SLCO1B1 Gene

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solute carrier organic anion transporter family member 1B1

genes

1. Normal Function

The *SLCO1B1* gene provides instructions for making a protein called organic anion transporting polypeptide 1B1, or OATP1B1. This protein is found in liver cells; it transports compounds from the blood into the liver so that they can be cleared from the body. For example, the OATP1B1 protein transports bilirubin, which is a yellowish substance that is produced when red blood cells are broken down. In the liver, bilirubin is dissolved in a digestive fluid called bile and then excreted from the body. The OATP1B1 protein also transports certain hormones, toxins, and drugs into the liver for removal from the body. Drugs transported by the OATP1B1 protein include statins, which are used to treat high cholesterol; heart disease medications; certain antibiotics; and some drugs used for the treatment of cancer.

2. Health Conditions Related to Genetic Changes

2.1. Rotor syndrome

Mutations in the *SLCO1B1* gene are involved in Rotor syndrome. This condition is characterized by elevated levels of bilirubin in the blood that can cause yellowing of the skin and whites of the eyes (jaundice). For this condition to occur, individuals must have mutations in the *SLCO1B1* gene and a related gene called *SLCO1B3*. This related gene provides instructions for making a protein called OATP1B3, which has a similar transport function to OATP1B1. In some cases, the condition is caused by a deletion of genetic material that removes parts of both the *SLCO1B1* and *SLCO1B3* genes, so no functional OATP1B1 or OATP1B3 protein is made. Most mutations that cause Rotor syndrome lead to abnormally short, nonfunctioning OATP1B1 and OATP1B3 proteins. Without the function of either transport protein, bilirubin is less efficiently taken up by the liver and cleared from the body. The buildup of this substance leads to jaundice in people with Rotor syndrome.

2.2. Other disorders

Certain common variations (polymorphisms) in the *SLCO1B1* gene are associated with a reduced ability to process certain drugs, including statins. The most widely studied polymorphism associated with this abnormality, which is

found in approximately 15 percent of the population, changes a single protein building block in the OATP1B1 protein: the amino acid valine at position 174 is replaced with the amino acid alanine (written as V174A or *SLCO1B1**5). The protein produced from this version of the *SLCO1B1* gene is less able to transport compounds into the liver, which leads to elevated levels of the compounds in the body.

When statins are not efficiently transported into the liver, they build up in the body, and can cause a condition known as statin-induced myopathy. This condition causes fatigue, pain, tenderness, weakness, and cramping in muscles. People with the V174A polymorphism who take statin drugs have an increased risk of developing statin-induced myopathy.

3. Other Names for This Gene

- HBLRR
- liver-specific organic anion transporter 1
- LST-1
- LST1
- OATP-2
- OATP-C
- OATP1B1
- OATP2
- OATPC
- SLC21A6
- SO1B1_HUMAN
- sodium-independent organic anion-transporting polypeptide 2
- solute carrier family 21 (organic anion transporter), member 6
- solute carrier family 21 member 6
- solute carrier organic anion transporter family, member 1B1

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