RETREG1 gene

reticulophagy regulator 1

Normal Function

The *RETREG1* gene provides instructions for making a protein that is involved in a cellular process called autophagy. Cells use this process to recycle worn-out or unnecessary cell parts and break down certain proteins when they are no longer needed. In particular, the RETREG1 protein helps direct autophagy of a cell structure called the endoplasmic reticulum, which is important in protein processing and transport. Autophagy may be a way for the cell to remove parts of the endoplasmic reticulum when they are no longer needed or to break down excess or abnormal proteins that are being processed within the structure.

The RETREG1 protein also appears to be important in the organization of another cell structure called the Golgi apparatus, which is important for distribution of proteins within the cell.

The RETREG1 protein is found in sensory and autonomic nerve cells (neurons). Sensory neurons transmit pain, touch, and temperature sensations. Autonomic neurons help control involuntary functions of the body such as heart rate and blood pressure.

Health Conditions Related to Genetic Changes

Hereditary sensory and autonomic neuropathy type II

Mutations in the *RETREG1* gene are responsible for one type of hereditary sensory and autonomic neuropathy type II (HSAN2) called HSAN2B; at least five mutations have been identified in affected individuals. People with HSAN2B lose the ability to feel pain or sense hot and cold. The *RETREG1* gene mutations may lead to an abnormally short and nonfunctional protein. The resulting lack of functioning RETREG1 protein impairs autophagy of the endoplasmic reticulum and alters the structure of the Golgi apparatus in sensory and autonomic neurons.

Researchers suspect that an inability to break down parts of the endoplasmic reticulum when they are no longer needed, and the subsequent accumulation of these structures and other proteins in cells, leads to cell death. The loss of sensory and autonomic neurons due to impaired autophagy results in the signs and symptoms of HSAN2B. It is unclear what role abnormalities of the Golgi apparatus play in the condition.

Other Names for This Gene

- F134B_HUMAN
- FAM134B
- FAM134B protein
- FAM134B protein isoform 1
- FAM134B protein isoform 2
- family with sequence similarity 134 member B
- family with sequence similarity 134, member B
- FLJ20152
- FLJ22155
- FLJ22179
- JK1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of RETREG1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=54463[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28FAM134B%5BTIAB%5D%29+OR+%28JK1%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- RETICULOPHAGY REGULATOR 1; RETREG1 (https://omim.org/entry/613114)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/54463)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=RETREG1[gene])

References

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Genomic Location

The *RETREG1* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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