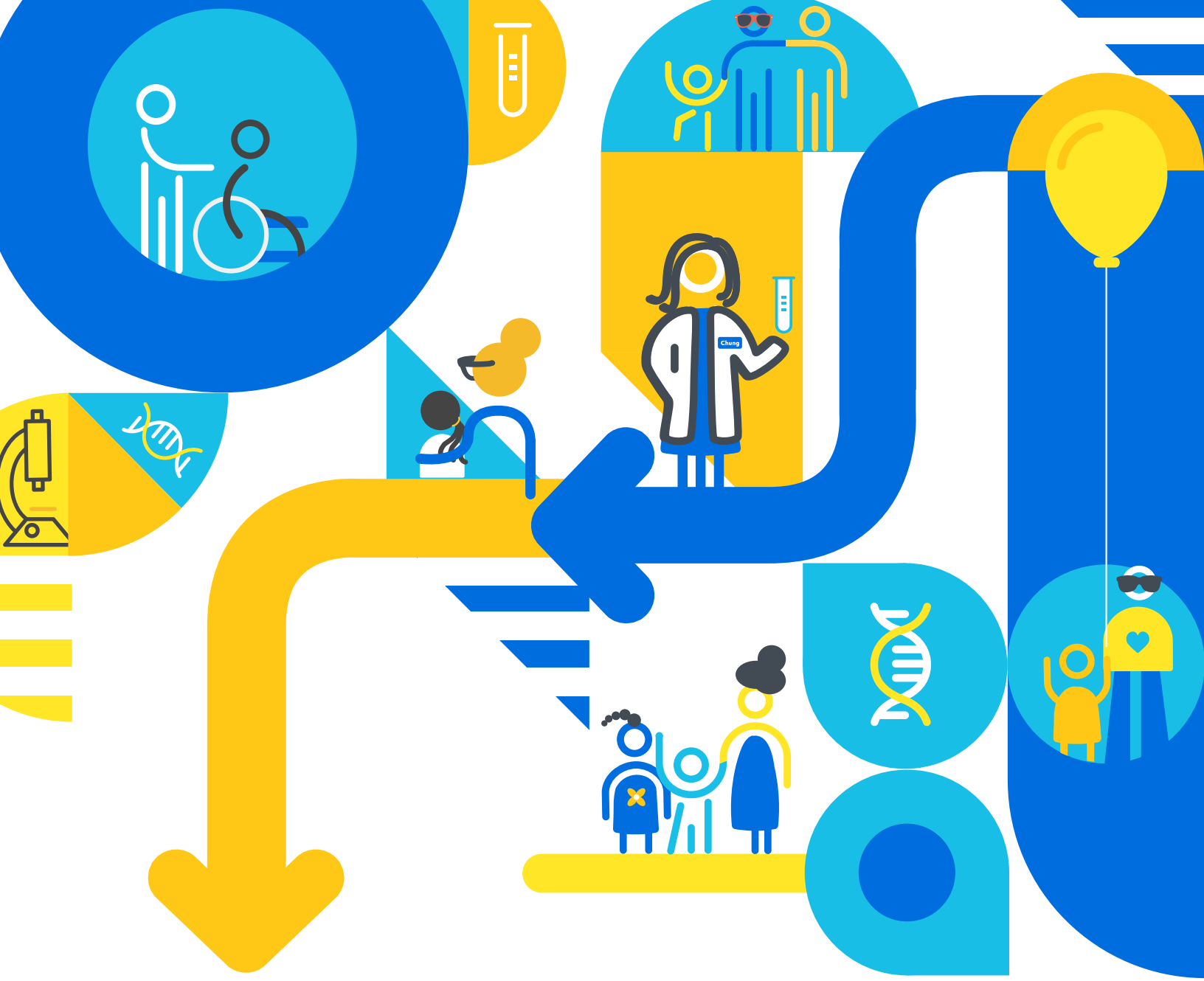

A Family Guide

Navigating Life after a Rare Genetic Diagnosis

Developed by

SIMONS SEARCHLIGHT

September 2023

Overview

This guide covers the steps from genetic diagnosis to finding resources and potential treatments for rare genetic neurodevelopmental disorders. It is designed to help caregivers and families navigate the complicated world of rare diseases.

