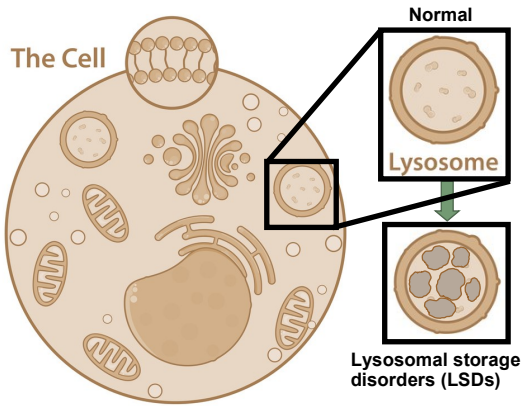
Kansas Department of Health and Environment

Patient Information
Pompe / MPS I

What are Lysosomal Storage Disorders?

Lysosomal Storage Disorders or LSDs, are a group of inherited metabolic disorders in which cells are unable to break down sugars and fats due to a lack of a specific enzyme or an improperly working enzyme. The type of LSD is determined by the type of enzyme that is missing or not working properly. When cells can't break down sugars and fats, these sugars or fats build up inside the cells, causing them to swell. Left untreated, this build up causes tissue damage, and can result in a variety of symptoms. These symptoms are not usually visible at birth, but newborn screening for these conditions helps to identify them before the condition can cause permanent disability.



What is Pompe?

Pompe is an LSD in which a gene mutation causes the body to not produce any or enough alpha-glucosidase enzyme. This causes a build up of a sugar called glycogen in the body's tissues, particularly muscle tissues, including the heart. The severity of symptoms depends on when symptoms begin. Symptoms can include:

- Delayed physical milestones
- Enlarged heart
- Feeding difficulties
- Muscle weakness
- Difficulty breathing
- Failure to thrive

What is Mucopolysaccharidoses I (MPS I or Hurler Syndrome)?

MPS I is an LSD in which a gene mutation causes the body to not produce any or enough alpha-L-iduronidase enzyme. This causes a build of a complex sugar called glycosaminoglycan in the body's tissues, particularly in the bones, joints, brain, liver, spleen or heart. The severity of symptoms depends on when symptoms begin. Symptoms include:

- Large head
- "Coarse" facial features
- Physical & cognitive delays
- Swollen abdomen
- Hearing loss
- Clouding of the eyes

Where can I find more information about these disorders?

These websites have a variety of information about the different types of LSD.

pompediseasenews.com
unitedpompe.com

babysfirsttest.org

mpssociety.org
mps1disease.com

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What does it mean if my baby has an abnormal screen for an LSD?

Note: Screening is not the same as diagnostic testing. A diagnostic test can help determine if your baby is affected by a disorder or just an active carrier.

An abnormal screen means that your baby's newborn screening specimen showed a decreased enzyme activity AND genetic markers for either Pompe or Mucopolysaccharidoses I (MPS I).

These results mean one of three things:

1. Your baby may be a carrier for one of these disorders.
2. Your baby may be diagnosed with one of these disorders.
3. You baby's result may be a false positive.*

Additional testing is needed to confirm the presence of the disorder or carrier status. Your baby's doctor will refer you to a specialist who will complete the additional testing.

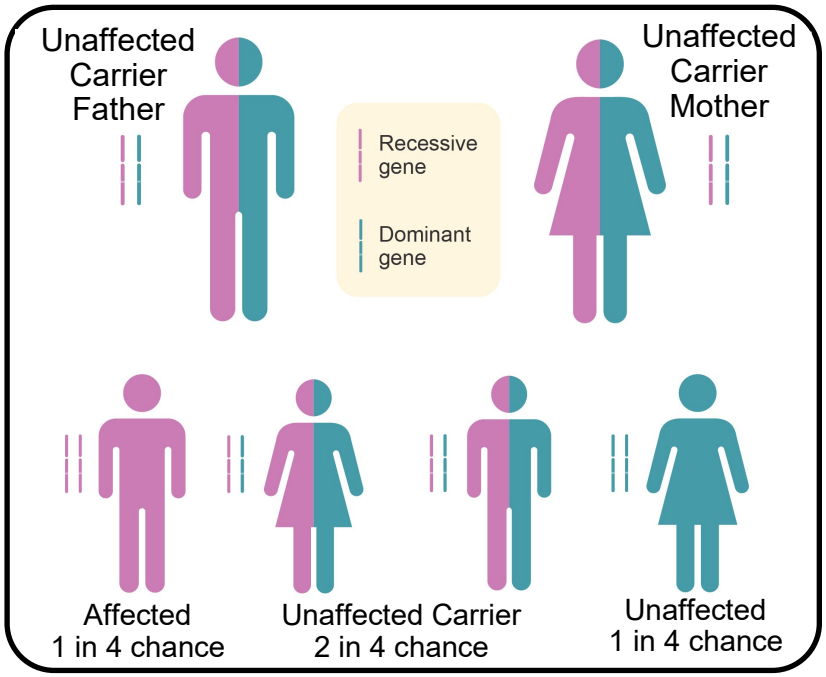
When the diagnostic test shows that the child is neither a carrier or affected, this is considered a false positive.

How are Pompe and MPS I treated?

Currently there is no cure for Pompe or MPS I, but there are treatment options. This is done through a multi-disciplinary approach that can include Enzyme Replacement Therapy (ERT), physical therapy, occupational therapy, and sometimes dietary modifications. These treatment options can helps prevent or reduce the symptoms of the disorder, leading to a better quality of life and longer life span.

How does someone get Pompe or MPS I?

Pompe and MPS I have an autosomal recessive inheritance pattern, meaning that both parents are unaffected carriers of the gene mutation. If both parents are carriers of the recessive gene, there is a 25% chance that their child will be affected.



Got Questions?

Visit us online at

kdheks.gov/newborn_screening/

or call 785-296-3363.

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