

Finding Your Way: A Parent's Guide to Rare Disease

A Step by Step Guide for Parents Navigating a Rare Disease Diagnosis

Written by parents, for parents



About Us



Adam is a passionate rare disease advocate, driven by his children Alfie and Lola, who were born with the rare genetic condition CRELD1 and participants in the 100,000 Genomes Project. To support other families navigating similar journeys and to drive vital medical research forward, Adam and his wife Jess founded the UK charity CRELD1 Warriors. Adam is also Vice-Chair for Rare Conditions on the Genomics England Participant Panel, where he represents the interests of Genomics England participants, ensuring that genomics research is clearly understood and directly benefits the patient and participant community.



Mel founded the charity Cure DHDDS after two of her three children were diagnosed with an ultra-rare DHDDS genetic variant, which causes both a neurodevelopmental and neurodegenerative condition. Recognising the lack of research and support, she established the organisation with her husband, Charlie Dixon, to drive awareness, fund research, and support affected families.

Now working full time as its CEO, Mel is dedicated to accelerating progress toward treatments. She is also on the Participant Panel at Genomics England, a Trustee of CDG UK, a member of UK Rare Epilepsies Together (UKRET), and the LSD Collaborative. Mel is a committed advocate for greater recognition and investment in rare and ultra-rare diseases.

Guide Rationale

While there are some existing resources about the rare disease pathway, the guide we're creating offers a unique and valuable perspective for several reasons:



A Personal, Action-Oriented Pathway:

The guide is structured as a clear, two-pathway model ("Finding an Existing Community" and "Creating Your Own Community"). A simple, actionable framework that is easy for overwhelmed parents to follow.



Inspired by Lived Experience:

The guide is grounded in our personal journeys with CRELD1 Warriors and CureDHDDS. This provides a powerful, relatable narrative that can inspire and empower other parents.



UK-Specific Focus:

While many resources are US-centric, our guide prioritizes UK-based organisations like Genetic Alliance UK, Rare Disease UK, and Beacon for Rare Diseases.



Direct and Simple Language:

The guide avoids jargon and presents information in a clear, concise and empathetic way, which is essential for parents who are already processing a great deal of complex medical information.

We are not reinventing the wheel but rather creating a more accessible and personalised "front door" to the vast world of Rare Disease resources.

The guide acts as a "quick start" guide, directing parents to the right places while providing a framework for their journey, all through the reassuring lens of a fellow parent who has walked the same path.

First Steps on Your Journey

Receiving a rare disease diagnosis for your child can be an overwhelming experience. While it marks the end of a journey that many refer to as the 'diagnostic odyssey,' it often marks the beginning of a new odyssey of uncertainty. It can be incredibly frustrating to find that even with a name for the condition, information is scarce, understanding among health professionals is limited, and many of your most critical questions remain unanswered.

Your GP, specialist clinician or genetics counsellor should provide you with information at the time of diagnosis, even if there is very little information available.

Please know that you are not alone. This guide offers a pathway to that journey; helping you to connect with other families who understand your experience and can provide invaluable support.

First Steps on Your Journey

Before you start looking for a community, it's important to give yourself a moment and take these initial steps:



Take a Moment to Process

Allow yourself time to absorb the diagnosis. It's okay to feel a range of emotions, from shock and grief to even relief at having an answer. This is a significant life event. Acknowledging your feelings is a crucial first step before you can move forward.



Gather Information

Work with your child's clinical geneticist, genetic counsellor or GP to understand the diagnosis. Ask for the exact spelling of the condition, how it might affect your child, and if there are different subtypes. Knowledge is empowering. The more you understand, the more confident you will become in advocating for your child.



Build Your Medical Team

Your child may need to see multiple specialists and it's important to seek multiple opinions to get a rounded view of what care is available to you, both within the NHS and privately. A strong, coordinated medical team is essential for your child's care and your own peace of mind.



