



SLC34A2 gene

solute carrier family 34 member 2

Normal Function

The *SLC34A2* gene provides instructions for making a protein called the type IIb sodium-phosphate cotransporter, which plays a role in the regulation of phosphate levels (phosphate homeostasis). Although this protein can be found in several organs and tissues in the body, it is located mainly in the millions of small air sacs (alveoli) in the lungs, specifically in cells called alveolar type II cells. These cells produce and recycle surfactant, which is a mixture of certain phosphate-containing fats (called phospholipids) and proteins that lines the lung tissue and makes breathing easy. The recycling of surfactant releases phosphate into the alveoli. Research suggests that the type IIb sodium-phosphate cotransporter normally helps clear this phosphate.

Health Conditions Related to Genetic Changes

Pulmonary alveolar microlithiasis

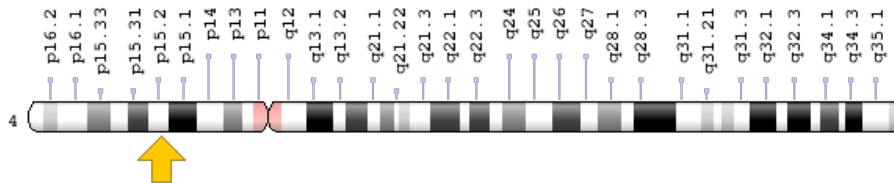
At least 18 *SLC34A2* gene mutations have been identified in people with pulmonary alveolar microlithiasis, a disorder in which many tiny fragments (microliths) of a compound called calcium phosphate gradually accumulate in the alveoli. These deposits eventually cause widespread lung damage (interstitial lung disease) that leads to breathing problems.

SLC34A2 gene mutations are thought to impair the activity of the type IIb sodium-phosphate cotransporter, resulting in the accumulation of phosphate in the alveoli. The accumulated phosphate forms the microliths that cause the signs and symptoms of pulmonary alveolar microlithiasis.

Chromosomal Location

Cytogenetic Location: 4p15.2, which is the short (p) arm of chromosome 4 at position 15.2

Molecular Location: base pairs 25,655,851 to 25,678,748 on chromosome 4 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- NAPI-3B
- NAPI-IIb
- NPTIIb
- sodium-dependent phosphate transport protein 2B
- sodium/phosphate cotransporter 2B
- solute carrier family 34 (sodium phosphate), member 2
- solute carrier family 34 (type II sodium/phosphate cotransporter), member 2
- solute carrier family 34 (type II sodium/phosphate cotransporter), member 2
- type II sodium-dependent phosphate transporter 3b

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Adjacent Cell Types Collaborate in the Alveoli of the Lungs
<https://www.ncbi.nlm.nih.gov/books/NBK26875/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC34A2%5BTIAB%5D%29+OR+%28%28NAPI-3B%5BTIAB%5D%29+OR+%28NAPI-IIb%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+3600+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 34 (SODIUM/PHOSPHATE COTRANSPORTER), MEMBER 2
<http://omim.org/entry/604217>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SLC34A2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLC34A2%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:11020
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:10568>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/10568>
- UniProt
<https://www.uniprot.org/uniprot/O95436>

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