# SLC2A9 Gene

Subjects: Genetics & Heredity
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solute carrier family 2 member 9

genes

## 1. Normal Function

The *SLC2A9* gene provides instructions for making a protein called glucose transporter 9 (GLUT9). This protein is found mainly in the kidneys, specifically in structures called proximal tubules. These structures help to reabsorb needed nutrients, water, and other materials into the blood and excrete unneeded substances into the urine. Within the proximal tubules, the GLUT9 protein helps reabsorb or excrete a substance called urate. Urate is a byproduct of certain normal biochemical reactions in the body. In the bloodstream it acts as an antioxidant, protecting cells from the damaging effects of unstable molecules called free radicals. When more urate is needed in the body, the GLUT9 protein helps reabsorb it into the bloodstream. Most urate that is filtered through the kidneys is reabsorbed into the bloodstream; about 10 percent is released into urine.

The GLUT9 protein also plays a role in reabsorbing and excreting the simple sugar glucose.

## 2. Health Conditions Related to Genetic Changes

### 2.1. Renal hypouricemia

At least 17 mutations in the *SLC2A9* gene have been found to cause renal hypouricemia. This condition results in a reduced amount of urate in the blood. Renal hypouricemia often does not cause any health problems but can lead to kidney stones, blood in the urine (hematuria), or pain and nausea after exercise. Most of the mutations that cause renal hypouricemia replace single protein building blocks (amino acids) in the GLUT9 protein and severely reduce or eliminate the protein's ability to reabsorb urate into the bloodstream. As a result, an excessive amount of urate is lost through the urine. While it is not clear how these changes in urate levels lead to the signs and symptoms of renal hypouricemia, it is likely that the loss of urate's antioxidant properties in combination with the increase in urate passing through the kidneys to be released in the urine contribute to the characteristic features of this condition.

### 2.2. Gout

Genetic changes in the *SLC2A9* gene are associated with a condition called gout, which is a form of arthritis. Gout develops when elevated urate levels in the blood (hyperuricemia) lead to the formation of urate crystals in joints, triggering an inflammatory response from the immune system.

*SLC2A9* gene changes associated with gout likely increase the production of a form of the GLUT9 protein that is 28 amino acids shorter than the full-length version. This shorter version of GLUT9 reabsorbs urate into the bloodstream more readily than the full length version. An increase in the short GLUT9 protein raises the levels of urate in the blood and reduces its release into the urine. As a result, urate can accumulate in the body's joints in the form of crystals, leading to painful arthritis.

While changes in the *SLC2A9* gene can alter urate levels in the body, they are likely not enough to cause gout by themselves. A combination of dietary, genetic, and other environmental factors play a part in determining the risk of developing this complex disorder.

## 3. Other Names for This Gene

- glucose transporter type 9
- GLUT-9
- GLUT9
- GLUTX
- human glucose transporter-like protein-9
- solute carrier family 2 (facilitated glucose transporter), member 9
- solute carrier family 2, facilitated glucose transporter member 9
- UAQTL2
- urate voltage-driven efflux transporter 1
- URATv1

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