



LIFE

MEDICAL

# Raregivers™ Emotional Journey Map Guidebook

Includes a **DETACHABLE POSTER** of the  
Raregivers™ Emotional Journey Map



## When life gets medical, people get emotional.

One in ten families worldwide are living with uncured rare, chronic and complex diseases - many more are undiagnosed. Our community is global, reaching every remote corner of every country on earth. Yet we are fragmented. There are more than 10,000 rare diseases and we are so often focused on the specific disease, we forget to focus on what is common - our feelings.

We have a shared experience. Every community suffers from the social, emotional, financial and physical impacts of disease. The suffering of the care providers are often unseen as precious resources are used for treatment and care of our loved ones. We also grapple with these facts: half of rare patients are children and a third of these children will not see their 5th birthday. There are only treatments for 5% of rare diseases. There are no cures.

This reality is unimaginable for most people. So our rare caregivers, patients and professionals tend to become isolated, treading in deeper and deeper waters, meeting each day with bravery and courage; often left feeling anger and disappointment. At Raregivers we believe in uplifting rare families by caring for the caregivers. Our Health & Wellness Practitioner Network brings a curated group of world-class healers to you and to your community. Through guided online training, YouTube workshops, experiential retreats, crisis support and a multilingual global network, the Raregivers Coalition is connecting hearts daily in a safe, nurturing environment.

### **We commend you. You have taken that first step. You have invested in yourself.**

As you explore the Raregivers™ Emotional Journey Map & Guidebook, you can be confident that the explanations and exercises were developed by a coalition of caring, global experts. Through 287 pieces of published research and deep insights from a leading practitioner, Dr. Laura Riordan, you will find the holistic, person-centered pathways and resources to sustain you when “life gets medical.” These journeys can be long and life-altering; they can churn your heart through cycles of hope and grief. We walk with you.

Sending strength and grace,



*Cristol Barrett O'Loughlin*

**Cristol Barrett O'Loughlin**  
Founder & CEO



*Laura Riordan*

**Laura Riordan, PhD**  
Psychologist & Transformational Coach

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# INTRODUCTION

## Who are the Raregivers™?

1. Those rare individuals who hold both hope and grief in the same human heart.
2. Caregivers, patients, family, friends, teachers, doctors, nurses, medical professionals, genetic counselors, researchers, patient advocates, payers, pharmaceutical employees, academics, gamers, entertainers, technologists, volunteers, donors and all others giving their talents, time and life-force to further and honor chronically medical lives.

## Mental health is as important as physical health

Often Raregivers downplay their emotional, physical, financial and social health and do not receive the support they need to continue caregiving in a sustainable way.

The caregiving journey in rare disease is an ever-changing, dynamic human experience and the emotional experiences and needs of rare caregiving are likely different for everyone. That said, chronic caregiving stress, driven by cycles of hope and grief is a common experience shared by many in our community.



*In a world where the journey with rare disease can feel isolating and overwhelming, Raregivers serves as a beacon of hope and support. This guidebook, along with the Emotional Journey Map, offers a lifeline to those navigating the uncharted waters of rare diseases. This guidebook not only acknowledges the shared experiences of rare caregivers but also provides practical exercises and resources to nurture their well-being along the way. Through exercises like identifying core values and practicing self-care, patients, caregivers and professionals are empowered to navigate their roles with strength and resilience. Raregivers™ Emotional Journey Map is not just a guidebook, but a testament to the strength and unity of the rare caregiving community.”*

**Donny W. Suh, MD, FAAP, MBA, FACS**, Chief and Professor Pediatric Ophthalmology and Strabismus Gavin Herbert Eye Institute (GHEI) & Fellowship Program & Eye Mobile Program University of California at Irvine



**NOTE:** As you work your way through this guidebook, please feel free to begin at the stage at which you most identify at this moment. The stages are often non-linear phases that overlap and repeat. Each stage includes a description of the common experience of the stage, Raregivers Relief Tips and exercises to help ease the journey. The guidebook will provide more skills and resources including a common Raregivers language.



# 1. NOTICING CHANGES

## “Something isn’t Right”

From the appearance of first symptoms to the point of diagnosis can be a long, painful journey for caregivers, patients and professionals supporting the family. Medical journeys often start with a gut feeling — something’s not quite right— that leads to questions. What’s wrong? What’s going on? How do we fix this? Who do we talk to?

## Desire for diagnosis

Many rare diseases are hard to recognize and diagnose. The desire for answers leads raregivers to consult with multiple primary care providers and different kinds of specialists in order to answer the question, “What’s wrong?” Finding answers, or a diagnosis, can take years (on average, 5-7 years) and in the process, raregivers become—need to become—experts in asking the right questions.

## Engage specialists / bureaucracy

Finding reasons for symptoms can lead to a range of exploratory tests, hospitalizations, travel to see disease specialists, surgeries, and even misdiagnosis. Communicating with health professionals can be challenging. Healthcare providers do not always explain why tests are necessary, or provide information about costs, insurance, and how to be a caregiver.

## Lack of disease - specific information

The diagnostic journey (or odyssey) can be circular. Tests might rule out one diagnosis but circle back to the unknown. Partial answers, the uncertainty that comes with waiting for test results, and lack of diagnosis can pull raregivers down the time-consuming and energy-sapping rabbit hole of online research, scouring the internet for symptom- and disease-related information, diagnostic tests, and treatment options.

## Emotions: *Fear, anxiety, shame, “Savior”*

Raregivers say that fear about the future, anxiety about the unknown, and fatigue from navigating health care and insurance are all common experiences in the early stages of noticing changes in their loved ones. Raregivers might also feel shame when they aren't able to find answers or provide solutions.



“*The Raregivers Emotional Journey Map is an important work whose time has come. All too often new Raregivers are unaware of the difficult path they are about to embark upon. This guide will help them start exploring what questions to ask of themselves and their medical team.*”

*J Lawrence Merritt II, MD, attending physician at Seattle Children's Hospital and Associate Professor in the Department of Pediatrics at the University of Washington School of Medicine.*



## Raregiver Support

- **Relief Tip:** Reflect and name your emotions through journaling or a conversation with a trusted resource.
- Access to mental health and wellbeing programs in the early stages of caregiving will create a foundation for building self-care skills
- Visit [Raregivers.global](https://Raregivers.global) to find details on free weekly and monthly emotional support groups for women, men and anyone in the care support system looking for support.



## EXERCISE 1: YOUR CORE VALUES

Getting clear on what matters most at the *Noticing Stage* is very important as raregivers have limited time and resources to find diagnostic answers and solutions. Prioritizing what matters most (which will be unique to each family) will help to streamline decisions and create a platform for moving through the stages of the raregiver journey.

Each of us has our own set of values that, like a moral code, help guide us through our decisions in life. Our values clarify who we are and articulate what we stand for. They help determine our tastes, our way of life, our social, political and religious affiliations. Some of them may be transitory or appropriate for a specific circumstance. Other values are more fixed and stay with us throughout our life, forming the basic elements of how we live and work: these are our core values. To start understanding these core values, we will do a writing exercise to define them and refine them for more clarity.

- Step 1** As you read through the following list of core values, circle or highlight each value that resonates with you. Do not overthink your selection. Circle as many as you wish that feel like your values and add any additional words that come to mind.
- Step 2** Group all similar values together from the list of values you just circled or highlighted. Group them in a way that makes sense to you.
- Step 3** Choose one word within each grouping that represents or best characterizes the theme for the entire group and circle that word. Do not overthink this choice, just circle the word that is the most important for you – there are no right or wrong answers.
- Step 4** Write each of the “most important” values that you circled in step 3 in a list.
- Step 5** Now try to narrow your Core Value list to 5 or less. These are your Core Values!