Knowing the cause of your child's neurodevelopmental diagnosis can be a relief. But, a new genetic diagnosis can also be stressful, scary, and overwhelming. It is normal to have a wide range of emotions – sometimes conflicting – about a new diagnosis. It is important to remember that there is no right or wrong way to respond, and we all adjust to information differently.

Here are suggestions from the Simons Searchlight genetic counselors to help you begin to adjust to your or your family member's diagnosis:

- Understanding the genetics of your child's condition can be a challenge. It's important to work with your medical team to develop a clear understanding of the new diagnosis. This will help you to develop a clear course of action.
- Every parent has their own way of dealing with stress and caring for their child. It may be helpful to share your feelings with a family member, close friend, or mental health provider.
- In addition to family and friends, online or in-person support groups can help you overcome feelings of isolation and allow you to connect with others that are caring for a child who has a similar condition.
- Focus on your child's overall well-being, not just on their genetic condition. Your child has a unique personality, strengths, and characteristics.

"Simons Searchlight exemplifies the power of community-engaged research done respectfully and responsively. Families are true partners on this journey to understanding."

Wendy Chung, Ph.D., M.D.
Simons Searchlight Principal Investigator

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Steps After Getting a Genetic Diagnosis

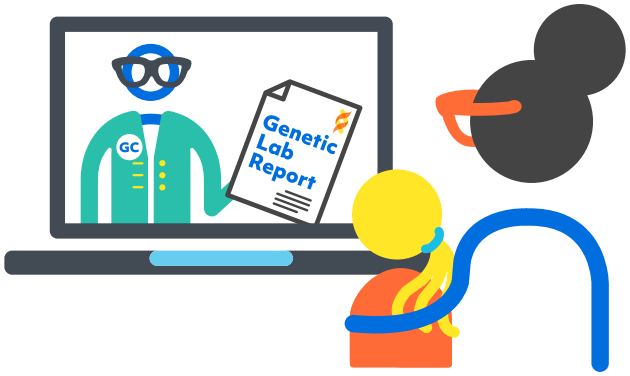
Step 1

Your Genetic Diagnosis

You may have received your genetic result from a medical geneticist or genetic counselor.

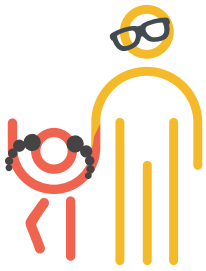
If you have questions about your genetic diagnosis, you can find a genetic counselor in the U.S. or Canada at NSGC.org.

You can also meet with one of the Simons Searchlight Board Certified Genetic Counselors. This is a free resource for families to better understand their genetic report findings and discuss complicated concepts and ideas. It is important to note that our team will not be able to provide specific medical advice.



Step 2

Where to Start



You Are Not Alone.

Parents or caregivers may find it beneficial to connect with others in their rare genetic community. Patient advocacy groups and online communities can provide support and access to other families that are facing similar journeys.

- At Simons Searchlight, we host online communities for all the genetic conditions we study, and we collaborate with over fifty patient advocacy communities. You can join our Facebook communities without enrolling in Simons Searchlight. We encourage you to **find your community** today!
- Rare disease parents and advocacy leaders have created **The Ultimate Resource Guide**.

Education and Information

Families may seek out reliable sources of information, such as reputable websites, books, or conferences, to better understand the disorder, its characteristics, and available resources.

