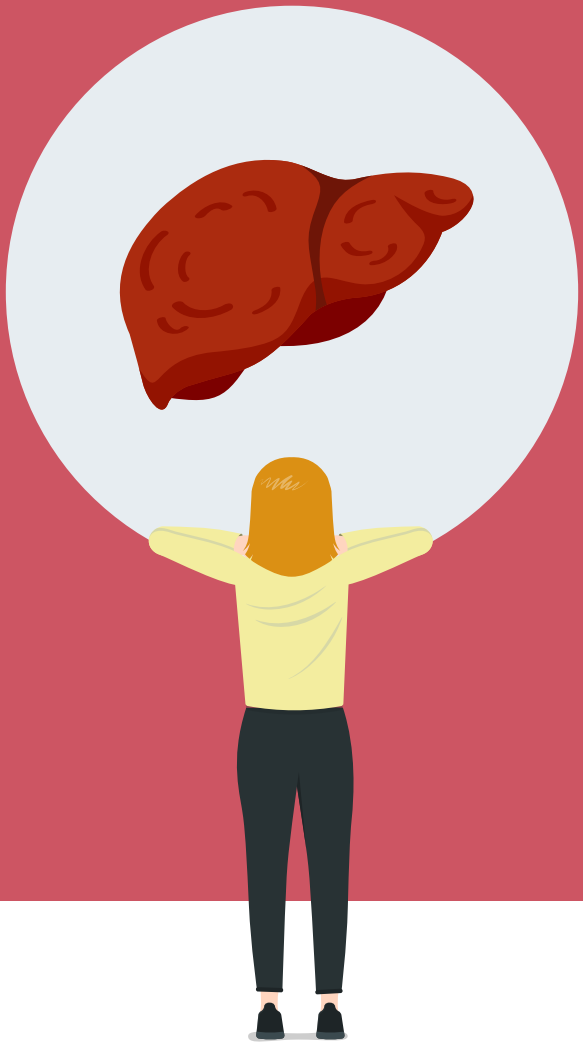
Uncover a Deeper Understanding of GSDIa

Knowledge is power, and staying informed and educated about glycogen storage disease type Ia (GSDIa) is essential.



Understanding GSDIa

What Is GSDIa?

Understand Glycogen Storage Diseases

Glycogen storage diseases (GSDs) are a group of rare genetic diseases that affect how the body stores glycogen and regulates blood glucose levels. Glycogen is the stored form of glucose (sugar), which is the body's main source of energy.

Glycogen storage disease type Ia (GSDIa) is one of the most common and severe types of GSD. It occurs when variants, or changes, in the *G6PC* gene are passed down from both parents. This causes a malfunction in the enzyme that is involved in both breaking down glycogen into glucose and the creation of new glucose.

This leads to potentially life-threatening hypoglycemia (low blood glucose levels) and a range of complications that affect many parts of the body.

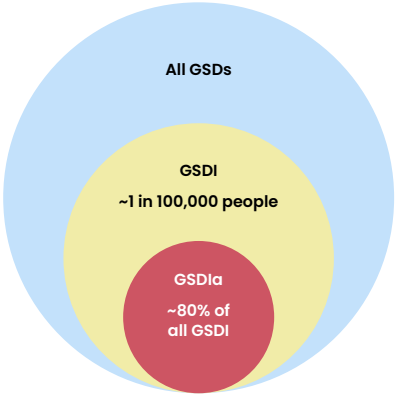


Image illustrative of disease types within the larger GSD hierarchy. Not to scale based on disease prevalence.

Spot the Possible Signs and Symptoms

Hypoglycemia is often one of the first noticeable signs of GSDIa. In many cases, symptoms may not appear until around the time that feedings for infants become more spaced out, when they start sleeping through the night, or if they have an illness lasting several days. In infants, hypoglycemia can cause feeding difficulties, irritability, seizures, or even death in severe cases. Other common signs of GSDIa in children include:

Distended abdomen

Developmental delays

Doll-like appearance (round face, full cheeks)

How Is GSDIa Diagnosed?

