

SFTPB gene

surfactant protein B

Normal Function

The *SFTPB* gene provides instructions for making a protein called surfactant protein-B (SP-B). 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-B 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. The SP-B protein plays a role in the formation of lamellar bodies and, thus, affects the processing of a surfactant protein called surfactant protein-C (SP-C).

Health Conditions Related to Genetic Changes

Surfactant dysfunction

More than 30 mutations in the *SFTPB* gene that cause surfactant dysfunction have been identified. Surfactant dysfunction due to *SFTPB* gene mutations (often called SP-B deficiency) causes severe, often fatal breathing problems in newborns. These mutations lead to partial or complete loss of mature SP-B, resulting in abnormal composition of surfactant and decreased surfactant function. In addition, lamellar body formation is impaired. The lack of normal lamellar bodies leads to abnormal processing of SP-C, resulting in a reduction of mature SP-C and a buildup of unprocessed forms of SP-C. The loss of functional surfactant raises surface tension in the alveoli, causing difficulty breathing and collapse of the lungs. The combination of SP-B and SP-C dysfunction may explain why the signs and symptoms of SP-B deficiency are so severe.

Other Names for This Gene

- 18 kDa pulmonary-surfactant protein
- 6 kDa protein
- PSP-B
- PSPB_HUMAN
- pulmonary surfactant-associated protein B
- pulmonary surfactant-associated proteolipid SPL(Phe)
- SFTP3
- SMDP1
- SP-B

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SFTP3 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6439\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6439[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SFTP3%5BTIAB%5D%29+OR+%28surfactant+protein+B%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- SURFACTANT, PULMONARY-ASSOCIATED PROTEIN B; SFTP3 (<https://omim.org/entry/178640>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6439>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SFTP3\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SFTP3[gene]))

References

- Nogee LM, Garnier G, Dietz HC, Singer L, Murphy AM, deMello DE, Colten HR. Amutation in the surfactant protein B gene responsible for fatal neonatal respiratory disease in multiple kindreds. J Clin Invest. 1994 Apr;93(4):1860-3.doi: 10.1172/JCI117173. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/8163685>) or Free

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- Nogee LM, Wert SE, Proffit SA, Hull WM, Whitsett JA. Allelic heterogeneity in hereditary surfactant protein B (SP-B) deficiency. *Am J Respir Crit Care Med*. 2000 Mar;161(3 Pt 1):973-81. doi: 10.1164/ajrccm.161.3.9903153. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/10712351>)
- Weaver TE. Synthesis, processing and secretion of surfactant proteins B and C. *Biochim Biophys Acta*. 1998 Nov 19;1408(2-3):173-9. doi:10.1016/s0925-4439(98)00066-0. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/9813310>)

Genomic Location

The *SFTPB* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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