THE CIRCLE OF CARE GUIDEBOOK FOR

CAREGIVERS OF CHILDREN WITH RARE AND/OR SERIOUS ILLNESSES

National Alliance for Caregiving

IN PARTNERSHIP WITH:

Global Genes®
Allies in Rare Disease

FOREWORD

Being a Caregiver for a child with a rare disease or other serious medical condition can be both enormously gratifying and extremely challenging. For most, the experience is life-altering, and for some, all-consuming. In particular, the long and winding diagnostic odyssey and daily complexity of navigating care, treatment and support services can be overwhelming.

Rare Caregivers often must advocate for equitable and timely access to diagnostic testing, medical specialists, trials and the latest treatments, and become experts in their disease, researchers, innovators and entrepreneurs, organizers, fundraisers, policy wonks and thought leaders – sometimes all simultaneously -- while being loving and available partners, parents and siblings.

The Circle of Care Guidebook is intended to help Caregivers navigate through the varied experiences and challenges of rare and serious medical conditions, guided by the insights, achievements, and learnings of other caregivers and experts.

The extensive array of topics covered in this Guidebook (nearly 100) underscores the many aspects of life and care that are impacted when caring for a child with a rare disease. Fortunately, there are a wide variety of helpful resources and organizations available to support and direct Caregivers, many of which are also highlighted in this document.

Global Genes would like to thank the National Alliance for Caregiving and all the other Caregivers, organizations and experts involved for calling attention to the unique needs of those caring for a child with a rare and serious illness, and for helping to develop this Guidebook as a resource.

Rare diseases affect nearly one in 10 Americans, and more than half of those affected are children. The urgency and pressures faced by parents, siblings or loved ones caring for a child with a rare disease are immense and constant. The Circle of Care Guidebook will help Caregivers more easily navigate through the needs and challenges of caring for children with rare and serious illnesses, and find the right next steps to take on their behalf.

Global Genes
www.globalgenes.org

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ACKNOWLEDGMENTS

In partnership with Global Genes, the National Alliance for Caregiving (NAC) is proud to present the *Circle of Care Guidebook for Caregivers of Children with Rare and/or Serious Illnesses*. This guidebook was made possible by Mallinckrodt Pharmaceuticals. This guidebook builds on the 2018 study, *Rare Caregiving in America*, by the National Alliance for Caregiving and Global Genes.

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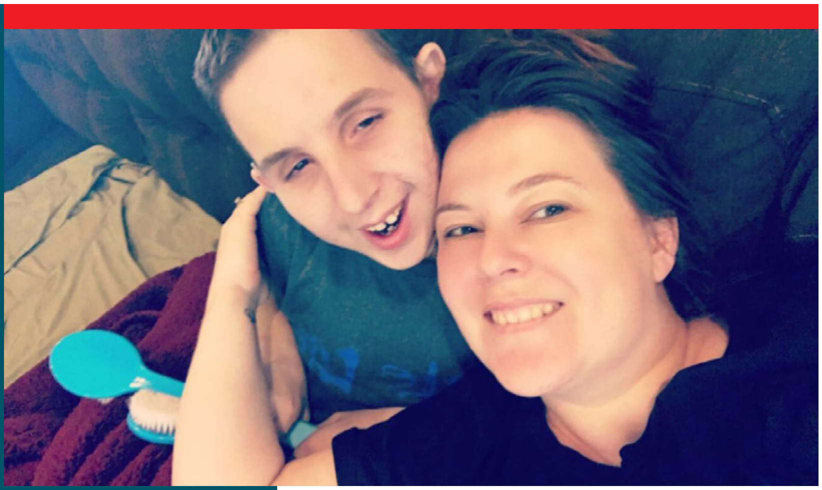
We'd like to thank all family caregivers who dedicate their time to caring for those in need. This guidebook is for them and for those who support them.



WELCOME

As a parent caregiver, advocate, and professional providing support to caregivers in the rare disease community, this book is especially meaningful to me. I became a caregiver to my now adult son long before I ever recognized I was indeed a caregiver myself. I am a mom. A mom to an extremely sick son who needed me to be so much more than I ever knew I could be for him.

I have felt the utter helplessness of not knowing what my own son had, and then learning it was a rare disease with no cure in sight. I have experienced the isolation of not having anyone to relate to and years of no support, or disease affiliated foundation to turn to. My son was 11 years old, several years past the previously expected life expectancy we had been given, when I first met others caring for someone with our son's rare disease.

The founding of the Global Foundation for Peroxisomal Disorders (GFPD) led me to meeting Dr. Mousumi Bose, a fellow mom and caregiver. Her son, Ilan, died at the age of 14 months from the same rare disease my son has. She was one of the first individuals to help me understand my role as a parent caregiver. My participation in focus groups led by her about the emotional experiences of caregivers validated the experiences I and so many other rare disease caregivers have had. A few of these research outcomes alongside many other highly regarded study findings are shared throughout this guidebook. They are essential to better understanding the experiences of the rare disease caregiver.

The combination of diverse experiences from Dr. Mousumi Bose, Kimberly Haugstad, and Theresa Smith have resulted in a guide with a vast range of resources for all caregivers. Rare disease is often complex and requires a multi-disciplinary approach to be effective, which is why this inclusive guidebook is a tool all caregivers should have access to. The care put into this book is representative of the care all caregivers need and deserve.

Rare disease caregiving comes with its own set of unique challenges. It is easy to feel you are alone in this, but you are not. This book serves as an understanding friend in the rare disease community, or maybe even a little bit of your future self, sending you some much needed guidance.

Caregivers themselves have created this book to not only pass the torch of advice, but to illuminate the path for you to know you are not alone in the dark. These authors have put so much more than words on these pages. Let their guidance walk with you and help you navigate all the twists and turns of your rare disease caregiving experience.

This is a tool I wish I had years ago. Whether you are a new or seasoned caregiver, you will find a fellow caregiver guiding the way throughout this guidebook.

Katie Sacra

Katie Sacra

IMAGE PROVIDED BY GLOBAL GENES

INTRODUCTION

For the purpose of this guidebook, rare and/or serious illnesses encompass medically complex conditions and diseases that might limit or shorten the life of a child. Family caregivers include parents, guardians, siblings, grandparents, other family members, friends, and other people who provide unpaid care to a child or children below the age of 18 with a rare and/or serious illness.

The impact on and needs of families affected by a rare and/or serious illness vary by disease. To best address these needs, we wanted to create a guidebook based on first-hand experienced from family caregivers. The topics presented were informed by caregivers through a series of focus group discussions, and the information is based on primary research.[4]

In the United States, rare diseases and conditions are those that affect fewer than 200,000 people.

It is estimated that more than 30 million Americans currently have a rare disease or condition.[1] Of these, 50 percent are children under the age of 18.[2] In 2018, The National Alliance for Caregiving (NAC) published *Rare Disease Caregiving in America* in partnership with Global Genes to describe the journeys, challenges, joys, and contributions of rare disease caregivers. The study found that rare diseases have a profound impact on the lives of caregivers and inspired the development of this guidebook. This *Circle of Care Guidebook for Caregivers of Children with Rare and/or Serious Illnesses* is designed to help patients, caregivers,[3] and families of children and young adults during their experience with a rare and/or serious illness by offering effective resources that have helped other caregivers in similar situations.

We hope this guidebook provides a comprehensive set of resources and guidance to address many of the challenges facing caregivers. Additionally, this guidebook provides information on awareness initiatives that emphasize and advocate for the needs of families who have a child with a rare and/or serious illnesses.

Each section provides an introduction, tools, and resources recommended by other caregivers, health professionals, and social workers from organizations that provide support to caregivers of rare and serious illnesses. Readers can select topics of interest from the table of contents based on current experiences or needs.

We welcome your feedback. If you find this guidebook helpful, or if you have recommendations for improvements, please email us at study@caregiving.org.

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[1] *National Human Genome Institute. (Jan., 2020). Rare Disease FAQ. Retrieved from https://www.genome.gov/FAQ/Rare-Diseases.*

[2] *Rare Genomics Institute. Get the Facts on Rare Diseases. Retrieved from https://www.raregenomics.org/rare-disease-facts.*

[3] *In general, our use of the term "family caregiver" aligns with the definition provided in the recently-enacted federal RAISE Family Caregivers Act. This legislation defines a family caregiver as "an adult family member or other individual who has a significant relationship with, and who provides a broad range of assistance to, an individual with a chronic or other health condition, disability, or functional limitation." (RAISE Family Caregivers Act of 2018 (P.L. No: 115-119), available at https://www.congress.gov/bill/115th-congress/house-bill/3759.) In research and in advocacy, "caregiver" may be described as: informal caregiver, care partner, care taker, and similar terminology. In an international context, the term "carer" is often used. It should be noted that in addition to adults, an estimated 3.4 million children under 18 may also have a caregiving role within their household (see, for example, Caregiving in the U.S. 2020, AARP and NAC, https://www.caregiving.org/caregiving-in-the-us-2020/).*

[4] *The inclusion criteria for the focus groups to inform guidebook content are: 1) a disorder that affects multiple organ systems; 2) a neuromuscular disorder; 3) a degenerative disorder; 4) a disorder that results in both cognitive and physical disabilities; or 5) a disorder that requires some form of life support. Focus group participants identified as either current or bereaved caregivers of children with various diagnoses, including Lennox-Gaustat syndrome, ataxia telangiectasia, Rett syndrome, Zellweger spectrum disorder, Niemann-Pick Type A and B, and Trisomy 21. Several participants are caregivers of a child(ren) with a variety of symptoms but no formal diagnosis. Common symptoms included epilepsy, cerebral palsy, intellectual disability, communication disability, cortical visual impairment, and various gastrointestinal symptoms.*

ABOUT CAREGIVING FOR CHILDREN WITH RARE AND/OR SERIOUS ILLNESSES

Caregiving for a child with rare and/or serious illnesses often brings mixed feelings—some positive, some negative. During our research, family caregivers talked about the positive impact of caring for a child with a rare disease:[5]

- Caregivers feel empowered when advocating for quality care on behalf of their child(ren), as well as supporting systemic change on behalf of the disease community. This empowerment often comes from developing the necessary medical skills to care for their child, being able to navigate systems and current research appropriate for their child's care, raising awareness and, at times, funding for the disease community.
- Caregivers report an improved ability to set boundaries in personal and professional interactions as a result of their heightened sense of empowerment, often resulting in healthier relationships with others.
- Caregivers often describe feeling more capable of managing their personal caregiving experiences and interactions with others.
- Some caregivers have found that their caregiving experiences changed their career trajectory, inspiring them to seek out professional opportunities related to their caregiving experience.
- Many caregivers say their experiences with their children have helped them better appreciate life and be more empathetic about the challenges facing others.
- Most rare caregivers report a sense of pride in improving the public's understanding of their child's condition, feeling close to their care recipient, and having a sense of purpose as a result of caregiving.

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[5] *Bose, M., Mahadevan, M., Schules, D. R., Coleman, R. K., Gawron, K. M., Gamble, M. B., & Rizzo, W. B. (2019). "Emotional Experience in Parents of Children with Zellweger Spectrum Disorders: A Qualitative Study." Molecular Genetics and Metabolism Reports, 19. https://doi.org/10.1016/j.ymgmr.2019.100459.*

These positive feelings about caregiving often coexist with the challenges. Many caregivers may say that caring for a child with a rare and/or serious illness feels like riding a roller coaster, with highs and lows..[6] Even simple activities in daily life can be full of emotional triggers. Some examples include:

- The difficult and lengthy process of navigating the health system.
- Lack of resources at local hospital institutions to effectively manage the underlying condition(s) of the child.
- Strenuous transitions from inpatient hospital care to home care and vice versa.
- The time, confusion, and frustration of coordinating with health insurance companies for reimbursement of services and equipment.
- Hard-to-access treatments or therapies for the underlying condition(s) or symptom management.
- Challenging and emotionally draining communications with the child's health care providers.
- The long-term commitment of caregiving for children with a rare and/or serious illness. On average, rare and/or serious illness caregivers provide care for an average of 8.9 years.[7]
- The often time-intensive and round-the-clock nature of rare caregiving, such as children who need constant health monitoring.
- The physical strain of certain tasks, especially as the caregiver and child grow older.
- The emotional toll of rare caregiving. Some parents and other caregivers experience anticipatory anxiety (worry about things that may happen) and grief, which adds to their level of stress.

Many caregivers may say that caring for a child with a rare and/or serious illness feels like riding a roller coaster, with highs and lows.

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[6] Ibid.

[7] National Alliance for Caregiving. (2018). Rare Disease Caregiving in America. Retrieved from https://www.caregiving.org/wp-content/uploads/2020/05/NAC-RareDiseaseReport_February-2018_WEB.pdf.

ADDRESSING EQUITY IN CAREGIVING FOR RARE AND/OR SERIOUS ILLNESSES

Disparities in healthcare and research have considerable consequences, including the reality that some communities do not have access to high quality care, support, and resources.

This guidebook aims to directly address the needs of all caregivers as effectively as possible given differing factors such as geography, socioeconomics, race and ethnicity, family composition, and others. These topics will be discussed here and throughout the guidebook. Additionally, we have provided information on where to inquire and access resources that are not readily available in a given area.

CAREGIVERS FROM UNDERREPRESENTED RACIAL AND ETHNIC GROUPS

Care for a condition that few people—including healthcare providers—have heard of is challenging. Communities of color face the added burden of deep and systemic inequities in access to healthcare, services, and treatment. Minorities are traditionally underrepresented in many studies and clinical trials. Since certain genetic conditions disproportionately affect communities of color, a lack of representation creates significant knowledge gaps that can directly affect patient care and treatments. In our focus groups, caregivers of color reported feeling that their concerns were not being taken seriously by the medical community. These interactions may make it difficult to find medical providers who can relate to families from diverse backgrounds and respectfully work within language needs and cultural competence.

In any situation, it is important that people feel comfortable with the help they are receiving; this is especially true when it comes to health and taking care of others. When caring for a child with a rare and/or serious illness, caregivers have a right to ask for the best care for themselves and their child. This includes access to medical providers who put caregivers at ease, both in medical practice and personal interactions. Caregivers from underrepresented communities should not be afraid to reach out to or search for available resources that can help them connect with doctors familiar and competent in more diverse community spaces.

In January 2021, the Black Women's Health Imperative (BWHI) formed the Rare Disease Diversity Coalition to focus on reducing racial disparities in the rare disease community. The group is made up of rare disease experts, health and diversity advocates, and industry leaders. It works to identify and advocate for evidence-based ways to ease the disproportionate burden of rare diseases on minority communities. www.bwhi.org

In May 2021, Global Genes, an alliance supporting the rare disease community, launched its RARE Health Equity Council (www.globalgenes.org/the-rare-health-equity-leadership-council) to aid in efforts to better understand and address diagnostic, access, and treatment gaps and challenges for underserved and underrepresented rare disease patient populations. Through a variety of programs and meetings, the RARE Health Equity Council will focus on improving outreach to Black, African American, Native American, Hispanic, LGTBQ, and other underserved and underrepresented patients about rare diseases, diagnostic testing, and the importance of participating in clinical trials. They will also work to improve cultural competency within and across rare disease patient communities through resources and meetings.

MALE CAREGIVERS

Most resources and support for caregivers of children with rare and/or serious illnesses tend to focus primarily on female caregivers, as women are typically the primary family caregiver. Fathers and male caregivers of children with rare and/or serious illnesses are in a unique role. Societal pressures such as toxic masculinity may influence male caregivers' ability to open up, instead making them feel they must project an image of confidence and control while secretly feeling afraid and stressed. Too often, male caregivers are less likely than female caregivers to open up to others when feeling stressed or overwhelmed by caregiving responsibilities.

Too often, male caregivers are less likely than female caregivers to open up to others when feeling stressed or overwhelmed by caregiving responsibilities.

It is important to recognize that everyone needs a support system, including male caregivers. Support may come from a spouse or from other relationships such as online or in-person male support groups. One nonprofit organization that specifically addresses the experience of male caregivers is Jack's Caregiver Coalition. Originally created to support male caregivers of cancer patients, Jack's current target audience focuses on male caregivers for individuals with a catastrophic illness. Jack's mission is to help male caregivers in their caregiver journey through informal get-togethers, a project-based support community, and a resource network. www.jackscaregiverco.org.

The most important first step is to reach out for help when needed.

SINGLE-PARENT CAREGIVERS

Single-parent caregivers face different struggles than two-parent families of children with rare and/or serious illnesses. These may include decreased communication and support between co-parents, or the loss of a co-parent entirely, and often, greater financial and task burdens compared to two-parent families. Additionally, it can be difficult to work while caring for a child with special medical needs. Peer-reviewed literature has shown that single parents experience greater stress than two-parent families affected by chronic illness.[8]

For a single parent to a child with a rare and/or serious illness, it is important to prioritize self-care and engage as much outside support for the child and household as possible. We recognize that single parents may have less access to a readily available care community of individuals who can share in caregiving responsibilities. The time, material resources, and advanced knowledge of disease processes that caregiving for a child with a rare and/or serious illness tends to require means single parents cannot do everything alone. This guidebook is intended to provide many resources meant to offer support, particularly if all of the formal and/or informal support options have been exhausted. The most important first step is to reach out for help when needed. If possible, involve close family or friends as a support system. Scheduling family time and counseling may also be helpful. There are many programs that offer counseling on a sliding scale, and sometimes counseling is covered by health insurance. Single parents may be eligible for Social Security benefits or other forms of financial assistance.

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[8] *Cousino, M. K., & Hazen, R. A. (2013). "Parenting Stress among Caregivers of Children with Chronic Illness: A Systematic Review." Journal of Pediatric Psychology, 38(8), 809–28. Doi: 10.1093/jpepsy/jst049.*

CHILDREN AS CAREGIVERS

While they often go unrecognized, there are approximately 3.4 million children in the United States who assist with the care of a friend or family member.[9] This number does not account for children under 18 years of age who are the primary caregiver. Many of these children are providing care to another child or sibling with a rare and/or serious illness. Support resources and additional information for young caregivers are available from the American Association of Caregiving Youth. www.aacy.org

To help children understand their sibling's rare and/or serious illness, it is helpful to include them in family decisions regarding care of the sibling living with the rare disease.[10] Children may engage in age-appropriate caregiving tasks—if and only if—they would like to do so, and if the tasks don't impede on their school or free time. Children should be educated and made aware of age-appropriate information regarding their sibling's rare disease. Adults need to remain sensitive to age-level comprehension and seek out child-friendly materials. Child caregivers may benefit from connections with other siblings of children with rare and/or serious illnesses. (For more information on supporting siblings, see Achieving Quality-of-Life as a Family Managing Rare and/or Serious Illness in this guidebook).

HELPFUL WEBSITES

American Association of Caregiving Youth
www.aacy.org

Black Women's Health Imperative
www.bwhi.org

Global Genes
www.globalgenes.org

Jack's Caregiver Coalition
www.jackscaregiverco.org

Rare Disease Diversity Coalition
www.rarediseasediversity.org

SibShops
www.siblingsupport.org/sibshops

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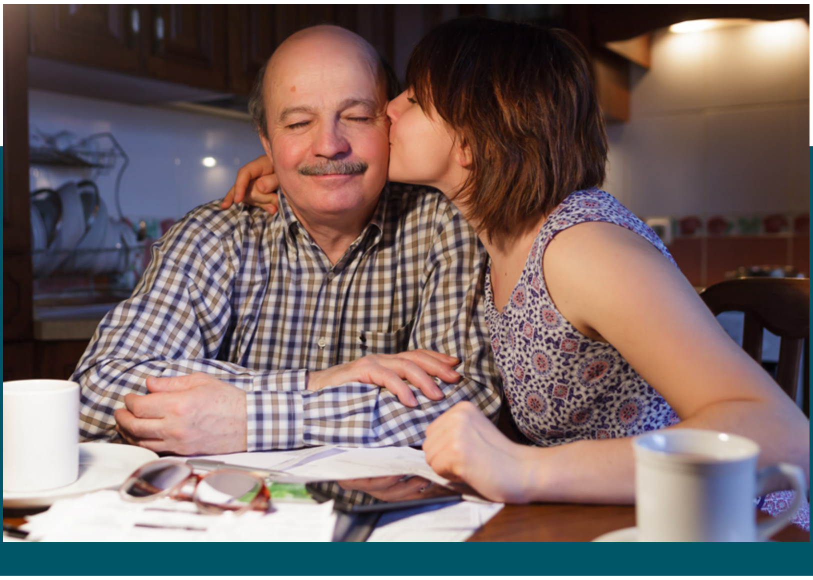

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[9] The National Alliance for Caregiving and AARP. (2020, May). Caregiving in the U.S. 2020. Retrieved February 2, 2021, from https://www.caregiving.org/wp-content/uploads/2020/06/AARP1316_RPT_CaregivingintheUS_WEB.pdf.

[10] Hirsch, L. (ed.). (2015, September). Caring for Siblings of Seriously Ill Children (for Parents). Retrieved February 2, 2021, from https://kidshealth.org/en/parents/sibling-care.html.

GETTING AN ACCURATE DIAGNOSIS

The path to diagnosis for patients and caregivers of rare and/or serious illnesses can be frustrating and chaotic and filled with roadblocks, especially at the beginning.