GSDIa is usually diagnosed in infancy or early childhood, once signs of hypoglycemia become apparent, usually around 3 to 6 months after birth. Signs and symptoms of GSDIa may not be the same for every individual, which can lead to challenges in diagnosing the disease properly. Diagnosis is typically confirmed by a test of the *G6PC* gene.

What Causes GSDIa?

Examine the Metabolism of GSDIa

The body naturally converts food into glucose to use for energy. Usually, when there is an excess of glucose, it gets stored as glycogen in the body.

Glycogen can be thought of as a sort of energy storage locker. When the body requires extra energy, such as between meals and during times of activity, glycogen gets broken down into glucose and released into the body.

Because of the non-working *G6PC* gene, individuals with GSDIa cannot properly break down glycogen into glucose and release it into the bloodstream for energy.

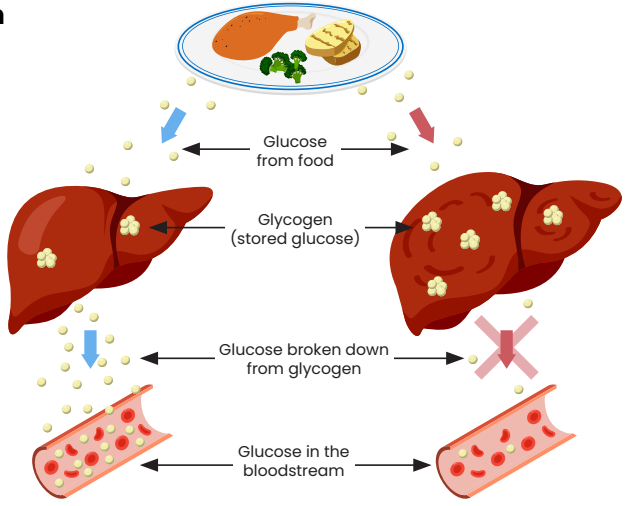
Typical Metabolism

Food is converted into glucose, which is used for energy throughout the body.

Excess glucose is stored as glycogen in the liver and other organs.

Glycogen in the liver is broken down into smaller glucose molecules.

Glucose is released into the bloodstream, providing energy wherever it is needed.



GSDIa Metabolism

Food is converted into glucose, which is used for energy throughout the body.

Excess glucose is stored as glycogen in the liver and other organs.

The body is unable to break down glycogen or to create new glucose molecules due to the non-working *G6PC* gene.

Glycogen builds up in the liver and glucose cannot be released into the bloodstream, causing acute and potentially life-threatening hypoglycemia.

Due to the fluctuations in blood glucose levels and the buildup of glycogen in the liver, individuals with GSDIa can experience long-term complications that negatively affect many parts of the body.

Managing GSDIa

Explore the Goals of GSDIa Management

Controlling glucose can be a full-time activity when managing GSDIa, but it's important to look beyond blood glucose levels and take a holistic approach to your health.

- Support glycemic and metabolic control (stabilize blood glucose levels)
- Limit disease signs and symptoms
- Lessen the impact of long-term complications
- Improve quality of life

Nutrition

Individuals with GSDIa must follow a strict medically prescribed nutritional plan as part of their overall management approach. GSDIa nutritional plans are complex and unique to the individual's needs and disease challenges. The chart below is an example of a typical nutritional plan that may be appropriate for managing GSDIa:

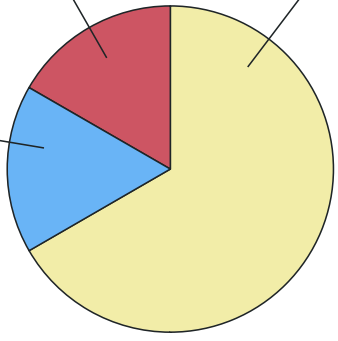
Typical GSDIa Nutritional Plan*

Fats

Generally avoid trans-fatty acids and saturated fats.

Protein

Most lean meat, poultry, and fish are allowed; generally avoid fatty and processed meats.



Carbohydrates

Generally avoid fruit and starchy vegetables, as well as grains, cereals, or breads sweetened with sugar, fructose, or sorbitol. Generally limit or avoid dairy.

