Family with Sequence Similarity 134, Member B

Alternative Names
FAM134B
JK1
RETREG1

Record Category
Gene locus

WHO-ICD
N/A to gene loci

Incidence per 100,000 Live Births
N/A to gene loci

OMIM Number
613114

Mode of Inheritance
N/A to gene loci

Gene Map Locus
5p15.1

Description
The FAM134B gene encodes a receptor protein that is anchored to the endoplasmic reticulum. This receptor is responsible for autophagy of the endoplasmic reticulum by binding to the autophagy modifiers LC3 and GABARAP. It is believed that FAM134B may be required for the long-term survival of certain cells, in particular, the nociceptive and autonomic ganglion neurons. This is reinforced by studies of the mouse ortholog gene as *Fam134b* disruption in mice has been shown to lead to the expansion of the endoplasmic reticulum, sensitize cells to stress-induced apoptotic death and result in the degeneration of sensory neurons.

Mutations in the FAM134B gene have been associated with Hereditary Sensory and Autonomic Neuropathy, Type IIB (HSAN2B), a neurologic disorder characterized by an impaired sensory perception of pain, temperature and touch in the distal extremities.

Molecular Genetics
The FAM134B gene is located on the short arm of chromosome 5. It spans a length of 144.2 kb of DNA and its coding sequence is contained within 12 exons. The protein product encoded by this gene has a molecular mass of 54.6 kDa and consists of 497 amino acids. An additional 39 kDa isoform exists due to alternative splicing. Apart from the nervous system, the gene is found to be expressed in the intestine, lung, kidney, muscle and heart. At least 5 different FAM134B gene mutations, including deletions and nonsense variants, have been implicated in Hereditary Sensory and Autonomic Neuropathy, type IIB.

Epidemiology in the Arab World
Saudi Arabia
Monies et al. (2017) described the findings of 1000 diagnostic panels and exomes carried out at a next generation sequencing lab in Saudi Arabia. One patient, a 9-year-old male suffering from behavioral changes, muscle weakness, frequent falls and brisk reflexes was suspected to have HSP. However, whole exome sequencing uncovered a homozygous mutation (c.694+2T>-) in exon 6 of the patient's FAM134B gene, associated with Hereditary Sensory and Autonomic Neuropathy, Type 2B. Given the atypical presentation of the patient, this case helped in the phenotypic expansion of the disorder.

References
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Related CTGA Records

Neuropathy, Hereditary Sensory and Autonomic, Type IIB

External Links

http://www.genecards.org/cgi-bin/carddisp.pl?gene=RETREG1

Contributors

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