Caregivers often consult several doctors and specialists, regularly receiving many misdiagnoses and differing opinions before a correct diagnosis is confirmed.

Pediatricians typically have limited knowledge of rare illnesses and often refer families to specialists for testing, diagnosis, and appropriate, detailed ongoing care. Even specialists with years of experience and training can struggle with diagnosing a rare and/or serious illness. Recent reports have found that it can take years of testing and visits to many different specialists to determine an accurate diagnosis. Many rare and/or serious illnesses do not have dependable diagnostic tools or even standard disease treatment plans. As a result, obtaining an accurate diagnosis of a rare and/or serious illness can be a lengthy, confusing, and frustrating process. *The National Economic Burden of Rare Disease Study* found that the average rare disease diagnosis spans more than six years and includes visits to more than seventeen healthcare specialists. Fortunately, there are resources and services for caregivers to explore in conjunction with their health care team to help reach a correct diagnosis.[11]

GLOBAL GENES RARE CONCIERGE SERVICE

RARE Concierge is a service for patients who are either seeking to find an accurate diagnosis of a rare disease or who have additional questions about their rare disease diagnosis. They can obtain immediate and personalized support from Global Genes' network of healthcare providers, researchers and leading patient advocates and foundations. The service is free for anyone seeking a diagnosis and/or information and resources on their rare disease.
www.globalgenes.org/rare-concierge/

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[11] *EveryLife Foundation for Rare Diseases. (2021). The National Economic Burden of Rare Disease Study. https://everylifefoundation.org/wpcontent/uploads/2021/02/The_National_Economic_Burden_of_Rare_Disease_Study_Summary_Report_February_2021.pdf*

Specialists can use genetic testing to confirm a child's diagnosis and help caregivers understand the cause of unexplained symptoms.

CENTERS OF EXCELLENCE (COE)

Centers of Excellence (COE) are programs within healthcare institutions that provide a high concentration of expertise and related resources focused on a particular area. These COEs deliver a holistic approach, allowing specialists across many disciplines to closely collaborate in patient care. Specialists benefit from each other's expertise, with the goal of achieving the best patient outcomes possible. The COEs also act as local, national, and even global experts, and as centers of knowledge that offer services (such as diagnostic testing), treatment, and research in a disease area. Within the context of rare and/or serious illnesses, COEs may effectively address issues in which care providers have limited knowledge. There are COEs for many diseases, including Duchenne muscular dystrophy, Spinal Muscular, Atrophy, and hemophilia; more are planned over time.

Children's National Rare Disease Institute at Children's National Hospital

In 2017, the National Organization for Rare Disorders (NORD) and the Children's National Hospital in Washington, D.C. established the first COE focused on advancing the holistic care and treatment of children and adults with rare diseases as a whole. The Children's National Rare Disease Institute (CNRDI) seeks to reduce the time to diagnosis, works with patients to manage their own care, and provides data to clinicians and researchers to further the diagnosis and development of treatments for rare disease. This group is also developing standards of care for treating rare disease. www.childrensnational.org/departments/rare-disease-institute

RESEARCH CENTERS

A research center is a structured entity within a university or academic institution, independent of a department or other academic organizational structure. Research centers are established to advance scholarly activity primarily through collaboration, training, and dissemination of research within a specific field. Centers are generally led by a director or coordinator; their role is crucial in facilitating interdisciplinary interactions among faculty, scholars, students, and industry to enable research opportunities, academic excellence, and real-world problem solving in these fields. Research centers may focus on a specific disease or disease group, or have a broader focus, such as the Orphan Disease Center.

Orphan Disease Center at the University of Pennsylvania Perelman School of Medicine

The Orphan Disease Center (ODC) was established in 2011 as a nonprofit research center housed within the infrastructure of the UPenn School of Medicine and focused on accelerating therapeutic development for rare diseases. Both researchers and clinicians at the ODC work closely with patient groups and foundations, industry, and the academic community to identify new treatments, as well as develop strategies to overcome the gaps and obstacles that currently exist in the clinical progression of new therapeutics. Initiatives include the JumpStart program, where ODC partners with patient groups and families by providing expert scientific guidance and network outreach to address gaps by identifying key opinion leaders and introducing new researchers to a disease. Specifically, the JumpStart program helps facilitate the development of animal models, establish a patient alliance or a patient registry, and organize symposia. The ODC has also established several research programs of excellence in various rare diseases, including mucopolysaccharidosis disorders and rare immune disorders. Additionally, the ODC has cultivated partnerships with multiple industry stakeholders in gene therapy, genome editing, and mRNA therapeutics. Although primarily a research-based organization, the ODC is committed to creating resources for the rare disease community to enable discovery and preclinical development of potential therapies, as well as the clinical translation of those efforts. www.orphandiseasecenter.med.upenn.edu

TESTING PROGRAMS

Increasing numbers of individuals are taking part in genetic testing as part of an accelerated path to accurate diagnosis. Specialists can use genetic testing to confirm a child's diagnosis and help caregivers understand the cause of unexplained symptoms. Additional testing also helps identify more patients who may be appropriate for trials or natural history studies.

There are many different genetic sequencing tests and genetic labs. A doctor or a genetic counselor can help determine which tests are the most appropriate for the child. Genetic counselors can often help keep costs down by carefully selecting test panels and laboratories, as well as helping caregivers navigate through insurance coverage. A list of genetic counselors is available at: www.nsgc.org/page/find-a-genetic-counselor

Some tests are covered by insurance; others are provided for free or low cost. Ultimately, these efforts support speeding up the pace of clinical trial research and bringing new therapies to market sooner.

Genetic screening may also be sponsored by various stakeholders, including academia, patient foundations, and industry. A few examples include:

- **The Genetic Testing Registry**, a database created by the National Health Institute to connect people to available genetic testing. www.ncbi.nlm.nih.gov/gtr/
- **The Lantern Project**, a partnering initiative between PerkinElmer Genomics and Sanofi Genzyme, provides diagnostic testing for multiple diseases including Fabry, Pompe, Gaucher, Niemann—Pick Type A and B, mucopolysaccharidosis disorders and others. www.perkinelmergenomics.com/lanternproject/
- **The Rare Genomes Project** of the Broad Institute at MIT and Harvard offers free testing and analysis of rare and undiagnosed conditions that are suspected to be related to a single gene. www.raregenomes.org

UNDIAGNOSED DISEASES

Rare and/or serious illnesses can be very difficult to diagnose. Some very rare diseases, called “ultra-rare,” may have just a few or even only one diagnosed instance in the entire world. Caregivers in this “undiagnosed limbo” who are searching for answers should not give up. Science continues to evolve and discover new treatments. There are also several available resources regarding undiagnosed diseases.

Rare & Undiagnosed Network

www.rareundiagnosed.org

RUN raises awareness for families with children affected with undiagnosed or rare diagnosed conditions. RUN priorities include urging insurance companies to reimburse genome sequencing in clinical settings, and networking with researchers, insurance providers, and industry stakeholders to meet families’ needs.

Undiagnosed Diseases Network

www.undiagnosed.hms.harvard.edu

UDN, funded through NIH Common Fund, is designed to accelerate discovery and innovation in the diagnosis and treatment of patients with previously undiagnosed diseases.

HELPFUL WEBSITES

Access to Critical Therapies:
Guiding Principles of Rare Disease Care and Patient Care
www.globalgenes.org/resources/guiding-principles-of-rare-disease-care-and-patient-access/

Children’s National Rare Disease Institute
www.childrensnational.org/departments/rare-disease-institute

Diversity of Odysseys:
The Diagnostic Challenges of Underserved People with Rare Diseases
www.globalgenes.happyfox.com/kb/article/272-diversity-of-odysseys/

Find a Genetic Counselor
www.nsgc.org/page/find-a-genetic-counselor

Genetic Testing Registry
www.ncbi.nlm.nih.gov/gtr/

Global Genes RARE Concierge
www.globalgenes.org/rare-concierge/

Lantern Project
www.perkinelmergenomics.com/lanternproject

Orphan Disease Center
www.orphandiseasecenter.med.upenn.edu

Rare and Undiagnosed Disease Network
www.rareundiagnosed.org

Rare Genomes Project
www.raregenomes.org

Undiagnosed Diseases Network
www.undiagnosed.hms.harvard.edu

> **Some very rare diseases, called “ultra-rare,” may have just a few or even only one diagnosed instance in the entire world. Caregivers in this “undiagnosed limbo” who are searching for answers should not give up. Science continues to evolve and discover new treatments.**

GETTING ACCURATE INFORMATION ON A CHILD'S DISEASE

Receiving an accurate diagnosis can provide an initial sense of relief. However, everything changes when a child is diagnosed with a rare and/or serious chronic medical condition or a life-threatening illness.

One of the first steps a caregiver may want to take after receiving a diagnosis is to find more information about the child's disease. This can be a daunting task considering the relatively limited amount of accurate information available from various sources, online and otherwise. Caregivers will likely find individual genetic counselors and social workers who work in medical care systems where children with rare and/or serious illnesses are diagnosed or treated to be key resources when learning about a specific disease. Caregivers may also wish to search independently for more information. There are many online resources and research organizations, as well as patient advocacy organizations, that can provide both information and support.

THE GENETICS AND RARE DISEASES INFORMATION CENTER

The Genetics and Rare Diseases Information Center (GARD) is a program of the National Center for Advancing Translational Sciences (NCATS) and the National Human Genome Research Institute (NHGRI) at the National Institute of Health (NIH). The GARD provides access to current, reliable, and easy-to-understand information about rare or genetic diseases, in both English and Spanish languages. It catalogs videos, brochures, and other informational resources by disease. Resources provided may include information on genetic testing and diagnosis, conferences, advocacy organizations, research studies, and FDA-approved treatments. www.rarediseases.info.nih.gov

These groups may provide information and resources for specific medical conditions but tend to focus most on helping the rare disease community.

UMBRELLA PATIENT AND ADVOCACY ORGANIZATIONS

Umbrella organizations represent rare and/or serious illnesses in general. These groups may provide information and resources for specific medical conditions but tend to focus most on helping the rare disease community. Umbrella groups often work together and with specific rare and/or serious disease groups on advocacy and policy initiatives.

Caregiver Action Network

www.caregiveraction.org

CAN is a national family caregiver group working to improve the quality of life for individuals who care for loved ones with chronic conditions and disabilities. It provides a Family Caregiver Toolbox for common issues in caregiving, informational resources for self-care, addressing caregiver depression, and managing the cost of care.

EveryLife Foundation for Rare Diseases

www.everylifefoundation.org

EveryLife is dedicated to empowering the rare disease community to advocate for impactful, science-driven legislation and policy that advances the equitable development of and access to lifesaving diagnoses, treatments, and cures.

EURORDIS

www.eurordis.org

EURORDIS is a European nonprofit alliance of over 900 patient organizations in 73 countries advocating for access and treatment for rare diseases.

Genetic Alliance

www.geneticalliance.org

Genetic Alliance advocates for health benefits in the accelerating field of genomic research. The organization is involved in research, advocacy, programs and workshops.

Global Genes

www.globalgenes.org

Global Genes provides patients, families, and specific rare disease organizations with information and resources worldwide and collaborates with all members of the rare disease community to share data and experiences and support advocates through programming, resources and events. Their RARE Concierge service (www.globalgenes.org/rare-concierge) is for patients who are either seeking to find an accurate diagnosis of a rare disease or who have additional questions about their rare disease diagnosis. Patients can obtain immediate and personalized support from Global Genes' network of healthcare providers, researchers and leading patient advocates and foundations.

The National Organization for Rare Disorders

www.rarediseases.org

NORD focuses on the identification, treatment, and cure of rare disorders through programs, advocacy, research, and patient services. This includes patient and family assistance programs and a Rare Caregiver Respite Program.

Patient Worthy®

www.patientworthy.com

This online publication provides relevant information to rare disease patients, caregivers, and advocates. Through awareness and humor, the group advocates for research for new treatment options and promotes awareness.

RareConnect

www.rareconnect.org

RareConnect promotes global conversation and collaboration via online communities for patients and families with rare medical conditions so they can connect with others and share their experiences. The project is a collaboration between EURORDIS and Care4Rare Canada.

EveryLife, Global Genes, and NORD work with individual rare disease advocacy organizations and support groups that may be useful resources in the identification of relevant, disease-specific support or advocacy. All three organizations house databases for various rare diseases that include descriptions of the diseases, as well as information on known disease-specific organizations and resources.

DISEASE-SPECIFIC SUPPORT GROUPS

Disease-specific support and advocacy groups are an important source of emotional and informational support to caregivers of people with rare and/or serious illnesses. A recent review found that rare and/or serious disease support groups help caregivers by:

- Providing interaction with other people with similar experiences.
- Sharing current information about diseases and related treatment options.
- Facilitating opportunities to give and receive emotional support.
- Providing a forum for caregivers to speak openly about their experiences and feelings.
- Teaching useful coping skills.
- Offering opportunities for learning about and participating in advocacy efforts.

Collectively, the impact of disease-specific support groups can help caregivers feel empowered and hopeful.[12] Our own focus groups revealed similar sentiments, with caregivers stating that disease-specific support groups can help address feelings of isolation and provide hope through directly interacting with others who share and subsequently understand the same experiences.

Caregivers in our focus groups also discussed the benefits of disease-specific advocacy groups in supporting research for treatments and therapies, navigating the best standards of care for their child, and in giving specific guidance for individual and whole-group advocacy initiatives.

Disease-specific organizations typically operate as charitable nonprofit entities or support groups. They are typically funded through direct sponsorships, grants, and donations from pharmaceutical, biotech, industry, specialty pharmacy, and other affiliated corporate organizations. Some receive direct grants and donations from other corporate or foundation organizations, government grants, and individual charitable giving.

Charities often raise a significant amount of money to support their causes through fundraising events. These may be walks, runs, game or trivia nights, or other fun and engaging activities that bring individual supporters of a cause together to raise money. These activities are important to the charities but can also be rewarding and inspiring to the attendees. In addition, they are also a great way for caregivers to network and meet, in a positive environment, others impacted by a disease.

Families living with rare and/or serious illnesses look to support and advocacy groups for different reasons. For some, it may be to find medical information, treatment options, the latest research, or financial aid resources. It may also offer ways to connect with other people who understand what having the illness means or to join local advocacy networks that are working to change policy. While there are benefits to interacting with communities of disease-specific support groups, it is possible that caregivers may not want to immediately connect with disease-specific groups or other supports. This is perfectly normal and should happen at the caregiver's own pace.

The websites for these disease-focused groups are a great place to start. They often list detailed information and current research about a given disorder, available resources, share information about conferences, meet-ups, and provide online support groups for individuals and families affected by a specific disorder. Following are three examples of disease-specific organizations and their initiatives.

> **The websites for these disease-focused groups are a great place to start.**

The Global Foundation for Peroxisomal Disorders (GFPD), founded in 2010 for those impacted by peroxisomal disorders in the Zellweger spectrum and related single enzyme protein deficiencies, is focused on family engagement. Activities like support groups, webinars, parent-to-parent support navigators, an equipment exchange program, a registry, a biennial family conference, regional meet-ups, as well as access to researchers and physicians help patients and caregivers cope with the challenges of living with a rare disease. The organization also supports publishing treatment guidelines and funding relevant research initiatives. www.thegfpd.org

The National MPS Society was founded in 1974 and exists to cure, support, and advocate for mucopolysaccharidosis (MPS) and mucolipidosis (ML). Members receive support through this nonprofit patient advocacy organization available to individuals with these diagnoses, their caregivers, and families. Through scholarship and grant programs, the Society enables members to access family conferences and obtain educational advancement, receive durable medical equipment and reimbursement for medical travel, among other resources. The Pathways program offers face-to-face connection with direct social work support throughout the first year of diagnosis. Services for families and individuals continue throughout the lifetime and into bereavement support. Funding for research is provided on an international level, bringing forth the most up-to-date science and information as the organization seeks to find cures for all syndrome types. www.mpssociety.org

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[12] *Delisle, V. C., Gumuchian, S. T., Rice, D. B., Levis, A. W., Kloda, L. A., Körner, A., & Thombs, B. D. (2017). "Perceived Benefits and Factors that Influence the Ability to Establish and Maintain Patient Support Groups in Rare Diseases: A Scoping Review. The Patient-Patient-Centered Outcomes Research, 10(3), 283–93. Doi: 10.1007/s40271-016-0213-9.*

The National Niemann-Pick Disease Foundation (NNPDF) is a nonprofit patient advocacy and family support organization dedicated to supporting and empowering patients and families affected by Niemann-Pick disease through education, collaboration, and research. Founded in 1993, NNPDF serves families throughout the nation at all stages of their Niemann-Pick journey. The NNPDF is the U.S. member organization of the International Niemann-Pick Disease Alliance (INPDA). www.nnpdf.org

CONFERENCES

Each year, organizations for rare and/or serious disease host events to bring together patients, caregivers, medical care providers, industry partners, government, and other stakeholders to discuss key initiatives pertaining to current issues and advancement opportunities in rare and/or serious disease diagnosis, treatment, care, drug development, and policy. Each of the national groups typically draw a thousand or more attendees and exhibitors. Many national disease-specific or local and state-based organizations also have conferences, workshops, or advocacy events.

Attending a conference or advocacy event can be a truly empowering experience and serve as a brief respite from daily caregiving. These advocacy events are often filled with networking opportunities and camaraderie. Many patients and caregivers find these events to highlight relatable experiences and to be a place where they find a sense of belonging and understanding.

Some annual national events include:

Living Rare, Living Stronger Patient & Family Forum

www.livingrare.org

Hosted by the National Organization for Rare Disorders (NORD), this Patient and Family Forum takes place in a major U.S. city every June.

Rare Disease Day

www.rarediseaseday.org

The main objective of Rare Disease Day is to raise awareness among the public and decision makers about rare diseases and their impact on patients' lives. Started in 2008 by EURORDIS, today there are numerous partners across the United States who hold events to promote awareness for rare diseases. The event is held on the last day of February each year.

Rare Disease Week

www.everylifefoundation.org/rare-advocates/rare-disease-week/

Rare Disease Week occurs on Washington, D.C.'s Capitol Hill, typically each February, in conjunction with Rare Disease Day. The week is filled with networking and scheduled advocacy appointments with members of Congress. It is hosted by the Rare Disease Legislative Advocates (RDLA), a program of the EveryLife Foundation.

Attending a conference or advocacy event can be a truly empowering experience and serve as a brief respite from daily caregiving.

Rare Diseases and Orphan Products Summit

www.nordsummit.org

Hosted by NORD, the Summit meets in Washington, D.C. each October to discuss critical rare disease policy issues.

RARE Drug Development Symposium

www.globalgenes.org/event/rare-disease-drug-development

Hosted by Global Genes and the Orphan Disease Center of the University of Pennsylvania, the RARE Drug Development Symposium includes workshops for rare disease community members in the earlier stages of their research efforts or careers and looking to build knowledge in core competencies. In addition, the event aims to address the current landscape of rare drug development and allow participants to interact with subject matter experts and other rare disease stakeholders to help them better understand and develop their impact and role in advancing potential treatments.

RARE on the Road

www.globalgenes.org/rare-on-the-road

Hosted by Global Genes and the EveryLife Foundation, RARE on the Road has united and activated the rare disease community on the local level. States are chosen each year with community feedback and with an aim to build a more inclusive and diverse rare disease community. Discussions focus on state-specific issues and resources relevant to patients, caregivers, and advocates with diverse backgrounds and needs impacted by a rare disease. Participants take part in robust conversations with other local rare disease community members about what it means to be an advocate, practice telling their own rare stories and learn about relevant policy updates.

RARE Patient Advocacy Summit
www.globalgenes.org/event/patient-summit

Hosted by Global Genes, this is the largest rare patient advocacy event in the United States. The event includes a variety of interactive and educational events, meet-ups, workshops, and networking opportunities. Patients, caregivers, and advocates have the opportunity to connect and engage with others in the rare disease community, experiencing a sense of community and belonging. The event allows participants to gain insights about the latest in rare disease innovations, best practices for advocating on an individual and organizational level, and actionable strategies they can implement immediately to accelerate change.

The cost of attending a conference is a common concern. There are numerous programs and support systems available to help pay for the cost of travel and housing while at a conference. Most of the national umbrella organizations and a great many of the disease specific groups provide a limited number of scholarships to families who would otherwise be unable to attend. For example, NORD offers a Rare Caregiver Respite Program, which assists caregivers with funding to either attend a conference or have a day away from their caregiving responsibilities.

Email CaregiverRespite@rarediseases.org
or find more on this patient assistance program at: www.rarediseases.org/wp-content/uploads/2019/05/NRD-1182-RespiteCare_SS_1up_FNL_NoCrops_Fixed.pdf.

See Economic Impact of Caregiving for a Child with a Rare and/or Serious Illness in this guidebook for more information on financial resources for families.

Again, it should be noted that, despite the opportunities that large conferences and events can offer caregivers for children with rare and/or serious illnesses, the desire to participate in activities will vary from caregiver to caregiver. If a caregiver feels that attending a large conference may not be appropriate for his or her needs, there are many other useful resources for obtaining support and accurate information.

Social media has made it easy to connect with others around the world experiencing the same rare and/or serious disease. Facebook groups allow caregivers to connect to special interest groups, including those focused on specific diseases.