Abundance	Credibility	Knowledge	Security
Acceptance	Curiosity	Leadership	Self-Awareness
Accomplishment	Daring	Learning	Service
Accountability	Decisiveness Empathy	Love	Simplicity
Achievement	Encouragement	Loyalty	Sincerity
Adventure	Enthusiasm	Mastery	Skillfulness
Advocacy	Environment Equality	Making a Difference	Solitude
Agility	Ethics	Mindfulness	Spirituality
Altruism	Excellence	Moderation	Stability
Ambition	Expressiveness	Modesty	Stewardship
Appreciation	Fairness	Motivation	Strength
Assertiveness	Fashion	Open-Mindedness	Structure
Attractiveness	Family	Optimism	Success
Autonomy	Fidelity	Order	Sustainability
Awareness	Fitness	Organization	Synergy
Balance	Flexibility	Originality	Talent
Being the Best	Friendships	Passion	Teamwork
Benevolence	Freedom	Patience	Thankfulness
Boldness	Fun	Peace	Thoughtfulness
Bravery	Generosity	Perfection	Tolerance
Brilliance	Grace	Performance	Traditionalism
Calmness	Gratitude	Perseverance	Transparency
Candor	Growth	Personal Development	Trustworthiness
Capable	Happiness	Philanthropy	Truth
Caring	Hard Work	Playfulness	Understanding
Charity	Harmony	Popularity	Uniqueness
Cheerfulness	Health	Positivity	Unity
Cleverness	Honesty	Power	Usefulness
Community	Honor	Preparedness	Variety
Commitment	Humility	Proactivity	Versatility
Compassion	Humor	Professionalism	Vigor
Cooperation	Imagination	Punctuality	Vision
Collaboration	Inclusiveness	Quality	Vitality
Competition	Independence	Realism	Warmth
Confidence	Individuality	Recognition	Wealth
Connection	Innovation	Relationships	Well-Being
Consciousness	Inspiration	Reliability	Wisdom
Consistency	Integrity	Resilience	Zest
Contribution	Intelligence	Resourcefulness	
Cordiality	Intuition	Responsibility	
Courage	Joy	Responsiveness	
Craftsmanship	Justice	Risk Taking	
Creativity	Kindness	Safety	

# NOTES



## Discussion Questions

**The following discussion questions, along with the Core Values exercise, are designed to help you reconnect with yourself and find your center again.**

- As you notice changes in your loved one, what changes do you notice in yourself?
- Even as things shift, what remains true about your child or loved one, about you, and about your bond?
- How do you typically cope with uncertainty and is that approach serving you right now?
- What kind of support would make the biggest difference for you in the next few weeks?



## 2. ADJUSTING +/- DIAGNOSIS



### Managing symptoms

Even while raregivers are waiting for test results, coordinating care with healthcare providers, and navigating insurance, they still have to find ways of managing their loved one's symptoms. Raregivers say they spend a lot of time and energy contacting and liaising with providers and specialists, sourcing and acquiring adaptive equipment, and seeking and accessing support services. Raregivers are often doing all of these tasks while also managing the disparate needs of other children and members of the family and planning for their loved one's future care.

### Learning about genetics

Once a diagnosis is made, cascade genetic testing and/or genetic counseling might be offered to other family members. Genetic testing identifies the genetic basis for a rare condition and provides information to help parents and raregivers make healthcare decisions. Raregivers say the learning curve about genetic testing is steep but necessary. However, not all raregiver's journeys have a diagnosis. This uncertainty can be very challenging for raregivers.

## Complex care coordination

Rare diseases are generally complex and require specialist care that involves coordinating input from health professionals across different disciplines, clinics, and hospitals. Few healthcare settings do this care coordination for patients. Depending on location, insurance, and other factors, raregivers often spend considerable time and energy finding a way through the healthcare system, including insurance and financial assistance, and coordinating care across different types of providers. Strain on family budgets may increase during this adjustment phase.

## Lack of treatment

Even when a diagnosis occurs, health professionals do not necessarily know about or are able to share information about rare diseases or their potential treatment options, and not all rare conditions are associated with effective or curative therapies.

## Emotions: *Overwhelm, fatigue, “Hero”*

Recognizing and acknowledging that treatment might not be available or effective while also trying to coordinate care for loved ones places a tremendous burden on raregivers to be a “hero.” But being a hero on a daily basis can easily lead to fatigue and overwhelm.



## Raregiver Support

### ***Let Go of the Hero's Cloak***

- Connection with disease-specific patient organizations can reduce stress by providing verified information on disease management, possible treatments and clinical trials. Utilize our referral partner organizations like NORD (National Organization for Rare Disorders), Global Genes, EURORDIS (European Organization for Rare Diseases), IndoUSrare (India) and RDI (Rare Diseases International).
- Give an Hour's Emotional Life Skills® provides accessible tools to help caregivers track their emotional health, identify signs of emotional suffering, and create healthy self-care habits.
- The National Society of Genetic Counselors (NSGC.org) is a cohort of clinically trained professionals who help navigate the genetic journey including Whole Genome Sequencing (WGS). Connecting with a trained expert can assist understanding the complexity of genetics and how that factors into rare, chronic and complex disease journeys.



## EXERCISE 2: LESSONS FROM YOUR PAST SELF

One of the challenging aspects of becoming a raregiver is that you lose touch with some of the things that you enjoy doing on a regular basis. As you include your “former self” in your caregiving, you will have a fuller experience of you as a rare caregiver.

The purpose of this exercise is two-fold: (1) to reflect on a time when you were at your best and bring those skills and strengths forward and (2) to acknowledge that your reality has changed and that your past self is evolving into a raregiver.

- Step 1**      Make a list of activities or ideals that used to feed your soul, make you smile or feel more alive.
- Step 2**      Find some old photos or create visual representations for each activity/ideal. These can be magazine ads, photos, drawings, art or other memento's you've kept that represent these ideals.
- Step 3**      Schedule time to do one of these activities on a regular basis (daily, weekly, monthly) and set the intention to do it. This can be a ten minute break or a two hour event. After engaging in a beloved activity, you will find yourself back in control, in charge and replenished with energy to continue with whatever the day brings you.
- Step 4**      Bonus work: Have fun creating a vision board of the images you selected. When you are finished, display it where you can see it every day. It will be a reminder of the things that fuel you.

# NOTES



## Discussion Questions

**The following discussion questions, along with Exercise 2: A Visit to Your Past Self, are intended to help you reconnect with your strengths and remember who you are beneath the uncertainty.**

- What does your child or loved one still need from you that has nothing to do with health or diagnosis?
- What assumptions you once held no longer feel true?
- Have there been moments, however brief, that felt grounding or stabilizing?
- What feels hardest to explain to friends or family right now?



EVERY DAY STRESS



## 3. SHIFTING RESPONSIBILITIES

### Role strain/identity change

Family life changes as the need for caregiving increases, especially for women (as mothers, relatives, and partners), who are the dominant raregivers for people with rare diseases. Caregiving is time-intensive and involves adopting multiple roles such as care coordinator, researcher, decision-maker, and advocate. Becoming the main caregiver can place strain on those giving care as they try to balance employment, family life, social life, and caregiving. Becoming a caregiver can also mean losing other meaningful sources of identity and lead to feelings of loneliness and abandonment.

### Learning about the disease

Diagnosis can bring relief, but scouring the internet for symptom and disease-related information, diagnostic tests, and treatment options is a constant for many rare disease raregivers, especially if there are no current treatment options.



**“**When caregiving responsibilities shift over time in the context of rare disease, it’s easy to lose your sense of self. Being in a community with other ‘raregivers’ offers a healing space to acknowledge these shifting responsibilities, practice self-nourishment, and sustain a vibrant sense of self.”

**Alexandra Howson PhD, Principal, Write Medicine and lead Data Analyst/**  
Co-Author of the Raregivers™ Emotional Journey Map

## Becoming the expert/advocate

Raregivers often spend a great deal of time researching their loved one's disease or condition, possible treatments and relevant clinical trials. As such, they become the "experts" in their loved one's rare disease. Raregivers often find themselves educating health professionals about the rare disease or condition.

