

## GEPHE SUMMARY

	Gephebase Gene	GephelD
EDAR ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase=^EDAR^#gephebase-summary-title)	GP00002360	Main curator
Published	Entry Status	Courtier

## PHENOTYPIC CHANGE

	Trait Category		
Morphology, Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category=^Morphology^/and+Trait Category=^Physiology^#gephebase-summary-title)	Trait		
Scales (reduced) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Scales">https://www.gephebase.org/search-criteria?/and+Trait=^Scales</a> (reduced)^#gephebase-summary-title)	Trait State in Taxon A		
reduced scales	Trait State in Taxon B		
reduced scales	Ancestral State		
Taxon A	Taxonomic Status		
Interspecific ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status=^Interspecific^#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
Sinocyclocheilus grahami ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sinocyclocheilus+grahami^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sinocyclocheilus+grahami^#gephebase-summary-title</a> )		Sinocyclocheilus anshuiensis ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sinocyclocheilus+anshuiensis^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sinocyclocheilus+anshuiensis^#gephebase-summary-title</a> )	
-	Common Name	-	Common Name
	Synonyms		Synonyms
Barbus grahami; Barbus grahami Regan, 1904; Sinocyclocheilus grahami (Regan, 1904); BMNH:1904.1.26.27		Sinocyclocheilus anshuiensis Gan, Wu, Wei & Yang, 2013; KIZ 12060239; KIZ 12070271; KIZ 12070276; KIZ 12070277; KIZ 12070280; KIZ:12060239; KIZ:12070271; KIZ:12070276; KIZ:12070277; KIZ:12070280	
species	Rank		Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleoste; Osteoglossocephalai; Clupeocephala; Otomorpha; Ostariophysi; Otophysi; Cypriniphysae; Cypriniformes; Cyprinoidei; Cyprinidae; Cyprininae; Sinocyclocheilus	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleoste; Osteoglossocephalai; Clupeocephala; Otomorpha; Ostariophysi; Otophysi; Cypriniphysae; Cypriniformes; Cyprinoidei; Cyprinidae; Cyprininae; Sinocyclocheilus	
Sinocyclocheilus () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75365">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75365</a> )	Parent	Sinocyclocheilus () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75365">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75365</a> )	Parent
75366 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75366">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75366</a> )	NCBI Taxonomy ID	1608454 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=1608454">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=1608454</a> )	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?		is Taxon B an Infraspecies?
	No		

## GENOTYPIC CHANGE

	Generic Gene Name		
EDAR	Synonyms	UniProtKB Homo sapiens	
DL; ED3; ED5; ED1R; EDA3; HRM1; EDA1R; ECTD10A; ECTD10B; EDA-A1R	String	GenebankID or UniProtKB	
9606.ENSP00000258443 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000258443">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000258443</a> )	Sequence Similarities	0	
-	GO - Molecular Function		
GO:0004888 : transmembrane signaling receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004888">https://www.ebi.ac.uk/QuickGO/term/GO:0004888</a> )			
GO:0038023 : signaling receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0038023">https://www.ebi.ac.uk/QuickGO/term/GO:0038023</a> )			

## GO - Biological Process

GO:0030154 : cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030154>)  
 GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)  
 GO:0010628 : positive regulation of gene expression  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0010628>)  
 GO:0001942 : hair follicle development  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0001942>)  
 GO:0042475 : odontogenesis of dentin-containing tooth  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0042475>)  
 GO:0043123 : positive regulation of I-kappaB kinase/NF-kappaB signaling  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0043123>)  
 GO:1901224 : positive regulation of NIK/NF-kappaB signaling  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:1901224>)  
 GO:0060662 : salivary gland cavitation  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0060662>)  
 GO:0006915 : apoptotic process (<https://www.ebi.ac.uk/QuickGO/term/GO:0006915>)  
 GO:0008544 : epidermis development  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0008544>)  
 GO:0046330 : positive regulation of JNK cascade  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0046330>)  
 GO:0033209 : tumor necrosis factor-mediated signaling pathway  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0033209>)

## 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:0045177 : apical part of cell (<https://www.ebi.ac.uk/QuickGO/term/GO:0045177>)

Presumptive Null

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

Molecular Type

Coding ([#gepheebase-summary-title](https://www.gepheebase.org/search-criteria?/and+Molecular+Type=^Coding))

Aberration Type

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

Deletion Size

-

Molecular Details of the Mutation

Deletion of a large part of the EDAR2 coding region.

Experimental Evidence

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

Main Reference

The Sinocyclocheilus cavefish genome provides insights into cave adaptation. (2016) (<https://pubmed.ncbi.nlm.nih.gov/26728391>)

Authors

Yang J; Chen X; Bai J; Fang D; Qiu Y; Jiang W; Yuan H; Bian C; Lu J; He S; Pan X; Zhang Y; Wang X; You X; Wang Y; Sun Y; Mao D; Liu Y; Fan G; Zhang H; Chen X; Zhang X; Zheng L; Wang J; Cheng L; Chen J; Ruan Z; Li J; Yu H; Peng C; Ma X; Xu J; He Y; Xu Z; Xu P; Wang J; Yang H; Wang J; Whitten T; Xu X; Shi Q

Abstract

An emerging cavefish model, the cyprinid genus *Sinocyclocheilus*, is endemic to the massive southwestern karst area adjacent to the Qinghai-Tibetan Plateau of China. In order to understand whether orogeny influenced the evolution of these species, and how genomes change under isolation, especially in subterranean habitats, we performed whole-genome sequencing and comparative analyses of three species in this genus, *S. grahami*, *S. rhinocerous* and *S. anshuiensis*. These species are surface-dwelling, semi-cave-dwelling and cave-restricted, respectively.

The assembled genome sizes of *S. grahami*, *S. rhinocerous* and *S. anshuiensis* are 1.75 Gb, 1.73 Gb and 1.68 Gb, respectively. Divergence time and population history analyses of these species reveal that their speciation and population dynamics are correlated with the different stages of uplifting of the Qinghai-Tibetan Plateau. We carried out comparative analyses of these genomes and found that many genetic changes, such as gene loss (e.g. opsin genes), pseudogenes (e.g. crystallin genes), mutations (e.g. melanogenesis-related genes), deletions (e.g. scale-related genes) and down-regulation (e.g. circadian rhythm pathway genes), are possibly associated with the regressive features (such as eye degeneration, albinism, rudimentary scales and lack of circadian rhythms), and that some gene expansion (e.g. taste-related transcription factor gene) may point to the constructive features (such as enhanced taste buds) which evolved in these cave fishes.

As the first report on cavefish genomes among distinct species in *Sinocyclocheilus*, our work provides not only insights into genetic mechanisms of cave adaptation, but also represents a fundamental resource for a better understanding of cavefish biology.

Additional References

## RELATED GEPHE

## Related Genes

No matches found.

## Related Haplotypes

1 ([#gepheebase-summary-title](https://www.gepheebase.org/search-criteria?/or+Gene+Gepheebase=^EDAR/and+Taxon+ID=^75366/or+Gene+Gepheebase=^EDAR/and+Taxon+ID=^1608454))

## EXTERNAL LINKS

## COMMENTS

Deleterious mutations in the EDAR1 gene also.