ONLINE RESOURCES AND SOCIAL MEDIA

The internet can be an extremely useful resource for caregivers of children with rare and/or serious illness, especially if support and expertise on a specific condition cannot be found through local clinicians. A recent literature review reported that online information and emotional support can be empowering to caregivers of children with special health care needs in their ability to care for their child.[13]

Our focus groups with caregivers found that online resources can offer:

- new knowledge
- support from individuals in similar situations
- hope through sharing and learning about the personal stories of other caregivers

Social media has made it easy to connect with others around the world experiencing the same rare and/or serious disease. Facebook groups allow caregivers to connect to special interest groups, including those focused on specific diseases. Most social media sites, such as Twitter and Instagram, also employ the use of tagging. Searching for a disease on these sites through

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[13] *DeHoff, B. A., Staten, L. K., Rodgers, R. C., Denne, S. C. (2016). "The Role of Online Social Support in Supporting and Educating Parents of Young Children with Special Health Care Needs in the United States: A Scoping Review." Journal of Medical Internet Research, 18(12), e333. Doi:10.2196/jmir.6722.*

its appropriate tag (#raredisease, #careaboutrare, #rarecancer, #chronicillness, #rarediseaseawareness, #rarediseaseadvocacy, to name a few) connects users who are engaging on the topic. The broader community can also be reached through tags for rare disease-specific events and days. Some rare and/or serious disease communities have created their own social media platforms. Sites such as Inspire were created specifically for the purpose of connecting individuals experiencing the same disease through message boards, so support and conversation on similar experiences can be shared. More information can be found at www.inspire.com.

Social media can also provide a platform to quickly release new information. New disease information, studies, and potential treatments for a disease can often be found on social media. Most rare disease advocacy and research groups can be followed on Facebook and Twitter, making their information easy to find. Many organizations distribute newsletters that provide relevant updates and are delivered through free subscriptions. Some doctors, patient advocates, and researchers can also be followed on social media, putting information at the caregiver's fingertips.

Although online resources can be highly useful, remember to be cautious when evaluating the use of internet-based resources in the management of care for a child. Social media content sourcing is primarily open to the public and may contain inaccurate or potentially biased information. Moreover, some inaccurate information about the child's condition may be upsetting in nature. Caregivers of rare and/or serious illnesses have expressed concerns about the quality of internet-based content as well as the potential for unfavorable social media interactions.[14,15] It may be challenging to know if internet and social media-based content is accurate. It's important to consult with the child's medical care team, especially when information may be related to the care and treatment of the child.

> **Although online resources can be highly useful, remember to be cautious when evaluating the use of internet-based resources in the management of care for a child.**

HELPFUL WEBSITES

Caregiver Action Network
www.caregiveraction.org

EveryLife Foundation for Rare Diseases
www.everylifefoundation.org

EURORDIS
www.eurordis.org

Genetic Alliance
www.geneticalliance.org

Genetics and Rare Diseases Information Center (GARD)
www.rarediseases.info.nih.gov

Global Foundation for Peroxisomal Disorders
www.thegfpd.org

Global Genes
www.globalgenes.org

Inspire
www.inspire.com

Living Rare, Living Stronger Patient & Family Forum
www.livingrare.org

National Niemann-Pick Foundation
www.nnpdf.org

National Organization for Rare Disorders
www.rarediseases.org

Patient Worthy
www.patientworthy.com

RareConnect
www.rareconnect.org

Rare Disease Day
www.rarediseaseday.org

Rare Diseases and Orphan Products Summit
www.nordsummit.org

Rare Disease Week
www.everylifefoundation.org/rare-advocates/rare-disease-week/

RARE Drug Development Symposium
www.globalgenes.org/event/rare-disease-drug-development

RARE on the Road
www.globalgenes.org/rare-on-the-road

RARE Patient Advocacy Summit
www.globalgenes.org/event/patient-summit

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[14] *Castro, A. R., Chougui, K., Bilodeau, C., & Tsimicalis, A. (2019). "Exploring the Views of Osteogenesis Imperfecta Caregivers on Internet-based Technologies: Qualitative Descriptive Study." Journal of Medical Internet Research, 21(12), e15924. Doi: 10.2196/15924.*

[15] *Knapp, C., Madden, V., Wang, H., Sloyer, P., & Shenkman, E. (2011). "Internet Use and eHealth Literacy of Low-income Parents whose Children have Special Health Care Needs." Journal of Medical Internet Research, 13(3), e75. Doi: 10.2196/jmir.1697.*

MANAGING HEALTH AND MEDICAL CARE FOR A CHILD

There have been many treatment advances in recent years, with hundreds of newly approved drugs for rare and/or serious illnesses.

However, it can be difficult to get an accurate diagnosis and find doctors with the right expertise to help navigate a child's ongoing care and treatment. There are few therapies on the market today for most rare and/or serious illnesses, but the potential for treatment grows as research, science, and technology continue to advance. For diseases with no therapy, symptom management may still be an important part of care for a child.

There can also be numerous challenges to accessing care and therapies. Some specialty care may be accessible only at certain hospitals and drugs only at certain pharmacies. Insurance companies vary widely in their in-network decisions about clinicians and drug cost coverage. Prior authorization requirements by an insurance provider that reviews whether a medication is appropriate for therapy can be confusing and take time to process.

All of these aspects of care management can be challenging for families with a child diagnosed with a rare and/or serious illness, and caregivers have a very important role as the primary decision maker in the care of the child. It is important to remember that the primary caregiver is the expert on the child's specific needs and typically will be the very best advocate for the child.

Nevertheless, it is common for all caregivers to be concerned about whether they are making the best decisions for their children. One study at Children's National Hospital found that among parents and clinicians of patients in the pediatric intensive care unit, decision making was best informed when parents were:[16]

- Putting their child's needs first.
- Advocating for their child with their child's clinical team.
- Focusing on their child's quality of life.
- Collaborating on strategies that promote effective communication with the clinical team.

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[16] *October, T. W., Fisher, K. R., Feudtner, C., & Hinds, P. S. (2014). "The Parent Perspective: 'Being a Good Parent' when Making Critical Decisions in the PICU." Pediatric Critical Care Medicine, 15(4), 291–98. Doi: 10.1097/PCC.0000000000000076.*

FINDING QUALITY MEDICAL CARE PROVIDERS, HOSPITALS, AND MEDICAL CENTERS

High quality care means receiving the right care, treatment, and support at the right time. It is not unusual to have follow-up care appointments with a specialist every six to twelve months in addition to urgent care needs. The best possible outcomes emerge when care is coordinated. This means doctors, nurses, laboratories, radiology services, specialty services, genetic counselors, geneticists, and hospital administrators need to be working together. Great care includes these medical providers, along with the caregiver, working as a united team to provide the best care possible in the most cost-effective and timely way. If team members and other providers are not talking with each other, the child may be subjected to unnecessary tests, medication errors, or costs. Consistent and coordinated communication is vital.

Unfortunately, the American healthcare system is highly fragmented. Many doctors and medical personnel believe deeply in coordinated care and are working to bring the system together. However, as the system currently exists today, it falls to the caregiver to be a strong, determined, and dedicated advocate for the child and to coordinate the various aspects of care. This is particularly true if the child is seeing many medical providers across multiple clinics and hospitals. Patient organizations are typically very familiar with these challenges and can be a great support resource on how to successfully coordinate care across clinicians, hospitals, and other service providers.

Approaches to healthcare and treatment options can change rapidly. Staying current on the latest care and therapies is important and can be a challenge for busy doctors. Doctors and specialists who work collaboratively as a team find this to be an effective way to stay current on new treatment and research. Technology sharing, including electronic medical records and interactive websites, provides information to the general public as well as healthcare professionals. This results in better care and treatment.

Miscommunication between healthcare professionals and families may also be a possible challenge in obtaining quality care for a child. One study found that parents had a hard time getting medical professionals to understand their experience caring for their child with a rare and/or serious illness at home. The medical professionals' misunderstanding of the situation or disease, along with the constant need to repeat themselves, would fatigue parents and cause them to feel that their situation had become impersonal.[17]

Despite these challenges, it is important to remember that the caregiver should be a key member of the healthcare team and insights from patients and caregivers should be central in shared decision making. A quality clinic will regard patients and caregivers as experts. Issues identified as needing change can help the clinic improve and ultimately better serve the child.

CHECKLIST TO CONSIDER WHEN LOOKING FOR A QUALITY HEALTHCARE TEAM

- ☐ Doctors and centers caring for your child have access to medical records including information about medications, labs results, urgent care visits, and after hours calls.
- ☐ A coordinated care team exists with doctors, specialists, and other medical providers.
- ☐ A commitment to good communication exists between all medical providers that care for your child's condition.
- ☐ Health care services, such as X-ray and imaging, lab, specialty, or services are at or nearby the child's medical center.
- ☐ Ongoing preventive care and clear treatment instructions are provided.
- ☐ Doctors and specialists keep themselves up to date on the latest in medical advancements and research findings being done.
- ☐ A backup doctor or specialist is available when a primary doctor is not. This provider has access to the child's medical records and history.
- ☐ Medical providers hear, listen, and respond to your questions and concerns.
- ☐ The medical team helps caregivers make informed decisions by discussing the pros and cons of various care and treatment options.
- ☐ Caregivers and patients have access to medical advice and guidance 24 hours a day.
- ☐ Caregivers and patients have easy access to medical records and lab test results online.

www.accountablecaredoctors.org

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[17] *Currie, G. & Szabo, J. (2019). "'It Would be much Easier if we were Just Quiet and Disappeared': Parents Silenced in the Experience of Caring for Children with Rare Diseases." Health Expectations, 22(6), 1251–59. https://doi.org/10.1111/hex.12958.*

While there is no central list of expert health professionals for all rare and/or serious illnesses, many patient advocacy groups will have knowledge of the best specialists. In specialty areas, medical doctors are often doing research studies (clinical trials) and may be looking for study participants. The Genetic and Rare Diseases Information Center (GARD) provides easy-to-understand information about rare diseases as well as tips on how to find a disease specialist.

www.rarediseases.info.nih.gov/guides/pages/96/patients-families-and-friends

Building A Medical Care Team

Finding knowledgeable specialists and learning to make informed treatment decisions while taking care of a child can be challenging. However, no one is alone. There are resources, services, and experts that can help. A child's care team may include many specialists and additional medical care providers. Depending on the child's diagnosis, experts may be available locally, or caregivers may need to travel to meet with a disease specialist. Specialists are beneficial as local doctors are often not as knowledgeable about rare and/or serious illnesses.

Rare and/or serious illnesses involve a range of services including assessment, diagnosis, patient care, treatment, testing, support services, and inpatient care. It is helpful to have a primary person on the team for communication; someone who is trustworthy, accessible, and who understands the child and his or her family's needs. This is often a case manager or nurse coordinator. By building a meaningful, trusting relationship, solutions are found and together, positive outcomes follow.

POTENTIAL CORE CARE TEAM MEMBERS

Case Manager	A case manager can help manage many aspects of the child's care. This may include coordinating providers and services or addressing insurance issues. Case managers are often social workers or nurses. Caregivers may find a case manager through the child's hospital, social services, or insurance provider at no cost. Case managers can also be hired privately.
Clinical Geneticist	A doctor who meets with patients to evaluate, diagnose, and manage genetic disorders and has specialized training in genetics.
Clinical Psychologist	A licensed, doctoral-level professional trained to evaluate mental health status using clinical interviews, psychological evaluation, and testing. Psychologists are qualified to diagnose mental health conditions and provide psychotherapy.
Disease Specialist	A doctor who has advanced training and experience in a specific field of medicine. Often these doctors are also involved in clinical research.
Genetic Counselor	Genetic counselors specialize in genetics and counseling to provide personalized help to families as they make decisions about genetic health.
Nurse and/or Financial Navigator	Financial navigators can help families understand out-of-pocket cost and what health insurance covers. They can also help families navigate expenses and financial difficulties in the health care system to reduce stress and hardship.
Nurse Coordinator	Often a nurse coordinator who works directly with a specialist doctor or clinic will be a primary and important contact and resource. This nurse supports communication among a child's care team.
Palliative Care Specialist	Palliative care specialists focus on providing relief from the symptoms and stress of serious illnesses. They often work with a team of doctors, nurses, and other care providers with an overarching goal of improving the quality of life for both the patient and the family.
Pediatrician	Generalist medical doctor who manages the physical, behavioral, and mental health for children until age 18.
Social Worker	Social workers can assist with locating resources, completing paperwork, and many other tasks as well as providing emotional support. Social workers are typically found in the hospital or university where the child's doctor works.

Hospitals and Medical Centers

An identified network of specialized disease care medical centers can greatly benefit patients with rare and/or serious illnesses by providing collective expertise and access for patients. Numerous medical centers and rare and/or serious disease foundations have collaborated to establish networks of accredited specialty clinics or comprehensive care centers focused on a specific disease area or have become affiliated with rare disease centers across the country. These centers bring doctors and their collective experiences together, and patients are able to get the high-quality care they need.

These centers also allow caregivers to connect and share experiences, information, and advice with each other. For patient families, both the convenience of specialists working together and sharing their expertise are significant benefits. A child's clinic appointments are often organized on a single day when all of their specialists are available. In addition to medical care, these clinics play an important role in educating both the caregiver and the child about the disease. They often work closely with patient advocacy groups and promote public awareness of a specific disease or disease area. In addition to the Rare Disease Institute at Children's National Hospital, several institutions have been designated Centers of Excellence (COE) for diseases relevant to rare and/or serious illnesses. The clinicians often work closely with patient advocacy groups and promote public awareness of a specific disease or disease area.

As mentioned earlier, these centers provide a high concentration of expertise and related resources focused on a specific area, delivering care in a comprehensive, interdisciplinary way. For rare and/or serious illnesses, important initiatives to achieve COE status likely include that the center demonstrates it can reduce the duration of the path to diagnosis, work collaboratively with patients and families in managing their own care, and provide data to clinicians and researchers to further advance the development of treatments, as well as develop standards of care for rare and/or serious illnesses.

Collaboration Across Professionals

Families will likely need to interact with multiple healthcare professionals, specialists, and agencies in the management of a child's rare and/or serious illness. As a result, the coordination of medical care for a child with a rare and/or serious illness often falls on the caregiver. This can take a toll on the caregiver, especially when considering the numerous other responsibilities caregivers have. Caregivers of children with different rare diseases have expressed frustration about having to actively facilitate communication and collaboration across multiple specialists in the care of their child.[19,20]

To address this, it may help to work with the child's medical care team to develop a care coordination plan. According to the Agency of Healthcare Research and Quality, care coordination consists of deliberately organizing patient care activities and sharing information among all of the stakeholders in the patient's care to achieve safer and more effective care. A care coordination plan may involve:

- Setting up a schedule for health care provider visits, medications, and/or treatments
- Providing resources to find local services or providers
- Referrals to specialists or other health care providers, community resources, and case managers who can assist in finding care and services
- Getting insurance approvals for care or services
- Helping coordinate care among various providers

Implementing a care coordination plan may be an effective strategy in helping improve a child's outcomes, fostering collaboration between providers, and empowering the caregiver to be part of the decision-making process in the child's care management.
www.ahrq.gov/ncepcr/care/coordination.html

The Children's Hospital of Philadelphia Leukodystrophy Center (CHOP)

Leukodystrophies are inherited disorders of the white matter of the brain and spinal cord that can cause problems with movement, speaking, vision, hearing, and overall development. These disorders are difficult to recognize and diagnose. The Leukodystrophy Center at Children's Hospital of Philadelphia (CHOP) provides comprehensive clinical care, diagnostic testing, and the most advanced treatments available to infants, children, and adolescents living with these inherited white matter diseases. Here, a child has access to doctors and nurses experienced in caring for individuals with leukodystrophies and who are actively involved in ongoing research to find new treatments.

www.chop.edu/centers-programs/leukodystrophy-center

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[19] bid.

[20] Khangura, S. D., Tingley, K., Chakraborty, P., Coyle, D., Kronick, J. B., Laberge, A. M., & Siddiq, S. (2016). *"Child and Family Experiences with Inborn Errors of Metabolism: A Qualitative Interview Study with Representatives of Patient Groups." Journal of Inherited Metabolic Disease, 39(1), 139–47. Doi: 10.1007/s10545-015-9881-1.*

HOME HEALTH CARE

It is quite common for caregivers of children with rare and/or serious illnesses to receive some hands-on training in the hospital or clinic before the child comes home. Typically, this is adapted to the needs of the child and the home setting. This may include medical tasks such as caring for a tracheostomy, managing a gastrostomy tube (G tube) and feedings, or other care, as well as the use of medical devices such as a suction machine. Once home, the caregiver may receive support and additional coaching from home health care staff, such as nurses or aides. Caregivers may also seek educational resources in different aspects of home medical care from organizations such as the Oley Foundation (www.oley.org) or the Global Tracheostomy Collaborative (www.globaltrach.org)

Pediatrics has been one of the fastest-growing areas of home health care for many years. Home care may reduce costs and inpatient hospitalizations. Families often prefer to manage pediatric medical care at home when possible. Through Early Periodic Screening, Diagnostic and Treatment (EPSDT), a federal Medicaid program, states are required to cover the necessary services to correct or improve physical and mental illnesses and conditions. Some services that may be covered are home health nursing, personal care services, and case management services.

Skilled Nursing Care at Home

Depending on the child's medical condition, a licensed practical nurse (LPN) or a registered nurse (RN) may be needed at home. Skilled nursing care might include intravenous infusions, tracheostomy care, managing gastrostomy tubes for feeding, or other care and procedures commonly done by a nurse or specially trained caregiver.

Before a nurse begins working in a home, it is helpful to arrange a "meet and greet." During this visit, caregivers can ask about the nurse's experience caring for children. It's important to be specific about the child's needs and share expectations regarding role and responsibilities. Often, a child will have several nurses sharing the needed care hours.

When caregivers decide to add a home care nurse to the child's care team, they want to be sure the nurse is experienced in meeting the child's disease-specific needs. Begin by checking whether the home health agency is accredited with an organization such as The Joint Commission (www.jointcommission.org) or the National Association for Home Care & Hospice (www.nahc.org).

Caregivers will also want to confirm with the child's insurance provider that home care is a covered benefit. This includes the number of hours per day/week/month nursing care covered and if there are any restrictions. Skilled home nursing may be covered by private insurance or by Medicaid depending on state eligibility criteria. If the child is in a hospital, there is often a representative or social worker at the hospital that can assist with this process of determining coverage.

Home health care aides provide help or respite during periods when the burden of caregiving tasks can become both physically and emotionally overwhelming.

Home Health Care Aide

Home health care aides provide help or respite during periods when the burden of caregiving tasks can become both physically and emotionally overwhelming. Home health care aides do not perform medical tasks or services that require a nurse; they provide aid that can help ease caregiver stress, which may allow caregivers to care for their children successfully and sustainably. Home health care services may provide respite care for the caregiver, including assisting in daily activities for the child such as bathing, grooming, feeding, and dressing. Aides can also provide respite care for the caregiver, light housekeeping, and run errands. Home health care services may be covered by private insurance or by Medicaid depending on state eligibility criteria.

TREATMENT AND RESEARCH

For many rare and/or serious diseases, there are no direct treatments for the disease itself but there are treatments that may exist for symptoms and complications resulting from the condition. Most rare and/or serious disease therapies today are small molecules or biologic medications, but future therapies are likely to use more innovative approaches, such as gene therapy and tissue and cell-based therapies. Creating therapies for rare and/or serious diseases is complicated, expensive, and takes time.

There is good reason to hope for the future due to the increased pace of new scientific research, discovery and understanding, and new technologies to support advancements. Many research networks and groups are actively studying rare and/or serious illnesses and advancing the understanding of diseases. The FDA Office of Orphan Products Development (OOPD) exists to advance the evaluation and development of promising products for rare diseases. www.fda.gov/industry/developing-products-rare-diseases-conditions

Many research networks and groups are actively studying rare and/or serious illnesses and advancing the understanding of diseases.

GROUPS AND PROGRAMS ADVANCING THE UNDERSTANDING OF RARE DISEASES

All of Us Research Program www.allofus.nih.gov	All of Us Research Program is an NIH initiative to build a diverse health database that includes one million people from all backgrounds across the United States. Data provided will help identify risk factors, develop better treatments, determine what treatments work best for people with different backgrounds, and find ways to prevent different diseases.
Data DIY www.globalgenes.org/data-diy	Data DIY is a program developed by Global Genes to support collaboration between patients, caregivers and partners, clinicians and researchers in order to accelerate results in drug development for rare diseases. The challenge for patient advocates and organization leaders is to be as technically savvy about data as the rest of these passionate stakeholders. Knowledge is power, and Data DIY wants its audience to be empowered data owners and stewards.
JumpStart at the Orphan Disease Center www.orphandiseasecenter.med.upenn.edu/jumpstart	Programs such as JumpStart at the Orphan Disease Center at the University of Pennsylvania's School of Medicine look to establish research agendas in emerging and neglected rare diseases.
Rare Diseases Clinical Research Network www.rarediseasesnetwork.org	RDCRN is an NIH-funded research network focused on groups of rare disorders. The program is designed to advance medical research on rare diseases by providing support for clinical studies and facilitating collaboration, study enrollment, and data sharing.
Rare Diseases Registry Program www.ncats.nih.gov/radar	RaDaR at NIH aims to define best practices for patient registries. RaDaR also strives to identify and adopt standards to support high-quality registries for rare diseases therapeutics development.
National Human Genome Research Institute www.genome.gov	NHGRI at NIH supports the development of resources and technology that will accelerate genome research and its application to human health. A critical part of the NHGRI mission continues to be the study of the ethical, legal, and social implications (ELSI) of genome research.

