



Nutrition and Dietary Therapy

Many people with Pompe disease have trouble with eating. Muscle weakness can make it hard to bite, chew, suck, or swallow food. This can make it hard for patients of all ages to eat comfortably and get the nutrition their bodies require. Breathing problems may also make people too tired to eat. As a result, many people with Pompe disease have trouble gaining weight, keeping it on, and getting proper nutrition. Weak swallowing muscles pose a risk of accidentally inhaling food or liquid into the lungs. Weak muscles may also slow the passage of food through the body. This can lead to digestion or bowel problems. Changing your diet or your eating habits or getting nutrition through tube feeding can help manage these problems. This handout explains the benefits of dietary therapy for people with Pompe disease.



Q: Why do people with Pompe disease have trouble with eating?

A: People with Pompe disease have feeding and swallowing difficulties and infants often fail to thrive. As glycogen builds up in the cells, the muscles that we use to eat and digest food get weaker and weaker. This affects people in different ways.

In children diagnosed with infantile Pompe disease, contributing factors for feeding and swallowing difficulties include weakness of the face or mouth muscles (facial hypotonia), enlarged tongue (macroglossia), tongue weakness, and decreased ability to achieve tongue cupping and lip seal for sucking. Oral stimulation and non-nutritive sucking for non-oral feeders should be provided to maintain normal oral sensory development and to develop emerging oral skills. Modified dietary textures should be re-evaluated on a regular basis to maintain safety in light of a changing disease process. Improvements in swallowing dysfunction have been noted in some infantile-onset patients receiving ERT and patients have been able to resume oral feeding. Growth parameters such as height, weight and head circumference should be followed closely.

In patients with late onset Pompe disease, fatigue of jaw muscles with difficulty swallowing and chewing food is often a first complaint and may contribute to inadequate intake (total calories, vitamins, and minerals) and muscle protein breakdown.

Nutrition and Dietary Therapy

In more serious cases, such as infants who are too weak to eat at all on their own, older patients who are severely underweight, or those whose breathing problems interfere with eating, tube feeding may be necessary.

Q: What steps can I take to improve my diet and eat better?

A: Foods that are too large, very thick, dry, or solid can be difficult to chew or swallow and you risk inhaling them into your lungs (aspiration). To make them easier to eat, change the size, texture, or thickness of foods by mashing them up, chopping them into smaller pieces, or by mixing them with some type of sauce or gravy. Take small mouthfuls of food and chew your food well. Eat several smaller meals instead of 3 large ones. Drink liquids slowly and be sure to drink enough liquids throughout the day. Drinking with a straw and staying upright for an hour or two after eating may also be helpful. Food and drinks that are thin (such as soup, broth, water, or milk) can also be hard to swallow. They can move through the throat too quickly and make you choke. Try thickening drinks and soups with baby rice cereal, cornstarch, or special powders that are made for this purpose. Work with a registered dietician who can plan well-balanced meals for you or your child in order to provide enough calories and nutrients that you need to have each day. You may be able to replace meals with nutritional drinks that supply needed vitamins and minerals. Special exercises may help strengthen eating-related muscles.

Your doctor may ask you or your child to see a speech-language pathologist, (also called a speech therapist). A speech therapist is able to assess, diagnose, treat, and help to prevent disorders related to speech, language, cognitive-communication, and swallowing. A speech therapist can teach you different ways to eat and chew that reduce the risk for aspiration.

Q: What is a videofluoroscopic swallow study and should people with Pompe disease have this study done?

A: A videofluoroscopic swallow study should be done at baseline for everyone diagnosed with Pompe disease as they are at an increased risk for aspiration (food or liquid entering the trachea). A videofluoroscopic swallow study (also commonly referred to as modified barium swallow study) is an objective assessment of swallow function. The purpose of the videofluoroscopic swallow study is to assess for aspiration with oral feedings. During the swallow study the patients are positioned in a typical feeding position consistent with their age and development. They are given a variety of food consistencies (thin liquid, thick liquid, puree, soft solid, hard solid) injected with barium. The study assesses the phases of swallowing:

- **The Oral Phase:** Swallowing starts with the oral phase, in which food is placed in the mouth and moistened and chewed with the aid of the muscles of mastication (chewing).
- **The Pharyngeal Phase:** As the bolus (a small round soft mass of chewed food) reaches the pharynx, special sensory receptors activate the involuntary part of swallowing. A critical part of the pharyngeal phase is the involuntary closure of

Nutrition and Dietary Therapy

the larynx by the epiglottis and vocal cords, and the temporary inhibition of breathing, both of which prevent food from going “down the wrong pipe” into the airway (trachea) and the lungs. The closure of the larynx by the epiglottis protects the lungs from injury, as food and other particles can lead to severe infections and irritation of the lung tissue. Lung infections caused by problems with the pharyngeal phase of the swallowing reflex are commonly known as aspiration pneumonia.