The patient advocacy organizations listed below are additional resources that support the rare disease community:

USA
- Genetic Alliance
- Global Genes
- National Organization for Rare Disorders (NORD)

Europe
- European Rare Diseases Organization (EURORDIS)
- Genetic and Rare Diseases Information Center (GARD)

Step 2 cont

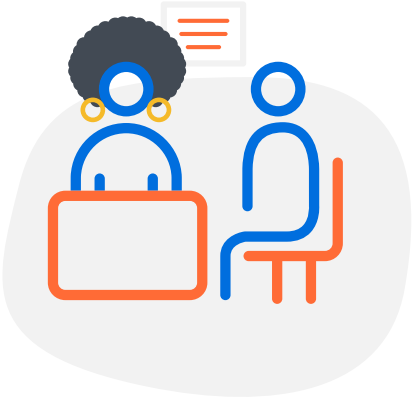
Where to Start

Therapy and Interventions

Families and individuals who have rare genetic conditions have the ability to contribute to meaningful research opportunities that could help develop future treatments.

In the meantime, you can access services that can greatly benefit overall development and well-being. Available resources will depend on where you live but may include early intervention, educational support, developmental programs, speech therapy, occupational therapy, physical therapy, and behavioral therapy. Make sure that you are accessing all the services that your loved one needs from your local government, school, or healthcare system. Reach out to local providers to learn more about what is available.

A local doctor may be able to evaluate your loved one's needs and recommend services like therapy, educational support, adaptive skill training, and assistive equipment that could help them thrive. While caring for someone with a rare disease is challenging, you have the power to improve their quality of life and advance research through your participation.



"We hope that with Simons Searchlight, we can really help families understand and use that genetic diagnosis to empower their child and to empower themselves."

Jamie Atondo and Rebecca Smith
Simons Searchlight Genetic Counselors

Step

3

Creating a Support Network

Medical Professionals

Collaborating with a team of medical professionals, including **medical geneticists or primary care providers, specialists for medical conditions, and therapists**, can provide comprehensive care and guidance for your or your child's specific needs.

To find optimal care for your condition, consult centers of excellence, specialized experts, and patient advocacy groups, or **contact us** for guidance on identifying knowledgeable providers tailored to your needs.

Step 3 cont

Creating a Support Network

Financial Assistance

Exploring financial resources, such as insurance coverage, government programs, grants, or foundations dedicated to rare genetic disorders, can help alleviate the financial burden associated with medical expenses and therapies. Need more help? In the U.S., you can request a social worker to assist you with applying for financial assistance programs based on your family's hospital and state. Explore more U.S.-based financial resources below (we hope to add more international resources in the future):

- **The Arc** is a community-based organization that helps with future planning and housing. They have many local chapters, for people who have intellectual and developmental disabilities. It is a great source for information on policy, disability rights, and services.
- The **Easter Seals** is a nonprofit organization with services that include early intervention, inclusive childcare, medical rehabilitation, and autism services for young children and their families; job training and coaching, employment placement, and transportation services for adults with disabilities, including veterans; transitioning military, veterans and their families, adult day services, and employment opportunities for older adults – and much more.

Step 3 cont

Creating a Support Network

Genetic Community Resources

Connecting with others through Facebook groups, WhatsApp, or gene-specific foundations and communities is vitally important to find other families like yours.

Patient advocacy organizations are instrumental in providing vital information, resources and a supportive community for families navigating rare genetic neurodevelopmental disorders. Simons Searchlight collaborates closely with over fifty organizations, ranging from formal non-profits to active Facebook groups. Their continuous support is essential in expanding and connecting genetic communities, making a lasting impact on research programs including Simons Searchlight. We extend our gratitude to these organizations, their leaders, board members, and dedicated volunteers, as their unwavering support strengthens Simons Searchlight's mission.

Check the "Support Resources" on your **Simons Searchlight gene page** to find your genetic community and other places to connect.

Step

4

Developing a Personalized Medical Care Plan

Individualized Management Plan

An Individualized Management Plan is a customized care plan made specially for one person's needs.

