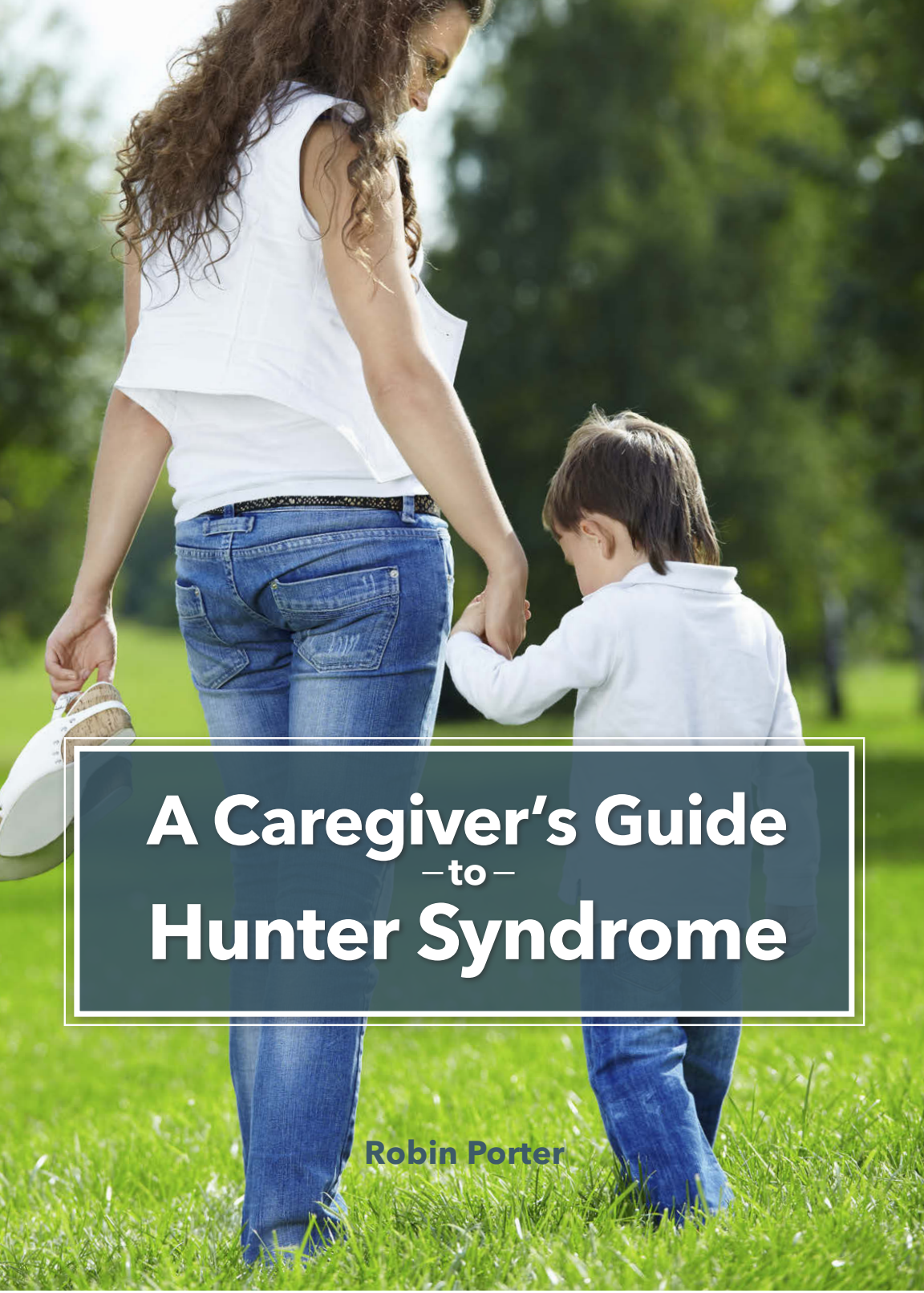

A Caregiver's Guide to Hunter Syndrome

Robin Porter

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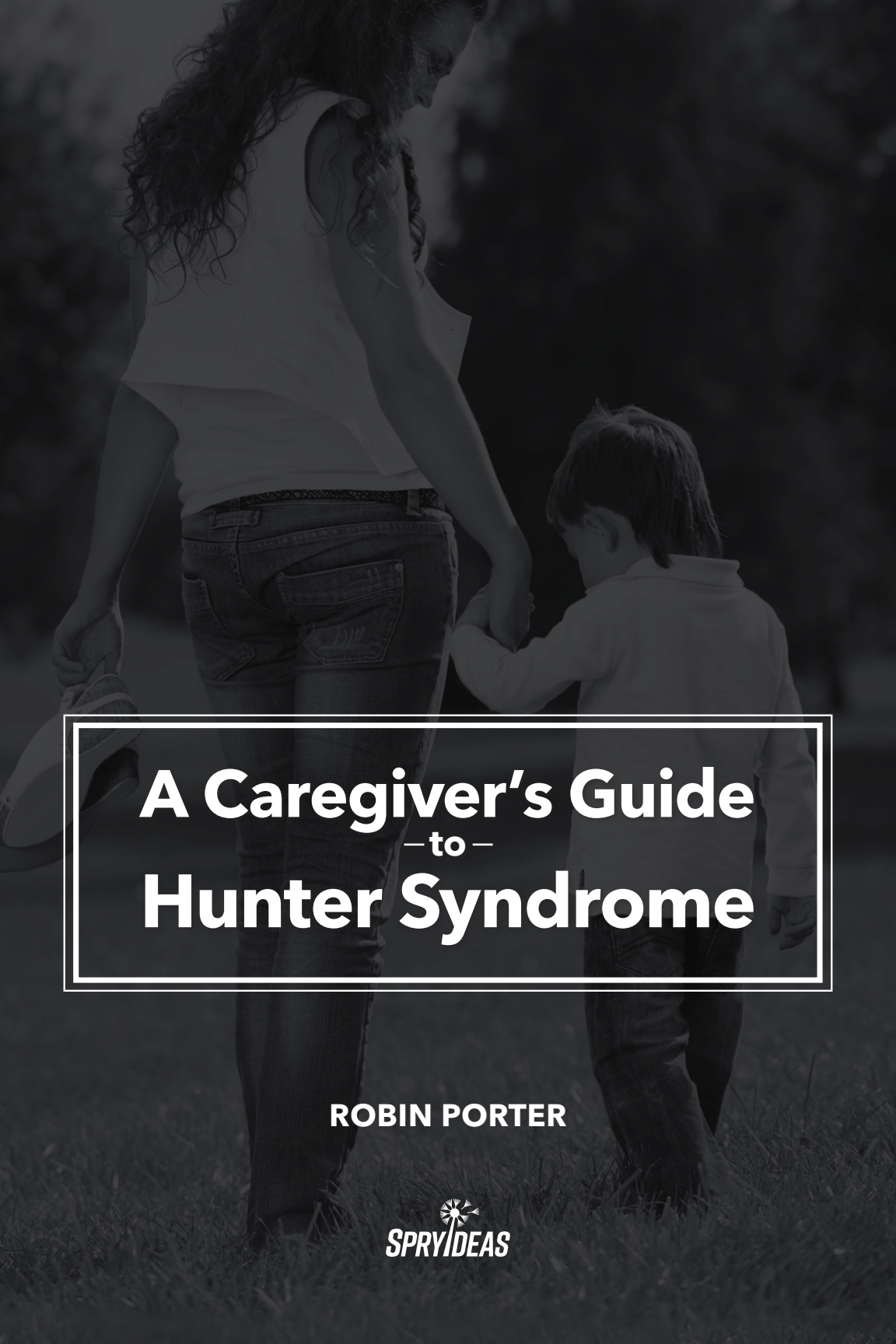

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ROBIN PORTER

SPRYIDEAS

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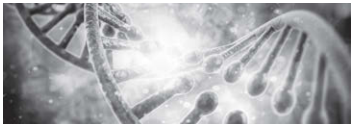
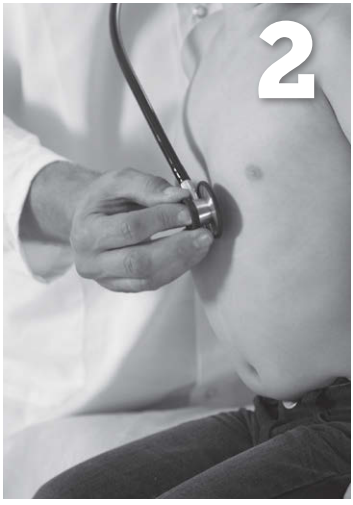




Contents

Introduction

Introduction

As a genetic counselor in a lysosomal storage disease center, my primary role for the past 8 years has been coordinating the care and treatment of individuals who have mucopolysaccharidosis (MPS). This often involves working closely with parents and caregivers of children who are affected with the disease. As it often happens, these families have also taught me a great deal – not only about the clinical aspects of this disease, but about care, compassion and courage.

MPS II, or Hunter syndrome, is a rare genetic disease affecting about one in 100,000 males. When a child receives a diagnosis of MPS II, families are launched into the world of a rare disease. While all serious childhood diseases are challenging, a rare disease presents a unique set of trials. For instance, parents quickly discover that not many people, including most doctors, have even heard of Hunter syndrome. Sometimes MPS II is passed down in a family, so a parent may have a brother or uncle with the disease, which provides important clues for making a diagnosis. Other times, however, MPS II is caused by a new genetic change in the family, and there is no family history. In this situation, parents often experience what's referred to as the "diagnostic odyssey."

This odyssey usually begins when a young, healthy boy starts showing developmental delays, signs of skeletal disease, hearing loss, and other symptoms. It's clear that something is wrong, but a diagnosis may take months or years because most physicians have never seen a child with MPS II before and do not know to test for it. The diagnostic odyssey can be a long, frustrating process of tests, some invasive, that do not provide an answer, and may result in misdiagnoses such as autism or ADHD.

Once a correct diagnosis is finally made, there is often relief that the odyssey is over. However, that is followed by the start of a new journey – managing MPS II. Receiving this news can be devastating for parents as they realize their child is at risk for multiple serious medical complications and a shortened lifespan. Children with the severe form of MPS II also have acute central nervous system disease that includes a regression, or loss, of all developmental and cognitive skills over time.

Managing the disease can easily feel like a full-time job as children need to be followed by several different specialists. Their care will also involve periodic assessments and evaluations to monitor for disease progression, and sometimes surgery is required to address medical issues. It's a lot for a child and his parents to endure. As a result, many parents and caregivers become emotionally exhausted helping their child battle Hunter syndrome.

Another challenge for families dealing with a rare disease is the lack of readily-available information, which is the reason this book was written. We hope *A Caregiver's Guide to Hunter Syndrome* will be a useful resource for caregivers. It offers practical information about the disease, its management and getting organized. It also contains suggestions about how to discuss MPS II with siblings, family members, and schools. Perhaps, most importantly, it provides tips and encouragement for self-care. The MPS II journey is overwhelming, and therefore requires caregivers to take steps to maintain their own physical and mental well-being. With that in mind, this book is intended to help caregivers feel like they have a plan for getting started and some good skills moving forward.

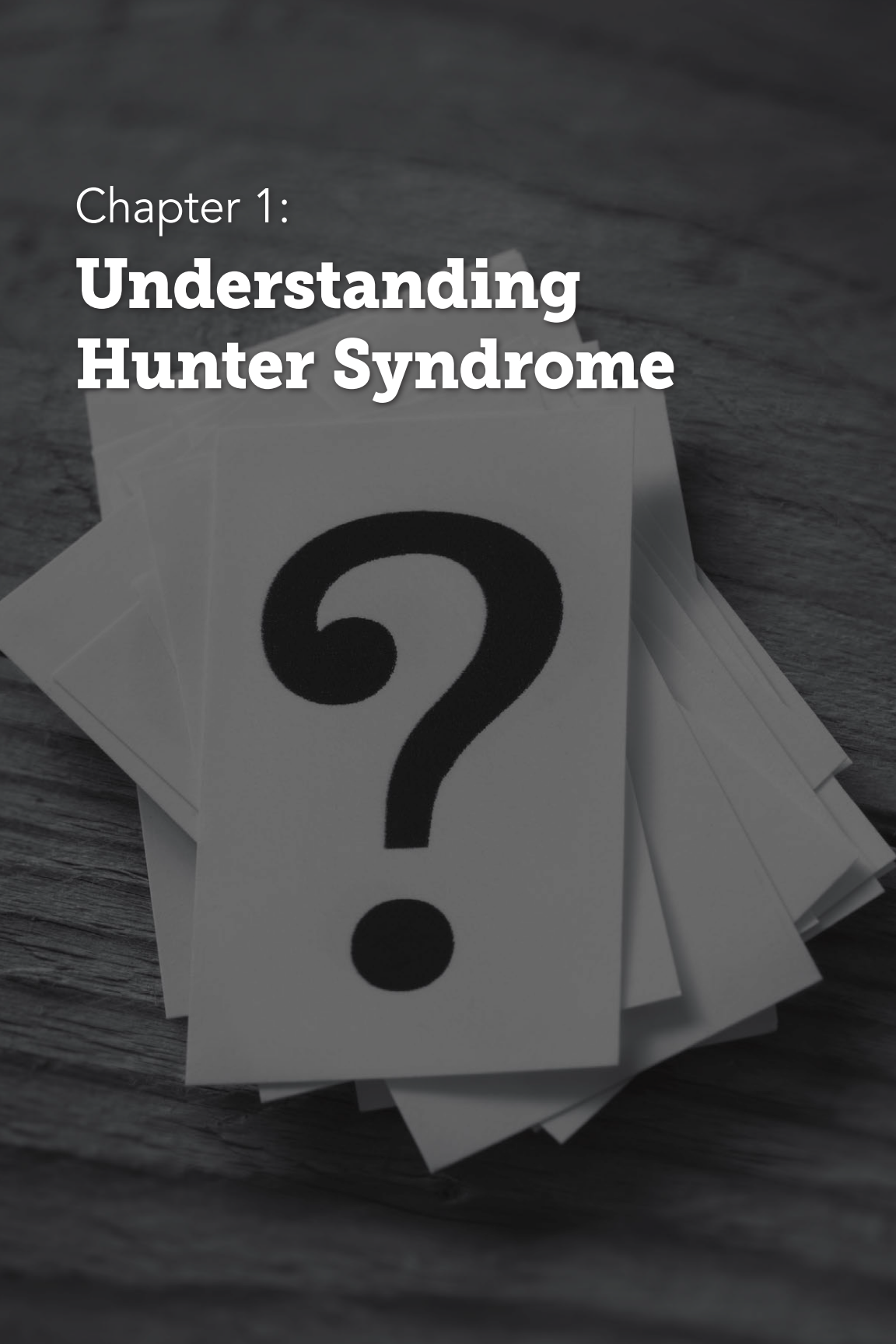
I can attest that parents and caregivers of children with Hunter syndrome are among the strongest and most resilient people I have ever met. They welcome newly diagnosed children and families with open arms, understanding, and love into the larger MPS II family, and so I encourage you to reach out and accept this support. Though MPS II is a rare disease, parents and caregivers should feel far from alone.

Sincerely,

Stephanie Cagle, MS, CGC

Certified Genetic Counselor Lysosomal Storage Disease Center
Department of Human Genetics
Emory Healthcare, The Emory Clinic

Stephanie Cagle is a certified genetic counselor in Emory's Lysosomal Storage Disease Center in Atlanta, GA. She received her Masters of Science in Genetic Counseling in 2009 from Mount Sinai School of Medicine in New York City. She moved to Atlanta after graduation, and her primary focus for the past 8 years has been mucopolysaccharidosis. Stephanie is also a study coordinator for several industry-sponsored clinical research trials for lysosomal storage diseases and other genetic conditions.



Chapter 1:

Understanding Hunter Syndrome

Understanding Hunter Syndrome

"Understanding the challenges you face with your illness, and then planning a life despite them, may be one of the healthiest decisions you will ever make."

– Lisa Copen

Your child's diagnosis of a serious disease is a life-changing event for you, your family, and of course, your child. It can be one of the most difficult challenges you will face as a parent. Many parents/caregivers describe the experience as a rollercoaster of emotions, from anger and frustration, to overwhelming love and compassion, and everything in between.

One of the most common complaints voiced by parents caring for a child with a serious disease is a feeling of helplessness. As parents, we want to help our children live the best life possible, and because a serious disease removes so much from our control, it's natural to feel powerless. While it's impossible to eliminate those feelings, learning as much as you can about the disease can help you feel more empowered.

Knowledge truly is power. The better you understand the illness, its symptoms and complications and how to manage them, as well as care options, the more equipped you will be to cope with the obstacles, make decisions, communicate effectively with others and improve the quality of life for your child. With that in mind, let's begin the learning process.

WHAT IS HUNTER SYNDROME?

Hunter syndrome, or mucopolysaccharidosis type II (MPS II), is a rare, inherited genetic disorder that primarily affects males. In the simplest terms, it's caused by a missing or malfunctioning enzyme in the body. Because the body doesn't have enough of the enzyme to break down certain complex molecules, the molecules build up in harmful amounts.

More specifically, the disorder interferes with the body's ability to break down and recycle certain mucopolysaccharides (mew-ko-pol-ee-sak-ah-rides), also known as glycosaminoglycans (gli-ko-sah-mee-no-gli-cans) or GAGs. In normal quantities, GAGs attract water and are useful to the body as a lubricant or a type of shock absorber. For instance, the fluid that lubricates your joints contains GAGs. However, in Hunter syndrome, GAGs build up in cells throughout the body due to a deficiency or absence of the enzyme iduronate-2-sulfatase (I2S). This buildup interferes with the way certain cells and organs in the body function and leads to a number of serious symptoms

involving the cells, blood and connective tissues, causing permanent and progressive damage (see Signs and Symptoms). The severity and type of symptoms can vary widely among individuals (see Chapter 2: Managing Symptoms and Complications).