## Learning new supportive care skills

Many rare conditions require extensive, long-term supportive care and specialist services such as physical therapy, occupational therapy, and speech therapy. Commercial health insurance, Medicare, or Medicaid do not always cover these services. As a result, raregivers say they have to learn new skills (e.g., medication or oxygen administration, tube feeds, manual handling, lifting) and develop supportive care and nursing expertise.

## Emotions: *Resistance to role, "Expert"*

Raregivers say they feel frustrated about having to become experts in their loved one's rare disease. The more that raregivers learn about their loved one's rare disease, the further they might feel from their "old normal" and the "normal" of other people's family lives.



## Raregiver Support

### **CRISIS SUPPORT: NEED HELP NOW?**

- The shifting responsibilities that come with managing disease in yourself, your loved one or as a professional can cause emotional distress and, for some, an existential identity crisis. It is critical that you ask for support, particularly if your thoughts turn to suicide.
- **United States:** For immediate help: Call, text or chat the 988 Suicide & Crisis Lifeline at 988. OR text RARE or RARAS to 741741 for the Crisis Text Line. Both services are free and available 24/7.
- **Europe:** Help lines in different European countries give support and contact information for rare diseases. The help lines offer social, psychological and information solutions. You can find a list of rare disease helplines here: <https://www.eurordis.org/information-support/rare-disease-help-lines/>
- **Global:** The United Nations and World Health Organization prioritize emotional health. For a list of country-specific resources you can utilize this Wikipedia list as a starting point: [https://meta.wikimedia.org/wiki/Mental\\_health\\_resources](https://meta.wikimedia.org/wiki/Mental_health_resources)



### EXERCISE 3: FIND YOUR NATURAL STRENGTHS

After acknowledging that your world has changed dramatically, most raregivers find themselves in a transitional state that offers the opportunity of great creativity, innovation, and renewal. You may find that you are blossoming into a different person with resilience, resources and strength you never knew you had. Who you are and who you are becoming can differ from your pre-raregiver self. This exercise will help you discover yourself as a rare caregiver.

Being able to give care from a place of strength and developing new patterns of behavior that align with natural strengths can help raregivers to maintain a sense of control and connect to their purpose. When a raregiver can define their areas of strength, they will own their tasks and create more flow in their daily lives. The tasks that do not fall into their strengths category need to be reassigned or let go of so that energy is directed to the most important tasks.

**NOTE:** Support systems help at this stage to pick up tasks that are outside of a raregiver's strengths zone and to give relief to raregivers. Emotional and companionship support can help raregivers to feel understood at a point in their caregiving journey when a sense of identity shift feels especially acute.

**Step 1** Think for a moment about what kinds of activities bring you joy. Some of you may love to cuddle and soothe small infants, some may be natural creators, some may be great at organizing, researching and fundraising, and others may focus on nutrition and be excellent cooks. These activities draw upon your natural strengths, what you do effortlessly that flows from within.

**Step 2** Make a list of the activities that you love to do. Here are some common strengths that may help you to identify some of your own: organizing, networking, creative play, strategic thinking and active listening.

**Step 3** Next, you can use some of these natural abilities to engage with your loved one in activities that you enjoy and you feel happy doing. If you like to dance, set up a dance party with some of everyone's favorite songs. If you love crafts, set up a space to be creative together. Even if your loved ones cannot participate fully in the activities you enjoy, they will benefit from being with you as you do them. The important thing is to find activities that you like to do together.

When you are using your natural strengths, you will feel more connected to yourself and your loved ones will feel more connected to you too!

# NOTES



## Discussion Questions

**The following questions are meant to support reflection and help you take stock of where you are now.**

- Is there a skill you've developed that surprised you? Is this something you enjoy or are particularly good at?
- What do you notice in your body or emotions when you think about this phase?
- Where do you feel confident, and where do you still feel lost?
- What does your child or loved one seem to need more of right now: comfort, normalcy, honesty, or distraction?



## 4. FULL-TIME CARE

Ongoing Health & Financial Navigation

### Costs Of Time Off Work/Loss of Income

Many raregivers are in paid employment when they start caregiving. Trying to balance caregiving with paid work or employment looks different for everyone. To find this balance, some raregivers reduce their working hours, seek work with flexible leave, or stop working altogether. These changes in access to work often reduce income, weaken work-placed ties and friendships, and can affect productivity at work. In other cases, raregivers have to increase their working hours to address new financial demands.

### Relationship Impact

Becoming the main caregiver in a family often redefines relationships between children, spouses, and wider family. In addition, many rare diseases are heritable but unpredictable, leading to anxiety about how and when the disease could affect other family members. The demands of caregiving also limit the time and opportunity for leisure, social activities, and contact with friends and relatives, which is likely to increase emotional stress and could lead to further feelings of isolation.

## Need for/seeking social support

Raregivers often provide ongoing care with little or no break. But the enormity of responsibilities as a rare caregiver can feel overwhelming, as can the burden of carrying the feelings and worries of other family members. Social and emotional support can be valuable in helping raregivers to emotionally recharge, build relationships with others, and find validation. Respite care from peers or community/ rare disease organizations can help to restore balance and open opportunities to cultivate leisure and social activities that bring pleasure and increase contact with friends and relatives.

## Emotions: *Isolation, Insecurity, “Advocate”*

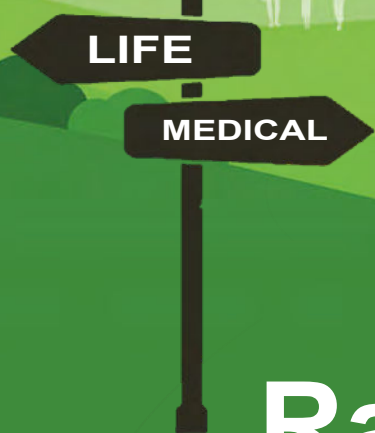
The loss of work and colleagues can have a profound impact on how raregivers see themselves, as well as how others see them. The time, emotions, and energy that caregiving requires can result in depression, social isolation, and relationship strain.



## Raregiver Support

### ***Developing Self-Care Skills***

- Healthcare providers are sources of information about available psychological support.
- Coping and self-care practices such as physical activity, journaling, and yoga have also been shown to have a positive and protective impact on mental and emotional health.
- Raregivers™ support groups are a great place to learn self-care skills. Learn more at [raregivers.global](http://raregivers.global)