It lists treatments, therapies, and other help that should be given to someone with a certain health condition. The plan is tailored to the specific symptoms and challenges of each individual patient.

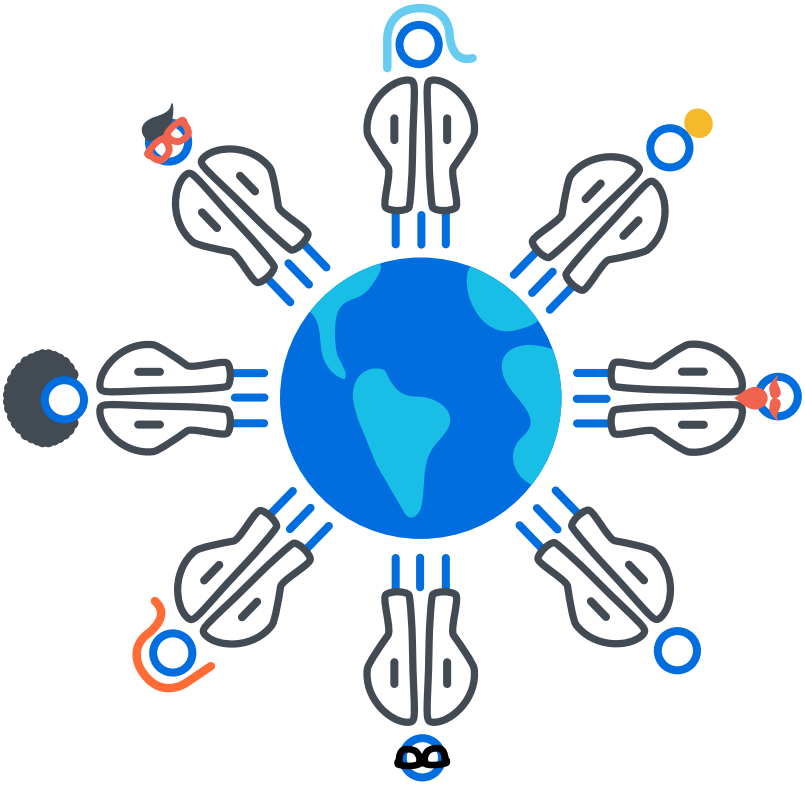
It's not a one-size-fits-all plan - it's made just for you!

The goal of an Individualized Management Plan is to give each person the best care and support for their unique situation.

Some genetic conditions have treatment recommendations available on **GeneReviews**. GeneReviews is a resource for doctors and includes relevant and medically actionable information created by experts. If your condition does not have a suggested guideline at this time, an individualized treatment plan is still created for a person based on their medical features. This may involve a combination of medical interventions, therapies (such as occupational, speech, or physical therapy), and educational supports that can help optimize your or your child's development and quality of life.

Step 4 cont

Developing a Personalized Medical Care Plan



Collaboration with Local and International Specialists

Work closely with your doctor and other specialists to create a care plan tailored to you or your dependent's symptoms and needs. Together you can find helpful therapies and treatments.

Since many conditions Simons Searchlight studies are rare, your doctor may not have treated someone with the exact same condition before. It is common for doctors to learn along with you. They can research the medical literature and contact experts internationally who know more about the specific condition. Learning together with your doctor is common.