Hunter syndrome, or MPS II, gets its name from Charles Hunter, the professor of medicine in Manitoba, Canada, who first described two brothers with the disease in 1917. It's a rare but serious disorder that primarily affects males. It interferes with the body's ability to break down and recycle specific sugar molecules called mucopolysaccharides, also known as glycosaminoglycans (GAGs). This process requires critical enzymes. If one of these enzymes is deficient, the large molecules build up in harmful amounts in the body's cells.

WHAT CAUSES HUNTER SYNDROME?

Hunter syndrome is an inherited genetic disorder, mostly passed down through families. The defective gene is on the X chromosome, which means the disease is X-linked recessive. Girls may be carriers of the disease, but except in very rare cases, only boys will be affected (see "How is Hunter Inherited?").

As a result of this defective gene, the body cannot produce an enzyme called iduronate-2-sulfatase (I2S). Without this enzyme, chains of sugar molecules cannot be broken down properly and build up in various body tissues, causing damage.

That, of course, is a simplified explanation of a very complex process. To understand fully, we must take a closer look at what happens in the body.

The human body depends on a vast array of biochemical reactions to support critical functions, including the production of energy, growth and development, communication within the body, and protection from infection. Another critical function is the breakdown of large biomolecules, which is the underlying issue in MPS II and other related storage disorders.

In Hunter syndrome, the problem begins in a part of the connective tissue of the body known as the extracellular matrix. This matrix is made up of a variety of sugars and proteins, and helps to form the framework of the body. You can think of it as a type of meshwork that holds the cells in place. One of the parts of this meshwork is a complex molecule called proteoglycan.

Like many components of the body, proteoglycans need to be broken down and replaced (similar to the way oil breaks down in your car over time and must be changed). When the body breaks down these molecules, one of the resulting products is mucopolysaccharides or glycosaminoglycans, otherwise known as GAGs.

During our lives, there is a continuous cycle of building new GAGs and breaking down old ones. This ongoing "recycling process" is required to keep the body healthy. The breakdown process occurs in a part of the cell called the lysosome and requires special enzymes. That's why Hunter syndrome or MPS II is considered one of approximately 40 different types of lysosomal storage diseases, which are all caused by the deficiency of individual enzymes.

There are several types of GAGs, each found in certain areas of the body:

GAG	LOCATION IN THE BODY
Hyaluronic acid	Connective tissues, skin, cartilage, synovial fluid
Chondroitin sulfate	Cartilage, cornea, bone, skin, arteries
Dermatan sulfate	Skin, blood vessels, heart, heart valves
Heparan sulfate	Lung, arteries, cell surfaces
Heparin	Certain immune system cells
Keratan sulfate	Cartilage, cornea, intervertebral discs

Source: www.hunterpatients.com

Hunter syndrome involves two GAGs – dermatan sulfate and heparan sulfate. These GAGs require a certain enzyme, called lysosomal enzyme I2S, to be broken down in the body. In people with Hunter syndrome, this enzyme is either missing or deficient.

Since the GAGs cannot be properly broken down, they build up in cells throughout the body, especially in tissues that contain large amounts of dermatan sulfate (skin, blood vessels, heart, heart valves) and heparan sulfate (lungs, arteries, cell surfaces). As this buildup continues, it interferes with the way certain cells and organs in the body function, which causes a variety of symptoms and complications.

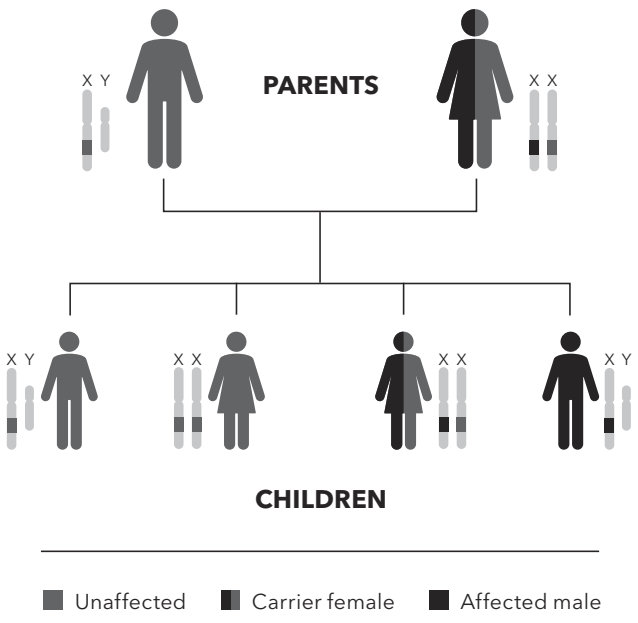
HOW IS HUNTER SYNDROME INHERITED?

Hunter syndrome is passed down from one generation to another in a specific way. It's known as an X-linked recessive disorder. Nearly every cell in the human body has 46 chromosomes – 23 from each parent. The I2S gene, which causes the disease, is located on the X chromosome. Females have two X chromosomes, one inherited from each parent, while males have one X chromosome from their mother and one Y chromosome from their father.

If a male is born with an abnormal copy of the I2S gene, he will develop Hunter syndrome. This can happen in two ways: 1) His mother is often a carrier, meaning she has one abnormal and one normal I2S gene, and she passes along this abnormal gene to her son. If a woman is a carrier, there is a 50 percent risk that any boy born to her will have the disease; 2) A mutation can develop in the I2S gene during the egg and sperm formation, which means the mother is not a carrier and the risk of a spontaneous mutation occurring again in a future sibling is low but not zero.

Females can carry one abnormal copy of the I2S gene and are usually not affected.

How Hunter Syndrome is Passed On



WHAT ARE THE RISK FACTORS?

There are two major risk factors for developing Hunter syndrome:

1. **Family history.** As we discussed, Hunter syndrome is caused by a defective gene or chromosome that is inherited. Hunter syndrome is known as an X-linked recessive disease, which means that women carry the disease-causing X chromosome, but women aren't affected by the disease themselves. However, as mentioned, the mutation can occur spontaneously with no previous family history.

2. **Sex.** Hunter syndrome occurs primarily in males. Girls are far less at risk of developing Hunter syndrome because they inherit two X chromosomes. If one X chromosome is defective, their normal X chromosome can provide a functioning gene. If the X chromosome of a male is defective, however, there isn't another normal X chromosome to compensate for the problem.

Hunter syndrome affects a calculated estimate of approximately 1 in 162,000 live births, making it a rare disorder.

WHAT ARE THE COMMON SIGNS AND SYMPTOMS?

The symptoms of MPS II are usually not apparent at birth. They may appear in infants as young as 18 months, but typically start to become noticeable between 2 and 4 years of age. Symptoms vary among individuals and range from mild to severe.

Common early symptoms of Hunter syndrome may include:

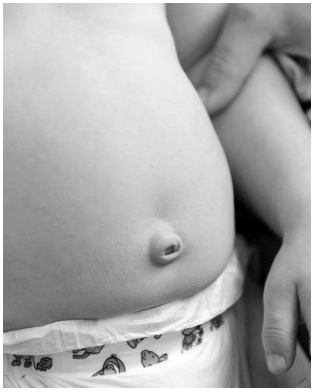
- Inguinal hernia[1]
- Recurrent ear infections
- Runny nose
- Colds
- Coarse facial features
- Hearing loss
- Recurrent watery diarrhea
- Joint stiffness
- Enlarged abdomen
- Enlarged tongue and tonsils

[1]An inguinal hernia occurs when tissue, such as part of the intestine, protrudes through a weak spot in the abdominal muscles.

Of course, many of these symptoms are very common among babies and toddlers, and therefore, are not likely to lead a physician to diagnose Hunter syndrome at an early stage. However, as the build up of GAGs continues, the signs of MPS II become more apparent.

Some later signs and symptoms may include:

- A distinctive coarseness in facial features, including a prominent forehead, a nose with a flattened bridge and flared nostrils*
- A large head
- Thickening of the lips
- An enlarged or protruding tongue*
- A deep, hoarse voice
- An enlarged or distended abdomen (resulting from enlarged internal organs)
- Frequent ear infections*
- Respiratory infections
- Cardiac/valvular heart disease
- Breathing problems while sleeping
- Limited lung capacity
- Joint stiffness and limited range of motion*
- Carpal tunnel syndrome
- Stunted growth or short stature
- Abnormal bone size or shape and other skeletal irregularities
- White skin growths that resemble pebbles on upper arms, legs and upper back
- Aggressive behavior
- Delayed development, such as late walking or talking



(*These signs and symptoms may also appear earlier in Hunter patients with severe cases.)

It's important to note that the number of symptoms and the degree of severity can differ dramatically for each child with Hunter syndrome, which can sometimes make the initial diagnosis difficult. Because many of the early signs and symptoms are so common among infants and toddlers, it can often take time to diagnose the disease.

Hunter syndrome is rare, but as a parent and a caregiver, it's important to be an advocate for your child. Learning about the disease and keeping track of symptoms and/or changes in your child, can help your pediatrician make a diagnosis. For instance, if you notice changes in your child's facial features, a loss of previously acquired skills, or a combination of any of the signs or symptoms listed above, it's time to consult your pediatrician or primary care doctor. He or she may refer you to a genetic specialist for further testing.

HOW IS HUNTER SYNDROME DIAGNOSED?

Infants born with Hunter syndrome almost always appear healthy at birth. As discussed, visible signs and symptoms are usually the first clues, and typically appear between the ages of 2 and 4. Because many of these symptoms overlap with common childhood complaints, it's usually a combination or cluster of symptoms that point to Hunter syndrome. Because Hunter syndrome progresses slowly, and its signs and symptoms overlap with other disorders, getting a definitive diagnosis may take some time. Of all the symptoms, coarse facial features are one of the strongest diagnostic indicators, even though they may be subtle.

If Hunter syndrome is suspected, your primary care physician or pediatrician may initially order laboratory tests, such as blood and urine samples. Urine is tested for increased levels of GAGs, which are present in patients with this disease. Blood, urine and tissue samples can be checked for the deficient enzyme or for excess amounts of GAGs. Children with a family history of Hunter syndrome may be referred directly to a genetic specialist (also referred to as medical geneticist or genetic counselor) for testing.

A genetic specialist will make a definitive diagnosis by measuring iduronate-2-sulfatase (I2S) activity. This requires taking blood from the patient and testing the I2S activity in the serum or white blood cells. It may also require a skin biopsy to test the I2S activity in the skin cells.

Early diagnosis is critical. The earlier Hunter syndrome is diagnosed, the sooner potential treatment options can be explored and care for complications can be administered, which may prevent some of the permanent damage caused by the disease.

ASSEMBLING A TEAM OF SPECIALISTS

Once a diagnosis is made, you may be referred to a number of medical specialists, depending on the particular complications your child is experiencing (see Chapter 2: Managing Symptoms and Complications). Over time, as the disease progresses, symptoms may change, requiring new or additional medical specialists.

Your medical team might include specialists in the following areas:

- **Audiology** – for identifying, diagnosing, treating and monitoring hearing and balance problems.
- **Cardiology** – for diagnosing and treating disorders of the heart.

- **Dentistry** – for diagnosing, prevention and treatment of diseases and conditions of the mouth, gums and teeth.
- **Genetics** – for initial diagnosis, as well as prescribing and managing enzyme replacement therapy.
- **Neurology** – for dealing with disorders of the nervous system.
- **Occupational and Speech Therapy** – for help with daily functions that may be affected, such as feeding, swallowing, cognition, movement and speech.
- **Ophthalmology** – for diagnosing and treating problems with vision and diseases of the eye.
- **Orthopedics** – for surgical and nonsurgical treatment of problems with the musculoskeletal system, such as the spine.
- **Otorhinolaryngology** – for dealing with conditions of the ear, nose and throat.
- **Pediatrics** – for the primary medical care of infants, children and adolescents.
- **Physiotherapy** – for restoring and maintaining movement and function when the body is affected by illness or injury.
- **Pneumology (or Pulmonology)** – for diagnosing and treating issues with the lungs, pulmonary or respiratory system.
- **Rheumatology** – for the detection and treatment of musculoskeletal and autoimmune conditions that affect the joints, muscles and bones.

While this list may seem overwhelming, it's important to take things one step at a time. The number and type of specialists your child may receive treatment from will depend on the individual symptoms he experiences over time. Keeping good records of visits with each specialist will make things more manageable (see Chapter 3: Getting Organized).

HOW TO FIND A SPECIALIST?

In many cases, your pediatrician or primary care physician will refer you to the appropriate specialists or make recommendations. You can also:

- Check with area hospitals – many offer referral services.
- Check with your county medical society.
- Check with Hunter syndrome support groups to connect with families in your area who can share their experiences with local specialists.
- Check with your geneticist or genetic counselor, if you have one.
- Ask your insurance company, health maintenance organization or managed care plan if they have preferred physicians to choose from.
- When choosing a specialist, consider the following:
  - Is the physician licensed and board certified? (You can call the American Board of Medical Specialties at 800-ASK-ABMS or 800-275-2267 or www.abms.org to confirm board certification.)
  - Has the physician ever evaluated children with Hunter syndrome?
  - Does the practice serve people with disabilities? If so, how many?

- Does the practice accommodate people with disabilities (i.e., reduced wait times, quiet waiting areas, etc.)?
- Are the location and office hours convenient? What type of after-hours service, emergency coverage and telephone consultations do they offer?
- Does the practice specialize in pediatric treatment?
- Does the practice accept your health insurance plan, or is it part of your insurance network?

Once you've found a specialist, you may want to set up an appointment to interview him or her. Many offices set aside time for brief interviews for new patients. Bring a list of questions and take notes so you can keep track of each specialist. Meeting a physician face-to-face is still the best way to determine if he or she is a "good fit" for you and your child. Keep in mind, many specialists have never seen a patient with Hunter syndrome. In this situation, make sure the specialist seems interested and dedicated to learning about the disease to provide your child with the best possible care.

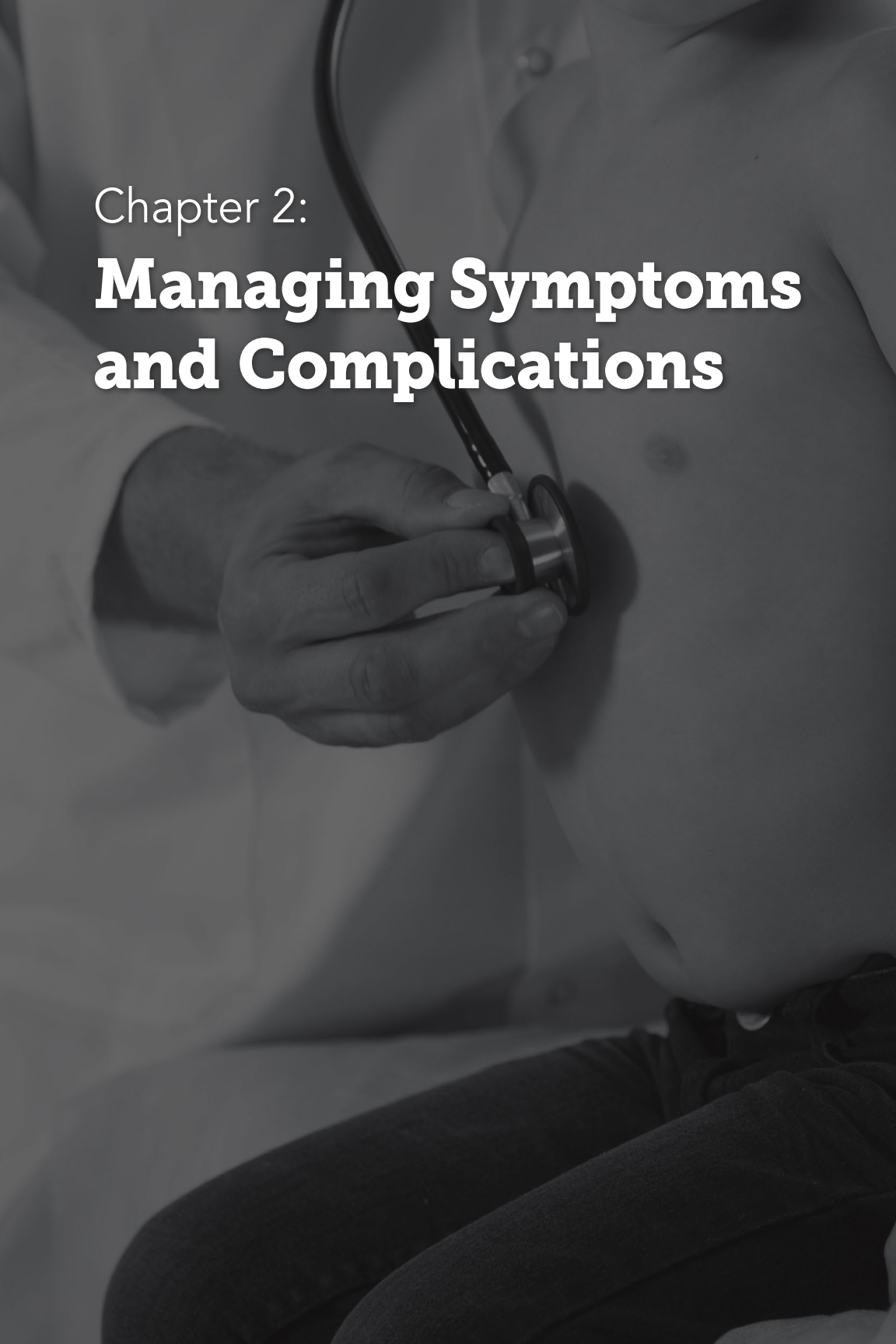
Now that you've completed a "crash course" on Hunter syndrome, you may want to make a list of additional questions you have for your pediatrician or primary care physician. In fact, it's a good idea to keep a notebook or diary handy so you can jot down questions and concerns as they come to mind. No doubt, the most pressing questions you have now are: what type of complications can we expect, and how can we manage them? This is the subject of our next chapter.

