GETTING THE RIGHT CARE FOR POMPE DISEASE

It takes a team of healthcare providers to meet the needs that may arise with Pompe disease. Each member of your healthcare team has unique skills to offer. One doctor may serve as team leader and coordinate the care given by all healthcare providers. With Pompe disease being so rare, it is common to see providers who have never treated anyone with this neuromuscular disorder. It will help to work closely with the medical team to get the treatment and services you or your child really need. Learning all about Pompe disease can help you take charge of your care. This brochure will help get you started.

Q: I know it's important to learn about Pompe disease, but where do I begin?

A: You can find good resources of information on the Internet. Much of it comes from groups that were started by patients or parents of children with Pompe disease. You can also learn a lot from groups that support research into other neuromuscular disorders, as well as from government agencies and major medical centers around the world. (The list of resources in Where to Learn More on the last page can help you get started).

Q: How is Pompe disease treated?

A: There are a number of effective Enzyme Replacement treatments (ERT) available worldwide and there are trials of Gene Therapy treatments also planned and ongoing. For more information on these direct treatments, please review the Pompe Connections: Treatment Edition.

In addition to direct interventional treatments like ERT, supportive therapies help people with Pompe disease keep up their strength and prevent health problems caused by muscle weakness. Supportive therapies are tailored to the specific needs of the individual patient. Respiratory therapy can provide the support needed for breathing. Physical therapy can help increase movement, lessen stiffness, strengthen muscle, and ease pain. Dietary therapy helps to maintain calories and prevent weight loss. Everyone with Pompe disease faces new challenges as their body mechanics' change. Getting the right therapy from a knowledgeable medical team combined with the support and care of those around you can help you live each day to its fullest.

Q: Who are the key people on my healthcare team?

A: It takes a wide range of skills to meet the needs of someone with Pompe disease. In addition to your family doctor or primary care provider, the chart below lists <u>possible</u> members of your medical team. The team will need to be kept informed of any and all health changes or new symptoms that occur. Be sure each team member knows the care you are receiving from other team members. If you have a child with Pompe disease, it is important to keep track of your child's symptoms and reactions to treatment, and assist with supportive therapies.

Primary Care Physician:	A physician who provides both the first contact for a person with an undiagnosed health concern as well as continuing care of varied medical conditions, not limited by cause, organ system, or diagnosis. Will also manage non-Pompe medical issues that arise.
Pediatrician:	A doctor that specializes in the medical care of children.
Medical Geneticist:	The study of diseases and disorders that is at least partially genetic in origin.
Neurologist:	A specially trained physician who diagnoses and treats disorders in the nervous system. This includes diseases of the brain, spinal cord, nerves, and muscles.
Cardiologist:	A specialist in the structure and function and disorders of the heart.
Pulmonologist or Respiratory Specialist:	The branch of medicine that deals with dysfunction of the respiratory system. Treats the breathing problems caused by muscle weakness.
Gastroenterologist:	A physician who specializes in the diagnosis and treatment of disorders of the gastrointestinal tract. Treats problems related to digestion and feeding difficulties.
Orthopedist:	A specialist in treating and correcting deformities of the skeletal system. An orthopedist treats scoliosis (curvature of the spine), contractures (muscle tightness), and other joint or bone problems related to muscle weakness.
Respiratory Therapist:	Works with the pulmonologist to treat patients who have difficulty breathing. They operate equipment such as respirators and ventilators.

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Dietitian:	Concerned with the promotion of good health through proper diet and with the therapeutic use of diet in the treatment of disease.
Genetic Counselor:	Deals with individuals and families who have a genetic disease or who are at risk for such a disease. Genetic counseling is designed to provide patients and their families with information about their condition, risks for future pregnancies, and treatments to help them make informed decisions.
Psychologist:	A person trained and educated to perform psychological therapy, testing and research. Helps people cope with their fears and worries and deal with practical concerns.
Social Worker:	Draws on the social and behavioral sciences to meet the needs of clients including financial, family and resources.
Physical Therapist	Help individuals with disabling conditions by providing services that restore function, improve mobility, relieve pain, and prevent or limit permanent physical disabilities. They restore, maintain, and promote overall fitness and health.
Occupational Therapist:	Help to enable people to participate in the activities of everyday life. They achieve this outcome by enabling people to do things that will enhance their ability to participate or by modifying the environment to better support participation.
Nurse:	Assists people, sick or well, in the performance of the activities contributing to health or its recovery that they would perform unaided if they had the necessary strength, will or knowledge and to do this in such a way as to help the person gain independence as rapidly as possible.

Q: How do I find a medical center that has experience in dealing with Pompe disease?

A: There is an increasing understanding of Pompe disease with a number of medical centers around the world specializing in treating Pompe disease. Clinics and rehabilitation centers that treat similar neuromuscular disorders or rare genetic diseases may also provide expert care for people with Pompe disease.

In the USA, Europe and other continents, there are many specialized centers for people with Pompe and neuromuscular disorders.

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Q: How can the family stay involved?

A: Recognizing that the “family” is the constant in a patient’s life, a plan of care should be developed by the family and treating physician or provider that centers on the needs identified by the family and health care providers working as a team. Families and patients should form a partnership with the professional team at all levels of care to allow for equal participation to establish the goals of care. Multiple needs which may include separation from core family members due to hospitalization or therapy, transportation, lodging costs, loss of work days, specialized equipment including an adapted vehicle, household assistance, regular medical procedures, vocational retraining, and educational programming could be addressed. Information about therapies, treatments, and resources can be continuously shared with the family and/or patient in an appropriate and supportive manner that is sensitive to your family relationships. Timely referrals to appropriate community agencies, including a social worker or other case workers, and advocating for the needed community services are essential support that can be requested of the individual coordinating the care of the patient. The family and all providers should update the care plan and patient progress at regular intervals. This will insure comprehensive coordinated care that is family centered creating a partnership in care.

Q: Beyond medical advice, where can I turn to for help with all the practical challenges I will face?

A: Pompe patient support organizations are an invaluable resource for you and your family. Many countries have national support organizations (a list by country can be found on the IPA’s Affiliates page). They can provide support on a wide-variety of topics, from emotional support to educational resources.

Depending on where you live, there may be services available to help with access to resources such as: groups that can help review your finances, apply for government benefits and financial aid, find medical and social services, set up an education plan for your child, arrange for home care, help you to learn your legal rights, and deal with employment issues and insurance problems. Your local Pompe patient support organizations, medical team, and other sources can help to put you in touch with these groups.

Parent to parent and patient to patient support has also been reported by many to be one of the most valued resources provided. Another consideration is to identify support for siblings, grandparents and other family members. Patient support organizations can assist with this, and so can connecting with other Pompe patients through social media, such as the Pompe specific Facebook groups.

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Q: I feel like I know more about Pompe disease than some of the healthcare providers who are treating my child. What can I do about this?

A: Many caregivers and patients with rare disorders like Pompe disease feel that the only way to be sure of getting the right treatment is to learn all they can about the disease. Knowledge can be a real advantage when you are dealing with a large number of healthcare providers. You should keep track of what everyone is doing. This may help prevent mistakes or errors from occurring. There may be times when it is necessary to educate a pediatrician, or family physician, or emergency room doctor who has not had much experience with Pompe disease. Think of your knowledge as a skill you have to share with the healthcare team. And when you have concerns, do not hesitate to offer your opinion or question why something is being done. Remember that no one has a greater stake in your care or your child's care than you do.

Where to Learn More: See Further information section