

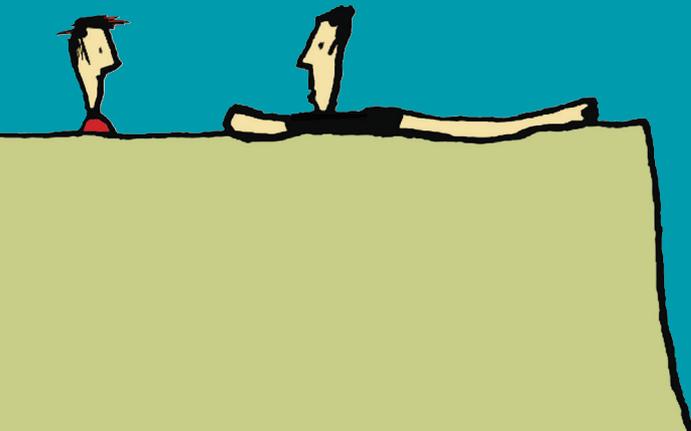
HOW DO I TALK TO MY FAMILY ABOUT
Pompe?



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This booklet will help you talk to your family about Pompe. Every person with Pompe is different, and every family is different. So this booklet includes several different approaches to speaking with your family. Choose an approach that you think will work best for you.



Introduction



What is family?

There is no one-size-fits-all family. Your family is the group of people who are important to you and your well-being. They might be related to you biologically, legally (through adoption or marriage), or emotionally (through affection, dependence, obligation, or cooperation).

Why should I talk to my family about Pompe?

Pompe is a rare condition (relative to other chronic health conditions). Most people do not know what it is and may never have heard of it. You may find that even some doctors and other healthcare professionals do not know much about it.

It is important to help both your family and your healthcare providers understand what Pompe is, what it means for you, and how your diagnosis might impact your family. Some topics to share are major signs and symptoms and how the condition is passed down in families. [Turn to page 2 for more information on Pompe.]

Sharing your story will help your relatives understand their family health history and possibly even their own personal history. Knowledge of your symptoms and the reason for them—Pompe disease—may help relatives make sense of their own physical experience, especially if they have had symptoms medical professionals have been unable to explain. In turn, it is helpful for you to know your whole family health history, including Pompe and other conditions that might run in your family. Putting the whole picture together will help you and your relatives:

- Identify risks due to shared genes.
- Talk to each other about health (including quality of life).
- Summarize health information to give to healthcare providers, as well as colleagues, teachers, and others.

Pompe Disease

What is Pompe?

Pompe disease is an inherited, neuromuscular condition that causes muscle weakness in people of all ages. Symptoms may first occur at any time from infancy to adulthood. Pompe is also known as acid maltase deficiency or glycogen storage disease type 2.

Our bodies contain thousands of active substances called enzymes. For example, your stomach has enzymes that help break down the food you eat. Similarly, the enzyme acid alpha-glucosidase (pronounced AL-fa glue-CO-sigh-days), also called GAA, breaks down glycogen, a form of sugar that is stored in a specialized compartment of muscle cells throughout the body. People with Pompe do not have enough GAA enzyme, so glycogen builds up in muscle cells. This leads to progressive loss of muscle function. Pompe mostly affects the skeletal (including heart) and respiratory muscles.

There are two types of Pompe: infantile-onset and late-onset. Infantile-onset is the result of complete or near complete deficiency of GAA. Late-onset is caused by partial deficiency of GAA.

How is Pompe passed on?

Pompe is an autosomal recessive condition. This means two non-working copies of the gene are needed for an individual to be affected by the condition. Individuals with one working copy and one non-working copy of the gene are called carriers.

- When both parents are carriers, there is a 1 in 4 (25%) chance that any child born will have Pompe.
- If one parent has the disease and the other parent is a carrier, there is a 1 in 2 (50%) chance that any child born will have Pompe.
- When both parents have Pompe, all of their children will inherit the disease.



