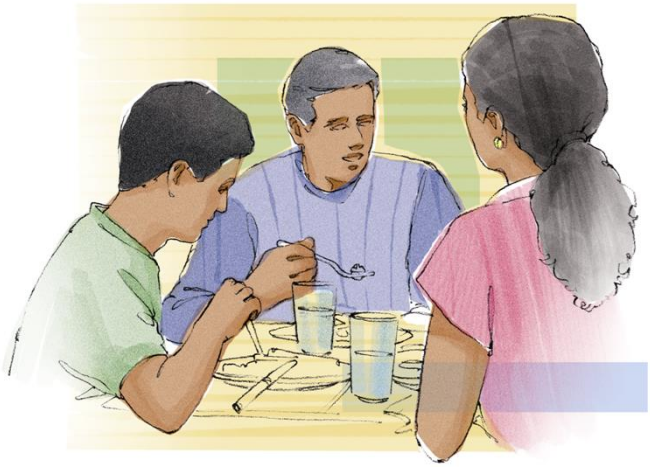
# Nutrition and Dietary Therapy



*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 that 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/or getting proper nutrition. Having weak swallowing muscles poses 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 diet or eating habits or getting nutrition through tube feeding can help manage these problems. This brochure 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 are used to eat and digest food, continues to weaken over time. This affects people in different ways.

In children diagnosed with infantile Pompe disease, there are many contributing factors for feeding and swallowing difficulties that 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 the 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.

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. More information on tube feeding is provided below.

**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 the 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 video-fluoroscopic swallow study and should people with Pompe disease have this study done?**

**A:** A video-fluoroscopic swallow study may be done at baseline for everyone diagnosed with Pompe disease as they can be at an increased risk for aspiration (food or liquid entering the trachea). A video-fluoroscopic swallow study (also commonly referred to as modified barium swallow study) is an objective assessment of swallow function. The purpose of the video-fluoroscopic 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 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 may need to be 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

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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 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. This is because you are limiting the amount of carbohydrates that are consumed (which Pompe patients are not able to break down) and replacing them with protein and fat that can be properly used for energy. 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; and
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. [Ref 1] 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 high protein diet may be a good adjunct to ERT. [Ref 1 and 2] 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.

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**Q: Are there any specific gastro-intestinal / nutrition suggestions that should be reviewed with your clinical team?**

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

- Video-fluoroscopic swallowing assessment and evaluation for GE reflux to guide management of feeding (oral/gavage feeding) at baseline and as clinically indicated.
- Provision of oral stimulation and non-nutritive sucking for infants who are nonoral feeders.
- Monitor growth parameters carefully.
- Providing adequate nutrition (high protein consisting of 20– 25% protein) with attention to vitamins and minerals.

**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.

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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.

| Name of Vitamin | Major Functions | Common Food Source |
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| 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. | **Vitamin A:** Liver, vitamin A fortified milk and dairy products, butter, whole milk, cheese, egg yolk<br>**Provitamin A:** Carrots, leafy green vegetables, sweet potatoes, pumpkins, winter squash, apricots, cantaloupe.<br>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. |
| 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 | 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.<br>Vitamin E is also important in the formation of red blood cells and helps the body to use vitamin K. | Good sources of vitamin E include sunflower and safflower oils, oil-based salad dressings, almonds, sunflower seeds, peanut butter, and dark leafy greens.<br>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). Products made from these foods, such as margarine, also contain vitamin E. |

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| Vitamin K | Vitamin K helps make four of the 13 proteins needed for blood clotting.<br>Vitamin K is also involved in building bone. | 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 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.<br>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.<br>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 | Good sources of vitamin B6 include fortified cereals, beans, |

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| | red blood cell formation; helps body use fats | 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. Too little folate, is linked to birth defects such as spina bifida and anencephaly. | Many foods are excellent sources of folate—fruits and vegetables, whole grains, beans, breakfast cereals, and fortified grains and grain products. |
| 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 See More Information section

Ref 1 Pompe disease diagnosis and management guideline
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3110959/

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Ref 2 Exercise training alone or in combination with high-protein diet in patients with late onset Pompe disease: results of a cross over study
https://pubmed.ncbi.nlm.nih.gov/32505193/

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