

GEPHE SUMMARY

Hps4 (#Gephebase=Hps4#Gephebase-summary-title)	Gephebase Gene	GP00002122	GepheID
Published	Entry Status	Santos	Main curator

PHENOTYPIC CHANGE

Morphology (#Gephebase-summary-title)	Trait Category		
Coloration (albinism) (#Gephebase-summary-title)	Trait		
wildtype coloration	Trait State in Taxon A		
albino type	Trait State in Taxon B		
Taxon A	Ancestral State		
Intraspecific (#Gephebase-summary-title)	Taxonomic Status		

Taxon A		Taxon B	
	Latin Name		Latin Name
Ictalurus punctatus (#Gephebase-summary-title)	Ictalurus punctatus	Ictalurus punctatus (#Gephebase-summary-title)	Ictalurus punctatus
channel catfish	Common Name	channel catfish	Common Name
Silurus punctatus; channel catfish; Ictalurus punctatus (Rafinesque, 1818); Silurus punctatus Rafinesque, 1818; Ictalurus punetaus; letalurus punetaus; Letalurus punetaus	Synonyms	Silurus punctatus; channel catfish; Ictalurus punctatus (Rafinesque, 1818); Silurus punctatus Rafinesque, 1818; Ictalurus punetaus; letalurus punetaus; Letalurus punetaus	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Otomorpha; Ostariophysi; Otophysi; Characiphysae; Siluriformes; Siluroidei; Ictaluridae; Ictalurus	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Otomorpha; Ostariophysi; Otophysi; Characiphysae; Siluriformes; Siluroidei; Ictaluridae; Ictalurus	Lineage
Ictalurus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7997)	Parent	Ictalurus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7997)	Parent
7998 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7998)	NCBI Taxonomy ID	7998 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7998)	NCBI Taxonomy ID
No	is Taxon A an Infrappecies?	No	is Taxon B an Infrappecies?

GENOTYPIC CHANGE

Hps4	Generic Gene Name	Q99KG7 (http://www.uniprot.org/uniprot/Q99KG7)	UniProtKB Mus musculus
le; BLOC-3; AU040608; mKIAA1667; 2010205O06Rik; C130020P05Rik; Le	Synonyms	()	GenebankID or UniProtKB
10090.ENSMUSP00000047920 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000047920)	String		
-	Sequence Similarities		
GO:0046983 : protein dimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046983)	GO - Molecular Function		
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)			
GO:0017137 : Rab GTPase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0017137)			
GO:0005085 : guanyl-nucleotide exchange factor activity			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0005085>)

GO - Biological Process

GO:0007596 : blood coagulation (<https://www.ebi.ac.uk/QuickGO/term/GO:0007596>)

GO:0030318 : melanocyte differentiation

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)

GO:0050821 : protein stabilization (<https://www.ebi.ac.uk/QuickGO/term/GO:0050821>)

GO:0006605 : protein targeting (<https://www.ebi.ac.uk/QuickGO/term/GO:0006605>)

GO:0007040 : lysosome organization

(<https://www.ebi.ac.uk/QuickGO/term/GO:0007040>)

GO:1903232 : melanosome assembly (<https://www.ebi.ac.uk/QuickGO/term/GO:1903232>)

GO:0006996 : organelle organization

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006996>)

GO - Cellular Component

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)

GO:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)

GO:0031410 : cytoplasmic vesicle (<https://www.ebi.ac.uk/QuickGO/term/GO:0031410>)

GO:0005764 : lysosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0005764>)

GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)

GO:0031085 : BLOC-3 complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0031085>)

GO:0042827 : platelet dense granule (<https://www.ebi.ac.uk/QuickGO/term/GO:0042827>)

Presumptive Null

Yes ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title](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](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](https://www.gephebase.org/search-criteria?/and+Aberration+Type=~Deletion^#gephebase-summary-title))

Deletion Size

-

Molecular Details of the Mutation

A 99bp deletion was identified spanning intron 2 and exon 3 junction of the Hps4 gene

Experimental Evidence

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

Main Reference

A deletion in the Hermansky-Pudlak syndrome 4 (Hps4) gene appears to be responsible for albinism in channel catfish. (2017) (<https://pubmed.ncbi.nlm.nih.gov/28289846>)

Authors

Li Y; Geng X; Bao L; Elasad A; Huggins KW; Dunham R; Liu Z

Abstract

Albinism is caused by a series of genetic abnormalities leading to reduction of melanin production. Albinism is quite frequent in catfish, but the causative gene and the molecular basis were unknown. In this study, we conducted a genome-wide association study (GWAS) using the 250K SNP array. The GWAS analysis allowed mapping of the albino phenotype in the Hermansky-Pudlak syndrome 4 (Hps4) gene, which is known to be involved in melanosome biosynthesis. Sequencing analysis revealed that a 99-bp deletion was present in all analyzed albino catfish at the intron 2 and exon 3 junction. This deletion led to the skipping of the entire exon 3 which was confirmed by RT-PCR. Therefore, Hps4 was determined to be the candidate gene of the catfish albinism.

Additional References

RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS