TET2 Gene

Subjects: Genetics & Heredity
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Tet methylcytosine dioxygenase 2: The TET2 gene provides instructions for making a protein whose function is unknown.

genes

1. Normal Function

The *TET2* gene provides instructions for making a protein whose function is unknown. Based on the function of similar proteins, researchers believe the TET2 protein is involved in regulating the process of transcription, which is the first step in protein production. Although this protein is found throughout the body, it may play a particularly important role in the production of blood cells from hematopoietic stem cells. These stem cells are located within the bone marrow and have the potential to develop into red blood cells, white blood cells, and platelets. The TET2 protein appears to act as a tumor suppressor, which is a protein that prevents cells from growing and dividing in an uncontrolled way.

2. Health Conditions Related to Genetic Changes

2.1. Essential thrombocythemia

Some gene mutations are acquired during a person's lifetime and are present only in certain cells. These changes, which are called somatic mutations, are not inherited. Somatic mutations in the *TET2* gene have been identified in a small number of people with essential thrombocythemia, which is a condition characterized by high numbers of platelets in the blood. Platelets are the blood cells involved in blood clotting.

TET2 gene mutations alter the TET2 protein in different ways; however, all of them appear to result in a nonfunctional protein. The role these mutations play in the development of essential thrombocythemia is unknown.

2.2. Polycythemia vera

Somatic mutations in the *TET2* gene are associated with polycythemia vera, a disorder characterized by uncontrolled blood cell production. These mutations are thought to result in a nonfunctional protein. Mutations in this gene have been found in approximately 16 percent of people with polycythemia vera. It is unclear what role these mutations play in the development of polycythemia vera.

2.3. Primary myelofibrosis

Somatic mutations in the *TET2* gene are associated with primary myelofibrosis. This condition is characterized by scar tissue (fibrosis) in the bone marrow, the tissue that produces blood cells. It is unclear what role the *TET2* gene mutations play in the development of primary myelofibrosis.

2.4. Other disorders

Somatic *TET2* gene mutations are also associated with certain types of cancer of blood-forming cells (leukemia) and a disease of the blood and bone marrow called myelodysplastic syndrome. These mutations are thought to result in a nonfunctional TET2 protein. A loss of TET2 protein in hematopoietic stem cells may lead to uncontrolled growth and division of these cells. Researchers are working to determine exactly what role *TET2* gene mutations play in the development of bone marrow disorders.

Systemic mastocytosis

3. Other Names for This Gene

- FLJ20032
- KIAA1546
- MGC125715
- probable methylcytosine dioxygenase TET2
- probable methylcytosine dioxygenase TET2 isoform a
- probable methylcytosine dioxygenase TET2 isoform b
- tet oncogene family member 2
- TET2_HUMAN

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