GEPHE SUMMARY

Gephebase Gene

Low-density lipoprotein receptor-related protein 2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Low-density lipoprotein receptor-related protein 2^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00002349

Main curator

Courtier

PHENOTYPIC CHANGE

Trait Category

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

Trait

Organ size (eye; enlarged) (https://www.gephebase.org/search-criteria?/and+Trait=^Organ size (eye; enlarged)^#gephebase-summary-title)

Trait State in Taxon A

eyes of standard size
wild goldfish and many strains of domesticated goldfish

Trait State in Taxon B

enlarged eyes
4 four Telescope-eye strains of domesticated goldfish

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

goldfish

Synonyms

Carassius carassius auratus; Cyprinus auratus; goldfish; Carassius auratus (Linnaeus, 1758); Cyprinus auratus Linnaeus, 1758; Carassius auratis

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Otomorpha; Ostariophysi; Otophysi; Cypriniphysae; Cypriniformes; Cyprinoidei; Cyprinidae; Cyprininae; Carassius

Parent

Carassius () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7956)

NCBI Taxonomy ID

7957 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7957)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

goldfish

Synonyms

Carassius carassius auratus; Cyprinus auratus; goldfish; Carassius auratus (Linnaeus, 1758); Cyprinus auratus Linnaeus, 1758; Carassius auratis

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Otomorpha; Ostariophysi; Otophysi; Cypriniphysae; Cypriniformes; Cyprinoidei; Cyprinidae; Cyprininae; Carassius

Parent

Carassius () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7956)

NCBI Taxonomy ID

7957 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7957)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

LRP2

Synonyms

DBS; GP330; LRP-2

String

9606.ENSP00000263816 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000263816)

Sequence Similarities

Belongs to the LDLR family.

GO - Molecular Function

GO:0005509 : calcium ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005509)
GO:0017124 : SH3 domain binding (https://www.ebi.ac.uk/QuickGO/term/GO:0017124)

UniProtKB Homo sapiens

P98164 (http://www.uniprot.org/uniprot/P98164)

GenebankID or UniProtKB

()

GO:0031994 : insulin-like growth factor I binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0031994)
GO:0051087 : chaperone binding (https://www.ebi.ac.uk/QuickGO/term/GO:0051087)
GO:0042562 : hormone binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042562)
GO:0005041 : low-density lipoprotein particle receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0005041)
GO:0038024 : cargo receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0038024)
GO:0140318 : protein transporter activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0140318)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
GO:0001843 : neural tube closure (https://www.ebi.ac.uk/QuickGO/term/GO:0001843)
GO:0006898 : receptor-mediated endocytosis
(https://www.ebi.ac.uk/QuickGO/term/GO:0006898)
GO:0001523 : retinoid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0001523)
GO:0008283 : cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008283)
GO:0008584 : male gonad development
(https://www.ebi.ac.uk/QuickGO/term/GO:0008584)
GO:0051897 : positive regulation of protein kinase B signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0051897)
GO:0001822 : kidney development (https://www.ebi.ac.uk/QuickGO/term/GO:0001822)
GO:0006897 : endocytosis (https://www.ebi.ac.uk/QuickGO/term/GO:0006897)
GO:0006629 : lipid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006629)
GO:0030900 : forebrain development
(https://www.ebi.ac.uk/QuickGO/term/GO:0030900)
GO:0003148 : outflow tract septum morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0003148)
GO:0030001 : metal ion transport (https://www.ebi.ac.uk/QuickGO/term/GO:0030001)
GO:0097242 : amyloid-beta clearance
(https://www.ebi.ac.uk/QuickGO/term/GO:0097242)
GO:0007605 : sensory perception of sound
(https://www.ebi.ac.uk/QuickGO/term/GO:0007605)
GO:0015031 : protein transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015031)
GO:0003281 : ventricular septum development
(https://www.ebi.ac.uk/QuickGO/term/GO:0003281)
GO:0050769 : positive regulation of neurogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0050769)
GO:0060068 : vagina development (https://www.ebi.ac.uk/QuickGO/term/GO:0060068)
GO:0030514 : negative regulation of BMP signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0030514)
GO:0071363 : cellular response to growth factor stimulus
(https://www.ebi.ac.uk/QuickGO/term/GO:0071363)
GO:0035904 : aorta development (https://www.ebi.ac.uk/QuickGO/term/GO:0035904)
GO:0060982 : coronary artery morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0060982)
GO:1904447 : folate import across plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:1904447)
GO:0140058 : neuron projection arborization
(https://www.ebi.ac.uk/QuickGO/term/GO:0140058)
GO:1905167 : positive regulation of lysosomal protein catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:1905167)
GO:0070447 : positive regulation of oligodendrocyte progenitor proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0070447)
GO:0061156 : pulmonary artery morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0061156)
GO:0044321 : response to leptin (https://www.ebi.ac.uk/QuickGO/term/GO:0044321)
GO:0003139 : secondary heart field specification
(https://www.ebi.ac.uk/QuickGO/term/GO:0003139)
GO:0045056 : transcytosis (https://www.ebi.ac.uk/QuickGO/term/GO:0045056)
GO:0150104 : transport across blood-brain barrier
(https://www.ebi.ac.uk/QuickGO/term/GO:0150104)
GO:0003223 : ventricular compact myocardium morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0003223)
GO:0042359 : vitamin D metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042359)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0016324 : apical plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016324)
GO:0070062 : extracellular exosome (https://www.ebi.ac.uk/QuickGO/term/GO:0070062)
GO:0043235 : receptor complex (https://www.ebi.ac.uk/QuickGO/term/GO:0043235)
GO:0005794 : Golgi apparatus (https://www.ebi.ac.uk/QuickGO/term/GO:0005794)

