YOUR GUIDE TO
TYPE 1 GAUCHER DISEASE

Being diagnosed with Type 1 Gaucher Disease begins a journey down the twisting, turning path of rare diseases. It's natural for patients, parents and caregivers to be fearful and filled with questions about what to expect, how to manage symptoms and complications, where to find the best possible care, and perhaps, most importantly, how to live the best life possible with this disease. In *Your Guide to Type 1 Gaucher Disease*, you'll find answers to many of your questions, as well as tips for communicating with others, staying organized, and navigating the often-confusing world of health insurance. When facing a rare, chronic disease, knowledge truly is power.

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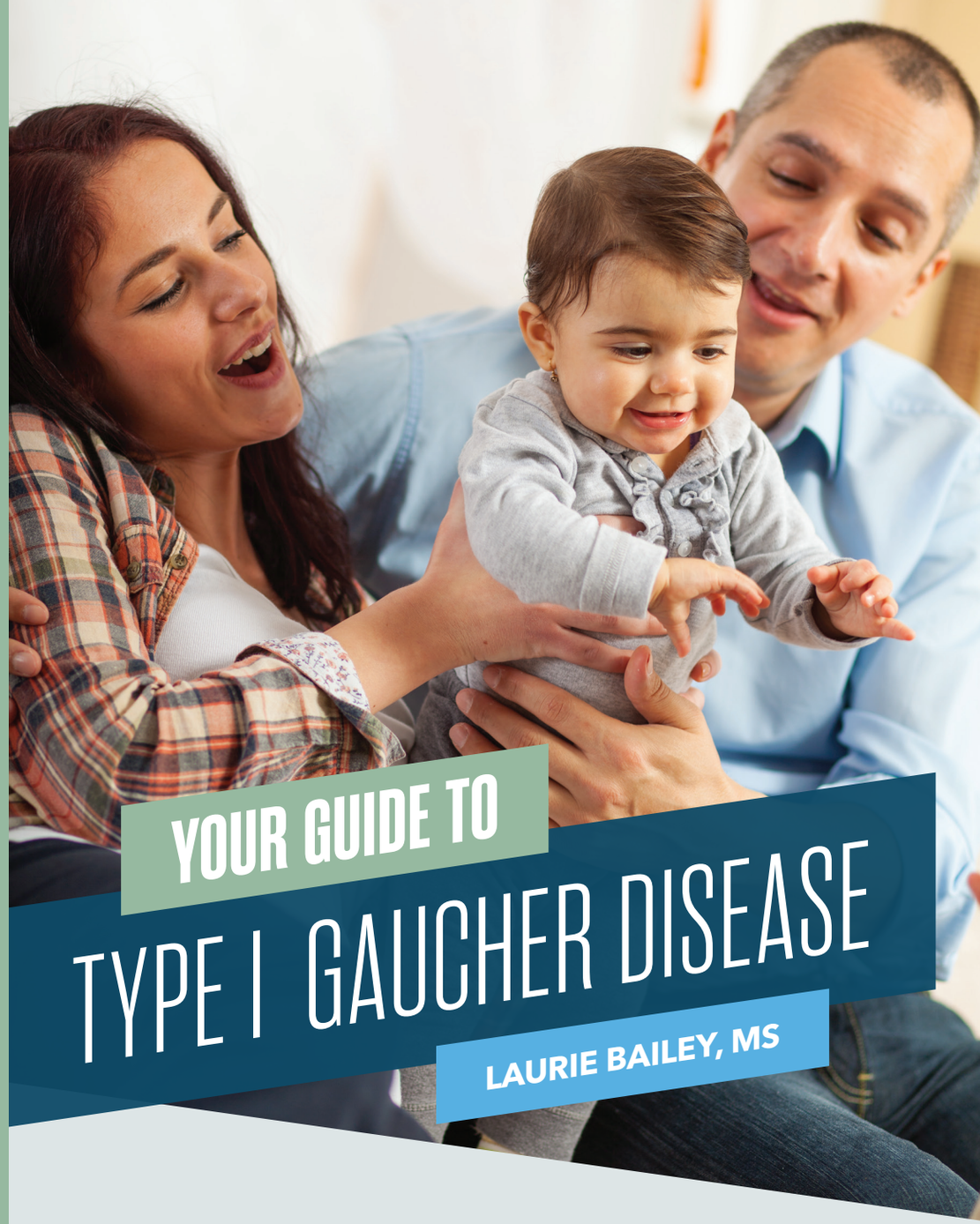
YOUR GUIDE TO TYPE 1 GAUCHER DISEASE LAURIE BAILEY, MS



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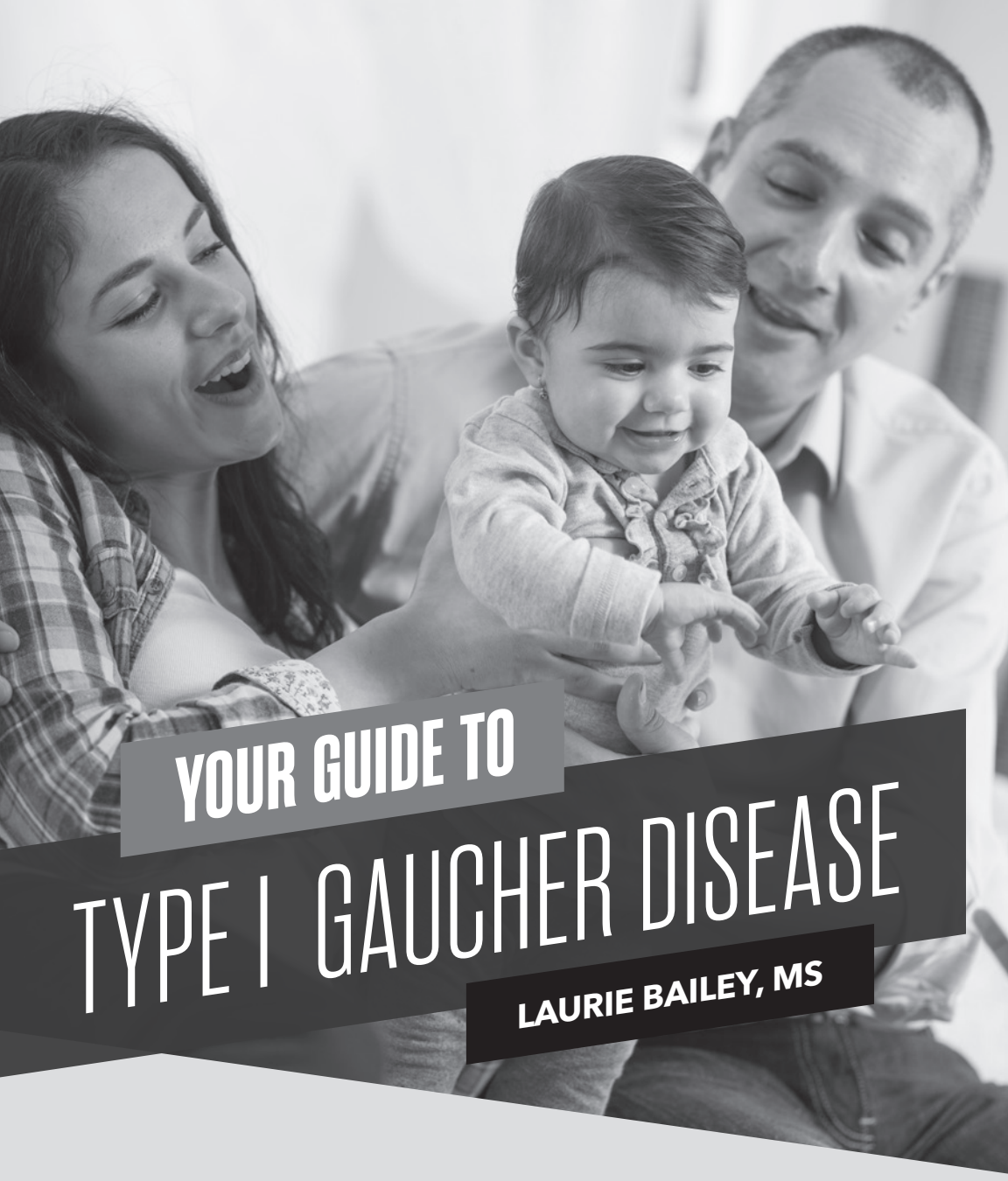

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This book is dedicated to all the patients with Gaucher disease and their families who took the time to share their stories of tragedy and triumph with me over the years. I have been humbled by the experience and learned more about Gaucher disease, as well as life, than anyone could imagine. Thank you for the opportunity to serve you.

ACKNOWLEDGMENT

I would like to acknowledge Dr. Greg Grabowski for sharing his passion with me for the treatment and management of people with Gaucher disease. His knowledge was invaluable, and his drive to make a difference in the lives of those affected inspired me to strive to learn more and always advocate for the patient and their families.

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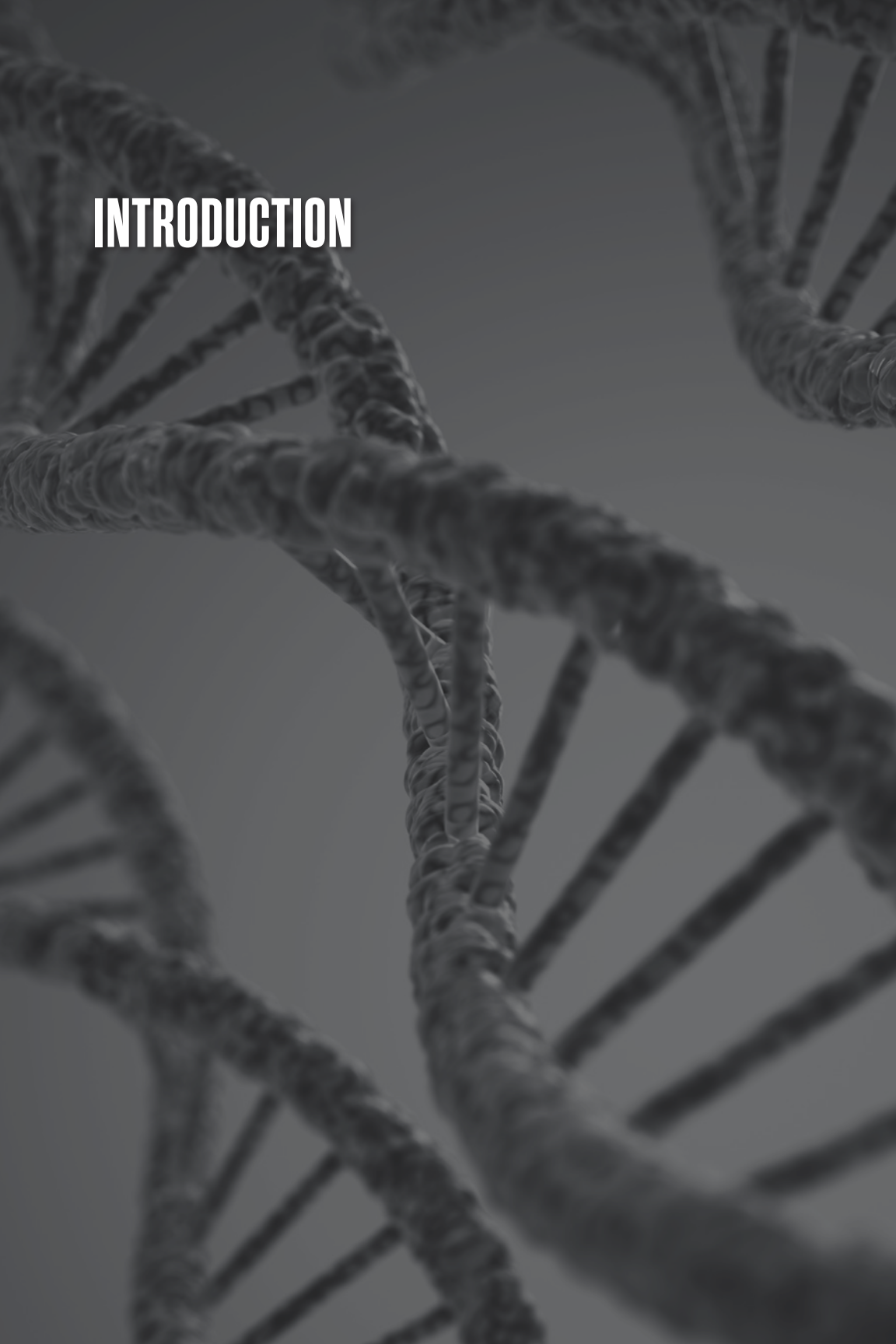


INTRODUCTION

INTRODUCTION

No matter how you say it, you've probably never heard of Gaucher (pronounced Go-shay) disease until you or someone you love is diagnosed with the condition. Although receiving news that you or a loved one has a rare disease can be difficult, the diagnosis of type 1 Gaucher brings relief to the majority of those who receive it in that they don't have something much worse, like cancer. In addition, it often ends a diagnostic odyssey that has progressed for many years or provides confirmation that a patient is not crazy — the symptoms they've been experiencing are valid complaints with a cause. Having an answer means that you can finally take steps toward feeling better.

Patients are also relieved to learn that there are effective treatments for type 1 Gaucher disease, which makes it unique among the majority of inherited diseases. Dr. Roscoe Brady, who served as scientist emeritus at the National Institutes of Health (NIH), spent more than 50 years conducting pioneering research on hereditary metabolic storage diseases, such as Gaucher. Dr. Brady designed the first enzyme replacement therapy (ERT) for patients with Gaucher, which was approved in 1991. Since then, it has stood as a paradigm of success for other treatments in lysosomal storage disease.

So, now that you have a diagnosis, what do you do next? Most general physicians have never had experience with Gaucher disease. As with other chronic conditions like diabetes and hypertension, your first step should be to find a specialist. To achieve the best outcome, it's important to have a specialist monitor your symptoms and formulate a treatment plan. There are many types of Gaucher specialists, including hematologists, pediatricians, liver specialists, and medical geneticists, among others, who are available to help. Resources such as this book are also invaluable for helping organize and facilitate the care of patients with type 1 Gaucher disease.

Of course, the patient and family are always the best advocates. For more than 5 years now, I have had the privilege of working with patients and families who are affected by Gaucher disease. They have welcomed me into their lives and together we have journeyed to achieving better health. I have learned about the disease by standing on the shoulders of the many brilliant counselors and physicians who have come before me, using this information to guide the care of individuals with this rare disease. And, I continue to learn each day through the interaction of patients and their families.

As you embark on this journey, please know that you are not alone. I urge you to join support groups and take advantage of the wealth of knowledge and experience in the Gaucher community. I have found that the people in this community are both welcoming and willing to help in many ways.

Sincerely,

Val Long, MS, CGC, CCRC

Certified Genetic Counselor
Certified Clinical Research Coordinator
Lysosomal Storage Diseases
Emory Healthcare
Division of Medical Genetics

Val Long is a certified genetic counselor and clinical research coordinator in Emory's Lysosomal Storage Disease Center in Atlanta, GA. After completing her undergraduate degree in human genetics at the University of Georgia, she went on to pursue her graduate degree in genetic counseling in Washington, DC. Val spent about 7 years as a prenatal genetic counselor before changing her focus to lysosomal storage diseases, particularly Gaucher disease, Late-onset Pompe disease, and Lysosomal Acid Lipase deficiency. She serves as a clinical coordinator for patients with these conditions, as well as a study coordinator for industry-sponsored clinical research trials regarding these diseases.

Chapter 1:

UNDERSTANDING TYPE I GAUCHER DISEASE

UNDERSTANDING TYPE I GAUCHER DISEASE

Receiving a diagnosis of a rare, incurable disease, such as type 1 Gaucher disease, is a life-changing event for everyone involved. While type 1 Gaucher can affect people of all ages, it can be especially challenging for parents to hear that their child is facing a serious disease. Many parents/caregivers describe it as a journey filled with twists and turns, unknown paths, and a wide range of emotions along the way.

In fact, the emotional experience is often compared to the five stages of grief – denial, anger, bargaining, depression and acceptance. It's important to realize that these emotions are normal. As difficult as it may seem, working through these stages can help you deal with your new reality in a healthy way. Remember, coping is an ongoing process.

Besides getting support, one of the best ways to begin this coping process is by learning as much as possible about the disease and its complications. This is important for two reasons:

1) As a parent/caregiver, you are your child's advocate. Becoming educated on what your child is going through will help you develop productive relationships with healthcare professionals, school staff, and others who are involved in the care of your child.

2) Knowledge truly is power – the better you understand the disease, its symptoms and complications and how to manage them, as well as care options, the more equipped you will be to deal with the obstacles, make decisions, communicate effectively with others, and improve the quality of life for you or your child.

While it's natural to feel helpless when you or your child is ill, educating yourself will help you feel more empowered. Let's start with a basic overview.

WHAT IS TYPE 1 GAUCHER DISEASE?

Overview

Gaucher (pronounced go-SHAY) disease is an inherited disorder. Though rare, it is one of the most common of about 50 lysosomal storage disorders. Lysosomes are tiny parts of a cell that contain enzymes. Enzymes are proteins that break large molecules into smaller pieces for the body to use or eliminate. When someone has a lysosomal storage disease like Gaucher, a specific enzyme doesn't work the way it is supposed to.

More specifically, a person with Gaucher disease lacks an enzyme called glucocerebrosidase (GLOO-ko-SER-e-bro-sy-dase) or GBA. This enzyme is needed to break down a fatty substance called glucocerebroside (GLOO-ko-SER-e-bro-side). Without enough of this enzyme, the fatty substance builds up in some cells, mainly in the spleen, liver, bone marrow and lungs. As the stored material continues to grow, these cells begin to enlarge. Cells that become enlarged due to the buildup of glucocerebrosides are referred to as Gaucher cells.

Over time, Gaucher cells collect in various organs, tissues and bone marrow, causing the signs and symptoms of Gaucher disease.

Individuals with type 1 Gaucher disease usually begin to exhibit symptoms during adolescence, but the age of onset ranges from early childhood to adulthood. The disease affects both males and females equally.

WHO GETS GAUCHER?