- **Esophageal Phase:** As food leaves the pharynx, it enters the esophagus, a tube-like muscular structure which leads food into the stomach due to its rhythmic contractions. The esophagus has two important sphincters, namely the upper and lower esophageal sphincters, which under normal conditions prevent food or saliva from being regurgitated toward the mouth. In doing so, the esophageal sphincters serve as a physical barrier to regurgitated food. Both esophageal sphincters, first the upper, and then the lower, open reflexively as food is brought down during swallowing.

If the risk for aspiration is high, oral feeds may need to be stopped and the patient tube fed.

Q: What are tube feedings and why do people diagnosed with Pompe disease sometimes need to have them?

A: Tube feeding provides complete nutrition for late-onset patients who are not able to take food by mouth because of chewing, swallowing, or breathing problems. Tube feedings are also used for babies with infantile-onset disease who are too weak to suck from a breast or bottle or are not gaining weight. Tube feeding also helps to prevent food from going into the lungs when food “goes down the wrong way”. The state of being fed by a feeding tube is called enteral feeding or tube feeding. In some instances a combination of oral and tube feeding may be recommended to allow for adequate intake of calories, and to allow for normal oral sensory development.

There are 2 types of feeding tubes:

- **A Nasogastric (NG) tube:** A NG tube is inserted through the nose and delivers nutrients directly into the stomach.
- **Gastrostomy tube (or G-tube):** A G-tube is surgically placed through an opening in the stomach wall and delivers nutrients directly into the stomach. A G-tube is a good option for people who may require tube feedings for a longer period of time.

Q: What can I do to manage digestion or bowel problems?

A: The best advice is to talk with your healthcare provider. Describe the problems you are having and ask for help in managing the symptoms. Weakness of the muscles that move the food toward the stomach may cause heartburn, or acid reflux also called gastroesophageal reflux disease (GERD). This occurs when swallowed food and stomach acids flow back toward the mouth through the esophagus (a muscular tube through which food passes from the pharynx to the stomach). Eating smaller, more

Nutrition and Dietary Therapy

frequent meals and remaining in an upright position during and after meals may be helpful. Weakness of the muscles in the chest or abdomen can make it hard to push waste out of the body when you have a bowel movement. This may lead to constipation. Constipation can also lead to diarrhea. Eating a diet high in fiber and drinking lots of fluids can help prevent constipation. Be sure to check with your healthcare provider before taking any medicines for digestion, diarrhea, or bowel problems.

Q: I've heard that a high-protein diet can help people with Pompe disease. What's known about it?

A: A high-protein diet does seem to help some children and adults with late-onset Pompe disease. The diet is based on the theory that eating more protein and fat (like meat, eggs, cheese, and butter) and fewer carbohydrates (like bread and pasta) may help slow the muscle weakness that occurs when too much glycogen builds up in the cells. There are some patients whose muscle strength and ability to walk have improved after following a high-protein diet. But many others showed no sign of improvement.

For patients with late-onset Pompe disease, the goal is intended to manage the:

- 1) Increasing accumulation of glycogen
- 2) Increase in amino acid utilization.

It has been observed that a high protein-low carbohydrate diet plus aerobic exercise may be beneficial to some of these patients. The rationale to this form of therapy is an attempt to decrease glycogen deposition, increase muscle fatty acid utilization, and at the same time compensate for the increased amino acid oxidation that has been shown to occur in Pompe disease. A study, failed to demonstrate improvement in the late-onset Pompe disease patients treated with a high protein diet and alanine supplementation. A high protein diet may be a good adjunct to ERT but randomized controlled studies are needed. Overall, maintaining good nutrition with attention to macronutrients (protein, fat, and carbohydrates) and micronutrients (vitamins) is important in the management of all patients with Pompe disease.

Q: How can alanine and other nutritional supplements help people with Pompe disease?

A: Alanine is an amino acid, one of the building blocks of protein. A few studies have suggested that alanine may give children and adults with Pompe disease more energy if taken in small amounts through the day. Though the scientific evidence is limited, healthcare providers who favor this approach feel that alanine supplements help to prevent muscle wasting when glycogen builds up in the cells. Alanine comes in a powdered form that can be mixed with food.

