GEPHE SUMMARY

Gephebase Gene

EDN3 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^EDN3^#gephebase-summary-title)

GepheID

GP00002377

Entry Status

Published

Main curator

Santos

PHENOTYPIC CHANGE

Trait Category

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait

Coloration (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration^#gephebase-summary-title)

Trait State in Taxon A

WT coloration; â€œdarkâ€ mottled green and black

Trait State in Taxon B

White coloration

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name

Ambystoma mexicanum (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ambystoma mexicanum^#gephebase-summary-title)

Common Name

axolotl

Synonyms

Gyrinus mexicanus; axolotl; Ambystoma mexicanum (Shaw & Nodder, 1798); Gyrinus mexicanus Shaw & Nodder, 1798

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amphibia; Batrachia; Caudata; Salamandroidea; Ambystomatidae; Ambystoma

Parent

Ambystoma (mole salamanders) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8295)

NCBI Taxonomy ID

8296 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8296)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Ambystoma mexicanum (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ambystoma mexicanum^#gephebase-summary-title)

Common Name

axolotl

Synonyms

Gyrinus mexicanus; axolotl; Ambystoma mexicanum (Shaw & Nodder, 1798); Gyrinus mexicanus Shaw & Nodder, 1798

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amphibia; Batrachia; Caudata; Salamandroidea; Ambystomatidae; Ambystoma

Parent

Ambystoma (mole salamanders) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8295)

NCBI Taxonomy ID

8296 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8296)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

EDN3

Synonyms

ET3; ET-3; WS4B; HSCR4; PPET3

String

9606.ENSP00000337128 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000337128)

Sequence Similarities

Belongs to the endothelin/sarafotoxin family.

GO - Molecular Function

GO:0005102 : signaling receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005102)
GO:0031708 : endothelin B receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031708)
GO:0005179 : hormone activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005179)

GO - Biological Process

UniProtKB Homo sapiens

P14138 (http://www.uniprot.org/uniprot/P14138)

GenebankID or UniProtKB

()

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)

Presumptive Null

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

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Experimental Evidence

Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)

Main Reference

Identification of Mutant Genes and Introgressed Tiger Salamander DNA in the Laboratory Axolotl, Ambystoma mexicanum. (2017) (https://pubmed.ncbi.nlm.nih.gov/28127056)

Authors

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

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

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS