

GEPHE SUMMARY

		Gephebase Gene		GepheID
Ldia2 - Diaphanous-related formin (#Ldia2 - Diaphanous-related formin ^#gephebase-summary-title)		GP00000538		
		Entry Status	Martin	Main curator
Published				

PHENOTYPIC CHANGE

		Trait Category	
Morphology (#Morphology ^#gephebase-summary-title)			
		Trait	
Shell chirality (coiling) (https://www.gephebase.org/search-criteria?/and+Trait=Shell chirality (coiling) ^#gephebase-summary-title)			
		Trait State in Taxon A	
Lymnaea stagnalis - dextral (DD or Dd)			
		Trait State in Taxon B	
Lymnaea stagnalis - sinistral (dd)			
		Ancestral State	
Taxon A		Taxonomic Status	
Intraspecific (#Intraspecific ^#gephebase-summary-title)			
Taxon A		Taxon B	
Lymnaea stagnalis		Lymnaea stagnalis	
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=Lymnaea stagnalis^#gephebase-summary-title)		(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=Lymnaea stagnalis^#gephebase-summary-title)	
		Common Name	
great pond snail		great pond snail	
		Synonyms	
great pond snail		great pond snail	
		Rank	
species		species	
		Lineage	
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Lophotrochozoa; Mollusca; Gastropoda; Heterobranchia; Euthyneura; Panpulmonata; Hygrophila; Lymnaeoidea; Lymnaeidae; Lymnaea		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Lophotrochozoa; Mollusca; Gastropoda; Heterobranchia; Euthyneura; Panpulmonata; Hygrophila; Lymnaeoidea; Lymnaeidae; Lymnaea	
		Parent	
Lymnaea () - (Rank: genus)		Lymnaea () - (Rank: genus)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6522)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6522)	
		NCBI Taxonomy ID	
6523		6523	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6523)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6523)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

		Generic Gene Name	UniProtKB Homo sapiens
DIAPH1		O60610 (http://www.uniprot.org/uniprot/O60610)	
		Synonyms	GenebankID or UniProtKB
DIA1; DRF1; DFNA1; LFHL1; SCBMS; hDIA1; DIAP1		KU341302 (https://www.ncbi.nlm.nih.gov/nuccore/KU341302)	
		String	
9606.ENSPP00000381565			
(http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000381565)			
		Sequence Similarities	
Belongs to the formin homology family. Diaphanous subfamily.			
		GO - Molecular Function	
GO:0003723 : RNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003723)			
GO:0005102 : signaling receptor binding			
(https://www.ebi.ac.uk/QuickGO/term/GO:0005102)			
GO:0003779 : actin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003779)			
GO:0044325 : ion channel binding (https://www.ebi.ac.uk/QuickGO/term/GO:0044325)			
GO:0017048 : Rho GTPase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0017048)			
		GO - Biological Process	
GO:0007010 : cytoskeleton organization			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0007010>)
GO:0008360 : regulation of cell shape
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008360>)
GO:0043312 : neutrophil degranulation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043312>)
GO:0030335 : positive regulation of cell migration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030335>)
GO:0007605 : sensory perception of sound
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007605>)
GO:0030036 : actin cytoskeleton organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030036>)
GO:0030041 : actin filament polymerization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030041>)
GO:0071420 : cellular response to histamine
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071420>)
GO:0035372 : protein localization to microtubule
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035372>)
GO:2000145 : regulation of cell motility
(<https://www.ebi.ac.uk/QuickGO/term/GO:2000145>)
GO:0051493 : regulation of cytoskeleton organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051493>)
GO:0032886 : regulation of microtubule-based process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032886>)
GO:0051279 : regulation of release of sequestered calcium ion into cytosol
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051279>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0030667 : secretory granule membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030667>)
GO:0101003 : ficolin-1-rich granule membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0101003>)
GO:0005815 : microtubule organizing center
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005815>)
GO:0072686 : mitotic spindle (<https://www.ebi.ac.uk/QuickGO/term/GO:0072686>)
GO:0032587 : ruffle membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0032587>)

Presumptive Null

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

Molecular Type

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

Aberration Type

Deletion (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion~#gephebase-summary-title>)

Deletion Size

1-9 bp

Molecular Details of the Mutation

1bp deletion resulting in frame-shift (pseudogenization)

Experimental Evidence

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

Main Reference

Formin Is Associated with Left-Right Asymmetry in the Pond Snail and the Frog. (2016) (<https://pubmed.ncbi.nlm.nih.gov/26923788>)

Authors

Davison A; McDowell GS; Holden JM; Johnson HF; Koutsovoulos GD; Liu MM; Hulpiau P; Van Roy F; Wade CM; Banerjee R; Yang F; Chiba S; Davey JW; Jackson DJ; Levin M; Blaxter ML

Abstract

While components of the pathway that establishes left-right asymmetry have been identified in diverse animals, from vertebrates to flies, it is striking that the genes involved in the first symmetry-breaking step remain wholly unknown in the most obviously chiral animals, the gastropod snails. Previously, research on snails was used to show that left-right signaling of Nodal, downstream of symmetry breaking, may be an ancestral feature of the Bilateria [1 and 2]. Here, we report that a disabling mutation in one copy of a tandemly duplicated, diaphanous-related formin is perfectly associated with symmetry breaking in the pond snail. This is supported by the observation that an anti-formin drug treatment converts dextral snail embryos to a sinistral phenocopy, and in frogs, drug inhibition or overexpression by microinjection of formin has a chirality-randomizing effect in early (pre-cilia) embryos. Contrary to expectations based on existing models [3, 4 and 5], we discovered asymmetric gene expression in 2- and 4-cell snail embryos, preceding morphological asymmetry. As the formin-actin filament has been shown to be part of an asymmetry-breaking switch in vitro [6 and 7], together these results are consistent with the view that animals with diverse body plans may derive their asymmetries from the same intracellular chiral elements [8].

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Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Loss-of-function mutation in a tandem-duplicated gene; functional redundancy may explain lack of deleterious pleiotropy; Recessive allele is due to Loss-of-function