Gephebase Gene GephelD Delta-1-pyrroline-5-carboxylate synthase A (https://www.gephebase.org/search-GP00001280 criteria?/and+Gene Gephebase=^Delta-1-pyrroline-5-carboxylate synthase A^#gephebase-Main curator summary-title) Courtier Entry Status Published PHENOTYPIC CHANGE Trait Category  $Physiology\ (https://www.gephebase.org/search-criteria?/and+Trait$ Category=^Physiology^#gephebase-summary-title) Trait Drought response (drought-induced proline accumulation) (https://www.gephebase.org/search-criteria?/and+Trait=^Drought response (droughtinduced proline accumulation)^#gephebase-summary-title) Trait State in Taxon A Arabidopsis thaliana- Ler0 Trait State in Taxon B Arabidopsis thaliana- Shahdara Ancestral State Taxon A Taxonomic Status Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=`Intraspecific`\*gephebase-summary-title)Taxon A Taxon B Latin Name Latin Name Arabidopsis thaliana Arabidopsis thaliana (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Arabidopsis (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Arabidopsis thaliana^#qephebase-summary-title) thaliana^#gephebase-summary-title) Common Name Common Name thale cress thale cress Synonyms Synonyms thale cress; mouse-ear cress; thale-cress; Arabidopsis thaliana (L.) Heynh.; Arabidopsis thale cress; mouse-ear cress; thale-cress; Arabidopsis thaliana (L.) Heynh.; Arabidopsis thaliana (thale cress); Arabidopsis\_thaliana; Arbisopsis thaliana; thale kress thaliana (thale cress); Arabidopsis\_thaliana; Arbisopsis thaliana; thale kress Rank Rank species species Lineage Lineage  $cellular\ organisms;\ Eukaryota;\ Viridiplantae;\ Streptophyta;\ Streptophytina;\ Embryophyta;$  $cellular\ organisms;\ Eukaryota;\ Viridiplantae;\ Streptophyta;\ Streptophytina;\ Embryophyta;$ Tracheophyta; Euphyllophyta; Spermatophyta; Magnoliophyta; Mesangiospermae; Tracheophyta; Euphyllophyta; Spermatophyta; Magnoliophyta; Mesangiospermae; eudicotyledons; Gunneridae; Pentapetalae; rosids; malvids; Brassicales; Brassicaceae; eudicotyledons; Gunneridae; Pentapetalae; rosids; malvids; Brassicales; Brassicaceae; Camelineae; Arabidopsis Camelineae; Arabidopsis Parent Parent Arabidopsis () - (Rank: genus) Arabidopsis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 3701) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 3701) NCBI Taxonomy ID NCBI Taxonomy ID (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 3702) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 3702) is Taxon A an Infraspecies? is Taxon B an Infraspecies? Yes Yes Taxon A Description Taxon B Description Arabidopsis thaliana- Ler0 Arabidopsis thaliana- Shahdara

## GENOTYPIC CHANGE

Generic Gene Name UniProtKB Arabidopsis thaliana P<sub>5</sub>CSA P54887 (http://www.uniprot.org/uniprot/P54887) GenebankID or UniProtKB Synonyms ATP5CS; delta1-pyrroline-5-carboxylate synthase 1; T5I7.10; T5I7\_10; P5CS1; At2g39800 818566 (https://www.ncbi.nlm.nih.gov/nuccore/818566) 3702.AT2G39800.1

 $(http://string-db.org/newstring\_cgi/show\_network\_section.pl? identifier=3702.AT2G39800.1$ Sequence Similarities

In the N-terminal section; belongs to the glutamate 5-kinase family.;In the C-terminal section; belongs to the gamma-glutamyl phosphate reductase family

GO - Molecular Function

GO:0005524: ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)

(https://www.ebi.ac.uk/QuickGO/term/GO:0004350) GO - Biological Process  $GO: 0009651: response \ to \ salt \ stress \ (https://www.ebi.ac.uk/QuickGO/term/GO: 0009651)$ GO:0048364: root development (https://www.ebi.ac.uk/QuickGO/term/GO:0048364) GO:0009414 : response to water deprivation (https://www.ebi.ac.uk/QuickGO/term/GO:0009414)GO:0042538 : hyperosmotic salinity response (https://www.ebi.ac.uk/QuickGO/term/GO:0042538) GO:0006979: response to oxidative stress (https://www.ebi.ac.uk/QuickGO/term/GO:0006979) GO:0055129: L-proline biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0055129)  $GO:0009555: pollen \ development \ (https://www.ebi.ac.uk/QuickGO/term/GO:0009555)$ GO:0006561: proline biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006561) GO - Cellular Component  $GO:0005737: cytoplasm \ (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)$ GO:0016020: membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016020)GO:0009507: chloroplast (https://www.ebi.ac.uk/QuickGO/term/GO:0009507)Mutation #1 Presumptive Null Yes (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) Aberration Type  $Insertion \ (https://www.gephebase.org/search-criteria?/and+Aberration \ Type=^lnsertion^{\#gephebase-summary-title})$ 1-9 bp Molecular Details of the Mutation insertion of three or four TA repeats in intron 2 - together with the G>T mutation it causes frequent exon 3 skipping Experimental Evidence  $Linkage\ Mapping\ (https://www.gephebase.org/search-criteria?/and+Experimental\ Evidence=`Linkage\ Mapping\ ^\#gephebase-summary-title)$ Intron-mediated alternative splicing of Arabidopsis P5CS1 and its association with natural variation in proline and climate adaptation. (2012) (https://pubmed.ncbi.nlm.nih.gov/22615385) Kesari R; Lasky JR; Villamor JG; Des Marais DL; Chen YJ; Liu TW; Lin W; Juenger TE; Verslues PE

