IMPORTANT INFORMATION TO SHARE WITH YOUR LOVED ONES

Pompe disease runs in families[1]

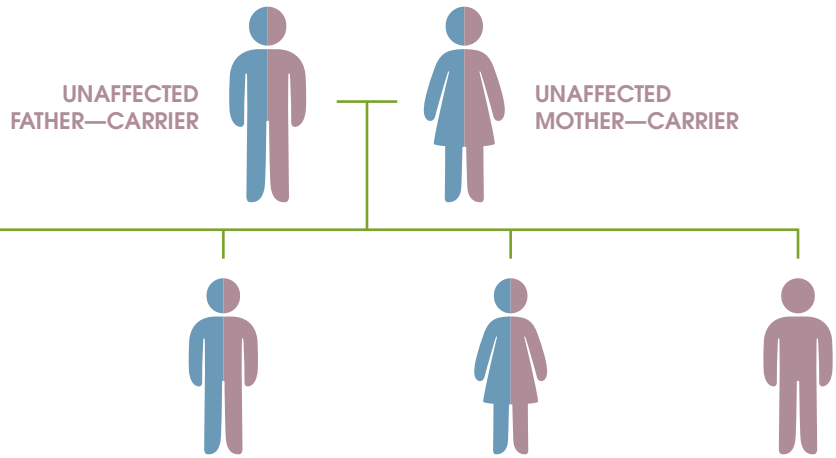
This guide explains why it is important for your brothers and sisters to get tested for Pompe disease, so they can get timely access to care if they need it. Note: Any words that appear in **bold** are defined in the glossary on the next page.

Pompe disease is a rare disease that is caused by differences in a person's **genes**. It is considered an **autosomal recessive** genetic disease, which means that it is acquired when a child inherits two affected (mutated) acid-α glucosidase (**GAA**) genes—one from each parent.[1,2]

Families share the majority of their genes, so it is possible for more than one family member to have Pompe disease, even if there are no signs or symptoms. Family members can also be healthy **carriers** of the disease, which means that they will not feel the effects of Pompe disease, but might pass the **mutation** on to their biological children.[1]

HOW POMPE DISEASE IS PASSED ON FROM PARENTS TO CHILDREN

This graphic shows a scenario in which two healthy individuals without Pompe disease are carriers of the affected GAA gene. These unaffected carriers can pass along the affected gene to their children.[1-3]



UNAFFECTED NON-CARRIER
1 in 4 (25%) chance that the child will be completely unaffected[2]

UNAFFECTED CARRIER
2 in 4 (50%) chance that the child will inherit only one affected gene and become a carrier, but be unaffected by Pompe disease[2]

AFFECTED PERSON
1 in 4 (25%) chance that the child will inherit two affected genes and be affected by Pompe disease[2]

Unaffected gene Affected gene

THE MORE YOU KNOW

Share this guide to help your loved ones understand why it's important to get tested for Pompe disease.

Testing for Pompe disease

HELP YOUR FAMILY TAKE THE NEXT STEP

Even if your brothers or sisters have never shown symptoms of Pompe disease, it is important to know whether they carry the affected GAA gene, which they could pass on to their own children. There are effective ways to manage Pompe disease, but early diagnosis is important to ensure timely attention to address the symptoms.[2,4,5]

There are several ways to check for Pompe disease; for example, healthcare professionals can use a **dried blood spot test**. This simple method only requires a few drops of blood for the laboratory to see any changes in GAA enzyme function that are related to Pompe disease. The results of this test can help healthcare professionals know what steps to take next.[2,6]

Healthcare professionals are the first and best source of information about Pompe disease. You and your family can also learn more by visiting websites that offer information, support, and resources for people living with Pompe disease. These include:

Australian Pompe Association (APA)

International Pompe Association (IPA)

Rare Voices Australia

GLOSSARY

Autosomal recessive: A condition that is present only when two copies of an affected gene are passed from parents to a child.[7]

Carrier: A person whose inherited genetic material contains one affected gene. This person is a healthy carrier of the gene and might pass that affected gene to his/her children.[8]

Dried blood spot test: A simple laboratory test that requires only a few drops of blood and can be used to help identify changes in the function of the GAA enzyme and/or genetic differences related to Pompe disease.[6,9]

GAA: Acid-α glucosidase—a critical enzyme that helps the body break down a type of sugar called glycogen.[1]

Gene: Instructions made of DNA sequences that tell the body how to grow and work. Each person has over 20,000 genes, and inherits a complete set of genes from each parent.[10]

Mutation: Difference in a gene that can affect how its specific job is done—these can alter the instructions in a gene and lead to problems in different parts of the body.[11]

THE MORE YOU CAN DO

Talk to your healthcare professional about getting your brothers and sisters tested for Pompe disease.

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A rare commitment to the Pompe community.

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