GO:0030669 : clathrin-coated endocytic vesicle membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0030669)
GO:0030425 : dendrite (https://www.ebi.ac.uk/QuickGO/term/GO:0030425)
GO:0005783 : endoplasmic reticulum
(https://www.ebi.ac.uk/QuickGO/term/GO:0005783)
GO:0030424 : axon (https://www.ebi.ac.uk/QuickGO/term/GO:0030424)
GO:0005764 : lysosome (https://www.ebi.ac.uk/QuickGO/term/GO:0005764)
GO:0009897 : external side of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0009897)
GO:0005765 : lysosomal membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005765)
GO:0031904 : endosome lumen (https://www.ebi.ac.uk/QuickGO/term/GO:0031904)
GO:0031526 : brush border membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0031526)
GO:0005905 : clathrin-coated pit (https://www.ebi.ac.uk/QuickGO/term/GO:0005905)

Mutation #1

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

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

SNP Coding Change

Nonsense

Molecular Details of the Mutation

-

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Tyr	STP	3425

Main Reference

The Genetic Basis of Morphological Diversity in Domesticated Goldfish. (2020) (https://pubmed.ncbi.nlm.nih.gov/32392470)

Authors

Kon T; Omori Y; Fukuta K; Wada H; Watanabe M; Chen Z; Iwasaki M; Mishina T; Matsuzaki SS; Yoshihara D; Arakawa J; Kawakami K; Toyoda A; Burgess SM; Noguchi H; Furukawa T

Abstract

Although domesticated goldfish strains exhibit highly diversified phenotypes in morphology, the genetic basis underlying these phenotypes is poorly understood. Here, based on analysis of transposable elements in the allotetraploid goldfish genome, we found that its two subgenomes have evolved asymmetrically since a whole-genome duplication event in the ancestor of goldfish and common carp. We conducted whole-genome sequencing of 27 domesticated goldfish strains and wild goldfish. We identified more than 60 million genetic variations and established a population genetic structure of major goldfish strains. Genome-wide association studies and analysis of strain-specific variants revealed genetic loci associated with several goldfish phenotypes, including dorsal fin loss, long-tail, telescope-eye, albinism, and heart-shaped tail. Our results suggest that accumulated mutations in the asymmetrically evolved subgenomes led to generation of diverse phenotypes in the goldfish domestication history. This study is a key resource for understanding the genetic basis of phenotypic diversity among goldfish strains.

Additional References

Mutation #2

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

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

SNP Coding Change

Nonsense

Molecular Details of the Mutation

-

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gly	STP	4480

Main Reference

The Genetic Basis of Morphological Diversity in Domesticated Goldfish. (2020) (https://pubmed.ncbi.nlm.nih.gov/32392470)

Authors

Kon T; Omori Y; Fukuta K; Wada H; Watanabe M; Chen Z; Iwasaki M; Mishina T; Matsuzaki SS; Yoshihara D; Arakawa J; Kawakami K; Toyoda A; Burgess SM; Noguchi H; Furukawa T

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Additional References

Mutation #3

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

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

Insertion Size

10-100 kb

Molecular Details of the Mutation

13-kbp retrotransposon insertion of a foamy-like endogenous retrovirus in intron 45 in the Black Telescope-eye strain. Almost no normal lrp2aL mRNA was expressed in the Black Telescope-eye strain.

Experimental Evidence

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

Main Reference

The Genetic Basis of Morphological Diversity in Domesticated Goldfish. (2020) (https://pubmed.ncbi.nlm.nih.gov/32392470)

Authors

Kon T; Omori Y; Fukuta K; Wada H; Watanabe M; Chen Z; Iwasaki M; Mishina T; Matsuzaki SS; Yoshihara D; Arakawa J; Kawakami K; Toyoda A; Burgess SM; Noguchi H; Furukawa T

Abstract

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Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@TE In mouse LRP2 plays the role of sonic hedgehog (shh) clearance receptor and regulates shh-induced cell proliferation at the retinal margin, resulting in a large eye phenotype.