A CAREGIVER'S PERSPECTIVE

Many parents/caregivers initially feel devastated by the news that their child has Hunter syndrome. Questions such as, "How did this happen?" and, "What can we do?" are normal. Though physicians often advise parents not to go home and look up all the possible symptoms and outcomes because it's simply too overwhelming, it's human nature to want to know as much as possible about the disease affecting your child.

However, experienced parents advise those who are newly diagnosed to not become bogged down by the information and to give yourself time to accept your "new normal." For example, in an effort to regain control, many parents will immediately schedule consultations with specialists until their calendar is jam-packed with medical appointments. This can quickly become "too much" for both caregivers and their children.

Ask your primary care physician which appointments to prioritize. For example, getting a baseline echocardiogram to check for heart valve disease is important, as physical symptoms of this problem may not be obvious until the heart is damaged. As for other specialists, unless there is an emergency, parents advise spreading out appointments over time — a year or more. In other words, be sure to leave time to live your life and love your child, and deal with things as they come.



Chapter 2:

Managing Symptoms and Complications

Managing Symptoms and Complications

"We must never forget that we may also find meaning in life even when confronted with a fate that cannot be changed. What matters is to bear witness to the uniquely human potential at its best, which is to transform a personal tragedy into a triumph…When we are no longer able to change a situation, we are challenged to change ourselves."

– Viktor E. Frankl

Hearing that there is no cure for your child's disease can feel like a punch to the gut. Even the strongest person can feel overwhelmed and frightened when receiving this kind of news. Sometimes it takes a while for the shock to subside and the reality to set in. Once it does, it's important to remember that having a flood of emotions is normal, and not something that you should be embarrassed or ashamed about expressing.

In fact, most people go through the five stages of grief when diagnosed with an incurable disease. They experience denial, anger, bargaining, depression and finally acceptance. (To read more on this topic, go to https://grief.com/the-five-stages-of-grief) As difficult as it may seem, working through these stages can help you process your feelings and better cope with your new reality.

One way to cope with a life-changing diagnosis is to channel your feelings into something productive, such as learning about treatment options, getting organized, talking to other parents, and joining organizations such as the National MPS Society, whose mission is to find a cure. It's also important to focus on the "now" (taking it one day at a time), instead of getting caught up in the future "what ifs," which is likely to leave you feeling anxious or depressed.

Once you've accepted the diagnosis, you can begin the journey of managing symptoms and complications, providing relief for your child as the disease progresses, and helping him (and you) to live the best life possible.

As we discussed in Chapter 1, the type and severity of symptoms and complications can vary widely among children with Hunter syndrome. The severity of symptoms can be viewed as a continuous spectrum, with the most severely affected individuals at one end, and the less severely affected (attenuated) individuals on the other end, with a wide range in between.

When receiving a diagnosis, you may also hear physicians refer to two specific types of MPS II: neuronopathic, which affects the nervous system and is more severe; or non-neuronopathic, which is the same as attenuated (less severe). As a general rule, individuals with cognitive impairment and developmental regression in childhood have the severe form of the disease (neuronopathic), while those who do not have cognitive impairment have the attenuated or non-neuronopathic form. However, it's important to note that individuals with attenuated Hunter syndrome may still have serious physical symptoms.

Your child may or may not experience the following complications, but knowing what they are can help you recognize problems and get the appropriate treatment.

PHYSICAL APPEARANCE AND DEVELOPMENT

Babies with severe Hunter syndrome may be quite large at birth and grow faster than normal during the first two years of life. After that, growth typically slows and becomes significantly less than normal, depending on the severity of the disease. For instance, those with severe MPS II may only reach a height of 4 feet to 4 feet 7 inches, while others with MPS II may grow to normal height. In addition, some patients with attenuated Hunter syndrome who have no cognitive or developmental impairments, may still have a short stature and be considered dwarfed.

Children with Hunter syndrome also tend to share physical attributes, including short, broad noses with a flattened bridge, flat faces, and large heads with prominent foreheads. They often have plump, rosy cheeks and thicker than normal lips. MPS II also causes tongues to be enlarged and hair to be thick and abundant. Beyond facial features, children with Hunter syndrome typically have protruding abdomens (due to enlarged organs), and stand and walk in a hunched position, which is caused by joint contractures (a shortening of the muscle or ligament connecting the joint).

Developmental delays are common in children with MPS II. Again, there is a wide range of intelligence and abilities, based on the severity of the disease. Some children with severe Hunter syndrome will experience serious learning disabilities and only learn to speak a few words, while others will be able to read at a basic level. On the other end of the spectrum, children and adults with attenuated MPS II may have no difficulty learning. In fact, some individuals with MPS II have successfully attended college and embarked on careers.

Medical professionals and experienced parents advise helping children with Hunter syndrome to learn as much as possible before the disease progresses. Reading to your child and engaging him in simple puzzles and games can be both enjoyable and beneficial.

RESPIRATORY COMPLICATIONS

Both the buildup of GAGs, which cause tissues in the airway to thicken, and changes in the anatomy, such as an enlarged tongue, tonsils and adenoids, can obstruct airways and lead to breathing problems. This is why many individuals with MPS II have frequent coughs, colds and throat infections. Children with Hunter syndrome also often experience noisy breathing and snoring, as well as suffer from sleep apnea. If parents notice significant episodes of interrupted breathing during sleep (sleep apnea), having your child evaluated by a sleep specialist may be advised.

The use of breathing devices such as CPAP (continuous positive airway pressure) or BIPAP (bilevel positive airway pressure), with or without oxygen, can improve sleep for some by keeping the airway open. CPAP, for example, uses a face mask or an endotracheal tube to assist breathing by pumping air into the airway, keeping it open and increasing lung volume. Maintaining an open airway can also help avoid low blood oxygen levels, which can be harmful over time.

In some cases, the removal of tonsils and adenoids can open a patient's airway and help relieve breathing problems, as well as reduce the incidence of strep throat. However, as the disease progresses, tissues may continue to thicken, causing these problems to reoccur. Consulting with your physician is the best way to determine which treatment is best for your child.

If the respiratory system becomes further compromised by pulmonary restriction due to changes in the thoracic skeleton (middle section of the spine), which restricts the lungs from fully expanding, it may be necessary to see a pulmonary specialist.

EARS/HEARING DIFFICULTIES

One of the first symptoms of Hunter syndrome is often ear infections, along with a runny nose and frequent colds. This is caused by both physical changes in the body and the thickening of tissues that make airways and

drainage tubes narrower. Without proper drainage, the ear becomes infected or clogged with fluid behind the eardrum, which is normally air-filled. While ear infections are a common childhood malady, children with Hunter syndrome do not "outgrow" these conditions as other children do. Frequent ear infections can cause permanent damage to the eardrum, so it's important to minimize them. When an infection occurs in the inner ear, both balance and hearing can be affected.

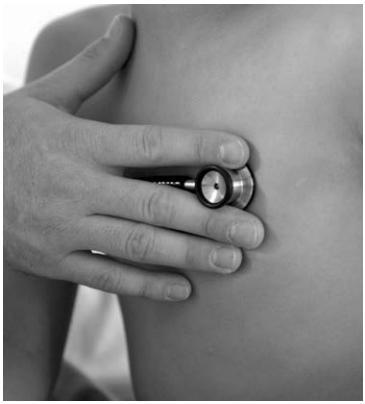
Antibiotics are necessary to clear ear infections. However, chronic infections often require a surgical procedure called myringotomy, which involves making a tiny incision in the eardrum to relieve pressure caused by the excessive buildup of fluid, or to drain pus from the middle ear. A tympanotomy tube is inserted into the eardrum to keep the middle ear aerated over time and prevent the fluid from reaccumulating. Without the insertion of a tube, the incision usually heals on its own in two to three weeks. If a tube is inserted, it will either fall out naturally in 6 to 12 months, or require removal by a physician, depending on the type of tube used. Tube removal is a minor procedure usually performed at an office visit. In some cases, tympanotomy tubes may also improve hearing.

It's important to note that, because Hunter syndrome patients have frequent ear infections, many children develop something called "copathogenicity," which means their bodies don't respond to prescribed medications, such as antibiotics. Therefore, it's imperative that parents schedule follow-up visits with their child's physician to make sure an infection has completely cleared.

Unfortunately, all Hunter patients experience some extent of hearing loss, from mild to severe, due to the constant buildup of fluids and chronic infections. Because communication is such a vital component in a child's development, the use of a hearing aid may be recommended. Caregivers should consult with an otorhinolaryngologist, a doctor who specializes in conditions of the ear, nose and throat (ENT) and related structures of the head and neck, as well as an audiologist, who specializes in hearing disorders.

It's important to note that medications often affect individuals with MPS II differently. Always consult your physician before using over-the-counter (OTC) medications such as decongestants, antihistamines, cough suppressants or other cold remedies.

CARDIAC CONCERNS



It's important for your physician to monitor your child closely for cardiovascular issues. The continued accumulation of GAGs in the heart can weaken the heart muscle, damage valves and narrow the arteries, leading to high blood pressure, heart murmur, leaky heart valves, and in severe cases, heart failure and heart attack.

Early detection of high blood pressure and treatment with antihypertensive medications can prevent or slow further damage. In extreme cases, damaged heart valves may be surgically replaced. Working with a cardiologist is the best way to monitor, diagnose and treat problems with the heart.

SKELETAL AND CONNECTIVE TISSUE ABNORMALITIES

As we've discussed, the buildup of GAGs in connective tissues can cause a wide range of complications from skeletal deformities and joint stiffness, to hernias. Certain conditions such as scoliosis (the abnormal curving of the spine), joint contractures (a shortening of a muscle or ligament connecting a joint), and carpal tunnel syndrome (the compression of nerves in the wrist, which typically causes pain, numbness and/or tingling), may be treated surgically.

Because most children with Hunter syndrome have bulging abdomens due to posture, weakened muscles and enlarged livers and spleens, hernias frequently occur. Hernias result when a part of the abdominal contents pushes out behind a weak spot in the wall of the abdomen. Hernias may also be repaired surgically, but because of weakness in connective tissues, results are not always ideal. Often, the procedure needs to be repeated. Another option to discuss with your physician is the use of supportive trusses instead of surgery.

Joint stiffness and limited range of motion is common in individuals with MPS II. Joint flexibility can often be improved by physical therapy, which reduces stiffness and helps maintain range of motion. While physical therapy cannot stop progressive damage to joints and the resulting decline in motion, it can often help improve joint function, reduce pain and increase stamina.

ANESTHESIA AND SURGERY

When considering any type of surgery, it's important to note that many children with Hunter syndrome don't heal well and often have complications after surgical procedures. This may limit certain surgical options for skeletal and connective tissue problems. For example, surgery to stabilize the spine using internal hardware is difficult when bones are fragile. And, because connective tissues are weakened, surgical repairs of hernias may need to be repeated, as discussed above.

There may also be risks associated with the use of anesthesia due to compromised airways. Children who experience shortness of breath, mouth breathing, snoring and sleep apnea are considered to have "difficult airways," which makes the use of anesthesia riskier. Because anesthesia reduces your ability to breath on your own, it often requires the use of an endotracheal tube, which is inserted into the trachea or throat (either through the mouth or nose) to ensure the proper exchange of oxygen and carbon dioxide. If a patient's airway is compromised or obstructed, this makes the procedure more difficult (and sometimes impossible). These conditions may also cause respiratory problems during the postoperative recovery period.

Therefore, consultations with a surgeon and anesthesiologist are necessary before deciding on surgical solutions. These medical professionals can help you weigh the benefits and risks associated with surgery.

NEUROLOGICAL ISSUES

As discussed, children with Hunter syndrome may be diagnosed with neuronopathic MPS II, which affects the nervous system and is more severe; or non-neuronopathic. About two-thirds of patients have neuronopathic Hunter syndrome, and will experience developmental delays (affecting language and motor skills) and cognitive impairment, such as memory and attention problems, which progressively worsen over time. In addition, sleep disturbances are three times more common in patients who have the neuronopathic disease than in non-neuronopathic patients.

While patients with non-neuronopathic Hunter syndrome are more likely to have normal cognitive abilities, they may still develop neurological issues, such as vision and hearing loss, carpal tunnel syndrome, spinal cord compression and hydrocephalus (fluid on the brain) due to the buildup of GAGs. As discussed earlier, carpal tunnel syndrome and spinal compression may be managed surgically. Hydrocephalus can also be treated by inserting a shunt or thin tube to drain fluid from the brain when the fluid pressure in the skull is too high.

Some children with MPS II also experience seizures, which usually occur early in the disease. If your child has seizures, your doctor may prescribe anticonvulsant medications. It's also a good idea to learn how to deal with seizures, such as the proper way to turn the patient onto his side and make sure his airway is clear.

Because of these complications, neurologists typically play an important role in the diagnosis and management of patients with Hunter syndrome.

BEHAVIORAL PROBLEMS

Often related to the neurological impacts of this disease, Hunter syndrome patients may develop behavior issues. In fact, problems with behavior are sometimes the first symptoms parents report. These problems may include tantrums, obstinacy, aggressive behavior and hyperactivity. Because these behaviors are similar to those found in children with attention deficit hyperactivity disorder, the condition is sometimes misdiagnosed. On the flip side, many children with Hunter syndrome are also cheerful and affectionate.

Although neurological changes play a role, many behavior problems may be a result of frustration – children who are dealing with hearing loss, the inability to verbally communicate wants and needs, and/or limited mobility can naturally become exasperated and irritable. Communicating with children in an age-appropriate way, and helping them to understand their limitations, can help lessen some of these feelings (see Chapter 4: Communicating Effectively).

Some families have found working with a child psychologist or behavior therapist to be very helpful. One parent shared this story about her son, Michael, who was diagnosed with Hunter syndrome at age 4 and is now 9 years old:

"The most helpful intervention was a Behavior Intervention Plan (BIP), which was implemented through and by Michael's school. They collected data over time and were able to pinpoint the antecedents or precursors to his behaviors. Based on this data, we could see that Michael mainly

experienced aggressive or disruptive behaviors during transitions, because he didn't understand what was happening. We began implementing 'first/then' statements to his day. For example, 'first colors, then toys.' In addition, we discovered that Michael needs breaks throughout the day while attending to tasks.

"His aggressive behaviors have decreased significantly since this plan was put in place three years ago. Overall, using school psychologists and behavioral therapists to help with assessments and offer modifications has been invaluable. These professionals can also connect families to additional resources that may help. My philosophy has been to meet with everyone once and then build your team from there. Just like sports, some will make the 'team' and some won't — and that's okay."

WHAT ABOUT MEDICATIONS?

Although medication is used to effectively treat other childhood behavioral problems, such as attention deficit disorder, treating behavioral issues in Hunter syndrome patients with medication has had limited success. Many medications have side effects that can exacerbate other complications of the disease, such as respiratory problems. Nonetheless, some families have found that the right medication can help with behavior issues.

If you are considering medication, it's important to work closely with the prescribing psychiatrist or physician in order to find the drug or combination of drugs that work best for your child, without causing dangerous side effects. Be very observant when starting a new medication for any changes in your child's health.

Another essential aspect to managing behavior challenges is to provide a safe home environment. An occupational therapist can help you make changes around the house to make it easier for your child to get around and do things for himself, as well as reduce opportunities for injuries. Additionally, a speech therapist can improve your son's ability to communicate, while a physical therapist can increase mobility — all of which can reduce frustration and improve quality of life.

It's also important to remember to allow your child to live a meaningful, rewarding life beyond his diagnosis. Include him in family activities whenever possible, encourage friendships, and find out what hobbies and activities interest him. Attending a play group that offers a variety of activities can also keep your child occupied and engaged.

Helping your child learn as much as possible while his brain is working well can also be very therapeutic. If your son goes to school, work with the staff to develop an individualized education program (IEP) for him. He may be eligible for one-on-one attention in the classroom or help for other issues, such as hearing problems.

Since behavior issues often change over time as the disease progresses, parents need to be flexible and make modifications. In other words, what works now may not be effective in the future. Although children with Hunter syndrome are sometimes difficult to manage, understanding, compassion and love can go a long way to bring out the best in your child.

SLEEPING, EATING AND TOILETING ISSUES

As discussed, children with the neuronopathic or severe form of Hunter syndrome typically have more sleep disturbances than those with the non-neuronopathic (attenuated) type. However, most MPS II patients experience some form of disrupted sleep patterns or restlessness, which can worsen over time. The use of medications, including melatonin and sedatives can often improve sleep, but it sometimes takes a bit of trial and error to find what works best for each child. Always discuss medications or supplements with your child's physician before use.