CLINICAL TRIALS

As therapies are developed, they undergo a process of evaluation and approval by the FDA before they are made available to the public. This requires conducting clinical trials on the new experimental therapies. This may be an option for some children, particularly if other treatments do not work or there are no other treatment options. Locating researchers and gaining access to appropriate clinical studies can require dedication and flexibility. It is important to know that trials typically have a limited number of seats available and have specific requirements. Several caregivers in our focus group noted that participating in clinical trials, regardless of the outcome, helped them feel actively involved in their child's care. A database of privately and publicly funded clinical studies conducted around the world can be found at www.clinicaltrials.gov.

Caregivers may feel hesitant to enroll a child in clinical trials for therapies, particularly if they are part of an underserved community. Trust in the medical system has long been a barrier to research participation among communities of color.[21] Despite national efforts led by the NIH and the FDA, research participation remains low for underrepresented groups designated by race, ethnicity, gender, socioeconomic status, and age. In addition, medical provider bias

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[21] *Scharff, D. P., Mathews, K. J., Jackson, P., Hoffsuemmer, J., Martin, E., & Edwards, D. (2010). "More than Tuskegee: Understanding Mistrust about Research Participation." Journal of Health Care for the Poor and Underserved, 21(3), 879. Doi: 10.1353/hpu.0.0323.*

is a systemic problem that contributes to underserved patients not being asked to participate in research.

Lack of diversity in research participation can have serious consequences, including being unable to generalize study results to advance effective therapies, and preventing some populations from experiencing the benefits of research innovations. Deciding to have a child participate in an available trial is a complicated decision. Resources are available to offer helpful tools to support parental decision making, including:

- Courageous Parent Network www.courageousparentsnetwork.org/topics/clinical-trial
- From Molecules to Medicine: How Patients Can Share Their Voices Throughout The Drug Development Process www.globalgenes.happyfox.com/kb/article/31-from-molecules-to-medicine-how-patients-can-share-their-voices-throughout-the-drug-development-process/
- Informed Consent: Important for Treatment Decisions and Advancing Research https://globalgenes.org/wp-content/uploads/2019/01/GG_toolkit_informed-consent_web-hyperlinked.pdf

Expanded Access/Compassionate Use

Patients with serious or life-threatening conditions for which there is no FDA-approved treatment but there is an investigational drug in development or under review for FDA approval may gain access from the manufacturer through the FDA Expanded Access process. There are, however, risks with these programs that caregivers and the child's doctor will need to carefully consider prior to participating. https://www.fda.gov/news-events/expanded-access/expanded-access-information-patients

Palliative Care

Pediatric palliative care is another option for care that focuses on the overall needs of a family and child with a rare and/or serious illness. Often, this is facilitated through a team of physicians, nurses, social workers, or other specialists. The team works along with the child's other care providers. Reducing the child's pain and suffering, as well as improving quality of life is the main focus of the palliative care team, as they provide relief and support to the entire family. Palliative care is based on the needs of the patient, not on the patient's prognosis. It is appropriate at any age and stage in a serious illness, and it can be provided along with curative treatment. www.ninr.nih.gov/sites/files/docs/palliative-care-brochure.pdf and www.globalgenes.org/wp-content/uploads/2014/11/Toolkit_12_Web.pdf

Pediatric Hospice Programs

Every caregiver's hope is for their child to get well. At the same time, it is important to consider the child's overall well-being when dealing with a life-limiting prognosis. Hospice care is focused on the child's quality of life while also supporting the family. There is an important emphasis of hospice care known as palliative care. While the majority of hospice patients are elderly, the focus on providing palliative care to children is growing. Hospice care can be provided in the home, in a facility, or in a hospital setting. In palliative care, controlling the child's symptoms, including pain, is a priority. It is also essential to address the family's fears about end of life so it does not interfere with caring for the child. Working with hospice provides support with how to talk to the child about what is happening and how to manage emotions such as anger and grief. Typically, hospice support for the family continues even after the patient passes away.

A key difference between hospice care services for children and for adults is that adults who are officially receiving hospice care must have a prognosis of fewer than six months and must usually stop curative treatment for the underlying disease. However, there is no expectation that hospice services are only available when a child approaches end of life. For some, the only difference between palliative and hospice care services is financial. Choosing to use a hospice benefit through insurance may designate that the majority or all fees for equipment, medications, and other expenses are billed through hospice versus traditional insurance. In a pediatric setting, this is often one of the most significant changes noted by caregivers when they move a child from palliative to hospice care. The services offered vary significantly from place-to-place across the country.

Some helpful resources are the National Hospice and Palliative Care Organization www.nhpco.org and Children's Hospice International www.chionline.org.

Every caregiver's hope is for their child to get well.

Mental Health Care

Access to mental health care for children can be challenging. Having a rare and/or serious illness can amplify mental health care needs. Behavioral issues, emotional exhaustion, anxiety, stress, and suicidal thoughts have all been recognized in the rare and/or serious disease population, with nearly one in five children having a mental, emotional, or behavioral disorder.[22]

It can be difficult to find pediatric mental health providers close to home, and families often travel long distances or have to endure long waiting lists to get care. Insurance coverage and costs add to the challenge of getting mental health care for a child. Patients and caregiver mental health concerns are sometimes not taken seriously or can be misdiagnosed. It is important that caregivers discuss mental well-being concerns with the child's pediatrician or specialist and advocate for additional support when needed. Caregivers know their child best and need to trust their judgment. They must be diligent and not lose hope. These care challenges may feel overwhelming but trying to maintain hope and diligence can help, along with the available tools and supports.

The National Alliance on Mental Illness (NAMI) is a leading voice on mental health. As a grassroots mental health organization aligned with over 600 local affiliates, NAMI works to educate, advocate, listen, and lead to improve the lives of Americans with mental illness and their loved ones. www.nami.org

IMPACT OF COVID-19

The COVID-19 pandemic has greatly impacted families with serious health conditions. It has affected the physical and mental well-being of caregivers and their families. Caregivers and patients already living with the ongoing stress and complexity of a rare and/or serious illness and balancing work while providing care have experienced months of lockdowns and social distancing. This has led to increased feelings of isolation, depression, and fear in families. Families have experienced the cancellation of medical appointments or have voluntarily held off in-person care. The pandemic has also disproportionately impacted low-income and diverse populations and raised systemic issues to the forefront of national dialogue.

Studies from the National Organization for Rare Disease (NORD) report disruptions for rare and/or serious illness patients, including increased prices and difficulties and delays accessing medication, medical supplies and devices, and personal protective equipment (PPE). Job loss, loss of income, and lost health insurance have all added to an increase in emotional and financial challenges.[23]

In addition, Global Genes found through their Continuity of Care RARE Patient Impact Grant Program[24] that the overwhelming portion of the funds given (82 percent) provided assistance with basic living expenses, caregiver expenses, medical transportation, and personal protective equipment. Grant recipients faced many challenges prior to the outbreak of COVID-19, but the pandemic intensified them by deepening the isolation rare disease patients and their families experience. The small grants addressed immediate needs of the recipients during the pandemic, and it let them know that there were sources of support out there for them. During the pandemic itself, the program, which sometimes granted funds for technology to connect with doctors, also opened access to the multiple organizations available online to support rare and/or serious illness families.

The COVID-19 pandemic has greatly impacted families with serious health conditions. It has affected the physical and mental well-being of caregivers and their families.

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[22] *O'Connell, M. E., Boat, T., & Warner, K. E. (2009). Preventing Mental, Emotional, and Behavioral Disorders among Young People: Progress and Possibilities (Vol. 7). National Academies Press. www.pubmed.ncbi.nlm.nih.gov/20662125.*

[23] *NORD Rare Insights. (2020, August). COVID-19 Community Follow-up Survey Report. Retrieved from http://www.rarediseases.org/wp-content/uploads/2020/08/NRD-2061-RareInsights-CV19-Report-2_FNL.pdf*

[24] *Chupka, R. (2021, May). Continuity of Care RARE Patient Impact Grant Program Report. Retrieved from https://globalgenes.happyfox.com/kb/article/273-continuity-of-care-rare-patient-impact-grant-program-report/*

Telemedicine

Use of telemedicine has dramatically increased during the COVID-19 pandemic and generally has been positively viewed as an alternative to in-person appointments. In part due to these virtual visits, recognition of the caregiver as an integrated member of the healthcare team has improved during the pandemic. Telemedicine also allows some clinical trials to continue despite the pandemic. This has provided families with feelings of safety and convenience.

Clinical Trials During the COVID-19 Pandemic

Clinical trials have experienced significant impacts from the COVID-19 pandemic. Patient visits to trial sites have been cancelled or delayed, some health care providers have moved to care for COVID-19 patients, and some research laboratories have closed or have been re-directed to work on the pandemic. Despite these interruptions, many trials have continued as a priority at the FDA during the pandemic, either through telemedicine or other approaches.

Use of telemedicine has dramatically increased during the COVID-19 pandemic and generally has been positively viewed as an alternative to in-person appointments.

HELPFUL WEBSITES

Agency of Healthcare Research and Quality
www.ahrq.gov/ncepcr/care/coordination.html

All of Us Research Program
www.allofus.nih.gov

Children's Hospital of Philadelphia
www.chop.edu/centers-programs/leukodystrophy-center

Clinical Trials
www.clinicaltrials.gov

Courageous Parent Network
www.courageousparentsnetwork.org/topics/clinical-trial

The Genetic and Rare Diseases Information Center
www.rarediseases.info.nih.gov/guides/pages/96/patients-families-and-friends

FDA Expanded Access
www.fda.gov/news-events/expanded-access/expanded-access-information-patients

FDA Guidance on Conduct of Medical Products during COVID-19 Public Health Emergency
www.fda.gov/regulatory-information/search-fda-guidance-documents/fda-guidance-conduct-clinical-trials-medical-products-during-covid-19-public-health-emergency

FDA Office of Orphan Products Development
https://www.fda.gov/industry/developing-products-rare-diseases-conditions

Global Genes COVID-19 Resources
www.globalgenes.org/coronavirus-covid-19-resources/

JumpStart at the Orphan Disease Center
www.orphandiseasecenter.med.upenn.edu

National Alliance on Mental Illness
www.nami.org

National Association for Home Care & Hospice
www.nahc.org

National Institute of Nursing Research
www.ninr.nih.gov/sites/files/docs/palliative-care-brochure.pdf

National Hospice and Palliative Care Organization
www.nhpco.org

Rare Diseases Clinical Research Network
www.rarediseasesnetwork.org

Rare Diseases Registry Program
www.ncats.nih.gov/radar

Rare Disease Therapy Development and Access Remain Top FDA Priorities During COVID-19
www.fda.gov/news-events/fda-voices/rare-disease-therapy-development-and-access-remain-top-fda-priorities-during-covid-19

The Joint Commission
www.jointcommission.org

ECONOMIC IMPACT OF CAREGIVING FOR A CHILD WITH A RARE AND/OR SERIOUS ILLNESS

Caregiving for serious illnesses and medical conditions often impact family finances.

Rare Disease Caregiving in America[25] found that over half of the responding caregivers had exhausted their personal savings due to costs associated with the care of their child, three in four caregivers worried about their ability to pay for care, and nearly all caregivers had experienced financial hardship.

Beyond the costs of medical care, the impact that rare and/or serious illnesses have on the income of the family is significant. Nearly all participating caregivers in *Rare Disease Caregiving in America* reported that they had to take some time off from work for their caregiving responsibilities. It is not uncommon for caregivers to make career changes to be able to manage the medical appointments, complex medical needs, and daily care necessary for a child. A considerable number of caregivers of rare and/or serious disease children reported that they have also cut back on their work hours, taken a leave of absence, turned down a promotion, or left the workforce altogether.[26] These options come with high costs that can financially devastate a family.

In the recently released *The National Economic Burden of Rare Disease Study*, it was found that the overall cost of rare disease in 2019 was nearly a trillion dollars, with the indirect costs (such as forced retirement, absenteeism, and reduction in community involvement) and nonmedical costs (such as paid daily care, transportation, education and healthcare not covered by insurance) that are absorbed directly by families accounting for 60 percent of that cost.[27]

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[25] *National Alliance for Caregiving. (2018). Rare Disease Caregiving in America. Retrieved from https://www.caregiving.org/wp-content/uploads/2020/05/NAC-RareDiseaseReport_February-2018_WEB.pdf.*

[26] *Ibid.*

[27] *EveryLife Foundation for Rare Diseases. (2021). The National Economic Burden of Rare Disease Study. https://everylifefoundation.org/wpcontent/uploads/2021/02/The_National_Economic_Burden_of_Rare_Disease_Study_Summary_Report_February_2021.pdf*

The cost of care and treatment for rare and/or serious illnesses can easily be staggering. There are many public and private programs and resources that may be accessible to assist families with the financial burden of caring for a child with a rare and/or serious illness. However, the paperwork and phone calls to navigate these resources can be time consuming and seem like a full-time job in itself.

MEDICAL INSURANCE

Insurance coverage is important to families experiencing rare and/or serious illnesses. Understanding these insurance benefits is also very important. Patients and caregivers living with rare and/or serious illnesses tend to be empowered, knowledgeable, and engaged by necessity. The reality is that the insurance company may not be familiar with the child's rare disease or its treatment. Caregivers and doctors may need to educate them on what the child needs. When needed, doctors, nurses, social workers, and navigators can often help navigate exclusions and exceptions.

Learning to navigate insurance may seem like learning a whole new language, but many resources exist to help. Always carefully review insurance plans before signing up for coverage. Resources that may provide guidance include:

- Family or friends with chronic conditions
- An employee's human resources department
- The insurer (the child may be assigned a case manager)
- Nurses, social workers, and case managers from hospital centers
- Patient advocacy organizations that provide support

Numerous co-pay and coverage assistance programs offered by pharmaceutical manufacturers and foundations exist for many conditions to help offset patient out-of-pocket costs.

Premium and Co-Pay Assistance

There are numerous premium, co-pay, and coverage assistance programs available to help offset patient out-of-pocket costs for families. Funding for these programs often comes from pharmaceutical manufacturers and foundations. Not all diseases are covered, and some funds open and then close during the year when funding is exhausted, so it is necessary to review each to see if the child's disease is supported by a program.

PREMIUM, CO-PAY AND COVERAGE ASSISTANCE PROGRAMS

The Assistance Fund www.tafcares.org	The Assistance Fund provides financial assistance for co-payments, coinsurance, deductibles, and other health-related expenses.
CancerCare www.cancercare.org/copayfoundation	CancerCare helps people with cancer overcome financial access and treatment barriers by assisting them with co-payments for their prescribed treatments.
Global Genes Financial Advocacy in RARE Educational Resources https://resource-hub.globalgenes.org/kb/section/22/	Global Genes provides educational resources focused on financial advocacy such as: • Financial Advocacy in RARE: Navigating the U.S. Health System for Young Adults Guide • Financial Advocacy Landscape Assessment • Navigating Health Insurance • Financial Advocacy RARE Impact Grant Program
Good Days www.mygooddays.org	Good Days provides resources for life-saving and life-extending treatments to people in need of access to care.
HealthWell Foundation www.healthwellfoundation.org	The HealthWell Foundation assists with co-pays, premiums, deductibles, and out-of-pocket expenses.
The National Organization for Rare Disorders www.rarediseases.org	NORD provides financial assistance for medications, insurance premiums and co-pays, diagnostic testing, and travel for clinical trials or consultation with disease specialists.
Patient Access Network Foundation www.panfoundation.org	The foundation helps the underinsured cover out-of-pocket costs associated with medications and treatments for specific diseases.
Patient Advocate Foundation www.patientadvocate.org	The foundation provides case management assistance for the uninsured or underinsured with life-threatening or debilitating illnesses. Includes access to care, co-pay assistance, social security disability applications and insurance appeals.
Patient Services, Inc. www.patientservicesinc.org	Patient Services, Inc. offers assistance programs for various conditions. They include help with co-payments, premiums, ancillary services, infusion, nursing services, and travel.

GOVERNMENT SUPPORT

There are state and federal programs that can assist in caring for an ill child such as Medicaid, Medicaid Waivers, Children's Health Insurance Programs, or other supplements or support. The Benefit Finder can help identify what benefits the child many qualify for. www.benefits.gov

Supplemental Security Income

Supplemental Security Income (SSI) payments for children with disabilities are monthly supplements to people with low income and limited resources. Children younger than 18 may qualify if they have a medical condition or combination of conditions that meet Social Security's definition of disability for children and if their income and resources fall within the eligibility limits. The SSI payments differ from state to state. Local and state Social Security Offices can provide more information on how to apply.

Social Security Disability Insurance

Social Security Disability Insurance (SSDI) benefits are for adults who have been disabled since childhood. This benefit is paid based on a parent/guardian's Social Security earnings record. To be eligible, a parent or guardian must be receiving Social Security retirement or disability benefits or have died but worked enough to qualify for Social Security. These SSDI benefits continue if the individual remains disabled, although marriage of the disabled adult may affect eligibility for this benefit. www.ssa.gov/disability

Medicaid Waiver Programs

Medicaid waivers are state-run programs that use federal and state funds to pay for health care for people with certain health conditions. The process for a Medicaid waiver will vary depending on the specific waiver and the state of residence. Most states have some type of Medicaid waiver program and there are many waiver types. www.medicaid.gov and www.kidswaivers.org

The Katie Beckett Program

Katie Beckett is an important program for children with a rare disease or serious illness. The Katie Beckett Program allows Medicaid to cover medical services for children in the home, regardless of the parents'/guardian's income, in cases where home-based treatment will cost less than or the same as treatment in a hospital. This may also be called a Katie Beckett Waiver or TEFRA Waiver.

Some states will pay for qualified caregivers to serve as home care providers to their own children, but they must be trained and employed through a home health care agency. While most parents or guardians are not RNs or LPNs, this is something to keep in mind. www.cga.ct.gov/2003/rpt/2003-R-0040.htm

The Tax Equity and Fiscal Responsibility Act

The Tax Equity and Fiscal Responsibility Act (TEFRA) is a 1982 act that expanded Katie Beckett. State criteria varies, so it is important to research the process by state. After the age of 19, a child with disabilities can transition to Medicaid Home and Home and Community-Based Services Waivers.

Children's Health Insurance Program

Children's Health Insurance Program (CHIP) is an insurance program for low-cost health coverage for children in families that earn too much money to qualify for Medicaid but not enough to buy private insurance. In some states, CHIP covers pregnant women. Each state offers CHIP coverage and works closely with its state Medicaid program. Families can apply any time. Coverage begins immediately, any time of year, for families that qualify. www.healthcare.gov/medicaid-chip/childrens-health-insurance-program/

Title V Funding and Catastrophic Illness Relief Funds

Several states use federal funding from the Title V Maternal and Child Health Services Block Grant Program to support the promotion and improvement of the health and well-being of children with special needs and their families. Catastrophic illness relief funds are one way this funding is utilized. The purpose of catastrophic illness or health relief funds is to help ease the burden of out-of-pocket health care expenses that are not covered by insurance. The financial support to caregivers may come from state or local funds. Catastrophic illness funds vary from state to state. Two examples of state websites for Catastrophic Illness Relief Programs are listed below.

Title V funding www.mchb.hrsa.gov/maternal-child-health-initiatives/title-v-maternal-and-child-health-services-block-grant-program

Catastrophic Illness in Children Relief Fund in New Jersey:

A financial assistance program for New Jersey families with catastrophic medical funds relating to a child's illness or condition. www.state.nj.us/humanservices/cicrf/home/

Catastrophic Illness in Children Relief Fund in Massachusetts:

Financial relief for Massachusetts families caring for children under the age of 22 with large medical and related expenses. www.mass.gov/catastrophic-illness-in-children-relief-fund-cicrf

Other Resources

Even for families with medical insurance and other financial support, a serious medical issue or disability can result in high medical bills. A loss of income, or miscellaneous expenses, such as food, travel, or other medical needs add up quickly. There are organizations available to assist with managing finances or providing support.

Medication Assistance

Healing the Children www.htcne.org	Healing the Children provides support for children with burn injuries, cleft palates, and other deformities whose families do not have access to or cannot afford treatment.
National Alliance on Mental Illness www.nami.org	NAMI has a list of free or low-cost mental health medications provided by pharmaceutical companies.
Needy Meds www.needymeds.org	Needy Meds provides information about government programs, low-cost or free medical and dental clinics, and prescription assistance and disease-specific financial aid programs.
RxAssist www.rxassist.org	RxAssist offers a database of patient assistance programs, as well as practical tools, news, and articles for health care professionals and patients.

Travel and Lodging Assistance

Often, a family will need to travel to receive specialist care or participate in research for the child's rare and/or serious illness. The cost of travel and lodging can be significant but numerous organizations provide support to help offset these expenses.