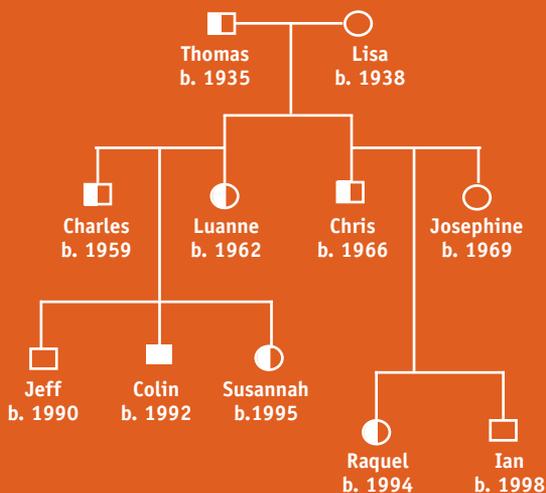
Colin's story



My name is Colin, and I have Pompe. No one else in my family has Pompe.

My grandfather, Thomas, was a carrier. He passed the gene to my mom and uncle Chris; they are both carriers, too. My uncle married Josephine, who does not have the gene change for Pompe. They have two children – my cousin Raquel is a carrier, and Ian is not.

My mom married another carrier—my dad. This means that each of their children—me, my brother and sister—had a 1 in 4 (25%) chance of having Pompe, a 1 in 2 (50%) chance of being a carrier, and a 1 in 4 (25%) chance of not having the Pompe gene at all. Jeff was born first—he doesn't have the Pompe gene change! I'm the middle child—I have Pompe. Susannah is a carrier.



○ = female

□ = male

Shaded = Pompe

Half Shaded = carrier

Pompe Disease

Infantile-onset

Infants affected by Pompe show symptoms soon after birth, and the disease progresses rapidly. Most babies die from cardiac or respiratory (breathing or lung-related) complications before their first birthday.

Major signs and symptoms

- Muscle weakness or a “floppy” appearance
- Poor muscle tone
- Enlarged heart (usually detected by X-ray)
- Breathing problems and lung infections
- Feeding problems and failure to grow at the expected rate (failure to thrive)



Kerri's story

My daughter Sarah was a short-lived, sweet, beautiful child. Four months after she was born in 1996 we found out she had Pompe. Five months after that she passed away. It's a genetic disease, and she had my genes, so I blamed myself. More than a decade later, I was having health problems. I was misdiagnosed with chronic fatigue syndrome, arthritis, depression, and more.



Late-onset

Though late-onset Pompe may appear more gradually than the infantile-onset form, it is always progressive (gets worse over time) and is often debilitating. The onset can be as early as the second year of life or as late as the sixth decade of adulthood. In general, the rate of disease progression is variable in the late-onset form, but a rapid decline can occur at any time and is hard to predict. The primary symptom is muscle weakness, which may include respiratory weakness and failure. The heart is usually not involved.

Major signs and symptoms

- Weakness in the limbs and trunk
- Weakness of the diaphragm and other respiratory muscles
- Weakness in the hip muscles, which can lead to difficulty walking and climbing stairs
- Fatigue (tiredness)
- Reduction in quality of life, such as the ability to work, perform household chores, and move independently outside the home



I told my doctor I could not go on living like this. He referred me to a neurologist, and I finally got a diagnosis that stuck: Pompe.

I was upset when I found out. I didn't want it to be Pompe. I was terrified to tell my family and friends, but it was comforting that I could reassure them that this was not the death sentence Sarah was given. I could not let myself have a pity party. I prayed a lot and did a lot of investigating. Someone told me early on that I would be my own best advocate. That was the best advice I was given.

Pompe Disease

Testing and Diagnosis

Accurate diagnosis and treatment are important for people with Pompe because the disease is always progressive. Because the severity of signs and symptoms, rate of disease progression, and age of symptom onset can vary widely among people with Pompe, it is often mistaken for other disorders with similar signs and symptoms such as:

- Dystrophies (e.g., limb-girdle muscular dystrophy)
- Congenital myopathies
- Motor neuron disorders (neurological disorders that destroy motor neurons, the cells that control voluntary muscle movement such as speaking, walking, breathing, and swallowing; e.g., spinal muscular atrophy)

Doctors may use a variety of tests to assess your condition, such as muscle and movement tests, breathing tests, and laboratory tests. All of these tests reveal some aspect of Pompe, but none of these tests alone can specifically identify the condition. Once your doctor suspects Pompe, he or she can screen for Pompe with a simple blood test that measures GAA enzyme activity in the blood. All people with Pompe have lower than normal GAA activity levels.