Besides medication, good sleep habits can make a positive difference. Try to keep a strict bedtime schedule and make sure the room is well darkened, as light can be disruptive. Creating a safe environment in your child's room can also help him (and you!) sleep better by reducing the chance of injury. Suggestions include putting the mattress on the floor, padding the walls, removing all hard furniture, keeping only soft, safe toys in the room, gating stairways and using a monitor. Some parents have found it helpful to use a special bed that helps contain their child, therefore preventing falls or other injuries during the night and helping them to rest easier. There are also pads available to place under the carpet outside the child's bedroom door that cause a bell to ring if he leaves the room. Remember, it's vital for parents/caregivers to get proper sleep in order to cope during the day and stay healthy!

When it comes to eating, there are usually very few problems in the early stages of the disease. As the disorder progresses, however, the ability to chew and swallow food may become difficult. In that case, foods need to be mashed or pureed to the right consistency to avoid choking. Sometimes sputtering and coughing while eating becomes a problem. Parents can learn how to make swallowing easier by moving their hand gently under the chin and slowly down the throat. An occupational therapist can provide guidance in this area.

Some children with Hunter syndrome will also develop gastrointestinal issues, including upset stomach, vomiting and diarrhea. Dietary changes and medications may improve these symptoms and should be discussed with your physician. Although there is no scientific evidence that any specific diet is helpful for MPS II, many parents have found that a change in their child's diet can ease symptoms such as excess mucus, diarrhea or hyperactivity. A dietitian may recommend reducing or eliminating certain foods that are known to exacerbate problems.

While proper nutrition is very important for all developing children, it is even more critical for those combatting disease. The body needs the right balance of nutrients to boost immunity and maintain healthy function. If your child becomes a picky eater or has trouble with certain foods, such as meat, you may want to consult a nutritionist to help you develop a special diet that fits your child's particular needs, while ensuring proper nutrition.

Children who experience cognitive problems will often have trouble with toilet training or eventually lose their skills in this area. Physical limitations may also make using the toilet more difficult. Working with an occupational therapist to outfit your home with raised toilet seats, grab bars and other special equipment can prolong independence. Incontinence issues and accidents can be minimized by using special undergarments.

TREATMENT OPTIONS

Although there is no cure for Hunter syndrome, certain treatments have had some success by slowing the disease's progress and lessening its severity. Additional information on potential treatment options is available at rarediseases.org.

CLINICAL TRIALS

Families can get the updated status of the latest research and ongoing clinical trials for patients with Hunter syndrome by searching the online database at www.clinicaltrials.gov. This is a registry of federally and privately supported clinical trials conducted in the U.S. and around the world.

(See Resources for a list of organizations that can help families stay informed about medical progress and advances in managing Hunter syndrome.)

A CAREGIVER'S PERSPECTIVE

In the beginning, parents/caregivers often find it difficult to navigate the complications of Hunter syndrome and still live life as normally as possible. The best advice experienced parents offer is this: Don't dwell on what might happen, and instead deal with things as they occur. In other words, take it one step at a time.

It's normal to kick into "fix it mode," focusing all your energy on finding solutions to every symptom or complication. But that can be a recipe for "burnout." While it's important to take care of medical issues and improve your child's quality of life, it's also critical to reserve some energy for simply living.

Children with Hunter syndrome can attend school, make friends, pursue hobbies and interests, and flourish in their own way. Finding a balance between caregiving and just being present for your child can be challenging, but once you find it, many parents begin to feel more at peace and better able to cope with complications as they arise.

Finally, experienced parents urge those who are dealing with a new diagnosis to trust their gut when it comes to symptoms and complications. Physicians are only human, after all, and may miss things that parents are aware of because they know their child so well. For example, one parent shared a story about how her son never exhibited the typical symptoms associated with ear infections such as pain, ear tugging or noticeable inflammation. Instead, she could tell he had an infection when he started asking for more water and having trouble sleeping through the night. Because these symptoms are atypical, she had to urge their physician to "double check" for infections. Her advice: Never be afraid to advocate for your child. You know him or her better than anyone. While it can be intimidating to second-guess medical professionals, it's worth the effort, and most physicians are willing to listen.

Chapter 3:

Getting Organized

Getting Organized

"For every minute spent organizing, an hour is earned."

– Benjamin Franklin

To say that caregivers have a lot to think about and even more to do, is an understatement! Most caregivers report high levels of stress. While stress can stem from many things, such as worry, guilt, and helplessness, much of the anxiety caregivers encounter is caused by having too many demands and too little time. Unfortunately, there is no way to eliminate stress from this role, but parents/caregivers can learn to reduce stress and find healthy ways to cope with it (see Chapter 5: Care for the Caregiver).

One of the best ways to reduce caregiver stress is organization. Now, your first reaction might be, "I have no time to get organized." However, experienced caregivers will tell you that getting and staying organized can help you save precious time, avoid stressful situations, and handle emergencies more easily. There are many aspects of this journey that you can't control, but organization is one area that can help you feel empowered. In fact, it can be a life saver!

GENERAL ORGANIZATIONAL TIPS

Some simple ways to get organized include:

Use a Daily/Weekly Planner – Whether you prefer a book-type planner, computer calendar or smartphone app, find an easy method for keeping track of appointments and reminders – and use it! Be sure to take your planner with you to doctor's appointments so you can easily schedule future visits. You can also use your planner to jot down reminders to refill medications, schedule tests, and take care of other necessities. Some caregivers have found that using a color-coded system (e.g., red for medical appointments, blue for family activities, green for school, etc.) can make planning at-a-glance easier. Whatever system you choose, a planner can help you avoid scheduling conflicts, unnecessary follow-up calls, and forgotten important appointments, and allow you to schedule time for yourself – all of which reduces stress.

Make Lists – Trying to keep everything in your head is nearly impossible. By writing things down, you eliminate the stress of trying to remember a million details. For example, keeping an ongoing grocery list so you can write down items as you run low or think of them, helps you to avoid forgetting items and making repeat trips to the store. Similarly, keeping a list of questions you

want to ask the physician at your next appointment (as issues arise) is much easier than trying to recall everything that occurred between visits.

Create a Schedule/Routine – Although schedules can become completely disrupted by emergencies, appointment changes or a child that is not feeling well, planning your days and weeks, and establishing a routine is a stress-reliever. Both schedules and routines can help you get more accomplished and feel in command of the situation (at least for a little while). Try creating a daily/weekly schedule of tasks (using that handy planner) and stick to it as much as possible. For example, Monday might be grocery day, while Sunday night is a good time to refill your child's pill box for the upcoming week. If you're sharing caregiving duties with others, make a master schedule of duties, along with who is responsible for each one.

Plan Weekly Meals Ahead of Time – When days are hectic, the last thing you need is to scramble around trying to put a nutritious meal on the table. Resorting to fast food or pizza delivery works in a pinch, but isn't a healthy solution over the long term. Many caregivers have found it helpful to plan a week's worth of meals, adding the necessary ingredients to the grocery list and doing some prep work ahead of time. Remember, nutritious meals need not be elaborate! When you have the time, try cutting up fruits and vegetables for snacks or quick meals, making a pot of stew or soup that can be reheated, or preparing extra portions that can be frozen for busy nights. If you're not sure where to start, there are plenty of cookbooks that feature quick, healthful, "make ahead" meals.

Learn to Say "No" – Many people feel obligated to do everything they're asked to do. Learning to say "no" to tasks that are low on the priority list – and not feel guilty about it – can not only alleviate stress, but free up time to do something you really want to do. At first, prioritizing tasks can be difficult, but with practice you can learn to let go of obligations that are not critical. For instance, is it necessary to make homemade cookies for the class party or will store-bought work just as well? Do you need to host the holiday gathering, or can someone else take over this task? (One word of caution here: saying "no" to social occasions can be detrimental to your mental well-being. When possible, it's important to stay connected with family and friends, and attend events that are enjoyable.)

Organize Supplies – By taking the time to organize caregiving supplies, you'll save valuable time and frustration later. After all, trying to find something when you need it in a hurry can leave you frazzled. You may want to put similar supplies in handy bins instead of scattered about the house in different drawers. For instance, all first-aid supplies (wound care essentials, thermometer, ibuprofen, etc.) should be contained in one place. Other "bins" may include: medications, bathing supplies, and even grab-and-go snacks.

DEALING WITH DOCTORS

Individuals with Hunter syndrome typically require the care of numerous specialists, which means you'll be spending a good deal of time at medical offices and sometimes hospitals. In fact, going to medical appointments and keeping track of information can sometimes feel like a full- time job! Thankfully, there are ways to make this aspect of caregiving a bit easier for both parents and their children.

The first step is to find a primary care physician or pediatrician with whom you can develop a good relationship. Studies show that people who have a good relationship with a physician report greater satisfaction with their care and enjoy better health. Most people start with a primary healthcare provider who manages overall care, treats routine aliments, schedules regular tests and refers you to specialists as needed. Because this person is central to your child's care, it's important to take time to carefully select a primary care physician or pediatrician. When choosing a physician, you can:

Ask for recommendations – In addition to healthcare providers, family, friends and other parents dealing with MPS II can often be a valuable source of information. After all, nothing beats firsthand experience.

Check with your insurance – Be sure the physicians/specialists you're considering are in your plan's network. If you're choosing a new insurance plan, you may want to pick your primary care physician first and then select the plan based on his or her affiliation. If you want to see a physician outside of your network, find out what the out-of-pocket costs will be for office visits and treatments ahead of time.

Do some research – Check the Administrators in Medicine (AIM) website

(http://docboard.org/aim) to find out whether a physician is in good standing with state licensing agencies. AIM provides information about disciplinary actions or criminal charges filed against physicians in many states. Other sites such as Healthgrades.com or RateMDs.com may also provide useful background.

Consider hospital affiliation – Find out which hospital the physician is affiliated with. Are you familiar and comfortable with this facility if hospitalization is necessary?

Interview the physician – Most physicians will allow you to schedule an initial consult or interview. As mentioned, developing a rapport with this individual is critical to good care, and there is no better way to "get a feel" for a physician than with a face-to-face meeting. While you're there, pay attention to other service indicators, such as: how long is the average wait to be seen; does the office offer same-day appointments for urgent issues; if you have a question, does the physician's assistant or nurse return your call promptly; is there an after-hours answering service; and who will be seeing your child when your primary care physician is unavailable or on vacation?

INTERVIEWING A PHYSICIAN

During an interview with a physician, here are some factors to consider:

1. Does he or she treat you and your child with courtesy and respect?
2. Is his or her personality compatible with you and your child? Do you feel comfortable talking with him or her?
3. Does he or she listen without interrupting?
4. Does he or she answer your questions completely and clearly?
5. Does he or she explain diagnoses and treatment options patiently and in a way that's easy to understand? Does he or she encourage follow-up questions?
6. Does he or she provide "patient-centered" care in which they value the input of parents/caregivers?
7. Is he or she open-minded enough to consider your opinions/ suggestions or possible alternative treatments?

Source: *The Complete Caregiver's Organizer: Your Guide to Caring for Others While Caring for Yourself.*

MAKING THE MOST OF YOUR APPOINTMENTS

Once you've found a primary care physician and assembled a team of specialists, it's important to keep communications open and make your appointments productive. The best way to do this is by being prepared with information and questions at each visit. Remember, physicians only have a short period allotted to see each patient. Being organized will help you make the most of that time.

Keeping a Symptom Diary

As mentioned, it's nearly impossible to remember everything, especially when you're dealing with multiple symptoms and complications. Jotting down notes in a journal or health diary is a great way to keep track of changes in your child's health, as well as questions you have for the physician.

For example, if a new medication makes your child feel nauseous or too drowsy, you may want to ask the physician for an alternative. Perhaps you've noticed that your child's sleep pattern has changed recently or his appetite has been off, and you'll want to run this by your physician at your next visit. By keeping a symptom diary and list of questions/concerns, you will not only be helping yourself to remember important details, but you'll also be providing the physician with the accurate information he or she needs to provide your child with the best care possible.

Over time, a symptom diary can provide clues to improve healthcare. For instance, you may notice that certain foods, such as dairy, exacerbate problems with excess mucus or cause gastrointestinal issues, thus leading to a discussion with your physician about possible dietary changes. Emerging patterns in bedtime routines or physical activity can also help you make positive adjustments. This is particularly important in a progressive disease like Hunter syndrome, when symptoms and complications continually change.

Don't think of this diary as "just one more thing to do," but rather as a helpful tool. 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 it's a regular spiral-bound notebook or a digital diary. Be sure to make daily entries, while symptoms/concerns are fresh in your mind. You can also use this diary to record positive events, such as, "Today my son had a lot of energy," or, "He really enjoyed a new game we played." In this way, the diary is not only useful from a medical standpoint, but may also offer therapeutic benefits.

Maintaining Good Records

While keeping official medical records is the responsibility of physicians, maintaining your own records regarding doctors, diagnoses, treatments and symptoms is very important. Having all the 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.

You will often find yourself answering the questions of one specialist with respect to other treatments. For instance, each new physician will require a list of previous surgeries and dates, along with a list of current medications. Having your own records will make it simple to answer these questions and keep track of multiple therapies. In addition, although it's something we'd rather not think about, if anything should happen to you, having organized, written records will be important to the continuing care of your child.

Some caregivers have found 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 (for those urgent questions that always seem to crop up in the middle of the night!). 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.

Sample Physician Record

Physician's Name:	
Specialty:	
Address:	
Phone:	
Answering Service:	
Date of First Visit:	**Reason:**

Visit Date	Recommendations / Notes	Medications Prescribed	Test/Results

Finally, since it's often difficult to listen attentively to a physician and take notes, while dealing with a child who may be overactive or disabled, it's a good idea to bring a family member or friend to these appointments with you, if possible.

MANAGING MEDICATIONS

It's not unusual for individuals with MPS II to be on multiple medications for a variety of health issues. Modern medications can be extremely beneficial when taken properly, but failing to take them as directed can cause serious consequences. 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 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 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 will 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 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 child's 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.

SAMPLE MEDICATION LIST

Medication List for:
Date updated:

Medication name:
Dosage:
Directions:
Reason for taking:
Prescribing doctor:

Medication name:
Dosage:
Directions:
Reason for taking:
Prescribing doctor:

Medication name:
Dosage:
Directions:
Reason for taking:
Prescribing doctor:

HOW TO MAKE THE MEDICINE GO DOWN

Giving medicine to young children can sometimes be tricky. Here are a few tips from parents and physicians to help the medicine go down — without that spoonful of sugar!

- **Put on a Happy Face** — Kids pick up on negativity and stress. If you act positive about "medicine time" and make it seem like an enjoyable thing, your children will feel less stressful. For very young children, playing a game like peekaboo or "here comes the choo-choo" can help children happily accept a dropper or syringe.
- **Bypass the Taste Buds** — Some kids will spit out bitter-tasting medicine. To avoid this, some parents choose to continue to use syringes and droppers even when their children are old enough to drink out of a cup. A syringe or dropper can administer the medicine along the inside of the cheek and off the tongue. However, NEVER squirt medicine down the back of the throat, which may cause choking or gagging.
- **Disguise the Taste** — Ask your pharmacist to add flavors such as chocolate, root beer or grape to liquid medicines. This can be especially helpful when children are on medication for prolonged periods. You can also give the medicine a fun name, such as Chocolate Chugger or something associated with a favorite character. Another trick for icky-tasting medicine is to have your child suck on ice chips before taking the medicine, which numbs the taste buds. (Note: This should only be used with older kids as it may be a choking hazard for young children.)
- **Give Choices (when possible)** — Taking their medicine is never a choice, but you can provide options such as using a dropper or a cup. You might let your child decide if he wants to take his medicine before or after an activity, as long as it's within the correct dosing time. Having some choice gives children a sense of empowerment.
- **Let Them Play Doctor** — Have your child pretend to give a stuffed animal medicine before he takes his. "If Mr. Bear can do it, then so can you!" This can make taking medicine a more comfortable experience.
- **Be Honest** — Don't tell children that medicine will taste good if it's won't. Once they're old enough to understand, you can explain that although taking medicine is no fun, it's necessary to help them feel better. By making it part of their daily routine, like brushing their teeth (we must brush to prevent cavities) or taking a bath (we must bathe to stay clean), taking medicine will eventually become normal and expected.

Adapted from "*9 Clever Ways to Help the Medicine Go Down,*" by Katie McDonald, American Baby.

HANDLING HOSPITAL VISITS

Whether your child is admitted through the emergency room or for a specific procedure, hospital stays can be stressful. This is especially true for children who may not understand what is happening and are fearful of the strange surroundings. MPS II patients with severe cognitive impairment can be particularly distressed by the hospital environment.

With this in mind, many hospitals offer a pediatric emergency room and a pediatric ward that caters to the special needs of children. They typically provide a less chaotic, calmer environment, along with a colorful, "fun" atmosphere that can ease fears. In addition, the doctors and nurses often specialize in pediatrics, which may result in better care.

This is where your record keeping really comes in handy! During admittance to a hospital, you can provide the personnel with a prepared list of medications, as well as a list of specialists and treatments, which can help the hospital staff provide more immediate and improved care (and make the process less stressful for you).

During your child's hospital stay, never be afraid to ask for special accommodations such as thicker mattresses (to relieve joint discomfort), safety rails, raised toilet seats, or other amenities that are necessary for the comfort and safety of your child. Because Hunter syndrome is a rare disorder, hospital staff may not be immediately familiar with your child's needs.

To alleviate some of the stress associated with a hospital stay, schedule a time with the physician/surgeon to find out as much as possible about the procedure, treatment plan, or surgery, including what to expect during recovery. You may only have a few minutes with a specialist or surgeon, so being prepared with a list of questions and concerns will ensure that you don't forget important details. While physicians can be difficult to pin down in a busy hospital, nurses can be a great resource. It's a good idea to get to know the nursing staff that will be caring for your child. Again, don't be afraid to ask questions and keep good notes. Open, two-way communication is necessary for quality care.

