

GEPHE SUMMARY

pot-2 (#gephebase-summary-title)	Gephebase Gene	GP00001317	GepheID
Published	Entry Status	Courtier	Main curator

PHENOTYPIC CHANGE

Trait Category		Trait	
Physiology (#gephebase-summary-title)			
Telomere length (#gephebase-summary-title)		Trait State in Taxon A	
C. elegans		Trait State in Taxon B	
C. elegans - long telomeres		Ancestral State	
Taxon A		Taxonomic Status	
Intraspecific (#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
Caenorhabditis elegans (#gephebase-summary-title)		Caenorhabditis elegans (#gephebase-summary-title)	
-	Common Name	-	Common Name
roundworm; Rhabditis elegans; Caenorhabditis elegans (Maupas, 1900); Rhabditis elegans Maupas, 1900	Synonyms	roundworm; Rhabditis elegans; Caenorhabditis elegans (Maupas, 1900); Rhabditis elegans Maupas, 1900	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Ecdysozoa; Nematoda; Chromadorea; Rhabditida; Rhabditina; Rhabditomorpha; Rhabditoidea; Rhabditidae; Peloderinae; Caenorhabditis	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Ecdysozoa; Nematoda; Chromadorea; Rhabditida; Rhabditina; Rhabditomorpha; Rhabditoidea; Rhabditidae; Peloderinae; Caenorhabditis	Lineage
Caenorhabditis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6237)	Parent	Caenorhabditis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6237)	Parent
6239 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6239)	NCBI Taxonomy ID	6239 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6239)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

pot-2	Generic Gene Name	UniProtKB Caenorhabditis elegans
CELE_F57C2.3; F57C2.3	Synonyms	GenebankID or UniProtKB
6239.F57C2.3 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 6239.F57C2.3)	String	NM_064616.3 (https://www.ncbi.nlm.nih.gov/nuccore/NM_064616.3)
-	Sequence Similarities	
GO:0035312 : 5'-3' exodeoxyribonuclease activity (https://www.ebi.ac.uk/QuickGO/term/GO:0035312)	GO - Molecular Function	
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>)

Presumptive Null

Yes ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=%27Yes%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive%20Null=%27Yes%27#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=%27Coding%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular%20Type=%27Coding%27#gephebase-summary-title))

Aberration Type

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type=%27SNP%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration%20Type=%27SNP%27#gephebase-summary-title))

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Phenylalanine-to-isoleucine (F68I)

Experimental Evidence

Association Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=%27Association Mapping%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental%20Evidence=%27Association%20Mapping%27#gephebase-summary-title))

	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; FÅ©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

Related Genes

No matches found.

Related Haplotypes

No matches found.

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.