Another testing option, called genotyping, uses DNA from a blood sample to see if the person has the gene mutation, or defect, that causes Pompe disease. This is especially useful within families in which the particular mutation is known, or to identify carriers of the disease.

Once Pompe disease is diagnosed, testing of all family members and a consultation with a genetics professional are recommended.



Family Health History & Genetics

What is family health history, how does it affect me?

Family health history is a collection of information about health conditions that run in your family, as well as the eating habits, activities, and environments that your family shares. Your family's health is one part of the entire history of your family. Knowing about health conditions in your family can help you make informed choices.

How can family health history affect my health?

You inherit many things from your parents and grandparents. They pass on culture and values through photos, recipes, stories, spiritual practices, and music. You also inherit some parts of how you look—for example, your height and the color of your eyes. Small structures in cells called genes carry information for these characteristics and guide your body through growth and development. Changes in genes may also lead to a risk in the family for developing certain health conditions, such as Pompe.



George's story



My son Phoenix was born healthy: he had a normal Apgar score, and the neonatologist remarked on how strong his heart was. He was released from the hospital, and everything was fine. But when we took him in for a check-up around four months, the pediatrician noticed a heart murmur. An ultrasound showed an enlarged heart (hypertrophic cardiomyopathy). There are a handful of causes for this, Pompe among them. It took a few months to get the definitive diagnosis, and in that time Phoenix became considerably weaker. Once you lose the muscle, there is no way to recover it. Early detection is key, so newborn screening for Pompe is important.

When you get this rare diagnosis, everyone says, Why me? My answer is, Sometimes you draw the short straw, but you have to accept it. My way to deal with it was research. I read journal articles four or five times each with a dictionary in hand to figure out all the medical jargon. I also joined an online network for patients and families with glycogen storage disorders. It helped me understand the disease and cope with it.

I don't pull any punches when I talk about Pompe. I tell anybody who will listen. I keep it as simple as possible at first, trying to connect all the dots.

Now, Phoenix is nine, he is active and with it. He goes to school, goes to Disney World, does things out there in his wheelchair. He can do it, he just does it a little differently. It helps you appreciate life a lot sooner when the possibility of premature death is present at the beginning of life.

Family Health History & Genetics

Tell me more about my genes

Genes are the instructions inside each of your cells. Since everyone has slightly different genes, everyone has a different set of instructions. Genes are one reason why you are unique!

- Genes carry instructions that tell your cells how to work and grow.
- A person has two copies of each gene, one from the mother and one from the father.
- A change in a gene is called a mutation.
- Genes are inside cells. Every part of your body is made up of billions of cells working together.
- Genes are arranged in structures called chromosomes. Most people have 23 pairs of chromosomes. Copies of the chromosomes are found in each cell.
- Chromosomes are made up of DNA. DNA is the special code that spells out the instructions in your genes.
- Pompe is caused by a mutation in a single gene.



John's story



In hindsight, I notice symptoms of Pompe I've had all my life. Though I was blessed to stay in athletics through college and stay active through my thirties, I was always nagged by a weak back and frequent back injuries. In my thirties, friends teased me about my odd gait.

Pompe showed itself with a vengeance in my 40s. My first response to the increasing weakness was a self-diagnosis that I was an aging, lazy guy. I started seeing a personal trainer and lifting heavy weights. I saw a neurologist, but was misdiagnosed with polymyositis, for which I was medicated with a powerful steroid. Basically, I was doing all the wrong things!

Before I was diagnosed, I hid my illness. My wife and I talked, but that was it. We didn't know what was wrong, except for the fact that I was becoming profoundly weaker. Though I couldn't do a lot of things – ride a bike, mow the lawn or pick up a golf ball – I still looked like a healthy guy, and the ruse held up.

When I finally learned I have Pompe, it was a relief of sorts, and I shared the diagnosis with my family. We put together an exhaustive family tree, but we still could not discern a genetic link. We've alerted the whole family and told our college-age son that he should talk about Pompe with his fiancée before he gets married.

Living with Pompe

How are individuals affected?

Receiving a diagnosis of a genetic condition can be a life-changing event for individuals and families. Fortunately, much has been learned about Pompe, like what causes it and how its symptoms can be managed.

