ATP8B1 Gene

Subjects: Genetics & Heredity
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ATPase phospholipid transporting 8B1

Keywords: genes

1. Normal Function

The *ATP8B1* gene (also known as *FIC1*) provides instructions for making a protein that is found throughout the body. It is thought to control the distribution of certain fat molecules known as aminophospholipids on the inner surface of liver cell membranes. Based on this role, the ATP8B1 protein is sometimes known as an aminophospholipid translocase. In particular, this protein performs its function in the membranes of liver cells that transport fat-digesting acids called bile acids into bile, and it likely plays a role in maintaining an appropriate balance of bile acids. This process, known as bile acid homeostasis, is critical for the normal secretion of bile and the proper functioning of liver cells.

2. Health Conditions Related to Genetic Changes

2.1. Benign Recurrent Intrahepatic Cholestasis

Mutations in the *ATP8B1* gene can cause benign recurrent intrahepatic cholestasis type 1 (BRIC1). People with BRIC1 have occasional episodes of impaired bile secretion that lead to severe itching (pruritus), and yellowing of the skin and whites of the eyes (jaundice). Most *ATP8B1* gene mutations that cause BRIC1 change single protein building blocks (amino acids) in the ATP8B1 protein. These mutations likely alter the structure or function of the ATP8B1 protein only moderately. Through unknown mechanisms, mutations in the *ATP8B1* gene result in the buildup of bile acids in liver cells, which leads to the signs and symptoms of BRIC1. It is unclear what causes the episodes to begin or end. On occasion, people with BRIC1 have been later diagnosed with a more severe condition called progressive familial intrahepatic cholestasis (described below) when their symptoms worsened.

2.2. Progressive Familial Intrahepatic Cholestasis

More than 50 mutations in the *ATP8B1* gene have been found to cause a severe form of liver disease called progressive familial intrahepatic cholestasis type 1 (PFIC1). Most mutations in the *ATP8B1* gene that cause PFIC1 remove large portions of the gene or lead to an abnormally short protein. These mutations are likely to severely alter the structure or function of the ATP8B1 protein. These mutations cause bile acids to build up in liver cells, damaging these cells and causing liver disease. Although the ATP8B1 protein is found throughout the body, it is unclear how a lack of this protein causes short stature, deafness, diarrhea, and other signs and symptoms of PFIC1.

3. Other Names for This Gene

- AT8B1_HUMAN
- ATPase, aminophospholipid transporter, class I, type 8B, member 1
- BRIC
- FIC1
- PFIC
- PFIC1

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