

GEPHE SUMMARY

IIL1 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~IIL1^#gephebase-summary-title)	Gephebase Gene	GP00001229	GepheID
	Entry Status		Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology^#gephebase-summary-title)		Trait Category	
Plant growth (pleiotropic growth abnormalities) (https://www.gephebase.org/search-criteria?/and+Trait=~Plant+growth+(pleiotropic+growth+abnormalities)^#gephebase-summary-title)		Trait	
Arabidopsis thaliana- Pf-0		Trait State in Taxon A	
Arabidopsis thaliana-Bur-0		Trait State in Taxon B	
Data not curated		Ancestral State	
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Intraspecific^#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Arabidopsis thaliana (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Arabidopsis+thaliana^#gephebase-summary-title)		Arabidopsis thaliana (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Arabidopsis+thaliana^#gephebase-summary-title)	
thale cress		thale cress	
thale cress; mouse-ear cress; thale-cress; Arabidopsis thaliana (L.) Heynh.; Arabidopsis thaliana (thale cress); Arabidopsis_thaliana; Arbisopsis thaliana; thale kress		thale cress; mouse-ear cress; thale-cress; Arabidopsis thaliana (L.) Heynh.; Arabidopsis thaliana (thale cress); Arabidopsis_thaliana; Arbisopsis thaliana; thale kress	
species		species	
cellular organisms; Eukaryota; Viridiplantae; Streptophyta; Streptophytina; Embryophyta; Tracheophyta; Euphyllophyta; Spermatophyta; Magnoliophyta; Mesangiospermae; eudicotyledons; Gunneridae; Pentapetales; rosids; malvids; Brassicales; Brassicaceae; Camelineae; Arabidopsis		cellular organisms; Eukaryota; Viridiplantae; Streptophyta; Streptophytina; Embryophyta; Tracheophyta; Euphyllophyta; Spermatophyta; Magnoliophyta; Mesangiospermae; eudicotyledons; Gunneridae; Pentapetales; rosids; malvids; Brassicales; Brassicaceae; Camelineae; Arabidopsis	
Arabidopsis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=3701)		Arabidopsis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=3701)	
3702 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=3702)		3702 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=3702)	
Yes		Yes	
Arabidopsis thaliana- Pf-0		Arabidopsis thaliana-Bur-0	

GENOTYPIC CHANGE

IIL1		Generic Gene Name		UniProtKB Arabidopsis thaliana	
ATLEUC1; isopropyl malate isomerase large subunit 1; T9E8.170; T9E8_170; At4g13430		Synonyms		GenebankID or UniProtKB	
3702.AT4G13430.1 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=3702.AT4G13430.1)		String		826975 (https://www.ncbi.nlm.nih.gov/nuccore/826975)	
Belongs to the aconitase/IPM isomerase family.		Sequence Similarities			
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)		GO - Molecular Function			
GO:0003861 : 3-isopropylmalate dehydratase activity (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 (<a +no`"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Presumptive Null=">https://www.gephebase.org/search-criteria?/and+Presumptive Null="+No`"#gephebase-summary-title)	
Cis-regulatory (<a +cis-regulatory`"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Molecular Type=">https://www.gephebase.org/search-criteria?/and+Molecular Type="+Cis-regulatory`"#gephebase-summary-title)	Molecular Type
	Aberration Type
Insertion (<a +insertion`"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Aberration Type=">https://www.gephebase.org/search-criteria?/and+Aberration Type="+Insertion`"#gephebase-summary-title)	
-	Insertion Size
	Molecular Details of the Mutation
expansion of a GAA/TTC trinucleotide repeat in the third intron	
	Experimental Evidence
Linkage Mapping (<a +linkage="" href="https://www.gephebase.org/search-criteria?/and+Experimental Evidence=" mapping`"#gephebase-summary-title"="">https://www.gephebase.org/search-criteria?/and+Experimental Evidence="+Linkage Mapping`"#gephebase-summary-title)	
	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) (<a +3702`"="" and+grouphaplotypes='true#gephebase-summary-title"' and+trait="Plant" growth="" href="https://www.gephebase.org/search-criteria?/or+Taxon ID=">https://www.gephebase.org/search-criteria?/or+Taxon ID="+3702`"/and+Trait=Plant growth/and+groupHaplotypes=true#gephebase-summary-title)	
	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