The following provides a snapshot of travel and lodging support. The NIH Genetic and Rare Disease Information Center maintains an updated list of resources to help. www.rarediseases.info.nih.gov/guides/pages/118/help-with-travel-costs

Air Charity Network www.aircharitynetwork.org	Provides access to member organizations that offer free air transportation to specialized health care facilities or distant destinations needed during family, community, or national crisis.
American Cancer Society www.cancer.org/treatment/support-programs-and-services/road-to-recovery.html	The American Cancer Society's Road to Recovery Program provides free transportation for cancer patients receiving treatment at nearby hospitals or medical facilities.
Children's Flight of Hope www.childrensflightofhope.org	Provides free air transportation to and from medical facilities in the eastern United States for seriously ill and injured children.
Children's Inn at NIH www.childrensinn.org	A nonprofit residence dedicated to serving the families of children involved in pediatric research at the NIH Clinical Center in Bethesda, MD. First-time reservations must be made by the medical team.
Hospitality Homes www.hosp.org	Hospitality Homes provides temporary housing in volunteer host homes and other donated accommodations for families and friends of patients seeking care at Boston-area medical centers.
Mercy Medical Angels www.mercymedical.org	Mercy Medical Angels provides both air and ground transportation support for patients in need of specialized medical care outside their local region.
Miracle Flights www.miracleflights.org	Miracle Flights helps children struggling with severe illnesses fly to specialized medical treatment centers anywhere in the United States.
Patient AirLift Services www.palservices.org	PALS arranges free air transportation based on need to individuals requiring medical care and for other humanitarian purposes.
National Cancer Institute https://supportorgs.cancer.gov/home.aspx?lang=1	The National Cancer Institute provides a list of more than 100 organizations nationwide that provide support for people with cancer and their families.
Ronald McDonald House Charities www.rmhc.org	Ronald McDonald House Charities is a nonprofit organization that provides nearby lodging for families with hospitalized children under 21 years of age. Depending on the affiliated hospital, accommodations may include targeted recreational activities and support services for child and family enrichment, sibling support services, and other programs.
Wings of Hope www.wingsofhope.ngo	The Wings of Hope Medical Relief & Air Transport (MAT) Program provides medical air transport services, free of charge, to individuals who need specialized medical care unavailable to them locally. Using aircraft specially outfitted with stretchers to accommodate fragile and non-ambulatory patients, they fly individuals to hospitals and treatment centers in 26 states within a 600-mile radius of their St. Louis, MO headquarters.

The Helping Hands Program

The Hemophilia Federation of America is a national nonprofit that assists the bleeding disorders community through support and advocacy. Their Helping Hands Program specifically assists with financial hardships that may occur due to medical expenses.

Some disease-specific patient advocacy organizations have charitable programs that can assist with travel and lodging, gym memberships, tutoring and emergency assistance during a crisis. An example of this is the Hemophilia Federation of America's Helping Hands Program. Be sure to contact the rare disease specific organization to determine if they have these types of support services.

Meal Trains

Meal Trains can be a helpful way to make sure a family is eating during difficult times, or when time or energy to cook might be low. This can include a period of frequent doctor visits, periods of surgery, or if caregivers are helping their child during a difficult pain flare-up. In a meal train, a small group of close family and friends coordinate a calendar to determine who will provide a meal on which night, based on when help is needed by the family. These calendars can be informal and entirely based on the group's preference, organized through text or email, or through websites that help with the coordination of meal trains:

- Care Calendar (www.carecalendar.org)
- Meal Train (www.mealtrain.com)
- Take Them a Meal (www.takethemameal.com)

Online Fundraising

Also known as crowdfunding, this is a method for consideration when facing financial hardship because of a rare disease. Some patients find that friends, family, coworkers, and community members are eager to contribute financially if they are aware of the difficult situation. Money can be raised for different purposes, including emergency support, equipment, or supportive needs like wheelchair ramps for homes or to fund research. Sites such as GoFundMe, Fundly, 99Pledges, and even Facebook can be used to crowdsource funding. Very large gifts could be taxable, but the donor typically pays the tax. However, it's recommended that caregivers consult with an accountant or tax attorney with questions.

HELPFUL WEBSITES

Air Charity Network www.aircharitynetwork.org

American Cancer Society www.cancer.org

The Assistance Fund www.tafcares.org

Benefits www.benefits.gov

CancerCare www.cancercare.org/copayfoundation

Care Calendar www.carecalendar.org

Catastrophic Illness in Children Relief Fund in Massachusetts www.mass.gov/catastrophic-illness-in-children-relief-fund-cicrf

Catastrophic Illness in Children Relief Fund in New Jersey www.state.nj.us/humanservices/cicrf/home/

Children's Flight of Hope www.childrensflightofhope.org

Children's Health Insurance Program www.healthcare.gov/medicaid-chip/childrens-health-insurance-program

Children's Inn at NIH www.childrensinn.org

The National Economic Burden of Rare Disease Study http://www.everylifefoundation.org/wp-content/uploads/2021/02/The_National_Economic_Burden_of_Rare_Disease_Study_Summary_Report_February_2021.pdf?eType=EmailBlastContent&eId=1d1fcfbd-10fc-4553-bf38-dfd73c7e7cd8

Global Genes www.globalgenes.org

Good Days www.mygooddays.org

Healing the Children www.htcne.org

HealthWell Foundation www.healthwellfoundation.org

Hemophilia Federation of America www.hemophiliafed.org

Hospitality Homes www.hosp.org

Kids Waiver www.kidswaivers.org

Meal Train www.mealtrain.com

Medicaid www.medicaid.gov

Mercy Medical Angels www.mercymedical.org

Miracle Flights www.miracleflights.org

National Alliance on Mental Illness www.nami.org

National Cancer Institute https://supportorgs.cancer.gov/home.aspx?lang=1

The National Organization for Rare Disorders (NORD) www.rarediseases.org

Needy Meds www.needymeds.org

NIH Genetic and Rare Disease Information Center www.rarediseases.info.nih.gov/guides/pages/118/help-with-travel-costs

Patient Airlift Services www.palservices.org

Patient Access Network Foundation www.panfoundation.org

Patient Advocate Foundation www.patientadvocate.org

Patient Services, Inc. www.patientservicesinc.org

Ronald McDonald House Charities www.rmhc.org

RxAssist www.rxassist.org

Social Security www.ssa.gov/disability

Take Them a Meal www.takethemameal.com

Title V funding www.mchb.hrsa.gov/maternal-child-health-initiatives/title-v-maternal-and-child-health-services-block-grant-program

Wings of Hope www.wingsofhope.ngo

EDUCATION AND RECREATION

Quality-of-life (QOL) is a multidimensional concept evaluating the impact of health status on physical, mental, emotional, and social functioning.[28]

Studies have reported that rare and/or serious disease populations generally have an overall lower quality of life compared to populations not affected by rare and/or serious diseases. Therefore, it is important to identify elements in a child's life that may have a positive effect on overall quality of life. Caregivers who are less worried about their child's quality of life may find more time to work on their own quality of life.

EDUCATION

Collaborating with the child's school to make accommodations for a good learning experience can be stressful and cause worry over ensuring the child has what they need. It is important to know that teachers, administration, and support staff within a school system have the child's best interests in mind and want them to succeed. Finding appropriate accommodations for a child may have challenges, but it is important to be open to possible solutions and work collaboratively with the school as a united team.

It is appropriate, and often necessary, to ask for accommodations. Several laws support students and their caregivers in this process. This includes the Individuals with Disabilities Act (IDEA), a federal special law for children with disabilities. Another important law is Section 504 of the Rehabilitation Act of 1973, a provision of federal civil rights law that aids to prevent discrimination against people with disabilities. The following are specific aspects of these laws that may be useful information when working to identify appropriate accommodations:

The Early Intervention Program for Infants and Toddlers with Disabilities is a federal grant program that assists states in running programs to provide services to infants and toddlers (birth to 3 years old) with disabilities. Once eligibility is determined,

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[28] *Related Quality of Life & Well-being. (n.d.). Retrieved from https://www.healthypeople.gov/2020/topics-objectives/topic/health-related-quality-of-life-well-being.*

services may include speech and feeding services, physical and/or occupational therapy, social work services, and many others. Often, early intervention services can be provided at home or at any early center/daycare in which the child is enrolled. For more information on specific services by state and statewide contacts for this program, visit www.ideainfanttoddler.org/state-contacts.php or www.zerotothree.org/.

Individualized Education Program (IEP) is a plan for children with medical or developmental disabilities, or other health impairments who require adaptations of modifications to their school day and environment in order to access the school building and curriculum. The plan provides a customized educational plan, including related services to meet a child's unique needs. The IEP creates an opportunity for school personnel, parents/guardians, and students to work together to improve the benefit and experience for the child. There is no cost to families to request or implement an IEP for their child.

A **Section 504 Plan** is a plan for how the school will provide support and remove barriers for students with disabilities. This can include services and changes to the learning environment to enable students to learn alongside their peers. It is for children with physical, emotional, or learning disabilities. There is no cost to families to request or implement a 504 plan.

Section 504 of the Rehabilitation Act of 1973 provides rights to students with disabilities. This federal regulation states that all students have the right to a "free, appropriate education." regardless of the type (mental or physical) or severity of the disability. A student must have a diagnosis that limits a major life function, but this alone may not make them eligible for school accommodations. A physician can help, but an evaluation by the school will also be necessary.

When a child is considered eligible by the school, barriers to their learning are removed by making accommodations for the student. This might include personalized assignments, alternative grading, or a classroom change. Schools need to provide (in writing) information about the child's assessment and what changes will be made. Parents or guardians have the right to review their child's records and request an impartial hearing if they disagree. Regular reviews and updates to the 504 Plan should be made. If a parent/guardian feels their child's rights are not being met, they may submit a complaint to the Office for Civil Rights enforcement office. More information about each state's office is available at www2.ed.gov/policy/rights/reg/ocr/edlite-34cfr104.html.

An **Individualized Health Care Plan (IHCP)** is a regularly updated and annually reviewed plan developed by the school nurse in collaboration with caregivers, medical providers, and others. It details additional medical care a child receives while at school. This might include receiving medication or providing additional nursing care, such as a one-to-one nurse, during the school day. www.nasn.org/advocacy/professional-practice-documents/position-statements/ps-ihps

Discipline. When necessary, students are disciplined at school even if they have learning differences. However, when reprimanding a student with special needs, it is important that the child's individual needs and accommodations are considered. A child with an IEP or Section 504 Plan has additional legal rights. Schools have their own code of conduct to keep students safe and ensure discipline is fair and appropriate. School discipline may include contacting a parent/guardian, counseling, extra schoolwork, or exclusion. The school needs to ensure the chosen method of discipline does not interfere with the services a student is receiving. Also, the child's learning differences could be a cause for the behavior and should be considered when trying to understand the child's actions. Parents/guardians can appeal the discipline imposed on their child if they believe it is unfair or if there is a method better suited to the child's needs.

Neglect. If a parent or guardian feels the school is not following the child's IEP or 504 Plan, they should address it to ensure the child has the best school experience possible. It is best to begin by speaking with the teacher(s) about concerns in a nonconfrontational manner. The teacher and parents/guardian can review the 504 or IEP together and identify where they think there could be improvements made that will enhance the child's learning experience. If need be, parents or guardians should follow-up with school administrators to ensure the plan is being followed. It can be helpful to document when the plan was not followed and any conversations with school staff. With this information, parents/guardians can request an IEP team meeting to determine if the plan is being ignored or if they feel their concerns are not being addressed. If all else fails, mediators can handle disputes or parents/guardians can file a complaint with the state. At times, a lawyer may be helpful in advising on and dealing with these issues.

Self-Esteem. As children with rare and/or serious illnesses grow older, they may begin to realize they are different from other children. Special accommodations at school, frequent doctor visits, and the need for medications, medical equipment, or nursing care may cause self-consciousness and a loss of self-esteem. Each child will be different, and some may never experience these issues, but it is important to remember that this can occur regardless of the illness the child is battling. It is important to praise the child's accomplishments and any positive attributes. There may be a time when caregivers will need to sit down with their child to discuss their differences and help them see their differences do not make them "lesser" than other students.

The guardian-child relationship is also important in matters of self-esteem. One study found that children with special needs who are cared for by their parents become more passive in decision-making processes and when facing challenges than children in traditional parent-child relationships.[29] Teaching children to speak up when they are underestimated by others is important. Parental confidence in caregiving skills and in the child can naturally help the child become more confident. The study concluded that allowing children to grow through the experience with the parent and keeping line of communication open makes both the caregiver and child feel supported; the child will likely begin taking a more active role in their needs. Having more control over their lives and choices will go a long way toward building self-esteem.

Supporting siblings. It's important to consider the experience of children who may share a classroom or school building with their ill sibling. Whenever possible, teachers should avoid taking children out of class to tend to their ill sibling, which can happen when a child is experiencing a distressing episode. Siblings will often know best in how to help or soothe their ill sibling, but their time in the classroom/play time must also be respected and protected. Instead, teachers should work preemptively with parents to ensure a proper care plan is in place for the school day, ensuring that both children with rare and/or serious illnesses—and their siblings—can thrive in the classroom.

CHILD RECREATION

The nature of rare and/or serious illnesses in children often presents challenges when it comes to participating in organized sports and other activities important for social and emotional growth. Several organizations have developed activities that promote both inclusive and adaptive sports and other activities for individuals with disabilities. Studies have proven the physical and psychosocial benefits of sports and recreational activities among children with intellectual and physical disabilities.[30,31]

Special Olympics

Special Olympics provides year-round sports training and athletic competition in a variety of Olympic-type sports for children and adults with intellectual and developmental disabilities. Their Young Athletes Program promotes early childhood development and inclusivity by introducing basic sport skills like running, kicking and throwing to children with and without intellectual disabilities, beginning at age 2. Special Olympics Unified Champion Schools brings together students with and without intellectual disabilities through sports, inclusive leadership, and whole-school engagement activities to create school climates of increased acceptance and inclusion. www.specialolympics.org

Studies have proven the physical and psychosocial benefits of sports and recreational activities among children with intellectual and physical disabilities.[30,31]

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[29] Lee, Y., Park, H. & Recchia, S. (2015). "Embracing Each other and Growing Together: Redefining the Meaning of Caregiving a Child with Disabilities." Journal of Child & Family Studies, 24(12), 3662–75. Doi: 10.1007/s10826-015-0175-x.

[30] Johnson, C. C. (2009). "The Benefits of Physical Activity for Youth with Developmental Disabilities: A Systematic Review. American Journal of Health Promotion, 23(3), 157¬–67. Doi: 10.4278/ajhp.070930103.

[31] Shapiro, D. R. & Malone, L. A. (2016). "Quality of Life and Psychological Affect Related to Sport Participation in Children and Youth Athletes with Physical Disabilities: A Parent and Athlete Perspective." Disability and Health Journal, 9(3), 385–91. Doi: 10.1016/j.dhjo.2015.11.007.

Athletes Serving Athletes

Maryland-based Athletes serving Athletes (ASA) promotes recreational sports for individuals with rare and/or serious illnesses. The mission of ASA is to improve the quality of life for individuals with limited mobility by empowering them to train for and participate in community running events. This initiative seeks to create programs and events for athletes with disabilities to pair them with volunteer athletes who provide assistance in athletic events, such as crossing finish lines in races. These experiences increase confidence, self-esteem, dignity, and the quality of life for individuals with disabilities and build positive community awareness for individuals with disabilities. www.athletesservingathletes.org

> **These experiences increase confidence, self-esteem, dignity, and the quality of life for individuals with disabilities and build positive community awareness for individuals with disabilities.**

Summer Camps

In addition to traditional summer camp activities, camps for children with rare and/or serious illnesses usually have medical care and specially trained staff on site. Typically, camps also have support groups and skill building opportunities for children affected by serious illnesses, their siblings, and their caregivers. A sample of these camps include:

Double H Ranch
www.doublehranch.org

Double H Ranch provides specialized programs and year-round support for children and their families dealing with life-threatening illnesses.

Hole in the Wall Gang Camp
www.holeinthewallgang.org

Hole in the Wall Gang provides camps and retreats for children and families affected by serious illnesses. Their hospital outreach program brings camp-style activities to hospitalized patients in the Northeast United States. All programs are free.

Serious Fun Children's Network
www.seriousfun.org

Serious Fun Children's Network is an international family of camps and programs that provide life-changing experiences to children with serious medical conditions, always free of charge.

Victory Junction
www.victoryjunction.org

Victory Junction enriches the lives of children with serious illnesses by providing life-changing camping experiences that are exciting, fun, and empowering, at no cost to children or their families.

Huntington's Disease Youth Organization
https://en.hdyo.org/you/events

HDYO is a nonprofit that provides support and education for young people impacted by Huntington's Disease. They often provide youth summer camps where children can meet others with HD, learn about HD through workshops and share their experiences through fun camp activities like hiking, swimming and archery.

MDA Summer Camp
www.mda.org/summer-camp

The Muscular Dystrophy Association provides a free, overnight summer camp to children with neuromuscular diseases where they can experience regular camp activities such as swimming, horseback riding and zip lining.

Wish-Granting Organizations

Wish-granting organizations provide wishes (gifts) primarily to children who have a severe or life-threatening diagnosis. Often, the fulfilled wish is beyond the means of the family, whether financial or otherwise. In general, wishes are for travel, adventure, or to meet a famous person. These wishes are often transformative experiences not only for the child receiving the wish but also the community network who has helped fulfill the wish. Two wish-granting organizations include the Make-A-Wish Foundation and Tim Tebow Foundation: Night To Shine.

The Make-A-Wish Foundation

Founded in 1980 and likely the most well-known wish-granting organization, Make-A-Wish grants life-changing wishes for children diagnosed with a critical illness. Examples include adventures where the child plays a superhero or another role of their choice, a new pet, home modifications, or a family vacation. Parents and other caregivers familiar with the child and the impacts of their illness (including medical professionals providing care for a child) can refer a child through the Make-A-Wish website www.wish.org/refer-a-child

Night to Shine (Tim Tebow Foundation)

Night to Shine is an international, faith-based organization sponsored by the Tim Tebow Foundation to offer special-needs adolescents (14 years and older) a “prom night” experience with the support of local churches. The event is free for attendees and their families. To learn more about Night to Shine events, please visit www.timtebowfoundation.org/ministries/night-to-shine

These wishes are often transformative experiences not only for the child receiving the wish but also the community network who has helped fulfill the wish.

HELPFUL WEBSITES

Athletes Serving Athletes
www.athletesservingathletes.org

Double H Ranch
www.doublehranch.org

Hole in the Wall Gang Camp
www.holeinthewallgang.org

Huntington’s Disease Youth Organization
https://en.hdyo.org/you/events

Individuals with Disabilities Act
www.ideainfanttoddler.org/state-contacts.php

Make-A-Wish Foundation
www.wish.org/refer-a-child

MDA Summer Camp
www.mda.org/summer-camp

National Association of School Nurses
www.nasn.org

Nondiscrimination on the Basis of Handicap in Programs or Activities Receiving Federal Financial Assistance
www.ed.gov/policy/rights/reg/ocr/edlite-34cfr104.html

Serious Fun Children’s Network
www.seriousfun.org

Special Olympics
www.specialolympics.org

Tim Tebow Foundation: Night to Shine
www.timtebowfoundation.org/ministries/night-to-shine

U.S. Department of Education: Nondiscrimination on the Basis of Handicap in Programs or Wrights Law
www.wrightslaw.com

Victory Junction
www.victoryjunction.org

Zero to Three
www.zerotothree.org

ACHIEVING QUALITY-OF-LIFE AS A FAMILY MANAGING RARE AND/OR SERIOUS ILLNESS

Managing effective relationships within families can be challenging when caring for a child with a rare and/or serious illness. However, strong relationships with immediate and extended family can be a valuable resource when managing care.

Family members, including partners/spouses, can play important roles in providing care to the child and easing the burden of the primary caregiver. Studies have found that the stress of managing day-to-day family responsibilities while caring for a child with a rare and/or serious illness plays a role in the caregiver's psychological and physical health. Functioning well can greatly reduce stress and help with the effects of self-perception and social support. Family support may allow the caregiver to accomplish tasks outside of caregiving, such as employment. Family members can also provide emotional support.[32]

To successfully achieve family support, it may be useful to address and clearly define roles that family members will play in providing support and sharing the responsibility of care management for the child. These conversations may be facilitated through family therapy and/or a social worker.

MARRIAGE/PARTNERSHIPS

Many relationships suffer from periods of stress, but there are additional stressors for caregivers of a child with a serious illness or life-threatening medical condition. These may be due to financial issues, lack of free time, or fatigue due to caregiving responsibilities. If these issues are not addressed, they can add additional strain or even contribute to the end of a marriage or relationship. Communication is particularly important for these caregivers.

In our focus group discussions, a caregiver shared frustration toward their spouse's seemingly indifferent approach to the care of their hospitalized son, only to realize many years later that the spouse was actually trying to support the caregiver's own well-being in their actions. This misunderstanding was likely due to poor communication about their individual emotional experiences.

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[32] *Parminder, R., O'Donnell, M., Rosenbaum, P., Brehaut, J., Walter, S. D., Russell, D., Swinton, M., Zhu, B. & Wood Ellen. (2005). "The Health and Well-being of Caregivers of Children with Cerebral Palsy. Pediatrics, 115(6), 626–36. Doi: 10.1542/peds.2004-1689.*

Research suggests that parents may differ in the way they understand their child's disease, and that their lives might also be impacted differently.[33]

Patience and awareness that emotional experiences in caregiving can manifest in different ways will improve communication between spouse/partners. It is beneficial to set aside regular "couple time" and accept offers of help from friends or family to schedule "time off." Open communication with partner/spouses is very important; it may be helpful to meet with a counselor to improve this communication.

