Mental Wellness

A Guide for People Living With Rare Diseases

The Unique Challenges to Mental Health When Living With Rare Diseases

Our society is witnessing an increase in problems related to **mental health**.[1,2] Stress of many kinds, such as heightened economic uncertainty and concerns about the future, has created a worldwide mental health crisis.[2,3] Depression, anxiety and other psychological disorders are on the rise.[4]

People living with a rare disease face the same challenges to mental health experienced across society. They also face the challenge of maintaining **mental wellness** while coping with their physical disease.[5-7]

It is important to seek help. The road to better mental health has two parts:

Awareness—understanding how a rare disease can affect mental health, and vice versa

Action—finding and using the mental health resources available to the rare disease community

Getting to mental wellness is a rewarding experience for people living with rare diseases, their families and friends.[2]

Amicus Therapeutics®

Developed by Amicus Therapeutics as part of our ongoing commitment to the patient community to enable education in health care and research.

Awareness—Rare Disease and Mental Health

When living with a rare disease, the goal is not merely surviving, but thriving.[8] Although each individual thrives in a unique way, based on individual strengths and limitations, there is growing awareness that all need mental wellness to thrive.[7,9,10]

People living with rare diseases learn to cope with many aspects of their disease, such as physical changes, the process of testing and treatment and special needs in their work, school and home environments.[7,10-13] But the challenges of living with a rare disease produce many points of stress[5]—as shown on the right-hand page.

FACTS TO KNOW

In People Living With Rare Disease...[6]

39.3%
will have **major depression** in their lifetimes

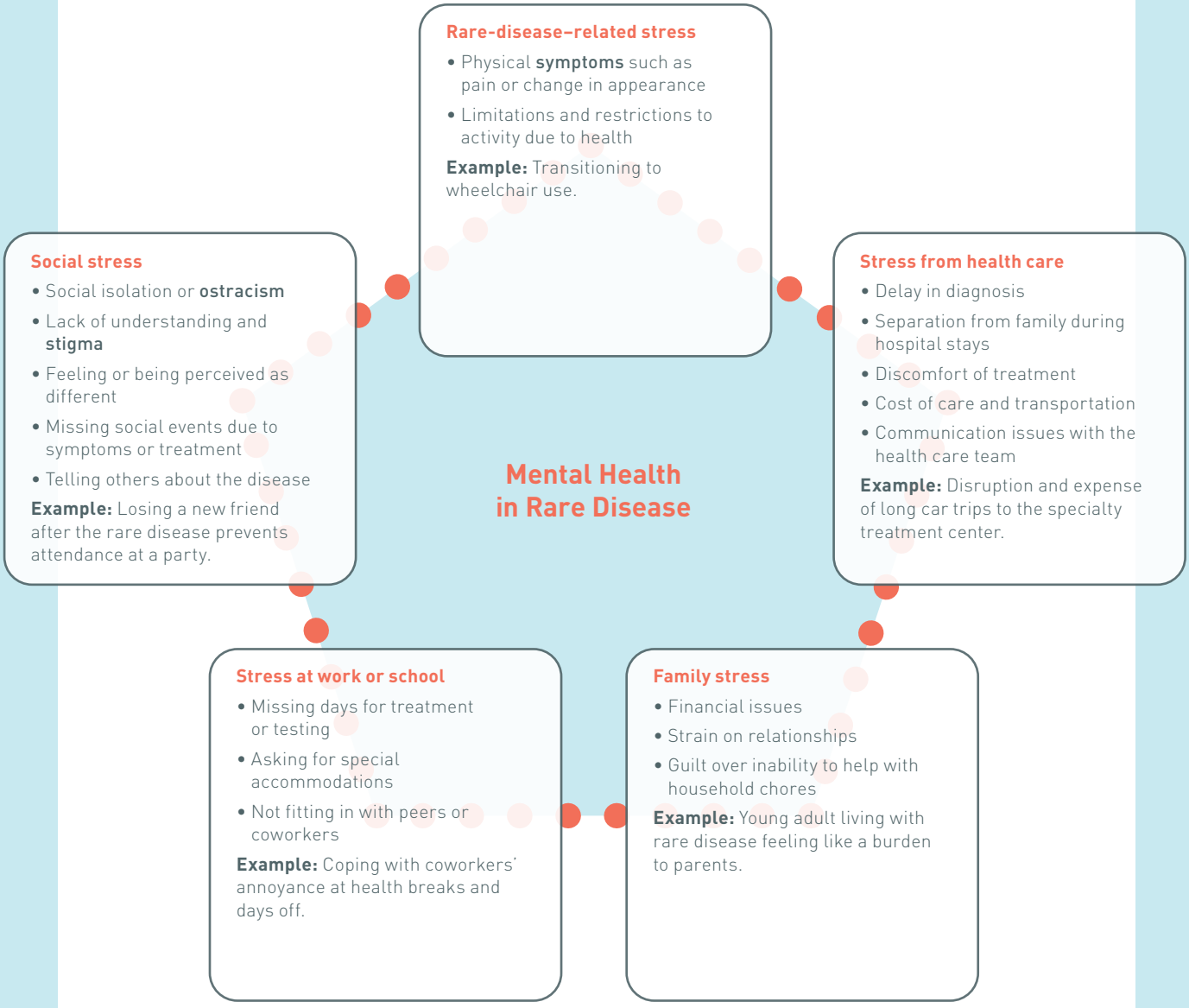
46.1%
will have a **mood disorder** in their lifetimes

