FAM111B gene

family with sequence similarity 111 member 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

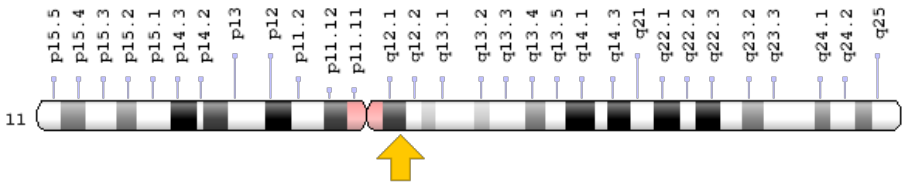
At least six mutations in the *FAM111B* gene have been identified in people with hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis (POIKTMP). This disorder affects many parts of the body, particularly the skin, muscles, lungs, and pancreas.

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.

Chromosomal Location

Cytogenetic Location: 11q12.1, which is the long (q) arm of chromosome 11 at position 12.1

Molecular Location: base pairs 59,107,185 to 59,127,415 on chromosome 11 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

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

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Cells Degrade Proteins by Several Pathways
  https://www.ncbi.nlm.nih.gov/books/NBK21750/#A559

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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
  http://omim.org/entry/615584

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_FAM111B.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=FAM111B%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:24200
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:374393
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/374393
- UniProt
  https://www.uniprot.org/uniprot/Q6SJ93

Sources for This Summary

- OMIM: FAMILY WITH SEQUENCE SIMILARITY 111, MEMBER B
  http://omim.org/entry/615584
- Mercier S, Küry S, Salort-Campana E, Magot A, Agbim U, Besnard T, Bodak N, Bou-Hanna C, Bréhéret F, Brunelle P, Caillon F, Chabrol B, Cormier-Daire V, David A, Eymard B, Faivre L, Figarella-Branger D, Fleurence E, Ganapathi M, Gherardi R, Goldenberg A, Hamel A, Igual J, Irvine AD, Israël-Biet D, Kannengiesser C, Laboisse C, Le Caignec C, Mahé JY, Mallet S, MacGowan S, McAleer MA, McLean I, Méni C, Munnich A, Mussini JM, Nagy PL, Odel J, O'Regan GM, Péréon Y, Perrier J, Piard J, Puzenat E, Sampson JB, Smith F, Soufir N, Tanji K, Thauvin C, Ulane C, Watson RM, Khumalo NP, Mayosi BM, Barbarot S, Bézieau S. Expanding the clinical spectrum of hereditary fibrosing poikiloderma with tendon contractures, myopathy and pulmonary fibrosis due to FAM111B mutations. Orphanet J Rare Dis. 2015 Oct 15;10:135. doi: 10.1186/s13023-015-0352-4.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/26471370
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4608180/
- Mercier S, Küry S, Shaboodien G, Houniet DT, Khumalo NP, Bou-Hanna C, Bodak N, Cormier-Daire V, David A, Faivre L, Figarella-Branger D, Gherardi RK, Glen E, Hamel A, Laboisse C, Le Caignec C, Lindenbaum P, Magot A, Munnich A, Mussini JM, Pillay K, Rahman T, Redon R, Salort-Campana E, Santibanez-Koref M, Thauvin C, Barbarot S, Keavney B, Bézieau S, Mayosi BM. Mutations in FAM111B cause hereditary fibrosing poikiloderma with tendon contracture, myopathy, and pulmonary fibrosis. Am J Hum Genet. 2013 Dec 5;93(6):1100-7. doi: 10.1016/j.ajhg.2013.10.013.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/24268661
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3853004/

- Seo A, Walsh T, Lee MK, Ho PA, Hsu EK, Sidbury R, King MC, Shimamura A. FAM111B Mutation Is Associated With Inherited Exocrine Pancreatic Dysfunction. Pancreas. 2016 Jul;45(6):858-62. doi: 10.1097/MPA.0000000000000529.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/26495788
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4841754/
- Takeichi T, Nanda A, Yang HS, Hsu CK, Lee JY, Al-Ajmi H, Akiyama M, Simpson MA, McGrath JA. Syndromic inherited poikiloderma due to a de novo mutation in FAM111B. Br J Dermatol. 2016 Jul 13. doi: 10.1111/bjd.14845. [Epub ahead of print]
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/27406236

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/FAM111B

Reviewed: February 2017
Published: June 11, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services