Q: Are there any specific gastro-intestinal / nutrition recommendations?

A: Yes, the following are gastro-intestinal / nutrition recommendations:

- Obtain videofluoroscopic swallowing assessment and evaluation for GE reflux to guide management of feeding (oral/gavage feeding) at baseline and as clinically indicated.

Nutrition and Dietary Therapy

- Provide oral stimulation and non-nutritive sucking for infants who are nonoral feeders.
- Monitor growth parameters carefully.
- Provide adequate nutrition (high protein consisting of 20– 25% protein) with attention to vitamins and minerals.
- Encourage appropriate exercise in consultation with a physical therapist with experience in Pompe disease.

Q: What is Osteopenia and osteoporosis and why should patients with Pompe disease be screened for this?

A: Osteopenia is the thinning of bone mass. A decrease in bone mass is considered a serious risk factor for the development of osteoporosis. The diagnostic difference between osteopenia and osteoporosis is the measure of bone mineral density.

Osteoporosis, the "fragile bone" disease is characterized by a loss of bone mass caused by a deficiency in calcium, vitamin D, magnesium and other vitamins and minerals. Many of the foods you eat contain these bone-building minerals.

Femur fracture and thoracic vertebral fractures have been identified in patients with infantile onset Pompe disease. Osteopenia has been seen in patients with Pompe disease as young as 4 months of age, which could be attributed to chronic immobilization and weakness, but osteopenia has also been identified in patients with Pompe disease with good motor strength and nutrition and needs to be further studied. The pathophysiologic mechanisms of osteopenia/osteoporosis in Pompe disease are not yet well understood so are managed generically.

Factors that have the potential to contribute to osteopenia and osteoporosis have therapeutic implications. Nutrition needs to be adequate especially in terms of intake of calcium and Vitamin D and attention given to medications (e.g., long term use of certain diuretics which could cause hypercalciuria, long term steroid use). Potential interventions may include the provision of weight bearing in physical therapy and in standing devices. There is insufficient evidence to suggest pharmacologic therapy such as bisphosphonates as a preventive treatment in Pompe disease at the current time.

Emerging reports of osteopenia, osteoporosis, and fracture, suggest that screening patients with Pompe disease for osteopenia is indicated.

Bone mineral density (BMD) is the measurement of calcium levels in bones, which can estimate the risk of bone fractures. It is also used to determine if a patient has osteopenia or osteoporosis. Bone mineral density tests are non-invasive and painless procedures usually done on the hip, spine, wrist, finger, shin bone, or heel. While osteopenia can be diagnosed using plain radiographs, the most common method for measuring BMD (and a way to definitively diagnose osteoporosis) is through Dual Energy X-ray Absorptiometry or DEXA. This scan uses low-energy x-rays that expose patients to much less radiation than standard x-rays and can assess calcium levels in

Nutrition and Dietary Therapy

bone. The results are measured as a "score" and are compared to those of healthy individuals.

What do the numbers mean? A patient's BMD is given a T-score, which is obtained by comparing it to an average score for a healthy 30-year-old of the same sex and race. The difference between the "normal young" score and the patient's score is referred to as a standard deviation (SD). T-scores can fall as low as -1 SD and still be considered healthy. Patients with T-scores between -1 SD and -2.5 SD are diagnosed with osteopenia and are considered at high risk for osteoporosis. Patients with T-scores lower than -2.5 SD are diagnosed with osteoporosis. For these patients, treatment may be necessary and may include the use of medications to help increase bone mass, as well as lifestyle changes such as diet and exercise.

Q: What are vitamins and why does our body need them?

A: The body uses vitamins for a variety of biological processes, including growth, digestion, and nerve function. There are 13 vitamins that the body absolutely needs: vitamins A, C, D, E, K, and the B vitamins (thiamine, riboflavin, niacin, pantothenic acid, biotin, vitamin B-6, vitamin B-12 and folate).

There are two categories of vitamins:

- **Water-soluble vitamins:** These vitamins are easily absorbed by the body.
 - a. Vitamins B complex and C are water-soluble vitamins.
 - b. Water-soluble vitamins dissolve in water and are not stored; they are eliminated by the kidneys.
 - c. Because these vitamins are not stored, people need a continuous supply of them in their diet.