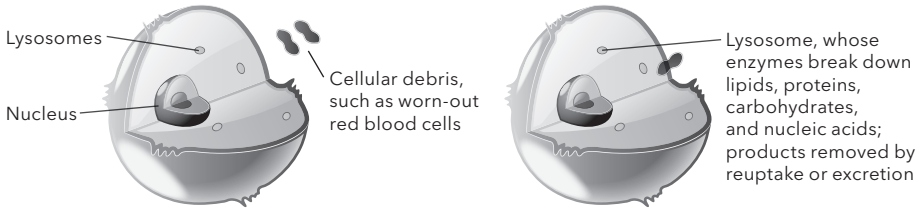
Among the general population, Gaucher disease occurs in roughly 1 in 50,000 to 1 in 100,000 people. However, it is most common among people of Ashkenazi (Eastern European) Jewish ancestry. In people of Ashkenazi Jewish ancestry, it occurs in approximately 1 in 500 to 1 in 1,000 people. (See "Why Are Some Genetic Conditions More Common in Particular Ethnic Groups?") It's estimated that there are approximately 6,000 individuals with Gaucher disease in the U.S.

Let's take a deeper look:

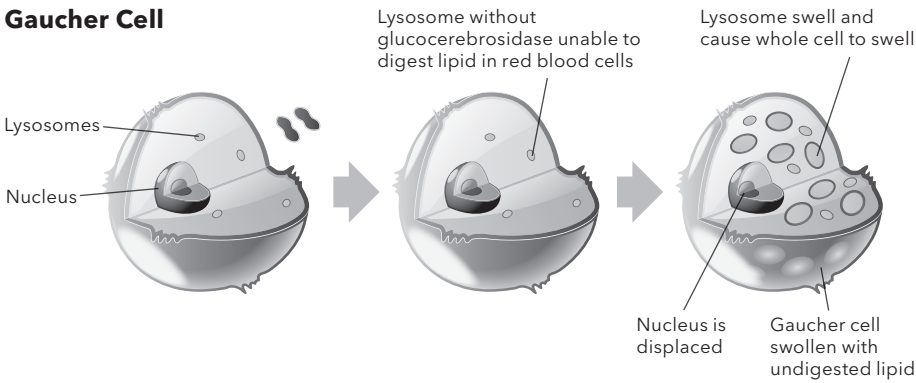
There are approximately 37.2 trillion cells in the human body. They provide structure for our bodies, take in nutrients from food, convert those nutrients into energy, and carry out specialized functions. Cells have many parts, each with a specific job.

Lysosomes are sac-like structures inside the cells. Think of them as "recycling centers" that break down unwanted substances into simpler ones so that the cell can use them to make new material or get rid of them. Glucocerebrosidase is an enzyme that exists in lysosomes and is necessary for breaking down this waste product. If this enzyme is lacking, waste builds up in the cells. If too much waste builds ups, the cells will no longer work properly.

Normal Cell



Gaucher Cell



Disease Spectrum

While there are other types of Gaucher disease, type 1 is the most common form. It accounts for more than 90% of all cases. Type 1 is the only form of Gaucher disease that is considered "non-neuronopathic," which means it does not usually affect the brain and central nervous system (spinal cord) like other forms of the disease.

Types 2 and 3 Gaucher are extremely rare and are known as neuronopathic forms of the disorder because they involve problems with the central nervous system, such as abnormal eye movements, seizures and cognitive impairment, in addition to the symptoms of type 1 Gaucher.

Type 2, also known as acute neuronopathic Gaucher disease, occurs in newborns and infants. It causes problems with the nervous system because the abnormal accumulation of glucocerebroside results in toxicity to the brain. The first symptom is often enlargement of the spleen (also known as splenomegaly), which may occur before 6 months of age. Infants with type 2 Gaucher often lose previously acquired motor skills (regression), have poor muscle tone and experience involuntary muscle spasms that result in slow, stiff movements of the arms and legs, and in many cases, have crossed eyes.

In addition, affected infants may have difficulty swallowing, as well as abnormal positioning of the neck, which results in feeding problems. These affected infants often fail to gain weight and grow at the appropriate rate (also known as failure to thrive). They may also have high-pitched breathing caused by the contraction of muscles in the voice box (laryngeal spasm). Anemia and a low blood platelet count (thrombocytopenia) may also occur, resulting in weakness and easy bruising. Sadly, type 2 Gaucher often progresses to life-threatening complications, such as respiratory distress or aspiration pneumonia caused by food in the windpipe or lungs. Newborns can be severely affected, having skin abnormalities and general swelling, with death occurring in the first few weeks of life. Other children with type 2 have significantly reduced lifespans, with death usually occurring between the ages of 1 and 3 years old.

Type 3 Gaucher, which is also known as chronic neuronopathic Gaucher disease, typically results in symptoms during the first decade of life. In addition to the blood and bone abnormalities discussed above, affected children develop neurological complications, such as mental deterioration, uncoordinated movements, and muscle spasms in the arms, legs or entire body (myoclonic seizures).

Some individuals with type 3 Gaucher have difficulty moving their eyes side-to-side (horizontal gaze palsy) or up and down (vertical gaze palsy). However, the symptoms and rate of progression of these symptoms can vary widely in patients and typically progress slower in type 3 Gaucher disease than in type 2 Gaucher disease. Those with type 3 Gaucher may live into their teens and early 20's, while others survive well into their 30's and 40's. Depending on the severity of complications, these individuals may require help with activities of daily living (ADLs), such as eating, bathing and getting around.

Because type 1 is by far the most common form of Gaucher, that is the focus of this book.

WHAT ARE THE SIGNS AND SYMPTOMS?

Even within type 1 Gaucher disease, the signs and symptoms vary widely among affected individuals – from mild to severe – and may appear any time from childhood to adulthood. Some patients develop few or no symptoms (they are considered asymptomatic), while others experience serious complications.

Typically, the most common signs and symptoms include:

Blood and Organs

- Spleen and liver enlargement (Hepatosplenomegaly) – When Gaucher cells build up in the spleen and/or liver, these organs become enlarged and can cause the abdomen to appear swollen and, and in some cases, be painful.
- Low blood platelet count (Thrombocytopenia) – A spleen that functions normally filters the blood and breaks down old blood cells. Because a person with Gaucher disease is missing an important enzyme needed to break down these old blood cells, the enlarged cells clog the spleen's filtering system and trap the newer cells. Ultimately, this results in low blood counts, including platelets.
- Bleeding and clotting problems – With fewer platelets, patients with Gaucher disease may have excessive bleeding even after minor injuries. Low platelet counts can also cause frequent nosebleeds, bleeding gums and easy bruising. Very low platelet counts can result in more serious bleeding issues, particularly after dental work, surgery, trauma and delivering a baby.
- Anemia – Gaucher cells in bone marrow reduce production of blood cells, and then the spleen quickly destroys the blood cells that are made. This combination can cause anemia, or low levels of red blood cells. Red blood cells carry energy-producing oxygen to all parts of the body, which means that patients with anemia often feel extremely fatigued. Anemia can also be caused by an iron deficiency or vitamin B_{12} deficiency, so it's important to work with a Gaucher specialist to identify and treat problems with anemia.
- Fatigue – As mentioned above, anemia causes fatigue, so it's common for individuals with type 1 Gaucher to feel excessively tired. However, not all fatigue in Gaucher disease is due to anemia.
- Respiratory problems – If glucocerebroside accumulates in the lungs, patients can experience respiratory problems. Respiratory issues may also cause fatigue, as well as shortness of breath.

Bone Involvement

- Bone pain and bone crisis – Patients often experience bone pain, including severe episodes called "bone crisis," which results from reduced blood flow to the bones.
- Bone infarction or avascular necrosis (AVN) – This condition occurs when parts of the bone don't get enough oxygen, causing bone tissue to deteriorate and die. Bone infarction often leads to hip or shoulder problems, severe arthritis and increased risk for fractures.
- Osteopenia and osteoporosis – Gaucher disease causes loss of calcium and minerals in the bones in patients of all ages, which weakens the bones. Smoking, excessive alcohol use, lack of physical exercise and certain medications can add to the risk of osteoporosis in patients with Gaucher.

- Spontaneous fractures – Bones that are weak are more likely to break. Since osteopenia (bone loss) and osteoporosis weaken the bones, the risk of fractures becomes higher. Some bone breaks may occur even without a trauma, which is considered a spontaneous fracture.

- Joint pain, arthritis and joint damage – It's common for patients with Gaucher to experience joint pain. The disease may cause severe arthritis and joint damage, which can be permanent if left untreated.

Although there is no cure for type 1 Gaucher disease, there are ways to manage the symptoms and treat the condition (see Chapter 3).

HOW DOES SOMEONE INHERIT GAUCHER DISEASE?

Gaucher is considered a genetic disease because it's inherited from a person's parents. In other words, it's passed down from generation to generation through genes.

What are genes? Genes are the segments of DNA (the blueprints for our bodies) that contain the instructions for making specific proteins. Basically, genes act as instructions to determine certain characteristics that are passed down from parents to their children, such as hair and eye color. Genes also tell the body how to produce important proteins called enzymes, which help break down specific compounds in the body. It's estimated that humans have between 20,000 and 25,000 genes.

Genes are carried within structures called chromosomes. Every human cell contains 23 pairs of chromosomes, one from each parent. Males and females have the same chromosomes except for one pair that determines the sex. Females have two X chromosomes, while males have one X chromosome and one Y chromosome.

While the genetic code is truly amazing, sometimes errors can occur. Information can be missing from the code or the code might contain too much information or have information in the wrong order. Some genetic problems, such as Gaucher disease, are caused by mutations, or changes, in one gene. A condition caused by mutations in one or more genes is called a genetic disorder.

As discussed, Gaucher disease is a genetic disorder caused by mutations in the GBA gene, which provides instructions for making an enzyme called glucocerebrosidase. The GBA gene is located on chromosome 1. There are two copies of the GBA gene, one from the mother and the other from the father.

Gene mutations can arise in two different ways:

- Hereditary mutations are inherited from a parent and are present throughout a person's life and are present in virtually every cell in the body.
- Acquired (or somatic) mutations occur at some time during a person's life and are found in only certain cells in the body. These changes may be caused by environmental factors or during cell division. These cannot be passed to the next generation.

Mutations resulting in Gaucher disease are hereditary. Gaucher disease can only develop when a person inherits two abnormal copies of the gene responsible for producing the enzyme glucocerebrosidase. If a person inherits only one abnormal copy of this gene, he or she is considered a carrier and could pass the gene that causes the disease on to his or her children, but will not actually have Gaucher disease.

INCIDENCE & INHERITANCE OF GAUCHER DISEASE

Both parents must carry the faulty gene to have an affected child, and men and women are affected equally.

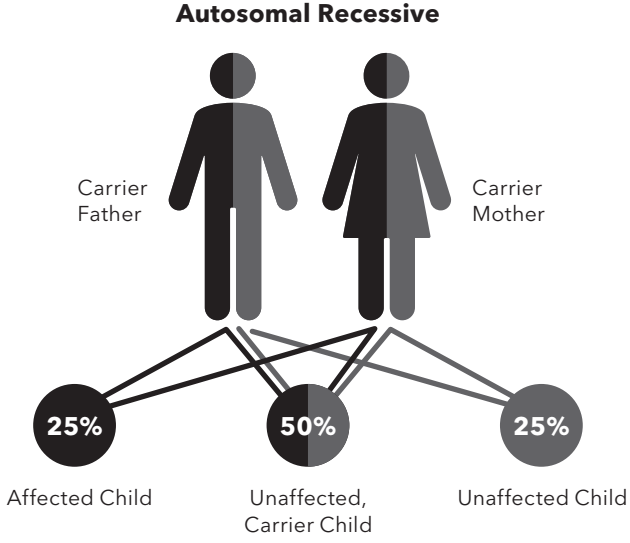
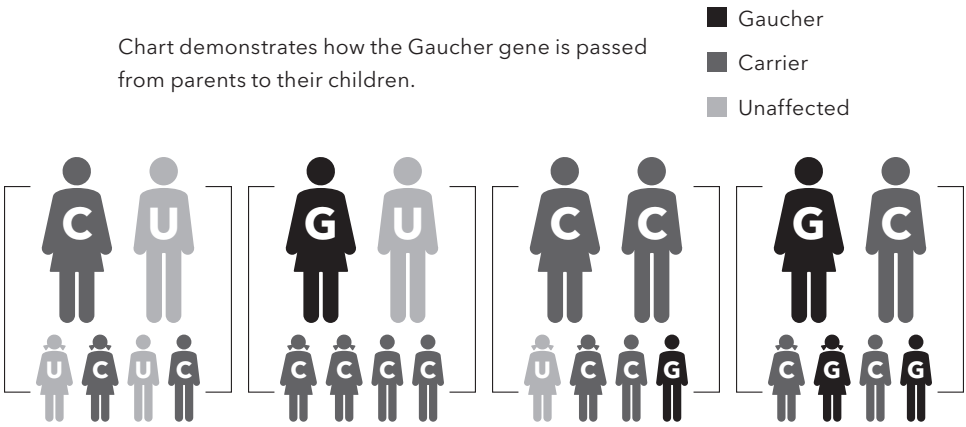


Chart demonstrates how the Gaucher gene is passed from parents to their children.



HOW IS GAUCHER DIAGNOSED?

If an individual is experiencing any of the symptoms of type 1 Gaucher disease, such as an enlarged liver and spleen, decreased blood counts, easy bleeding and bruising, and/or bone problems, it's important to get tested. This is especially true if there is a family history of the disease.

Because Gaucher disease is rare, patients must often request to be tested. Most healthcare professionals are not aware of Gaucher disease and may not suspect it unless a patient suggests it.

Testing for type 1 Gaucher disease involves a blood test for adults or a less invasive heel stick (a smaller prick) for babies. Testing to measure the activity level of the enzyme glucocerebrosidase is the most accurate way to diagnose Gaucher disease. Those with type 1 Gaucher will have very low levels of this enzyme activity. A second type of laboratory test involves DNA analysis of the GBA gene to check for specific mutations.

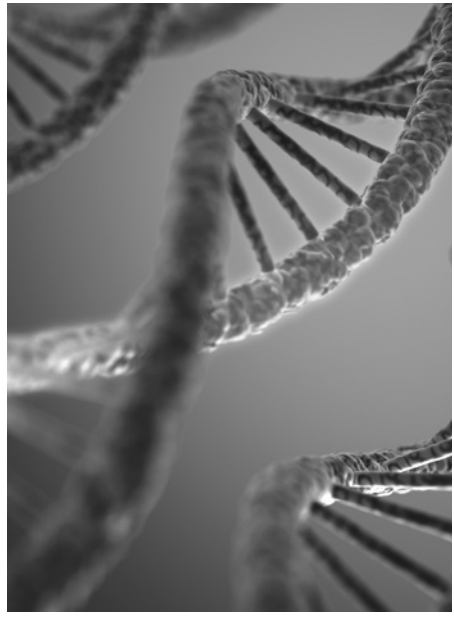
Gaucher cells may also be seen on a bone marrow or liver biopsy, suggesting a diagnosis of Gaucher disease, but demonstration of the enzyme deficiency and/or confirmation of DNA mutations is needed to make a diagnosis of Gaucher disease.

It's important to note that early diagnosis is critical for proactive management of the disease, which can help minimize long-term effects.

Genetic Screening for Carriers

Carrier screening for Gaucher disease is possible in people with Ashkenazi Jewish ancestry or if there is a family history of the disease. DNA testing can be very helpful in determining carrier status, which cannot be determined by enzyme analysis. Two carrier parents are at risk of having a child with Gaucher disease. However, DNA testing cannot predict how severely the child will be affected by Gaucher disease.

The DNA test involves a simple blood draw or saliva sample. It is also possible to do DNA testing prenatally.



WHY ARE SOME GENETIC CONDITIONS MORE COMMON IN PARTICULAR ETHNIC GROUPS?

Some genetic disorders are more likely to occur among people who trace their ancestry to a particular geographic area. People in an ethnic group often share certain versions of their genes, which have been passed down from common ancestors. If one of these shared genes contains a disease-causing mutation, a particular genetic disorder may be more frequently seen in the group. It's important to note, however, that these disorders can occur in any ethnic group.

Source: The National Library of Medicine, Genetics Home Reference, "Your Guide to Understanding Genetic Conditions."

FAMILY TO FAMILY

"Fear, sadness, and lots of questions…that's what I remember most about the day we received my son's diagnosis of type 1 Gaucher disease. He was only 8 years old at the time. In the beginning, there are often more questions than answers, and it's easy to become overwhelmed with the 'what ifs.' As hard as it may be, my advice is to take things one step at a time and focus on the present.

Also, remember that because the enzyme deficiency varies from person to person, the symptoms can also be very different.

It's also important to see a Gaucher specialist, because most general healthcare practitioners have no experience with this rare disease. With proper treatment, the disease can often be managed.

Most of all, I recommend seeking support. This may be a rare disease, but there is an entire community of Gaucher patients and caregivers to help you live the best life possible."

Mary – Mother of a 15-year-old boy with type 1 Gaucher

Chapter 2:

MONITORING YOUR CONDITION

MONITORING YOUR CONDITION

WHAT YOUR DIAGNOSIS MEANS

As discussed in Chapter 1, because the enzyme deficiency varies from person to person, the symptoms of type 1 Gaucher disease can also be different. Some individuals may be relatively symptom-free, while others experience more serious complications. In some cases, symptoms may not appear until adulthood, and even then, those symptoms can range from mild to severe.

Over the years, there have been significant advances in managing this disease. (See Chapter 3 for Treatment Options.) The key to effective disease management is to see a specialist familiar with Gaucher disease, along with healthcare providers that treat the specific symptoms you or your child are experiencing.

ASSEMBLING A TEAM OF SPECIALISTS

One of the most important aspects of living well with type 1 Gaucher disease is seeing a Gaucher specialist on a regular basis – at least once a year. Because this disease is rare, many general practitioners and even specialists have never treated a patient with Gaucher disease. Working with a Gaucher specialist can help you:

- Accurately diagnose type I Gaucher disease and evaluate the type and severity of symptoms.
- Create an individualized treatment plan based on your symptoms.
- Manage your symptoms to improve quality of life.
- Track key health indicators, such as blood counts, spleen and liver volume, and bone density (see Disease Monitoring).
- Coordinate your care with your primary care physician and other specialists.
- Proactively monitor for conditions related to Gaucher disease.

How can you find a Gaucher specialist?