SIBLINGS/OTHER CHILDREN

Siblings of children with rare and/or serious illnesses are often exposed to unique experiences distinct from other children. This can have positive or negative impacts on their well-being.[34] While many siblings have described increased empathy, cooperation, and appreciation as a result of their experience, siblings of children with chronic illness are more likely to suffer from depression, anxiety, or other negative psychological outcomes.[35] Siblings may resent the amount of time and energy taken up by the sibling with a serious medical condition. They may be fearful of their and their family's future, or they may feel overwhelmed by the responsibilities and obligations they have to their sibling.[36] Moreover, siblings may feel they cannot express these feelings to their those who care for them given the caregiving burden of responsibility.

While caregivers may feel they should focus all of their energies on the child with a rare and/or serious illness, it is important to provide time and support to other children as well. The siblings are experiencing caregiving challenges along with the rest of the family. Caregivers can share information on a level they can understand. Age-appropriate books about the sibling's diagnosis may be helpful.

Courageous Parents Network (www.courageousparentsnetwork.org) provides learning modules to better understand a sibling's experience. To prevent resentment, it is important to create a balance among the children through chores and responsibilities. It is also important to have family activities that do not revolve around the needs of only one child. To prevent siblings from feeling lonely, work in periods of one-on-one time and encourage activities with their friends or look for sibling support groups, such as Sibshops.

Sibshops is a global program created to help the siblings of children with special needs. Sibshops create fun, recreational events where siblings will meet others like them, children who have experienced life as the sibling of a special needs child. The events provide games and fun, as well as an open environment to talk about the good (and not-so-good) parts of being a sibling of a child with special needs. The events also provide an opportunity for the kids to learn about the services and supports their special needs siblings receive. While Sibshops is not therapy, the events can be therapeutic for the siblings who attend. The Sibshops website has a list of events and information about how to create a Sibshop.
www.siblingsupport.org

While caregivers may feel they should focus all of their energies on the child with a rare and/or serious illness, it is important to provide time and support to other children as well.

The Sibling Leadership Network is a group with the purpose of providing siblings of those with disabilities with the resources, tools, and support they need in order to advocate alongside their brothers and/or sisters for issues that are important to them and their family. The Sibling Leadership Network helps siblings build a network in order to connect with the people and resources they need to become better advocates. The site itself provides policy news, research, and information on how to get involved.
www.siblingleadership.org

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[33] *Cardinali, P., Migliorini, L. & Rania, N. (2019). "The Caregiving Experiences of Fathers and Mothers of Children with Rare Diseases in Italy: Challenges and Social Support Perceptions." Frontiers in Psychology. Doi: 10.3389/fpsyg.2019.01780.*

[34] *Fleary, S. A., & Heffer, R. W. (2013). "Impact of Growing Up with a Chronically Ill Sibling on Well Siblings' Late Adolescent Functioning." ISRN Family Medicine, 2013.*

[35] *Sharpe, D., & Rossiter, L. (2002). "Siblings of Children with a Chronic Illness: A Meta-analysis." Journal of Pediatric Psychology, 27(8), 699–710. Doi: 10.1093/jpepsy/27.8.699.*

[36] *Ibid.*

EXTENDED FAMILY, FRIENDS, AND OTHER SOCIAL INTERACTIONS

In our focus group discussions, many caregivers felt as if they could not be honest about their situation with those outside of their immediate family. One caregiver described being fearful in public that their child was going to have a seizure; another described difficulty in activities such as using a public restroom with a child with a serious medical condition. The discomfort associated with being open about the nature of the family situation often results in feelings of isolation. Additionally, although many friends and extended family want to be helpful to their friends who are caregivers, they may not know how to effectively support caregivers.

Ways to support caregivers include:

- **Reaching out**: Even if you don't know what to say, ask what you can do to help.
- **Being respectful**: Check with the family before visiting and postpone the visit if you are ill.
- **Offering support**: Invite caregivers to coffee or to go on a walk. Volunteer to pick up groceries or deliver a meal.
- **Coordinating support**: Organize volunteers to provide meals, transportation, or help with household chores.

COMMUNITY SUPPORT

The community surrounding a caregiver for a child with a rare and/or serious illness is crucial to addressing caregiver needs and supporting overall quality of life in the family. Community resources can be local or remote and can offer support through both informational and logistical guidance as well as emotional support.

Although it may seem initially challenging, consider asking trustworthy friends and family members for help, being specific on how they can best provide support. If possible, explain to them the isolation, exhaustion, stress, grief, or other emotions being experienced. Being open, honest, and clear about caregiver needs may provide guidance and directions to friends and family who feel otherwise helpless. Depending on the nature of the relationship, friends and family may serve as an effective support system for both emotional needs and for activities to ease the burden of everyday tasks for the caregiver. Ultimately, accepting this support can improve a caregiver's well-being, which helps everyone.

Both advocacy organizations as well as disease-specific organizations (see Getting Accurate Information on a Child's Disease in this guidebook) may provide resources for community support or respite for caregivers and their immediate family. The following list describes programs and organizations that can be beneficial to families affected by a child with a rare and/or serious illness.

Talking to friends and family:

- **Acknowledge that the child's rare and/or serious illness has changed the family's life.**
- **Provide some very basic facts about the condition.**
- **Show gratitude for their caring, interest, and support.**
- **Share honestly with those who care about the caregiver and family.**
- **Take help that is offered! Family can be a wonderful support system.**

Although it may seem initially challenging, consider asking trustworthy friends and family members for help, being specific on how they can best provide support.

Organizations That Provide Supports and Services for Caregivers of Children with Rare and/or Serious Illnesses

CanCare www.cancare.org	CanCare provides emotional support, counseling, and information on financial resources to ensure that no one who is experiencing cancer goes through the experience alone.
Center for Parent Information and Resources www.parentcenterhub.org	Parent Training and Information Centers (PTI) and Community Parent Resource Centers (CPRC) are state-based centers that provide direct services for children and youth with disabilities, families, professionals, and other organizations that support them. The organization helps parents participate effectively in their children's development, partnering with professionals and policy makers to improve outcomes for all children with disabilities.
Courageous Parents Network www.courageousparentsnetwork.org	Created to provide support for those caring for children with serious illnesses, this organization delivers programs to families as well as resources to providers delivering family-centered care. The goal is that families have confidence in their role caring for the child. Information and tools that address caregiver psychological and emotional issues and enable informed decision-making in healthcare is provided through videos, podcasts, discussion forums and guides. Resources include working with the medical team, making difficult decisions, caring for the siblings, tending to marriage, coping with grief, and anticipating end of life.
Easterseals www.easterseals.com	Easterseals provides state programs including early intervention, inclusive childcare, medical rehabilitation, and autism services for young children and their families, mental health programs, assistive technology, camp and recreation, and caregiving support including respite.
Family Voices www.familyvoices.org	Family Voices is a national network of families and friends of children and youth with special health care needs and disabilities. The organization promotes partnership with families–including those of cultural, linguistic, and geographic diversity. They offer training and share best practices and resources on family engagement and advocacy.
Parent to Parent USA www.p2pusa.org/parents	The network of state-based programs offers parent-to-parent support as a core resource for families with children who have a special health care need, disability, or mental health concern. The organization matches and connects trained support parents to other caregivers dealing with the same or similar conditions to offer support via phone calls and email. Support parents are trained to provide a safe environment for the caregiver to share their experiences. They do not give any medical or professional counseling.

The goal is that families have confidence in their role caring for the child.

Rays for Rare

Rays for Rare is a local organization in Idaho that provides support to medically fragile children and their families through respite family support, advocates for pediatric palliative and hospice care resources for children, and helps with daily tasks such as housekeeping, meal service, and snow removal. www.raysforrare.org

VACATION AND TRAVEL

Children with a rare and/or serious illness and their families can (and need to) take vacations! Vacations can be a great way to manage emotional strain, even when there is grief. There are many ideas and options with different expense levels available. Families should not be afraid to take a vacation. It just takes research and planning.

Planning is key. Successful travel and outings with a child with a rare and/or serious illness needs even more preparation. By preparing ahead, trips can build wonderful memories for the entire family. Caregivers should not be shy about calling ahead to ask about travel needs, lodging, restroom and meal accommodations, and any other necessities. If flying, find out ahead of time what items can be packed by confirming the latest Transportation Security Administration (TSA) guidelines at www.tsa.gov/travel/special-procedures.

It is helpful to carry a doctor's letter outlining the child's condition, medications, and medical needs in case of an emergency, as well as phone numbers of the child's doctors and specialists. The child's doctor may be able to recommend physicians or local hospitals in the area in case of an emergency. Take insurance cards and review policies before the trip in case prior approval is necessary to visit an out-of-town doctor or emergency room.

Finally, it is important to recognize that the very best plans sometimes get interrupted by life with a rare and/or serious illness. Do not expect perfection. If the trip goes sideways from the plan, flexibility and adaptability will help families enjoy their time together.

Theme Parks with Special Needs Access

Many amusement and theme parks welcome families with disabilities and make special accommodations for them. Some are well-known institutions with specialized accessibility programs, others are smaller and family-owned.

Disney Parks
https://disneyparks.disney.go.com/blog/disney-parks-disability-access-service-card-fact-sheet/

Disney theme parks offer Disability Access Service Cards through Guest Relations without documentation from a doctor. Visitors will receive ride return times to avoid long wait times.

Morgan's Wonderland
www.morganswonderland.com

Morgan's Wonderland and Morgan's Inspiration Island Splash Park in San Antonio, TX comprises the only fully accessible theme park, featuring more than twenty-five elements, including rides, playgrounds, and other colorful attractions. Admission is free for children with special needs.

Sesame Place
www.sesameplace.com

Sesame Place, based on the children's television show, is a theme park with specialized accessibility programs in Philadelphia, PA. Team members receive special training on topics such as sensory awareness and motor skills to support visitors with special needs.

Six Flags
www.sixflags.com

Six Flags' Attraction Access Program is designed to accommodate guests with disabilities or certain other qualifying impairments so they may participate in the enjoyment of the parks. Each attraction at Six Flags has been evaluated for the criteria necessary for an individual to ride safely.

Universal Studios
www.universalstudioshollywood.com
www.universalorlando.com

Universal Studios provides an Attractions Assistance Pass to visitors requesting such at Guest Services. This pass provides ride return times and allows your child's stroller the same access as a wheelchair.

HELPFUL WEBSITES

CanCare www.cancare.org

Center for Parent Information and Resources
www.parentcenterhub.org

Courageous Parents Network
www.courageousparentsnetwork.org

Easterseals
www.easterseals.com

Family Voices
www.familyvoices.org

Global Genes Support for the Rare Disease Family
Caring for Siblings of Kids with Rare Diseases
https://resource-hub.globalgenes.org/kb/article/180-track-1-support-for-the-rare-disease-family-caring-for-siblings-of-kids-with-rare-diseases/

Morgan's Wonderland
www.morganswonderland.com

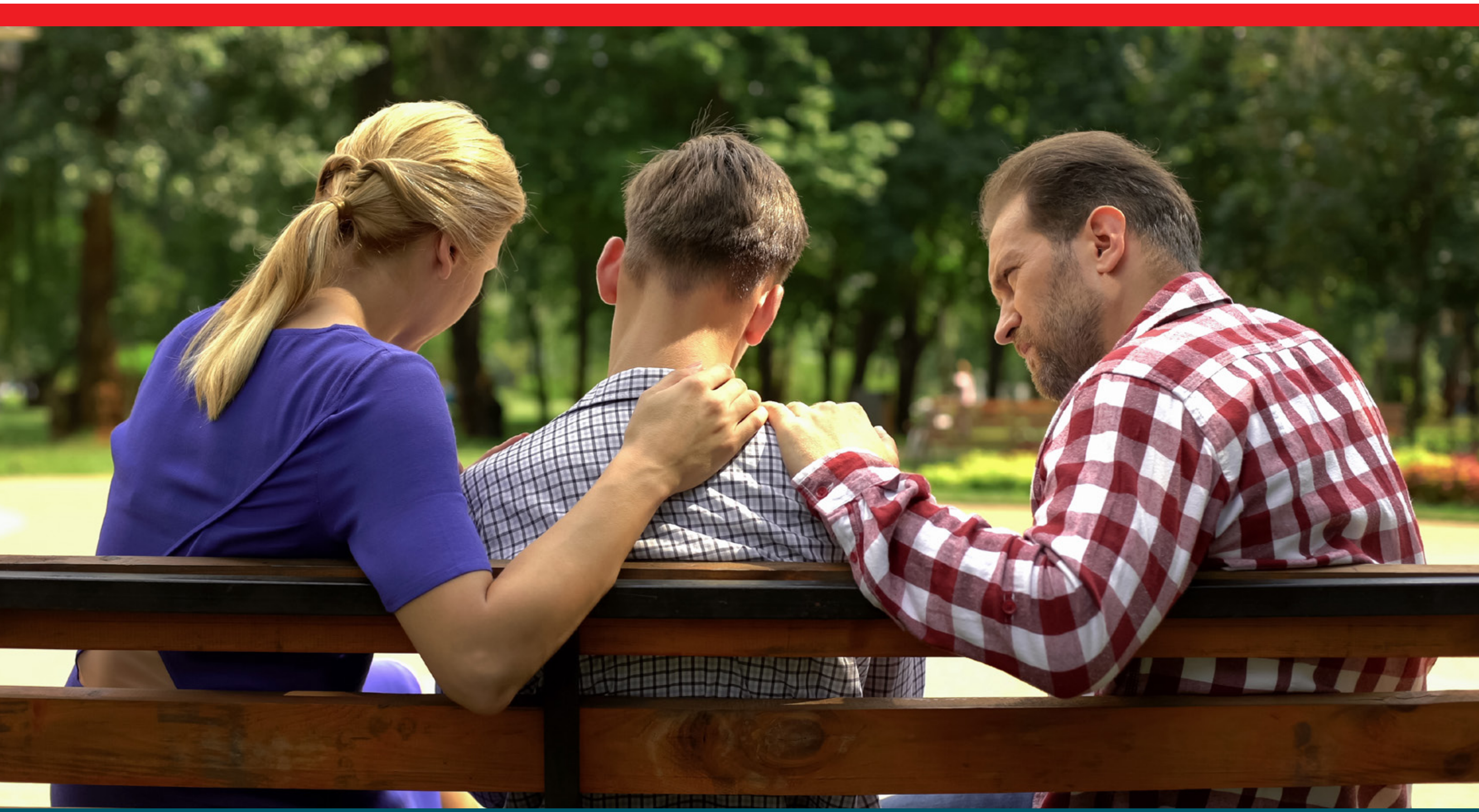
Parent to Parent USA
www.p2pusa.org/parents

Rays for Rare
www.raysforrare.org

Sesame Place
www.sesameplace.com

Sibshops
www.siblingsupport.org

Travel with Disabilities and Medical Conditions
www.tsa.gov/travel/special-procedures



LIFE TRANSITIONS AND FUTURE CARE

Until a child turns 18 years old, a parent or caregiver is considered their legal guardian. Before the child's eighteenth birthday, caregivers should consider if they need to continue making decisions for their child.

The child may keep the right to make some decisions. For example, they might vote or be able to handle a certain amount of money. The court (probate or surrogate) appoints a guardian for an incapacitated person. In some states, a separate person (a conservator) is assigned to handle the child's financial matters.

For older caregivers, it is important to have a plan in place for the child's future care. Caregiving becomes more difficult with age, and older populations are more likely to need caregivers themselves. Having a plan in place early on will ensure the child continues to receive needed care. Further, there can be the unspoken expectation that other members of the family, such as a sibling, will help care for the child when the main caregiver becomes too old or passes on. It is important the entire family sits down and discusses future care plans so the rights and wishes of all involved are honored.

GUARDIANSHIP

Sometimes a court may assign one or more caregivers. Depending on a family's situation, an independent guardian may be assigned. The child's best interest is the priority. Caregivers should also consider who to appoint as a guardian after their death. Anyone over 18 years of age may be a guardian, but caregivers should think through this choice carefully and talk to the person they have in mind before deciding. If everyone agrees, this arrangement should be documented in writing with the help of a lawyer.

RESIDENCE

An adult child with special needs may wish to live in the least restrictive community setting. Some adult children may feel comfortable living with their parent(s) or a family member. Many housing options are available to families. These vary depending on the level of care needed and the family's ability to pay.

Adult children may choose to live in a group home with other special-needs individuals. These residents may not be capable of living on their own and need some support from the staff. A group

Adult children may choose to live in a group home with other special-needs individuals.

home is funded through personal funds or through state programs. When a high level of care is needed, an assisted living facility is a better option. Residents live in apartments and receive help with cooking, bathing, or managing medications. If a higher level of medical care is needed, a skilled nursing facility is the best option. Due to the high cost, families usually choose this as a last resort. Some may qualify for Medicaid coverage.

If an adult child is able to live on their own, they can apply for Section 8 housing. Many disabled people receive Social Security Disability Insurance or Supplemental Security Income. Often, they can get help through the Section 8 program. A trust may cover the cost of a home or rent. This may also include services needed to maintain a home.

Section 8 Housing (The Housing Choice Voucher Program)

Section 8 is a federal program run by the Department of Housing and Urban Development. The program was created to assist low-income families, the elderly, and the disabled in affording safe and decent housing on the private market. Those who participate are not limited to subsidized housing projects and can apply to live in any housing, including single-family homes, townhouses, and apartments, that meet the criteria of the program. Eligibility for Section 8 Housing is determined locally by public housing agencies (PHAs). A family or individual can submit to their PHA, who will determine eligibility based on total gross annual income and family size. If deemed eligible, the PHA will supply a housing choice voucher, and the family is then responsible for finding suitable housing where the owner agrees to rent under the program. It is important to keep in mind that housing assistance is in high demand and often exceeds available resources. Regardless of eligibility, there may be long waiting periods in a given area.

RETIREMENT AND ESTATE PLANNING

Retirement and estate planning may be necessary for both the caregiver and the child. Saving is a challenge for all families, but planning for a child who will remain dependent as an adult is even more challenging. In addition to current medical costs, there are also future support costs. As well as financial support, caregivers should consider their emotional needs and financial needs, legal rights, and government benefits during their lifetime.

Estate planning for a child with an illness or physical disability is important. There are many choices when planning ahead. Three options are:

- A special needs trust (sometimes called a supplemental needs trust)
- A life insurance policy (whole or term), and
- A savings account, such as the ABLE (Achieving a Better Life Experience) account

Health and life insurance, wills, and advanced directives for both the caregiver and child are all items to also be considered. Consulting with a financial, legal or accounting advisor for guidance can be very helpful. They can help create a plan around specific needs and help navigate the financial and tax implications of different options.

Special Needs Trust

Special Needs Trusts (SNTs) are accounts that allow caregivers to save for a family member with disabilities. These SNTs may be established at any time in the child's life. These accounts may pay for anything that benefits the beneficiary alone, with the exception of food and housing, and it will also allow the option of receiving disability benefits provided by Social Security. The design of the trust will outline the restrictions. A trustee may assist the beneficiary in managing their finances. A trust is separate from income when applying for aid, such as Medicaid or food stamps.

There are two types of SNTs to consider: first-party and third-party trusts. Third-party SNTs are the most commonly used when planning in advance for a child with special needs. These SNTs are typically funded upon the death of the beneficiary's caregivers or the individual who established the SNT. First-party SNTs are more often used when a person with a disability inherits money or property or receives a court settlement.

If it is not possible to set up a trust for the special needs the child requires, another option is a last-to-die, or survivorship, insurance policy. Because payments are made over two lifespans, the premiums are smaller and can provide larger benefits down the road. With this type of policy, taxes are paid upon the first person's death to reduce costs later on.

Achieving a Better Life Experience Accounts

Achieving a Better Life Experience Accounts (ABLE) allow families to save for a family member with disabilities without reducing eligibility for disability benefits provided by Social Security. The account must be established before the child turns 26 years old. Other requirements for these savings accounts vary by state. The money can be used for a broadly defined list of qualified disability expenses which can include medical treatment, job training, assistive technology, housing, and legal administrative fees.

www.irs.gov/government-entities/federal-state-local-governments/able-accounts-tax-benefit-for-people-with-disabilities

HELPFUL WEBSITES

Achieving a Better Life Experience Accounts
www.irs.gov/government-entities/federal-state-local-governments/able-accounts-tax-benefit-for-people-with-disabilities

The Center for Complicated Grief
www.complicatedgrief.columbia.edu

The Compassionate Friends
www.compassionatefriends.org

Financial Advocacy in RARE: Navigating the U.S. Health System for Young Adults
www.globalgenes.happyfox.com/kb/article/270-financial-advocacy-in-rare-navigating-the-u-s-health-system-for-young-adults/

Parenting a Child with a Life-Limiting Illness
www.globalgenes.org/wp-content/uploads/2014/02/GG_toolkit_4final_print.pdf

BEREAVEMENT AND GRIEF SUPPORT

Rare and/or serious illnesses are often life-limiting, and as a result, caregivers of children with these conditions may face the loss of their child. Some may experience anticipatory grief and not only mourn the future loss of a child, but also milestones or achievements not experienced along the way. To manage these devastating circumstances and associated trauma, bereavement support groups and resources can help in establishing and acknowledging the grief and other emotions associated with the loss of a child. Some bereavement and grief support resources include:

The Center for Complicated Grief www.complicatedgrief.columbia.edu	The Center for Complicated Grief provides current research about complicated grief through support via social media, other online and published media, and community presentations, as well as resources for therapeutic options.
The Compassionate Friends www.compassionatefriends.org	The Compassionate Friends has chapters in all fifty states to deliver highly personal comfort, hope, and support to every family experiencing the death of a child, as well as to help others better assist the grieving family. Resources include informational books and other media, online and in-person support groups, and crisis telephone lines.
Courageous Parents Network www.courageousparentsnetwork.org/guides/coping-with-anticipatory-grief	Courageous Parents Network provides various resources to parents and caregivers, including information on topics such as anticipatory grief and shared decision making.
Parenting a Child with a Life-Limiting Illness www.globalgenes.org/wp-content/uploads/2014/02/GG_toolkit_4final_print.pdf	Offered by Global Genes, this toolkit is a helpful starting point for parents and caregivers of children who have a life-limiting illness.

