward potential treatments.
Hope4Harper	Organ Donation and CBD advocacy are my top two messages. Acceptance is not defeat. It is the place you need to get to in order to move forward from where you are.
International Foundation for CDKL5 Research	It has a wide phenotype and affects both boys and girls. It can also be hereditary although most cases are currently presumed to be de novo
Jeavons Syndrome Facebook Group	There is a variety of traditional and non-traditional ways to deal with the symptoms of Jeavons.
KCNMA1 Channelopathy International Advocacy Foundation (KCIAF)	That there is no precision medicine solution and we need practitioners to communicate to our organization and to each other what the defining symptoms and the best evidence-based therapies are.
KIF1A.ORG	Genetic testing is required to discover/confirm diagnosis.
LGS Foundation	It evolves. Target it early to stop evolution. Research needs to go that direction.
Lissencephaly Foundation Inc	There is often time a gap in DX as other options are looked into first when seizure first present. It is also often time missed during pregnancy, as it's not screened for during ultrasound.
Mickie's Miracles	If a child fails the first form of epileptic treatment, they must be seen by a Pediatric Epileptologist. These doctors have a higher level of understanding epilepsy than a neurologist or pediatrician. When the pediatrician, specialists and neurologists exhaust their medical expertise, a child must be referred immediately to a higher level of care at a level 4 epilepsy center.
NORSE Institute	Early diagnosis of NORSE/FIRES. Importance of early treatment.
Phelan-McDermid Syndrome Foundation	That they need to come to us, the patient advocacy organization for information. When they have updates they need to provide them to us to we can update our families. Having publications does not mean families are getting the information!
Project 8p	We don't know enough and the only way to better understand the disease is to participate in research and by sharing our experiences in our natural history study and exchange useful information.
PVNH Support & Awareness	That PVNH is a very wide spectrum that creates many, sometimes life-threatening comorbidities in a high number of patients, and therefore proper referrals are critical for patient's safety.
RASopathies Network	don't be "exclusive" about your knowledge of one of the RASopathies to support a different RASopathy... the ideal doctors acknowledge not knowing about the syndrome and being willing to learn with the family
Ring Chromosome 20 Alliance	Ring 20 has been seen in as few as 5% of cells, and we recommend requesting a screen for chromosomal mosaicism. Since r(20) syndrome can present as a mosaic with the ring in only a small number of cells, a minimum of 50 cells must be analyzed. Newer array technology (CGH or SNP arrays) will NOT detect the ring chromosome and we recommend standard metaphase chromosome analysis.

Ring14 USA	That ring chromosome are best diagnosed with a karyotype. When we have an effective therapy, this message will undoubtedly change.
SynGAP Research Fund	- Who to send patients to (organization/support group) - High risk of Epilepsy - if there is no epilepsy diagnosis - monitor closely for seizures
The Brain Recovery Project: Childhood Epilepsy Surgery Foundation	Delay to surgical evaluation is still a problem.
The Cute Syndrome	Treatment guidelines
Tuberous Sclerosis Alliance	quicker diagnosis, appropriate treatment, better outcome