PATHWAY 1: FINDING AN EXISTING COMMUNITY

For many rare diseases, there are already established communities of families who are on a similar path. Here's where to find them:



Genetic Alliance UK

Genetic Alliance UK is an alliance of over 240 organisations, charities and support groups working together to improve the lives of everyone in the UK living with genetic, rare and undiagnosed conditions. They are a great starting point for finding a specific support group. They have also published [Rare Resources guides](#) for families who have recently received a diagnosis of a genetic or rare condition, are on the journey to a diagnosis or who have been told their child's condition is so rare they might not get a diagnosis.



Unique

Unique provides support, information and networking to families affected by rare chromosome and gene disorders.



SWAN UK (Syndromes Without A Name)

Some people having genetic testing will not receive a diagnosis. This can be very difficult and it is important that you have support.

Run by Genetic Alliance UK, SWAN UK is a dedicated support community for families of children and young adults with undiagnosed genetic conditions in the UK. It also provides a range of information for families. If you don't have a definitive diagnosis, this is the community for you.



Contact

This UK charity supports families with disabled children and runs a "Rare Conditions Network." They offer a helpline, a "Listening Ear" service for emotional support, and networking opportunities.



Beacon for Rare Diseases

Beacon is a UK-based organisation that helps connect patients and families with rare disease organisations.



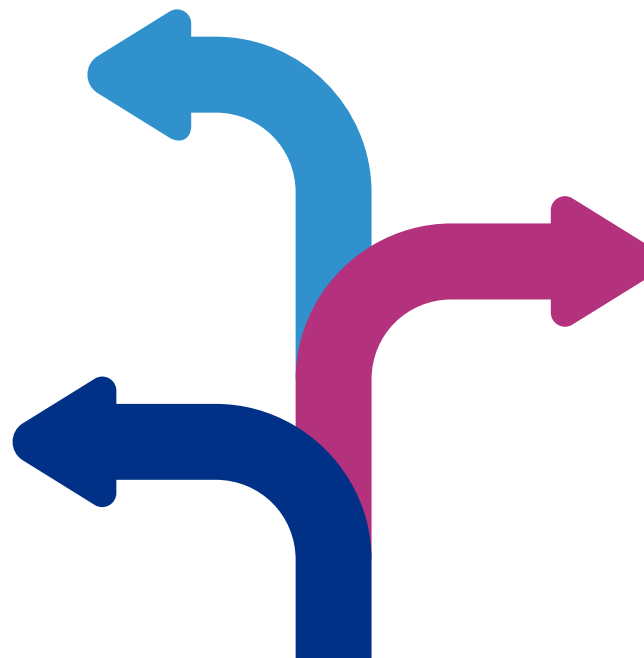
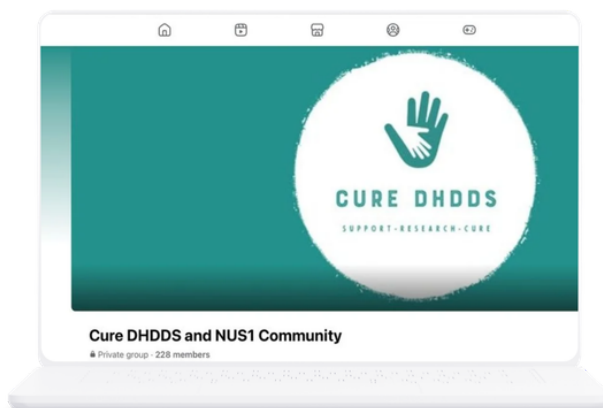
Online Support Groups

As a free informal platform, Facebook is home to a number of online support groups. Facebook is often the first place where parents seek community support and connect. These groups offer a space to share experiences and knowledge, ask questions, and receive emotional support from people who truly understand.

You don't need to have a Facebook account to search for groups, although if you do find a relevant group you will need to sign up to join and talk to people. If there isn't already a Facebook group for the condition, you can always set one up for the next person searching to find. It can be a great way to connect with others directly.

There are many groups such as 100,000 Genomes Project Support, SWAN, and numerous epilepsy support groups, where you can find other parents to connect with.