# Raregivers™ Emotional Journey Map

Detachable Poster

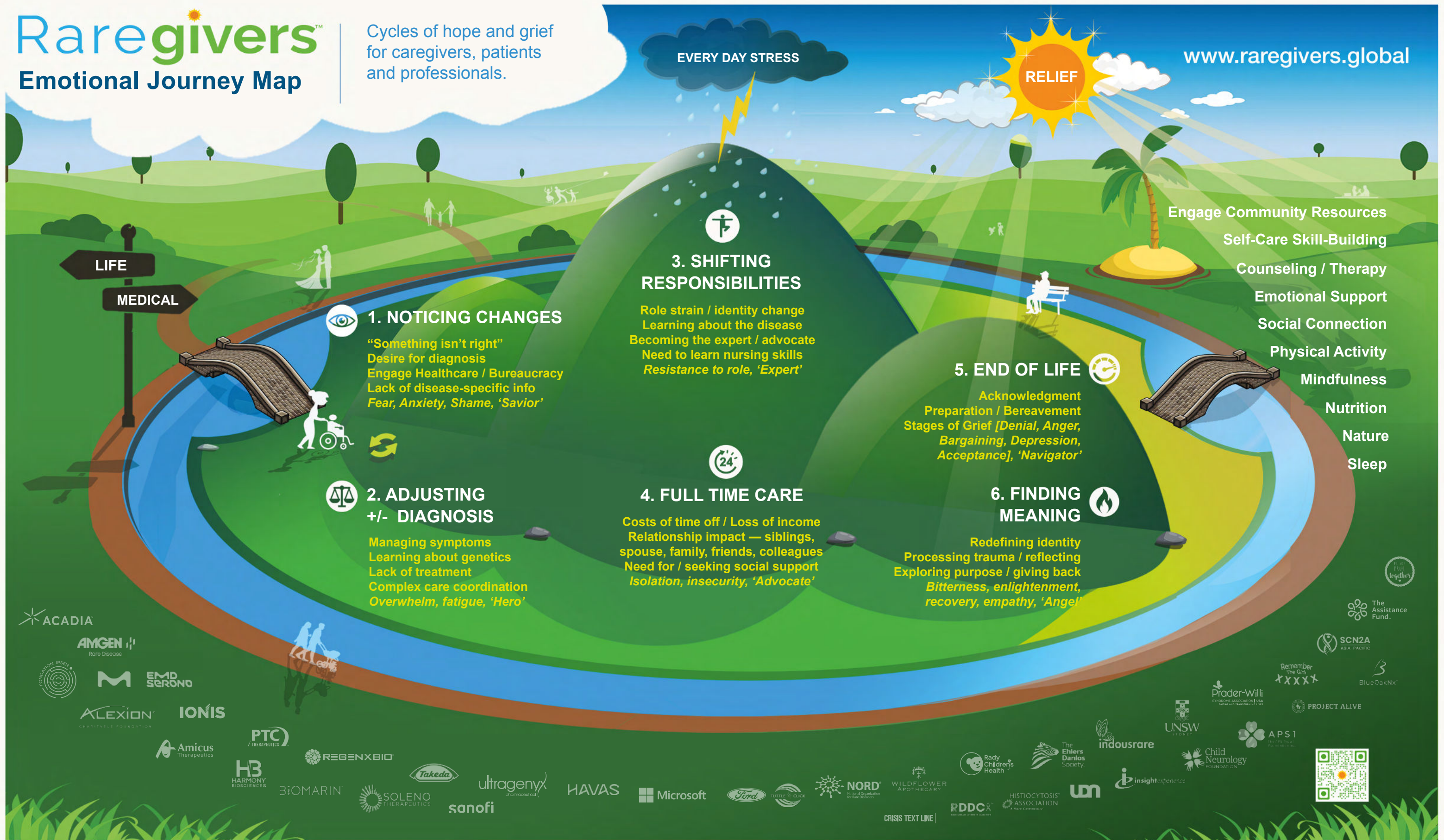


# Raregivers™

## Emotional Journey Map

Cycles of hope and grief for caregivers, patients and professionals.

[www.raregivers.global](http://www.raregivers.global)





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**Raregivers™** | *Ease the journey.*



## EXERCISE 4: THE IMPACT OF RAREGIVING ON YOUR WHEEL OF LIFE

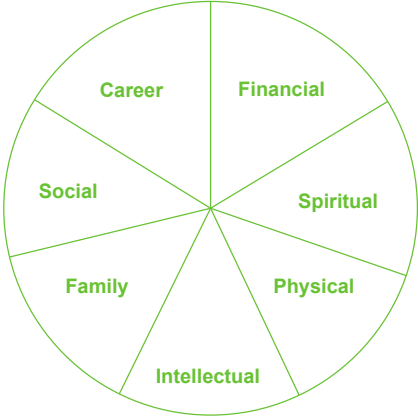
If an individual formerly worked outside the home and now serving as a full caregiver, the shift in identity and social opportunities can feel confusing and isolating. It can help to spread the workload throughout the family system (and beyond) and to help the individual to see where their caregiving demands are most affecting their overall life satisfaction. This next exercise is especially important to take stock of where the caregiving cycle is impacting you most.

This simple writing exercise evaluates how being a rare caregiver most impacts your family system, your work life and your personal well-being.. It can help determine where to ask for help and which resources to prioritize.

**Step 1** Begin by looking at the Wheel of Life diagram below. Ask yourself how satisfied you are in the following seven areas of your life: Career, Financial, Spiritual, Physical, Intellectual, Family, and Social. Rank your satisfaction on a 1-10 scale for each area with 1 being totally unhappy and 10 as highly satisfied.

**Step 2** Now ask yourself how much time and energy you devote to each area. Use the same 1-10 scale to measure your time spent in these areas. 1 is not very much time spent and 10 a lot of time spent. (Be honest with your evaluation and honor that your environment has drastically changed).

**Step 3** Typically, time and satisfaction correlate, but not always when you are a rare caregiver. A raregiver could be spending too much time and energy on family and not enough in other areas, creating an unbalanced wheel. Take note of the places where these two rankings are out of sync and pinpoint where you can use your resources more wisely and where



# NOTES



## Discussion Questions

**Alongside the Wheel of Life exercise, the following discussion questions are designed to help you stay attuned to yourself and your experience during this stage.**

- How can you stay connected to who your child or loved one is, not just what is happening to them?
- What has helped you get through this so far?
- Is there anything about how this experience is affecting you that you wish someone on the medical team would ask but hasn't?
- What roles have you taken on that you never expected?



## 5. END OF LIFE

### Acknowledgement

Caregiving in rare disease involves a delicate balance between trying to create and live a “normal” life and grieving for the life that could have been - for themselves, their loved ones or those they serve professionally. At some point, raregivers must grapple with how to best prepare for the end of life, or how to move forward, leaving behind their dreams of an idyllic life. For some, this involves the experience of bereavement while the loved one being cared for is still living. This experience, or anticipatory grief, can last many years.

### Preparation / Bereavement

Anticipatory grief can be supported through compassionate care, decision support, and counseling, as well as by finding opportunities to discuss the future, death, and dying with others. As a raregiver it's difficult to know what the future will look like after the passing of a loved one to whom intensive care has been given.



**“ I lost three sons Randy (12), David (18) and Jared (19) to Mucopolysaccharidosis or Hunter Syndrome. I was shut down emotionally for many years. These women - Rare Mothers - sharing their stories, feelings, thoughts, concerns, fears, solutions, challenges and hugs brings a degree of joy, solace and peace to me that I have never felt before.”**

**Phyllis Barrett - Rare Mother**, Journalist, Owner/The Bear Facts, Raregivers™  
Board of Directors

## Emotions: Stages of Grief\*, “Navigator”

Raregivers say that it’s easy to become, and be seen as, the Navigator at this stage—the person who is the in-the-moment problem-solver trying to find a way through their own feelings of loss, as well as the feelings of others. We can support your navigation with training and resources.

	Can look like:	Can feel like:
<b>Denial</b>	Avoidance, forgetting, distraction	Shock, numbness, confusion
<b>Anger</b>	Pessimism, cynicism, irritability	Frustration, impatience, rage
<b>Bargaining</b>	Over-thinking, comparing self to others, judgment about self/others	Guilt, shame, blame
<b>Depression</b>	Sleep/appetite changes, reduced energy, reduced social interest	Sadness, despair, hopelessness
<b>Acceptance</b>	Being present in the moment, able to tolerate emotions, adapting	Felling “good enough,” self-compassion, validation
<b>Finding Meaning</b>	Remembering those who have died with more love than pain	Honoring loved ones who have died

\*Kübler-Ross E. *On Death and Dying. What the Dying have to Teach Doctors, Nurses, Clergy, and their Own Families.* New York: Scribner. 2011. Kessler D. *Finding Meaning: The Sixth Stage of Grief.* New York: Scribner. 2019.



## Raregiver Support

### **Practices for Being in the Moment**

- Talking about end-of-life with trusted sources can help caregivers adjust to life without their loved one, consider a different future for themselves, and explore how family roles and routines will change.
- Self-care practices such as journaling can help to process the grieving process before and after death.



## EXERCISE 5: REPLENISH WITH SELF CARE

If you are feeling overwhelmed or very depleted, even thinking of something to do for yourself can seem like another notch on your expanding to-do list. However, you have to start somewhere to break the cycle of stress and depletion. It takes practice to put our own needs first, but as we get stronger, we will have more energy to care for ourselves and for others.

