# KCNB1 gene

potassium voltage-gated channel subfamily B member 1

## Normal Function

The *KCNB1* gene belongs to a large family of genes that provide instructions for making potassium channels. These channels, which transport positively charged atoms (ions) of potassium in and out of cells, play a key role in a cell's ability to generate and transmit electrical signals.

The *KCNB1* gene provides instructions for making one part of a potassium channel called Kv2.1. These channels are found primarily in nerve cells (neurons) in the brain where they transport potassium ions out of neurons. The flow of ions through potassium channels in neurons plays a role in regulating the activity of neurons and sending electrical signals in the brain, allowing communication between these cells.

## Health Conditions Related to Genetic Changes

KCNB1 encephalopathy

More than 45 mutations in the *KCNB1* gene have been found to cause a condition called *KCNB1* encephalopathy. This condition is characterized by abnormal brain function (encephalopathy), recurrent seizures (epilepsy), and developmental delay.

Most *KCNB1* gene mutations change single protein building blocks (amino acids), altering the proteins. Other mutations introduce a premature stop signal in the instructions for making the protein, resulting in production of a shortened, nonfunctional protein. All *KCNB1* gene mutations impair Kv2.1 channel function. Channels made with the abnormal proteins are unable to function, which reduces Kv2.1 channel function in the brain. As a result, the channels cannot regulate the flow of potassium ions in neurons, which disrupts normal communication between these cells. Impaired channel function disrupts normal brain development and leads to seizures, intellectual disability, and other features of encephalopathy that occur in this condition.

## Other Names for This Gene

- DEE26
- DRK1
- Kv2.1

- potassium channel, voltage-gated, shab-related subfamily, member 1

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of KCNB1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3745[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=kcnb1+%5Btiab%5D)

Catalog of Genes and Diseases from OMIM

- POTASSIUM CHANNEL, VOLTAGE-GATED, SHAB-RELATED SUBFAMILY, MEMBER 1; KCNB1 (https://omim.org/entry/600397)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/3745)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=KCNB1[gene])

## References


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