TMEM127 Gene

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Transmembrane protein 127 (TMEM 127): Mutations in the TMEM127 gene increase the risk of developing a noncancerous tumor associated with the nervous system called paraganglioma or pheochromocytoma (a type of paraganglioma).

genes

1. Normal Function

The *TMEM127* gene provides instructions for making a protein that acts as a tumor suppressor protein, which means it prevents cells from growing and dividing too quickly or in an uncontrolled way. The TMEM127 protein controls a signaling pathway that leads to cell growth and survival. Research shows that this pathway, regulated by a protein complex called mTORC1, is blocked (inhibited) by the TMEM127 protein, although the specific action of the TMEM127 protein is unknown.

2. Health Conditions Related to Genetic Changes

2.1. Nonsyndromic paraganglioma

Mutations in the TMEM127 gene increase the risk of developing a noncancerous tumor associated with the nervous system called paraganglioma or pheochromocytoma (a type of paraganglioma). *TMEM127* gene mutations occur most commonly in people with pheochromocytoma, and they are rarely found in people with other paraganglioma. Specifically, *TMEM127* gene mutations are associated with nonsyndromic paraganglioma or pheochromocytoma, which means the tumors occur without additional features of an inherited syndrome. At least 19 *TMEM127* gene mutations have been identified in people with one of these tumors. A *TMEM127* gene mutation increases the risk of tumor formation. The *TMEM127* gene mutations associated with paraganglioma or pheochromocytoma change single protein building blocks (amino acids) in the TMEM127 protein sequence or result in a shortened protein.

Most people with *TMEM127*-related paraganglioma or pheochromocytoma acquire an additional mutation that deletes the normal copy of the gene. This second mutation, called a somatic mutation, is acquired during a person's lifetime and is present only in tumor cells. Together, the two mutations lead to reduced or absent TMEM127 protein. As a result, the cell growth pathway controlled by the TMEM127 protein is abnormally active, leading to tumor formation.

3. Other Names for This Gene

- FLJ20507
- FLJ22257
- TM127_HUMAN

References

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