

## KCTD1 gene

potassium channel tetramerization domain containing 1

### Normal Function

The *KCTD1* gene provides instructions for making a protein that acts as a transcriptional repressor, which means that it turns off (represses) the activity of certain genes when they are not needed. A region of the KCTD1 protein called the BTB domain is essential for the protein's transcriptional repressor function.

The KCTD1 protein is thought to control (regulate) the activity of genes involved in the development of an embryonic cell layer called the ectoderm. Within the developing embryo, the ectoderm gives rise to several body tissues including the skin, hair, nails, and teeth.

### Health Conditions Related to Genetic Changes

#### Scalp-ear-nipple syndrome

At least 10 mutations in the *KCTD1* gene have been identified in people with scalp-ear-nipple syndrome; as its name suggests, this condition is characterized by abnormalities of the scalp, ears, and nipples. The mutations associated with scalp-ear-nipple syndrome affect the BTB domain of the KCTD1 protein and impair its transcriptional repressor function. This impairment results in abnormal regulation of genes involved in ectodermal development. The altered gene activity disrupts normal development of the tissues that arise from the ectoderm (ectodermal dysplasia) and leads to the signs and symptoms of scalp-ear-nipple syndrome.

### Other Names for This Gene

- C18orf5
- potassium channel tetramerisation domain containing 1
- potassium channel tetramerization domain-containing protein 1
- SENS

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of KCTD1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=284252\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=284252[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28KCTD1%5BTIAB%5D%29+OR+%28potassium+channel+tetramerization+domain+containing+1%5BTIAB%5D%29+OR+%28potassium+channel+tetramerisation+domain+containing+1%5BTIAB%5D%29+OR+%28potassium+channel+tetramerization+domain-containing+protein+1%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%29%29>)

### Catalog of Genes and Diseases from OMIM

- POTASSIUM CHANNEL TETRAMERIZATION DOMAIN-CONTAINING PROTEIN 1; KCTD1 (<https://omim.org/entry/613420>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/284252>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=KCTD1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=KCTD1[gene]))

## References

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## **Genomic Location**

The *KCTD1* gene is found on chromosome 18 (<https://medlineplus.gov/genetics/chromosome/18/>).

**Last updated April 1, 2017**