> **To manage devastating circumstances and associated trauma, bereavement support groups and resources can help in establishing and acknowledging the grief and other emotions associated with the loss of a child.**

SELF-CARE FOR THE CAREGIVER

Caring for a child with a rare and/or serious illness takes a toll on the mental and physical health of caregivers. *The Caregiving in the U.S. 2020* report published by the NAC and AARP revealed that family caregivers across all diseases and conditions are in worse health compared to caregivers in 2015.[37]

It can be a vulnerable time as caregivers struggle to provide care and adjust to the loss of initial expectations and hopes for their child, adapt their career plans, and make other monumental and significant life changes. After learning a child has a rare and/or serious illness, a caregiver begins the lifelong process of adjusting. Even after a diagnosis, so many unknowns exist that it can be very stressful. Caregivers often express feelings of isolation in the care of their child, due to both the burden of caregiving tasks as well as their inability to relate to other parents.

In addition to the usual parenting responsibilities, caregivers of children with special needs often have to manage frequent doctor or therapy appointments, medical treatments, hospitalizations, and school issues that can be overwhelming while trying to integrate the family routine. There may be added stress from worry about the child's vulnerability, pain level, and explaining health needs to those outside of the family.

Among the caregivers that participated in our focus groups, feelings of fear and anxiety related to the child's health and mortality, both anticipatory and post-traumatic, were common. The caregiver's own mortality in the context of future care for the child was also of concern. Caregivers reported feelings of uncertainty as well as guilt when making decisions about their child's care, their own ability in being an effective caregiver, and in exploring options for self-care.

Grief and distress were common emotions expressed by caregivers of children with rare and/or serious illnesses. The causes for grief

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[37] AARP and National Alliance for Caregiving. (2020). Caregiving in the U.S. Retrieved from https://www.aarp.org/content/dam/aarp/ppi/2020/05/full-report-caregiving-in-the-united-states.doi.10.26419-2Fppi.00103.001.pdf

can vary, but include the loss of a child, loss of expectations for their child, and anticipatory grief for future circumstances. Overall, caregivers described their grief as varied but chronic, often triggered by specific events and without an end in sight.

Taking care of a child with a rare and/or serious illness can be both physically and emotionally taxing but can also be incredibly rewarding and provide a sense of purpose. Families will need to find new ways of doing things and create a new sense of normal. There is an adjustment period during which caregivers will often feel vulnerable and struggle to believe that things will ever get better.

The time and effort that goes into caring for a child with a rare and/or serious illness can make it seem hard or nearly impossible for caregivers to take a step back and focus on themselves. Days may be filled with caregiving responsibilities, family obligations, and sometimes work duties, leaving no time to unwind or rest. This can add stress to an already difficult situation. For everyone's sake—the caregiver, the child, and the family—it is important for caregivers to take care of themselves mentally, physically, and emotionally. It is important to remember that caregiving is a tough job that takes a toll on body, mind, and spirit. Like anyone performing a tough job, a caregiver's mind and body deserve breaks and rewards.

Seeking out someone to talk to, such as a family member, a friend, or a professional counselor, can be helpful in working through the initial adjustment period. A support system or network of other caregivers in similar circumstances, closed Facebook groups, or support groups can be beneficial. There is hope, and caregivers need to know they are not alone.

Professional counseling may be useful, both individually and for the family, to help address the emotions and concerns that often impact family dynamics and interactions. See section below on "Emotional Health" for more recommendations.

Caregivers cannot forget to carve out time for themselves. Even if it only ten minutes a day, it is essential to step back and recharge. It is possible to accept the situation and be empowered, but everyone needs some down time.

HOW CAREGIVERS CAN TAKE CARE OF THEMSELVES

Being an effective caregiver can feel overwhelming due to a seemingly endless to-do list that involves an ill child as well as other personal and family responsibilities. It can be especially difficult for caregivers to meet the challenge of taking care of themselves. There may be barriers to caring for themselves, such as dedicating personal time, finding helpful resources, and determining the kind of activities that work best for the child. Caregivers tend to put themselves last, but self-care is the most important thing for all involved. Family caregivers carry the weight of not only providing personal and instrumental care (e.g., running errands, medications), but they often must manage other responsibilities: *What do I do when I get tired? Keep going? What do I do when I need help? Keep going? What happens when I cannot go anymore?* Self-care includes recognizing when to get help with balancing physical, emotional, financial, social, and spiritual needs.

> **Being an effective caregiver can feel overwhelming due to a seemingly endless to-do list that involves an ill child as well as other personal and family responsibilities.**

There are several ways for caregivers to care for themselves in ways that fit their busy schedules.

Physical Health

Physical health includes regular exercise, sufficient sleep, and a healthy diet. Physical activity has many proven benefits. For caregivers, however, it may be difficult to find the time. With planning, a short walk can bring many benefits and can be accomplished with a mindful approach. Getting outside is good for the spirit and soul. Following are some tips to include regular physical activity:

- Set a reminder to take a walk.
- Plan a route ahead of time.
- Wear walking shoes or keep them in the car.
- For smartphone users, consider downloading an app such

as "Map My Walk" (free) to help provide motivation related to calories burned, distance, and so on. www.mapmywalk.com/app

- Set a goal for the walk, such as twenty minutes: ten minutes one way, ten minutes back.
- If leaving the home is a barrier to physical activity, look for opportunities to exercise at home. Climb stairs if available, run in place, or do floor stretches. Several online programs provide guidance for in-home exercises.
- Although it can be challenging at times, eating well is important. Eating a balanced diet is essential to overall health, and to the ability to care for a someone else.
- Stay hydrated by drinking plenty of water and minimize alcohol intake during stressful times.
- Sleep is important. Get as much uninterrupted sleep as possible. When sleeping through the night isn't possible, find time to nap, even for short periods of time.

Emotional Health

Caregiving can be an emotional journey. Often, caregivers need emotional support from friends, family, or others on a similar journey. Knowing that there are others with similar experiences who are available for support can provide much-needed comfort. Emotional self-care provides the strength to continue the caregiving journey. There are many ways to improve emotional health:

- Caregiver support groups are a wonderful way to connect with others who have similar experiences. There are many types of groups such as condition-specific, gender-specific, online, or in-person groups.
- Social support is critical to emotional health. Set aside time on a regular basis to meet a friend for coffee or have a friend visit, if only for a short time.
- Mental health is important and requires looking after for everyone, not just caregivers. Counselors and therapists provide critical support and help you work through your everyday life and the challenges you encounter.
- Practice self-care by doing something solely for oneself. Go to a movie or dinner. Finding the time may be a challenge but it is important to realize that taking good care of someone else depends on taking good care of oneself. Caregivers must find some time for their own needs so they can continue to care for the care recipient.

Financial Health

Self-care means tending to finances. Caring for a child with serious illness can often mean out-of-pocket expenses that quickly add up. Meeting with a financial advisor can provide a sense of financial control and help make informed decisions about the care situation. See Economic Impact of Caregiving in this Guidebook for more.

Often, caregivers need emotional support from friends, family, or others on a similar journey.

Spiritual Health

Many believe that humans are hardwired to seek the ground of their being. Humans in distress find attending to their spiritual needs essential. Attending to spiritual health can take many forms, including prayer, meditation, yoga, spending time in nature, personal writing, or attending worship services. An additional benefit of spiritual practice for caregivers is the dedicated alone time. One thing we know about caregiving is that it leaves very little time for the caregiver. There are many ways to infuse spiritual activities into daily life:

- Begin each day with a few moments of spiritual focus.
- Take a walk in a peaceful or quiet place.
- Go online for a few minutes to participate in an online yoga class, meditation lesson, or writing group.
- Escape to the local library, even if only for a moment. There are many books with topics related to spirituality and many libraries offer online reading.
- Find local groups through www.meetup.com. Community centers can often provide resources.

Some caregivers of children with a rare and/or serious illness embrace spirituality to make sense of the suffering, to overcome loneliness, anxiety, and depression, and to improve overall emotional well-being. Focus group participants reported a need for emotional and spiritual support throughout the caregiving experience and when dealing with bereavement or other grief. Further research agrees with this finding. As many as nine in ten (94.7 percent) of parents caring for children with progressive conditions experienced a high degree of spirituality.[38,39] Research suggests that spirituality effectively mitigates some of the adverse emotional outcomes that rare caregivers face.[40,41]

Recognizing that caregivers are diverse in their belief systems, this guidebook does not prescribe a spiritual method. Instead, it encourages caregivers to lean on their core spiritual beliefs for hope, strength, and courage. Health providers and communities might also suggest helpful resources. Chaplains or clergy members in health centers are accessible spiritual resources who have received training to support patients and families. Chaplains are typically trained to work across faiths and cultures and to advocate for patients and their families. Patients and families have a choice to access this service, and many find it helpful. Caregivers can, at any point, choose to discontinue the relationship or to seek other supports, such as those offered by their faith communities. The key is to access support that agrees with one's core beliefs.

Caregivers might explore some evidence-based spiritual practices, such as prayer, assigning a purpose or meaning to one's hardship,[42] and maintaining a gratitude journal.

RESPITE

From time to time, caregivers will need a break from caring for a child with a rare and/or serious illness. Even if the care of the child is shared with others, caregivers who take "me time" often feel refreshed and better prepared to care. The more effectively caregivers care for themselves, the stronger they will be to care for their child. There are different types of respite care available. These include an alternate caregiver coming into the home, daily or weekly programs hosted by a family, residential facility, or a sleepaway camp for the child. Respite care can be expensive, but there are public and private programs that can help with funding. Access to Respite Care and Help (ARCH) is a respite care assistance organization that can be a useful resource to locate respite services and caregiver supports in a community and offers funding guidance. More information is available at www.archrespite.org.

From *How Caregivers Can Cultivate Moments of Positivity* :[43]

- Noticing positive events: Being aware of the good things in your life, an antidote to narrowly focusing on the negative.
- Capitalizing on positive events: A way of extending and savoring your good feelings, such as by telling someone else, writing them down, or remembering the event later.
- Mindfulness: Being aware of the present moment without judgment. Participants learn a guided mindful breathing exercise and incorporate mindfulness into daily activities like brushing teeth and washing dishes.
- Noting personal strengths: Recognizing the personal resources you have at your disposal and how you enact them in your life, an antidote to feeling helpless.
- Setting attainable goals: Choosing aims that are challenging but not too challenging, allowing you to feel accomplished and successful.

https://greatergood.berkeley.edu/article/item/how_caregivers_can_cultivate_moments_of_positivity

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[38] Siden, H. & Steele, R. (2015). "Charting the Territory: Children and Families Living with Progressive Life-threatening Conditions." Pediatrics & Child Health 20(3), 139–44. Doi: 10.1093/pch/20.3.139.

[39] Anandarajah, G., & Hight, E. (2001). "Spirituality and Medical Practice: Using the HOPE Questions as a Practical Tool for Spiritual Assessment." American Family Physician, 63(1), 81. https://www.aafp.org/afp/2001/0101/p81.html

[40] Tarakeshwar, N., Vanderwerker, L. C., Paulk, E., Pearce, M. J., Kasl, S. V., & Prigerson, H. G. (2006). "Religious Coping is Associated with the Quality of Life of Patients with Advanced Cancer." Journal of Palliative Medicine, 9(3), 646–657. https://doi.org/10.1089/jpm.2006.9.646.

[41] CAPC. (2020). Addressing the Spiritual Care Needs of Patients with Serious Illness.
https://www.capc.org/toolkits/addressing-spiritual-care-needs-patients-serious-illness/

[42] Frankl, V.E. (1962). Man's Search for Meaning: An Introduction to Logotherapy. Beacon Press.

[43] Newman, K. N. (2015). How Caregivers Can Cultivate Moments of Positivity. Retrieved from https://greatergood.berkeley.edu/article/item/how_caregivers_can_cultivate_moments_of_positivity.

In addition to the programs and camps described previously (see Education and Recreation in this guidebook), the following are examples of respite retreats specifically for caregivers:

Caregiver-Specific Retreats

ANGEL AID www.angelaidcares.org	ANGEL AID is a nonprofit organization dedicated to source research, relief, and inspiration for mothers of children with rare diseases.
David's Refuge www.davidsrefuge.org	David's Refuge is a faith-based organization located in upstate New York that provides respite, resources, and support to parents and caregivers of children with special needs or life-threatening medical conditions. Additionally, they offer virtual support and resources on health and wellness as well as spirituality.
A Mother's Rest www.amothersrest.org	A Mother's Rest is a nonprofit charity that specializes in coordinating therapeutic respite retreats for parents of children with extra and special health care needs. It offers retreats to mothers and fathers to get away and address caregiver fatigue, both physiological and mental. The organization partners with nationwide hotels, inns and lodging providers to make recuperative respite available and affordable for caregivers.

HELPFUL WEBSITES

American Counseling Association: Find a Counselor
www.counseling.org/aca-community/learn-about-counseling/what-is-counseling/find-a-counselor

A Mother's Rest
www.amothersrest.org

Access to Respite Care and Help
www.archrespite.org

ANGEL AID
www.angelaidcares.org

David's Refuge
www.davidsrefuge.org

Map My Walk
www.mapmywalk.com/app

Meet Up
www.meetup.com

Caregiving in the U.S. 2020
www.caregiving.org/wp-content/uploads/2021/01/full-report-caregiving-in-the-united-states-01-21.pdf

Psychologist Locator
https://locator.apa.org

Rare Caregiver Respite Program
www.rarediseases.org/wp-content/uploads/2019/05/NRD-1182-RespiteCare_SS_1up_FNL_NoCrops_Fixed.pdf

From time to time, caregivers will need a break from caring for a child with a rare and/or serious illness. Even if the care of the child is shared with others, caregivers who take "me time" often feel refreshed and better prepared to care.

GETTING INVOLVED IN CAREGIVING ADVOCACY

The nature of being a caregiver to a child with a rare and/or serious illness means that the caregiver will more likely than not become an advocate in some way.

When the child is young, the caregiver is their voice to their health care team, advocating for tests and treatments; as the child grows older, the caregiver supports them in their decisions and needs. While caring for a child with a rare and/or serious illness can take up a lot of time and energy, sharing the family's experiences and perspectives as a caregiver can help advance advocacy efforts to improve the experiences of families across the rare disease community. Actions such as telling the family's story, making others aware of the child's illness, or simply exchanging useful tips to other caregivers are all forms of advocacy that can encourage broader support and positively impact other families.

There are several ways for caregivers to engage in advocacy. Sharing a life story creates awareness and highlights what needs to be changed by policy. Defining goals or specific changes to advocate for provides focus to begin supporting and engaging in work that will create positive change. Acknowledging the unique challenges facing rare and/or serious illness caregivers, specific changes that caregivers can elevate both personally and with policy makers include:

- Expanding and enhancing Centers of Excellence (COE) programs within healthcare institutions to provide expertise care in rare and serious diseases.
- Ensuring that health care for rare and/or serious illnesses is not negatively impacted by overt or implicit bias based on race, color, sex, national origin, religion, orientation, age, disability, or socioeconomic status.
- Increasing funding for clinical research for rare and/or serious illnesses, including the development of innovative therapies and increased participation in clinical trials by underrepresented populations, such as minorities.
- Increasing services and supports to help caregivers efficiently navigate the health system, including providing qualified respite, skilled nursing and home care options, and developing a centralized resource for identifying expert health professionals in rare and/or serious illnesses.

- Streamlining the insurance claim process for reimbursement of services and equipment, including improving the caregiver's understanding of treatment procedures and availability for rare and/or serious illnesses.
- Facilitating health care coordination and collaboration across providers for rare and/or serious illnesses by eliminating silos in care.
- Training health care providers to: 1) communicate with parents and guardians with respect; 2) acknowledge their roles as caregivers; and 3) deliver health care and social support services using culturally competent approaches.
- Increasing caregivers' access to appropriate training in providing proper health care for rare and/or serious illnesses within the home environment.
- Helping parents and guardians of children with rare and/or serious illnesses locate services and support by encouraging them to recognize their caregiving roles and to identify as caregivers.
- Assisting caregivers in navigating the health care system by expanding networks of qualified case managers, health professionals, and health care facilities to have the necessary expertise and resources to manage rare and serious diseases.
- Protecting families' financial security through increased eligibility for and access to state and federal programs, health insurance coverage, and payment assistance programs to cover or offset the high costs of care and treatment for rare and/or serious illnesses.
- Reducing the financial impacts of caregiving through refundable tax credits, helping families cover out-of-pocket expenses, paid family and medical leave policies, and flexible work arrangements.

Connecting with advocacy groups provides support and resources. It also provides an avenue for caregivers, if and when they are ready, to connect with lawmakers and elected officials who have the ability to make progressive change.

The following are good starting points for advocacy resources:

Genetic and Rare Diseases Information Center Advocacy Organizations Database
www.rarediseases.info.nih.gov/search?keyword=contentType%3Dorganization

Global Foundation for Peroxisomal Disorders
www.thegfpd.org/

National Alliance for Caregiving
www.caregiving.org

National Organization for Rare Disorders
www.rarediseases.org/for-patient-organizations/ways-partner/advocacy/

Rare Diseases Clinical Research Network
www.rarediseasesnetwork.org/pags

Rare Disease Day

www.rarediseaseday.org

The main objective of Rare Disease Day is to raise awareness among the public and decision makers about rare diseases and their impact on patients' lives. Started in 2008 by EURORDIS, today there are numerous partners across the United States who hold events to promote awareness for rare diseases. The event is held on the last day of February each year.

Rare Disease Week

www.everylifefoundation.org/rare-advocates/rare-disease-week/

The main objective of Rare Disease Day is to raise awareness among the public and decision makers about rare diseases and their impact on patients' lives. Started in 2008 by EURORDIS, today there are numerous partners across the United States who hold events to promote awareness for rare diseases. The event is held on the last day of February each year.

