SFTPC Gene

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1. Normal Function

The *SFTPC* gene provides instructions for making a protein called surfactant protein-C (SP-C). This protein is one of four proteins (each produced from a different gene) in surfactant, 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 the delivery of oxygen to the body is impaired. Surfactant lowers surface tension, easing breathing and avoiding lung collapse. The SP-C protein helps spread the surfactant across the surface of the lung tissue, aiding in the surface tension-lowering property of surfactant.

The phospholipids and proteins that make up surfactant are packaged in cellular structures known as lamellar bodies, which are found in specialized lung cells. The surfactant proteins must go through several processing steps to mature and become functional; some of these steps occur in lamellar bodies.

2. Health Conditions Related to Genetic Changes

2.1. Surfactant dysfunction

More than 35 mutations in the *SFTPC* gene have been identified in people with surfactant dysfunction. When this condition is caused by mutations in the *SFTPC* gene (sometimes called SP-C dysfunction), it can cause severe breathing problems in newborns or gradual onset of milder breathing problems in children or adults.

SFTPC gene mutations associated with surfactant dysfunction affect the processing of the SP-C protein. Many of the mutations occur in a particular region of the gene called the BRICHOS domain, which appears to be involved in the processing and cellular placement of the SP-C protein.

Mutations in the *SFTPC* gene result in a reduction or absence of mature SP-C and a buildup of abnormal forms of SP-C. It is unclear which of these outcomes causes the signs and symptoms of SP-C dysfunction. Lack of mature SP-C can lead to abnormal composition of surfactant and decreased surfactant function. The loss of functional surfactant would raise surface tension in the alveoli, causing difficulty breathing and collapse of the lungs. Alternatively, research suggests that abnormally processed SP-C proteins form the wrong three-dimensional shape and accumulate inside lung cells. These misfolded proteins may trigger a cellular response that results in cell damage and death. This damage may disrupt surfactant production and release, leading to the breathing problems associated with surfactant dysfunction.

3. Other Names for This Gene

- BRICD6
- PSP-C
- PSPC_HUMAN
- pulmonary surfactant apoprotein-2 SP-C
- pulmonary surfactant-associated protein C
- pulmonary surfactant-associated proteolipid SPL(Val)
- SFTP2
- SMDP2
- SP-C

- SP5

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