Resource Guide for Patients Living with Rare Diseases

This Guide is a starting point for your individual journey. It contains information and links to resources to get you started, including advocacy organizations, podcasts, health system navigation, and clinical trial information.

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Where to Start After Being Diagnosed with a Rare Disease

Advocacy Organizations

It's important to understand your diagnosis. Get information from reliable sources. These could include advocacy organizations specific to the disease/condition.

You can contact the GARD to get help with:

- Finding or understanding information about a rare disease
- Navigating information on getting a diagnosis
- Learning about disease experts
- Finding clinical research studies for a rare disease

Meet with a Genetic Counselor to Learn More About the Disease

Check out this animated video in NORD's Rare Disease Video Library on genetic testing for rare and undiagnosed diseases.

It is important to be prepared with questions to ask your genetic counselor. Check out this resource published by Global Genes.

Helpful resources can be found at:

Global Genes

Orphanet

Genetic and Rare Disease (GARD) Information Center

Connect with the Rare Disease Community

Connecting with people can help you learn more about the challenges and offer support. Consider joining Facebook groups specific to the indication.

Connect & Learn from Community Advocates

Having a connection and resource in the rare community is critical to navigating challenges and accessing services and support. Patient advocates, caregivers and care partners play a critical role in connecting the community. This is apparent amongst two incredible "rare disease moms" who are trailblazers in the rare disease community. Their work and resources are highlighted below:

EFFIE PARKS; PODCAST HOST

Effie Parks, originally from beautiful Montana, has become a guiding light in the rare disease community following her son Ford's diagnosis with CTNNB1 syndrome. Settling in Washington, she transformed her family's journey into a crusade for advocacy, support, and empowerment for families navigating similar challenges. As the host of the "Once Upon a Gene" podcast, Effie has been recognized for several awards including WEGO Health and Podcast Magazine for her impactful storytelling and resource-sharing in the realm of rare genetic disorders. Effie extends her advocacy through speaking engagements at medical and patient advocacy conferences, sharing her experiences and insights from her work to bridge the gap between all rare disease stakeholders. Her skill in community engagement, developed through her advocacy, empowers her efforts in building a supportive network and raising awareness. With a mission to leave the world better than she found it, Effie is dedicated to fostering a more informed and empathetic environment for those impacted by rare diseases. Her work embodies resilience and compassion, inspiring and uniting the rare disease community. Effie's journey is not just about sharing stories, it's about driving change and creating a lasting impact in the world of rare genetic conditions.

The Once Upon a Gene Podcast is an excellent resource for the rare disease community and serves to inspire so many.

SIERRA PHILLIPS: CO-FOUNDER OF LIBRAREY: A FREE RARE DISEASE AND DISABILITY RESOURCE DATABASE

Sierra is a twin mom, rare mom, genetics enthusiast, and relentless advocate and the co-founder of Librarey. In 2021 her son was diagnosed with Warsaw Breakage Syndrome and at the time there were less than 25 documented cases worldwide. During the short time she has been a caregiver on this journey, she found that the best resources and tips have come from caregivers walking a similar journey within the rare and disability communities. The doctors, therapists, case workers, service facilitators often didn't even know about resources she would bring to them. These experiences are what sparked the passion to create a comprehensive resource guide for families and she has been working bringing Librarey to life ever since. Librarey is a rare disease and disability resource database that is completely free. Librarey is a place where families can find resources and recommend resources to the community, making it "family sourced". Sierra's motto is "knowledge is power and sharing knowledge empowers us all" and her hope is that families, friends, & professionals can utilize Librarey to find and spread awareness about all of the amazing resources that are out that support rare disease and disability communities.

Navigating Health Systems

- Keep a notebook to keep track of all questions to ask your care provider
- Advocate for yourself or your loved one. It's important to ask questions and to ensure you get the information you need to understand the diagnosis
- For Primary or Supplemental health care coverage see NORD's resource on Medicaid Financial Eligibility.

A rare disease affects fewer than 200,000 people in the USA and only one in every 2,000 people in Europe.

Is a Clinical Trial an Option for Me?

Participating in a clinical trial is often the only hope for treatment for an individual with a rare disease. Unfortunately, enrolling eligible patients for a rare disease clinical trial is quite difficult as each rare disease affects fewer than 200,000 people in the USA and only one in every 2,000 people in Europe. To help make the process a little more manageable, here are some important steps to help you navigate a clinical trial.