GO:0017084 : delta1-pyrroline-5-carboxylate synthetase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0017084)

GO:0004349 : glutamate 5-kinase activity

(https://www.ebi.ac.uk/QuickGO/term/GO:0004349)GO:0004350: glutamate-5-semialdehyde dehydrogenase activity

Abstract

Authors

Molecular Type

Insertion Size

Main Reference

Drought-induced proline accumulation is widely observed in plants but its regulation and adaptive value are not as well understood. Proline accumulation of the Arabidopsis accession Shakdara (Sha) was threefold less than that of Landsberg erecta (Ler) and quantitative trait loci mapping identified a reduced function allele of the proline synthesis enzyme  $\hat{I}^{"}(1)$ pyrroline-5-carboxylate synthetase1 (P5CS1) as a basis for the lower proline of Sha. Sha P5CS1 had additional TA repeats in intron 2 and a G-to-T transversion in intron 3 that were sufficient to promote alternative splicing and production of a nonfunctional transcript lacking exon 3 (exon 3-skip P5CS1). In Sha, and additional accessions with the same intron polymorphisms, the nonfunctional exon 3-skip P5CS1 splice variant constituted as much as half of the total P5CS1 transcript. In a larger panel of Arabidopsis accessions, low water potential-induced proline accumulation varied by 10-fold and variable production of exon 3-skip P5CS1 among accessions was an important, but not the sole, factor underlying variation in proline accumulation. Population genetic analyses suggest that P5CS1 may have evolved under positive selection, and more extensive correlation of exon 3-skip P5CS1 production than proline abundance with climate conditions of natural accessions also suggest a role of P5CS1 in local adaptation to the environment. These data identify a unique source of alternative splicing in plants, demonstrate a role of exon 3-skip P5CS1 in natural variation of proline metabolism, and suggest an association of P5CS1 and its alternative splicing with environmental adaptation.

Additional References

Mutation #2			
			Presumptive Null
No (https://www.gephebase.org/search-criteria?/and+Presump	otive Null=^No^#gephebase-summ	ary-title)	Molecular Type
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)			Molecular Type
SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title)			Aberration Type
SINF (https://www.gepnebase.org/search-criteria:/and+Aberra	ition Type= 311P #gepnebase-sum	mary-title)	SNP Coding Change
-			Molecular Details of the Mutation
G-to-T in intron 3 causing alternative splicing			Molecular Details of the Mutation
	V 15		Experimental Evidence
Linkage Mapping (https://www.gephebase.org/search-criteria	//and+Experimental Evidence=^Linl	kage Mapping^#gephebase-summary-title	<del>)</del> )
	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	_	_	_

Main Reference

Intron-mediated alternative splicing of Arabidopsis P5CS1 and its association with natural variation in proline and climate adaptation. (2012) (https://pubmed.ncbi.nlm.nih.gov/22615385)

Authors

Kesari R; Lasky JR; Villamor JG; Des Marais DL; Chen YJ; Liu TW; Lin W; Juenger TE; Verslues PE

Abstract

Drought-induced proline accumulation is widely observed in plants but its regulation and adaptive value are not as well understood. Proline accumulation of the Arabidopsis accession Shakdara (Sha) was threefold less than that of Landsberg erecta (Ler) and quantitative trait loci mapping identified a reduced function allele of the proline synthesis enzyme Î"(1)-pyrroline-5-carboxylate synthetase1 (P5CS1) as a basis for the lower proline of Sha. Sha P5CS1 had additional TA repeats in intron 2 and a G-to-T transversion in intron 3 that were sufficient to promote alternative splicing and production of a nonfunctional transcript lacking exon 3 (exon 3-skip P5CS1). In Sha, and additional accessions with the same intron polymorphisms, the nonfunctional exon 3-skip P5CS1 splice variant constituted as much as half of the total P5CS1 transcript. In a larger panel of Arabidopsis accessions, low water potential-induced proline accumulation varied by 10-fold and variable production of exon 3-skip P5CS1 among accessions was an important, but not the sole, factor underlying variation in proline accumulation. Population genetic analyses suggest that P5CS1 may have evolved under positive selection, and more extensive correlation of exon 3-skip P5CS1 production than proline abundance with climate conditions of natural accessions also suggest a role of P5CS1 in local adaptation to the environment. These data identify a unique source of alternative splicing in plants, demonstrate a role of exon 3-skip P5CS1 in natural variation of proline metabolism, and suggest an association of P5CS1 and its alternative splicing with environmental adaptation.

Additional References

## **RELATED GEPHE**

Related Genes

No matches found.

Related Haplotypes

No matches found.

## **EXTERNAL LINKS**

## **COMMENTS**

@Splicing neither the small intron 2 TA-insertion nor the intron 3 polymorphism by itself are sufficient to cause frequent exon 3-skip transcript formation. The high exon 3-skip P5CS1 accessions (with exception of Pt-0) all have both the intron 3 G-to-T transition and insertion of three or four TA repeats in intron 2. Arabidopsis thaliana- Shahdara allele is a reduced functional allele