

## GEPHE SUMMARY

EDN3 ( <a +edn3+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+EDN3+"#gephebase-summary-title</a> )	Gephebase Gene	GP00002377	GepheID
Published	Entry Status	Santos	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title</a> )	Trait Category		
Coloration ( <a +coloration+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration+"#gephebase-summary-title</a> )	Trait		
WT coloration; æœdarkâ€ mottled green and black	Trait State in Taxon A		
White coloration	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated ( <a +domesticated+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated+"#gephebase-summary-title</a> )	Taxonomic Status		
	Taxon A		Taxon B
Ambystoma mexicanum ( <a +ambystoma+mexicanum+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Ambystoma+mexicanum+"#gephebase-summary-title</a> )	Latin Name	Ambystoma mexicanum ( <a +ambystoma+mexicanum+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Ambystoma+mexicanum+"#gephebase-summary-title</a> )	Latin Name
axolotl	Common Name	axolotl	Common Name
Gyrinus mexicanus; axolotl; Ambystoma mexicanum (Shaw & Nodder, 1798); Gyrinus mexicanus Shaw & Nodder, 1798	Synonyms	Gyrinus mexicanus; axolotl; Ambystoma mexicanum (Shaw & Nodder, 1798); Gyrinus mexicanus Shaw & Nodder, 1798	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amphibia; Batrachia; Caudata; Salamandroidea; Ambystomatidae; Ambystoma	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amphibia; Batrachia; Caudata; Salamandroidea; Ambystomatidae; Ambystoma	Lineage
Ambystoma (mole salamanders) - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8295">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8295</a> )	Parent	Ambystoma (mole salamanders) - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8295">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8295</a> )	Parent
8296 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8296">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8296</a> )	NCBI Taxonomy ID	8296 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8296">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8296</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

## GENOTYPIC CHANGE

EDN3	Generic Gene Name	P14138 ( <a href="http://www.uniprot.org/uniprot/P14138">http://www.uniprot.org/uniprot/P14138</a> )	UniProtKB Homo sapiens
ET3; ET-3; WS4B; HSCR4; PPET3	Synonyms	()	GenebankID or UniProtKB
9606.ENSP00000337128 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000337128">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000337128</a> )	String		
Belongs to the endothelin/sarafotoxin family.	Sequence Similarities		
GO:0005102 : signaling receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005102">https://www.ebi.ac.uk/QuickGO/term/GO:0005102</a> )	GO - Molecular Function		
GO:0031708 : endothelin B receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031708">https://www.ebi.ac.uk/QuickGO/term/GO:0031708</a> )			
GO:0005179 : hormone activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005179">https://www.ebi.ac.uk/QuickGO/term/GO:0005179</a> )			
	GO - Biological Process		

GO:0007275 : multicellular organism development  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0007275)  
 GO:0007165 : signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0007165)  
 GO:0030182 : neuron differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030182)  
 GO:0001755 : neural crest cell migration  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0001755)  
 GO:0006874 : cellular calcium ion homeostasis  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0006874)  
 GO:0007186 : G protein-coupled receptor signaling pathway  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0007186)  
 GO:0008284 : positive regulation of cell proliferation  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0008284)  
 GO:0043406 : positive regulation of MAP kinase activity  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0043406)  
 GO:0007267 : cell-cell signaling (https://www.ebi.ac.uk/QuickGO/term/GO:0007267)  
 GO:0045597 : positive regulation of cell differentiation  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0045597)  
 GO:0010460 : positive regulation of heart rate  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0010460)  
 GO:0048070 : regulation of developmental pigmentation  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0048070)  
 GO:0030593 : neutrophil chemotaxis (https://www.ebi.ac.uk/QuickGO/term/GO:0030593)  
 GO:0010468 : regulation of gene expression  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0010468)  
 GO:0008015 : blood circulation (https://www.ebi.ac.uk/QuickGO/term/GO:0008015)  
 GO:0007166 : cell surface receptor signaling pathway  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0007166)  
 GO:0010961 : cellular magnesium ion homeostasis  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0010961)  
 GO:0048016 : inositol phosphate-mediated signaling  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0048016)  
 GO:0030318 : melanocyte differentiation  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0030318)  
 GO:0030072 : peptide hormone secretion  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0030072)  
 GO:0046887 : positive regulation of hormone secretion  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0046887)  
 GO:0002690 : positive regulation of leukocyte chemotaxis  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0002690)  
 GO:0045840 : positive regulation of mitotic nuclear division  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0045840)  
 GO:1901381 : positive regulation of potassium ion transmembrane transport  
 (https://www.ebi.ac.uk/QuickGO/term/GO:1901381)  
 GO:0003100 : regulation of systemic arterial blood pressure by endothelin  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0003100)  
 GO:0019229 : regulation of vasoconstriction  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0019229)  
 GO:0042310 : vasoconstriction (https://www.ebi.ac.uk/QuickGO/term/GO:0042310)  
 GO:0014826 : vein smooth muscle contraction  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0014826)

GO - Cellular Component

GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)  
 GO:0005623 : cell (https://www.ebi.ac.uk/QuickGO/term/GO:0005623)  
 GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)	Presumptive Null
Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)	Molecular Type
Unknown (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Unknown^#gephebase-summary-title)	Aberration Type
-	Molecular Details of the Mutation
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)	Experimental Evidence
Identification of Mutant Genes and Introgressed Tiger Salamander DNA in the Laboratory Axolotl, <i>Ambystoma mexicanum</i> . (2017) (https://pubmed.ncbi.nlm.nih.gov/28127056)	Main Reference
Woodcock MR; Vaughn-Wolfe J; Elias A; Kump DK; Kendall KD; Timoshevskaya N; Timoshevskiy V; Perry DW; Smith JJ; Spiewak JE; Parichy DM; Voss SR	Authors

Abstract

The molecular genetic toolkit of the Mexican axolotl, a classic model organism, has matured to the point where it is now possible to identify genes for mutant phenotypes. We used a positional cloning-candidate gene approach to identify molecular bases for two historic axolotl pigment phenotypes: white and albino. White (d/d) mutants have defects in pigment cell morphogenesis and differentiation, whereas albino (a/a) mutants lack melanin. We identified in white mutants a transcriptional defect in endothelin 3 (edn3), encoding a peptide factor that promotes pigment cell migration and differentiation in other vertebrates. Transgenic restoration of Edn3 expression rescued the homozygous white mutant phenotype. We mapped the albino locus to tyrosinase (tyr) and identified polymorphisms shared between the albino allele (tyr) and tyr alleles in a Minnesota population of tiger salamanders from which the albino trait was introgressed. tyr has a 142â€‰bp deletion and similar engineered alleles recapitulated the albino phenotype. Finally, we show that historical introgression of tyr significantly altered genomic composition of the laboratory axolotl, yielding a distinct, hybrid strain of ambystomatid salamander. Our results demonstrate the feasibility of identifying genes for traits in the laboratory Mexican axolotl.

Additional References

## RELATED GEPHE

No matches found.

No matches found.

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## EXTERNAL LINKS

## COMMENTS