LIPH Gene

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Lipase H

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1. Introduction

The *LIPH* gene provides instructions for making an enzyme called lipase H. This enzyme is found in many cells and tissues, where it breaks down the molecule phosphatidic acid into lysophosphatidic acid (LPA) and free fatty acid. LPA is a ligand, which means that it attaches (binds) to certain proteins called receptors. A ligand and its receptor fit together like a key in a lock. LPA has multiple receptors and is involved in many cellular functions, such as cell growth and division (proliferation), cell movement (migration), and the self-destruction of cells (apoptosis).

One of LPA's receptors, the LPA_6 protein, regulates the proliferation and maturation (differentiation) of cells within hair follicles, which are specialized structures in the skin where hair growth occurs. These cell processes are important for the normal development of hair follicles and for hair growth; as the cells in the hair follicle divide, the hair strand (shaft) is pushed upward and extends beyond the skin, causing the hair to grow. Lipase H is also found in the outermost layer of skin (the epidermis) and glands in the skin that produce a substance that protects the skin and hair (sebaceous glands).

2. Health Conditions Related to Genetic Changes

2.1. Autosomal Recessive Hypotrichosis

More than 15 mutations in the *LIPH* gene have been found to cause autosomal recessive hypotrichosis, a condition that results in sparse hair growth (hypotrichosis) on the scalp and, less frequently, other parts of the body. Some mutations are specific to groups with Pakistani or Japanese ancestry, or in the Mari and Chuvash populations of Russia. *LIPH* gene mutations lead to the production of a lipase H enzyme with little or no function. Without functional lipase H, LPA is not produced. A lack of LPA impairs many cellular functions, including the proliferation and maturation of the cells that make up hair follicles. As a result, hair follicles are structurally abnormal and often underdeveloped. Irregular hair follicles alter the structure and growth of hair shafts, leading to fragile hair that breaks easily. A lack of lipase H function in the epidermis likely contributes to the skin problems sometimes seen in individuals with autosomal recessive hypotrichosis.

2.2. Other Disorders

Mutations in the *LIPH* gene can also cause a hair condition called autosomal recessive woolly hair. People with this condition have hair that is unusually coarse, dry, fine, and tightly curled. Woolly hair typically affects only scalp hair and is present from birth. In some cases, affected individuals develop hypotrichosis as they get older. Certain *LIPH* gene mutations cause autosomal recessive woolly hair in some people and autosomal recessive hypotrichosis (described above) in others, even among members of the same family. Because of a shared genetic cause and overlapping features, it is uncertain whether these two conditions are separate disorders or part of the same disease spectrum.

3. Other Names for This Gene

- lipase member H
- lipase, member H
- LIPH_HUMAN
- LPD lipase-related protein

- LPDLR
- membrane-associated phosphatidic acid-selective phospholipase A1-alpha
- membrane-bound phosphatidic acid-selective phospholipase A1
- mPA-PLA1
- mPA-PLA1 alpha
- phospholipase A1 member B
- PLA1B

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