

## SIL1 gene

SIL1 nucleotide exchange factor

### Normal Function

The *SIL1* gene provides instructions for producing a protein located in a cell structure called the endoplasmic reticulum. Among its many functions, the endoplasmic reticulum folds and modifies newly formed proteins so they have the correct 3-dimensional shape. The SIL1 protein works with BiP, a protein that helps fold newly produced proteins into the proper shape and refold damaged proteins. To start this process, BiP attaches (binds) to a molecule called adenosine triphosphate (ATP). When BiP folds a protein, the ATP is converted to a similar molecule called adenosine diphosphate (ADP). Then the SIL1 protein releases ADP from BiP so that it can bind to another molecule of ATP and start the protein folding process again. Because of its role in helping BiP exchange ADP for ATP, the SIL1 protein is called a nucleotide exchange factor.

### Health Conditions Related to Genetic Changes

#### Marinesco-Sjögren syndrome

More than a dozen mutations in the *SIL1* gene have been found to cause Marinesco-Sjögren syndrome. Most of these mutations result in the production of a protein that has little or no activity. Defective SIL1 protein cannot remove ADP from BiP. BiP is then unable to bind to ATP and re-start the protein folding process. A disruption in protein folding impairs protein transport and causes proteins to accumulate in the endoplasmic reticulum. This accumulation likely damages and destroys cells in many different tissues, leading to poor coordination, muscle weakness, and the other features of Marinesco-Sjögren syndrome.

### Other Names for This Gene

- BAP
- BiP-associated protein
- MSS
- SIL1 homolog, endoplasmic reticulum chaperone (*S. cerevisiae*)
- SIL1 protein
- SIL1\_HUMAN

- ULG5

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of SIL1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=64374\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=64374[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28SIL1%5BALL%5D%29+OR+%28BiP-associated+protein%5BALL%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- SIL1 NUCLEOTIDE EXCHANGE FACTOR; SIL1 (<https://omim.org/entry/608005>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/64374>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SIL1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SIL1[gene]))

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## Genomic Location

The *SIL1* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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