RIT1 gene

Ras like without CAAX 1

Normal Function

The *RIT1* gene provides instructions for making a protein that helps cells survive during periods of cellular stress, such as unusually high energy demands. As part of a signaling pathway known as the RAS/MAPK pathway, the RIT1 protein relays signals from outside the cell to the cell's nucleus. These signals instruct the cell to grow and divide (proliferate) or to mature and take on specialized functions (differentiate). The RIT1 protein is a GTPase, which means it converts a molecule called GTP into another molecule called GDP. To transmit signals during periods of cellular stress, the RIT1 protein is turned on by attaching (binding) to a molecule of GTP. The RIT1 protein is turned off (inactivated) when it converts the GTP to GDP. When the protein is bound to GDP, it does not relay signals to the cell's nucleus.

The *RIT1* gene belongs to a class of genes known as oncogenes. When mutated, oncogenes have the potential to cause normal cells to become cancerous. The *RIT1* gene is in the Ras family of oncogenes, which also includes three other genes: *KRAS*, *HRAS*, and *NRAS*. These proteins play important roles in cell division, cell differentiation, and the self-destruction of cells (apoptosis).

Health Conditions Related to Genetic Changes

Noonan syndrome

At least 14 mutations in the *RIT1* gene have been found to cause Noonan syndrome. This condition is characterized by mildly unusual facial characteristics, short stature, heart defects, bleeding problems, skeletal malformations, and many other signs and symptoms. People with Noonan syndrome caused by *RIT1* gene mutations often have swelling caused by a buildup of fluid (lymphedema). Rarely, people with Noonan syndrome caused by a *RIT1* gene mutation develop cancer, including a blood cancer called acute lymphoblastic leukemia.

The *RIT1* gene mutations associated with Noonan syndrome change single protein building blocks (amino acids) in the RIT1 protein. The mutations lead to the production of an altered RIT1 protein that is either continuously turned on (active) or has prolonged activation, rather than promptly switching on and off in response to other cellular proteins. The abnormally active protein alters normal RAS/MAPK signaling and leads to

abnormal cell proliferation, which disrupts the development of organs and tissues throughout the body, resulting in the signs and symptoms of Noonan syndrome.

Lung cancer

MedlinePlus Genetics provides information about Lung cancer

Cancers

Mutations in the *RIT1* gene have been found in several different types of blood cell cancer (leukemia), including acute myeloid leukemia (AML), chronic myelomonocytic leukemia (CMML), and a bone marrow disease called myelodysplastic syndrome (MDS). Similar to the *RIT1* gene mutations that cause Noonan syndrome (described above), the mutations involved in these cancers likely result in a constitutively active protein. As a result, cell growth is continuously promoted and cells grow and divide uncontrollably leading to cancer formation. Unlike the Noonan syndrome mutations, the *RIT1* gene mutations associated with leukemia are somatic mutations, which means they occur during a person's lifetime and are not inherited.

Other Names for This Gene

- GTP-binding protein Roc1
- MGC125864
- MGC125865
- Ras-like without CAAX 1
- ras-like without CAAX protein 1
- RIBB
- RIT
- ROC1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of RIT1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6016[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RIT1%5BTIAB%5D%29+OR+%28Ras+like+without+CAAX+1%5BTIAB%5D%29%29+OR+%28Ras-like+without+CAAX+1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- RIC-LIKE PROTEIN WITHOUT CAAX MOTIF 1; RIT1 (https://omim.org/entry/609591)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6016)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=RIT1[gene])

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Genomic Location

The *RIT1* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

Last updated May 1, 2016