NCSTN Gene

Subjects: Genetics & Heredity
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nicastrin

genes

1. Introduction

The *NCSTN* gene provides instructions for making a protein called nicastrin. This protein is one part (subunit) of a complex called gamma- (γ-) secretase. Nicastrin plays a critical role in the assembly and stability of this complex.

The γ-secretase complex is located in the membrane that surrounds cells, where it cuts apart (cleaves) many different proteins that span the cell membrane (transmembrane proteins). This cleavage is an important step in several chemical signaling pathways that transmit signals from outside the cell into the nucleus. One of these pathways, known as Notch signaling, is essential for the normal maturation and division of hair follicle cells and other types of skin cells. Notch signaling is also involved in normal immune system function.

2. Health Conditions Related to Genetic Changes

2.1. Hidradenitis suppurativa

At least 11 mutations in the *NCSTN* gene have been found to cause hidradenitis suppurativa, a chronic skin disease characterized by recurrent boil-like lumps (nodules) under the skin that develop in hair follicles. The nodules tend to become inflamed and painful, and they produce significant scarring as they heal.

NCSTN gene mutations reduce the amount of functional nicastrin produced in cells, so less of this protein is available to act as part of the γ-secretase complex. The resulting shortage of normal γ-secretase impairs cell signaling pathways, including Notch signaling. Although little is known about the mechanism, studies suggest that abnormal Notch signaling may promote the development of recurrent nodules in hair follicles and trigger inflammation in the skin.

3. Other Names for This Gene

- anterior pharynx-defective 2

- APH2
- ATAG1874
- KIAA0253
- NICA_HUMAN
- nicastrin precursor
- RP11-517F10.1

References

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