TSC2 Gene

Subjects: Genetics & Heredity
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TSC complex subunit 2

genes

1. Normal Function

The *TSC2* gene provides instructions for producing a protein called tuberin, whose function is not fully understood. Within cells, tuberin interacts with a protein called hamartin, which is produced from the *TSC1* gene. These two proteins help control cell growth and size. Proteins that normally prevent cells from growing and dividing too fast or in an uncontrolled way are known as tumor suppressors. Hamartin and tuberin carry out their tumor suppressor function by interacting with and regulating a wide variety of other proteins.

2. Health Conditions Related to Genetic Changes

2.1. Lymphangioleiomyomatosis

Mutations in the *TSC2* gene cause most cases of a disorder called lymphangioleiomyomatosis (LAM). This destructive lung disease is characterized by the abnormal overgrowth of smooth muscle-like tissue in the lungs. It occurs almost exclusively in women, causing coughing, shortness of breath, chest pain, and lung collapse.

LAM can occur alone (isolated or sporadic LAM) or in combination with a condition called tuberous sclerosis complex (described below). Researchers suggest that sporadic LAM is caused by a random mutation in the *TSC2* gene that occurs very early in development. As a result, some of the body's cells have a normal version of the gene, while others have the mutated version. This situation is called mosaicism. When a mutation occurs in the other copy of the *TSC2* gene in certain cells during a woman's lifetime (a somatic mutation), she may develop LAM.

2.2. Tuberous sclerosis complex

More than 1,100 mutations in the *TSC2* gene have been identified in individuals with tuberous sclerosis complex, a condition characterized by developmental problems and the growth of noncancerous tumors in many parts of the body. Most of these mutations insert or delete a small number of DNA building blocks (base pairs) in the *TSC2* gene. Other mutations change a single base pair in the *TSC2* gene or create a premature stop signal in the instructions for making tuberin.

People with *TSC2*-related tuberous sclerosis complex are born with one mutated copy of the *TSC2* gene in each cell. This mutation prevents the cell from making functional tuberin from that copy of the gene. However, enough tuberin is usually produced from the other, normal copy of the *TSC2* gene to regulate cell growth effectively. For some types of tumors to develop, a second mutation involving the other copy of the gene must occur in certain cells during a person's lifetime.

When both copies of the *TSC2* gene are mutated in a particular cell, that cell cannot produce any functional tuberin. The loss of this protein allows the cell to grow and divide in an uncontrolled way to form a tumor. A shortage of tuberin also interferes with the normal development of certain cells. In people with *TSC2*-related tuberous sclerosis complex, a second *TSC2* gene mutation typically occurs in multiple cells over an affected person's lifetime. The loss of tuberin in different types of cells disrupts normal development and leads to the growth of tumors in many different organs and tissues.

3. Other Names for This Gene

- PPP1R160
- TSC2_HUMAN
- tuberin
- tuberous sclerosis 2

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