

## FAM111B gene

FAM111 trypsin like peptidase B

### Normal Function

The *FAM111B* gene provides instructions for making a protein whose function is not well understood. The FAM111B protein, which is found in many parts of the body, contains a functional region called a peptidase domain. Similar proteins containing such a domain are able to break down other proteins. However, the types of proteins the FAM111B protein interacts with and the roles it plays in the body are unknown.

### Health Conditions Related to Genetic Changes

Hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis

At least six mutations in the *FAM111B* gene have been identified in people with hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis (POIKTMP). This disorder affects many parts of the body, particularly the skin, muscles, lungs, and pancreas.

The *FAM111B* gene mutations that cause POIKTMP result in production of an abnormal FAM111B protein from one copy of the gene in each cell. Because most of the *FAM111B* mutations identified in people with POIKTMP change single protein building blocks (amino acids) in the peptidase domain, researchers think that the mutations alter the protein's function, and that these changes in FAM111B function underlie the varied signs and symptoms of POIKTMP.

### Other Names for This Gene

- cancer-associated nucleoprotein
- CANP
- POIKTMP
- protein FAM111B isoform a
- protein FAM111B isoform b

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of FAM111B ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=374393\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=374393[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28FAM111B%5BTIAB%5D%29+OR+%28family+with+sequence+similarity+111+member+B%5BTIAB%5D%29%29+OR+%28%28CANP%5BTIAB%5D%29+OR+%28POIKTMP%5BTIAB%5D%29+OR+%28cancer-associated+nucleoprotein%5BTIAB%5D%29+OR+%28protein+FAM111B+isoform+a%5BTIAB%5D%29+OR+%28protein+FAM111B+isoform+b%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

- FAMILY WITH SEQUENCE SIMILARITY 111, MEMBER B; FAM111B (<https://omim.org/entry/615584>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/374393>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=FAM111B\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=FAM111B[gene]))

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## Genomic Location

The *FAM111B* gene is found on chromosome 11 (<https://medlineplus.gov/genetics/chromosome/11/>).

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