44.2%
will have an **anxiety disorder** in their lifetimes

Major depression is a disorder that causes a persistent feeling of sadness and loss of interest. Mood disorders, also called affective disorders, include depression and bipolar disorder, in which symptoms of depression alternate with impulsiveness and other signs of elevated mood and energy. Anxiety disorders involve repeated episodes of intense fear, worry, or sense of danger.

Points of Stress That Affect Mental Health

People living with a rare disease experience many unique stressors.[5,7-17] The **stressors** can diminish **mental resilience** and increase a person's risk of depression or anxiety.[6,11-14,17]



The Impact of Mental Health Problems

What the Research Shows

Recent research has shown that people living with rare diseases have high rates of depression, strong fears (called **phobias**) and anxiety.[6,7,10,17] They may also experience complex post-traumatic stress disorder (**c-PTSD**) related to the ongoing stress of having a rare disease.[18]

Of course, people living with rare diseases also confront the same risks to their mental health as people without a rare disease. It is important to note that the risk of developing mental health issues may be especially great in teenagers, so that teens living with a rare disease are especially vulnerable.[5, 34, 35]

Mind and Body Work Together in Complex Ways

Just as the symptoms of a rare disease can affect mental health, mental health can affect the symptoms of a rare disease. Depression, for example, affects many body **systems**, as shown on the facing page.[19] Physical symptoms, such as aches and pains, are common in depression. Many people with depression who seek treatment in primary care report only physical symptoms.[20] The figure on page 5 shows how a rare disease can affect mind and body.

