LPAR6 Gene

Subjects: Genetics & Heredity
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Lysophosphatidic acid receptor 6

Keywords: genes

1. Introduction

The *LPAR6* gene provides instructions for making a protein called lysophosphatidic acid receptor 6 (LPA_6). This protein functions as a receptor. Receptor proteins have particular sites into which certain other proteins, called ligands, fit like keys into locks. A specific fat called lysophosphatidic acid (LPA) is the ligand for the LPA_6 protein. LPA can attach to many receptors, but LPA_6 is the only LPA receptor found in hair follicles. Hair follicles are specialized structures in the skin where hair growth occurs. 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. The LPA_6 protein is also found in the outermost layer of skin (the epidermis). Attachment of LPA to LPA_6 helps regulate the growth and division (proliferation) and maturation (differentiation) of cells in the hair follicle.

2. Health Conditions Related to Genetic Changes

2.1. Autosomal Recessive Hypotrichosis

More than 30 *LPAR6* gene mutations 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 populations with Pakistani ancestry. *LPAR6* gene mutations lead to the production of an abnormal LPA_6 protein that cannot bind to LPA to regulate cell proliferation and differentiation within 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 LPA_6 protein 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 *LPAR6* 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 *LPAR6* 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

- G-protein coupled purinergic receptor P2Y5
- LPA receptor 6
- LPA-6
- LPAR6_HUMAN
- oleoyl-L-alpha-lysophosphatidic acid receptor
- P2RY5

- P2Y purinoceptor 5
- P2Y5
- purinergic receptor 5
- purinergic receptor P2Y G protein-coupled protein 5
- purinergic receptor P2Y, G-protein coupled, 5
- RB intron encoded G-protein coupled receptor

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