

GEPHE SUMMARY

pot-2 (<a +pot-2+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+pot-2+"#gephebase-summary-title)	Gephebase Gene	GepheID
	GP00001317	
	Entry Status	Main curator
Published	Courtier	

PHENOTYPIC CHANGE

Physiology (<a +physiology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Physiology+"#gephebase-summary-title)		Trait Category		
Telomere length (<a +telomere+length+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Telomere+length+"#gephebase-summary-title)		Trait		
C. elegans		Trait State in Taxon A		
C. elegans - long telomeres		Trait State in Taxon B		
Taxon A		Ancestral State		
Intraspecific (<a +intraspecific+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Intraspecific+"#gephebase-summary-title)		Taxonomic Status		
Taxon A		Taxon B		
Caenorhabditis elegans (<a +caenorhabditis+elegans+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Caenorhabditis+elegans+"#gephebase-summary-title)		Caenorhabditis elegans (<a +caenorhabditis+elegans+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Caenorhabditis+elegans+"#gephebase-summary-title)		
-		-		
roundworm; Rhabditis elegans; Caenorhabditis elegans (Maupas, 1900); Rhabditis elegans Maupas, 1900		roundworm; Rhabditis elegans; Caenorhabditis elegans (Maupas, 1900); Rhabditis elegans Maupas, 1900		
species		species		
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Ecdysozoa; Nematoda; Chromadorea; Rhabditida; Rhabditina; Rhabditomorpha; Rhabditoidea; Rhabditidae; Peloderinae; Caenorhabditis		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Ecdysozoa; Nematoda; Chromadorea; Rhabditida; Rhabditina; Rhabditomorpha; Rhabditoidea; Rhabditidae; Peloderinae; Caenorhabditis		
Caenorhabditis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=6237)		Caenorhabditis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=6237)		
6239 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=6239)		6239 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=6239)		
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?		
No		No		

GENOTYPIC CHANGE

pot-2		Generic Gene Name	UniProtKB Caenorhabditis elegans
CELE_F57C2.3; F57C2.3		Synonyms	O45595 (http://www.uniprot.org/uniprot/O45595)
6239.F57C2.3 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=6239.F57C2.3)		String	GenebankID or UniProtKB
-		Sequence Similarities	NM_064616.3 (https://www.ncbi.nlm.nih.gov/nuccore/NM_064616.3)
		GO - Molecular Function	
GO:0035312 : 5'-3' exodeoxyribonuclease activity (https://www.ebi.ac.uk/QuickGO/term/GO:0035312)			
GO:0061730 : C-rich strand telomeric DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0061730)			
GO:0003684 : damaged DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003684)			
GO:0043047 : single-stranded telomeric DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043047)			

GO - Biological Process

GO:0006303 : double-strand break repair via nonhomologous end joining
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006303>)
GO:0036297 : interstrand cross-link repair
(<https://www.ebi.ac.uk/QuickGO/term/GO:0036297>)
GO:1904357 : negative regulation of telomere maintenance via telomere lengthening
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904357>)
GO:0031848 : protection from non-homologous end joining at telomere
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031848>)

GO - Cellular Component

GO:0000784 : nuclear chromosome, telomeric region
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000784>)

Yes ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~Yes^#gephebase-summary-title))

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding^#gephebase-summary-title))

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=~SNP^#gephebase-summary-title))

Nonsynonymous

Phenylalanine-to-isoleucine (F68I)

Association Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Association Mapping^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Association+Mapping^#gephebase-summary-title))

Presumptive Null

Molecular Type

Aberration Type

SNP Coding Change

Molecular Details of the Mutation

Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

The Genetic Basis of Natural Variation in *Caenorhabditis elegans* Telomere Length. (2016) (<https://pubmed.ncbi.nlm.nih.gov/27449056>)

Authors

Cook DE; Zdraljevic S; Tanny RE; Seo B; Riccardi DD; Noble LM; Rockman MV; Alkema MJ; Braendle C; Kammenga JE; Wang J; Kruglyak L; F lix MA; Lee J; Andersen EC

Abstract

Telomeres are involved in the maintenance of chromosomes and the prevention of genome instability. Despite this central importance, significant variation in telomere length has been observed in a variety of organisms. The genetic determinants of telomere-length variation and their effects on organismal fitness are largely unexplored. Here, we describe natural variation in telomere length across the *Caenorhabditis elegans* species. We identify a large-effect variant that contributes to differences in telomere length. The variant alters the conserved oligonucleotide/oligosaccharide-binding fold of protection of telomeres 2 (POT-2), a homolog of a human telomere-capping shelterin complex subunit. Mutations within this domain likely reduce the ability of POT-2 to bind telomeric DNA, thereby increasing telomere length. We find that telomere-length variation does not correlate with offspring production or longevity in *C. elegans* wild isolates, suggesting that naturally long telomeres play a limited role in modifying fitness phenotypes in *C. elegans*.

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Additional References

RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

The F68I mutation is found in 12 strains of *C. elegans*. The haplotypes that contain this variant are rare and not geographically restricted.