How Other People May Be Affected

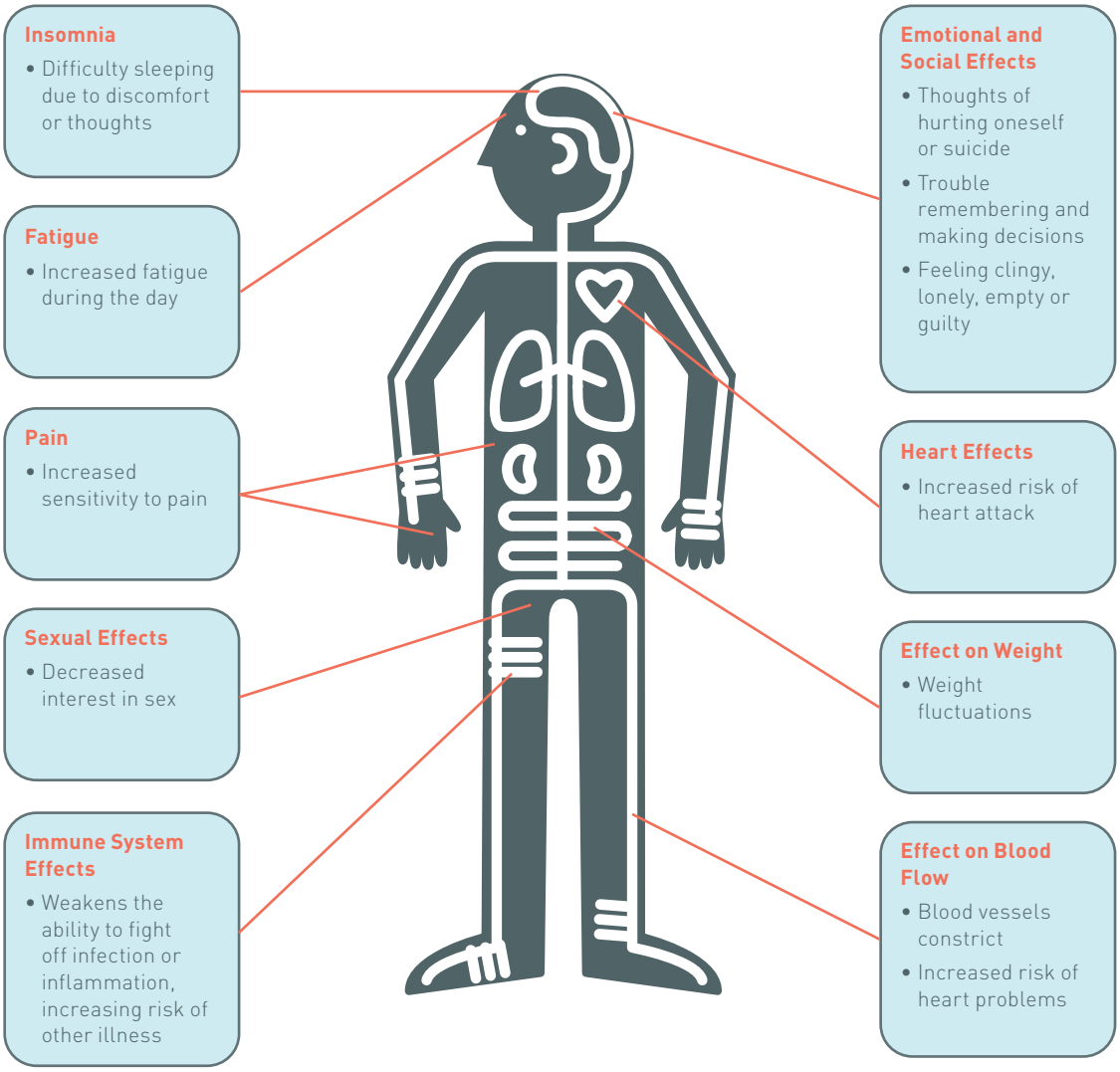
When people living with a rare disease experience mental health challenges, those around them may try to support their loved one's mental health journey. These supports may include parents, caregivers, siblings, grandparents and friends. In giving support, these people may feel stress in relating to their loved one.[8,21-23]

Examples of How Rare Diseases Affect Mental Health

- People living with **Pompe disease** have reported that pain, physical changes, medical tests and treatment and restrictions to their daily activities can affect their mental health,[15,16] causing fear of being judged, depression and anxiety.[15]
- People living with **Fabry disease** have reported disease-related stressors linked to mental health, such as pain, economic problems, relationship difficulties and, due to symptoms such as inability to sweat or fatigue, limitations on recreational activities.[10,11,14,17]

  The changes caused by Fabry disease are often invisible to others, and this can affect mental health.[24,25] When a disease is invisible, friends, family and even health professionals may not believe the disease is serious. This may cause the person living with the disease to feel hopeless and depressed.[25]

Mind-Body Impact of Depression[19]



Action—Getting Support to Achieve Mental Wellness

By partnering with their health care teams, people living with rare disease, their families and friends can find mental health support through counseling, advocacy groups, and the local and regional rare disease community.[26,27]

This support has never been more available than it is today. In the past, the concept of wellness, including mental wellness, was not part of society's definition of health.[7] But today, views have shifted and society now accepts that:

- People seek to be supported, restored and empowered by effective mental health care[3]
- Good mental health can have a positive effect on physical health and self-management of disease[28]
- Mental health care supports all five aspects of health and wellness: physical, emotional, social, spiritual and intellectual

FACTS TO KNOW

Reaching Out for Help

Because society recognizes the importance of mental wellness, there are now many kinds of support services and resources available. Finding this help starts by reaching out to the health care team. Physicians, nurses and other team members, in tandem with medical and community organizations, can recommend professionals and groups that can provide services.[26,27] Seek out **allies**—the professionals and community members who listen, validate concerns and continue to learn about resources for mental wellness.[30]

Professionals who support mental wellness include a range of therapists, such as **counselors, social workers, psychotherapists** and **psychiatrists**. These professionals may be part of community health and social service agencies or medical practices, or may be independent practitioners. The services they offer are shown on the next page of this brochure. When setting up appointments with these professionals, it is helpful to be patient. The need for mental health care is very great, and there may be a short (not long) wait for services. Use **self-care** tools to help in the meantime. There are also many local mental health hotlines and, in the United States, a national mental health help line: 1-800-662-HELP (4357) and online support at https://findtreatment.samhsa.gov/.