LIVING WITH A CHRONIC DISEASE



It takes time to adjust to and accept the realities of a long-term illness. You might feel affected not just physically, but also emotionally, socially, and sometimes even financially. But understanding more about your condition, and doing your part to manage it, can help you take health challenges in stride. Many people find that taking an active part in the care of a chronic health condition can help them feel stronger and better equipped to deal with lots of life's trials and tribulations.

The symptoms and challenges of Pompe will inevitably require changes in daily routine and lifestyle. But these changes don't always mean you have to give up your independence or the activities you enjoy. Instead, you may just need to learn new strategies and approaches to adapt to your situation. These can be as simple as adjusting expectations about what you can do in a single day. Or, they may be changes such as modifying your home or workstation to better fit your needs. Keeping up with regular routines can also help you feel better, both physically and mentally.



Remember that Pompe is progressive, so your needs will likely evolve over time. A positive attitude, creative problem-solving skills, and the support of others will help you meet new challenges head on.

COORDINATING CARE

Individuals with Pompe have a wide range of symptoms and needs. It is best to be managed by a multidisciplinary team of specialists (such as, neurologist, cardiologist, respiratory therapist, physical therapist and nutritionist) who are knowledgeable about the disease and can offer supportive therapies as well as symptomatic care. Your primary care physician can put the pieces together to make sure your treatment and disease management plans are good for the whole you.

However, you don't have to leave everything to your doctor. You can seek out other people with Pompe. You can also listen to your body and track its changes. This kind of home monitoring might help you spot potentially harmful changes before they become bigger problems.



Living with Pompe

QUALITY OF LIFE

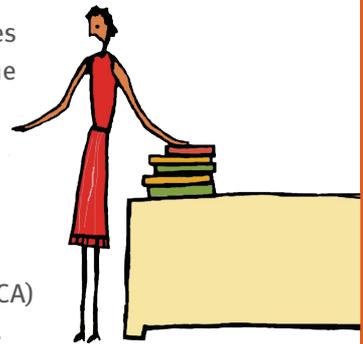
For some patients, discomfort, pain, and disability due to muscular impairment can seriously impact quality of life. Today, with early diagnosis and optimal care, one can plan to manage symptoms and make lifestyle adjustments.

EMPLOYER AND INSURANCE ISSUES

People often hesitate to share their diagnosis, or that of their child, with employers because of fears they will be discriminated against. A law passed in 2008, the Genetic Information Nondiscrimination Act, protects people from genetic discrimination by employers and health insurers based on genetic information, including family health history.

If you have Pompe, employers may make accommodations at work to help you keep doing your job. These may include switching you to part-time or flexible hours, or making changes to your physical work environment. However, there are still reasons to be cautious—not all types of insurance provide coverage, and there are some exemptions for small employers—so you should base your decision whether or not to share your diagnosis or that of your child on your individual circumstances.

The Patient Protection and Affordable Care Act (ACA) offers additional protections from discrimination. Under the law, you cannot be denied insurance coverage or dropped from your plan because of a pre-existing condition. [See the Resources section for links to more information on GINA and the ACA.]





Monique's story



Years before I went to work for a large resort company, I saw an interview with a senior executive talking about his particular degenerative condition. He said it's an inconvenience, but he doesn't let it slow him down. I adopted that as my mantra. We were opening new resorts, working long hours. Everyone around me was exhausted, too, so it just seemed like the norm. But when everyone started to bounce back, I didn't. My breathing was so bad that I would wake up with horrible headaches. I took intermittent medical leave, expecting the doctors to just figure it out and fix me up.

Because I was having so many flare-ups, I decided to file FMLA (Family Medical Leave Act) papers to protect my job. It was not an easy decision—you don't want to be that sick person at work—but it was right to put my health first. There can be a lot of denial and stubbornness, saying "I'm not going to let it beat me." But I knew that if I didn't start taking care of myself, I could get a lot worse and possibly die.

At the time of my Pompe diagnosis I was so ill that finally identifying the cause was a relief. My aunt came to all my appointments with me. From my experience, whether it's a family or a friend, if you have one person to stand by you, it will be ok.

I started a blog to keep family and friends informed, but it also gives me a sense of control. Instead of being a victim of Pompe, I embrace it, live with it, and fight it – mentally, physically, politically, whatever I need to do.