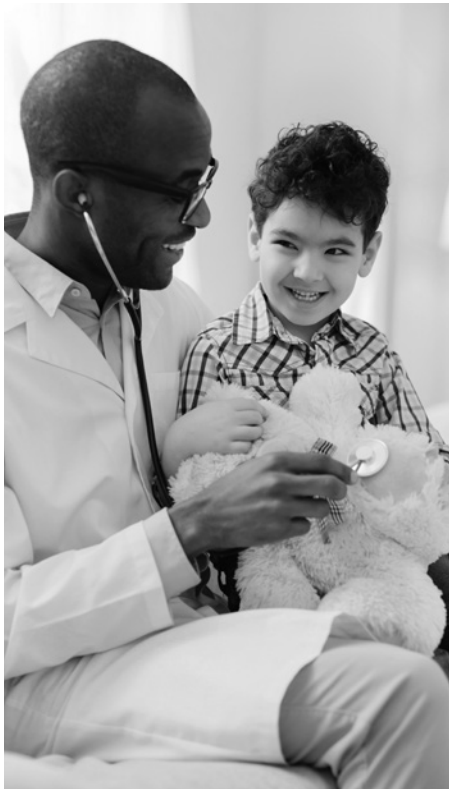
- The National Gaucher Foundation offers a "Treatment Finder" tool on their website. Visit www.gaucherdisease.org/gaucher-diagnosis-treatment/treatment-finder/.
- Consult with your primary care physician or genetic counselor, who may be able to help you locate a specialist in your area.
- Talk to other families living with Gaucher and ask for a recommendation.

Other organizations that offer resources, including lists of specialists, include:

Center for Jewish Genetics
30 South Wells St.
Chicago, IL 60606
Telephone: 312-357-4717
Fax: 312-855-3295
E-mail: jewishgeneticsctr@juf.org
Website: https://www.jewishgenetics.org

Children's Gaucher Research Fund
P.O. Box 2123
Granite Bay, CA 95746-2123
Telephone: 916-797-3700
Fax: 916-797-3707
E-mail: research@childrensgaucher.org
Website: http://www.childrensgaucher.org

Jewish Genetic Disease Consortium (JGDC)
450 West End Ave., 6A
New York, NY 10024
Toll-free: 866-370-GENE (4363)
Telephone: 855-642-6900
Fax: 212-873-7892
E-mail: info@jewishgeneticdiseases.org
Website: http://www.JewishGeneticDiseases.org



What should you look for in a Gaucher specialist?

Most Gaucher specialists work in a related specialty, such as hematology, orthopedics, genetics or pediatrics. Be sure to confirm that the specialist you work with is aware of all the problems someone with Gaucher disease can have. For example, a hematologist may not be an expert in bone complications. You might select a specialist based on the particular symptoms that you or your child are experiencing. It's also important to interview specialists when making a selection since you'll be developing a long-term relationship with this person. Most physicians will schedule time for an initial consultation or interview. To make the most of this time, come prepared with a list of questions, such as:

- How many years have you been treating Gaucher patients?
- How many Gaucher patients are you currently treating? How many of these patients are children (if applicable)?
- Have you authored or coauthored any research papers on Gaucher?
- Do you belong to any Gaucher organizations or advocacy groups?
- Are you familiar with the latest disease management and treatment options?
- Are you open to exploring alternative therapies or clinical trials?

Of course, as with any physician, the most important criteria for selecting a Gaucher specialist is how comfortable you feel with this person. Studies show that developing a good rapport with a physician, which involves trust and open communication, is critical to receiving the best possible care. (See "Developing Good Rapport with Your Physician.")

In addition to a Gaucher specialist, it's important to assemble a team of healthcare professionals to help manage and treat individual symptoms.

DEVELOPING GOOD RAPPORT WITH YOUR PHYSICIAN

1. Good communication – Communication should be two-way, allowing patients ample opportunities to ask questions and express concerns. The physician should make direct eye contact and face the patient rather than looking at a laptop during the entire conversation. Patients should come prepared for appointments with questions and accurate information (see #4). Patients should also take notes during the appointment, so they can accurately follow instructions given by the physician.

2. Interest in the whole person – A physician should be interested in more than symptoms. Knowing about a patient's family dynamics, job/school situation, relationships, etc. can lead to better diagnostics and treatment decisions. Patients should be willing to share this type of information.

3. Emotional presence – Making a genuine connection with a patient requires a physician to be "emotionally present." Does he or she seem genuinely interested and concerned with all changes in a patient's condition, even those that may seem small?

4. Honesty – Patients must be open and honest about all symptoms, medications, and lifestyle. Patients should be prepared to accurately answer questions regarding any changes in health. Keeping a symptom diary can help patients keep track of these changes and allow the physician to make better assessments. Physicians must also be willing to answer all questions fully and candidly.

5. Clarity – Physicians should avoid jargon and overly-complex explanations. Patients should feel comfortable asking for clarification. Taking notes can help patients remember important details regarding their care, especially when dealing with a number of specialists.

Depending on symptoms and complications, your healthcare team may also include:

- **Geneticist** – to evaluate, diagnose and counsel patients with hereditary conditions, such as Gaucher. Often these healthcare professionals work with genetic counselors and can answer questions about family risks and help coordinate care with other specialists.
- **Hematologist** – to diagnose and treat diseases related to blood, such as anemia. More specifically, they treat diseases that affect the production of blood and its components, such as blood cells, hemoglobin, blood proteins, bone marrow, platelets, the spleen and ability of blood to coagulate or clot, all of which may be affected by type 1 Gaucher.
- **Orthopedist** – to treat problems with the musculoskeletal system, using both non-surgical and surgical means. Bone conditions are some of the main complications of type 1 Gaucher disease. An orthopedist can repair fractures and deteriorated joints, as well as treat avascular necrosis, where bone tissue dies due to lack of blood supply.
- **Primary care physician** – to coordinate your treatment plan among specialists, ensure medications being prescribed do not interact negatively, diagnose and detect other issues related to Gaucher, as well as treat non-related health issues.
- **Pediatrician** – to provide medical care to infants, children and adolescents. The American Academy of Pediatrics recommends that individuals under the age of 21 be treated by a pediatrician.
- **Hepatologist (liver specialist)** – to diagnose and treat diseases of the liver, gallbladder and pancreas. The liver is one of the organs most often affected by type 1 Gaucher disease, which makes a hepatologist an important member of your team. A liver specialist can help diagnose and treat conditions often associated with type 1 Gaucher, such as portal hypertension (high blood pressure in the portal vein that transports blood to the liver), gallstones and gallbladder disease, and liver cancer.
- **Neurologist** – to diagnose and treat disorders of the nervous system, including the brain, spinal cord and muscle function. Neurologists are vital for individuals with type 3 Gaucher disease.
- **Pain management specialist** – to treat and manage chronic pain associated with bones and joints, as well as other painful conditions. These specialists typically take a multi-pronged approach to ease suffering and improve quality of life for those experiencing chronic pain.
- **Endocrinologist** – to treat the osteoporosis associated with Gaucher disease.
- **Physical therapist** – also known as physiotherapists, these healthcare professionals use mechanical force, movements, manual therapy, exercise therapy and electrotherapy to improve mobility, increase flexibility and reduce joint pain, thereby improving a patient's quality of life.

- **Psychologist or psychiatrist** – to help patients and caregivers cope with the often-overwhelming emotional demands associated with a chronic illness.
- **Oncologist** – to prevent, diagnose and treat cancer. Since type 1 Gaucher patients have a slightly increased risk of developing certain cancers of the blood and liver, seeing an oncologist for regular monitoring or changes in health may be advised.
- **Dentist or oral surgeon** – to diagnose and treat dental issues, including defects in the head, neck, face and jaws. Because Gaucher disease affects the bones, many patients also have dental problems related to bone and tooth hardness. In particular, some patients have problems with the mandible or jawbone. Routine x-rays can help detect problems early. Also, Gaucher disease may affect a patient's post-procedure bleeding and risk of infection. Therefore, your dentist should be aware of possible dental complications associated with Gaucher.

DISEASE MONITORING

One of the most important ways type 1 Gaucher patients can achieve optimal health and prevent permanent damage is through continued disease monitoring by a Gaucher specialist. These healthcare professionals will keep an eye on key health markers (measurements) to ensure the best possible care and outcomes. The following health markers are of particular concern for type 1 Gaucher patients:

Blood

As discussed in the first chapter, Gaucher cells that accumulate in bone marrow and the spleen make it hard for the body to produce blood cells normally and maintain healthy blood counts. Therefore, type 1 Gaucher patients may experience a number of blood-related complications, including low red blood cell counts (anemia), as well as low platelet counts that can lead to easy bruising and bleeding problems.

It's recommended that a complete blood cell count, including platelets and liver enzymes, be done at initial diagnosis (baseline) and then every 12 months in untreated patients. Patients who receive enzyme replacement therapy (ERT) should have blood cell counts performed every three months and at treatment changes until all values have been optimized.

Bones

Bone problems are common in people with type 1 Gaucher. Gaucher may cause the weakening of bones (osteoporosis) and bone loss (osteopenia), as well as bone pain due to reduced blood flow to the bones. If left untreated, these conditions can lead to avascular necrosis (the deterioration and death of bone tissue), increased risk of fractures, and spontaneous fractures. In addition, type 1 Gaucher may cause joint pain, arthritis and joint damage, which can become permanent if not treated properly.

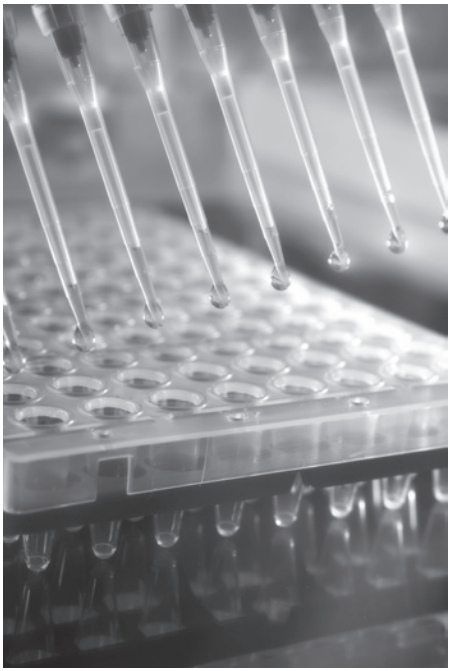
Therefore, it's critical for patients to be tested regularly for bone issues. This typically involves an initial MRI (magnetic resonance imaging) of the femur or thigh bone, spine and areas that may be stiff or painful, such as shoulders and hips. Periodic follow-up assessments should be considered and discussed with your physician.

Additionally, type 1 patients should have regular bone density measurements. A bone density test uses X-rays to measure how many grams of calcium and other bone minerals are packed into a segment of bone. The bones typically tested are in the hip and spine, but sometimes bones in the forearm, wrist and heel are also assessed. Bone density tests should be done every 1-2 years.

Biomarkers

A biomarker is a measurable indicator of the presence or severity of diseases in the body. In other words, a biomarker is anything that can be used to determine if a disease is present or how the disease is progressing. Biomarkers can be biological properties or molecules that can be detected and measured in parts of the body, such as blood or tissue. They can also be specific cells, genes, enzymes or hormones that affect the body.

One of the simplest examples of a biomarker is body temperature, which indicates a fever and possible infection. Blood pressure is another common biomarker, which is used to determine the risk of a stroke.



In addition to helping physicians diagnose and prevent diseases, biomarkers are useful in measuring the progression of diseases. Some drug-related biomarkers can be assessed to help determine whether a certain medication may be effective in a specific patient, as well as how the patient's body will react to or process the drug.

In the case of type 1 Gaucher, several biomarkers are used to assess the severity of the disease and monitor its progression, as well as determine a patient's response to therapy, such as ERT. Biomarkers cannot be used independently to monitor Gaucher disease, but together with other assessments, such as MRI and blood counts, they help healthcare providers make decisions about treatments and the doses that are most appropriate. The following are some common blood biomarkers used to monitor individuals with type 1 Gaucher disease:

Chitotriosidase (CHITO): CHITO is measured in the blood. It's an enzyme produced and secreted in activated macrophages (a type of white blood cell) and is involved in breaking down things that contain "chitin," like bacteria and certain fungi. There are more activated macrophages when there is chronic inflammation. Since type 1 Gaucher disease causes chronic inflammation, there are more activated macrophages, and therefore, more CHITO is produced. Individuals with untreated Gaucher disease can have highly elevated levels of CHITO.

Tartrate Resistant Acid Phosphastase (TRAP): TRAP is another type of enzyme which is stored in lysosomes. TRAP is secreted by the bone, liver, spleen and kidney. More of this enzyme is secreted in the presence of activated macrophages and chronic inflammation, as is found in people with type 1 Gaucher disease. Therefore, elevated levels of TRAP are often observed in those with Gaucher disease.

Angiotensin Converting Enzyme (ACE): ACE is a non-specific indicator of lipid storage, which means that the level of ACE may be elevated as a result of Gaucher-related lipid storage. However, levels of ACE could also be higher than normal due to other lipids, such as cholesterol being stored in the body. People with diabetes and hyperthyroidism may also have elevated levels of ACE. If an individual with Gaucher disease is taking a class of drug known as ACE inhibitors (e.g., Lisinopril or Enalapril), which are primarily used to treat high blood pressure, then the ACE measurement will be reduced to zero and it will not be a useful biomarker.

Glucosylsphingosine (Lyso-Gb1): In an individual without Gaucher disease, glucocerebroside is broken down into glucose and ceramide for the body to process. Remember, a person with type 1 Gaucher disease lacks the enzyme to break down this fatty substance, allowing it to build up in some cells. Those with type 1 Gaucher disease have elevated levels of Lyso-Gb1.

Type 1 Gaucher patients should have their biomarkers monitored on a regular basis by a Gaucher specialist in order to help assess progression of the disease and its treatment.

It's also important for those living with Gaucher to take good care of themselves, including eating well and exercising, which will help strengthen bones and ensure optimal health.

FAMILY TO FAMILY

"It took nearly three years before we received a diagnosis of type 1 Gaucher disease for our daughter. When she was 14 years old, she began getting frequent nosebleeds, and we noticed that she bruised easily. We thought the bruises were caused by soccer, but she seemed to get more than other kids. A blood test showed that she was anemic, and she was prescribed iron pills, which didn't help. Eventually, she became so fatigued that she could barely get through the day.

At age 17, we finally saw a hematologist who ordered a bone marrow biopsy, which revealed that she had type 1 Gaucher. He sent us to another hematologist who is also a Gaucher specialist.

When we first met with our Gaucher specialist, we definitely had more questions than answers. Honestly, we didn't know where to start as it relates to specialists, testing and treatment. But, after our initial visit, we felt much more in control. She did a thorough assessment and outlined a treatment plan for our daughter that was based on her symptoms. She began receiving bi-weekly infusions and started feeling better. It was very reassuring to finally talk to someone who knew what we were going through and could walk us through the process.

We've been working with this physician for over 5 years now and have formed a wonderful relationship with her. Thanks to regular visits, diligent monitoring and treatment, our daughter has been able to live an active life. In fact, she will soon be graduating from college!

We're thankful that she has a relatively mild form of type 1 Gaucher, but we also understand the importance of keeping an eye on changes in her health. She will need monitoring and treatment for the rest of her life. She also follows a diet and exercise program designed to help prevent bone loss and stay healthy.

Our advice to other families living with this disease is to find a great specialist and explore disease management options with him or her. Our daughter also found lots of support and encouragement from other Gaucher patients on the Gaucher Disease blog (www.gaucherstories.wordpress.com) and the NGF website. She has met Gaucher patients from all over the world. It's nice to know that we're not alone."

Rebecca – Mother of a 22-year-old daughter with type 1 Gaucher disease

Chapter 3:

TREATMENT OPTIONS

TREATMENT OPTIONS

"Incurable" is a scary word. It means your life has been changed forever, which can be difficult to process even for the strongest person. As discussed earlier, it's normal for patients and loved ones to go through a flood of different emotions as you learn to cope with this new reality.

While there's no cure for type 1 Gaucher disease, there are a variety of treatment options that are intended to help control symptoms. Because this disease affects each person differently, it's important to work with a Gaucher specialist to monitor symptoms and complications and recommend treatment options. Some people have such mild symptoms that they don't require treatment, while others require a more aggressive approach.

The good news is there are treatment options available to help manage this disease. It's important to understand, however, that treatment may not address all the symptoms associated with type 1 Gaucher disease and could result in negative side effects. Deciding which treatment will be most effective, along with the correct dosage, will depend on the type and severity of symptoms you're experiencing, as well as other factors, such as age and overall health. Because choosing a treatment is such a significant decision, you should work with your Gaucher specialist to determine which option is best for you or your child.

There are two types of Gaucher disease treatments currently available: enzyme replacement therapy (ERT) and substrate reduction therapy (SRT).

ENZYME REPLACEMENT THERAPY (ERT)

How it Works:

Remember, people with Gaucher disease do not have enough active enzyme. Enzyme replacement therapy (ERT) provides the body with an artificial enzyme. Essentially, it balances low levels of the glucocerebrosidase (GCase) enzyme that Gaucher patients have with a modified version. The artificial enzyme helps break down glucocerebroside (GL1), the fatty chemical
that accumulates in the cells of patients with Gaucher. This may help to compensate for the defective enzyme.

ERT is given in a procedure called an infusion. An infusion involves a drug being administered intravenously (IV) through a vein in the arm. Infusions are usually given every 2 weeks. However, dosages and timing may vary depending on the patient and recommendations of your specialist.

What to Expect with Your Infusion:

Each infusion typically takes 1 to 2 hours to complete. Patients can receive ERT in a clinical setting, such as an infusion center, a physician's office, or a specialty pharmacy infusion center. A patient can also receive an infusion at home with the help of a home health nurse. Your insurance company may have preferences regarding infusion location, and your managing physician may have preferences about starting infusions in a clinical setting before moving to a home setting. Pharmaceutical case managers work with your physician's office, as well as your insurance company, to determine the best options (see Chapter 6 for more regarding health insurance).

- Infusion Centers – These are medical facilities specializing in intravenous (IV) infusions. Infusion centers may be part of a medical center, specialty pharmacy or a stand-alone facility. Once a patient arrives at an infusion center, medical staff prepare the medication. This process can take an hour or more, although some centers will start the preparation when patients are on their way. Depending on the infusion center, you may be in a large room, sitting in a reclining chair, with several other patients getting infusions. Patients may be getting ERT for Gaucher disease, but could also be receiving chemotherapy for a cancer diagnosis or another IV drug. Sometimes a curtain can be drawn between infusion chairs for privacy.

  Some patients find traveling to infusion centers and waiting for the procedure, as well as receiving the infusion itself, time-consuming and inconvenient. It can also be difficult for young children, who often miss activities and events due to infusions. On the other hand, many patients prefer infusion centers because they are familiar and have medical staff on site.