It's also worth doing an internet search for any support groups associated with your diagnosed condition. For example, it may be useful to you to search the name of your condition and the words 'patient support UK' in an internet search engine. Be careful though, as it is easy to scare yourself as often the results you find might be the worst-case scenario.



PATHWAY 2: CREATING YOUR OWN COMMUNITY



If a community for your child's condition doesn't exist, you have the power to create one. It often starts with a single family reaching out to find others.

Here are the steps you can take:

1

Start with Social Media

Create a Facebook group for your child's condition. This is a simple but powerful way to find other families who may be searching for connection.

2

Build a Central Hub

A simple website can serve as a central point for information, resources, and a place to share family stories. Creating a website for your condition will also appear in the Google search rankings for anyone searching for relevant information about the condition. Anyone can create a website and there are a number of simple and affordable website builder tools such as Wix and GoDaddy. They provide simple step by step support in purchasing a web domain and building your site.

3

Establish a Patient Registry

A patient registry is a database of information about individuals with a specific condition. It is one of the most powerful tools you can create to drive scientific research. It allows researchers to study the condition, identify potential participants for clinical trials, and develop a deeper understanding of its natural history.

You don't have to build this from scratch. Organisations like [Sanford CoRDS \(Coordination of Rare Diseases at Sanford\)](#), and NORD's [IAMRARE](#) provide the infrastructure and guidance to create and manage a patient registry for free.

By collecting patient data, you transform your community from a support network into a research-ready powerhouse, making it easier for scientists to engage with you.

4

Engage with Researchers & Industry

Build relationships with the key players who can accelerate research. Parents are the experts in their child's condition, and your lived experience is invaluable. A good place to start is by searching online for any researchers or scientists related to your rare condition.

A helpful place to begin is at relevant academic conferences related to genetics, rare diseases, or your child's specific symptoms. You can also engage with pharmaceutical and biotech companies at rare disease industry events. However, for ultra-rare conditions, attracting industry interest can be more challenging, as drug development is often influenced by patient numbers and available data. A strong starting point is to bring together the patient and academic research community, support natural history studies, and strengthen the evidence base. These steps can help build momentum and increase the chances of meaningful industry engagement over time.

The platforms below are excellent for identifying experts who have published work related to your condition or have expertise in a relevant biological pathway.



This is the go-to database for biomedical literature from MEDLINE, life science journals, and online books. Search for your child's condition or gene and see who is publishing on it. The author affiliations will tell you which institutions they work at. Look for the "corresponding author" on papers, often marked with an asterisk or envelope icon. This is typically the senior researcher leading the study and a great person to contact.



Similar to PubMed, but with a broader reach that includes patents and citations. It can help you see which researchers are most frequently cited, indicating a high level of influence in their field. Use the "Cited by" feature to find more recent papers that have referenced a key study on your condition. This can lead you to currently active researchers.



An essential resource for rare diseases, Orphanet provides a directory of expert centres, medical laboratories, and research projects across Europe. It's a fantastic way to find established clinical and research hubs. Search for your condition and then explore the "Research & Trials" section, which lists research projects, patient registries, and biobanks.

It's also a good way to learn all the names that people use for a condition. When you have found the right page for your condition you should see that in the blue table there is a list of other names for the condition.



These are social networking sites for scientists and researchers. You can follow researchers, see their publications, and even contact them directly through the platform.

These platforms often have pre-print versions of papers available, giving you access to the latest research before it's formally published.

You can also join broader groups related to your child's symptoms, like [UKRET \(UK Rare Epilepsies Together\)](#), to gain wider awareness and connect with relevant organisations. If you're ready to explore medical research, organisations like Aspire Biosciences can help connect you with pharmaceutical partners.

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Formalise Your Group

As your community grows, you may want to register as a UK charity. Organisations like Beacon for Rare Diseases offer training and support for setting up a patient group. By registering as a charity you can also get access to certain grants to aid with research (see details below).

The organisations below are significant funders of medical research in the UK and have specific programmes or a strong interest in rare diseases. This list is not exhaustive, but it provides a strong starting point for any UK-based rare disease charity looking to fund vital research.

