

GEPHE SUMMARY

IIl1 (#gephebase-summary-title)	Generic Gene Name	GP00001229	GephelD
	Entry Status	Arnoult	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category
Morphology (<a)<="" a="" href="https://www.gephebase.org/search-criteria?/and+Trait Category='Morphology'^#gephebase-summary-title">	Trait
Plant growth (pleiotropic growth abnormalities) (<a)<="" a="" href="https://www.gephebase.org/search-criteria?/and+Trait='Plant growth (pleiotropic growth abnormalities)^#gephebase-summary-title">	Trait State in Taxon A
Arabidopsis thaliana- Pf-0	Trait State in Taxon B
Arabidopsis thaliana-Bur-0	Ancestral State
Data not curated	Taxonomic Status
Intraspecific (<a)<="" a="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic Status='Intraspecific'^#gephebase-summary-title">	

Taxon A		Taxon B	
Arabidopsis thaliana (<a)<="" a="" href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms='Arabidopsis thaliana'^#gephebase-summary-title">	Latin Name	Arabidopsis thaliana (<a)<="" a="" href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms='Arabidopsis thaliana'^#gephebase-summary-title">	Latin Name
thale cress	Common Name	thale cress	Common Name
thale cress; mouse-ear cress; thale-cress; Arabidopsis thaliana (L.) Heynh.; Arabidopsis thaliana (thale cress); Arabidopsis_thaliana; Arbisopsis thaliana; thale kress	Synonyms	thale cress; mouse-ear cress; thale-cress; Arabidopsis thaliana (L.) Heynh.; Arabidopsis thaliana (thale cress); Arabidopsis_thaliana; Arbisopsis thaliana; thale kress	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Viridiplantae; Streptophyta; Streptophytina; Embryophyta; Tracheophyta; Euphyllophyta; Spermatophyta; Magnoliophyta; Mesangiospermae; eudicotyledons; Gunneridae; Pentapetalae; rosids; malvids; Brassicales; Brassicaceae; Camelinae; Arabidopsis	Lineage	cellular organisms; Eukaryota; Viridiplantae; Streptophyta; Streptophytina; Embryophyta; Tracheophyta; Euphyllophyta; Spermatophyta; Magnoliophyta; Mesangiospermae; eudicotyledons; Gunneridae; Pentapetalae; rosids; malvids; Brassicales; Brassicaceae; Camelinae; Arabidopsis	Lineage
Arabidopsis () - (Rank: genus) (<a)<="" a="" href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 3701">	Parent	Arabidopsis () - (Rank: genus) (<a)<="" a="" href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 3701">	Parent
3702 (<a)<="" a="" href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 3702">	NCBI Taxonomy ID	3702 (<a)<="" a="" href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 3702">	NCBI Taxonomy ID
Yes	is Taxon A an Infraspecies?	Yes	is Taxon B an Infraspecies?
Arabidopsis thaliana- Pf-0	Taxon A Description	Arabidopsis thaliana-Bur-0	Taxon B Description

GENOTYPIC CHANGE

IIl1	Generic Gene Name	UniProtKB Arabidopsis thaliana
	Synonyms	GenebankID or UniProtKB
ATLEUC1; isopropyl malate isomerase large subunit 1; T9E8_170; T9E8_170; At4g13430	String	
3702.AT4G13430.1 (<a)<="" a="" href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 3702.AT4G13430.1">	Sequence Similarities	
Belongs to the aconitase/IMC isomerase family.	GO - Molecular Function	
GO:0046872 : metal ion binding (<a)<="" a="" href="https://www.ebi.ac.uk/QuickGO/term/GO:0046872">		
GO:0003861 : 3-isopropylmalate dehydratase activity (<a)<="" a="" href="https://www.ebi.ac.uk/QuickGO/term/GO:0003861">		

GO:0051539 : 4 iron, 4 sulfur cluster binding

(<https://www.ebi.ac.uk/QuickGO/term/GO:0051539>)

GO:0050486 : intramolecular transferase activity, transferring hydroxy groups

(<https://www.ebi.ac.uk/QuickGO/term/GO:0050486>)

GO - Biological Process

GO:0019761 : glucosinolate biosynthetic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0019761>)

GO:0046686 : response to cadmium ion

(<https://www.ebi.ac.uk/QuickGO/term/GO:0046686>)

GO:0009098 : leucine biosynthetic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0009098>)

GO - Cellular Component

GO:0009507 : chloroplast (<https://www.ebi.ac.uk/QuickGO/term/GO:0009507>)

GO:0009570 : chloroplast stroma (<https://www.ebi.ac.uk/QuickGO/term/GO:0009570>)

GO:0009536 : plastid (<https://www.ebi.ac.uk/QuickGO/term/GO:0009536>)

Presumptive Null

No ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=^No))

Molecular Type

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

Aberration Type

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

Insertion Size

-

Molecular Details of the Mutation

expansion of a GAA/TTC trinucleotide repeat in the third intron

Experimental Evidence

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

Main Reference

A genetic defect caused by a triplet repeat expansion in *Arabidopsis thaliana*. (2009) (<https://pubmed.ncbi.nlm.nih.gov/19150812>)

Authors

Sureshkumar S; Todesco M; Schneeberger K; Harilal R; Balasubramanian S; Weigel D

Abstract

Variation in the length of simple DNA triplet repeats has been linked to phenotypic variability in microbes and to several human disorders. Population-level forces driving triplet repeat contraction and expansion in multicellular organisms are, however, not well understood. We have identified a triplet repeat-associated genetic defect in an *Arabidopsis thaliana* variety collected from the wild. The Bur-0 strain carries a dramatically expanded TTC/GAA repeat in the intron of the ISOPROPYL MALATE ISOMERASE LARGE SUB UNIT1 (IIL1; At4g13430) gene. The repeat expansion causes an environment-dependent reduction in IIL1 activity and severely impairs growth of this strain, whereas contraction of the expanded repeat can reverse the detrimental phenotype. The Bur-0 IIL1 defect thus presents a genetically tractable model for triplet repeat expansions and their variability in natural populations.

Additional References

A Polynucleotide Repeat Expansion Causing Temperature-Sensitivity Persists in Wild Irish Accessions of *Arabidopsis thaliana*. (2016) (<https://pubmed.ncbi.nlm.nih.gov/27630650>)

RELATED GEPHE

Related Genes

5 (EARLY FLOWERING 3(ELF3) [possible pseudo-replicate], Enhanced shoot growth under mannitol stress 2 (EGM2), TZP, FUMARASE 2, ICARUS1)

([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=^3702#/and+Trait=Plant+growth/and+groupHaplotypes=true))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Pleiotropy - triplet expansion causes IIL transcript decrease ; growth defect dependent on high temperature or high UV-B; Two genetic modifiers detected by QTL in 27630650