Who knows how to take care of you better than your pre-caregiver self? Take some time to connect with who you were before you became a rare caregiver. What types of activities gave you pleasure, rest, fulfillment or a sense of ease? Ask your former self to make a list of suggested activities to get you back in touch with the things that you enjoy doing.

**Here are some examples of activities pulled together from fellow raregivers:**

- Say “yes” to yourself out loud
- Watch your favorite movie
- Practice yoga
- Read a favorite book
- Walk on the beach
- Play your own music in the car
- Take a 10 minute “time out”
- Go to lunch with a friend
- Go for a solo hike
- Browse through magazines
- Do your meditation practice
- Dance
- Go to a museum or art show
- Take a morning exercise class
- Play a favorite game
- Make yourself a non-kid-friendly meal

# NOTES



## Discussion Questions

**These discussion questions may be especially meaningful to explore with trusted friends, counselors, or spiritual advisors.**

- Are there moments when this feels especially heavy?
- What losses are you grieving, even if they're difficult to name?
- What do you wish others understood about this stage of the journey?
- Which emotions surprise you the most?



## 6. FINDING MEANING

### Processing trauma / reflecting

When caregiving has been at the core of someone's experience and identity, the end of caregiving can trigger not only distress, but also guilt and uncomfortable and unanticipated feelings of relief. Similar to individuals who have lost a loved one to suicide, raregivers might experience a period of prolonged or complicated grief. Raregivers who have lost a child might re-experience feelings of loss and grief for many years after their child's death, especially during times that would have been milestones for their child. It can also be difficult to feel motivated to engage in life or connect with people who have not experienced a similar loss. These feelings are normal.

### Redefining identity

Following the death of a loved one, many raregivers suffer identity loss and find it difficult to adjust to life without their caregiving role, still also living with an acute sense of loss. For some raregivers, grieving involves not only the loss of their loved one, especially a child, but also involves grieving for the life they hoped to live with their loved one. Over time, it is possible for raregivers to feel stronger and develop a new identity. But it can take time to find a new sense of purpose in life, to re-enter the world of paid work, and to make new relationships with people who are not raregivers.

## Exploring purpose / Giving back

It is possible to reconnect with life and reinvent one's sense of purpose and identity following the loss of a loved one with rare disease. It is valuable for raregivers to find and embrace sources of emotional support that affirm their relationship as a raregiver and encourage motivation to continue living. Caregivers of people without rare disease say that providing support to other caregivers and finding ways to honor and keep bonds with their deceased loved ones are helpful ways to explore purpose and give back. Many raregivers who have survived this stage find purpose and meaning in drawing on their lived experience to support research, advocacy, or fundraising, or by mentoring rare families or professionals.

## Emotions: Bitterness, recovery, enlightenment, empathy, 'Angel'

There is potential for raregivers to feel bitter and experience feelings of shame, blame, and isolation following the death of a loved one with a rare disease. As a result, it can be extremely difficult for raregivers to talk about or, at times, even acknowledge their grief and loss. At the same time, family members, friends and acquaintances might be reluctant to talk about death and how the raregiver's life has changed. Once they are able to find meaning, it sows seeds of empathy and makes it possible for heart-to-heart connection with others going through this experience.



*The Survivorship part of the journey - what we call 'Finding Meaning' - can initially be a very complicated mix of grief and relief, accompanied by an overwhelming sense of 'Who am I now?' But it can also become an opportunity to redefine one's identity and to find new purpose in life."*

**Randy Phelps, Ph.D.**, Clinical Psychologist, Rare Disease Family Member and co-founder and Chief Clinical and Science Officer of WeBe Life Inc



## Raregiver Support

Every raregiving emotional journey is unique and the types of support that raregivers need following loss will vary, but might include:

- Opportunities to talk about the experience of loss with other raregivers who can provide empathy and acknowledge their experiences.
- Support to lessen negative feelings and find purpose.
- Space and time to make practical and lifestyle adjustments.



## EXERCISE 6: REMEMBERING WHAT MAKES YOU HAPPY

*For the meaning of life differs from man to man, from day to day and from hour to hour. What matters, therefore, is not the meaning of life in general but rather the specific meaning of a person's life at a given moment. - Viktor E. Frankl*

Transitions never truly end. The end of a physical life is the beginning of a transition to a new world where caregiving may no longer be your primary focus. This is a time where self-care can really make a difference in your wellbeing. Try this simple exercise and discover more about what you enjoy. You will need a piece of paper, a writing instrument and a timer to complete this exercise.

- Step 1** Start by noting how you are feeling right now. Are you excited, depressed, anxious, tired, etc.?
- Step 2** Set the timer for four minutes. Make a list of everything that makes you happy. Write down (as fast as you can) anything that comes to mind without judgment, or correction.
- Step 3** When the timer goes off, stop writing and make a brief note of how you feel. You might be surprised to notice that the simple act of writing down the things that make you happy will elevate your mood.
- Step 4** Keep this sheet as a starting point to finding your own happiness. Enjoying a moment of happiness can provide a bit of respite and over time, perhaps create meaning as well.

# NOTES



## Discussion Questions

**At its core, the Raregivers Emotional Journey Map exists to help hearts find common ground—and to support one another through shared understanding.**

- How has my Raregivers journey changed how I see myself and my closest relationships?
- What parts of myself feel neglected or missing?
- What are the self-care practices that work best for me?
- What feels possible now that didn't before?



# 10 ESSENTIAL TIPS FOR SUSTAINABLE RAREGIVERS™

**1. Take Sleep Seriously:** Sleep deprivation takes a toll on your body and can affect your mood. If you are chronically sleep-deprived, consider how enjoyable and productive your time awake will be. What's the benefit of reducing your sleep just so you can drift through the next day feeling like a zombie? Four hours of uninterrupted sleep is essential and 8 to 9 hours each day is ideal. Schedule your sleeping hours and stick to them. Make sure you are sleeping in a dark room with no electronic equipment, no mobile phones and no TV!

**2. Schedule time in nature:** Sunrises, sunsets, moon cycles, weather patterns, the four seasons ~ nature is an infinite source of energy. Schedule time in nature on your calendar each week and make sure that you have the resources in place to cover you. It takes practice to put our own needs first, but as we get stronger, we will have more energy to care for ourselves and for others.

**3. Focus on your own nutrition:** This is an important daily exercise, especially when time is short. Make sure that you have the proper fuel to burn and reserves for when you need an extra boost. Take charge of what you eat and you will start to notice a difference in how you feel each day. Even simple things like eating less processed food or giving up quick service meals makes a huge difference in overall attitude and energy. If you need additional help in this area, seek out a nutritionist or ask your doctor for guidance. Try to include healthy snacks for yourself, so that you avoid junk food cravings when you're hungry.

**4. Be mindful and allow yourself to receive:** The simple mantra of gratitude is a good start as it places you in the present moment by appreciating all that you have in your life. It is only in the present that we can truly experience happiness and the wonderment of being alive. It takes practice to put our own needs first, but as we get stronger, we will have more energy to care for ourselves and for others.

**5. Get physical:** Take time to do something just for yourself, even if it is only for 10 minutes a day. Light a candle over coffee and practice deep breathing, take a long shower, walk to do that errand. Make sure you schedule the time and set the intention to do it. After doing this, you will find yourself back in control, in charge and replenished with energy to continue with whatever the day brings.

**6. Connect socially with others:** You are part of a global community of 350 million people worldwide! Broadening your outreach beyond a specific disease can connect you with a much broader community of families, advocates and experts in rare, chronic and complex diseases. Consider joining our weekly support groups.

**7. Name your emotions and write them down:** Journaling is a self-reflective practice that allows you to notice your feelings and what is really going on inside of you. Begin by writing for 10 minutes each day and see what appears.

**8. Ask directly for what you need from family, friends and professionals:**

Raregivers are constantly getting less than what they require, because they are too often focused on others and too polite to ask directly for help, including counseling or therapy. Asking for what you need is an act of service - for yourself and those around you - because it allows you to sustain your emotional and physical health.