Living with Pompe

How are families affected?

Pompe is an inherited condition. A diagnosis of Pompe in any person may reveal an increased risk that others in the family may carry the gene, or in fact have Pompe. This includes siblings, parents, and children as well as more distant relatives such as cousins. Genetic counseling for families with Pompe is important to help address who should be tested to determine

if they have Pompe or if they are carriers.



Knowing about Pompe can help you make informed choices for yourself and your family. For example, you can:

- Make sure you and your family members are tested for Pompe.
- Get accurate information before making important reproductive choices.
- Learn about treatment options.
- Educate employers, teachers, camp counselors and other members of your care community

EMOTIONAL AND PSYCHOLOGICAL EFFECTS

Learning that you or a family member carry the gene mutation can lead to feelings of depression, hopelessness, or guilt. This might lead to denial that the individual may be affected and can cause someone to delay seeking medical treatment. Be prepared for different reactions and the emotional investment that goes into getting tested.

Knowing about Pompe can help you
make informed choices for yourself
and your family.



Talking about Pompe

Why should I talk to my family about Pompe?

You share a lot with your family—including what can make you sick. Family members can have genes, habits, diet, and environment in common, so they share risk for the same health problems.

Most common diseases (heart disease, diabetes, etc.) are known as “multifactorial” conditions. They are caused by a combination of genes, lifestyle, and environment. In these cases, people with similar genes may not develop the same illness if they make different choices or live in a different environment.

On the other hand, some conditions are caused by a specific change in the DNA of a single gene, called a mutation. Many of these conditions, like Pompe, are rare. In these cases, genes play a much bigger role than lifestyle and environmental influences. These conditions usually develop when an individual is born with a specific gene mutation(s).

It is important to discuss all types of health conditions—common, rare, mild, fatal, acute, and chronic—with your family and doctors. Knowing about the conditions that run in your family can help you make informed decisions about your health.

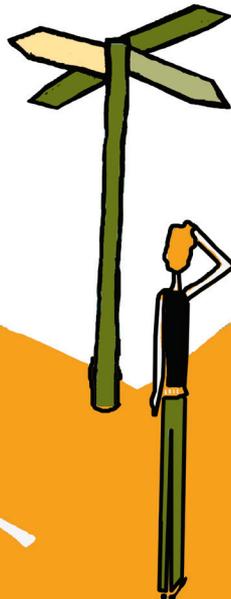
How do I prepare for the talk?

GO THROUGH THE ACCEPTANCE PROCESS YOURSELF

Most people go through stages when learning to cope with a chronic illness. You will have mixed emotions—sadness, guilt, confusion, vulnerability. Everyone’s reaction is different. Seek the help you need to come to terms with your condition or your child’s condition.

KNOW THE FACTS

Know the facts about Pompe so you can answer questions your family members have or refer them to other resources. Use your own experiences and information you learned from your doctor (or your child’s doctor) and others. Remember, Pompe varies from person to person, so talk to healthcare providers for medical advice. [Turn to page 2 for more details on Pompe that you can share with family.]



Talking about Pompe

How do I talk to my family about Pompe?

Pompe disease may cause different symptoms than more common conditions like heart disease, diabetes, and cancer, but the steps you take to talk to your family about it are very similar.

TALK TO YOUR FAMILY

Your relatives are the best source of information about your family. This means you are a source of information for them in return. Family history is often shared during conversations at events like birthday parties, weddings, reunions, religious gatherings, holiday dinners, and funerals. These events provide an opportunity to talk to family members about their lives. Share your purpose.



Explain that the information you are sharing can help not only you but the whole family. You can ask family members questions about their health and tell them what you know about yours.

USE WHAT YOU HAVE

Test results, letters from your doctor, or other information you received about your diagnosis or your child's diagnosis can be shared with interested family members to help them understand your disease. It may explain their chances of having Pompe or the chance of children inheriting Pompe from their parents. Websites and brochures that have been useful for you may also be useful for your family members. This booklet can be passed along to help explain family health history and Pompe.