- Gaucher Disease Treatment Centers – Some hospitals and physicians' offices are equipped with infusion centers for Gaucher patients. This allows Gaucher specialists to easily monitor key health markers (see Chapter 2) and adjust a patient's medication dosage. Patients must still travel to these appointments and wait for medication to be prepared. However, some patients find it more convenient if they can combine infusions with appointments to see their specialist. Having a Gaucher specialist on hand who is familiar with the drug and the disease can be reassuring if a reaction occurs during an infusion. Some physicians' offices provide private rooms for the infusions.

- Home Infusions — For some patients, receiving infusions at home is more convenient and comfortable. It requires the help of a home health nurse, who prepares the medication and assists with the IV in your home. Home infusions are typically offered only after a patient has received several infusions in a clinical setting without complications. Home infusions may be done in the evenings or on weekends, depending on the nurse's availability. Because you don't have to travel or spend time waiting at an infusion center, home infusions may save time. This can help children lead a more "normal" life by allowing them to more easily attend school and other activities. In addition, some younger children might be more comfortable in a home setting. However, there are no additional medical staff on hand if complications occur, such as difficulty inserting the IV.

Regardless of the setting, it's important to work with your Gaucher specialist to monitor these treatments and stick to your treatment plan.

Currently, the FDA has approved several enzyme replacement therapy drugs. Again, your Gaucher specialist can help determine which of these drugs may be right for you and can counsel you on the potential risks and benefits of treatment.

SUBSTRATE REDUCTION THERAPY (SRT)

How it works:

Substrate reduction therapy (SRT) takes a different approach to treating type 1 Gaucher disease. Instead of providing an enzyme to break down glucocerebroside, SRT reduces the production of the substance that people with Gaucher disease cannot break down. Since everyone with Gaucher disease has a tiny bit of natural functioning enzyme, the hope is that if there isn't as much material to break down then the natural enzyme can keep up. If we go back to our "recycling" analogy, ERT helps the body recycle more waste (glucocerebroside), while SRT helps the body produce less of it to begin with.

Administration:

SRT is administered as an oral medication, which does not require an infusion. However, SRT cannot be used by women who are pregnant or breastfeeding, nor is it approved for use in children. These drugs are also not recommended for very elderly patients or people with kidney or liver disease.

Because every person reacts differently to these drugs, they may not be effective for certain patients. Again, a Gaucher specialist can help you decide which type of medication is right for you.

OTHER POTENTIAL TREATMENTS

ERT and SRT are intended to treat the underlying cause of type 1 Gaucher disease to prevent problems that require additional treatments. A Gaucher specialist will help you determine if additional treatments may be necessary. The following are a few therapies that may be discussed:

Medication for Osteoporosis

For reasons that are not completely understood, people with type 1 Gaucher disease develop osteopenia (bone loss) and osteoporosis (weakened bones). Depending on the results of MRIs and bone density testing, your specialist may prescribe additional treatment for osteoporosis.

There are a number of drugs used to help strengthen and rebuild bones.

The most common type of medication prescribed for osteoporosis is a "bisphosphonate." Healthy bones continually break down and rebuild. Biophosphonates work by slowing down the cells which break down bone. By slowing down bone loss, they allow the bone-building cells to work more effectively. Your physician can help you decide if this type of treatment would be beneficial, and if so, which drug is right for you. Biophosphonates can be administered orally, either once a week or once a month, or in some cases, through a quarterly or yearly intravenous infusion.

Some physicians may recommend calcium and vitamin D supplementation. **Never start taking a medication or supplement without first consulting your physician, as any medication or supplement could have side effects or interact with other medications you are taking.**

Don't rely on medication alone. Most experts agree that medication is more effective when combined with lifestyle changes. In fact, the following recommendations will not only make treatment more effective, but they may prevent osteoporosis from occurring:

- **Get Some Exercise:** Any physical activity that is "weight bearing" (see "8 Exercises to Strengthen Bones"), along with exercises that improve balance can strengthen bones and reduce your risk of a fracture. This is especially true as you age because bones naturally weaken over time.
- **Eat a Healthy Diet:** An overall balanced diet is essential for good health, but getting enough calcium and vitamin D is particularly important for strong bones.
- **Quit Smoking:** Smoking cigarettes speeds up bone loss.
- **Limit Alcohol:** If you choose to drink alcohol, do so in moderation. For healthy women, that means up to one drink a day and up to two drinks per day for healthy men.

Bone Marrow Transplant

A bone marrow or stem cell transplant is a risky procedure that involves getting rid of a person's bone marrow with chemotherapy and replacing it with a donor's bone marrow that does not have Gaucher disease. If successful, the new bone marrow produces the enzyme deficient in Gaucher disease and may eliminate the need for ERT and SRT. The procedure can make patients quite sick and may result in death. Therefore, most physicians agree that the risks of the procedure outweigh the benefits and reserve this option for individuals with severe disease for whom ERT and SRT may not be effective.

Splenectomy

Before enzyme replacement therapy became available, removing the spleen (splenectomy) was a common treatment for Gaucher disease. When dangerously low levels of hemoglobin and platelets cause a massively enlarged spleen, a splenectomy can correct the problem by removing the spleen, which is trapping the blood cells. This procedure, however, does not address the underlying problem of storage of material the body cannot break down, so other problems associated with Gaucher disease continue to progress, such as bone disease and liver enlargement. Today, this procedure is usually not performed unless there is a serious Gaucher-related complication.

(Note: For information on joint replacements, pain management and medications for related diseases, such as Parkinson's, please see Chapter 4: Managing Symptoms and Complications.)

Every Case is Different

When it comes to type 1 Gaucher disease, there is no "one-size-fits-all" treatment plan. There are many variables, including when the disease is diagnosed, and the severity and type of symptoms, to consider when developing a course of treatment. However, there are some commonalities:

- Early diagnosis and proactive treatment is important.
- Working with a Gaucher specialist can help you determine the most effective treatment plan.
- Taking a holistic approach to your health will result in the best outcomes.

8 EXERCISES FOR STRONG BONES

Exercises that put stress on bones and muscles are called "weight bearing." Because weight-bearing exercises help bones stay strong, they should be incorporated into your everyday life. Thankfully, you don't have to spend hours in a gym to reap the benefits! Of course, be sure to talk to your doctor before starting any new exercise routine to make sure it's safe for you. Once you get the okay, try the following workouts:

1. **Tai Chi** – This practice, which uses a series of slow, graceful movements, builds both coordination and bones. A study reported in *Physician and Sports Medicine* found that tai chi could slow bone loss in postmenopausal women, which was confirmed with bone density tests. Tai Chi is also a good stress reliever, which can benefit those dealing with the emotional stress associated with a chronic disease such as Gaucher.
2. **Yoga** – A study reported in *Yoga Journal* found an increase in bone mineral density in the spines of women who did yoga regularly. Whether you prefer a slow, precise style of yoga (Iyengar), or a more vigorous form (Ashtanga), yoga can build bone in your hips, spine and wrists, which are the bones most vulnerable to fracture. As an added bonus, yoga also improves balance, coordination, concentration and body awareness, all of which helps prevent falls and relieve stress.
3. **Brisk Walking** – Walking is still one of the best ways to exercise – not to mention it's free and doesn't require any special equipment. A study of nurses found that walking 4 hours a week resulted in a 41% lower risk of hip fractures, compared to those who walked less than 1 hour a week. Brisk walking, which increases your heart rate, is best, but you can adapt your speed to your current fitness level or physical abilities.

4. **Golf** – This is good news for golfers. Recent studies show that carrying a golf bag and walking (no carts) around the course, as well as swinging clubs, can add up to a good workout. While it may not be good for your golf game, chasing down those errant balls improves the benefits!

5. **Dancing** – Who says that exercise can't be fun? Dance classes are making a comeback these days, and for good reason – not only is dancing enjoyable, it provides both a cardio workout, as well as bone-building hip and leg action.
6. **Hiking** – If you love getting outside and enjoying nature, hiking could be the best bone-building exercise for you. Because hiking is done on trails, which include rougher terrain and hills, it provides a better workout than walking on flat surfaces. It's also a great way to relax.
7. **Racquet Sports** – These include tennis, squash and paddleball. Swinging the racquet impacts your arm, wrist and shoulder, while all that running around chasing balls works your hips and spine.
8. **Strength Training** – Lifting weights, using the weight machines at a fitness center, or doing calisthenics are all forms of strength or resistance training. You're working against some type of resistance – whether it's free weights or your own body weight – to stress a sequence of muscles and bones. The surgeon general recommends strength training at least twice a week to stimulate bone growth.

For the best results, try combining a few of these workouts into your routine. Invite some friends to join you, which will not only help you stay on track, but provide the added benefit of socializing.

If you've been told that your bones are weakened or have been diagnosed with osteoporosis, it's important to take a few precautions. Because you have a higher risk for fractures, sports that have the potential for serious falls, such as downhill skiing or ice skating, or that require high-impact activity, are not advisable. Also, if you have thinning in your spine, you should avoid deep backbends in yoga. Again, it's wise to check with your physician before starting any new exercise program.

Source: WedMD, Rebecca Buffum Taylor, 2018.

BEYOND TREATMENT

"The good physician treats the disease; the great physician treats the patient who has the disease." **- William Osler**

There is both a physical and mental aspect to coping with a chronic disease. Aside from the physical symptoms, which are treated with medical therapies, many patients become depressed or anxious, which can lead patients to stop taking care of themselves, as well as stress-related health issues. However, studies show that taking a holistic approach to treatment results in the best outcomes, which means that patients need to incorporate mental wellness, stress reduction, proper diet and exercise for optimal health.

Even when patients are limited physically due to joint and bone pain, it's important to exercise daily to whatever extent is possible. As little as 30 minutes of physical activity can provide tremendous benefits. Stretching, walking, bike riding and pool therapy, which are easy on joints, can not only reduce stress, but improve flexibility and reduce pain. If you haven't been physically active, you should begin by consulting a physician or working with a physical therapist.

A healthy diet is also essential. Think of food as your secret weapon in the battle against disease of all types. Proper nutrition gives your body the resources it needs to build muscle, fight infection, repair cellular damage and stay strong, as well as prevent a wide range of health issues, such as diabetes and heart disease. If you're not sure where to start, you might consult with a nutritionist or check out www.heart.org/HealthyLiving/HealthyEating/Nutrition, which provides guidelines for a heart-healthy diet that offers overall benefits.

Finally, don't neglect your mental well-being. Stress reduction techniques such as meditation and breathing exercises can be very beneficial. For those with strong religious or spiritual beliefs, tapping into the power of these practices can also be very helpful. Staying physically active, maintaining social ties and activities, and engaging in hobbies are also effective ways to reduce stress and improve mental well-being. Any activity that provides a respite from your worries and gives you a brief mental break will help you get through a rough day.

Focusing on today is also very helpful. It's easy to get caught up in the "what ifs," but dwelling on future treatments and possible complications is counterproductive. It can leave you overwhelmed and depressed.

If you or your child experience symptoms of depression, including withdrawal, lack of interest in activities, loss of appetite, fatigue, or thoughts of hurting oneself, it's important to seek professional help. In fact, seeing a professional therapist or participating in a support group can ward off depression and improve your coping skills. Involving family members in this type of therapy can also be beneficial. The goal is to live the best life possible.

FAMILY TO FAMILY

"When I was 32, I began to feel severely fatigued. Since I was working full time and raising two busy middle-schoolers, I thought being tired was normal! But, as the fatigue grew worse, I also noticed some tenderness and swelling in my abdomen, which turned out to be an enlarged spleen. Because the disease does run in our family, the physician suspected Gaucher, which blood tests confirmed.

As soon as I was diagnosed, I began working with a Gaucher specialist, who emphasized the importance of beginning treatment right away. After a series of tests, he recommended ERT. While the bi-weekly infusions are a bit inconvenient, I was willing to do whatever it took to feel better and prevent the disease from causing more damage to my body. Instead of getting frustrated by the time the infusions take, I now use this time to catch up on my reading.

I've been receiving infusions for several years now, and I feel good. I have some days that are better than others – days when I have more energy – but overall, I've been able to continue working and attend my daughter's dance classes and soccer games.

To reduce the risk of fractures, I did replace my daily jog with walking and started swimming at our local recreation center, which helps me stay in shape, but is easier on bones and joints.

One of the things I've found most helpful is talking to other people with type 1 Gaucher. I joined an online community, which fits into my busy schedule. These folks have provided great advice and tons of support. I think of them as my extended family."

Emily – Age 35, Type 1 Gaucher patient

Chapter 4:

MANAGING SYMPTOMS AND COMPLICATIONS

MANAGING SYMPTOMS AND COMPLICATIONS

Though the treatments we covered in the last chapter can be highly effective at minimizing or even eliminating certain symptoms and complications related to Gaucher, many patients receiving therapy will continue to experience some conditions, such as fatigue and pain. As we've discussed, the type and severity of these symptoms can vary dramatically from one person to another.

Why doesn't treatment "fix" all the issues patients face with type 1 Gaucher? To answer this question, let's first review how ERT and SRT work. Both therapies focus on the enzyme deficiency and resulting accumulation of glucocerebroside, which is the underlying cause of Gaucher. However, they are therapies, not a cure. Response to therapy can vary from one individual to the next, and some problems may not respond at all.

Working with your Gaucher specialist, along with other members of your medical team, is the best way to monitor your condition and treat complications as they arise. But, patients should also be aware of the most frequent complaints associated with this disease and how to manage them, as well as the potential risks of developing related conditions.

COPING WITH FATIGUE

Chronic fatigue is a very common symptom of type 1 Gaucher. For some patients, extreme fatigue is their most debilitating symptom. Unfortunately, the cause of this fatigue is still unclear, and may result from a number of problems, including anemia, a weakened immune system, pain, depression and sleep disturbances.

While ERT can reduce fatigue in some patients, others find that they still feel abnormally tired, especially right before they are due for their next ERT infusion. In some cases, increasing the dosage or frequency of ERT can reduce symptoms of fatigue. However, if blood counts are still abnormal after being on therapy for a while, then you or your child may be referred to a hematologist, who specializes in blood disorders. He or she may prescribe iron supplements, iron injections, or, in severe cases, blood transfusions.

In addition to fatigue, anemia may cause weakness, dizziness, shortness of breath, increased thirst and visibly pale skin. If you or your child are experiencing any of these symptoms, consult your Gaucher specialist or hematologist.

Obviously, more research needs to be done regarding the causes and treatment of fatigue in Gaucher patients. Until we learn more, patients should be sure to eat a nutritious diet, rich in iron, and maintain good sleep habits (see "Tips for Better Sleep"). While it may be difficult to get motivated when you're feeling so tired, physical activity can actually boost your energy and help you sleep more soundly at night. Even if you are experiencing severe fatigue, aim for at least 30 minutes of exercise each day, such as walking.

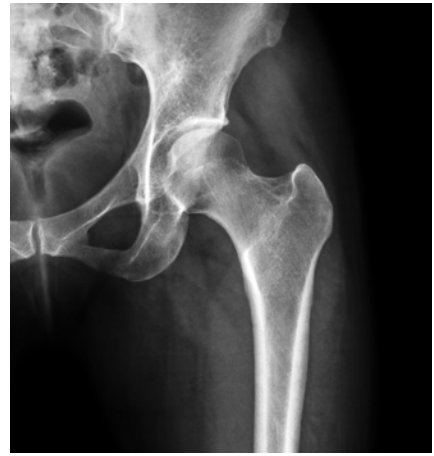
TIPS FOR BETTER SLEEP

Studies show that developing these healthy habits can help you get a better night's sleep:

- Follow a consistent sleep schedule. Try to go to bed and get up at the same time every day, even on weekends and vacations. It can be tempting to "catch up" on sleep on the weekends, but this can disrupt your body's internal clock.
- Set a bedtime that is early enough to get at least 7 hours of sleep. Trying to get all those "last minute" things done before bedtime only leads to sleep deprivation!
- Don't go to bed unless you feel sleepy. And, if you don't fall asleep after 20 minutes, get out of bed. Lying awake in bed will only make you more anxious. Get up and do something relaxing, such as reading under a soft light. Avoid television, cell phones and computers, which emit blue light. Studies show that exposure to bluish light can sabotage your sleep. In fact, electronic devices should be turned off at least 1 hour before bedtime.
- Establish a relaxing bedtime routine that signals your body that it's time for sleep.
- Make your bedroom quiet and relaxing. Keep the room at a comfortable, cool temperature.
- Limit exposure to bright light (and blue light) in the evenings.
- Don't eat a large meal before bedtime. If you're hungry at night, eat a light, healthy snack.
- Exercise regularly and maintain a healthy diet.
- Avoid consuming alcohol before bedtime.
- Avoid consuming caffeine in the late afternoon or evening.
- Reduce your fluid intake before bedtime to avoid getting up in the middle of the night to use the bathroom.

Source: American Alliance for Healthy Sleep, Sleep Education, http://www.sleepeducation.org/essentials-in-sleep/healthy-sleep-habits

DEALING WITH BONE AND JOINT PAIN



Besides fatigue, dealing with bone and joint pain is one of the most difficult aspects of living with type 1 Gaucher disease. As we've discussed, bone and joint problems are common among Gaucher patients. The first step in treating pain associated with bones and joints is identifying the root cause, which is not always straightforward. Often, it takes testing and a team approach from your physicians.

Discovering the source of pain typically begins with x-rays, an MRI (magnetic resonance imaging) and/or bone scans. Depending on the findings, you may work with your primary care physician, an orthopedist, a neurologist and/or a pain management specialist to help you effectively treat and manage your pain.

The course of treatment often depends on the patient's age, the duration of symptoms and the type of bone complications. In children and young teens with type 1 Gaucher (who are unlikely to have age-related arthritis, osteoporosis or long-term bone and joint damage), bone pain could be related to their type 1 Gaucher disease. Treatment for type 1 Gaucher may help with bone pain in these cases.

Treatment becomes more complicated as patients age. Individuals who had Gaucher long before newer treatments were available may have irreversible bone and joint damage that worsens with age. In addition to Gaucher disease-related pain, adults with type 1 Gaucher disease can have pain from arthritis or osteoporosis associated with the normal aging process.

Physicians often prescribe acetaminophen or non-steroidal anti-inflammatory drugs (NSAIDs) for pain relief. In severe cases, opioid medications may be prescribed. However, since there are certain health risks associated with these drugs, including the potential for addiction, patients or caregivers should discuss the risks and benefits of long-term use with their physician.

