

MYBPC1 gene

myosin binding protein C1

Normal Function

The *MYBPC1* gene provides instructions for making one version of a protein called myosin binding protein C. Several versions of myosin binding protein C are produced from different genes; these proteins are found in muscles used for movement (skeletal muscles) and in heart (cardiac) muscle. The version produced from the *MYBPC1* gene, which is known as the slow skeletal isoform, is found primarily in skeletal muscles.

The slow isoform of myosin binding protein C is active during the development of skeletal muscles. Researchers believe that this protein helps regulate the tensing of muscle fibers (muscle contraction). Myosin binding protein C interacts with other muscle proteins, including myosin, actin, and titin. These proteins play essential roles in muscle cell structures called sarcomeres, which generate the mechanical force needed for muscles to contract. Studies suggest that myosin binding protein C contributes to the stability and maintenance of sarcomeres.

Health Conditions Related to Genetic Changes

Distal arthrogryposis type 1

At least one mutation in the *MYBPC1* gene has been found to cause distal arthrogryposis type 1, a disorder characterized by joint deformities (contractures) in the hands and feet. The mutation changes a single protein building block (amino acid) in the slow isoform of myosin binding protein C. Specifically, the mutation replaces the amino acid tryptophan with the amino acid arginine at protein position 236 (written as Trp236Arg or W236R). It is unclear how the defective protein leads to contractures in people with distal arthrogryposis type 1, or why the joint problems are typically limited to the hands and feet. However, researchers speculate that contractures may be related to problems with muscle contraction that limit the movement of joints before birth.

Other Names for This Gene

- C-protein, skeletal muscle slow isoform
- MYBPCC
- MYBPCS

- MYPC1_HUMAN
- skeletal muscle C-protein
- slow MyBP-C

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MYBPC1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4604\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4604[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28MYBPC1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+OR+%28%28myosin+binding+protein+C%5BTIAB%5D%29+AND+%28Slow%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D%29>)

Catalog of Genes and Diseases from OMIM

- MYOSIN-BINDING PROTEIN C, SLOW TYPE; MYBPC1 (<https://omim.org/entry/160794>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4604>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MYBPC1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MYBPC1[gene]))

References

- Ackermann MA, Kontogianni-Konstantopoulos A. Myosin binding protein-C slow:an intricate subfamily of proteins. J Biomed Biotechnol. 2010;2010:652065. doi:10.1155/2010/652065. Epub 2010 Apr 8. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20396395>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2852610/>)
- Gurnett CA, Desruisseau DM, McCall K, Choi R, Meyer ZI, Talerico M, Miller SE, Ju JS, Pestronk A, Connolly AM, Druley TE, Weihi CC, Dobbs MB. Myosin bindingprotein C1: a novel gene for autosomal dominant distal arthrogryposis type 1. HumMol Genet. 2010 Apr 1;19(7):1165-73. doi: 10.1093/hmg/ddp587. Epub 2010 Jan 2. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20045868>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2838534/>)
- Weber FE, Vaughan KT, Reinach FC, Fischman DA. Complete sequence of

humanfast-type and slow-type muscle myosin-binding-protein C (MyBP-C).
Differential expression, conserved domain structure and chromosome assignment.
Eur J Biochem.1993 Sep 1;216(2):661-9. doi: 10.1111/j.1432-1033.1993.tb18186.x.
Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/8375400>)

Genomic Location

The *MYBPC1* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

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