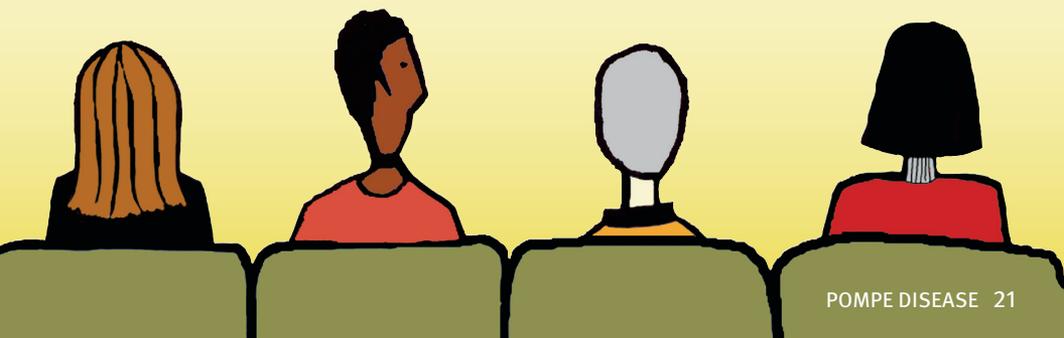
PLAN AN INDIVIDUAL CONVERSATION

After you have brought up Pompe, you may want to talk with certain family members in more detail. Some people respond better to one-on-one talks rather than conversations in a large group setting. This will give you the chance to address questions they have and ask questions of your own.

These individual talks should take place when both you and your family member feel up to it: you should not feel rushed or tired. Give the conversation the energy and attention it deserves. Also make sure your family member does not feel threatened or pressured to talk about everything immediately. Let him or her know you are open to questions and that it is not a one-time conversation; the discussion can continue in the future.

BRING SUPPORT

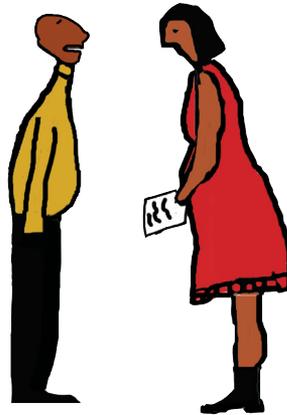
If you have a friend or family member who is a medical professional or who understands Pompe and has been a help to you throughout your (or your child's) diagnosis and treatment, consider whether it would be useful to have him or her there for support during the conversation.



Talking about Pompe

PUT IT IN WRITING

Some people may be more comfortable sharing and receiving health information in a letter or email. You may wish to send your family a message with an update on your/your child's health information and information about Pompe. Your doctor or genetic counselor may be able to help you write this letter and identify who in the family would benefit from being informed. In addition to the initial letter, you might consider starting a blog to keep family and friends informed. This way, you don't have to call or write everyone individually every time you have an update. Services like [CaringBridge.com](https://www.caringbridge.com) provide a space for you to describe your health challenges and progress and for friends and family to offer support.



Crowley Family story



Matt: In the beginning, communication with family about Maddie's illness was limited. The doctors mentioned elevated CK enzyme levels, and we figured it was mononucleosis, so we didn't want to concern anyone. However, once it got more serious, with muscle biopsies and eventually a diagnosis of Pompe, we started to share. My sister has a medical background and was able to explain a lot of the concepts. Our relatives were not going to open a genetics textbook, so getting the information in the family context made more sense. We got a lot of questions from friends and family, so we started a blog to keep everyone informed.

At first, we tried to share information with Maddie slowly. It's a lot to absorb, and she was only 11 at the time. It was tough because we didn't know how much of what we were learning would actually apply to her. What's on the Internet was the worst stuff; they tell you not to look there, so of course you immediately look.

Maddie: I don't talk much about it because I don't want people to worry. A majority of my friends know, but not just random kids at school.

Emma: I was diagnosed a few months after Maddie. I also don't talk about it very often because I don't feel the need to flaunt it. It just happens and it's normal to me. My close friends know, and word has traveled through parents and kids. My friend was in a biology class that had to do a rare disease project; he chose mine and asked me to speak to the class. I love speaking in front of people. If someone has a question, I don't mind answering. I just don't like to initiate the conversation. But I'm very comfortable with my disease. If you're not, it makes it harder to cope and makes it awkward for everyone else.

Talking about Pompe

How do I talk to my children about Pompe?