> **"Counseling has made such a difference in learning to cope with chronic illness, family and work-related issues...learning comfortable coping tools has been the key to self-help."**[10]
>
> —A person living with Fabry disease

Types of Mental Health Support[10,11,31-33]

Support Groups and Meetings

Group and individual meetings to provide social support and reduce isolation

In-Person, Telehealth or Online Counseling

Regular check-ins with a mental health counselor or social worker to help with stress and emotions

Care from a Psychotherapist

- A therapy called **CBT (cognitive behavioral therapy)** is often suggested when physical health affects mental health
- There are many other types of therapy, including **PDT (psychodynamic therapy), humanistic therapy and holistic therapy**

Care from a Psychiatrist

Appointments to assess mental health and plan treatment

Self-Care

Tools and activities that support mental wellness, such as music therapy or meditation

Resources

Information and support for mental wellness may be available from the health care team, online, and through advocacy organizations and public health agencies. Information abounds during mental health awareness months that are held in many countries. In the United States, the month of May is National Mental Health Month. Many communities and health care centers have in-person and online events during this month.

Many organizations support mental health needs of people living with rare diseases, including some Fabry disease and Pompe disease advocacy organizations, and some have resources that can help anyone living with any rare disease find mental health support.

National Organization for Rare Disorders
https://rarediseases.org/
1-800-999-6673

Global Genes
https://globalgenes.org/
949-248-RARE (7273)

EveryLife Foundation
https://everylifefoundation.org/
202-697-RARE(7273)

Lysosomal Disease Network
https://lysosomaldiseasenetwork.org/

National Alliance on Mental Illness
https://nami.org/Home
1-800-950-6264

EURORDIS – Rare Diseases Europe
http://www.eurordis.org

Maintain mental wellness. Become aware of mental health issues. Take action to get support. These simple steps open the way to living well with rare disease.

What Do These Words Mean?

allies: health care professionals and people in the community who support a person living with a rare disease. Allies listen, validate concerns and continue to learn so they can be of help.

anxiety disorders: involve repeated episodes of intense fear, worry or sense of danger.

CBT: cognitive behavioral therapy, a form of mental health therapy that works to change negative patterns of thinking and behavior.

c-PTSD: complex posttraumatic stress disorder, a mental health problem caused by repeated, multiple experiences of trauma. A person living with a rare disease can have repeated trauma from pain, the uncertainty of the course of the disease, medical treatment, ostracism and many other events. The symptoms of c-PTSD may include feelings of hopelessness, anger or anxiety.

counselor: a professional who listens and advises on mental health, such as people with degrees in counseling or clergy people.

depression: a disorder that causes a persistent feeling of sadness and loss of interest.

Fabry disease: a rare genetic disorder caused by a variant of a gene (called the *GLA* gene). This gene variant can be passed down by either parent. Fabry disease affects many systems of the body, including the eyes, kidneys, skin, nervous system, heart and digestive system.

holistic therapy: mental health therapy that combines approaches from multiple kinds of therapy, tailored to the needs of the individual.

humanistic therapy: a form of mental health therapy that focuses on making rational choices, achieving maximum potential and expressing concern and respect for others.

mental health: a person's overall emotional and psychological condition.

mental resilience: a person's ability to adjust or recover from mental health stressors.

mental wellness: a state of emotional and psychological well-being, resilience and realization of one's own abilities.

mood disorders: include depression and bipolar disorder, in which symptoms of depression alternate with impulsiveness and other signs of elevated mood and energy.

ostracism: exclusion from a social group.

PDT: psychodynamic therapy, a form of mental health therapy that seeks to discover and talk about deep-seated feelings, memories or childhood experiences, as a way to change life in the present.

phobia: an overwhelming fear of an object or living thing, place or situation. Examples of phobias include fear of heights (acrophobia) and fear of being in closed spaces (claustrophobia).

Pompe disease: a rare genetic disorder that causes loss of an enzyme, called GAA. It is inherited if both parents have the gene variant that codes for the disease. Pompe disease affects muscle throughout the whole body, not just the muscles that help people move, but also in the heart, the lungs and the digestive system.

psychiatrist: a medical doctor who specializes in mental health treatment.

psychologist: a trained professional who treats a person's mental health issues by regular, structured sessions of listening and talking.

PTSD: see c-PTSD.

self-care: activities people can do themselves as they work toward mental wellness, such as meditation.

social worker: a professional trained to help with social and emotional problems to improve a person's well-being.

stigma: belief that a disease, condition, or way of life is shameful and should be avoided.

stressor: something that causes stress, strain or tension.

symptom: subjective evidence of a disease or condition that can be recognized by the person living with a disease.

system: a grouping of organs and structures in the body that work together.

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