Harrington Discovery Institute UK

Harrington UK Rare Disease Scholar Award: This award provides £100,000, with the potential for an additional £400,000. They also offer invaluable drug development expertise and project management support. This is a highly competitive award for researchers in the UK. They are looking for "breakthrough discoveries defined by innovation, creativity and potential for clinical impact."

Medical Research Council (MRC) & National Institute for Health and Care Research (NIHR)

These two bodies are the largest public funders of medical research in the UK. They have jointly funded the UK Rare Disease Research Platform with a £14 million investment. Keep an eye on the UKRI (UK Research and Innovation) funding finder for opportunities related to rare diseases. The NIHR also offers support with planning and delivering research, as well as recruiting patients for studies.

Action Medical Research

They offer Project Grants and Research Training Fellowships focused on child health, including rare diseases. Their emphasis is on clinical research or research at the interface between clinical and basic science. Applications are assessed by rigorous peer review. They do not fund work undertaken outside the UK, and there are other exclusions, so it's important to read the guidance carefully.

Barts Charity

They offer a range of grants, including Research Project Grants (from £75,000 to £600,000) and Research Seed Grants (up to £75,000) for innovative, health-related research that could benefit the health of people in East London. While focused on East London, their funding can support research with a broader impact. They are particularly interested in projects that could form the basis of ongoing support from other funders.

The Medical Research Foundation

This independent charity supports vital medical research in underfunded areas, giving hope to people with overlooked health conditions. They are open to a wide range of medical research and are a good option for conditions that may not receive as much attention from larger funders.

LifeArc

LifeArc is a UK-based medical research charity that provides grants and funding aimed at translating academic research into viable diagnostics, therapies or devices - especially in areas of unmet medical need.

GOSH Partnership Grant

The GOSH Partnership Grant supports collaborations between families, charities, and researchers to advance understanding and treatment of rare and complex childhood diseases. It provides funding to kick-start early-stage research projects connected to Great Ormond Street Hospital and the UCL Great Ormond Street Institute of Child Health.

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Drive Fundraising and Research

Fundraising can feel daunting, but there are many organisations that can help. [Tree of Hope](#), for example, supports families with fundraising for medical needs.

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Influence National Research

Use your voice to shape the UK's genomic medicine strategy by joining the Participant Panel at Genomics England. The panel ensures that the views and experiences of patients and their families are at the heart of Genomics England's work. You are eligible if you or your child have contributed your genome data to the National Genomic Research Library.

You can also join your local Genomic Medicine Service Patient and Public Voice Panel to help raise awareness of your child's condition and influence the wider Genomics Unit through projects taking place within your Genomic Medicine Service.

By taking these steps, you are not only building a supportive community but also creating the essential infrastructure to attract researchers and drive the development of future treatments.

This journey is a marathon, not a sprint. By connecting with others, you can find the strength, support, and hope you need to navigate the path ahead. Remember, you are your child's greatest advocate, and you are not alone.



JOURNEYS OF HOPE: FROM A SINGLE DIAGNOSIS TO A GLOBAL COMMUNITY

On the journey with a rare disease, it can often feel like you are navigating an uncharted path alone. But you are not the first to walk this path. The most powerful communities are often born from a single family's determination to turn a diagnosis into a movement of hope and action.

In this section, we share the stories of two such pioneers. These are not just case studies; they are powerful, relatable narratives meant to inspire and empower you by showing what is possible.

You will read about CRELD1 Warriors and CureDHDDS, two organisations that each started with a personal diagnosis and grew into global forces for change. Their journeys provide a real-world look at how families can:

- Build a supportive global network from the ground up.
- Partner with scientists and clinicians to drive and fund critical research.
- Advocate for their condition and transform the future for everyone affected by it.

See for yourself how these families have successfully navigated this path. Their stories can be your blueprint for making a difference.



CRELD1 Warriors: Forging a Path from Isolation to Impact



When our children, Alfie and Lola, were finally diagnosed with CRELD1, we discovered that very little was known about CRELD1 or its link to epilepsy. There was nothing published in medical literature, no established community (other than a few families on Facebook), and no clear path forward. We were the first family in the UK to get a CRELD1 diagnosis, the other families were based in Canada and the U.S. We were, in effect, "on our own" once we received the diagnosis.