Some questions may include:

- What test is being performed, why is it necessary and what will it tell us?
- What treatment is needed and for how long?
- What are the benefits and risks of the treatment or surgery?
- When can the patient expect to be discharged?
- When the patient goes home, will there be any special instructions regarding activities or diet?
- What type of follow-up care is needed?
- Who should we call if we have additional questions?

IN CASE OF EMERGENCY

You never know when an emergency will occur, so it's wise to be prepared with the following:

- Personal identification
- Primary physician's name and phone number
- Family members' names and phone numbers
- Health records and list of medications
- List of allergies, if applicable
- Health insurance information

It's also handy to keep a "hospital bag" packed and ready to go. This can be used for doctors' visits, as well. Things to include:

- Paper and pen for taking notes
- Books or magazines, along with games, cards or puzzles for children (electronics are not allowed in many areas of the hospital)
- Protein/energy bars or other snacks
- Diapers/wipes
- Bottled water
- A sweater or jacket
- An extra charger

The National Institute of Health, which is part of the U.S. Department of Health and Human Services, offers the following recommendations regarding hospital stays:

What to Bring

It's best to bring as little as possible to the hospital. Leave valuables, such as jewelry, cash, credit cards and your checkbook at home. Also, don't bring electronic shavers, hair dryers, curling irons or similar equipment. If you do bring electronics, such as cell phones, tablets, or laptops, keep in mind that hospitals will not be responsible for the loss or theft of these items. When packing for a hospital stay, it's best to stick with the necessities, such as the patient's insurance card and photo ID, a robe and non-skid slippers (most hospitals provide bed clothes and may not allow pajamas from home), toiletries (i.e., toothbrush, toothpaste, deodorant, brush), hearing aids, glasses, a warm sweater, and books or magazines.

Admission

If a hospital stay is planned, you may be able to complete pre-admission questions over the phone. This will save time when you get to the hospital, although forms must still be signed. Also, be sure to check admission forms for accuracy, including personal information, what test, surgery or other procedure is being done, and the name of the physician/surgeon. You will also be asked about advance directives, including a DNR (do not resuscitate) order. If you don't have these prepared, the hospital can provide forms.

Once forms are completed, the patient will receive a hospital bracelet or ID. Read the bracelet to make sure the information is accurate. Many hospitals now use a digital scanning system to ensure patient information is accurate and will ask you to verify this data.

Going Home

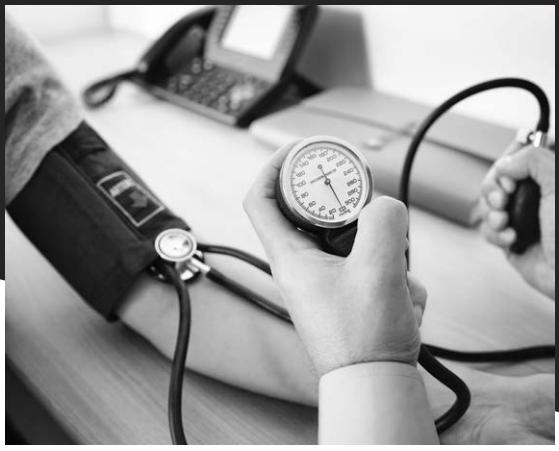
Depending on the patient/procedure, a social worker may be assigned to assist the family with home care, rehabilitation or palliative care. Additionally, an occupational therapist may be assigned to assess the safety of the home environment and suggest changes, or recommend follow-up therapies. Be sure to read and follow all discharge instructions, including follow-up treatments. Ask questions about anything that is unclear before leaving the hospital.

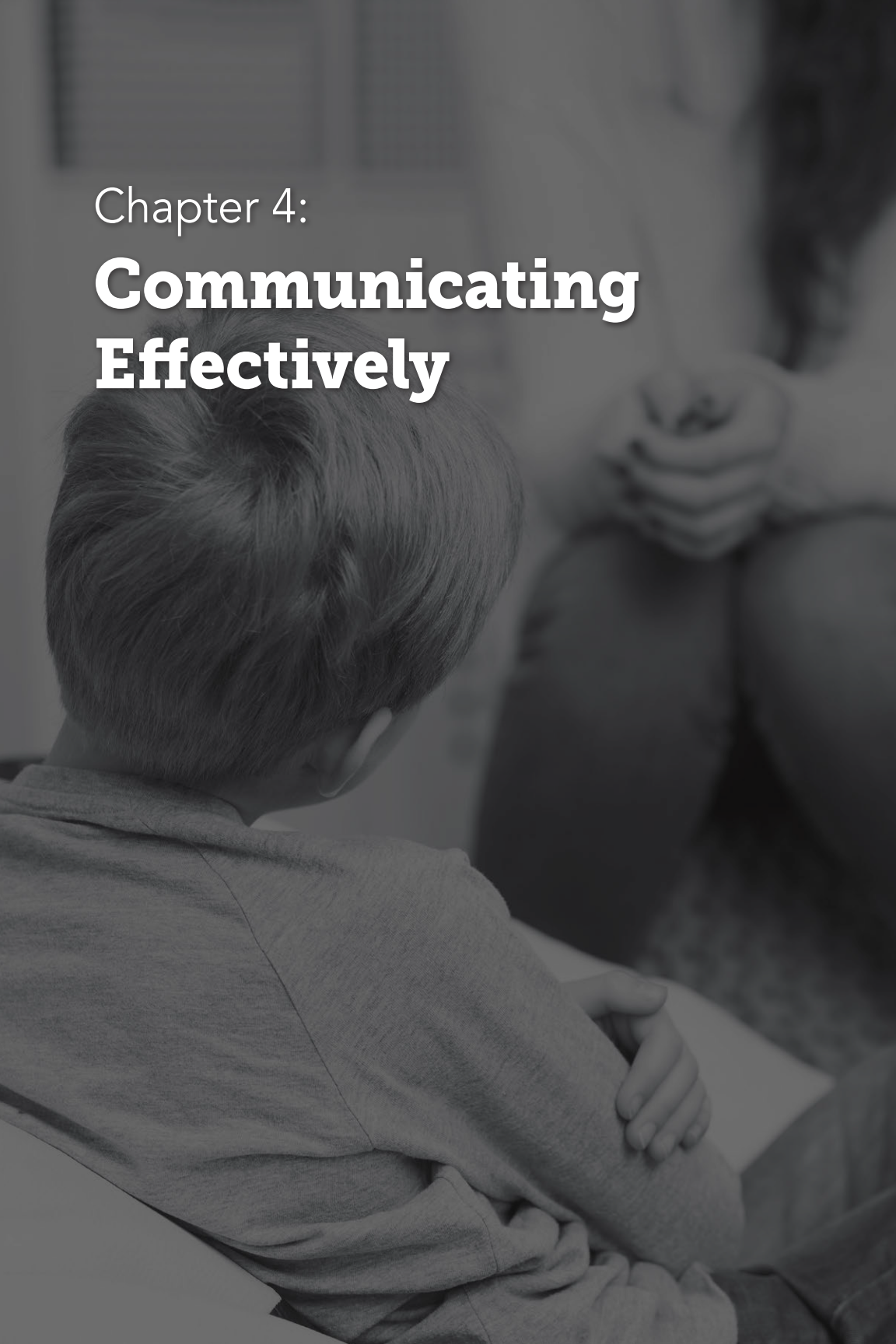
A CAREGIVER'S PERSPECTIVE

Although caregivers typically spend a great deal of time visiting physicians for their child, they never seem to have time to check in with their own doctor. This is understandable, because if you do have precious free time, the last thing you want to do is schedule yet another medical appointment! However, experienced caregivers (and physicians) stress the importance of caring for yourself, which includes regular check-ups and routine testing. Neglecting your own health will not help anyone, including the person under your care.

An essential part of being organized is scheduling annual physicals and routine testing for yourself to monitor weight, blood pressure, cholesterol and any changes in your health. Be sure to tell your doctor about your child's condition and your role as a caregiver. It's not uncommon for caregivers to experience weight gain (from lack of exercise or poor diet), weight loss (from skipping meals), insomnia, anxiety or depression. Regular appointments with your physician can help you get a handle on these health issues, prevent worsening symptoms, avoid more serious complications, and most importantly, help you feel better.

Also, don't forget the importance of mental health. Talking to a therapist or psychologist can be very beneficial when dealing with the stress associated with caregiving.





Chapter 4:

Communicating Effectively

Communicating Effectively

"The two words 'information' and 'communication' are often used interchangeably, but they signify quite different things. Information is giving out; communication is getting through"

– Sydney J. Harris

All humans have a desire to be heard and understood. It sounds simple enough, but of course, we all know that communicating effectively can sometimes be difficult – especially when you're trying to make sense of a complicated disease like Hunter syndrome. And yet, communication is important to many aspects of living with this disease, from receiving quality care, to maintaining bonds with family and friends.

In the last chapter, we discussed ways to deal with doctors and hospitals, including tips for improving two-way communication (e.g., keeping a symptom diary, asking questions and taking notes at appointments, maintaining good records, and trusting your gut instincts when advocating for your child). In this chapter, we'll focus on ways to communicate with your child, school staff, family members and others who will share this journey with you.

The first step in this process is to educate yourself about your child's disease. As mentioned, information can be empowering – helping you and your family prepare for and manage the long road ahead. It can also help you communicate more effectively. Keep in mind, that learning about Hunter syndrome is a process: You can read all the information in the world and still not know everything there is to know until you experience it! Parents and caregivers find that they continually learn new things as they talk to other parents, join support groups and seek advice from specialists. All this knowledge will help you answer questions your child and others will have about this disease.

TALKING TO YOUR CHILD

The first challenge for many parents is deciding how much information to share with their child. Children with neuronopathic or severe MPS II are incapable of understanding what is happening, and therefore, communicating specifics is not necessary. For these children, it's more about making them feel secure and comforted. For those with non-neuronopathic or attenuated MPS II, communication plays a more important role. (The following recommendations apply primarily to children who are able to comprehend their diagnosis.)

As parents, we are naturally protective of our children and their feelings, so it's normal to worry about causing undue anxiety and stress. However, children are very perceptive and usually know when their parents are not being truthful with them. It's best to be open and honest with your child regarding his disease, in an age-appropriate way. Children process information differently at each stage in their development. Understanding these stages can help you communicate with your child.

Infants and Toddlers

At this age, children have little to no understanding of their disease. Because they're just beginning to develop trust and a sense of security, they experience pain, restriction of motion and separation from parents as challenges to this development. Children at this age are often more afraid of being separated from their parents and the medical procedure they don't understand than any symptoms or complications. Therefore, a parent should be present for procedures to hold, sooth and interact with your child as much as possible.

You can also reduce the fear of strange environments by bringing along familiar, comforting items, such as blankets and favorite toys. Distraction is often effective for children at this age.

Pre-School Children

At this age, children are beginning to develop a sense of independence. While they understand what it means to be sick, they probably won't comprehend the cause or nature of their disease. Fear of pain or being hurt is very common at this age. Again, it's best to be honest and avoid saying things like, "This won't hurt," if the procedure will cause some discomfort. Instead, try explaining that the test or procedure may hurt or sting, but that the discomfort will be temporary and is being done to help make them better. Assure them of your presence and support.

Many children may begin to "act out" at this age, testing their limits and challenging parental control, because they lack control over their world. All children at this age love the word "No!" However, children dealing with health challenges may be more likely to be obstinate. Never ask your child,

"Do you want to take your medicine?" which gives him a chance to refuse and may lead to a battle of wills. Instead, try offering some options, such as, "Which medicine do you want to take first?" or, "Would you like to sit on my lap during this test or in the chair with me holding your hand?" This way, your child will feel like he has some control over the situation.

Early School-Aged Children

Children at this age are busy discovering their environment and trying to master it. Those children with normal cognitive development will begin to understand the reasons for their symptoms and complications. However, many children will struggle to make sense of the situation and therefore try to come up with their own "reasons" (e.g., believing they are somehow responsible). It's important for parents to reassure children that this disease is not due to something they've done.

School-aged children are also more likely to understand that they'll need to take medicine and receive treatments to feel better. Using books (see sidebar) and simple visuals/images can help you explain the challenges they are experiencing.

It's important for a child to know that he has a disease and needs extra care. Explain that doctors' visits, tests, medicine, and other potentially scary procedures are "good things" because they're helping to make sure he's okay. Again, be honest about procedures that may cause discomfort, but assure your child it will be temporary, and you'll be there to offer support. It can be helpful to prepare your child for an upcoming procedure, which will reduce the fear of the unknown. Both parents and physicians should explain what the procedure is, why it's being done, and what to expect (in an age-appropriate way).

Books can be great resources for school-aged children and parents coping with chronic conditions. They can help you explain things in simple language and with the help of images. Although there is not currently a book available that deals specifically with Hunter syndrome, many touch on the same challenges, questions and feelings that arise with all chronic diseases, such as "When Will I Feel Better?: Understanding Chronic Illness," by Robin Prince Monroe. For a list of books to consider, visit https://www.goodreads.com/list/show/8536.Books_for_Children_with_Chronic_Illness

Keep in mind, physicians and social workers can help parents speak to children about a life-changing diagnosis. They have developed effective ways to explain the situation in a way that children understand, so don't be afraid to rely on their expertise.

Older School-Aged Children

Even though children with attenuated MPS II are more capable of understanding their disease and the necessary treatments at this age, they should not be expected to react like an adult. They're recognizing that they're different from other children and may feel left out when they're not able to interact with their peers. It's natural for parents to "protect" their children from hurtful remarks by limiting activities with other children, but this can often do more harm than good. It's important to a child's independence and self-esteem to allow them to participate in school and social activities as much as they are able.

Even children with severe cognitive impairment can enjoy supervised activities and interactions with others. Look for groups that offer special activities designed for children with developmental challenges.

As children get older, the questions often become tougher to answer. For instance, it's common for a child to ask, "Why me?" It's okay to offer an honest answer: "I don't know." You can explain the basics of what causes Hunter syndrome, but reassure your child that it's not due to anything he has done. It's not anyone's fault. If your child says, "It's not fair," you can acknowledge that he's right – it's not fair. Assure him that having feelings of anger and frustration are okay, as long as those feelings are processed in a healthy way.

In fact, children will naturally have many feelings about the changes affecting their bodies, and should be encouraged to express those emotions. Sometimes simply taking the time to ask what your child is experiencing and listening without offering "solutions" is enough. Keep in mind, not all communication needs to be verbal. Kids can express themselves through music, drawing, writing and other activities.

Perhaps the toughest question you'll ever have to answer is, "Am I going to die?" The response will depend on the medical prognosis, as well as the age and maturity of your child. It can help to ask your child what specific concerns they have and address them individually. If you have religious, spiritual or cultural beliefs about death, these can be very comforting. However, therapists caution against using euphemisms such as "going to sleep," which can cause irrational fears about going to bed at night or other people falling asleep. Again, there are some good, age-appropriate books that address the subject of death.

Adolescents and Young Adults

Most adolescents begin to develop their own identity and seek greater independence. At this stage, your child will probably be concerned about the impact of his symptoms and complications on his daily life, such as school activities and interactions with friends. If there are no cognitive issues, teenagers are capable of understanding more complex explanations and becoming more involved with their treatments. Encourage your child to monitor and manage their own treatments as much as possible and involve them in some of the decision-making. They may be able to ask physicians their own questions and express concerns, which should be supported.

All adolescents become very aware of and concerned about self-image. This can be especially difficult for teenagers with MPS II due to the physical changes it causes. It's important to talk openly and honestly about these issues. Listening to your child's concerns or feelings without judgment and offering support is the best way to help him cope with a difficult situation.

Older children will also need to learn to communicate with others, if possible. You can help by suggesting simple explanations they can use when explaining the disease to others and role-playing situations. Help him to anticipate and answer possible questions about his disease.

It's normal for a child's thoughts and feelings to change over time. For children with severe Hunter syndrome, mental awareness will decrease as the disease progresses. In fact, these children often begin regressing early enough that they don't understand what's happening. Therefore, the impact of these changes is felt by parents rather than their children.

For children who do understand the changes they're experiencing, consulting with a therapist can be very helpful, not only for your child, but for advice on how to communicate your own feelings and emotions. Many parents feel they

must hide their feelings or maintain a "brave face," but it's okay to share some of your concerns and emotions.

Of course, you can't always promise that everything is going to be fine. The best you can do is to listen to your child, acknowledge his feelings, which may include sadness, anger, fear or depression — and assure him that having those emotions is normal. In the end, knowing that he has your love and support, as well as the love and support of family and friends, is the best comfort.

(Note: If your child begins to show signs of clinical depression, such as loss of appetite, loss of interest in things he normally enjoys, sleeping excessively or withdrawing socially, it's best to seek professional counseling.)

Limits and Responsibilities

It's important for parents to help their children lead as normal a life as possible, which includes treating a child with Hunter syndrome like any other child, as much as possible. Of course, your child will have special healthcare needs, but resist the urge to spoil or coddle a child simply because he has MPS II. For instance, you can provide some normalcy by setting limits in the same way you would another child, such as siblings. Stick to set bedtimes, enforce time limits on television or electronics, and demand acceptable behavior, to the extent that is possible.