Step 4 cont

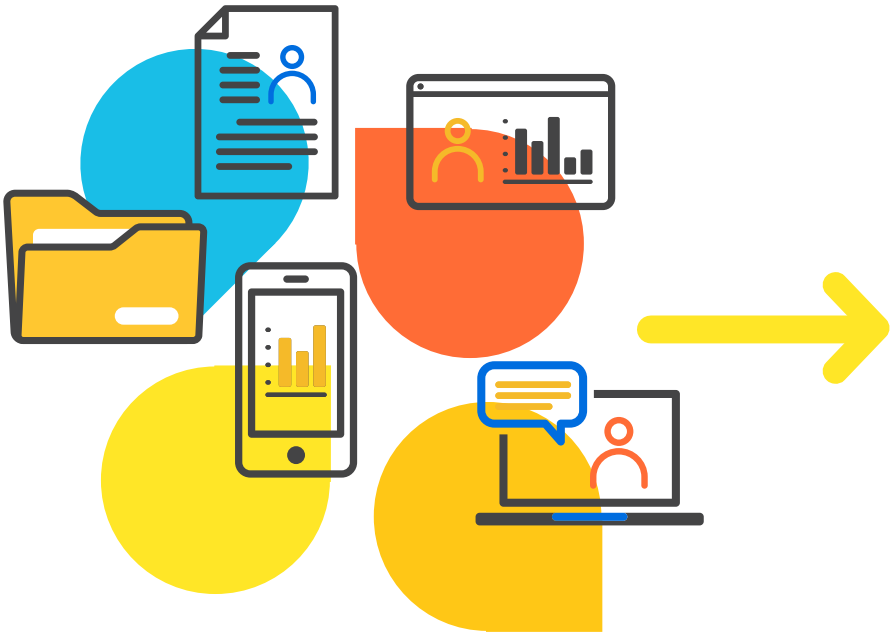
Developing a Personalized Medical Care Plan

Accessing Medical Records

Rare diseases can be hard to diagnose and even harder to treat and manage. Over the years, people who have genetic conditions may see many primary care doctors, case managers, and specialists at different hospitals. Keeping track of medical records can be a challenge, even for the most organized people.

There are ways to create and maintain a personal medical file.

Some people like paper files and others prefer electronic files or a combination of both.



"The team at Simons Searchlight guides every family with care and compassion. We strive for our work to transform sparse understanding into meaningful insights that make a real difference."

Cora Taylor, Ph.D.
Simons Searchlight Principal Investigator

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Step 4 cont

Developing a Personalized Medical Care Plan

Accessing Medical Records cont

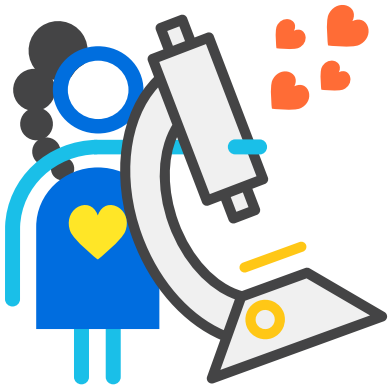
Choose whatever works best for you. The most important thing is that the records are easy to access and are up to date.

- **Electronic Medical Records**: For those who are comfortable with online tools, there are several personal electronic health record management systems that can help. We do not endorse a specific product. You can also store scanned paper documents in **DropBox** or **Google Docs**.
- **CareZone** is a simple and private space that provides a shared calendar and journal for family members and caregivers to coordinate and stay organized.
- **WebMD personal health record** allows you to securely gather, store, manage, and share your own and your family's health information.
- **Paper Records**: If you prefer to keep paper files in a binder or folder, we recommend organizing them by categories, such as laboratory work, genetic tests, consultation notes from specialists, school evaluations, etc. We also recommend making copies and having back up files available.
  - The **Caring for Rare Disease Caregivers** has guides for organizing your patient file, keeping an up-to-date medication list, and coordinating care across providers.
  - **Care Notebook** is a way to organize your medical files and can include specific information about your strengths, goals of care, and limitations.
  - **Care Plans** can also be used to explain your condition and health history.

Step

5

Get Involved in Research



Better treatment guidelines, an understanding of the condition, and a pathway to a cure are only possible with ongoing research participation from patients.

Rare disease research is hard because access to patient information is limited due to the small number of people who have each condition.

Simons Searchlight is an international research program funded by the Simons Foundation Autism Research Initiative, also known as SFARI.