**9. Use self-care rituals to create structure:** Rituals allow us to pause and be present in a moment that has been planned specifically for a purpose. It is a beautiful way to connect with one another and it is decidedly underused in today's society. Some easy rituals include lighting a candle, taking three deep breaths, walking in nature or enjoying the night sky.

**10. Enlist a predictable flow of community resources that meet your needs:**

Due to the chaotic nature of disease management, it is important to create structure and downtime for yourself by enlisting the help of others that you trust. You can create a schedule with your partner, trade childcare with a friend, hire a qualified caregiver or family members/ neighbors/work colleagues to help. You need to be able to count on these caregivers for a few hours each day, or more if you continue working.

[Raregivers.global](https://raregivers.global)

# EPILOGUE

## Not everyone's journey is the same.

But we do believe that there is a common baseline of chronic caregiving stress and landmark events during the journey that most raregivers share, regardless of the disease. That is why we have created the Raregivers™ Emotional Journey Map.



*If you love and care for someone with a rare disease, you are likely to find yourself in this map and feel a sense of comfort from the simple fact that you are not alone - there is a supportive community of professionals and other Raregivers that can relate to the heartache you've already felt, understand the utter exhaustion you may be currently facing, and offer guidance and resources to navigate the uncertainty of where you're headed next."*

**Jana Benesh PhD**, Neuroscientist, Rare Mother & Clinical Strategy & Patient Advocacy Lead Scientist, Center for Rare Diseases - Scientific Affairs, ICON PLC

Practicing holistic, person-centered self-care as a caregiver, patient or professional can help to avoid depression and other mental health issues. The National Alliance on Mental Illness provides mental health education and access to resources, such as support groups and information about evidence-based psychological therapies including Cognitive Behavioral Therapy and Acceptance and Commitment Therapy.

This Raregivers™ Emotional Journey Map is for raregivers, patients, professionals, and other individuals and organizations involved in supporting raregivers in rare disease. A rare life is a long, life-altering journey that is characterized by cycles of grief and hope. To find more information about holistic, person-centered pathways and support resources please visit Raregivers™ Coalition Global Mental Health & Wellness Program for Rare Disease.

[Raregivers.global](https://raregivers.global)

# THANK YOU

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To all those affected by or working in rare, chronic and complex diseases, we would like to *Thank You* very much for the opportunity to share this important research, website, discussion guide and interactive mental health and wellness tools. Download the poster, discussion guide and explore the interactive version the Raregivers™ Emotional Journey Map at Raregivers.global.

# REFERENCE RESEARCH

**Raregivers™ Emotional Journey Map. Program Strategist: Cristol Barrett O’Loughlin.  
Lead Data Analyst: Alex Howson. Press Release July 12, 2022.**

**<https://www.globenewswire.com/news-release/2022/7/12/2478336/0/en/ANGEL-AID-Unveils-Raregivers-Global-Mental-Health-Initiative-for-Caregivers-Patients-and-Professionals.html>**

1. Antoniadis AM, Galvin M, Heverin M, Hardiman O, Mooney C. Prediction of caregiver quality of life in amyotrophic lateral sclerosis using explainable machine learning. *Sci Rep* 2021; 11(1): 12237.
2. Applebaum AJ, Polacek LC, Walsh L, et al. The unique burden of rare cancer caregiving: caregivers of patients with Erdheim-Chester disease. *Leuk Lymphoma* 2020; 61(6): 1406-17.
3. Baumbusch J, Mayer S, Sloan-Yip I. Alone in a Crowd? Parents of Children with Rare Diseases' Experiences of Navigating the Healthcare System. *Journal of Genetic Counseling* 2019; 28(1): 80-90.
4. Beck AF, Solan LG, Brunswick SA, et al. Socioeconomic status influences the toll pediatric hospitalisations take on families: a qualitative study. *BMJ Qual Saf* 2017; 26(4): 304-11.
5. Boele FW, van Uden-Kraan CF, Hilverda K, et al. Neuro-oncology family caregivers' view on keeping track of care issues using eHealth systems: it's a question of time. *J Neurooncol* 2017; 134(1): 157-67.
6. Boettcher J, Boettcher M, Wiegand-Grefe S, Zapf H. Being the Pillar for Children with Rare Diseases-A Systematic Review on Parental Quality of Life. *Int J Environ Res Public Health* 2021; 18(9).
7. Bryson BA, Bogart KR. Social support, stress, and life satisfaction among adults with rare diseases. *Health Psychol* 2020; 39(10): 912-20.
8. Bygum A, Aygören-Pürsün E, Caballero T, et al. The hereditary angioedema burden of illness study in Europe (HAE-BOIS-Europe): background and methodology. *BMC Dermatol* 2012; 12: 4.
9. Cardinali P, Migliorini L, Rania N. The Caregiving Experiences of Fathers and Mothers of Children With Rare Diseases in Italy: Challenges and Social Support Perceptions. *Front Psychol* 2019; 10: 1780-.
10. Chowdhury SF, Sium SMA, Anwar S. Research and Management of Rare Diseases in the COVID-19 Pandemic Era: Challenges and Countermeasures. *Front Public Health* 2021; 9: 640282.
11. Craig TJ, Banerji A, Riedl MA, et al. Caregivers' role in managing hereditary angioedema and perceptions of treatment-related burden. *Allergy Asthma Proc* 2021; 42(3): S11-s6.
12. Currie G, Szabo J. '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 Expect* 2019; 22(6): 1251-9.
13. Currie G, Szabo J. "It is like a jungle gym, and everything is under construction": The parent's perspective of caring for a child with a rare disease. *Child Care Health Dev* 2019; 45(1): 96-103.
14. Currie G, Szabo J. Social isolation and exclusion: the parents' experience of caring for children with rare neurodevelopmental disorders. *Int J Qual Stud Health Well-being* 2020; 15(1): 1725362.
15. Davidson M, Stevenson M, Hsieh A, Ahmad Z, Crowson C, Witztum JL. The burden of familial chylomicronemia syndrome: interim results from the IN-FOCUS study. *Expert Rev Cardiovasc Ther* 2017; 15(5): 415-23.
16. Delisle VC, Gumuchian ST, Rice DB, et al. Perceived Benefits and Factors that Influence the Ability to Establish and Maintain Patient Support Groups in Rare Diseases: A Scoping Review. *Patient* 2017; 10(3): 283-93.
17. Dellve L, Samuelsson L, Tallborn A, Fasth A, Hallberg LR. Stress and well-being among parents of children with rare diseases: a prospective intervention study. *J Adv Nurs* 2006; 53(4): 392-402.
18. Denton CP, Laird B, Moros L, Luna Flores JL. Things left unsaid: important topics that are not discussed between patients with systemic sclerosis, their carers and their healthcare professionals-a discourse analysis. *Clin Rheumatol* 2021; 40(4): 1399-407.
19. Doyle M. Peer Support and Mentorship in a US Rare Disease Community: Findings from the Cystinosis in Emerging Adulthood Study. *Patient* 2015; 8(1): 65-73.
20. Dufresne H, Hadj-Rabia S, Méni C, Sibaud V, Bodemer C, Taïeb C. Family burden in inherited ichthyosis: creation of a specific questionnaire. *Orphanet J Rare Dis* 2013; 8: 28.