It's important for parents to understand that behavioral problems in children with severe MPS II stem from the disease and are not the result of bad parenting! Behavioral issues can be one of the most difficult aspects of the disease to manage. When behaviors are unacceptable, don't be afraid to discipline in a manner that's appropriate and understandable. For children with cognitive impairment, providing a distraction from the problematic behavior can be effective. For all children, try to be consistent. Many parents find it difficult to set limits and discipline children with Hunter syndrome or other chronic illnesses due to feelings of guilt and pity, but setting limits can be very reassuring for a child. Meanwhile, overindulgence can make behavioral issues worse and lead to insecurity.

Giving your child responsibilities within their abilities is also a good way to make a child feel like a normal part of the family. It can boost self-esteem and go a long way toward keeping the peace with siblings! Try assigning simple household chores, if possible, and remember to praise his efforts or provide consequences if chores are not completed. All children need to feel like they belong and contribute.

Balancing your child's illness and abilities with creating a "normal" life can be very challenging, but is beneficial for both your child and your family.

- Be sensitive to signs that your child wants to talk about the disease, and don't change the subject or distract him. On the other hand, if he isn't ready to talk, don't force the conversation.
- Encourage questions, even though it may be difficult to answer them.
- Use comforting language to make your child feel safe.
- Let your child be a kid. Let him play and interact with others, as much as possible.
- Continue to be a parent, setting appropriate limits and responsibilities, as you would if your child was well.
- Don't feel guilty about celebrating special occasions (denying yourself or other family members happiness due to guilt); encourage family interaction and participation in activities.
- Get siblings involved in the care of the child with Hunter syndrome, without it becoming a burden or worry.

TALKING WITH SIBLINGS

Caring for a child with a chronic disease like MPS II demands a lot of your time. If you have other children, it can leave them feeling jealous, angry, lonely, left out and even unloved. In some cases, siblings may wish they were sick, too, to get more attention. They may also seek attention by acting out in various ways. It's important for parents to reassure siblings that they're loved, but that inappropriate "attention-seeking" behavior is not acceptable.

As difficult as it may be, try to set aside time to devote to siblings. One-on-one dates, such as an afternoon spent engaging in an activity of their choice can go a long way toward making a sibling feel special. Don't feel guilty about hiring someone or asking a friend to watch your child with Hunter syndrome so you can attend activities, such as sporting events, with his siblings.

Siblings can also become stressed out from worry (i.e., worrying about their sick brother, their parents, about developing the same disease, etc.). Siblings may also suffer from feelings of guilt. Since it's common for children to keep these feelings to themselves, it's important for parents to be cognizant of their potential concerns. Find a quiet time to ask them how they're feeling and what they might be worried about, and then address these concerns

individually. You may have to initiate the conversation with specific questions, such as, "What are you worried about?"

Parents can help siblings by providing as much information as possible. The amount and type of information you provide will, of course, depend on the age and maturity level of your children. The same rule applies to siblings regarding honesty – be as open and honest with your explanations as possible. Encourage siblings to ask questions and express their concerns. Be aware that young children process information slowly. They may not have any questions initially, but think of things over the days and weeks following your explanation. They may want to attend physician visits with you to alleviate fears of the unknown. Meeting with the doctors, nurses and therapists that care for their brother can help reduce fears and increase understanding.

If possible, get siblings involved in the care of their brother. Children like to feel helpful and important to the family. Look for appropriate tasks that siblings can do, and be sure to praise their efforts. Siblings can also be included in counseling or therapy sessions, where they may be more comfortable expressing their emotions.

All children do best when their daily routines are predictable and consistent. This is not always possible when dealing with a child with special needs. However, maintaining family routines (mealtimes, bedtimes, regular activities) as much as possible will make life easier on siblings, as well as the rest of the family. Siblings should continue to attend school and their usual activities, even if it means enlisting friends and other family members to help with transportation. Continue to celebrate special occasions and schedule time together, such as game night or movie night, in which everyone can participate.

Finally, you may want to talk to your other children's teachers, school counselors or coaches and let them know about your child with Hunter syndrome and how the disease has impacted your family. They can keep an eye out for signs of stress or behavioral issues.

COMMUNICATING WITH FAMILY AND FRIENDS

Telling family members and friends about your child's diagnosis can be emotionally exhausting — and in some cases therapeutic. Repeating the same information to multiple people, all of whom will have questions and concerns, is like reliving the news over and over. And yet, talking to others can provide some relief. How do you find the right balance?

There is no one right way to tell loved ones about the fact that your child has Hunter syndrome, but it's important that close family members and friends 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:

- Tell one trusted family member or friend and ask that person to spread the word among your loved ones.
- Meet with family members and friends individually to talk about the basics, and provide written information for them to review.
- Hold a "family meeting" to explain the news so you don't have to explain multiple times.
- Ask a doctor, nurse or social worker to talk to your family or to be present when you do.

Expect everyone you inform to react differently. Some people will be emotional, some numb or uncomfortable, and others will leap into action to try to assist. Most people will ask what they can do to help. Don't brush off these offers. If you know what they can do to help, don't be afraid to tell them. If you're not sure what you need, tell them you'll get back to them when you have a better handle on things. Either way, accept help when it's offered. (See "*A Caregiver's Perspective.*")

WORKING WITH YOUR CHILD'S SCHOOL

Many children with Hunter syndrome will be able to attend school, even if it's for a limited time. However, sending a child with a serious disease to school can be nerve-racking. You want him to be safe and well-cared for, 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.

Depending on your school district, you may or may not have a choice as to which school your child can attend. In some cases, accommodations regarding school choice can be made for children with special healthcare

needs. If you have a choice, it's a good idea to do some background research and then meet with the staff to find out which school offers the best resources for children dealing with medical or cognitive issues. You might ask how many other children with special healthcare needs they have in the school. Do they offer behavioral therapists or other specialized staff? How willing are they to learn about your child's disease?

Although some parents may choose a private school or homeschooling, the advantage of public schools is that they must comply with Section 504 of the Rehabilitation Act of 1973, which is a federal law that ensures that those with disabilities are not discriminated against. The law states that children with disabilities, including medical conditions, need to be given accommodations so that 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 oversees these plans and outline the considerations that are applicable to your child's particular needs. Keep in mind that negotiating the details of your child's plan may take time and involve a good deal of education on your part. One parent said it took over 4 months to work out the details of her child's IEP!

You will also need to meet with the school principal, counselors and teachers to explain your child's disease and the potential impact on the school, such as developmental issues, limited mobility, frequent absences, behavior problems, fatigue, etc. Because MPS II is rare, it's highly likely that your school's staff will not be familiar with the disease or its complications. Be prepared by bringing as much information with you to that initial meeting as possible.

In some cases, this will also involve providing medications that need to be administered during the day, along with other supplies that the school may require for proper care. In the beginning, many parents spend a few days or partial days at the school to monitor how things are going.

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

- Don't demand — request. It's important to get off on the right foot with school staff. You want them to feel positively toward you and your child by having an encouraging attitude. Developing long-term personal relationships with school staff is important for success.
- Make communicating easy. School staff should be able to call or email you 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. Try to stay levelheaded and work to together to find a solution. Preface your requests with "For my child's safety…" instead of making accusations or demands.
- Be patient, but firm. It's common to have a "rough start" when children with special healthcare needs begin school. It can take time for everyone involved to get comfortable with the 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 your child's disease progresses, his needs regarding school may change. At some point, attending school may become impossible.

Overall, developing good relationships with the staff members who care for your child is important not only for your child's well-being and safety, but also for your peace of mind.

A CAREGIVER'S PERSPECTIVE

The first thing many family members and close friends will ask after learning about your child's diagnosis is, "What can I do to help?" At first, you may be too overwhelmed to answer that question! But, as time goes by, it's necessary to let others help you. First, you can't do this alone — it really does take a village. Second, allowing the people in your life to help will make them feel better. After all, they love you and want to contribute! Some ways that family and friends can help, may include:

- Running simple errands, such as picking up dry cleaning, groceries, prescriptions or other necessities. (Many grocery stores offer online ordering, which allows you to select your groceries and have them picked up outside the store.)
- Dropping off and picking up other children from events when you have conflicting appointments.
- Watching your other children when you have medical appointments.
- Spending a few hours watching your child with Hunter syndrome so you can take a mental health break or go on a date with your spouse or significant other!
- Prepare a meal once a week on "clinic days," or stock your freezer with a few prepared meals for busy weeknights.
- Come to medical appointments with you to take notes and be an extra set of hands.
- Do a few basic household chores, such as laundry or vacuuming once a week.
- Stop by for a coffee and offer a sympathetic ear.

It can be helpful to keep a running list of things that need to be done, so when someone asks what they can do to help, you can offer a suggestion. Remember, this is a long journey, so take opportunities to lighten your load whenever possible!

Chapter 5:

Care for the Caregiver

Care for the Caregiver

"Life's challenges are not supposed to paralyze you; they're supposed to help you discover who you are."

– Bernice Johnson Reagan

Anyone who has traveled by airplane has heard the directive of putting your own oxygen mask on first, before attending to others. This makes sense because if you pass out from lack of oxygen, you're unable to help anyone, including yourself! And yet, attending to your own needs is very difficult when you're overwhelmed by caring for a child with a serious disease. The emotional and physical demands associated with caregiving can take a toll on even the most resilient person.

The first step in the process of coping well is to acknowledge your feelings and realize that it's normal to have a wide range of emotions, from sympathy and worry, to frustration and anger. You may feel helpless, inadequate, guilty or even grief-stricken at times throughout this journey. This emotional rollercoaster, along with the demands of caring for your child can be very stressful. In fact, studies show that most caregivers report high levels of stress.

Unfortunately, stress cannot be completely avoided in this role (or in life, for that matter). There are ways, however, to manage and alleviate stress and ensure that you, as the caregiver, remain mentally and physically healthy.

ACCORDING TO STATISTICS:

- Family caregivers who reside with those they provide care spend 40.5 hours per week caring for this person.
- Those caring for a child under age 18 spend an average of 29.7 hours per week performing caregiving tasks.
- 22% of higher-hour caregivers are more likely to say their health is fair or poor.
- The toll on caregiver health appears to increase over time.
- Chronic or long-term conditions among care recipients seem particularly likely to cause emotional stress for caregivers and negatively impact health.
- More than half of those caring for someone find the situation to be either highly or moderately stressful.

Source: National Alliance for Caregiving and AARP, "Chronic or long-term conditions among care recipients seem to be particularly likely to cause emotional stress for caregivers. Caregiving in the U.S." 2015)

THE IMPORTANCE OF CARING FOR YOURSELF

Why is stress a problem? Stress can not only negatively affect your thoughts, feelings and behaviors, if left unchecked it can contribute to many serious health problems, such as high blood pressure, heart disease, obesity and diabetes. Stress symptoms may be affecting your health without you even realizing it. Headaches, muscle pain, chest pain, fatigue, stomach upsets and insomnia are common symptoms of stress on your body. Stress may also impact your mood by causing anxiety, restlessness, lack of motivation or focus, irritability and anger. Finally, stress has been shown to cause negative behaviors, such as overeating/undereating, angry outbursts, drug or alcohol abuse, lack of physical activity and depression.

So, as you can see, it's not just something that can or should be ignored.

Caring for someone with a chronic disease is a marathon: To make it to the finish line with your health intact, you must pace yourself – keeping your mind and body in good shape. This can be challenging considering the demands on your time, but it's not impossible. Let's start with the basics.

FIND TIME FOR EXERCISE

Physical activity is a great way to reduce stress and stay healthy. The Mayo Clinic, as well as hundreds of studies on exercise, has shown that regular physical activity can not only control your weight, but also combat hypertension and high cholesterol, both critical factors in avoiding heart disease. It's also been shown to prevent or manage type 2 diabetes, stroke, several types of cancer, arthritis and depression. Perhaps most importantly for caregivers, it stimulates various brain chemicals that leave you feeling happier and more relaxed, therefore reducing stress hormones, such as cortisol.

Exercise can also boost your energy, which you need to keep up with your caregiving duties and enjoy daily activities. Finally, it can help you fall asleep faster and deepen your sleep, leaving you feeling more refreshed and energetic.

If you can schedule regular trips to the gym, a daily walk or run, or an exercise class, that's great. But, even if you can't find large chunks of time to exercise every day, you can still benefit by being more active on a daily basis – take the stairs instead of the elevator, play active games with your children, go for a bike ride, take an evening walk, etc. Consistency is important.

To reap the best health benefits, aim for at least 150 minutes of moderate-intensity exercise or 75 minutes of vigorous exercise each week. Try to combine aerobic exercises, such as running, walking or swimming, with strength training at least twice per week. Strength training or resistance exercises help maintain muscle mass and bone density.

Instead of viewing exercise as just one more chore to fit into an already busy schedule, find an activity that you enjoy, such as a dance class or hiking outdoors. Look at exercise as a way to unwind and get away from things for a while. Finding a friend to exercise with is a great way to stay motivated, help you stick to an exercise schedule and provide social engagement. When finding childcare is impossible, you can still get daily exercise by using a special jogging stroller or pushing your child's wheelchair. Some fresh air and a change of scenery can be beneficial for both of you.

Need more encouragement? The American Heart Association notes that for every hour of regular exercise you perform, you'll gain about two hours of additional life expectancy and that those added hours are likely to be healthier!

(Note: Before beginning any exercise program, be sure to consult your physician, especially if you've been sedentary.)

EAT FOR OPTIMAL HEALTH

Just as nutrition is important for your child and his battle against disease, it's critical in your regime to stay healthy. In addition to being tasty and comforting, food can serve as preventative medicine, helping you stave off numerous health problems. Think of food as fuel: without the proper fuel, your car will not run efficiently or at all. The same applies to your body. Without proper nutrition, your body cannot run at optimal levels and becomes prone to disease.

Unfortunately, the busier we become with caregiving duties, the more likely we are to take shortcuts when it comes to meals. While the occasional take-out meal or pizza delivery is fine, it's not a recipe for good health over the long-term.

When it comes to a healthy diet, there have been volumes written on the subject, with wide variance on advice. Every day there seems to be a new "fad diet" promising miracles. The best advice, however, is to follow a heart-healthy diet. Experts agree this is the best overall diet to lower blood pressure, reduce cholesterol, prevent diabetes, maintain a healthy weight, and provide a variety of essential nutrients. You can find guidelines at www.heart.org (American Heart Association), but in general, a heart-healthy diet includes:

- **Reducing sodium** to prevent hypertension (high blood pressure). Remove the salt shaker from your table and check labels on prepared food for sodium content – you'll be surprised by what you find! The recommended guideline for sodium intake for healthy adults is 2,300 milligrams or less per day (those with hypertension should lower their daily intake to 1,500 milligrams). On average, Americans consume double or triple that amount!
- **Reducing saturated fats**, which are found in red meat, poultry skin, full-fat dairy products, butter and cheese.
- **Avoiding trans-fats**, which are artificial fats used to extend the shelf life of many packaged and processed foods, such as margarine, crackers, and bakery goods. Many deep-fried and fast foods also contain trans-fats.
- **Adding more good fats** (monounsaturated and polyunsaturated), which are found in foods such as avocados and nuts and some cooking oils (olive, canola and sunflower).
- **Including omega-3 fatty acids in your diet** by eating fish, particularly deep-water fatty fish (salmon, herring, trout, sardines, mackerel and albacore tuna) at least two times per week.
- **Increasing fiber** with whole grain, high-fiber foods, such as whole-grain breads and cereal, brown rice, couscous, quinoa, lentils, beans, nuts, oat bran and oatmeal.
- **Avoiding refined flour products and foods with added sugar** (simple carbs), such as white bread, cakes, cookies, sugar-added cereals, and soda. Beware of ingredients such as corn syrup, high-fructose corn syrup, dextrose and corn sweetener, among others, which are really sugar.
- **Loading up on fresh fruits and vegetables** (five or more servings per day), which not only increase fiber, but provide essential nutrients. A diet high in fruit and vegetables has been linked to the prevention of certain cancers.

Changing your eating habits, especially as a busy caregiver, can be difficult and even overwhelming. Instead of trying to incorporate all these tips at once, try adopting one or two at a time. For instance, you might begin by serving fish once a week and cutting down on processed food. As this becomes a habit, you can work on adding more fruits and vegetables. Every step, even if it's small, is a move in the right direction.

Being organized (see Chapter 3) is the best way to make sure you and your family are eating healthy meals. Planning meals ahead of time, making extra portions that can be frozen and reheated on busy nights, finding quick, healthful recipes to rely on, and doing prep work in advance are some ways to make mealtimes easier. If you are having trouble preparing meals while managing severe complications with your child, don't be afraid to ask for help. Allow family and friends to take turns providing meals.

GET YOUR ZZ'S

While it's tempting to stay up late to get everything done, being overly tired and irritable will only make the situation worse. Not getting enough sleep can leave you more than just drowsy and grumpy, however – the long-term effects of sleep deprivation can be serious. Studies have linked lack of sleep to a number of health problems, from weight gain to a weakened immune system. Specifically, sleep deprivation can cause:

- Memory issues, both short, and long-term.
- Mood changes, such as irritability, anger, sadness, inability to cope and depression.
- Decreased coordination and focus, which increases risks for accidents.
- Poor immunity – While you sleep, your immune system produces protective, infection-fighting substances like cytokines, which are used to combat bacteria and viruses. It may take you longer to recover from illnesses, as well.
- Impulsive behavior, such as overeating, and alcohol and drug abuse.
- Obesity, due to the disruption of two hormones, leptin and ghrelin, which control feelings of hunger and fullness.
- Type 2 Diabetes – Sleep deprivation causes your body to release higher levels of insulin after you eat, which controls your blood sugar level and increases your risk for type 2 diabetes.
- Heart disease – Sleep affects processes that keep your heart and blood vessels healthy, including your blood sugar, blood pressure, and inflammation levels. It also plays a vital role in your body's ability to heal and repair the blood vessels and heart.