If you have Pompe, you will need to consider explaining the condition to your children. If you have a child with Pompe, you likely will need to explain the condition to him or her, as well as to any other children you have. In today's world, the Internet provides easy access to an abundance of information with the click of a mouse. However, if possible, it can be beneficial to present the information to your children slowly and simply. Just as you need time to cope with the information and accept it, so will your children. If they are young, it might be difficult for them to understand the implications of the diagnosis. Put it in terms they can understand. For example, "your muscles are sick."

Kids are perceptive to parental stress, and they will have many of the same reactions adults do, such as hopelessness, or guilt if a sibling has Pompe and they do not. Emphasize the



Rachel's story

My symptoms first started when my daughter, Courtney, was four months old. Being a new mom, I thought the weakness was normal. As she grew and I remained weak and tired, I realized something was wrong. We got my Pompe diagnosis 18 months later.

Courtney is in pre-school now. Every once in a while she asks me if we'll ever be able to go on walks together without my scooter or wheelchair. When I tell her I can't walk a lot, she seems ok with it. She knows her own muscles are strong and healthy, but there are

importance of staying positive, and try to keep life as normal as possible. If needed, seek age-appropriate counseling to explore these issues fully as a family.

Conversation starters

There is not one right way to talk to your family about health and Pompe. After you tell your family your story, you may want to let them know what symptoms people can have that might be signs of Pompe.

HEALTH PROBLEMS CAUSED BY POMPE

- Weakness in the limbs and trunk
- Weakness of the diaphragm and other respiratory muscles
- Weakness in the hip muscles, which can lead to difficulty walking and climbing stairs
- Fatigue (tiredness)

some things I cannot do. So we do other things together, like crafts and reading books.

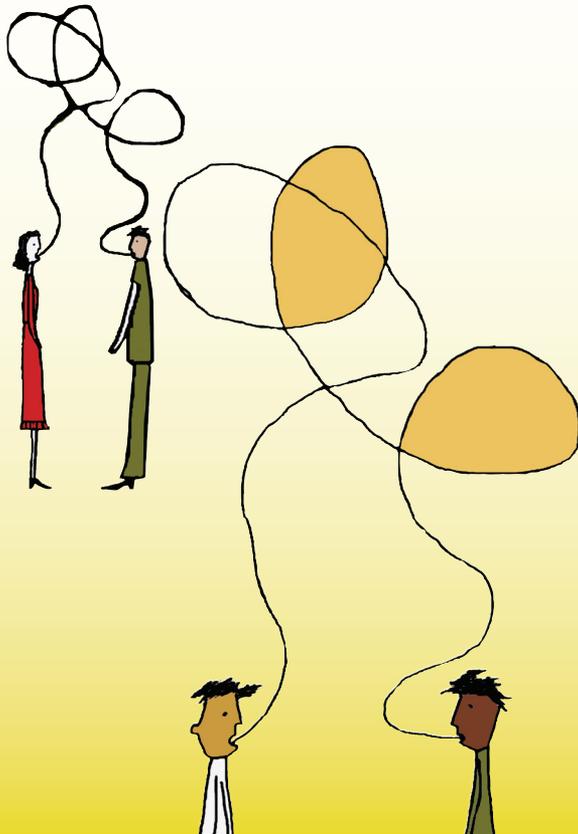
Her friends at school ask her, “Your mom is not an old lady, why does she need a cane?” Courtney tells them, “Mommy’s muscles are sick.” For her, it’s not a big deal; it’s just the way it is. She passes this acceptance on to her classmates, as well, teaching them not to be afraid of people with disabilities.



Talking about Pompe

QUESTIONS ABOUT FAMILY TO HELP BUILD A FAMILY HEALTH HISTORY

- Do you know if your parents or grandparents took medicine on a regular basis? If so, what kinds and for what?
- What kinds of things have people in your family died from?
- Has anyone had problems in pregnancy or childbirth? What kinds of problems?
- Are there any diseases that run in our family?
- Is there anything else you would like to tell me about your life or health concerns in our family?



What should I do if my family does not want to talk about Pompe?

Some family members might not understand why it is important to know about Pompe. Others might be nervous about receiving a diagnosis of Pompe themselves. It is normal to feel anxious or uncomfortable if something is unfamiliar. We are all at different emotional places at different times and will process the information differently.