- **Fat-soluble vitamins:** These vitamins are absorbed into the body with the use of bile acids, which are fluids used to absorb fat. The body stores these vitamins for use as needed.
 - a. Vitamins A, D, E and K are fat soluble vitamins.
 - b. Fat soluble vitamins dissolve in fat and are stored in the liver and fatty tissues, and are eliminated much more slowly than water-soluble vitamins.
 - c. These vitamins are stored, so they are not needed in your diet every day.
 - d. Fat-soluble vitamins are stored for long periods, so they generally pose a greater risk for toxicity than water-soluble vitamins when consumed in excess.

You can usually get all your vitamins from the foods you eat and your body can also make vitamins D and K. People who eat a **vegetarian diet** may need to take a vitamin B12 supplement.

Nutrition and Dietary Therapy

Name of Vitamin	Major Functions	Common Food Source
Vitamin A	Vitamin A does much more than help you see in the dark. It stimulates the production and activity of white blood cells, takes part in remodeling bone, helps maintain the health of endothelial cells (those lining the body's interior surfaces), and regulates cell growth and division. Beta carotene is an antioxidant and may protect against cancer.	<p>Vitamin A: Liver, vitamin A fortified milk and dairy products, butter, whole milk, cheese, egg yolk</p> <p>Provitamin A: Carrots, leafy green vegetables, sweet potatoes, pumpkins, winter squash, apricots, cantaloupe.</p> <p>It's best to choose a multivitamin supplement that has all or the vast majority of its vitamin A in the form of beta-carotene.</p>
Vitamin D	Vitamin D helps ensure that the body absorbs and retains calcium and phosphorus, both critical for building bone. Laboratory studies also show that vitamin D keeps cancer cells from growing and dividing, and plays a critical role in controlling infections	Very few foods naturally contain vitamin D. Good sources include dairy products and breakfast cereals (both of which are fortified with vitamin D), and fatty fish such as salmon and tuna.
Vitamin E	<p>Vitamin E is an antioxidant that protects body tissue from damage caused by unstable substances called free radicals. Free radicals can harm cells, tissues, and organs. They are believed to play a role in certain conditions associated with aging.</p> <p>Vitamin E is also important in the formation of red blood cells and helps the body to use vitamin K.</p>	<p>Good sources of vitamin E include sunflower and safflower oils, oil-based salad dressings, almonds, sunflower seeds, peanut butter, and dark leafy greens.</p> <p>Vitamin E is found in the following foods: Wheat germ, corn, nuts, seeds, olives, spinach and other green leafy vegetables, asparagus, vegetable oils (corn, sunflower, soybean, and cottonseed).</p> <p>Products made from these foods, such as margarine, also contain vitamin E.</p>
Vitamin K	<p>Vitamin K helps make four of the 13 proteins needed for blood clotting.</p> <p>Vitamin K is also involved in building bone.</p>	Vitamin K is found in many foods, especially green, leafy vegetables (kale, collard greens, broccoli, Brussels sprouts, parsley) and commonly used cooking oils. Some, but not all, multivitamins contain a small

Nutrition and Dietary Therapy

		amount of vitamin K.
Vitamin C (Ascorbic Acid)	Vitamin C plays a role in controlling infections. It's also a powerful antioxidant that can neutralize harmful free radicals, and it helps make collagen, a tissue needed for healthy bones, teeth, gums, and blood vessels.	Excellent food sources of vitamin C are citrus fruits or citrus juices, berries, green and red peppers, tomatoes, broccoli, and spinach. Many breakfast cereals are also fortified with vitamin C.
Thiamin (B-1)	Helps release energy from foods; promotes normal appetite; important in function of nervous system.	Dietary sources of thiamin include beef, brewer's yeast, legumes (beans, lentils), milk, nuts, oats, oranges, pork, rice, seeds, wheat, whole grain cereals, and yeast. In industrialized countries, foods made with white rice or white flour is often fortified with thiamin.
Riboflavin (B-2)	Riboflavin works with the other B vitamins. It is important for body growth and red blood cell production and helps in releasing energy from carbohydrates.	Lean meats, eggs, legumes, nuts, green leafy vegetables, dairy products, and milk provide riboflavin in the diet. Breads and cereals are often fortified with riboflavin.
Niacin (B-3)	Energy production from foods; aids digestion, promotes normal appetite; promotes healthy skin, nerves. Niacin assists in the functioning of the digestive system, skin, and nerves. It is also important for the conversion of food to energy.	Liver, fish, poultry, meat, peanuts, whole and enriched grain products. Niacin (also known as vitamin B3) is found in dairy products, poultry, fish, lean meats, nuts, and eggs. Legumes and enriched breads and cereals also supply some niacin.
Vitamin B-6 (pyridoxine)	Vitamin B-6 aids in protein metabolism, absorption; aids in red blood cell formation; helps body use fats	Good sources of vitamin B6 include fortified cereals, beans, poultry, fish, and some fruits and vegetables.
Folate (folic acid)	Aids in protein metabolism; promotes red blood cell formation; prevents birth defects of spine, brain; lowers homocystein levels and thus coronary heart disease risk.	Many foods are excellent sources of folate—fruits and vegetables, whole grains, beans, breakfast cereals, and fortified grains and grain products.