The bottom line is your body needs sleep as much as it needs good food to function at its best. During sleep, your body heals itself and restores chemical balances in your brain, as well as the rest of your body.

While it's normal to have an occasional night of tossing and turning, chronic insomnia or sleep disruptions should be discussed with your physician. Develop good sleep habits (visit The National Sleep Foundation at https://sleepfoundation.org/sleep-tools-tips/healthy-sleep-tips) and make it a priority to get a good night's sleep (7-9 hours per night).

Of course, this is easier said than done. Hunter syndrome often impacts a child's sleep patterns, which, in turn, disrupts your sleep. As we discussed in Chapter 1, good sleep habits and a safe environment can help both of you sleep better. You may also consider the use of a special bed that helps contain your child or sensor pads placed under the carpet outside his room. If the situation is not improving, you may want to discuss the use of medication or supplements with your physician.

MAINTAIN HOBBIES, ACTIVITIES AND SOCIAL CONNECTIONS

Unfortunately, enjoyable undertakings are often the first to take a back seat to your caregiving duties, because they are mistakenly considered unnecessary – but they are essential to your mental well-being and overall health. While you may not have hours to devote to gardening or golf, finding just 30 minutes (or whatever time you can sneak in) to prune your flowers or hit some balls at the driving range can be beneficial. Time spent painting, reading, scrapbooking, or whatever you find enjoyable and relaxing is not frivolous or selfish, but necessary to provide a much-needed mental boost.

Likewise, staying connected to family and friends is important for reducing stress, warding off depression, and boosting happiness. Did you know that a positive social network is strongly linked with better health and longevity? Don't wait until "you have time" to make plans, because that may never happen! Set dates to have dinner with your spouse or grab a coffee with a friend. Just seeing the notation on the calendar can give you a lift. Enlist family and friends to stay with your child or children. If necessary, hire an aide or take advantage of respite care (see Home Health Aides and Respite Care).

FIND YOUR SPIRITUAL SIDE

It doesn't matter what faith or spiritual practice you engage in, as long as you connect regularly. Studies show that praying and meditating are highly-effective stress relievers. In fact, just 20 minutes a day of prayer or meditation can lower blood pressure and reduce levels of cortisol (the stress hormone). If you've tried meditation and found it difficult to shut out distractions and focus, don't despair. Try some guided imagery or meditations – and keep practicing. It may take some time to get the hang of it, but the benefits are worth the effort.

CONSIDER A THERAPIST

Talking to a therapist has proven very helpful for many caregivers. Your physician can recommend a therapist or help you locate one. You may also ask other parents for referrals to someone they've had success with. It's important to find a therapist with whom you feel comfortable, so be sure to schedule an initial consultation. Keep in mind, you may need to see several before you find the right fit. You may also consider getting other family members involved in therapy sessions. After all, this type of diagnosis affects the entire family and its dynamics.

The benefit of talking to a therapist is that he or she will listen without judgment, allowing you to express any or all feelings you encounter without fear. He or she can also offer invaluable coping mechanisms to help you reduce stress and deal with difficult situations.

LEARN TO ACCEPT AND ASK FOR HELP

Many people have a hard time asking for and accepting help. In fact, it often takes a complete meltdown or health crisis before a person will admit to needing assistance. Caregivers, in particular, have a hard time requesting help, which has prompted a number of studies to understand the reasons behind the reluctance. Do any of these sound familiar?

- **Obligation** – We may feel it's our sole responsibility to care for our loved one. It's not uncommon to think, "No one can do it better than we can."
- **Guilt** – We may feel that we are somehow responsible for our loved one's disease, or we may feel guilty about asking someone to help with "our responsibility."
- **Worry** – If we allow others to help care for our child, there is always the possibility that he will not be cared for properly.

- **Reluctance to Bother Others** – Even though family and friends have offered to help, it can feel like an imposition to ask for assistance.
- **Absence of Family Help** – On the other end of the spectrum, you may be in a situation in which you don't have family or family members who are able or willing to assist. In this case, relying on hired help may feel uncomfortable.
- **Lack of Financial Resources** – Unfortunately, hiring aides to help care for someone or hiring help for other household duties can be cost prohibitive.

If you're finding yourself reluctant to seek help, you may want to talk to a therapist (see above) about the underlying reasons or fears. Also, talking to members of a support group can help you take positive steps and find resources. Some research into community centers and local family resources may also provide low-cost alternatives or financial aid for care. Finally, having open and honest discussions with your spouse, family members and friends can open the door to getting the help you need.

Some advice from experienced caregivers includes:

- **Let go of the guilt, and accept the reality of the situation.** For the most part, guilt is a useless emotion that keeps us stuck in an unproductive rut. No one is perfect, and to expect perfection of yourself or others will only cause constant disappointment. Do the best you can under the present circumstances.
- **Accept your limitations, and don't compare yourself to others.** You may know others who makes caring for a child with a serious disease look easy. From the outside, they may look like they have everything under control. However, there is no way to know what struggles they've encountered. Everyone has a different threshold for stress, and every situation is unique. Comparisons are useless. Simply do the best you can.
- **Don't be afraid to hire caregivers.** There are many reliable, caring aides available. You may have to try a few before you find one who meshes with you and your child, but don't give up. A good hired aide can not only take some of the burden off you, he or she may become a friend to you and your child. (See Home Health Aides and Respite Care.)
- **Accept help from others.** Chances are, if friends or family have offered help, they mean it. Sure, everyone is busy with their own lives, but dividing small tasks among several people is not a burden. Often the person who has volunteered his or her services is genuinely looking for a way to contribute and will feel better if given the opportunity.
- **Be realistic about your child's condition.** You may be able to handle all the caregiving tasks for a time, but as your child's disease progresses, the responsibilities can become too much to manage without help. Recognizing and accepting these changes can be difficult, but it's important to remain flexible.

HOME HEALTH AIDES AND RESPITE CARE

Once you've decided that you need some help, how do you find it? There are two options to finding a home health aide – you can hire someone through an agency or on your own. Agencies have licensing and standards, which means employees are screened for education and employment experience, and undergo background checks. Depending on the agency, they are often required to take random drug tests, and better agencies will provide some training and supervision, such as unannounced visits.

In addition, if you hire through an agency, you're covered if an aide can't make it due to illness or other issues. The agency will send a replacement. Also, if an aide doesn't work out, you can request another. However, these services come with a price, as agencies are generally more expensive than a private hire.

Private hires tend to be less costly and more flexible regarding hours or duties. If you do hire privately, you take on all the responsibility, including drawing up contracts, conducting background checks, and following up on references. If a private hire doesn't show up, there is no replacement, so a family member must be available. When hiring on your own, be sure the aide is registered self-employed, is fully insured, and can provide you with references. Sometimes the best way to find a private hire is through a recommendation from family or friends, your local community center or church.

Either way, having a home health aide provide help a few days a week can offer a caregiver some much-needed time to run errands, get some exercise, engage in social activity or simply relax. The amount and type of care will depend on the severity of your child's symptoms and complications. Keep in mind, that while home health aides can help with ADLs (activities of daily living), such as bathing, feeding, using the toilet and doing basic housekeeping duties, they can't administer medication or perform medical treatments.

If you're considering hiring an aide, it's important to conduct a thorough interview and spend time with him or her in the presence of your child. You

may suggest a trial period and letting him or her interact with your child while you're present. As you talk to the aide and observe the interactions, ask yourself the following questions:

- Do I get a good feeling about him or her? Is he or she warm and supportive?
- Does the aide treat my child with respect?
- Is he or she patient and tactful when interacting with my child?
- Does he or she anticipate needs or wait to be asked?
- Does he or she listen carefully and respect my care instructions, or interject with his or her own opinions, or ignore my instructions?
- Is he or she reliable (i.e., shows up on time, takes notes, maintains the agreed, upon schedule)?
- Are the necessary tasks being completed? (Make a specific list of duties.)
- How well does the aide get along with my child? Do they seem compatible?
- Does my child feel safe and comfortable with this person?

QUESTIONS TO ASK A HOME HEALTH AGENCY

- Are you licensed and insured?
- Do you perform background checks?
- Are employees trained and supervised?
- What is the policy for replacing aides?
- What services are the aides allowed to perform, and what are they prohibited from doing?
- What are the costs per hour, and how will payment be made?
- How do I handle complaints or concerns?
- Are any of your employees experienced with the care of children?
- Have any of your employees worked with a child who has a serious disease such as Hunter syndrome?

PROTECTING YOUR CHILD

Whenever you bring someone into your home, it's wise to be cautious, even if this person comes with glowing references. You can take some simple precautions to protect your child and your valuables:

- To prevent theft, keep cash, jewelry, personal identification and extra medication out of sight or locked up.
- Occasionally return home early to check on activities.
- Use remote monitoring technology to keep an eye on things when you're not there.
- Talk to your child privately about the activities they did with the aide.
- If your child is unable to communicate clearly, look for changes in mood or behavior, or signs of physical abuse, such as bruises or red marks.

RESPITE CARE

Another option for assistance is respite care, which provides parents and other caregivers with short-term care services. Respite care can be planned or offered during emergencies. Again, when your child has special needs, it can be challenging to find proper care – it's not the same as hiring a babysitter! Luckily, there are affordable, trustworthy resources you can use to take a well-deserved break. And, your child will benefit from interacting with other people. They may form new friendships, experience new environments, and have fun.

Respite care can come in several forms: a caregiver who comes to your home for a few hours daily, weekly or as needed; drop-off day programs (at a school, healthcare facility or volunteer agency) that provides appropriate activities; or daily or weekly respite programs offered by community-based agencies or camps.

Another option to consider is a parent "co-op," in which families of kids with special needs take turns watching each other's children. Support groups for parents of children with Hunter syndrome are a good place to meet other families. The benefit of this type of arrangement is that families are experienced with the type of symptoms and complications your child may be having.

To find local care, you can take advantage of referral services, such as:

- The Lifespan Respite Care Program, a federally funded grant program which assists states with improving access to respite care and may provide financial assistance. This program is offered in many states. Contact the ARCH National Respite Network and Resource Center (www.arch.respite.org) to see if there's a program in your state. If not, there might be a State Respite Coalition that can help.
- For private respite programs in your area, go to the ARCH National Respite Network. Tell them what kind of care you need and get a list of providers that may include visiting nurses, childcare centers, and sleepaway camps.

Of course, the same rules apply with respite care as hired home health aides. Be sure to conduct interviews and check references. Make a list of your child's needs, from communicating to toileting, and be sure the caregiver has the skills/experience to handle each task. Watch how they interact with your child.

Like home health aides, respite care can be costly. However, there are some programs that can help. If your state has a Lifespan program, call and ask for information on financial aid. Most children with special needs qualify for home, and community-based Medicaid waivers that can cover the cost of respite care. Keep in mind, many of these programs have waiting lists for waivers, so be sure to apply early.

Whether you choose a hired home health aide or respite care, don't underestimate the importance of taking a break. No one is superhuman — capable of doing everything all the time. The ability to run errands, go to an appointment, have dinner with your spouse, or just take a break to do something nice for yourself can go a long way toward preserving your mental health. Remember, when you're happy and healthy, you'll be better equipped to take care of your child and other family members.

SIGNS OF CAREGIVER BURNOUT

All caregivers experience some degree of exhaustion and feelings of "I've had enough." However, if these feelings persist, they may be signs of caregiver burnout, which is defined as "a state of physical, emotional and mental exhaustion that may be accompanied by a change in attitude, from positive and caring, to negative and unconcerned." The symptoms are similar to those of depression, and may include:

- Withdrawal from family and friends
- Loss of interest in activities that used to be enjoyable
- Feelings of inadequacy or helplessness
- Being easily irritated or angered
- Difficulty concentrating and forgetfulness
- Difficulty sleeping or sleeping too much
- A change in appetite (either loss of appetite or overeating)
- Frequent illnesses or worsening of health conditions
- Thoughts of hurting oneself or the person for whom you are caring

If you are experiencing any of the symptoms, it's a good idea to talk to your physician before the situation escalates. In some cases, a physician may prescribe medication or recommend therapy. It's also time to get some help with your caregiving duties!

SUPPORT GROUPS

It's normal for families and children diagnosed with a serious disease to feel different from others and isolated, which is why support groups are so valuable. Being around others who understand what you're going through can be very comforting and offer many benefits. For instance, sharing similar experiences and problems can not only make you feel better; you might discover solutions that you hadn't thought of before. Members of support groups can also recommend helpful resources and provide meaningful interactions for your child.

While there is no substitute for face-to-face interaction, you may also consider participating in an online group, which allows you to chat and share concerns whenever it's convenient.

A good place to start is with The MPS Society (https://mpssociety.org), which exists to support families, as well as advocate for MPS and support research for a cure. They provide a wide range of programs from continuing education scholarships to medical travel expense assistance. The site also lists events and social networks where families can find emotional support and make connections.

FOCUS ON THE POSITIVE

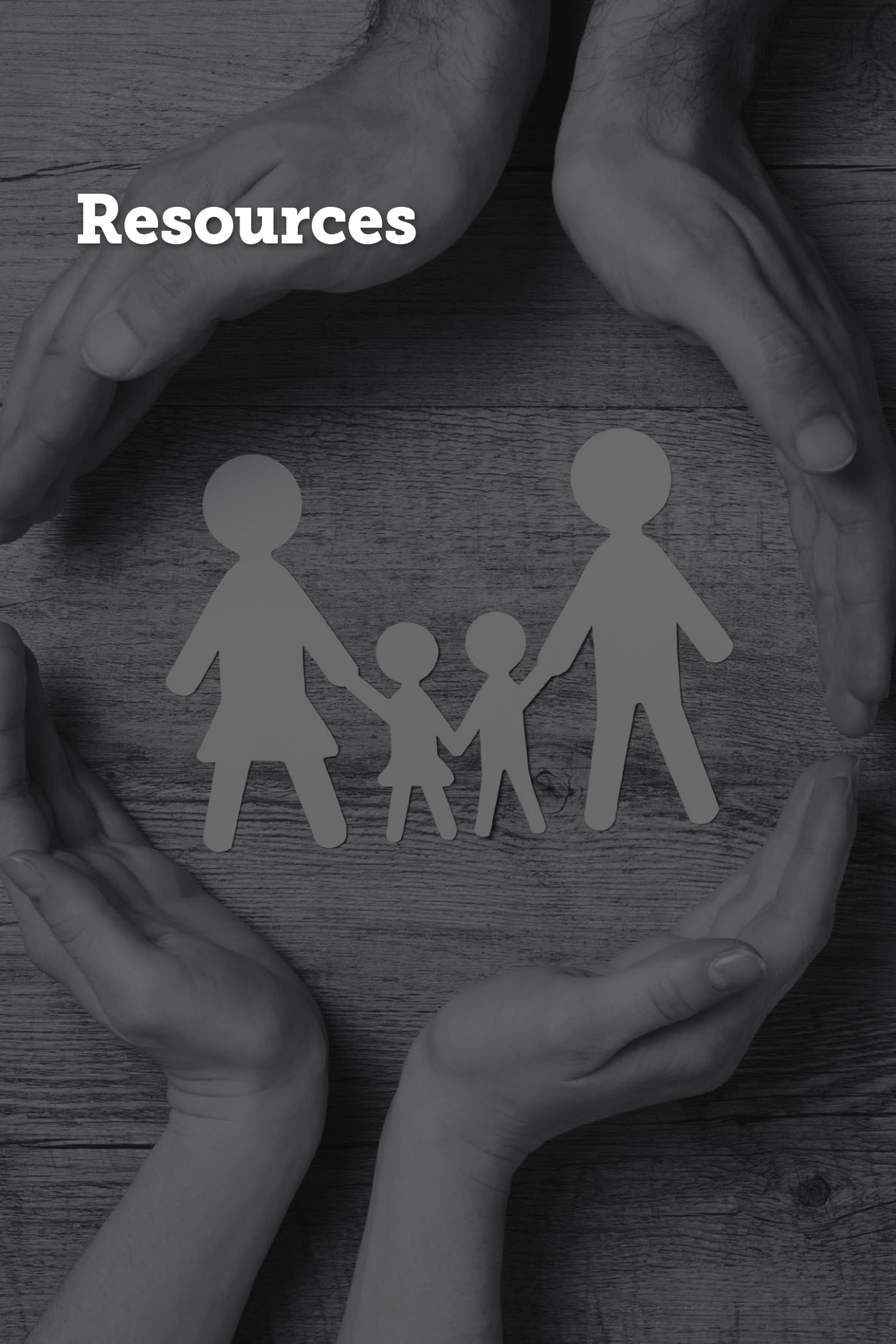
Parenting a child with Hunter syndrome is a challenge. The journey through your child's medical care will be difficult for you and your family, with many ups and downs, and twists and turns along the way. While life changes dramatically when your child is diagnosed with a serious disease, it doesn't have to be one of despair. It can often be hard to see the positive in this situation, but "finding the light" whenever possible will help you and your family cope.

While every caregiver has his or her own personal experience, some of the life lessons or rewards that may result include:

- Forming a closer bond or special relationship with your child and creating special memories.
- Strengthening bonds with your family. In fact, the families that are the most successful at coping with this type of diagnosis are those that are able to work together as a team.
- Teaching siblings important lessons about empathy, responsibility and kindness.
- Enriching your own life by helping someone you love.
- Becoming more organized and efficient.
- Appreciating the everyday things in life and the unexpected joys that we sometimes take for granted.
- Deepening of one's faith.
- Learning not to worry about the little things.
- Discovering who your true friends and support network are — often leading to close, life-long friendships.
- Recognizing how strong you really are.