Uncooked cornstarch is a slow-release carbohydrate that helps to keep blood glucose levels stable overnight and between meals. It is typically a sizable portion of total daily carbohydrate intake.

*The Typical GSDIa Nutritional Plan chart is for illustrative purposes only. A nutritional plan is part of a comprehensive approach to long-term management. Individuals should always talk with a doctor or dietitian about a nutritional plan that helps with their specific needs.

Managing GSDIa (cont'd)

Vitamins and Supplements

Because the GSDIa nutritional plan is restrictive, certain deficiencies are common. Supplements can lessen the impact of complications from insufficient nutrients in the body. Some common vitamins and supplements recommended to help manage complications caused by GSDIa include a multivitamin, vitamin D, and calcium.

Health Monitoring

Lifelong monitoring is needed to make sure that a GSDIa nutritional plan is working to control levels of blood glucose and other metabolic markers and to check for signs of long-term complications.

Blood glucose levels are checked multiple times a day, typically by using methods such as:

Finger-prick testing

Continuous glucose monitoring

GSDIa affects many parts of the body, and working with specialists can help support various needs as they arise. It is important to work with your care team to develop a management plan that suits your unique needs and health goals.

GSDIa Over Time

Uncover the Challenges of Management

Nutritional management is essential for individuals with GSDIa, but there are additional challenges to keep in mind:

Malnutrition is common due to the restrictive nature of typical GSDIa nutritional plans, potentially affecting growth and development.

Strict nutritional management can be difficult to follow, often causing sleep deprivation and anxiety.

Hyperglycemia can occur with overmanagement, leading to other problems, such as headaches, blurred vision, or fatigue.

Understand the Complications of GSDIa

GSDIa can cause complications throughout the body and lead to significant damage over time. When thinking about complications related to GSDIa, it's helpful to break them into 2 categories: immediate and long term.

Immediate Complications

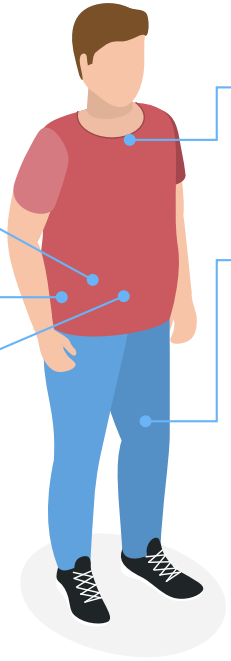
Hypoglycemia is the most immediate complication of GSDIa, and can lead to tremors, lightheadedness, seizures, and coma.

Other metabolic complications caused by glycogen buildup can include elevated levels of cholesterol, triglycerides, lactic acid, and uric acid.

GSDIa Over Time (cont'd)

Long-Term Complications

Over time, hypoglycemia and metabolic instability compound, contributing to long-term complications in many parts of the body:



Liver

- Hepatomegaly (enlarged liver)
- Hepatocellular adenomas (liver lesions)

Kidneys

- Nephromegaly (enlarged kidneys)
- Reduced kidney function

Intestines

- Bloating and diarrhea

Growth and Development

- Delayed growth
- Short stature

Bones

- Osteoporosis (weakened bones)

Beyond Physical Complications

GSDIa can also greatly impact the social, emotional, and mental well-being of both individuals with GSDIa and their caregivers.

Each individual and family will have their own unique experience with GSDIa. By working together with your doctor and dietitian, you can build a customized plan to help manage the disease and address your specific health goals.

Understanding GSDIa

Glossary of Common GSDIa Terms

Enzyme	A protein in the body that helps to speed up chemical reactions and is essential to keep them working properly
Glucose	The sugar that the body uses for energy
Glycemic	The level of glucose (or sugar) in one's blood
Glycogen	A form of glucose that is stored in the body and broken down to be used for energy when it's needed
Hypoglycemia	A condition that occurs when there is too little glucose in the bloodstream
Hyperglycemia	A condition that occurs when there is too much glucose in the bloodstream
Metabolic	Refers to anything related to the chemical changes that take place inside of cells when the body creates energy or other building blocks of life

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MRCP-DTX401-00037 09/2025