Additionally, type 1 Gaucher patients may experience joint pain, arthritis and joint damage, which can become permanent if not treated properly. Working with your Gaucher specialist, as well as an orthopedist, can help treat and manage these complications. In some cases, orthopedic surgery is necessary to replace painful, damaged joints.

Patients who experience nerve pain should discuss which treatment, including medications, may be right for you.

Bone Complications

As discussed in the last chapter, osteoporosis (weakening of the bones) and osteopenia (bone loss) are common among type 1 Gaucher patients. If you are diagnosed with osteoporosis, your physician may prescribe medication to help strengthen your bones and prevent further weakening.

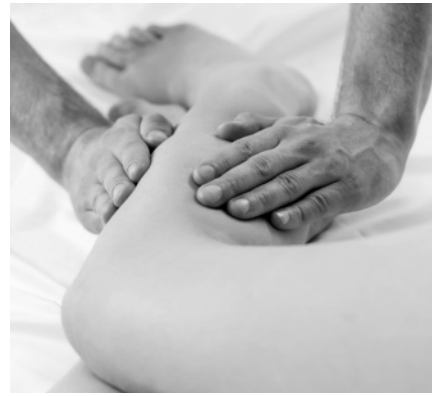
In more severe cases, type 1 Gaucher may lead to avascular necrosis, a condition caused by lack of blood flow to the bone tissue that eventually results in deterioration and possible death of these tissues. Reduced blood flow to bone tissue can cause acute, disabling bone pain, referred to as "bone crises." A classic Gaucher-related bone crisis is most often defined as a period of severe, intense pain in a very specific location, often in the hip or femur. Patients who experience bone crises should discuss pain management options with their physician.

Complementary Treatments

Additional, non-pharmacologic treatment options may help manage pain, including:

- **Physical therapy:** The Centers for Disease Control and Prevention recommend physical therapy for the management of most types of painful conditions. While medications mask the sensation of pain, physical therapists treat the source through movement, hands-on therapy and education. Physical therapists can prescribe specific exercises to build strength, improve flexibility (range of motion) and relieve stiffness (frozen joints). Meanwhile, manual therapy can relieve pain by reducing inflammation, stiffness, and soreness through manipulation and massage. This type of treatment also works to help the body heal itself by encouraging the production of natural pain-relieving chemicals and increased blood flow.
- **Exercise:** Though it may be difficult to engage in physical activity when you're experiencing pain, it's important for patients to keep moving. As mentioned, a physical therapist can recommend exercises that will be most beneficial and safe for your particular complications. However, even taking a daily walk can improve joint stiffness and pain.

- **Massage therapy:** Many type 1 Gaucher patients find that massage helps alleviate their pain. Massage increases blood flow to sore, stiff joints and muscles. Studies show that massage can also trigger the release of natural painkillers in the brain and increase the flow of oxytocin, a hormone that relaxes muscles and encourages feelings of contentment. It has been shown to ease chronic pain in a variety of conditions, as well as reduce anxiety.



- **Nerve stimulation:** This treatment involves tiny electrical pulses to reduce pain. Transcutaneous electrical nerve stimulation (TENS) uses small electrodes or devices that conduct electricity on the skin over the area of the body that's in pain. The electrodes are attached to a machine that releases small waves of electricity, sending tiny electrical impulses to the painful area. These impulses are thought to interrupt pain signals or messages from the brain. TENS may also encourage the body to produce more endorphins, which are natural pain relievers.
- **Acupuncture:** While this technique has been used in Asia for centuries to treat many conditions and relieve pain, it's now becoming more prevalent in the U.S. and other Western countries. It involves the insertion of very fine needles into the skin at specific "acupoints," which relieve pain by releasing endorphins, the body's natural painkilling chemicals. Acupuncture also affects the part of the brain that controls serotonin, a chemical that improves our mood. There are different types of acupuncture, as well as acupressure — a similar technique that uses deep manual pressure instead of needles. Although some people report great success, evidence is mixed on whether these techniques work well to relieve pain. If you choose to try this approach, be sure to find an experienced, licensed practitioner. (Look for a practitioner with a certification from the National Certification Commission for Acupuncture and Oriental Medicine — www.nccaom.org)

Important Note: Before beginning any complementary therapies or taking any vitamins or supplements for pain relief, consult with your Gaucher specialist or primary care physician.

CONDITIONS RELATED TO GAUCHER DISEASE[1]

Having type 1 Gaucher disease may increase your risk of developing other disorders (referred to as co-morbidities), including Parkinson's disease and certain cancers of the blood and liver. While most patients do not develop Parkinson's disease or cancers related to Gaucher disease, patients should understand the potential risks:

- Research suggests that patients with Gaucher disease have a slightly increased risk of developing Parkinson's disease later in life. Specifically, individuals older than 60 have a 2% to 4% chance of developing Parkinson's disease, which increases to approximately 5% by age 70 and 8% by age 80.
- Studies show that Gaucher disease carriers have a 3 percent risk of developing Parkinson's disease by age 70, which increases slightly by age 80.

Research also shows that careful testing and imaging studies can identify early-onset Parkinson's disease in patients with Gaucher. Prompt diagnosis is important because some patients may benefit from Parkinson's disease medications.

Gaucher disease may also increase your risk of developing certain cancers, including:

- Blood cancers, such as myeloma and lymphoma.
- Liver cancer (hepatocellular carcinoma). It is not yet known if treatment for Gaucher reduces the risk of developing this type of cancer.

Because type 1 Gaucher patients have an increased risk of developing these conditions, it's important to proactively monitor your health. Patients should receive special screening and blood tests to identify and treat problems early.

[1]Source: National Gaucher Foundation - www.gaucherdisease.org

SHOULD YOU CONSIDER A PAIN CLINIC?

If you or your child is suffering from chronic pain due to arthritis or bone involvement caused by type 1 Gaucher disease, you may want to consider working with a pain management specialist or pain clinic. These centers typically take an integrative approach to pain management that may include injections to deal with specific areas of pain, non-narcotic medications, physical and behavioral therapy, nerve blocks, biofeedback, cognitive behavioral therapy, hypnosis, water therapy and more.

A 2009 issue of *Baylor University Medical Center Proceedings* evaluated data from 108 people and found that after 4 weeks of this kind of comprehensive pain management care, patients saw improvement in pain, emotional distress and function. The approach was most effective when therapies were tailored to a patient's individual needs.

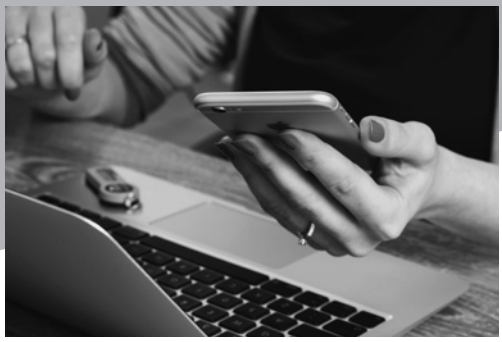
Because many drugs that treat pain can be addictive and only mask the problem, these complementary therapies are becoming more popular. In fact, in some cases, narcotics can increase pain because the medications change the way the body's endorphin system works.

Experts in pain management say pain clinics are most helpful when they encourage people to become active partners in their pain relief by focusing on self-management techniques, such as adopting an anti-inflammatory diet, doing low-impact exercises, developing a support system, and making self-care a priority.

If you are considering a pain clinic, your primary care physician may provide a referral, or you can do some research on your own. When you call or visit the clinic, be sure to ask:

- What kinds of therapies and treatments do you offer? (You want the answer to be wide ranging and more than just oral and injectable medications.)
- Do you have physical therapists, occupational therapists and psychologists at your clinic?
- What non-drug treatments, such as cognitive behavioral therapy, meditation, physical therapy and occupational therapy, do you offer?
- Are your pain doctors board-certified, trained in fellowships and accredited?
- What type of conditions do you commonly treat?
- Do you organize online and in-person patient support groups?
- Can I speak with patients with arthritis and related diseases who have completed your program?

Source: The Arthritis Foundation: https://www.arthritis.org/living-with-arthritis/pain-management/chronic-pain/pain-clinic.php



When it comes to managing symptoms and complications, as well as early diagnosis and treatment of related conditions, working closely with a Gaucher specialist is key. He or she will track important health indicators, such as spleen and liver volume, bone density and blood counts. Monitoring these indicators is necessary for managing medications, tracking changes in your health, and avoiding further complications. Keeping good records, which we'll discuss in the next chapter, can also help you and your physician on this journey.

FAMILY TO FAMILY

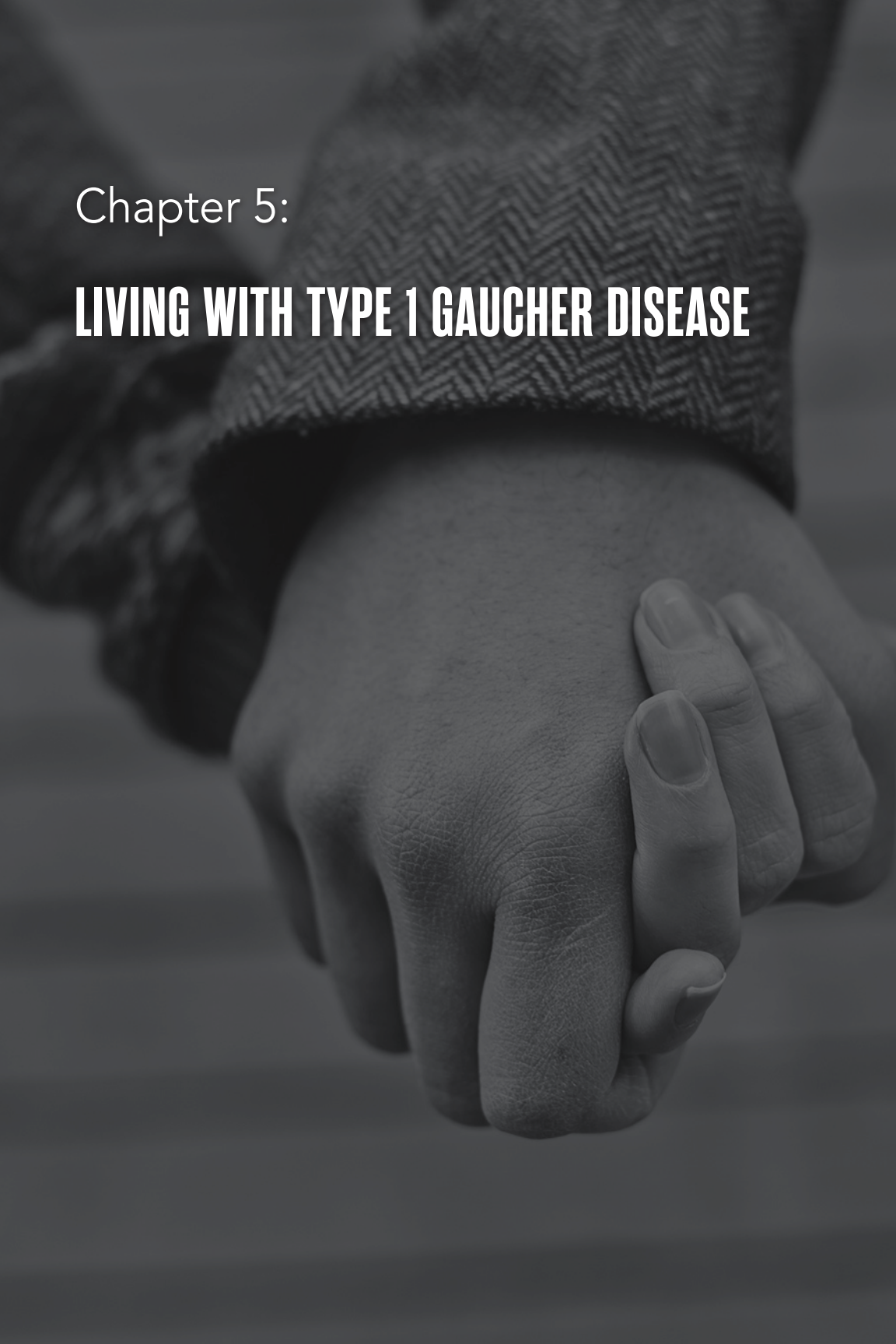
"We have two people in our family with type 1 Gaucher – my uncle and my son, Josh. When Josh was 4 years old, he complained of "tummy pain" and had a slightly extended abdomen. Because of my uncle's diagnosis, my first thought was Gaucher. Sure enough, Josh had an enlarged spleen, and a blood test confirmed that he had the disease.

I had seen my uncle go through some pretty scary, serious complications over the years, including removal of his spleen and orthopedic surgery to replace damaged hip joints. So, I was naturally very concerned for my son. However, I quickly learned that treatments for type 1 Gaucher have improved a lot since my uncle was diagnosed. Josh was started on therapy almost immediately and his condition has improved. He's an active, normal kid, who likes to play baseball and is on the swim team.

Josh has had some bouts of fatigue and some bone and joint pain. He needs to be monitored regularly to make sure the disease isn't progressing or causing bone/joint damage, and we make sure he eats well and gets proper rest, which isn't always easy with a kid! I mean what 9-year-old likes to eat spinach and go to bed on time?

Josh has found physical therapy to be very helpful, especially since he plays sports. His therapist has helped him improve his flexibility, and relieve sore muscles and joints, which helps him stay active."

Lena – Mother of a 9-year-old son with type 1 Gaucher disease



Chapter 5:

LIVING WITH TYPE 1 GAUCHER DISEASE

LIVING WITH TYPE 1 GAUCHER DISEASE

Living with a chronic illness can be difficult, but having a rare disease like Gaucher can present additional challenges. Very few people know about the disease or the symptoms and complications patients are likely to experience, which is why communication is an important part of coping effectively with type 1 Gaucher.

Whether you are informing family and friends, talking to your child's teachers, educating employers and colleagues, or working with medical specialists, communicating is essential. These conversations can't eliminate symptoms, but they can make your life easier! Effective communications can also help you receive the support you need – from getting quality care, to maintaining bonds with the important people in your life.

The first step in this process is to educate yourself about type 1 Gaucher (reading this book is a good first step!). Information can be empowering – helping you and your family manage the long road ahead. It can also help you communicate more effectively. Keep in mind, that learning about Gaucher is a continual process: Patients and caregivers find that they continually learn new things as they talk to other patients/parents, join support groups, and seek advice from specialists.

COMMUNICATING WITH OTHERS

Informing Family and Friends

Telling loved ones about this diagnosis can be both therapeutic and exhausting. Repeating the same information to multiple people can be emotionally draining, and yet, sharing your concerns can also be an important part of the coping process. It's natural for family and friends to have lots of questions and worries of their own. There is no right way to tell loved ones about your or your child's diagnosis, but it's important for people who are close to you to hear the news and become educated on what to expect. Experts at Capital Caring, which serves people living with advanced illness, suggest the following options for telling others:

- Tell one trusted family member or friend, and ask that person to share the news with others. You might even prepare "talking points" or notes for them to use.
- Meet with family members and friends individually to discuss the basics, and provide written information for them to review for more details.
- Hold a family meeting to explain the news so you don't have to repeat yourself multiple times.

- Ask a doctor, genetic counselor, nurse or social worker to talk to your family or to be present when you do. Many of these professionals are experts at explaining a diagnosis and answering questions. He or she may be able to help prepare the "talking points" or other materials, as well.

Expect everyone you inform to react differently. Some people might be emotional, some shocked or uncomfortable, and others will leap into action to try to assist. When people ask what they can do to help, take them up on their offer. If you don't know what they can do to help initially, tell them you'll get back to them when things settle down. Accepting support will not only lift some of the burden off your shoulders, it will allow loved ones to feel as though they've contributed in a positive way.

Talking to Teachers and School Staff

If your child has type 1 Gaucher disease, it's important for the school staff to be aware of his or her condition and the type of symptoms and/or complications to be expected. As a parent, you want your son or daughter to be safe and well-cared for at school, but also treated as normally as possible. To ensure academic success, social happiness and proper care, you need to do some planning and be actively involved.

The hope of treatment for type 1 Gaucher disease is that it restores the lives of affected people to as "normal" as possible. In fact, many children have fewer symptoms of the disease with therapy and may receive home infusions at times that don't interfere with school. In this case, it may not be necessary to have many accommodations made at school. However, initially this may not be the situation, and adjustments may be needed for infusions or other disease-related complications.

While some parents may choose a private school or homeschooling, there is an advantage to public education. Public schools must comply with Section 504 of the Rehabilitation Act of 1973, which is a federal law that ensures that those with disabilities or special needs are not discriminated against. The law states that children with disabilities, including medical conditions, need to be given accommodations so they receive an education comparable to that of other children who do not have disabilities.

To put the 504 plan or IEP (individualized education plan) in place, parents must schedule a meeting with the school personnel who oversee these plans, and outline the considerations that are applicable to the child's particular needs. Keep in mind that negotiating the details of your child's plan can take time (up to several months) and involve a good deal of education on your part.

You will also need to meet with the school principal, counselors and teachers to explain your child's disease and its potential impacts, such as frequent absences due to infusions, fatigue or pain; problems concentrating due to bouts of fatigue; restricted activity due to bone issues; and possible side effects of treatment. Remember, because Gaucher is a rare disease, it's highly likely that your school's staff will not be familiar with the condition or its complications. Be prepared for that meeting by bringing as much information with you as possible.

When it comes to working with school staff, experienced parents offer this advice:

- Request instead of demand. It's important to get off on the right foot. You want them to feel positively toward you and your child and develop an encouraging attitude. Developing long-term personal relationships with school personnel is important for your child's success.
- Keep communications open. School staff should be able to contact you easily by phone or email throughout the day with questions or concerns.
- Stay calm when problems arise. There are bound to be issues at some point. Getting upset over things will only make the situation worse. Focus on finding a solution together. Preface your requests with "For my child's safety…" rather than making accusations or demands.
- Be patient, but firm. It's common to have a "rough start" as everyone learns how to cope with a new situation and learn how to handle specific needs. Be reassuring and helpful, but don't be afraid to stand firm on the issues that are most important to you.
- Be flexible. No plan is cast in stone. As symptoms and complications arise, or treatments change, you may have to adjust your expectations.

Developing good relationships with the school staff is not only important for your child's well-being and academic success, but also for your peace of mind.

Discussing Your Condition with Employers and Colleagues

If you're the one with type 1 Gaucher, the conversations are more likely to be with an employer and colleagues. While you're entitled to your privacy by law, and therefore, not obligated to disclose your disease, it's a good idea to inform the people you work with so they can be aware of your challenges and provide support. This is especially true if your symptoms and complications could interfere with your performance. It's better if your employer understands the reasons for your behavior, and in most cases, he or she will appreciate your honesty.