How to Identify a Trial

Almost 50 percent of patients currently learn about clinical trials through their own research on the internet.[1] Although you may start with an inquiry to your personal physician, the truth is that fewer than 10 percent of physicians participate in research. You may find that your own physician may not even be aware of clinical trials available for your rare disease.

You can start your own online search for applicable clinical trials by first looking at advocacy groups to find any listed on their websites. Many have searchable databases and other resources for clinical research. NORD has a resource, Find A Rare Disease Organization and a section, Find Clinical Trials & Research Studies. These are both good starting points.

For a more general search, try ClinicalTrials.gov. This is a central database hub of clinical trials for all conditions. It is a valuable resource for finding a clinical trial specific to your diagnosis. Once you find a potential trial, contact the site listed for more information or talk to your doctor to see if they have more information about the trial and can connect you to the site.

Addressing Concerns about Participating in a Clinical Trial

Patients are often reluctant to participate in a clinical trial due to multiple unanswered questions or misconceptions. Here are some frequently answered questions about clinical trials.

WHAT IF THE EXPERIMENTAL TREATMENT IS NOT EFFECTIVE FOR MY DISEASE?

You will be closely monitored by the study team and your personal physician will be informed of your treatment and response. Every trial has the potential to advance understanding of the disease so that better treatment options might become available.

CAN I ENROLL IN A TRIAL AND STILL RECEIVE TREATMENT FROM OTHER PHYSICIANS?

Yes, you can be in a trial and continue to see your personal physician(s) or other specialties.

WHAT IF I EXPERIENCE UNPLEASANT OR SERIOUS SIDE EFFECTS RELATED TO THE STUDY TREATMENT?

Throughout the trial you will be assessed carefully for any side effects and treated immediately. Before you begin the trial, the study coordinator will review medication precautions and will give you a contact number to reach a coordinator at any time to discuss concerns or issues you may have.

CLINICAL TRIALS REQUIRE A LOT OF STUDY SITE VISITS. WHAT IF I'M TOO BUSY TO PARTICIPATE?

The time commitment for studies varies greatly. In addition, study sites are always willing to accommodate patient needs. You can ask for assistance with timing, transportation, and flexibility with scheduling.

DO ALL TRIALS INCLUDE A PLACEBO GROUP?

Placebos are not used in all trials. If a placebo is used it is likely because no standard treatment exists. In some cases a placebo is used in a trial that compares standard treatment plus a placebo, You will always will be told ahead of time if a study uses a placebo.

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WHY AM I NOT ALLOWED TO KNOW IF I AM RECEIVING THE TREATMENT MEDICINE OR PLACEBO?

Knowledge of whether you are receiving the treatment medicine or placebo can impact the effect of the treatment Not knowing is important to maintain the integrity of the clinical trial. Both the physician and the participant may not know this information - this is called a double-blind trial. *NOTE: In some trials, patients are permitted to add the experimental treatment to their current treatment.*

Diverstiy, Equity and Inclusion for Rare Diseases

Embracing diversity and inclusion in rare disease clinical trials is essential for advancing medical research and improving patient outcomes. The benefits of a diverse participant pool are numerous, ranging from enhanced scientific rigor to improved generalizability of study findings. Navigating participation in clinical trials requires a proactive approach, leveraging education, advocacy, and support networks. By collaborating with patients, researchers, and healthcare providers, we can foster an environment that ensures equitable access to promising treatments for all, irrespective of the rarity of their conditions.

Helpful resources can be found at:

RDDC (rarediseasediversity.org)

Diversity, Equity, and Inclusion for Rare Disease – RARE-X

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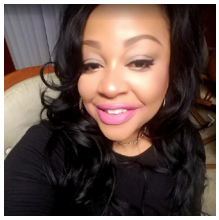

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References

1. https://www.ciscrp.org

About UBC

United BioSource LLC (UBC) is the leading provider of evidence development solutions with an expertise in uniting evidence and access. UBC helps biopharma mitigate risk, address product hurdles, and demonstrate safety, efficacy, and value under real-world conditions. Underpinned by our scientific expertise, data and analytics, and innovative technologies, we offer our customers flexible solutions generating the relevant real-world data necessary to make more informed decisions earlier, meet stakeholder requirements, and, ultimately, drive better patient outcomes.

To find out more about how UBC can help you on your patient journey, reach out to us at contact@ubc.com.

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