BSND gene

barttin CLCNK type accessory beta subunit

Normal Function

The *BSND* gene provides instructions for making a protein called barttin. This protein is found primarily in the kidneys, where it attaches (binds) to two specific chloride channels: ClC-Ka (produced from the *CLCNKA* gene) and ClC-Kb (produced from the *CLCNKB* gene). The ClC-Ka and ClC-Kb channels transport charged atoms of chlorine (chloride ions) out of kidney cells.

Barttin is essential for the normal placement of ClC-Ka and ClC-Kb channels in the cell membrane. It also regulates the channels' stability and function. The transport of chloride ions is part of the mechanism by which the kidneys reabsorb salt (sodium chloride or NaCl) from the urine back into the bloodstream. The retention of salt affects the body's fluid levels and helps maintain blood pressure.

Barttin, ClC-Ka, and ClC-Kb are also found in the inner ear, where they play a role in normal hearing.

Health Conditions Related to Genetic Changes

Bartter syndrome

More than a dozen mutations in the *BSND* gene have been identified in people with Bartter syndrome type IV. This form of the disorder causes severe or life-threatening health problems that become apparent before or soon after birth. Affected individuals also have hearing loss caused by abnormalities in the inner ear, which is why Bartter syndrome type IV is also known as antenatal Bartter syndrome with sensorineural deafness.

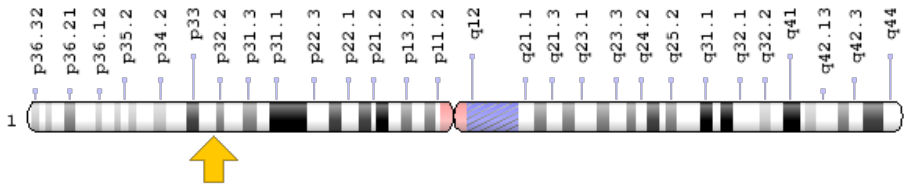
BSND gene mutations impair barttin's ability to regulate the ClC-Ka and ClC-Kb channels. Some mutations keep the channels from ever reaching the cell membrane. Other mutations allow the channels to reach the cell membrane but prevent them from transporting ions properly. As a result, the kidneys cannot reabsorb salt normally and excess salt is lost through the urine (salt wasting). The abnormal salt loss disrupts the normal balance of ions in the body. This imbalance underlies many of the major features of Bartter syndrome, including a failure to grow and gain weight at the expected rate (failure to thrive), dehydration, constipation, and increased urine production (polyuria). A loss of ClC-Ka and ClC-Kb function in the inner ear is responsible for the hearing loss characteristic of Bartter syndrome type IV.

Nonsyndromic hearing loss

Chromosomal Location

Cytogenetic Location: 1p32.3, which is the short (p) arm of chromosome 1 at position 32.3

Molecular Location: base pairs 54,998,944 to 55,008,792 on chromosome 1 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BART
- Bartter syndrome, infantile, with sensorineural deafness (Barttin)
- barttin
- barttin CLCNK-type chloride channel accessory beta subunit
- BSND_HUMAN
- deafness, autosomal recessive 73
- DFNB73

Additional Information & Resources

Educational Resources

- National Institute of Diabetes and Digestive and Kidney Diseases: The Kidneys and How They Work
  https://www.niddk.nih.gov/health-information/kidney-disease/kidneys-how-they-work

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28BSND%5BTIAB%5D%29+OR+%28barttin%5BTIAB%5D%29%29+OR+%28DFNB73%5BTIAB%5D%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

- BSND GENE
  http://omim.org/entry/606412

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_BSND.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=BSND%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:16512
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:7809
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7809
- UniProt
  https://www.uniprot.org/uniprot/Q8WZ55

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