Depending on your employer, this can be a stressful decision. You may be worried about maintaining your job and supporting yourself or your family. Unfortunately, protections for chronically ill workers in the U.S. are somewhat vague. However, there are two laws to be aware of. First, the Family and Medical Leave Act allows employees to take 12 weeks off each year for medical or family emergencies, but without pay. Second, the Americans with Disabilities Act requires employers to make reasonable adjustments for disabled workers, often in the form of additional time off. It often comes down to the individual policies of employers, which may include short- and long-term disability plans and/or paid sick days. Before scheduling a meeting with your employer, you should educate yourself on the company's policies regarding medical conditions.

The next step is educating the people you work with. Again, because Gaucher is rare and often an "invisible" disease, you'll need to provide as much information as possible. Where you begin this process depends on the company. In some cases, companies have a health representative or human resources contact who specializes in this type of discussion. In smaller companies, you may go directly to your supervisor or manager.

When explaining your diagnosis, it's helpful to bring information from your doctor or pamphlets and other resources on Gaucher and its possible symptoms and complications. Points to cover may include:

- Adjusting your schedule to accommodate infusions if you are receiving ERT.
- Working from home (if your job allows) on days that you're in pain or fatigued.
- A possible increase in sick days due to fatigue or pain.
- Time off for physical therapy or other treatments, if necessary.
- Changing roles or responsibilities based on physical abilities.
- Training another employee to cover your responsibilities in your absence.
- The possibility of reduced hours or job sharing.
- Making accommodations to your job or work environment, such as sitting instead of standing. According to the Americans with Disabilities Act, your employer is required to make reasonable accommodations to your job or work environment. Even though your illness may be episodic or controlled by medications/treatments, it may still be considered a disability according to a recent amendment to the law. If you are not sure what type of accommodations you're entitled to or how to ask for them, contact the Job Accommodation Network at 800-526-7234.

Remember that you only need to give your employer information about how your condition may impact you on the job, so simply reveal what you're comfortable with. It's not necessary for you to disclose any specifics about your treatments or medications unless you choose to do so.

You may want to take notes or record your conversation (ask permission or check consent laws in your state first) to ensure clarity on both sides. It's also a good idea to know the laws in your state regarding sick leave. Sick leave is not mandated federally, but may be at the state level. You can find out more through the Equal Employment Opportunity Commission.

Keep in mind, protections may be available if an employer tries to terminate your employment, decrease pay, or change your position without your input based on your condition.

It's also your decision who else in the workplace needs to know about your illness. Be sure to set some boundaries with your direct supervisor as to who should be informed or if you prefer to guard your privacy with others. Disclosing your disease to one or two people does not mean that you want it to be common knowledge throughout the company. When informing colleagues, it's also up to you how much information you would like to share and how you would like it to be conveyed. Some people may want to hold a departmental meeting or send out an email to inform everyone who may be impacted, while others are more comfortable with one-on-one conversations.

Overall, it's important to remember that your health is a priority, and by law, your place of employment is required to work with you on appropriate accommodations for your illness, especially if it does not interfere with the quality of your work. Don't be afraid to make your employer aware of your needs, and don't feel the need to apologize – having type 1 Gaucher is not your fault.

If your symptoms prevent you from performing work entirely, you can apply for Social Security Disability insurance. Because this involves a great deal of paperwork and meeting certain requirements, you may want to consult with an attorney to help you with this process.

KEEPING A HEALTH DIARY

Individuals with type 1 Gaucher disease typically require the care of numerous specialists, which means you need to keep information regarding symptoms, treatments and medications organized. Keeping an ongoing health diary is the best way to do this. Health diaries can also help you:

- **Make your appointments more productive.** Physicians are only allotted a short period to see each patient. Being organized with information and questions will help you make the most of that time.

- **Reduce stress levels.** It's nearly impossible to remember everything, especially when you're dealing with multiple symptoms and a team of physicians. Jotting down notes in a journal or diary is a good way to keep track of your or your child's health, as well as questions that arise. For instance, if a new medication causes nausea, or fatigue has gotten worse before treatments, you may want to address that at your next visit.
- **Receive better treatment.** Keeping a diary of symptoms and complications will not only help you remember important details, but also provide the physician with the accurate information he or she needs to provide the best possible care. If, for example, fatigue is worsening, your treatment plan may need modification. Other changes in health, such as increased bone pain, may warrant additional scans that lead to early intervention.
- **Identify patterns.** Over time, a health diary can provide important clues to improve care. For example, you may note that certain foods interact negatively with medication or treatments, which can lead to beneficial dietary changes. Emerging patterns regarding fatigue or pain before treatments or after physical activity, can help you and your physician make positive adjustments. This is especially important in chronic diseases like Gaucher in which symptoms are likely to change over time.

Don't think of this diary as "just one more thing to do," but rather a helpful tool for you and your team of medical professionals. You don't need to go into great detail – simple notes will do. The important thing is to use a system that works best for you, whether that's a notebook or a digital diary. Be sure to make entries while the information is fresh in your mind.

You may think that keeping official medical records is the responsibility of physicians – and it is. However, maintaining your own records regarding physicians, treatments and medications is very important. Having this pertinent information on-hand and easy to access will not only make your life easier, it will help healthcare professionals monitor care more accurately and adjust treatments if necessary.

In addition, you will often find yourself answering the questions of one specialist with respect to other treatments and procedures (e.g., date of previous surgeries, current medications, etc.). Having your own records will make it simple to answer these questions and keep track of multiple therapies.

Some people find it helpful to use a binder with separate tabs for each specialist. The first page of each binder should contain the physician's name and specialty, along with contact information, including the after-hours answering service. The subsequent pages would consist of records of each visit – date of appointment, recommendations/notes, medications prescribed, test results, questions to discuss at the next visit and other details. For those who prefer electronic recordkeeping, this information can be easily stored on a mobile device or flash drive.

Sample Physician Record

Physician's Name: _____

Specialty:_____

Address: _____

Phone: _____

Answering Service: _____

Date of First Visit:_____ Reason: _____

Visit Date	Recommendations/ Notes	Medications Prescribed	Test/Results

FATIGUE AND PAIN SCALES

Fatigue Scale

Fatigue can be hard to define, because it involves more than simply "feeling tired." It can affect your ability to concentrate, reduce motivation to get things done, and make decisions difficult. Fatigue can also range in severity, from requiring more rest after physical activity, to being completely debilitating. As you're keeping notes in your health diary, it can be helpful to use the following "Fatigue Impact Scale" to measure the effect that fatigue is having on your or your child's daily life.

Fatigue Scale

Score each of the following statements using the number that best describes how you feel:

Never = 0 Rarely = 1 Sometimes = 2 Often = 3 Almost Always = 4

Score:	1. I have been less alert.	Score:	2. I have had difficulty paying attention.
Score:	3. I have been unable to think clearly.	Score:	4. I have been clumsy and uncoordinated.
Score:	5. I have been forgetful.	Score:	6. I have had to pace myself in my physical activities.
Score:	7. I have been less motivated to do anything that requires physical activity.	Score:	8. I have been less motivated to participate in social activities.
Score:	9. I have been limited in my ability to do things.	Score:	10. I have had trouble maintaining physical effort for long periods.
Score:	11. I have had difficulty making decisions.	Score:	12. I have been less motivated to do anything that requires thinking.
Score:	13. My muscles have felt weak.	Score:	14. I have been physically uncomfortable.

Score:	15. I have had trouble finishing tasks that require thinking.	Score:	16. I have had difficulty organizing things.
Score:	17. I have been less able to complete tasks that require physical effort.	Score:	18. My thinking has been slowed down.
Score:	19. I have had trouble concentrating.	Score:	20. I have limited my physical activities.
Score:	21. I have need to rest more often or for longer periods of time.	Final Score:	

Points: 0-84
Physical: 0/36
Cognitive: 0/40
Pyschosocial: 0/8
Total = Physical + Cognitive + Psychosocial

Source: MDCalc, Modified Fatigue Impact Scale (MFIS)

This is a very comprehensive scale, which can provide useful information during an initial consultation. However, there is a shorter, simpler version called the "Fatigue Severity Scale," which can be found below and downloaded at: http://geriatrictoolkit.missouri.edu/fatigue/Fatigue-Severity-Scale.pdf.

Fatigue Severity Scale

Please circle the number between 1 and 7 that you feel best fits the following statements. This refers to your usual way of life within the last week. 1 indicates "strongly disagree," and 7 indicates "strongly agree."

Read and circle a number. Strongly Disagree → Strongly Agree

1. My motivation is lower when I am fatigued.
   1 2 3 4 5 6 7

2. Exercise brings on my fatigue.
   1 2 3 4 5 6 7

3. I am easily fatigued.
   1 2 3 4 5 6 7

4. Fatigue interferes with my physical functioning.
   1 2 3 4 5 6 7

5. Fatigue causes frequent problems for me.
   1 2 3 4 5 6 7

6. My fatigue prevents sustained physical functioning.
   1 2 3 4 5 6 7

7. Fatigue interferes with carrying out certain duties and responsibilities.
   1 2 3 4 5 6 7

8. Fatigue is among my most disabling symptoms.
   1 2 3 4 5 6 7

9. Fatigue interferes with my work, family, or social life.
   1 2 3 4 5 6 7

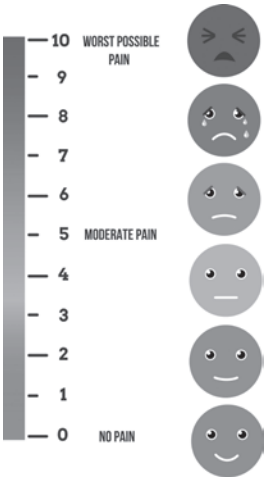
Visual Analogue Fatigue Scale (VAFS)

Please mark an "X" on the number line that describes your global fatigue with 0 being worst and 10 being normal.

0 1 2 3 4 5 6 7 8 9 10

Pain Scale

Pain is one of the hardest symptoms to quantify and describe to others. It is very personal – everyone feels pain differently. And, there is no test or gauge to show how intense the feelings are. Besides asking where it hurts, physicians will often have you rate your pain on a scale of 0 to 10, with 0 being "no pain," and 10 representing "the worst pain imaginable." Using this type of scale, especially one with cartoons or graphics (i.e., smiling faces vs. crying faces), is particularly helpful for young children to communicate with physicians.



Of course, pain scales can still be subjective. A person with a high tolerance for pain might describe their pain as a 2 or 3 on the pain scale, while another would describe the same pain as a 6 or 7. Beyond assigning a number, it's important for your physician to get a good sense of your chronic pain, such as how the pain hurts: Is it a stabbing pain or a constant ache? Does it throb or is it a shooting pain? Would you describe it as burning or buzzing? This type of information can help pinpoint the cause. Variations are equally important: How does the pain change during the day? What makes it feel worse? What makes it hurt less? When talking to your physician or pain management specialist, you should be prepared to answer these types of questions to the best of your ability.

Finally, physicians need to know how the pain is affecting your life and how it changes over time. Therefore, using a more detailed pain scale (see below) is often used to accurately assess and manage pain.

Global Pain Scale

Please indicate your level of pain by circling a number from 0 to 10.

Your Pain:

My current pain is:
No pain: 0 1 2 3 4 5 6 7 8 9 10 :Extreme pain

During the past week, the best my pain has been is:
No pain: 0 1 2 3 4 5 6 7 8 9 10 :Extreme pain

During the past week, the worst my pain has been is:
No pain: 0 1 2 3 4 5 6 7 8 9 10 :Extreme pain

During the past week, my average pain has been:
No pain: 0 1 2 3 4 5 6 7 8 9 10 :Extreme pain

During the past 3 months, my average pain has been:
No pain: 0 1 2 3 4 5 6 7 8 9 10 :Extreme pain

Your Feelings: During the past week I have felt:

Afraid:
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

Depressed:
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

Tired:
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

Anxious:
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

Stressed:
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

Your Clinical Outcomes: During the past week:

I had trouble sleeping:
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

I had trouble feeling comfortable :
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

I was less independent:
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

I was unable to work (or perform normal tasks):
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

I needed to take more medication:
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

Your Activities: During the past week I was NOT able to:

Go to the store :
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

Do chores in my home:
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

Enjoy my friends and family:
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

Exercise (including walking):
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

Participate in my favorite hobbies:
Strongly Disagree: 0 1 2 3 4 5 6 7 8 9 10 :Strongly Agree

Scoring: Add up the total score and divide by 2. Each subset is worth 25 points. The maximum total score is 100.

MANAGING MEDICATIONS

It's not unusual for Gaucher patients to be on multiple medications for a variety of health conditions. Modern medications can be extremely beneficial when taken properly, but failing to take them as directed can result in serious problems. According to estimates by the U.S. government, more than 125,000 people die each year due to improper use of medications, and many more are hospitalized. Improper use may include forgetting or skipping doses, taking more than directed, ingesting medication on an empty stomach when it should be taken with food or vice versa, or stopping medication due to unpleasant side effects without talking to your physician.

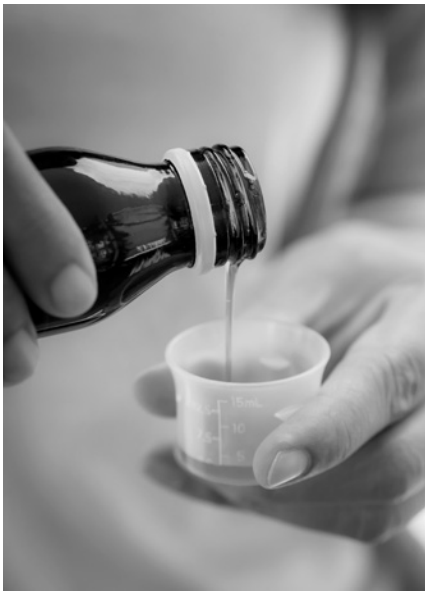
To prevent these issues, consider the following tips:

1. When you or your child is prescribed a new medication, make sure you understand what the medication is and what it is being prescribed for. Other questions to ask include: How does it work? How many times is it taken each day and at what intervals? Should it be taken on an empty stomach or with food? Are there any dangerous interactions with other drugs (including OTC drugs, vitamins and supplements), or certain foods/drinks? What are the possible side effects? How long will it need to be taken?

2. Use an organized pill box with separate sections for each day of the week. Some pill boxes also have separate sections for AM and PM. Fill the box up at the beginning of each week, making note of any medications that need to be refilled.

3. Many children's medications come in liquid form. Liquid medicines are generally easier to swallow, but measurement is important. Always check the label for dosage instructions and keep the dosing device

together with the bottle. Be sure to check whether the medication needs to be refrigerated. If not, the best place to keep liquid medications is on a counter where you'll easily remember it (but out of direct sunlight and away from heat sources). Always use the appropriate dosing device for exact measurement (i.e., dropper, syringe, medicine cup or dosing spoon). Don't mix dosing devices for different products, which may cause you to administer the wrong amount. Don't just fill the dropper or cup — look carefully at the lines and numbers to dispense the correct amount.



4. For medications that must be taken at the same time each day, set an alarm for the scheduled time. There are several medication alarms available, but many people find setting an alarm on their smartphone works just as well. Also, taking the medication with the same meal each day or making it part of your or your child's morning or bedtime routine are good ways to avoid forgetting a dose.

5. If you know you're going to be out of the house, don't forget to bring your medication in a travel case so it can be taken at the appropriate time. If the medication needs to be taken with food, be sure to bring a snack.

6. Never discontinue a medication or change dosages without consulting your physician.

7. Make a list of medications and keep it updated. Keep a copy of this list at home, as well as in your purse or wallet. This list will come in handy when seeing a new physician or during an emergency. One of the first questions emergency personnel ask is, "What medication is the patient taking?" Simply handing them a list, instead of trying to remember a long list of complicated medication names and dosages, makes an already stressful situation much easier — EMS staff and nurses will also appreciate the convenience.

Medication Log

PATIENT:

1. ALWAYS KEEP THE MEDICATION LOG WITH YOU. You may want to fold it and keep it in your wallet along with your driver's license. Then it will be available in case of an emergency.
2. Take this log to ALL doctor visits, tests and ALL hospital visits.
3. WRITE DOWN ALL CHANGES MADE TO YOUR MEDICINES on this log. If you stop taking a certain medicine, draw a line through it and write the date it was stopped. If help is needed, ask your doctor, nurse, pharmacist or family member to help you to keep it up-to-date.
4. In the REASON column, write down the name of the doctor who told you to take the medicine(s). You may also write down why you are taking the medicine (Examples: high blood pressure, high blood sugar, high cholesterol).

HOW DOES THIS LOG HELP YOU?

1. This log helps you and your family members remember all of the medicines you are taking.
2. Provides your doctor(s) and others with a current list of ALL of your medicines. Doctors need to know the herbals, vitamins, and over-the-counter medicines you take!
3. Concerns may be found and prevented by knowing what medicines you are taking.

Medication Log

Personal & Medical **CONTACT INFORMATION**	Date:

Personal Information	
Name	
Address	
Phone	
Date of Birth	
Emergency Contacts	Name (Phone)
	Name (Phone)

List all Medicines, Vitamins and Supplements

Name (Brand)	Start Date	Stop Date	Dosage	Frequency	Reason or Dr. Name

Living the best life possible with type 1 Gaucher involves more than just managing symptoms and complications, it requires good communication with the people in your life – from family and friends, to school staff and colleagues, and perhaps most importantly, your medical team. Being organized can not only improve communication, but also reduce stress – which we could all use less of, especially when dealing with a chronic illness.

FAMILY TO FAMILY

"With four busy kids and two full-time jobs, I didn't think life could get more hectic for our family. But, our lives changed forever when our youngest daughter, Emma, went in for her 2-year physical. She had been very lethargic and 'out of sorts.' It's hard for a toddler to describe how they're feeling, but my wife and I knew something was wrong.

The doctor noticed her pot belly, which we just assumed was baby fat, but was actually an enlarged spleen. He ordered a scan and a series of blood tests. At first, she was misdiagnosed with leukemia, but after being referred to a hematologist, we learned that she had type 1 Gaucher – a disease that we had never heard of. We have no family history of this disease and we aren't part of the ethnic group that is at highest risk, so it was quite a shock to learn that she had this rare disease.