21. Dufresne H, Hadj-Rabia S, Taieb C, Bodemer C. Development and validation of an epidermolysis bullosa family/parental burden score. *Br J Dermatol* 2015; 173(6): 1405-10.
22. Edgley A, Sykorova M, Stasi E, et al. "I Cry. I Simply Cry." An Ethnography of a Lymphedema Summer Camp. *Lymphat Res Biol* 2021; 19(5): 479-87.
23. Ford L, Rudge P, Robinson K, Collinge J, Gorham M, Mead S. The most problematic symptoms of prion disease - an analysis of carer experiences. *Int Psychogeriatr* 2019; 31(8): 1181-90.
24. Germeni E, Vallini I, Bianchetti MG, Schulz PJ. Reconstructing normality following the diagnosis of a childhood chronic disease: does "rare" make a difference? *Eur J Pediatr* 2018; 177(4): 489-95.
25. Godfrey EM, Kazmerski TM, Brown G, et al. Educational Needs and Preferences for Patient-Centered Outcomes Research in the Cystic Fibrosis Community: Mixed Methods Study. *JMIR Form Res* 2021; 5(3): e24302.
26. Gómez-Zúñiga B, Pulido R, Pousada M, Armayones M. The Role of Parent/Caregiver with Children Affected by Rare Diseases: Navigating between Love and Fear. *Int J Environ Res Public Health* 2021; 18(7).
27. Granero-Molina J, Sánchez-Hernández F, Fernández-Sola C, Jiménez-Lasserotte MDM, Antequera-Raynal LH, Hernández-Padilla JM. The Diagnosis of Hereditary Angioedema: Family Caregivers' Experiences. *Clin Nurs Res* 2020; 29(2): 117-26.
28. Grant N, Von Handorf R, Karaa A, Skotko BG. The experiences and support needs of siblings of people with mucopolysaccharidosis. *Am J Med Genet A* 2021; 185(11): 3418-26.
29. Grimstvedt TN, Miller JU, van Walsem MR, Feragen KJB. Speech and language difficulties in Huntington's disease: A qualitative study of patients' and professional caregivers' experiences. *Int J Lang Commun Disord* 2021; 56(2): 330-45.
30. Guilabert M, Martínez-García A, Sala-González M, Solas O, Mira JJ. Results of a Patient Reported Experience Measure (PREM) to measure the rare disease patients and caregivers experience: a Spanish cross-sectional study. *Orphanet J Rare Dis* 2021; 16(1): 67.
31. Holm KG, Neville AJ, Pierini A, et al. The Voice of Parents of Children With a Congenital Anomaly - A EUROlinkCAT Study. *Front Pediatr* 2021; 9: 654883.
32. Karantzoulis S, Heuer K, Sparling N, Teynor M. The patient experience of Wilson disease: a conceptual model based on qualitative research. *Orphanet J Rare Dis* 2021; 16(1): 437.
33. Kasparian NA, Rutstein A, Sansom-Daly UM, et al. Through the looking glass: an exploratory study of the lived experiences and unmet needs of families affected by Von Hippel-Lindau disease. *Eur J Hum Genet* 2015; 23(1): 34-40.
34. Kjoelaas S, Jensen TK, Feragen KB. 'I knew it wasn't normal, I just didn't know what to do about it': adversity and caregiver support when growing up in a family with Huntington's disease. *Psychol Health* 2022; 37(2): 211-29.
35. Lauder B, Sinclair PM, Maguire J. Mothers' experience of caring for a child with early onset scoliosis: A qualitative descriptive study. *J Clin Nurs* 2018; 27(7-8): e1549-e60.
36. Lima AGT, Sales C, Serafim WFL. Burden, depression and anxiety in primary caregivers of children and adolescents in renal replacement therapy. *J Bras Nefrol* 2019; 41(3): 356-63.
37. Litzkendorf S, Frank M, Babac A, et al. Use and importance of different information sources among patients with rare diseases and their relatives over time: a qualitative study. *BMC Public Health* 2020; 20(1): 860.
38. Lopes MT, Koch VH, Sarrubbi-Junior V, Gallo PR, Carneiro-Sampaio M. Difficulties in the diagnosis and treatment of rare diseases according to the perceptions of patients, relatives and health care professionals. *Clinics (Sao Paulo)* 2018; 73: e68.
39. Lyn N, Pulikottil-Jacob R, Rochmann C, et al. Patient and caregiver perspectives on burden of disease manifestations in late-onset Tay-Sachs and Sandhoff diseases. *Orphanet J Rare Dis* 2020; 15(1): 92.
40. Magliano L, Obici L, Sforzini C, et al. Psychosocial burden and professional and social support in patients with hereditary transthyretin amyloidosis (ATTRv) and their relatives in Italy. *Orphanet J Rare Dis* 2021; 16(1): 163.
41. Magliano L, Patalano M, Sagliocchi A, et al. "I have got something positive out of this situation": psychological benefits of caregiving in relatives of young people with muscular dystrophy. *J Neurol* 2014; 261(1): 188-95.
42. Maksymowicz S, Libura M, Malarkiewicz P. Overcoming therapeutic nihilism. Breaking bad news of amyotrophic lateral sclerosis-a patient-centred perspective in rare diseases. *Neurol Sci* 2022.

43. McMullan J, Crowe AL, Bailie C, et al. Improvements needed to support people living and working with a rare disease in Northern Ireland: current rare disease support perceived as inadequate. *Orphanet J Rare Dis* 2020; 15(1): 315.
44. McMullan J, Crowe AL, Downes K, McAnaney H, McKnight AJ. Carer reported experiences: Supporting someone with a rare disease. *Health Soc Care Community* 2021.
45. Mengel E, Patterson MC, Chladek M, et al. Impacts and Burden of Niemann pick Type-C: a patient and caregiver perspective. *Orphanet J Rare Dis* 2021; 16(1): 493.
46. Mooney J, Graham K, Watts RA. Impact of caring for someone with a rare rheumatic condition, views from patients and informal carers-the need for cat-like vigilance. *Rheumatol Adv Pract* 2019; 3(1): rkz003.
47. Moretti A, Cianci P, De Paoli A, et al. Burden of care in families of patients with rare genetic diseases: analysis of a large Italian cohort. *Eur J Med Genet* 2021; 64(7): 104230.
48. Palacios-Ceña D, Famoso-Pérez P, Salom-Moreno J, et al. "Living an Obstacle Course": A Qualitative Study Examining the Experiences of Caregivers of Children with Rett Syndrome. *Int J Environ Res Public Health* 2018; 16(1).
49. Pasquini TLS, Goff SL, Whitehill JM. Navigating the U.S. health insurance landscape for children with rare diseases: a qualitative study of parents' experiences. *Orphanet J Rare Dis* 2021; 16(1): 313.
50. Pelentsov LJ, Fielder AL, Laws TA, Esterman AJ. The supportive care needs of parents with a child with a rare disease: results of an online survey. *BMC Family Practice* 2016; 17(1): 88.
51. Pokrzywinski R, Hareendran A, Nalysnyk L, et al. Impact and burden of acid sphingomyelinase deficiency from a patient and caregiver perspective. *Sci Rep* 2021; 11(1): 20972.
52. Pompilus F, Ciesluk A, Marquis P, Griebisch I, Voorhaar M. Understanding the Patient Experience in NUT Carcinoma: Qualitative Interviews with Patients and Caregivers to Develop a Conceptual Framework. *Oncol Ther* 2021; 9(2): 591-605.
53. Porter KA, O'Neill C, Drake E, et al. Parent Experiences of Sanfilippo Syndrome Impact and Unmet Treatment Needs: A Qualitative Assessment. *Neurol Ther* 2021; 10(1): 197-212.
54. Prather AA, Epel ES, Portela Parra E, et al. Associations between chronic caregiving stress and T cell markers implicated in immunosenescence. *Brain Behav Immun* 2018; 73: 546-9.
55. Rice DB, Cañedo-Ayala M, Carboni-Jiménez A, et al. Challenges and support service preferences of informal caregivers of people with systemic sclerosis: a cross-sectional survey. *Disability and Rehabilitation* 2020; 42(16): 2304-10.
56. Siddiq S, Wilson BJ, Graham ID, et al. Experiences of caregivers of children with inherited metabolic diseases: a qualitative study. *Orphanet J Rare Dis* 2016; 11(1): 168.
57. Simpson A, Bloom L, Fulop NJ, et al. How are patients with rare diseases and their carers in the UK impacted by the way care is coordinated? An exploratory qualitative interview study. *Orphanet J Rare Dis* 2021; 16(1): 76.
58. Smith J, Damm K, Hover G, Chien J. Lessons from an Experiential Approach to Patient Community Engagement in Rare Disease. *Clin Ther* 2021; 43(2): 421-9.
59. Spillmann RC, McConkie-Rosell A, Pena L, et al. A window into living with an undiagnosed disease: illness narratives from the Undiagnosed Diseases Network. *Orphanet J Rare Dis* 2017; 12(1): 71.
60. Theodore-Oklotka C, Hartman DS, Hoffman DL, Björnsson HT. A Qualitative Study to Characterize the Humanistic Burden of Kabuki Syndrome in the United States and Canada. *Adv Ther* 2022; 39(1): 619-31.
61. Verrecchia E, Sicignano LL, Massaro MG, et al. Caregivers' and Physicians' Perspectives on Alpha-Mannosidosis: A Report from Italy. *Adv Ther* 2021; 38(1): 1-10.
62. Waldboth V, Patch C, Mahrer-Imhof R, Metcalfe A. Living a normal life in an extraordinary way: A systematic review investigating experiences of families of young people's transition into adulthood when affected by a genetic and chronic childhood condition. *Int J Nurs Stud* 2016; 62: 44-59.
63. Weaver R, O'Connor M, Carey Smith R, Sheppard D, Halkett GKB. "We're on a Merry-Go-Round": Reflections of Patients and Carers after Completing Treatment for Sarcoma. *Curr Oncol* 2021; 28(4): 3003-14.
64. Wu YH, Sun FK, Lee PY. Family caregivers' lived experiences of caring for epidermolysis bullosa patients: A phenomenological study. *J Clin Nurs* 2020; 29(9-10): 1552-60.
65. Yoo J, Halley MC, Lown EA, et al. Supporting caregivers during hematopoietic cell transplantation for children with primary immunodeficiency disorders. *J Allergy Clin Immunol* 2019; 143(6): 2271-8.