You may wish to start the conversation by mentioning more common conditions that run in your family, like high blood pressure or asthma. Then move on to Pompe. Make it clear that although there is no way to lower your risk for Pompe, you can still lead your life, with hobbies, a career, and a family.

Try to get family members involved, even if they express initial reluctance or denial. If they are not willing to take steps to learn more on their own behalf, they might be willing to do it for their children or the rest of their family.

Denial can be a way for some people to cope. Just because someone is not listening to you doesn't mean they do not hear what you are saying. Give them the time and space to think about what you have said and become more comfortable with the idea of Pompe.

Talking about Pompe

You may find that some family members won't know how to respond to you about Pompe right now, but they may be able to have the discussion in the future. If this happens, they will be better prepared and able to respond if they have already heard some of the messages about Pompe or have been directed toward information sources they can explore privately on their own in the meantime.

Let them know you are available to talk when they are ready and if they have questions, and give them some resources (on the opposite page) for when they are ready to learn more.

When family members do not want to talk about Pompe, you may feel hurt, upset or alone. Try to understand that it is not unusual for family members to react to and cope with the news differently. When this happens, seek support from doctors, friends, other family members or other people you know with Pompe. There are also online groups with Pompe patients from around the world ready to listen, help, and welcome newly diagnosed Pompe patients and families. Blogs, Facebook groups, Twitter feeds, and other forums dedicated to Pompe where all you to share your own experiences and learn from others.



RESOURCES

LEARN MORE AND GET INVOLVED

Acid Maltase Deficiency Association: www.amda-pompe.org

Muscular Dystrophy Association:
www.mda.org/disease/amd.html

Pompe Community: www.pompe.com

United Pompe Foundation: www.unitedpompe.com

DIAGNOSTIC TESTING AND REGISTRY

GeneTests: www.genetests.org

Pompe Registry: www.pomperegistry.com

MORE INFORMATION

Genetic Alliance: www.geneticalliance.org

Genetic Information Nondiscrimination Act (GINA):
www.ginahelp.org

Genzyme Medical Information: 800.745.4447

National Organization for Rare Disorders:
www.rarediseases.org

Patient Protection and Affordable Care Act (ACA):
www.healthcare.gov/law



Glossary

Acid alpha-glucosidase (GAA)—An enzyme that is missing, ineffective, or present in inadequate amounts in people with Pompe disease. It is normally found in the lysosomes.

Autosomal recessive—A pattern of inheritance where two non-working copies of the gene are needed in order to have the condition. Autosomal refers to the 22 pairs of autosomes, or non-sex chromosomes. Autosomal recessive conditions affect males and females equally.

Carrier—A person who has a change in only one gene of a pair, with the other gene of the pair working normally. Carriers typically do not display symptoms of the condition, but can pass on the mutation to offspring.

Chromosome—A threadlike strand of DNA and protein in every plant and animal cell.

Chromosomes carry the genes that define a person's bodily makeup (for example: hair and eye color, disease expression)

DNA—An abbreviation for deoxyribonucleic acid, the storehouse of all hereditary characteristics. All chromosomes are made up of genes, and genes are made of DNA.

Enzyme—A protein produced by the body that acts to chemically change other substances. Enzymes are involved in breaking down or chemically altering substances so that the body can use or excrete them. Enzymes are typically named by adding an “---ase” to the end of a word that describes them.

Gene—a piece of DNA that codes for a particular substance. Each gene occupies a specific location on a chromosome, which defines a person’s bodily makeup and function

Genetic—Affecting or affected by genes (genetic disorder)

Genetic Information Nondiscrimination Act of 2008 (GINA)—A federal law that protects individuals from genetic discrimination in health insurance and employment. Genetic discrimination is the misuse of genetic information.

Lysosome—A small structure in most cells acting as the chemical plant of the cell.

Lysosomes contain and make various enzymes that digest or break down substances. In Pompe disease, glycogen builds up in the lysosomes.

Lysosomal storage disease—A disease resulting from the storage or accumulation of material in the lysosomes. Pompe disease is a lysosomal storage disorder

Pompe disease— A genetic disorder caused by a deficiency of the enzyme acid alpha-glucosidase (GAA).

Protein—the building blocks of cells involved in all essential life functions (for example: cell growth, energy production, metabolism).



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