When we finally saw the Gaucher specialist, he recommended that we have our other three children tested. Our second oldest son, Ethan, was also diagnosed with type 1 Gaucher. He was 8 years old at the time and had never shown any symptoms, so this was shock number two!

Both Emma and Ethan began individualized treatments. They are both doing well, but Emma shows more signs of the disease, including occasional joint pain. It's really hard to see your child in pain, but we've received a lot of support from our medical team, as well as other families dealing with Gaucher. We've also found that physical therapy and massage have helped to ease Emma's discomfort.

Once we got over the initial shock and got a handle on treatments, we began to focus on living life as normally as possible. We still have weekly game night and the kids are still busy with activities. Because of Emma's infusions and other medical appointments, we've come to rely on other family members to help us out, and are truly thankful for their love and support.

We've also learned the importance of being organized! We keep a health diary for both Emma and Ethan, including medication charts and treatment records. With so much going on, this gives us some much-needed peace of mind."

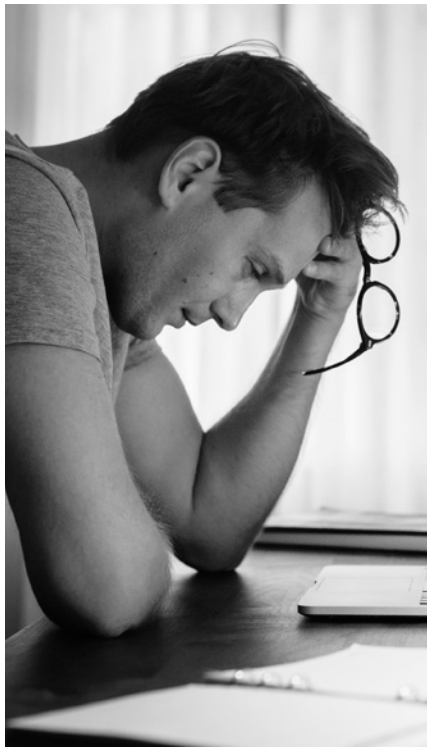
Joseph – Father of two children with type 1 Gaucher

Chapter 6:

NAVIGATING HEALTH INSURANCE WITH A CHRONIC CONDITION

NAVIGATING INSURANCE WITH A CHRONIC CONDITION

Living with a chronic illness is difficult enough without navigating a complicated healthcare system. Understanding insurance plans, industry terminology, coverage, and complex forms can be a daunting task, but there are some tips to help guide you through this process. The more you know, the more effective you can be at advocating for yourself and getting the best care (and coverage) possible.



The process of choosing the right insurance plan can be challenging for anyone, but for those living with a rare disease such as Gaucher, it often becomes overwhelming. There are special considerations such as making sure your Gaucher specialist is within your plan's network and ensuring that specialty medications and treatments, such as infusions, are covered.

For example, some specialty pharmacies that provide ERT and SRT drugs require two prescriptions – one from your physician and another on their own form. Also, insurance companies may have restrictions on where you can receive infusions (hospital vs. home). The goal of this chapter is to help you understand the basics and find your way through an often-confusing maze of information.

Throughout the process, it's important for patients to have a voice as it relates to their access to treatment and overall care.

THE PATIENT CHARTER

1. The patient experience is at the heart of medicine; thus, the patient must be at the center of all medical decision making.
2. The medical process should stay between the patient and their care provider.
3. The patient should have access to all treatments deemed appropriate by their care provider.
4. Access to care should not be limited by external forces, financial or otherwise.
5. Patients should be empowered and educated with the tools needed to make their voices heard.
6. Elected officials, insurance providers, drug manufacturers, and all those associated with the healthcare system shall make it their goal to ensure the patient is the focus of all decisions.
7. The medical team should strive not only to do no physical harm, but to do no emotional, mental, or financial harm to the patient.
8. Patients should be treated with dignity, transparency, and respect by everyone involved in the healthcare process.

Source: The Patient Charter was adopted by the CreakyJoints Patient Council during its annual in-person meeting in Chicago, Illinois in 2016. Reprinted in "A Patient's Guide to Healthcare," by the GLHF, 2018.

UNDERSTANDING THE BASICS

According to studies, 90% of Americans have chosen the wrong insurance plan based on their family situation. It's no wonder, when you consider how confusing the information can be! Whether you're covered by an employer-based healthcare insurance plan, individually-purchased healthcare insurance, Medicare or Medicaid, there are some basic terms to understand as you enroll in or reevaluate a plan that can help you make a more informed decision:

1. **Premiums** – This refers to the fee or cost of purchasing a plan. It's typically paid monthly. There are two main types of insurance plans: those with high premiums and low deductibles; and those with lower premiums and higher deductibles. It's important to look closely at how much you typically spend on health care when deciding which type of plan is best for you and your family.

2. **Deductibles** – This is the amount you must pay out-of-pocket before the insurance plan takes over and provides coverage. So, for instance, if you have a deductible of $2,000, you must pay for all treatments, medications and other health services until you reach that amount. Once you've spent $2,000, the insurance plan will cover all or a certain percentage of (depending on your plan) further healthcare costs. Again, you may initially be "turned off" by high-deductible plans, but it's important to map out what you spend over a year – you may be better off with a high-deductible plan.

   When deciding on a plan, one thing to consider is that Health Savings Accounts (see #6) allow you to pay your deductibles with pre-tax dollars. Many insurance plans will provide a simple chart outlining the type of plans to choose from, but experts advise that these charts are often "too simplified." Comparing plans can involve a lot of math, as well as additional information on what is covered and what is not, and how you use healthcare services. If you are having difficulty sorting out the details, take advantage of professional services. Community centers, online "insurance help" and customer service representatives can help you make sense of these plans and guide your choice. If you're employed, an HR specialist can be a good resource. It's important to understand these plans fully, because you can often realize substantial cost savings by choosing the one that's right for you. Don't be afraid to ask for help!

3. **Co-Pays** – These are the costs you incur and must pay out-of-pocket for various services, such as office visits, specialist visits, lab tests and diagnostic tests. In some plans, these are set fees, while others require you to pay a percentage of the cost. In some cases, once you reach your deductible, co-pays are covered by your plan. If you see a number of specialists, which many Gaucher patients do, it's important to look at the co-pays for these visits when deciding on a plan.

4. **In- and Out-of-Network** – Most insurance plans provide better coverage for services provided by physicians in their network of providers. Some plans have gone a step further and added "Preferred In-Network," which are covered at the highest rate; "Regular In-Network," which are covered at a lower rate; and "Out-of-Network," which may not be covered at all. Because your primary-care physician (PCP) typically plays an important role in coordinating your overall care, it's important to make sure your PCP is in your insurance network. When selecting a plan, also be sure your Gaucher specialist and other specialists that you've been working with are

part of the plan's network for the best coverage. It's often very difficult for Gaucher patients to switch physicians due to the rarity of this disease. So, if you must pay out-of-network fees to keep your preferred specialist, be sure to calculate these costs when determining which plan is most cost-effective.

5. **Flexible Spending Accounts (FSAs)** – These accounts are offered by some employers to help pay for healthcare costs. You select a percentage of your pre-tax earnings to set aside in a special account that can only be used for healthcare costs. While useful, these accounts can be cumbersome, as they require you to submit receipts for reimbursement. Also, if you don't spend the money in these accounts by year-end, you lose it.

6. **Health Savings Accounts (HSAs)** – These accounts are a newer and often better option to FSAs. With HSAs, you can set aside pre-tax earnings through your employer or on your own (through a bank or credit union). You must have a low-premium/high-deductible plan to open one of these accounts, as the funds are used to cover your deductibles. Some employers and banks offer the convenience of using a special debit card to pay for health services, eliminating the need to submit receipts. The main benefit of these plans, however, is that funds roll over – you don't lose the money you've set aside. In fact, some plans allow you to save the money in these accounts until retirement. There is often a maximum amount allowed each year. If you work with an accountant or financial planner, you may want to discuss these plans.

7. **Medicare** – Formed in 1965, this program covers people age 65 and older, along with those with certain disabilities. There are many options within Medicare:
   a. Part A – Covers hospital services
   b. Part B – Covers physician services
   c. Part C (Advantage) – Allows participants to choose coverage from insurance companies that are contracted by the government to provide more comprehensive coverage.
   d. Part D – None of the above covers everything, so many people choose to purchase supplemental coverage through various insurance companies. Depending on which plan you choose, there are many different levels of coverage.

Again, as each plan is different, it requires a lot of research and comparison to make a good decision. Some plans within Medicare are like an HMO, which offers preventive care, while others have more restrictions regarding specialists. For more information, as well as resources to help you understand these choices, go to www.medicare.gov.

8. **Medicaid** – While Medicare is run by the federal government, Medicaid is a state-run program. It's funded by both federal and state governments, but each state has a different program. Medicaid is typically available for pregnant women, children, people with disabilities and those with limited financial means. Medicaid is considered a "payer of last resort,' meaning if you're eligible for any other insurance, the provider will seek payment from Medicaid last. To learn more, go to the specific Medicaid website for your state.

CASE MANAGERS AND PATIENT ASSISTANCE PROGRAMS

As mentioned, for people with rare diseases, such as Gaucher, getting the proper coverage takes on heightened meaning. ERT and SRT require expensive drugs, which must be obtained from specialty pharmacies. Insurance plans have different requirements regarding how to obtain these medications. In addition, certain plans may have restrictions on where patients receive infusions. Therefore, if you prefer to have infusions done in your home, it's important to check with your insurance plan as to whether these are covered – many only cover infusions done in a hospital setting.

Because Gaucher patients receive such specialized treatments, the pharmaceutical companies that provide these medications typically provide patient resources, such as "case managers," who are representatives that help patients with financial support and medication delivery. For instance, if you are having trouble getting your insurance company to pay for SRT, dedicated case managers can help you understand your coverage and advocate on your behalf. They may also offer suggestions about which type of coverage is best for you and how to navigate numerous insurance plans. If you are working with a pharmaceutical case manager, it's recommended that you consult him or her before selecting an insurance plan.

Additionally, when you're prescribed any new medication, be sure to ask your physician or nurse about any co-pay assistance, coupons or rebates that may be available through the pharmaceutical company. You may also check the pharmaceutical company's website for information on assistance.

Even with health insurance, paying for Gaucher disease treatments can be a challenge for many families. Fortunately, there are resources available to help ease your burden. Patient Assistance Programs are designed to help patients with co-pays, deductibles, medication, travel expenses and other costs, depending on a family's income and financial need. Some examples of these nonprofit programs include:

- **The National Gaucher Care Foundation (NGCF)** – This program provides financial assistance to people with Gaucher disease and their families who are experiencing financial hardship. For those who qualify, the NGCF can help with insurance premiums, infusion charges that are not covered by insurance, travel expenses, OTC medications prescribed for Gaucher disease and related therapies, as well as emergency services.
- **NeedyMeds** – A nonprofit information resource that helps people find assistance affording their medication and related healthcare costs.
- **The National Organization for Rare Disorders (NORD) RareCare℠** – This program helps patients get lifesaving or life-sustaining medication they cannot otherwise afford. They provide:
  - Medication
  - Financial assistance with insurance premiums and co-pays
  - Diagnostic testing assistance
  - Travel assistance for clinical trials or consultation with disease specialists
- **The Patient Access Network (PAN) Foundation** – is an independent nonprofit organization dedicated to helping insured people who have chronic, critical and rare diseases. They provide financial support for out-of-pocket costs for prescribed medications.
- **Patient Services Inc.** – provides financial support and guidance for qualified patients with specific, rare chronic diseases. They also offer free legal services for certain rare disease communities.

Even if you're not eligible to receive financial assistance, many of these programs have professionals on staff who can help you make sense of insurance programs (i.e., what's covered and what's not, which specialists are in-network, restrictions on treatments, etc.). It's a good idea to get these folks involved in the decision-making process right from the start.

In short, deciding on the right insurance plan can be overwhelming, but know that you are not alone. There are people available to help you make sense of these choices and select the plan that is right for you.

THE FOUR P'S OF DEALING WITH HEALTH INSURANCE

"A Patient's Guide to Healthcare," published by The Global Healthy Living Foundation, recommends the following tips when navigating the often-winding road of health insurance coverage:

1. **Persistence**

Let's face it, dealing with insurance companies can sometimes be difficult and frustrating. However, no matter how much red tape you're dealing with, it's important to be persistent. Because healthcare is very personal and the coverage decisions made by your insurer can have lifelong impacts on your health, you must continue to self-advocate for the treatments your physicians recommend. You can go to your insurer's website or speak directly to a customer service representative if you have questions or concerns. It may take several interactions to effectively deal with complex issues, which brings us to our second "P"…

2. **Patience**

Healthcare decisions are typically big and costly, which means they tend to move slowly. It can be very stressful on patients when trying to get coverage for a necessary procedure and having to wait days or weeks to receive an answer – unfortunately, this is often the case. To expedite the process, make sure the insurance company has all necessary paperwork from the physician (e.g., recommendations for treatment, prior authorizations, referrals to specialists, etc.). If you have questions about any paperwork you might need, start with your physician's office. If the paperwork has been submitted, call the claim support center of your insurance provider to verify that they have received the proper forms.

3. **Paperwork**

Insurance claims are formal requests to insurance companies to cover certain treatments under your policy. Unfortunately, these forms aren't standardized and can be quite complex, which may cause delays. If you have questions about any paperwork that must be submitted, start with your physician's office. There is usually a "claim specialist" in the office who is experienced with many insurance providers and their required forms. These professionals can be an invaluable resource! The next step is contacting the claim support center of your insurance provider.

4. Politeness

Filling out paperwork and making calls can take time and may be fraught with frustration, so it's important to take a deep breath, and remain calm and polite. Getting upset and yelling at representatives rarely helps the situation! The people you speak with on the phone are only authorized to do so much. While being persistent is necessary, being rude is not. In fact, treating these representatives with courtesy may motivate them to work a bit harder on your behalf.

(For more information, please see Resources: Health Insurance.)

GLOSSARY OF TERMS FOR PATIENT ADVOCACY

To help you understand your insurance options and work more effectively with healthcare professionals, "A Patient's Guide to Healthcare" also offers the following list of terms and definitions:

Acute Care: Medical care administered, frequently in a hospital or by nursing professionals, for the treatment of a serious injury or illness or during recovery from surgery. Medical conditions requiring acute care are typically periodic or temporary in nature, rather than chronic.

Ambulatory Care: Medical care rendered on an outpatient basis and which may include diagnosis, certain forms of treatment, surgery and rehabilitation

Ambulatory Setting: Medical facilities such as surgery centers, clinics and offices in which healthcare is provided on an outpatient basis.

Benefit: This refers to medical services covered by your health plan. This word is also used to describe your health plan in general. It can also mean payment received under a plan.

Carrier: Any insurer, managed care organization, or group hospital plan, as defined by applicable state law.

Chronic: In healthcare and insurance terminology, a chronic condition is one that is permanent, recurring or long lasting, as opposed to an acute condition.

Claim: A bill for medical services rendered, typically submitted to the insurance company by a healthcare provider.

Closed Benefit Formulary: A formulary is a list of prescription drugs the health plan covers. If the plan has a closed formulary, it only covers drugs that are on that list. It will not cover any part of the cost of non-formulary drugs. However, in some instances, a plan may be willing to make an exception. To get one, you need to contact the plan and tell them why the drug is needed.

Coinsurance: This is the percentage of health care expenses you pay after your deductible. Your health plan pays the rest up to any benefit or lifetime maximum.

Copay: This is the dollar amount you pay for health care expenses. In most plans, you pay this after you meet your deductible limit. For example, you pay a set dollar amount to your doctor for an office visit. So, if your copay is $25, you pay that amount when you go to your doctor. In prescription drug plans, it is the amount you pay for covered drugs.

Deductible: The amount that the patient needs to pay before the insurance benefits activate.

Drug Utilization Review (DUR): The process by which health insurance companies evaluate or review the use of prescription drugs for appropriateness in the treatment of a patient.

Durable Medical Equipment (DME): Medical equipment used in the course of treatment or home care, including such items as crutches, knee braces, wheelchairs, hospital beds, prostheses, etc. Coverage levels for DME often differ from coverage levels for office visits and other medical services.

Dependent: A dependent (usually spouse or child) of an insured person who is eligible for insurance coverage

Drug Coverage (also covered benefits or covered expenses): These are services or supplies your health plan covers. They are eligible to be paid by your plan.

Drug Tiers: These are groups of different drugs. Usually, the plans group the drugs by price. Each group or tier requires a different copay. You might see the groups listed as generic, brand-name, or preferred brand-name drugs. Generic drugs often have lower copays. Brand-name drugs have higher copays.

Explanation of Benefits (EOB): This is a statement a health plan sends to a health plan member. It shows charges, payments and any balances owed. It may be sent by mail or e-mail.

Effective Date: The date on which health insurance coverage comes into effect.

Enrollee: An eligible person or eligible employee who is enrolled in a health insurance plan. Dependents are not referred to as enrollees.

Enrollment Period: The period of time during which an eligible employee or eligible person may sign up for a group health insurance plan.

Essential Benefits: The Patient Protection and Affordable Care Act (PPACA) requires all health insurance plans sold after 2014 to include a basic package of benefits including hospitalization, outpatient services, maternity care, prescription drugs, emergency care, and preventive services, among other benefits. It also places restrictions on the amount of cost-sharing that patients must pay for these services.

Formulary: This is a list of prescription drugs the health plan covers. It can include drugs that are brand name and generic. Drugs on this list may cost less than drugs not on the list. How much a plan covers may vary from drug to drug. An open formulary provides a greater choice of covered drugs. It is also called a "preferred drug list."

Formulary Exclusion List: This is a list of prescription drugs not covered by a health plan. It applies to closed formulary plans. If a member needs a drug on this list, the doctor must ask the plan to cover it as an exception. The plan will only do so if use is medically necessary.

Generic Drug: A drug that is exactly the same as a brand-name prescription drug, but that can be produced by other manufacturers after the brand name drug's patent has expired. Generic drugs are usually less expensive than brand-name drugs.

Grievance Procedure: The procedure by which a member or healthcare provider is allowed to file a complaint with a health insurance company and seek a remedy.

Group Health Insurance: A health insurance plan that provides benefits for employees of a business or members of an organization, as opposed to individual and family health insurance.

Inpatient: A term used to describe a person admitted to a hospital for at least 24 hours. It may also be used to describe the care rendered in a hospital when the duration of the stay is at least 24 hours.

Lapse: The termination of insurance coverage due to lack of payment after a specific period of time.

Limitations: A term referring to any maximums that a health insurance plan imposes on specific benefits.