This meant we had to carve our own journey to find care, build our community [CRELD1 Warriors](#), and drive research. We had to become the experts that didn't exist. Below are the steps that we took.

The Challenge	The Action We Took
Small Community	We found a Facebook group called 'CRELD1 Warriors and Loved Ones' which had around 10 families that had been diagnosed with CRELD1, all living in the U.S. and Canada. This became our new family, providing us with a fountain of knowledge about the condition. We have now grown to over 37 families worldwide.
No Online Resource	We built the CRELD1 Warriors website so the next family would have somewhere to turn. This grew into a global community.
No Guidance for Parents	Based on the key learnings of the lived experiences with CRELD1, we developed personalised information guides for parents to use when engaging with their consultants, to help provide them with all the information that can help inform their care plans.
No Published Medical Literature	A few of the U.S. families were part of a small medical study being run by Yale School of Medicine, exploring the link between CRELD1 and epilepsy. Lola and Alfie were enrolled in the CRELD1 patient cohort and we collaborated closely with the researchers to improve their understanding of the symptoms. By contributing our children's data, sharing detailed insights into the symptoms, and co-authoring the research paper, we helped produce the first published medical literature linking CRELD1 to epilepsy. This publication was the key. It provided the robust, peer-reviewed scientific evidence required to apply for a change in the UK's diagnostic system. With the paper published, we could then have the CRELD1 gene mutation added to Genomics England's PanelApp. Think of it as the official diagnostic "playlist" for genomic testing in the UK. The Genomics England PanelApp is a list of genes that have a strong enough evidence base to be included in routine diagnostic testing for specific conditions (or "panels") within the NHS.

The Challenge	The Action We Took
No Published Medical Literature	Getting a gene added to a panel means it is officially recognised by the NHS as a significant cause of a particular condition. This has a profound impact on future diagnoses. Any child in the UK who presents with epilepsy and undergoes a genomic test will now have the CRELD1 gene analysed as standard. This can end the diagnostic odyssey for future families, providing answers in months instead of years, or preventing them from being missed entirely. It formally establishes the CRELD1 gene as a significant cause of epilepsy within the NHS framework. Clinicians across the country now have a recognized, evidence-based reason to make a diagnosis, moving the condition out of the unknown and onto the map.
No Scientific Direction	We found and assembled our own Scientific Advisory Board, bringing together critical expertise from around the world to guide our efforts. This includes researchers, geneticists, neurologists and patient advocates.
No Data for Researchers	We set up a patient registry with Sanford CoRDS, creating a vital dataset for the research community to use when investigating the condition.
No Funding	We launched a registered UK charity, CRELD1 Warriors, to create a formal entity that could fundraise for and drive scientific research. We have undertaken some fundraising activities such as hosting charity karaoke fundraisers, running the great south run, forming a team to cycle from London to Brighton, and running 7 marathons in 7 days. This has helped us kickstart our own scientific research.
No Active Research	We found and partnered with a dedicated researcher at the University of Birmingham and are pioneering the first-ever research project into the CRELD1 condition, turning our community's hope into a scientific reality. By collecting skin fibroblasts from our CRELD1 children, we are building a CRELD1 cell model to better understand how the condition manifests. We have also partnered with Unravel Biosciences (RAREshift) to gather RNA data from CRELD1 children around the world, using AI to potentially identify therapeutics for drug repurposing.
No Seat at the Table	Adam joined the Genomics England Participant Panel to better understand the UK's genomic ecosystem, make new connections, and influence national strategy.



CureDHDDS: Building a research community from the ground up

Like many rare disease families, we began this journey feeling completely lost. Two of our three children have an ultra-rare genetic mutation, and at the time of diagnosis, there was almost no information available. We were simply told that the condition was ultra-rare, progressive, and that no research was planned into it in the UK.

