ABCA3 Gene

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Contributor: Bruce Ren

ATP binding cassette subfamily A member 3

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1. Normal Function

The *ABCA3* gene provides instructions for making a protein involved in surfactant production. Surfactant is a mixture of certain fats (called phospholipids) and proteins that lines the lung tissue and makes breathing easy. Without normal surfactant, the tissue surrounding the air sacs in the lungs (the alveoli) sticks together after exhalation (because of a force called surface tension), causing the alveoli to collapse. As a result, filling the lungs with air on each breath becomes very difficult, and delivery of oxygen to the body is impaired.

The ABCA3 protein is found in the membrane that surrounds lamellar bodies, which are the cellular structures in which the phospholipids and proteins that make up surfactant are packaged. The ABCA3 protein transports phospholipids into the lamellar bodies where they interact with surfactant proteins to form surfactant. The ABCA3 protein also appears to be involved in the formation of normal lamellar bodies. In addition to packaging, lamellar bodies are important for the correct processing of surfactant proteins, which is necessary for the proteins to mature and become functional.

2. Health Conditions Related to Genetic Changes

2.1 Surfactant dysfunction

More than 100 *ABCA3* gene mutations that cause surfactant dysfunction have been identified. Surfactant dysfunction due to mutations in this gene (often called ABCA3 deficiency) can cause severe, often fatal breathing problems in newborns or gradual onset of milder breathing problems in children or adults.

Some mutations in the *ABCA3* gene lead to the production of a protein that is not inserted into the lamellar body membrane. Other mutations lead to the production of an abnormal protein that is found in the lamellar body membrane but has little or no function. Without ABCA3 protein function, the transport of surfactant phospholipids is decreased. In addition, lamellar body formation is impaired, which causes abnormal processing of surfactant proteins. *ABCA3* gene mutations result in abnormal surfactant composition and function. The loss of functional surfactant raises surface tension in the alveoli, causing difficulty breathing and collapse of the lungs. It has been suggested that mutations that eliminate ABCA3 protein function cause severe forms of surfactant dysfunction, and mutations that leave some residual ABCA3 activity cause milder forms of the condition.

2.2 Idiopathic pulmonary fibrosis

MedlinePlus Genetics provides information about Idiopathic pulmonary fibrosis

3. Other Names for This Gene

- ABC transporter 3
- ABC-C
- ABC-C transporter
- ABC3
- ABCA3_HUMAN
- ATP-binding cassette sub-family A member 3
- ATP-binding cassette transporter 3
- ATP-binding cassette, sub-family A (ABC1), member 3

- SMDP3

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