## Independent Review Board (IRB), Ethics Review & University of New South Wales (UNSW) Data Study References

1. Lingam, R. Palmer, EE. Roberts, N. Beavis, E. McIntosh, R. Barrett O'Loughlin, C. Loden, M. Bullock, S. Kahn, JR. Robertson, EG. Murphy, JB. Pierce, K. (2024) Raregivers™ Wellness Retreat for Australia Caregivers: Wellbeing, Quality of Life & Social Inclusion Study. University of NSW (UNSW) Sydney Human Research and Ethics Committee: number HC220624.
2. Butler J & Kern M L (2015). The PERMA-Profler: A brief multidimensional measure of flourishing.
3. Wilson C, Secker J. Validation of the Social Inclusion Scale with students. *Social Inclusion*. 2015;3(4):52–62. doi:10.17645/si.v3i4.121.
4. Palmer, EE, Sachdev R, Kandula T, Macintosh R, Kirk E, & Bye . (2017). Genetics of Epileptic Encephalopathies. In eLS: John Wiley & Sons, Ltd.
5. Gessler S, Low J, Daniells E, Williams R, Brough V, Tookman A, Jones L. (2008). Screening for distress in cancer patients: is the distress thermometer a valid measure in the UK and does it measure change over time? A prospective validation study. *Psycho-oncology*, 17(6), 538–547. <https://doi.org/10.1002/pon.1273>
6. Stüntzner-Gibson D, Koren PE, DeChillo N (1995). The Youth Satisfaction Questionnaire: What kids think of services. *Families in Society*, 76(10), 616–624.
7. Scheffer IE, Berkovic S, Capovilla G, Connolly MB, French J, Guilhoto L, Hirsch E, Jain S, Mathern GW, Moshé SL and Nordli DR, 2017. ILAE classification of the epilepsies: position paper of the ILAE Commission for Classification and Terminology. *Epilepsia*, 58(4), pp.512-521.
8. Khan S, Al Baradie R. Epileptic encephalopathies: an overview. *J Epilepsy Res Treat* 2012;2012: 403592
9. Nevin SM, Wakefield CE, Schilstra CE, McGill BC, Bye A, Palmer EE. The information needs of parents of children with early-onset epilepsy: A systematic review. *Epilepsy Behav*. 2020 Nov;112:107382. doi: 10.1016/j.yebeh.2020.107382. Epub 2020 Aug 24. PMID: 32854014.
10. Rodenburg R, Meijer AM, DekovićM, Aldenkamp AP. Parents of children with enduring epilepsy: predictors of parenting stress and parenting. *EpilepsyBehav* 2007;11(2):197–20.
11. Jakobsen AV, Møller RS, Nikanorova M, Elklit A. The impact of severe pediatric epilepsy on experienced stress and psychopathology in parents. *Epilepsy Behav* 2020;113:107538
12. Reilly C, Atkinson P, Memon A, Jones C, Dabydeen L, Das KB, Gillberg C, Neville BGR, Scott RC. Symptoms of depression, anxiety, and stress in parents of young children with epilepsy: A case controlled population-based study. *Epilepsy Behav*. 2018 Mar;80:177-183. doi: 10.1016/j.yebeh.2017.12.020. Epub 2018 Feb 3. PMID: 29414549.
13. Mafutha, NN, Abeasi DA, Suglo J. Well-being focused interventions for caregivers of children with developmental disabilities-a scoping review protocol. medRxiv 2022.06.29.22277042; doi: <https://doi.org/10.1101/2022.06.29.22277042>
14. McKeon G, Palmer EE, Macintosh R, Nevin SM, Wheatley L, Rosenbaum S. Feasibility of a mental health informed physical activity intervention for the carers of children with developmental and epileptic encephalopathy. *Epilepsy Behav*. 2021 Aug;121(Pt A):108022. doi: 10.1016/j.yebeh.2021.108022. Epub 2021 May 18. PMID: 34020203.

## Raregivers, Inc. (former DBAs: ANGEL AID, Angel Aid Cares, A Nonprofit Group Enriching Lives)

1. Miodragovic J, Ghidinelli J, Jang Y. Needs Assessment for the Rare Disease Community [Internet]. Indiana: Indiana University School of Social Work; 2019 [cited 2022 Jul 26]. Available from: [angelaidcares.org/s/Indiana-University-ANGEL-AID-Needs-Assessment-for-the-Rare-Disease-Community-April-2019.pdf](https://angelaidcares.org/s/Indiana-University-ANGEL-AID-Needs-Assessment-for-the-Rare-Disease-Community-April-2019.pdf)
2. Riordan L, O'Loughlin C. The Sustainable Mom: Guidebook for Rare Mothers. Building a Stronger Family through Self-Care. [Released 2020] The Sustainable Caregiver: A Guide for Rare Families. [Released 2022] Raregivers™ Emotional Journey Map: A Guide to Sustainable Self-Care [Released 2024].
3. Cristol Barrett O'Loughlin, Alexandra Howson PhD, J. Lawrence Merritt II, MD, Grace Whiting, Alison Skrinar PhD, Randy Phelps PhD, Sandra Mulhinch MCs, PhD, Reena Kartha, PhD, Sharon King, Jessica Shiles, Taylor Kane, Nisha Venugopal, Geoffrey Frankel, Laura Riordan PhD, Elissa Epel PhD, Paige Rivard . Raregivers™ Emotional Journey Map & Discussion Guide [Internet]. Rare Caregivers Coalition; 2022, 2023, 2024.
4. Pierce K, Murphy JB, Robertson EG, KhanJR, Bullock S, Barrett O'Loughlin C, Loden M, McIntosh R, Beavis E, Roberts N, Palmer EE, Lingam R. "Just realising that I wasn't alone... was profound": A mixed-1 methods evaluation of a pilot peer-to-peer wellbeing program for 25 carers of children with rare epilepsies. Published: 21 October 2025. Volume 20, article number 524, (2025). <https://link.springer.com/article/10.1186/s13023-025-04036-0>

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