Simons Searchlight is an online research registry for more than 175 genetic disorders that are associated with neurodevelopmental conditions, including autism spectrum disorder. Participants share detailed medical and developmental information through online surveys and can provide an optional blood sample which is used to learn about biological changes. Our data scientists analyze the information and share de-identified data with researchers, making it easier for them to access the information they need to advance rare disease research. Simons Searchlight developed this guide to help families throughout this process. As a long-running, trusted research program working to better understand rare genetic disorders, we provide information and opportunities for families to contribute to the growing knowledge in this field.

Learn more and register for **Simons Searchlight**.

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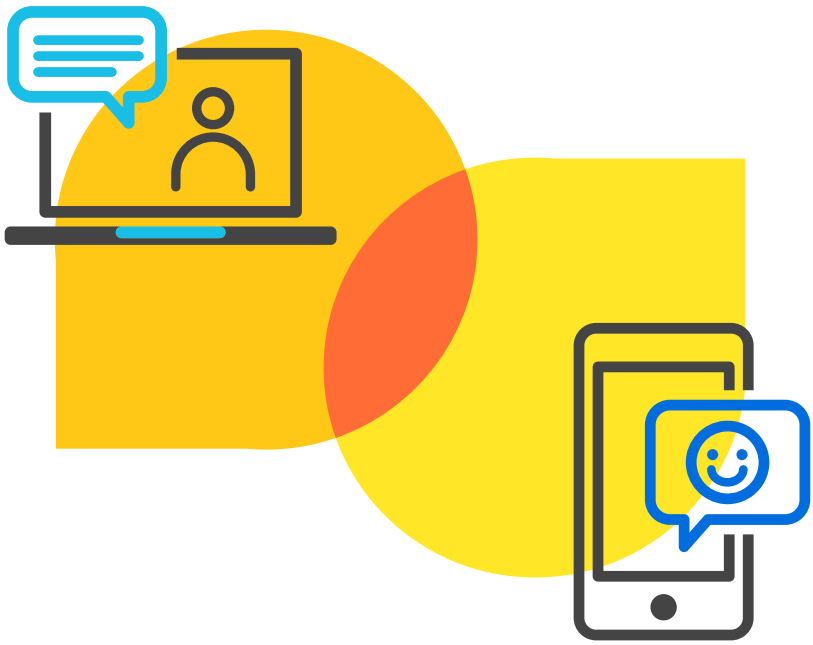
Step 5 cont

Get Involved in Research

Visit your patient advocacy community's website for information on other potential research studies or clinical trials.

Also, many medical technologies are available through hospitals and specialists, but some of the newer treatments and therapies are only offered through research and clinical trials. Listed below are some online resources that you can use to find ongoing clinical trials.

- **ClinicalTrials.gov** is a database that provides the public with current information on clinical research studies.
- The **International Rare Diseases Research Consortium** connects researchers and the organizations working on rare disease research as they develop new therapies and methods for diagnoses.



Step 6

Continued Learning

The resources below include information on genetic counselors, research, policy, clinical trials, and personal stories from patients and doctors.

- Simons Searchlight genetic counselors care deeply about you and are dedicated to helping participants understand their rare genetic neurodevelopmental disorder. **Learn more** about these amazing team members who are here to support you.
- **RARE Daily** is a news bulletin published by Global Genes that aims to eliminate the challenges of rare diseases through education and activism.
- You may want to set up a **Google Alert** to receive email notifications when Google finds new results on a topic that interests you.
- You can also set up alerts from **PubMed** to be notified when a new journal article is published on a topic of interest.

"I'd do anything to help my son have the best possible chance for a long and happy life, including contributing his medical history and completing surveys in Simons Searchlight!"

Vanessa
Simons Searchlight Participant and Parent

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Thank you!

Register at **SimonsSearchlight.org**

Coordinator@SimonsSearchlight.org

facebook.com/SimonsSearchlight

@s_searchlight.org

youtube.com/SimonsSearchlight