APPENDIX L. CLINICAL GUIDELINES

Organization	Link to Guidelines	Describe the development of guidelines (diagnosis, evaluation, treatment or other) for your disease. Were they patient or professional driven? Are they adopted across state and international borders? Are they recognized by insurance other regulatory agencies?
Batten Disease Support and Research Association	The above only exist for 1 of 13 forms of the disease	The guidelines for 1 of 13 forms of the disease were spearheaded by industry with input from professionals for around the world.
BPAN Warriors	https://www.ncbi.nlm.nih.gov/books/NBK424403/ There are "treatment" guidelines, however they are for the treatment of some of the symptoms, rather than the disease. As for diagnosis and evaluation guidelines, they exist, however we cannot really say whether or not clinicians actually use them.	Not sure how the guidelines where developed, however the patient community has not been part of the process (other than we have provided the data via patient visits at clinic and patient family conferences). There is very little interaction between OHSU and patient community outside of their clinic. This is an area of great concern to our organization as there has been very little patient community input/interaction outside of the clinic and conference. As for international adoption, we have not been able to assess, however clinical care overall is abysmal for our patient population. And a resounding NO for insurance and regulatory agency adoption of guidelines, due in part to the range in symptoms and severity of symptoms, the fact that the disease is relatively newly discovered (2012) and that our patient population was less that 100 two years ago - now we are somewhere between 500-2000.
Bridge the Gap - SYNGAP Education and Research Foundation		Professional driven, they are new and we are currently trying to spread awareness. We are still working on our ICD-10 Code to have insurance cover the cost of this disorder
CFC International	https://rasopathiesnet.org/wp-content/uploads/2014/01/CFC-Parent-Guide_R4.pdf	The guidelines were professional driven and mostly led by our Medical Advisory Board and Geneticists. They are recognized globally and may be used for coverage to treatment for insurance purposes.
Chelsea's Hope		Dr. Delgado-Escueta (UCLA) is working on establishing clinical guidelines. To my knowledge the group is currently debating whether all patients presenting with myoclonic seizures in adolescence should be submitted to genetic testing. Since the vast majority of these patients will prove to have JME and genetic testing is expensive, they are currently debating whether or not they should increase specificity by adding criteria such as occipital seizures or occipital spikes on EEG.
CureSHANK	https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4362650/ This paper includes guidelines for monitoring. There are no approved treatments, though the practice parameters paper has some general recommendations about things like referral to speech, OT, ABA, etc. Diagnosis: https://www.ncbi.nlm.nih.gov/books/NBK1198/	The guidelines for diagnosis and monitoring were driven by professionals. They are well-accepted internationally. The monitoring guidelines do not meet the formal requirements of "consensus guidelines," but to the best of my knowledge they have not been challenged by payors.
Doose Syndrome Epilepsy Alliance		I am not sure if these exist for Doose. I think that there are clinical guidelines, the research suggests EEGs indicative of diagnosis, etc.
Dravet Syndrome Foundation	https://www.pedneur.com/article/S0887-8994(16)31037-2/fulltext	They were driven by our clinicians, led by Dr. Elaine Wirrell, and funded by DSF through an educational grant. Dr. Wirrell did include some patients in the project and it was developed for all of North America.

Glut1 Deficiency Foundation	https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5983110/	The ketogenic diet guidelines were done by a professional consortium. There are current consensus guidelines specific to Glut1 Deficiency underway now with a select group of medical professionals and patient representatives (I serve on the committee).
Hope4Harper		The development of guidelines is both patient and professionally driven. It's time that stands in the way for both. Guidelines are being developed but the process is slow.
International Foundation for CDKL5 Research		there are clinical review papers but insufficient evidence for guidelines at this time
LGS Foundation		Different for various countries. No international consensus.
Mickie's Miracles	https://www.uptodate.com/contents/clinical-features-and-diagnosis-of-infantile-spasms	The development of guidelines included/includes patient centered research studies as well as clinical trials for drugs and other treatment options. The AAP and CNN have guidelines that recognize certain treatments and evaluations for Infantile Spasms that other countries (like Canada) do not have. For instance, Acthar Gel is recognized as a front line treatment for IS in the USA but in Canada, it is not yet recognized as a treatment for IS at all.
Phelan-McDermid Syndrome Foundation		We are in the process of doing this and we are using the model that Tuberous Sclerosis provided to us to guide our process.
PVNH Support & Awareness		They are now being prepared.
RASopathies Network		Clinical Tx guidelines for Noonan (in works of being updated - UK), NF1 (US) and CS (international); not for CFC (and using Noonan one for the time being)
Ring14 USA	Rinaldi et al. Orphanet Journal of Rare Diseases (2017) 12:69 DOI 10.1186/s13023-017-0606-4	The guidelines were developed by professionals but reviewed by parents and edits to include topics we thought were relevant. So I really do think it was a collaboration. I don't think they 'recognized' by anyone, especially since they don't really offer any solutions.
The Brain Recovery Project: Childhood Epilepsy Surgery Foundation	https://onlinelibrary.wiley.com/doi/full/10.1111/j.1528-1167.2006.00569.x	Re surgical evaluation, they are generally developed by ILAE. For functional outcomes, we are developing best practices guidelines re which clinicians should be following these children post-operatively.
The Cute Syndrome	https://www.thecutesyndrome.com/reference-guide.html	Diagnosis is done by genetic testing. The treatment guidelines have been patient driven as we discovered a largely negative reaction to a certain frontline epilepsy medication. The guidelines are generally adopted internationally.