It's important to remember that even in the most difficult of circumstances, it's possible to take away something meaningful.

Resources



Resources

BOOKS FOR FURTHER READING

Hunter Syndrome - A Medical Dictionary, Bibliography, and Annotated Research Guide to Internet References, Icon Health Publications, 2004.

Hunter Syndrome Medical Guide, Qontro Medical Guides, 2008.

Hunter Syndrome, Jesse Russell and Ronald Cohn, Books on Demand, 2012.

The Complete Caregiver's Organizer: Your Guide to Caring for Yourself While Caring for Others, Robin Porter, Spry Publishing, 2015.

When Your Child is Sick: A Guide to Navigating the Practical and Emotional Challenges of Caring for a Child Who is Very Ill, Joanna Breyer, PhD, 2018.

With Angel's Wings, Stephanie A. Collins, 2018.

FOUNDATIONS/ORGANIZATIONS

Angel's Hands Foundation The mission of this foundation is to improve the quality of life for individuals living with rare diseases. It supports families by assisting with lingering medical expenses, funding medical equipment not covered by insurance, funding family visits to educational events or to see medical specialists, sponsoring fundraising events, and hosting social outings for families. Visit www.angelshands.org.

Family Voices is a national grassroots network of families, friends, and advocates for healthcare services that are family-centered, community-based, comprehensive, coordinated, and culturally competent for all children with special healthcare needs. Family Voices promotes the inclusion of all families as decision makers at all levels of healthcare, and supports essential partnerships between families and professionals. Visit www.familyvoices.org.

The Genetic Alliance is a nonprofit health advocacy organization committed to transforming health through genetics and promoting an environment of openness centered on the health of individuals, families, and communities. Genetic Alliance's network includes more than 1,200 disease-specific advocacy organizations, as well as thousands of universities, private companies, government agencies, and public policy organizations. Visit www.geneticalliance.org.

GenesInLife.org is a place to learn about all the ways genetics is a part of your life. On this site, you can learn how genetics affect you and your family, why you should talk to your healthcare providers about genetics, how to get involved in genetics research, and much more.

The Global Genes Project is a leading rare and genetic disease patient advocacy organization with over 500 global organizations. The non-profit organization is led by Team R.A.R.E. and promotes the needs of the rare and genetic disease community. The Global Genes Project's mission is centered on increasing rare disease awareness, public and physicians, building community through social media, and supporting research initiatives to find treatments and cures for rare and genetic diseases. Visit www.globalgenes.org.

The Individuals with Disabilities Education Act (IDEA) is a law ensuring services to children with disabilities throughout the nation. IDEA governs how states and public agencies provide early intervention, special education, and related services to more than 6.5 million eligible infants, toddlers, children, and youth with disabilities. Learn more at www.idea.ed.gov

National Discrimination Center for Children with Disabilities is a centralized resource for families, educators, administrators, journalists, and students. The organization's special focus is children and youth (birth to age 22). Learn more at www.nidcd/gov.

WEBSITES/GENERAL INFORMATION

Family Caregiver Alliance – Information and online support groups – www.caregivers.org.

National Alliance for Caregiving – Advocacy, research and resources – www.caregiving.org.

National MPS Society – www.mpssociety.org
- National MPS Society on Twitter - @MPSSociety
- National MPS Society on Facebook – www.facebook.com/NationalMPSSociety (Sign up for webinars, family conferences, connect with families and more.)

National Organization for Rare Disorders (NORD) – https://rarediseases.org

What is Hunter Syndrome? – HunterPatients.com – www.hunterpatients.com

Hunter syndrome – Symptoms and causes – Mayo Clinic – www.mayoclinic.org/diseases-conditions/hunter-syndrome

BLOGS/ARTICLES

"Rare Disease Day: What MPS type 2 Hunter Syndrome means to us," Ger Symth – http://community.fireflyfriends.com/blog/article/rare-disease-say-what-mps-type-2-hunter-syndrome-means-to-uscommunity.fireflyfriends.com/type-2-hunter-syndrome

"Growing up with Hunter Syndrome," by Christopher Dutcher - https://globalgenes.org/raredaily/christopher-dutcher-on-hold

DIET AND EXERCISE

"The American Heart Association's Diet and Lifestyle Recommendations" – http://www.heart.org/HEARTORG/HealthyLiving/HealthyEating/Nutrition/The-American-Heart-Associations-Diet-and-Lifestyle-Recommendations_UCM_305855_Article.jsp#.Wo7ldlPwapo

"Quick & Easy Healthy Meal Recipes" – https://www.foodnetwork.com/healthy/packages/healthy-every-week/quick-and-simple

HEALTH AIDS AND TOOLS

www.assistedlivingtechnologies.com

www.caregiverproducts.com

HOME HEALTH CARE AGENCIES AND RESPITE CARE

Home Health Care Agencies – www.homehealthcareagencies.com

The Lifespan Respite Care Program – contact ARCH National Respite Network and Resource Center at archrespite.org

Respite Care Programs – Child Welfare Information Gateway https://www.childwelfare.gov/topics/preventing/prevention-programs/respite

"The Special Needs Respite Care Guide: Your Care Options" – https://www.care.com/c/stories/10269/the-special-needs-respite-care-guide-your-care-options

PHYSICIAN RESEARCH AND LOCATOR

American Board of Medical Specialties – www.abms.org

American Medical Association Doctor Finder – www.apps.ama-assn.org/doctorfinder

Administrators in Medicine – www.docboard.org

www.healthgrades.com

www.rateMDs.com

Visiting Nurse Association of America – www.vnaa.org

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Better with Age, by Robin Porter, Spry Publishing, 2015

"Caring for a Seriously Ill Child," Kids Health from Nemours – http://kidshealth.org/en/parents/seriously-ill.html#

"Caregiver Statistics: Demographics," Family Caregiver Alliance/National Center on Caregiving – https://www.caregiver.org/caregiver-statistics-demographics

"Chronic or long-term conditions among care recipients seem to be particularly likely to cause emotional stress for caregivers." Caregiving in the U.S., 2015

"How Parents Can Help Children Cope Chronic Illness," DBA Foundation – http://dbafoundation.org/wp-content/uploads/2012/11/4ChronicIllnessInsert.pdf

"Hunter Syndrome – Symptoms and Causes" – Mayo Clinic – https://www.mayoclinic.org/diseases-conditions/hunter-syndrome

"Hunter Syndrome (Mucopolysaccharidosis II) – The Signs and Symptoms a Neurologist Needs to Know," Touch Neurology – http://www.touchneurology.com/articles/hunter-syndrome-mucopolysaccharidosis-ii-signs-and-symptoms-neurologist-needs-know

"Life-Threatening Illness: What to Tell Family, Friends," WebMD – https://www.webmd.com/palliative-care/life_threatening_illness_what_to_tell_family_friends#3

Mucopolysaccharide & Related Diseases Society, Ltd. – http://www.mpssociety.org.au/hunter-syndrome2/living-with-hurler-syndrome-brain-involvement

"Respite Care for Children with Special Needs," Kids Health – https://kidshealth.org/en/parents/respite-care.html#

"Stress Symptoms: Effects on your body and behavior," Mayo Clinic – https://www.mayoclinic.org/healthy-lifestyle/stress-management/in-depth/stress-symptoms/art-20050987

The Complete Caregiver's Organizer: Caring For Yourself While Caring for Others, by Robin Porter, Spry Publishing, 2015.

"The Effects of Sleep Deprivation on Your Body," Healthline.com – https://www.healthline.com/health/sleep-deprivation/effects-on-body#1

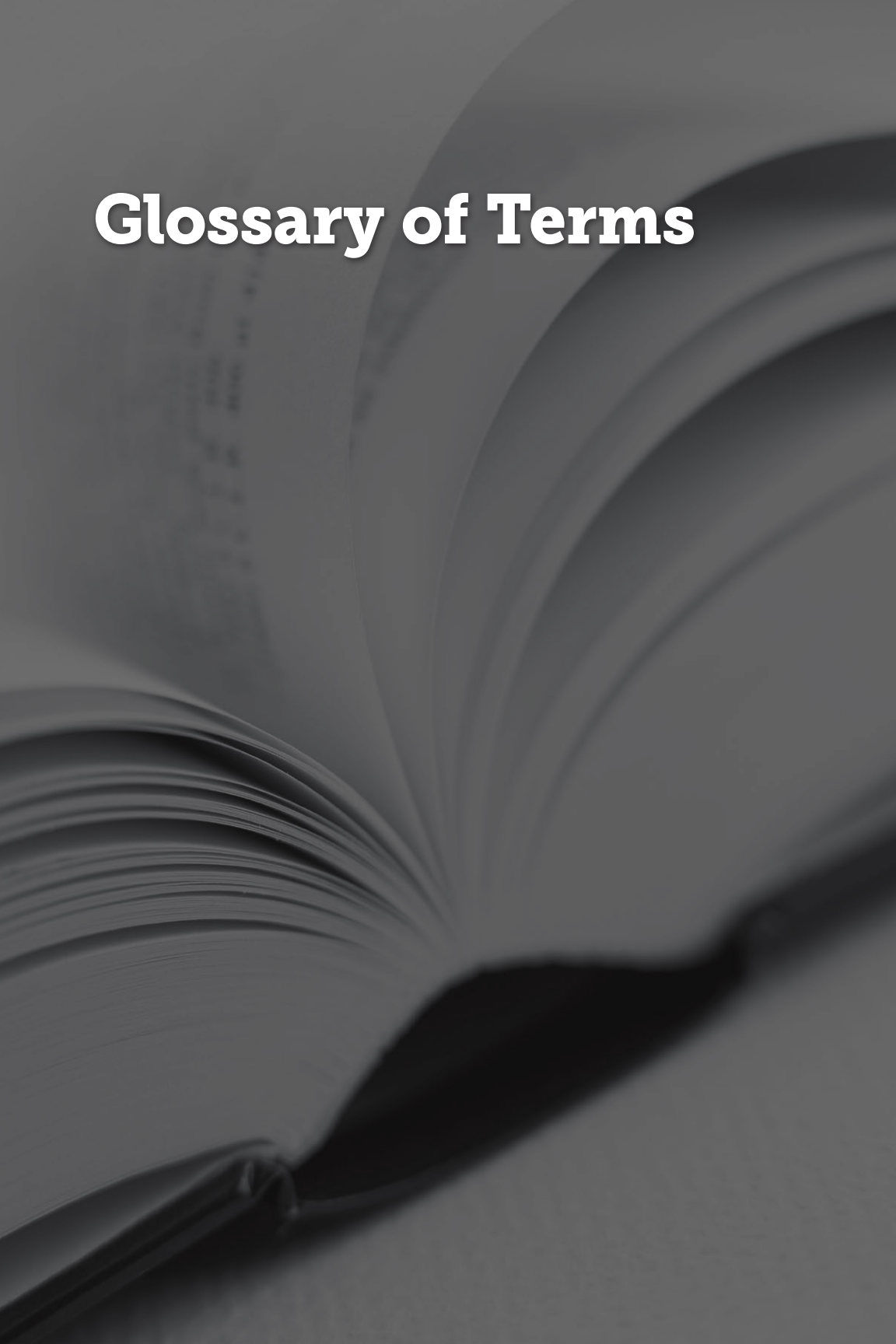
U.S. National Library of Medicine/National Institute of Health – https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3306562

"What is Hunter Syndrome?" – HunterPatients.com – www.hunterpatients.com

"9 Clever Ways to Make the Medicine Go Down," by Katie McDonald, *American Baby*.

"10 Ways to Cope with a Child's Chronic Illness," Parenting.com – http://www.parenting.com/child/health/10-ways-to-cope-childs-chronic-illness

Glossary of Terms



Glossary of Terms

Attenuated – weakened; diminished in force or intensity. Attenuated MPS II is a less severe form of the disease. (Also referred to as non-neuronopathic.) While those with attenuated Hunter syndrome do not experience cognitive impairment, they may still have serious physical symptoms.

Chondroitin sulfate – is a glycosaminoglycan or chain of alternating sugars. It is usually attached to proteins as part of a proteoglycan (a type of protein that is heavily glycosylated). Chondroitin sulfate is an important structural component of cartilage, and is also found in the cornea, bone, skin and arteries.

Copathogenicity –when antibiotics fail to clear an infection, such as strep throat, this is referred to copathogenicity; the body does not respond effectively to medications due to repeated infections.

Dermatan sulfate – is a glycosaminoglycan (or chain of sugar molecules) found mostly in skin, but also in blood vessels, heart valves, tendons and lungs.

Extracellular matrix – is a meshwork of proteins and carbohydrates that binds cells together or divides one tissue from another. The extracellular matrix is the primary product of connective tissue.

Geneticist – someone who studies and works to apply his or her knowledge of genetics, a brand of biological science that involves genes, heredity and natural variation in living organisms. They focus primarily on the passage of traits from parents to offspring, as well as how genes become mutated or involved in disease and aging.

Glycosaminoglycans (gli-ko-sah-mee-no-gli-cans) or GAGs – are long chains of sugar molecules that are found throughout the body, often in mucus and in fluid around the joints. Glycosaminoglycans are highly polarized and attract water, and are therefore useful to the body as a lubricant or as a shock absorber. (The former term for glycosaminoglycans is mucopolysaccharides.)

Heparan sulfate – are proteoglycans (a type of protein) found at the cell surface in the extracellular matrix or connective tissue. They are primarily found in the lungs, arteries and cell surfaces.

Heparin – is a naturally occurring anticoagulant or blood thinner which prevents the formation of clots within the blood.

Hydrocephalus – is a buildup of fluid on the brain. The excess fluid puts pressure on the brain, which can damage it.

Iduronate-2-sulfatase (I2S) – a sulfatase enzyme associated with Hunter syndrome. I2S is required to break down chains of sugar molecules (i.e., the lysosomal degradation of heparan sulfate and dermatan sulfate). Sulfatase enzymes are found in tissues throughout the body.

Inguinal hernia – occurs when tissue, such as part of the intestine, protrudes through a weak spot in the abdominal muscles.

Keratan sulfate – any of several glycosaminoglycans or chains of sugar molecules that have been found, especially in the cornea, cartilage and bones. It's also necessary in the formation of scar tissue following an injury. These large, highly-hydrated molecules can act as a cushion to absorb shock.

Lysosomal storage diseases – a group of about 50 rare inherited metabolic disorders that result from defects in lysosomal function. Lysosomes are sacs of enzymes within cells that digest molecules and pass the fragments on to other parts of the cell for recycling. This process requires critical enzymes. If one of these enzymes is absent or defective, the large molecules accumulate within the cell, eventually killing it. Hunter syndrome is a lysosomal disease.

Mucopolysaccharides (mew-ko-pol-ee-sak-ah-rides) – are long chains of sugar molecules that are found throughout the body, often in mucus and in fluid around the joints. (They are more commonly called glycosaminoglycans.)

Mucopolysaccharidosis type II (MPS) – also known as Hunter syndrome or Hunter disease, is a rare genetic disorder that primarily affects males and is caused by a missing or malfunctioning enzyme in the body (iduronate-2-sulfatase or I2S).

Myringotomy – a surgical procedure which involves making a small incision in the eardrum to relieve pressure caused by the buildup of fluid or to drain pus from the middle ear.

Neuronopathic and non-neuronopathic – refers to the two types of MPS II. Neuronopathic MPS II affects the nervous system and is more severe. This form results in cognitive impairment and developmental regression in childhood. Non-neuronopathic MPS II, also referred to as attenuated MPS II, is less severe. While individuals with the non-neuronopathic form do not experience cognitive impairment, they may still have serious physical symptoms and complications.

Proteoglycan (pro-te-o-gli-can) – a complex molecule or group of glycoproteins found primarily in connective tissue that play an important role in cell interaction. Proteoglycans help form the matrix of connective tissue.

Tympanotomy tube – also known as a grommet or myringotomy tube, is a small tube inserted into the eardrum in order to keep the middle ear aerated and prevent accumulation of fluid in the middle ear.

X chromosome – is one of the two sex-determining chromosomes in humans (the other is the Y chromosome) and is found in both males and females. Females have two X chromosomes.

X-linked recessive – X-linked recessive genetic conditions are caused by mutations in genes on the X chromosome. Males are more frequently affected than females.

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About the Author

Robin Porter has been writing content for books, websites and a variety of other materials for over 30 years. Working with physicians, parents and other professionals, she has co-authored several books on health-related topics, including women's heart health, migraines, juvenile arthritis and Type 1 diabetes. Robin has also penned two original titles: *Better With Age—Your Blueprint for Staying Smart, Strong, and Happy for Life and The Complete Caregiver's Organizer – Your Guide to Caring for Yourself While Caring for Others*. Though she has developed an expertise in medical topics, she enjoys researching and writing about many subjects.

Robin lives in Canton, Michigan, with her husband of 31 years and their college-aged son, along with their dog and cat.

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A Caregiver's Guide —to— Hunter Syndrome

A diagnosis of Hunter syndrome brings you and your family into the often confusing world of rare diseases. It's natural for parents/caregivers to be fearful and filled with questions about what to expect, how to manage symptoms and complications, where to find specialists to care for your child, and, perhaps, most importantly, how to live the best life possible with this new reality. In *A Caregiver's Guide to Hunter Syndrome*, you'll find answers to many of your questions about the disease, as well as tips for staying organized, dealing with doctors and hospitals, communicating effectively, and caring for yourself. When facing a serious, life-changing diagnosis, knowledge truly is power.

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