APPENDIX: HELPFUL WEBSITES

99Pledges 34
A fundraising site that allows caregivers to set up a donation page for free.
www.99pledges.com

A Mother's Rest 52
This respite foundation provides respite retreats for caregivers of children with disabilities or rare and/or serious diseases. The foundation runs a series of therapeutic retreats that are meant to provide true rest, and can be specifically geared toward fathers, veterans, couples, and families.
www.amothersrest.org

Achieving a Better Life Experience Accounts (ABLE) 47
ABLE accounts create tax breaks for people with disabilities, allowing states to designate savings for eligible people. Visit the IRS for more information, details, and forms.
www.irs.gov/government-entities/federal-state-local-governments/able-accounts-tax-benefit-for-people-with-disabilities

Air Charity Network 33
Serving all fifty states, the Air Charity Network provides free plane rides to those in need of specialized health care facilities or other destinations that need to be visited due to family, community or national crisis. The service provides transport to those who are financially or otherwise unable to travel on public transportation.
www.aircharitynetwork.org

Agency of Healthcare Research and Quality (AHRQ) 24
A division of the Department of Health and Human Services, AHRQ works to ensure health care is made safer, higher quality, more accessible, equitable and affordable, and that the reasoning and ability to do so is backed by research and evidence.
www.ahrq.gov

All of Us Research Program 26
This National Institute of Health program is working to create one of the most diverse, comprehensive health database in history. Researchers will use the data collected in order to understand more about how biology, environment and lifestyle may affect health, in an effort to treat and prevent disease in the future.
www.allofus.nih.gov

American Cancer Society 33
The American Cancer Society is a nonprofit with the goal of eradicating the world of cancer. In support of this goal, they fund and conduct research, spread information on prevention and provide help and support to patients.
www.cancer.org

American Association of Caregiving Youth 11
The only group dedicated solely to caregiving youth (caregivers 18 years and under), the American Association of Caregiving Youth advocates for and educates on this important division of caregivers through the Caregiving Youth Institute and the Caregiving Youth Project.
www.aacy.org

Athletes Serving Athletes (ASA) 38
ASA's goal is to help those with limited mobility to train for and be able to run in mainstream running events. They do so through their WingMan Program, which connects runners from the community to those in need in order to support and empower their own running.
www.athletesservingathletes.org

The Assistance Fund 31
The Assistance Fund is a nonprofit that helps others in managing their health by assisting in covering financial needs, including co-payments, co-insurance, and deductibles. The Assistance Fund covers seventy specific disease programs, all of which cover the FDA approved medication for that disease.
www.tafcares.org

Benefits.gov 32
Run by the U.S. government, Benefits.gov is a single source for information on all benefits offered by the government. The site can tell caregivers what benefits they or their children may be eligible for and how to apply to these benefits.
www.benefits.gov

Black Women's Health Imperative (BWHI) 10
BWHI is dedicated to improving the health and wellness of Black women and girls, which includes emotional health, physical health and financial health. They do so by investing in evidence-based strategies that create new programs geared towards their goal and by advocating for health-promoting policy.
www.bwhi.org

CanCare 43
CanCare is a community of those who have had experience with cancer in the past to support and inspire current cancer patients and their caregivers. It offers emotional support and resources for cancer patients.
www.cancare.org

CancerCare Copay Assistance 31
CancerCare's Copay Assistance can help with the treatment barriers to cancer that can be caused by need for financial assistance. The assistance will provide co-payments for prescribed treatments or, if they cannot help, will offer resources that may be able to.
www.cancercare.org/copayfoundation

Care Calendar 34
Care Calendar is a site that allows friends, neighbors and family members to coordinate care for others during times of illness, need, or large life changes. Tasks that need to be done can be listed for those on the calendar to claim and assist with.
www.carecalendar.org

Caregiver Action Network (CAN) 16
CAN is an organization working to improve the lives of family caregivers through education, peer support and resources. Caregivers can turn to CAN to connect with other caregivers and rare disease caregivers can access CAN's rare disease specific site for resources and support.
www.caregiveraction.org

Catastrophic Illness in Children Relief Fund (CICRF) 32
CICRF, available in Massachusetts and New Jersey, is a fund that financially assists families in their own respective states in order to help children with an illness or condition that is otherwise not covered by insurance.
Massachusetts: www.mass.gov/catastrophic-illness-in-children-relief-fund-cicrff
New Jersey: www.state.nj.us/humanservices/cicrf/home/

The Center for Complicated Grief 47
The center helps clinicians and the general public to learn how to recognize complicated grief in order to support and help those experiencing it. Increased awareness of complicated grief will ensure those suffering from it can get the help they need.
www.complicatedgrief.columbia.edu

Center for Parent Information and Resources (CPIRs) 43
CPIRs provide services and supports for children with disabilities, as well as to the families and health professionals that support them.
www.parentcenterhub.org

Children's Flight of Hope (CFOH) 33
CFOH provides free flights to children and families with specialized medical care needs who might otherwise be unable to access this care.
www.childrensflightofhope.org

Children's Health Insurance Program (CHIP) 32
CHIP gets low-cost medical care coverage for families who make too much to qualify for Medicaid but still need financial assistance. CHIP is offered in every state.
www.healthcare.gov/medicaid-chip/childrens-health-insurance-program/

Children's Hospital of Philadelphia (CHOP)........24
The first hospital in the nation devoted solely to the care of children, CHOP has been the home of countless medical innovations and discoveries that have improved pediatric healthcare and the lives of children.
www.chop.edu

Children's Inn at NIH...33
This nonprofit provides a place to stay for children and families who are being diagnosed or treated in an NIH clinical study. The Inn also provides a wide range of programs to children, teens, and young adults with a rare and/or serious illness.
www.childrensinn.org

Children's National Rare Disease Institute (CNRDI) 13
The rare disease institute at Children's National focuses on providing resources, support, and treatment to children with rare and/or serious diseases. The center is solely focused on advancing the care and treatment of children and adults with rare genetic diseases.
www.childrensnational.org/departments/rare-disease-institute

Clinical Trials...26, 29
This database provides a comprehensive list of clinical studies conducted around the world. Knowledge of current and past clinical studies can help a caregiver learn more about the child's rare and/or serious illness.
www.clinicaltrials.gov

Council of Accountable Physician Practices (CAPP) 22
CAPP is a physician-led coalition that works to elevate the voices of physicians and their organizations through research, education and communication in order to improve American health care. They stand for coordinated care, outcomes-based payment, health information technology, physician leadership, and quality improvement.
www.accountablecaredoctors.org

Courageous Parents Network (CPN)...... 41, 43, 47
CPN supports caregivers caring for a child with a serious illness through various programs that include education, community, and advocacy surrounding palliative care. CPN hopes to instill confidence in caregivers in their times of decision making and in their interactions with medical personnel.
www.courageousparentsnetwork.org/

David's Refuge...52
David's Refuge provides a place of community and respite for caregivers of children with special needs or serious life-threatening conditions. They host weekend respites, caregiver night outs, and events throughout the year to ensure caregivers are relaxed and taking care of themselves.
www.davidsrefuge.org

Disney Parks..44
Disney Parks are a series of family-friendly amusement parks located both nationally and internationally. Disney provides accommodations to children who may have specific needs and their families, making it a manageable vacation destination.
https://disneyparks.disney.go.com

Double H Ranch...38
Located in the Adirondacks, Double H Ranch is a camp for children facing serious illness. Their goal is to provide a camp experience that is memorable, fun, and physically and medically safe for the children attending.
www.doublehranch.org

Early Intervention 35
These are services and supports, such as speech therapy and physical therapy, that can be offered to babies and young children exhibiting developmental disabilities and delays and their families. Services are available for free or at a reduced cost to any eligible child.
https://www.cdc.gov/ncbddd/actearly/parents/states.html#:~:text=Is%20the%20term%20used%20to,of%20the%20child%20and%20family

Easterseals..43
Easterseals is a nonprofit that provides services, education, outreach, and advocacy to and for people with disabilities, veterans, seniors, and caregivers. Their goal is to make it possible for all to participate in the wider community.
www.easterseals.com

EURORDIS Rare Diseases of Europe 16
EURORDIS is a nongovernmental alliance of patient organizations and individuals involved in the European rare disease space, with the goal of creating a pan-European community to be the voice of those with rare disease at the European level.
www.eurordis.org

EveryLife Foundation..16
EveryLife is a nonprofit with the mission of empowering the rare disease community to advocate for themselves and others for science-driven legislation and policy that will lead to equitable access to health care.
www.everylifefoundation.org

Facebook Fundraising 34
Facebook Fundraising allows users to post a cause on the Facebook platform so friends, family, and others can donate toward a financial goal.
www.facebook.com/fundraisers

Family Voices...43
Family Voices is an organization of families and friends of children with special health care needs and disabilities who have created a network of family-led organizations across the nation. They aim to work with families at an individual and healthcare level to improve health care for children.
www.familyvoices.org

FDA COVID-19..29
These pages provide FDA information on the COVID-19 pandemic and the vaccines being created and approved to fight the pandemic.
www.fda.gov/news-events/fda-voices/rare-disease-therapy-development-and-access-remain-top-fda-priorities-during-covid-19
www.fda.gov/emergency-preparedness-and-response/coronavirus-disease-2019-covid-19/covid-19-vaccines

FDA Expanded Access for Patients...................27
Expanded Access is an opportunity for patients to try investigational medical products outside of a clinical trial when there are no other therapies available. The FDA provides information on this practice and the considerations they take on access requests.
www.fda.gov/news-events/expanded-access/expanded-access-information-patients

FDA Office of Orphan Products Development (OOPD) 26
OOPD's goal is to advance the evaluation of drugs and treatments that may help with the diagnosis and treatment of rare diseases. They do so by creating programs and offering sponsorships for the development of products for rare disease.
www.fda.gov/industry/developing-products-rare-diseases-conditions

Fundly..34
Fundly is a fundraising site that allows a user to tell their story as part of a crowdsourcing fundraising campaign for such needs as medical treatments and other health care issues.
www.fundly.com

Genetic Alliance...16
This organization works in research, advocacy, and policy to represent the voices of millions with genetic conditions. They encourage access to quality resources and fostering leadership from those in the community.
www.geneticalliance.org

Genetics and Rare Diseases Information Center (GARD) 15
GARD is joint funded by the National Institute of Health to provide the public with up-to-date and comprehensive data on rare or genetic diseases. The information is provided in both English and Spanish.
www.rarediseases.info.nih.gov

Global Foundation for Peroxisomal Disorders (GFPD)..17
GFPD provides support to patients and families impacted by peroxisomal disorders by connecting those in the community. They hold family and scientific conferences biannually to provide patients, families, and scientists in the field a space to collaborate on ways to improve the lives of those with peroxisomal disorders.
www.thegfpd.org

Global Genes..................10, 12, 16, 26, 28, 31, 47
Global Genes serves patients and families impacted by rare diseases by building connected communities, providing access to information and resources, encouraging sharing data and stories, and creating links to scientists, researchers, and government.
www.globalgenes.org

GoFundMe...34
GoFundMe is a crowdsourcing platform that allows users to raise money from friends, family, and the general public to fund potential medical needs.
www.gofundme.com

Good Days...31
Good Days is a charity that offers financial assistance for those with rare and or serious diseases so that they can afford the treatment they need. The charity also provides emotional support to patients through its Patient Care Navigators.
www.mygooddays.org

Healing the Children 33
Healing the Children is an international charity that provides medical and dental services to children who cannot afford or have no access to them.
www.htcne.org

HealthWell Foundation..31
HealthWell provides financial assistance for prescription co-pays, pediatric treatment costs, travel costs and health insurance premiums, and deductibles and co-pays for patients with chronic or life-altering diseases and their families.
www.healthwellfoundation.org

Hemophilia Federation of America (HFA) 34
HFA is focused on the bleeding disorders patient and caregiver community and provides patient education, services, and advocacy.
www.hemophiliafed.org

Hole in the Wall Gang Camp 38
Hole in the Wall Gang is a camp for children with serious medical conditions. Free of charge, the camp runs safe and appropriate traditional camp activities for children to engage in, providing a fun community experience.
www.holeinthewallgang.org

Hospitality Homes..33
Located in the Boston area, Hospitality Homes is a nonprofit that provides free short-term housing to patients and families receiving medical care in the Boston area.
www.hosp.org

HRSA Maternal & Child Health Title V Block Grant Program..32
As one of the largest federal block grant programs, Title V funding supports promoting and improving the lives of mothers and their children, including children with special needs, through access to quality health care.
www.mchb.hrsa.gov/maternal-child-health-initiatives/title-v-maternal-and-child-health-services-block-grant-program

Inspire..20
Inspire is an online message board platform that allows patients, their friends, and families to connect with others experiencing the same kind of illness or rare disease. This allows them to ask questions and share stories.
www.inspire.com

Jack's Caregiver Coalition 10
Jack's is a support group for male caregivers that aims to improve the way they think and feel about their caregiving journey. Jack's holds a variety of programs to help men become more comfortable as caregivers
www.jackscaregiverco.org

The Compassionate Friends 47
The Compassionate Friends is a group that understands how the loss of a child can deeply affect a family. Their goal is to provide highly personalized support to families who have experienced such loss, as well as to help those around them learn how to support the family in their time of need.
www.compassionatefriends.org

The Joint Commission 25
The Joint Commission evaluates health care organizations to ensure they're providing safe, effective and quality care. They envision a world where everyone experiences the safest, highest quality medical care.
www.jointcommission.org

The Lantern Project 14
Sanofi Genzyme and PerkinElmer Geonomics have partnered to create the Lantern Project, a complimentary genetic testing program. They offer diagnostic testing for specific disorders. These disorders and how to test for them can be found on their website.
www.perkinelmergenomics.com/lanternproject/

Make-A-Wish Foundation 39
Make-A-Wish is a nonprofit that aims to grant the wishes of children with chronic, critical, and life-ending illnesses. The organization strives to grant whatever the child wishes for in fullest way possible.
www.wish.org

Meal Train 34
Meal Train is a site that enables family, friends, and neighbors to coordinate a meal train schedule for others in times of high stress or need. The site provides scheduling and the ability to donate gift cards or money to the meal train effort.
www.mealtrain.com

Mercy Medical Angels..33
Mercy Medical is the largest charitable medical transportation system and provides bus passes, train tickets, and flights with volunteer or commercial pilots in order to remove the barrier of transportation to medical care.
www.mercymedical.org

Medicaid 25, 32, 46
Medicaid is a federal and state program that helps with health care costs for those with limited income and resources. Medicaid covers some benefits that Medicare does not.
www.medicaid.gov

Miracle Flights..33
Miracle Flights provide free flights to those in need of life-changing medical care that they cannot access locally. Miracle Flights will pay to fly the caregiver as well as the patient to wherever they need to go.
www.miracleflights.org

Morgan's Wonderland..44
Morgan's Wonderland is an amusement park created for children with disabilities, providing a safe, inclusive, and fun experience for the whole family.
www.morganswonderland.com

National Alliance on Mental Illness (NAMI) ... 28,33
NAMI is an organization providing advocacy, support, education, and public awareness so those affected by mental illness can live improved and healthy lives.
www.nami.org

National Association for Home Care & Hospice (NAHC) 25
NAHC is a trade association that represents the nation's homecare and hospice organizations as well as the chronically ill and disabled patients and caregivers they serve.
www.nahc.org

National Cancer Institute 33
The National Cancer Institute provides support to cancer patients and their families through emotional, practical, and financial services
www.cancer.gov

National Hospice and Palliative Care Organization (NHPCO) 27
NHPCO champions person-centered care that focuses on the needs for comfort, peace and dignity for patients and their families. They represent hospice and palliative care workers so they can provide a better experience for patients and caregivers during this time.
www.nhpco.org

National Human Genome Research Institute (NHGRI) 15, 26
NHGRI collaborates with scientific and medical communities to contribute to research and the study of diseases, to find treatments, and to apply new genomic discoveries to the betterment of human health.
www.genome.gov

National Institute of Health (NIH) National Institute of Nursing Research (NINR) Palliative Care 27
This resource from NIH describes palliative care, its benefits, and the logistics of going into palliative care.
www.ninr.nih.gov/sites/files/docs/palliative-care-brochure.pdf

National Niemann-Pick Disease Foundation (NNPDF) 18
The NNPDF is a patient advocacy and family support organization that supports those affected by Nieman Pick Disease through education, research and collaboration.
www.nnpdf.org

National Organization for Rare Disorders (NORD) 13, 16, 31, 54
NORD is an organization dedicated to those affected by rare disorders and the organizations that support them. They are committed to the identification and treatment of rare disorders through programs of education, advocacy, and research.
www.rarediseases.org

NeedyMeds...33
NeedyMeds is a nonprofit that connects patients to services and organizations that can help them afford their health care and medical costs.
www.needymeds.org

Office for Civil Rights (enforcement office)........36
The Office for Civil Rights protects those who may be discriminated against for race, ethnicity, religion, or disability in their education. They enforce civil rights within the schooling system to ensure equal access to education.
www.hhs.gov/ocr/index.html

Orphan Disease Center................................ 13, 26
The Orphan Disease Center works to develop therapies that can be used to combat multiple rare diseases but focuses especially on diseases with high unmet needs. They also work to ensure equal access to these therapies.
www.orphandiseasecenter.med.upenn.edu

Parent to Parent USA...43
Parent to Parent USA supports the national Parent to Parent network providing emotional support to families and caregivers of those with disabilities and special health care needs. The parent leadership and member organizations of the group help serve as strong advocates and mentors.
www.p2pusa.org/parents

Patient Access Network Foundation (PAN) 31
PAN is a nonprofit organization that helps insured patients living with life-threating, chronic, and rare diseases with the out-of-pocket costs for their medications.
www.panfoundation.org

Patient Advocate Foundation (PAF).................... 31
PAF is a nonprofit that connects patients with chronic, life-threatening, and debilitating diseases with access to care and treatment as recommended by their doctor.
www.patientadvocate.org

Patient Airlift Services (PALS)............................ 33
PALS arranges free flights for patients seeking treatment, diagnosis, or follow-up and who are unable to fly commercially due to finances or other complications.
www.palservices.org

Patient Services, Inc.. 31
Patient Services is funded by patients for patients, and helps pay for medication, provides assistance with health insurance whether it be navigation or co-pay, and provides legal advice and support.
www.patientservicesinc.org

Patient Worthy® 16
Patient Worthy is an online publication that acts as a source of information for rare disease patients, caregivers, and advocates. They provide news updates, new research and other rare disease information to connect those within the community.
www.patientworthy.com

RareConnect 16
RareConnect allows rare disease patients and organizations to connect globally as an online community through their platform. RareConnect is run and moderated by patients and patient advocates and is completely safe to use, with privacy respected and trusted information shared via real patient representatives.
www.rareconnect.org

Rare Diseases Clinical Research Network......... 26
This network is funded by the NIH and connects teams of researchers, patients, and clinicians focused on a specific group of rare disorders. The network encourages collaboration among research so a better understanding of rare diseases, their diagnosis, and their treatment can be reached.
www.rarediseasesnetwork.org

Rare Disease Day.. 18, 54
Rare Disease Day takes place on the last day of February each year to raise awareness among the general public about rare diseases and their impact on patients and caregivers.
www.rarediseaseday.org

Rare Diseases Registry Program (RaDaR) 26
RaDaR is an online resource that advises patient groups on how to begin and maintain online registries for rare diseases. These registries can be of use to research programs and treatment development.
https://registries.ncats.nih.gov

Rare Disease Week...................................... 18, 54
Rare Disease Week is hosted every year by EveryLife Foundation on Capitol Hill. The week gives rare disease community members a chance to come together, learn about federal legislation, meet other advocates, and share their stories.
www.everylifefoundation.org/rare-advocates/rare-disease-week/

Rare & Undiagnosed Network (RUN).................. 14
RUN is an organization that focuses on undiagnosed patients and empowers both rare and undiagnosed patients and their families through education, advocacy, and community.
www.rareundiagnosed.org

Ronald McDonald House Charities 33
RMHC is a charitable group that internationally supports programs that directly improve the health of children and their families. Local chapters provide housing, food, and support to children and families traveling for medical care.
www.rmhc.org

RxAssist.. 33
RxAssist is an active database of patient assistance programs that provide free medication through pharmaceutical companies. The database also includes updated news, articles, and other tools.
www.rxassist.org

Serious Fun Children's Network 38
Serious Fun is a series of camps around the world that serve children with rare and/or serious diseases and their families. The camps create a fun and safe environment where children can experience independence and life beyond their medical condition.
www.seriousfun.org

Sesame Place... 44
Sesame Place is an amusement park for young children and their families based on the children's show Sesame Street. The park provides accommodations for children with serious conditions and needs and has been certified as an Autism Center.
www.sesameplace.com

Sibling Support... 41
Sibling Support is a group dedicated to the siblings of children with serious health concerns or disabilities. The group is responsible for SibShops, are peer support groups for siblings, as well as provide other resources and programs that support siblings.
www.siblingsupport.org

Six Flags 44
Six Flags America is a national amusement and roller coaster park that provides accessibility and accommodation to those with disabilities or other health conditions. They have been designated as a Certified Autism Center.
www.sixflags.com

Social Security Disability Insurance (SSDI)........ 32
This federal program provides assistance to those with disabilities and pays benefits to those who have contributed to the program while employed.
www.ssa.gov/disability

Special Olympics 37
Special Olympics is a global initiative that provides year-round sports training and athletic competition in numerous Olympic-type sports for children and adults with intellectual and developmental disabilities. They also run a Young Athletes Program that promotes childhood development in athletic skills.
www.specialolympics.org

Take Them a Meal... 34
Take Them a Meal is a site that allows users to provide food to families and friends who may not have time to cook for themselves. Users can create a meal schedule for an intended recipient with others, look up the schedule of a recipient if they have expressed need themselves, and even directly send them a meal through the site's online ordering system.
www.takethemameal.com

Tim Tebow Foundation: Night to Shine 39
Night to Shine, hosted by the Tim Tebow Foundation, provides older children with disabilities a prom night experience. The Tim Tebow Foundation also has several other programs and initiatives geared toward helping children with complex medical needs.
www.timtebowfoundation.org

Transportation Security Administration (TSA).... 44
TSA protects airlines and airports, but its security requirements can create challenges for caregivers carrying medications and supplies for a child with complex medical needs. This page explains the procedures and rules related to these concerns.
www.tsa.gov/travel/special-procedures

Undiagnosed Diseases Network (UDN)............. 14
The UDN is a research study group that brings together researchers and experts from across the United States in an attempt to solve medical mysteries using advanced technology and evidence-based support.
www.undiagnosed.hms.harvard.edu

Universal Studios... 44
Universal Studios is a family amusement park that can accommodate children with disabilities or medical needs. The site provides comprehensive guidelines on how to make the trip safe and enjoyable for everyone.
www.universalstudioshollywood.com
www.universalorlando.com

Wings of Hope 33
Wings of Hope is a global nonprofit that uses planes to ensure people around the world have the resources and access they need to meet their developmental goals.
www.wingsofhope.ngo

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National Alliance for Caregiving

About the National Alliance for Caregiving

Established in 1996, the National Alliance for Caregiving is a non-profit coalition of national organizations focusing on advancing family caregiving through research, innovation, and advocacy. NAC conducts research, does policy analysis, develops national best-practice programs, and works to increase public awareness of family caregiving issues. Recognizing that family caregivers provide important societal and financial contributions toward maintaining the well-being of those they care for, NAC supports a network of more than 80 state and local caregiving coalitions and serves as Secretariat for the International Alliance of Carer Organizations (IACO). Learn more at **www.caregiving.org**.

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