HPS1 Gene

Subjects: Genetics & Heredity
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HPS1, biogenesis of lysosomal organelles complex 3 subunit 1

Keywords: genes

1. introduction

The *HPS1* gene provides instructions for making a protein that forms part of a complex called biogenesis of lysosome-related organelles complex-3 (BLOC-3). This complex plays a role in the formation of a group of cellular structures called lysosome-related organelles (LROs). In particular, BLOC-3 helps turn on the process by which necessary proteins are transported to LROs during their formation. LROs are very similar to compartments within the cell called lysosomes, which digest and recycle materials. However, LROs perform specialized functions and are found only in certain cell types.

Within pigment-producing cells (melanocytes), LROs called melanosomes produce and distribute melanin, which is the substance that gives skin, hair, and eyes their color. A different type of LRO is found in platelets, the blood cells involved in normal blood clotting. These LROs, called dense granules, release chemical signals that cause platelets to stick together and form a blood clot. LROs are also found in other specialized cells, including certain cells of the lungs.

2. Health Conditions Related to Genetic Changes

2.1. Hermansky-Pudlak Syndrome

At least 31 mutations in the *HPS1* gene have been found to cause Hermansky-Pudlak syndrome type 1. Individuals with this form of the condition typically have oculocutaneous albinism, a condition characterized by fair skin, light-colored hair and eyes, and poor vision. They may also have bleeding problems and a severe lung disease called pulmonary fibrosis. The *HPS1* gene mutations that cause Hermansky-Pudlak syndrome type 1 impair the normal function of BLOC-3, disrupting the size, structure, and function of LROs in cells throughout the body. The most common mutation causes a duplication of genetic material within the *HPS1* gene and is found in people from northwest Puerto Rico. Specifically, this mutation results in an extra 16 DNA building blocks (nucleotides) within the gene (written as 1470_1486dup16).

Because the abnormal melanosomes do not distribute melanin properly, people with Hermansky-Pudlak syndrome have unusually light coloring of the skin, hair, and eyes. The absence of dense granules within platelets leads to bleeding problems in affected individuals. Pulmonary fibrosis may also develop due to abnormal LROs in certain lung cells.

3. Other Names for This Gene

- BLOC3S1
- Hermansky-Pudlak syndrome 1
- Hermansky-Pudlak syndrome 1 protein
- Hermansky-Pudlak syndrome 1 protein isoform a
- Hermansky-Pudlak syndrome 1 protein isoform c
- Hermansky-Pudlak syndrome type 1
- HPS
- HPS1_HUMAN

- MGC5277

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