Much like Adam, our efforts began with a Facebook group, where we initially connected with just one other family in Canada affected by DHDDS. From that small start, our community has grown to over 220 families worldwide.

As a first step we also reached out to every author of the few scientific papers on DHDDS to learn more and build connections with their research. From those first online conversations, we gradually formalised our efforts and founded the charity Cure DHDDS.

Since then, we have established a Scientific Advisory Board, added DHDDS to genetic testing panels, created a patient registry, attended multiple scientific conferences and, hosted five Cure DHDDS Scientific Exchange Meetings. Like Adam, I have also joined the Genomics England Participant Panel — turning uncertainty into action and connection.

On the next page, you can find a few tips that helped us along the way.



CURE DHDDS
SUPPORT • RESEARCH • CURE

Practical Tips for Families

Reaching out to researchers:

Early on, I learned from another rare parent that in scientific research papers, the first and last authors listed are usually the key researchers. If you're looking to make contact or learn more about a study, those are the names to start with.

Learn about research models:

Research models, such as mice or cell models, are laboratory versions of a disease that help scientists understand how it works and test potential treatments safely before trying them in people:

- **Collecting samples:** If your child or loved one has a rare or little-researched condition, it's incredibly valuable to request blood samples and skin biopsies. These samples are powerful tools that can help researchers better understand the condition. From them, scientists can grow stem cells, brain organoids, and other cellular models, which are used to study how the disease works and to test potential treatments.
- **Exploring Mouse Models:** Check whether your condition already has a mouse model — a research tool that helps scientists study the disease and test potential treatments. If one doesn't exist, you can apply to The Jackson Laboratory (JAX) to create one. JAX specialises in developing custom mouse models for rare diseases and supports families and researchers through the process. Learn more at www.jax.org.

Building a Research Community

Because our research community is so small, we wanted to make sure that everyone's efforts complemented rather than duplicated each other.

To support collaboration, we host online biannual conferences, kindly supported pro bono by Costello Medical. These meetings give scientists a chance to:

- Share new findings
- Discuss research ideas
- Provide feedback on each other's work
- Work together to speed up progress

This open, collaborative approach helps us move research forward much faster.



Fundraising for Research

When we received our diagnosis, there was no existing research into DHDDS in the UK. Once we connected with scientists abroad studying DHDDS and those in the UK studying related pathways, we began to understand what kinds of projects could help us make progress.

To fund these early studies, we turned to platforms like JustGiving and GoFundMe. We've organised a variety of fundraising events, including:

- Two 50km charity walks bringing together family, friends, and local supporters
- A tandem cycle from Land's End to John O'Groats
- Charity auctions, social events, and golf days



Through these efforts, we've been able to fund initial research projects, which we hope will pave the way for larger scientific grants and long-term studies.

Searching for a Treatment

It's important to find out whether your disease shares similarities with other, better-studied conditions. This can open doors to existing treatments and drug repurposing opportunities — where medicines developed for one condition may also help another.

- More information on drug repurposing can be found on the [Beacon](#) website and [Remedi4all](#) website

When you connect with scientists, one key question to ask is:

"Is my mutation a loss-of-function or gain-of-function mutation?"

This information helps determine which types of future novel therapies such as Gene therapy or Antisense oligonucleotide (ASO) therapy might be suitable for your condition.

Another useful website is [The Rare Therapies Launch Pad \(RTLTP\)](#), which is a UK-based initiative designed to create a sustainable pathway for delivering individualised therapies, such as antisense oligonucleotides (ASOs), to children with ultra-rare and life-threatening genetic conditions.



OUR VISION: A CLEAR PATHWAY FOR EVERY FAMILY

This list is by no means exhaustive, and you shouldn't feel you have to do everything on it. Progress takes time. But if you find yourself facing a rare disease and want to help drive research, we hope this guide will point you in the right direction and save you some of the trial and error we experienced along the way.

Because of our journeys, we are now calling for clear frameworks and guidance to be established. We hope that our journey can provide guidance for other newly diagnosed families, ensuring that no one ever has to feel "abandoned" or be forced to carve out their own path alone again.



Adam



Mel

