KIF1B Gene

Subjects: Genetics & Heredity
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Kinesin family member 1B

genes

1. Introduction

The *KIF1B* gene provides instructions for making a protein called kinesin family member 1B, part of the kinesin family of proteins. These proteins are essential for the transport of materials within cells. Kinesin proteins function like freight trains that transport cargo, and their structure is suited for this cargo-carrying function. One part of the protein, called the motor domain, provides the power to move the protein and its cargo along a track-like system made from structures called microtubules. Another part of the kinesin protein, which varies among members of this protein family, binds to specific materials for transport.

Research suggests that the kinesin family member 1B protein specializes in carrying two types of cargo. In nerve cells (neurons), this protein transports small, sac-like structures called synaptic vesicles, which contain materials necessary for the transmission of nerve impulses. In other cell types, the kinesin family member 1B protein carries energy-producing structures called mitochondria.

In addition to its transport functions, the kinesin family member 1B protein appears to be involved in programmed cell death (apoptosis). Apoptosis is a common process throughout life that helps the body get rid of cells it does not need.

2. Health Conditions Related to Genetic Changes

2.1. Neuroblastoma

Deletion of a region of chromosome 1 containing the *KIF1B* gene, designated 1p36, has been identified in some people with neuroblastoma, a type of cancerous tumor composed of immature nerve cells (neuroblasts). 1p36 deletions are somatic mutations, which means they occur during a person's lifetime and are present only in the cells that become cancerous. In addition, several inherited *KIF1B* gene mutations have been identified in families with a history of neuroblastoma. These mutations change single protein building blocks (amino acids) in the kinesin family member 1B protein. Studies suggest that deletion or mutation of the *KIF1B* gene may disrupt apoptosis,

allowing cells to grow and divide too quickly or in an uncontrolled way. This kind of unregulated cell growth can lead to the formation of tumors.

2.2. Nonsyndromic Paraganglioma

KIF1B gene mutations have been reported in individuals with a type of paraganglioma called pheochromocytoma. Paragangliomas are noncancerous (benign) tumors of the nervous system. Pheochromocytomas specifically affect the adrenal glands, which are small hormone-producing glands located on top of each kidney. These tumors often cause no symptoms, but in some cases they can produce an excess of hormones that cause dangerously high blood pressure. *KIF1B* gene mutations are associated with nonsyndromic pheochromocytoma, which means the tumors occur without additional features of an inherited syndrome.

The *KIF1B* gene mutations identified in nonsyndromic pheochromocytoma change single amino acids in the kinesin family member 1B protein. Studies suggest that the mutations may disrupt apoptosis, allowing cells to grow and divide too quickly or in an uncontrolled way and potentially leading to tumor formation.

3. Other Names for This Gene

- CMT2
- CMT2A
- HMSNII
- KIAA0591
- KIAA1488
- KIF1B_HUMAN
- KLP

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