Nutrition and Dietary Therapy

	Too little folate, is linked to birth defects such as spina bifida and anencephaly.	
Vitamin B-12	Aids in building of genetic material; aids in development of normal red blood cells; maintenance of nervous system.	Found only in animal foods: meats, liver, kidney, fish, eggs, milk and milk products, oysters, shellfish.
Pantothenic Acid	Pantothenic acid is needed to form coenzyme-A (CoA), and is critical in the metabolism and synthesis of carbohydrates, proteins and fats.	Small quantities of pantothenic acid are found in nearly every food, with high amounts in whole-grain cereals, legumes, eggs, meat, and royal jelly.
Biotin	Biotin is necessary for cell growth, the production of fatty acids, and the metabolism of fats and amino acids.	Liver, kidney, egg yolk, milk, most fresh vegetables, also made by intestinal bacteria.

Where to Learn More

Information for this brochure was obtained from the following resources:

- American College of Medical Genetics (ACMG) Practical Guideline: Pompe Disease Diagnosis and Management Guideline 2006. Vol. 8. No. 5. The ACMG guidelines were designed as an educational resource for physicians and other health care providers.
- The Genzyme Corporation's Pompe Community website offers the Pompe community comprehensive information on the disease, as well as resources and support to help manage the challenges it may bring www.pompe.com.
- Center for Lysosomal and Metabolic Diseases Erasmus MC University Medical Center www.erasmusmc.nl/
- Genetics Home Reference: Your Guide to Understanding Genetic Conditions. <http://ghr.nlm.nih.gov/gene/GAA>

Information, Advice, and Support

For more advice and information about dietary therapy for people with Pompe disease, these sources may be helpful:

- The International Pompe Association (IPA) is a global federation of Pompe disease patient groups. The IPA helps patients, family members, and healthcare providers from around the world share their experiences and knowledge across continents and cultures. To find the contact for your country, visit the IPA Web site at www.worldpompe.org

Nutrition and Dietary Therapy

- Acid Maltase Deficiency Association (AMDA): The AMDA was formed to assist in funding research and to promote public awareness of Acid Maltase Deficiency, also known as Pompe's Disease. Visit the website www.amda-pompe.org
- Caring for a child with Pompe's disease offers tips on feeding a baby with infantile-onset Pompe disease. It is available online at www.pompe.org.uk/agsdarne.html.
- Medline Plus – Vitamins: <http://www.nlm.nih.gov/medlineplus/vitamins.html>.
- FDA - Fortify Your Knowledge About Vitamins
<http://www.fda.gov/ForConsumers/ConsumerUpdates/ucm118079.htm#why>
- Video Fluoroscopic Swallow Study: For more information refer to Children's Hospital Boston website:
<http://www.childrenshospital.org/clinicalservices/Site2145/mainpageS2145P4sublevel15.html>
- GSDNet: Join the GSDNet Listserv to exchange emails with people around the world living with Pompe disease. To subscribe to GSDNet by email:
"To" Line: Type the following onto the "To" Line: listserv@listserv.icors.org
"CC" Line: Leave the CC line blank
"Subject" Line: Leave the subject line blank.
Email Message: Type the following message into the body of the email:
Subscribe GSDNet <add your name>



- Understanding Pompe Disease is a free booklet that gives a good introduction to Pompe disease. It is available on the Pompe Community Web site at www.pompe.com.
- The Physicians Guide to Pompe Disease can be ordered from the National Organization for Rare Disorders (NORD) at www.rarediseases.org/programs/pompe_brochure.html

Nutrition and Dietary Therapy

- **Medical Centers Specializing in Neuromuscular Disorders:**
To locate medical centers that specialize in treating neuromuscular disorders, contact the Neuromuscular disorders (NMD) association for your country.
 - In the United States, contact the Muscular Dystrophy Association (MDA) at www.mdausa.org/clinics
 - In Europe, contact the European (EAMDA) at Web site www.eamda.net
 - In other continents, visit the World Alliance of Neuromuscular Disorder Associations (WANDA) at Web site www.wandaweb.org and click on Your Country

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