Long-term Care: Care provided on a continuing basis for the chronically ill or disabled. Long-term care may be provided on an inpatient basis (at a long-term care facility) or in the home setting.

Medically Necessary: Also known as "Necessary." Health plans usually pay only for care that is "necessary." They decide this by using medical standards or research that states what care is most effective. Care can mean health services or supplies. This also is called "medically necessary,""medically necessary services" or "medical necessity."

Medicaid: A state-funded healthcare program for low-income and disabled persons, children and pregnant women, and the elderly.

Medicare: A national, federally-administered health insurance program authorized in 1965 to cover the cost of hospitalization, medical care, and some related health services for most people over age 65 and certain other eligible individuals.

Medicare Supplement Insurance: Health insurance provided to an individual or group that is intended to help fill in the gaps in the coverage provided by Medicare.

Member: Anyone covered under a health insurance plan, including enrollees and eligible dependents.

Outpatient: A term referring to a patient who receives care at a medical facility, but who is not admitted to the facility overnight, or who stays for 24 hours or less. The term may also refer to the health care services that such a patient receives.

Over-the-counter (OTC) Drugs: Drugs that may be obtained without a prescription.

Open Formulary: Some prescription benefits plans cover all eligible prescription drugs. This means they have an "open formulary." In these plans, people might have lower copays for drugs on the preferred drug list. They might have higher copays for drugs that are not on this list.

Out-of-pocket Costs: These are medical costs that a member must pay. Copays and deductibles are examples.

Preferred Drug List: Also known as "Formulary." This is a list of prescription drugs the health plan covers. It can include drugs that are brand name and generic. Drugs on this list may cost less than drugs not on the list. How much a plan covers may vary from drug to drug. An open formulary provides a greater choice of covered drugs. It is also called a "preferred drug list."

Premium: The total amount paid by the patient for insurance coverage. Usually calculated at an annual rate and paid monthly.

Preauthorization/Precertification: These are terms that are often used interchangeably, but which may also refer to specific processes in a health insurance or healthcare context.

Pre-existing Condition: A health problem that existed or was treated before the effective date of your health insurance coverage.

Prescription Medication: A drug that may be obtained only with a doctor's prescription and which has been approved by the Food and Drug Administration.

Primary Care: Basic health care services, generally rendered by those who practice family medicine, pediatrics or internal medicine.

Provider: A term commonly used by health insurance companies to designate any healthcare provider, whether a doctor or nurse, a hospital or clinic.

Rating Process: The process by which a premium or rate for a group is determined. Items that may be considered in the rating process include age, sex, type of industry, benefits and administrative costs. The current health care system enforces community rating, where members of the same community are charged similar prices. Previously, insurers were able to use experience rating, and set premiums based on personal past health care "experiences."

Referral: The process through which a patient under a managed care health insurance plan is authorized by his or her primary care physician to a see a specialist for the diagnosis or treatment of a specific condition.

Renewal: Renewal occurs when a member continues coverage under a health insurance plan beyond the original time frame of the contract. At the end of each benefit year, a plan member is generally invited to renew his or her coverage.

Reimbursement: This is money you get back from your health plan for covered costs you paid to your doctor.

Specialty Medication: Specialty drugs are prescription medications that require special handling, administration or monitoring. These drugs are used to treat complex, chronic and often costly conditions, such as multiple sclerosis, rheumatoid arthritis, hepatitis C, and hemophilia.

Specialist: A doctor who does not serve as a primary care physician, but who provides secondary care, specializing in a specific medical field.

Underwriting: The process by which an insurer determines whether it will accept an application for insurance based upon risks and projections, and through which a determination on a monthly premium is made.

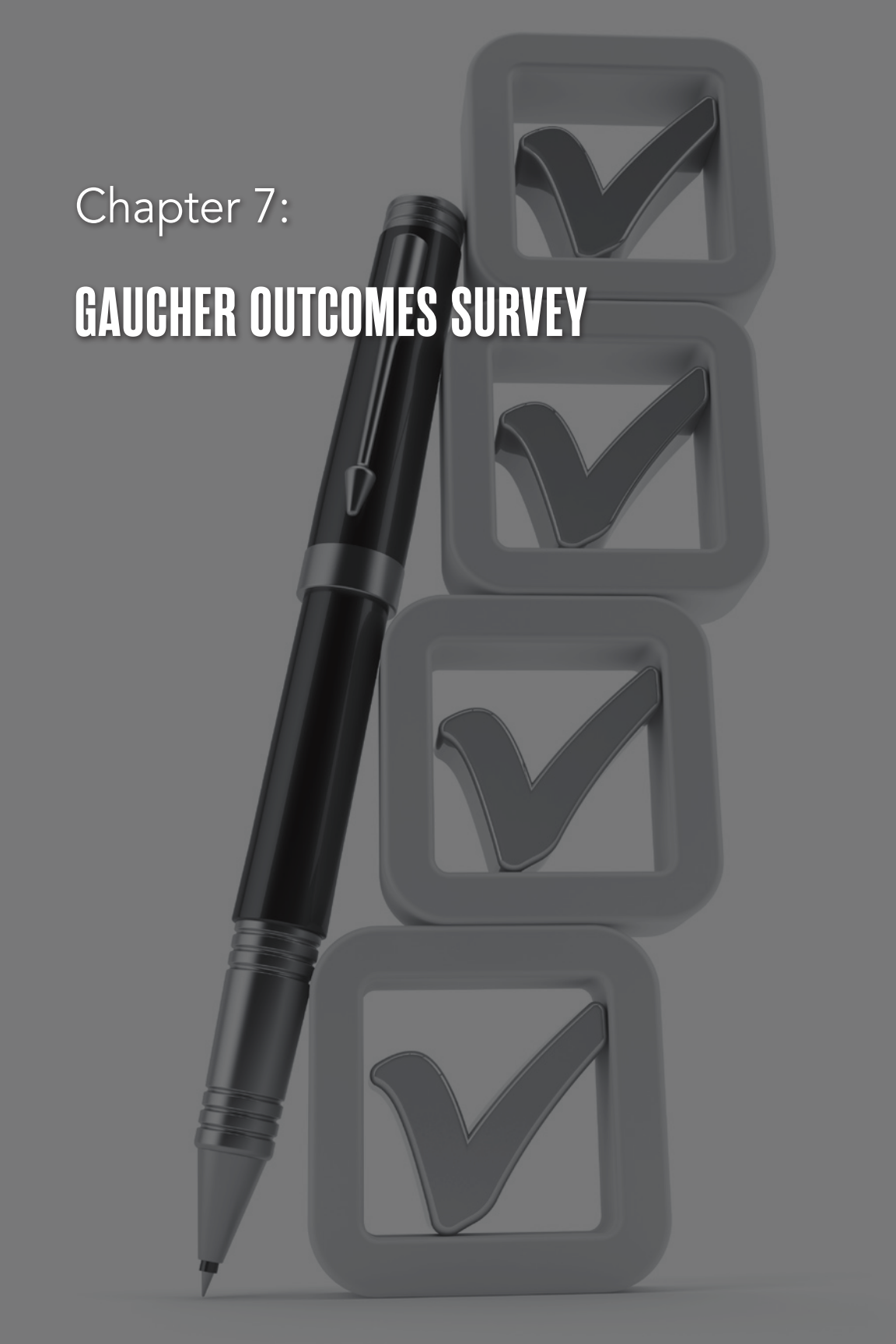
Utilization: This term refers to how frequently a group uses the benefits associated with a particular health insurance plan or healthcare program.

Utilization Management/Review: This term is often used to describe a group (or the work performed by a group) of nurses and doctors who work with health insurance plans to determine if a patient's use of healthcare services was medically necessary, appropriate and within the guidelines of standard medical practice. Utilization Management/Review may also be referred to as Medical Review.

Waiver (Exclusion Endorsement): An agreement under which a member agrees to waive coverage for specific pre-existing conditions or for specific future conditions.

Chapter 7:

GAUCHER OUTCOMES SURVEY



GAUCHER OUTCOMES SURVEY

WHAT IS THE GAUCHER OUTCOMES SURVEY?

The Gaucher Outcomes Survey (GOS) is an ongoing observational, international, multi-center, long-term registry of patients with Gaucher disease. It is one of several worldwide surveys, and includes patients receiving various types of treatment. The objectives of this registry are to: evaluate the safety and long-term effectiveness of Gaucher disease treatments; to gain a better understanding of the natural history of the disease; and to serve as a database for evidence-based management of Gaucher disease over time in real-life clinical practice. In other words, results from this study help researchers, pharmaceutical companies and physicians better understand the natural history of Gaucher disease, the population distribution of the disease and responses to treatment, in order to improve outcomes for all Gaucher patients.

WHAT HAS THE STUDY REVEALED?

These types of registries or surveys can provide a valuable source of real-world data for rare conditions, such as Gaucher, which would otherwise be challenging to collect. Initiated in 2010, the study currently has over 1,200 participants, representing 11 countries, although most participants reside in Israel and the United States. The most recent data showed that 44.3% of the patients enrolled in the study were from Israel and 31.4% were from the U.S. Approximately 44% of the patients in the survey were male and 56% were female. The average age of participants was 40.4 years of age. The vast majority of registered patients had type 1 Gaucher (91.5%), but patients with types 2 and 3 are also included. Approximately half the patients (55.8%) were of Ashkenazi Jewish ethnicity.

The survey also assesses:

- Infusion-related reactions
- Hemoglobin levels
- Platelet counts
- Decreases in liver volume
- Decreases in spleen size
- Bone mineral density

WHO CAN PARTICIPATE IN THE STUDY?

The study is voluntary and open to all Gaucher patients, regardless of your treatment status or type of treatment. Patients who participate in the GOS receive care determined by their physicians, as they normally would (there is no experimental intervention involved). While being treated, patient data from routine visits and clinical assessments is entered into the study's database through a secure online application.

While this survey compiles important information, participating in any study is a very personal decision. If you're interested in participating, please talk with your Gaucher specialist or primary care physician about whether it's right for you.

(If you are interested in learning more about the Gaucher Disease Outcome Survey (GOS), visit https://clinicaltrials.gov/ct2/show/NCT03291223.)

[1]*Author's Note: These are the mutations observed in primarily Israeli and U.S. populations. In addition to this study, there are a number of publications that can help patients stay up-to-date on the latest care and management of type 1 Gaucher disease.*

Chapter 8:

RESOURCES AND SOURCES

RESOURCES

GENERAL

National Gaucher Foundation (NGF) – an independent nonprofit dedicated to serving U.S. patients with Gaucher and their families. The NGF provides educational programs, financial support, and a variety of patient services.
www.gaucherdisease.org
5410 Edson Ln #220, Rockville, MD 20852
(800) 504-3189

ThinkGenetic – is a cutting-edge tool that empowers patients who want to know about possible genetic causes for their medical issues or obtain real-life answers to their questions about the impact of living with a genetic disease. ThinkGenetic will help you understand what questions to ask physicians in order to improve your medical care.
www.thinkgenetic.com

Center for Jewish Genetics – The Norton & Elaine Sarnoff Center for Jewish Genetics is an advanced educational resource for hereditary cancer and genetic disorders. The center works with healthcare professionals, clergy, support organizations and other individuals to inform community members and raise awareness of available resources. They also provide subsidized genetic counseling and screening programs.
https://www.jewishgenetics.org
30 S Wells St., Chicago, IL 60606
(312) 357-4718

National Organization for Rare Disorders (NORD) – an American nonprofit organization aiming to provide support for individuals with rare diseases by advocating and funding research, education, and networking among service providers.
www.rarediseases.org
PO Box 1968, 55 Kenosia Avenue, Danbury, CT 06813-1968
(203) 744-0100 or (800) 999-NORD

Children's Gaucher Research Fund – a 501(c)(3) charitable organization that raises funds to coordinate and support research to find a cure for Type 2 and Type 3 Gaucher Disease. Find family stories, fundraising ideas and recent news on this website.
www.childrensgaucher.org
8110 Warren Court, Granite Bay, CA 95746
(916) 797-3700

Genetic and Rare Diseases Information Center (GARD) – a program of the National Center for Advancing Translational Sciences (NCATS), funded by the National Institutes of Health (NIH). GARD provides the public with access to current, reliable and easy-to-understand information about rare or genetic diseases.
https://rarediseases.info.nih.gov
PO Box 8126, Gaithersburg, MD 20898-8126
(888) 205-2311

National Society of Genetic Counselors (NSGC) – a resource for patients to find genetic counselors, as well as educational opportunities and advocacy.
www.nsgc.org
330 North Wabash Avenue, Suite 2000, Chicago, IL 60611
(312) 321-6834

HEALTH INSURANCE

Regarding Employer-sponsored Plans:

"3 Important Questions to Ask: How much will it cost me, what is covered, and who is in my network"
www.monster.com/career-advice/article/health-insurance-benefit-open-enrollment-1016

Regarding the Individual Insurance Exchange:

5 tips about the Individual Exchange –
www.healthcare.gov/quick-guide/ one-page-guide-to-the-marketplace

The Marketplace Made Clear by UnitedHealth Care –
www.uhc.com/individual-and-family/understanding-health-insurance/how-insurance-works/ health-insurance-marketplace

Regarding Medicaid:

www.medicaid.gov/medicaid

Medicaid Eligibility – www.medicaid.gov/medicaid/eligibility

Regarding Medicare:

www.medicare.gov

Part D FAQs
www.health.ny.gov/health_care/medicaid/program/medicaid_transition/faq.htm

AARP Medicare Information
www.aarp.org/health/medicareinsurance/medicarebasics/?intcmp=AE-HEA-MEDI-TERTNAVBASICS

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https://kidshealth.org/en/parents/about-genetics.html

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https://www.capitalcaring.org/care-support/care-navigators/

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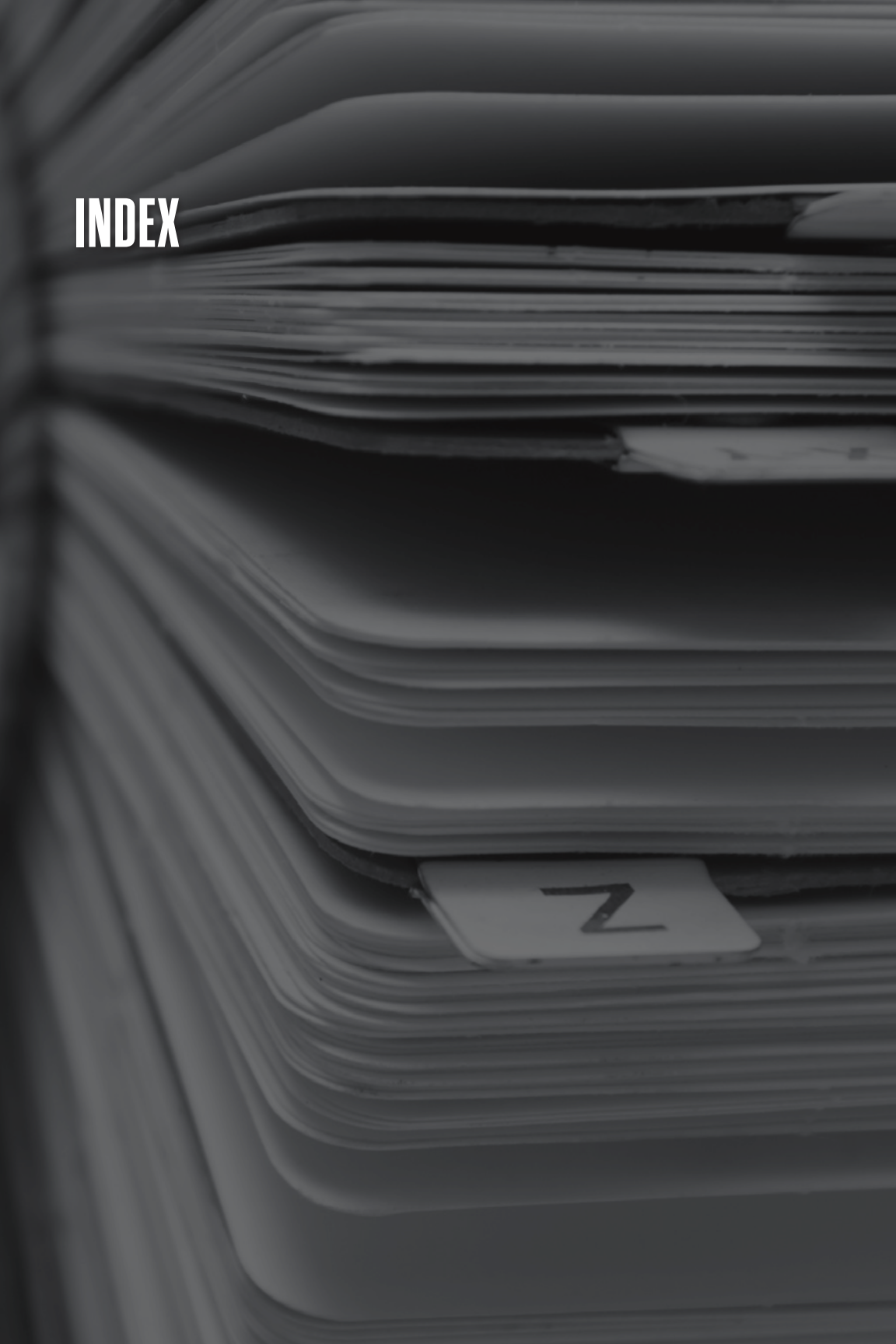

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ABOUT THE AUTHOR

Laurie Bailey is currently the Clinical Research Manager at Cincinnati Children's Hospital in Cincinnati, Ohio, although she was trained as a genetic counselor, receiving a Master's in Medical Genetics from the University of Cincinnati in 1995. She became involved with and interested in the study of Gaucher disease in 1999 when she took a position coordinating the Comprehensive Gaucher Disease Treatment Center under the leadership of Dr. Gregory Grabowski.

Dr. Bailey has participated in the management of over 100 children and adults with all types of Gaucher disease, and she has coordinated both interventional treatment trials and clinical research to advance the knowledge of Gaucher disease and strive for better outcomes for those affected by the disease. She has been an advocate for those with Gaucher disease, providing education on the treatment and management of the disorder by speaking at patient meetings, national meetings, and advisory boards.

Dr. Bailey currently oversees The Rare Disease Treatment and Management Team within the Division of Human Genetics where they promote the qualities of their STAR Program to provide Support, Treatment, Advocacy, and Research for both children and adults with rare genetic diseases, including Gaucher disease.