

## GEPHE SUMMARY

opsin - rhodopsin1 (RH1) ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> )		Gephebase Gene	GP00000791	GepheID
Gephebase="opsin - rhodopsin1 (RH1)"#gephebase-summary-title)				Main curator
Published	Entry Status		Courtier	

## PHENOTYPIC CHANGE

Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> )		Trait Category		
Category="Physiology"#gephebase-summary-title)				
Color vision (blue-shift) ( <a color"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="Color</a> )		Trait		
vision (blue-shift)"#gephebase-summary-title)				
Other mammals	Trait State in Taxon A			
Cetaceans	Trait State in Taxon B			
Data not curated	Ancestral State			
Intergenic or Higher ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> )		Taxonomic Status		
Status="Intergenic or Higher"#gephebase-summary-title)				

Taxon A		Taxon B	
Latin Name	Latin Name	Latin Name	Latin Name
Mammalia	Cetacea		
( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> )	( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> )		
Synonyms="Mammalia"#gephebase-summary-title)	Synonyms="Cetacea"#gephebase-summary-title)		
mammals	Common Name	whales	Common Name
mammals	Synonyms	whales; cetaceans; whale; whales, dolphins, and porpoises	Synonyms
class	Rank	order	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla	Lineage
Amniota (amniotes) - (Rank: no rank)	Parent	Cetartiodactyla (whales, hippos, ruminants, pigs, camels etc.) - (Rank: no rank)	Parent
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=32524">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=32524</a> )	NCBI Taxonomy ID	( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=91561">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=91561</a> )	NCBI Taxonomy ID
40674		9721	
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=40674">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=40674</a> )	is Taxon A an Intraspecies?	( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9721">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9721</a> )	is Taxon B an Intraspecies?
No		No	

## GENOTYPIC CHANGE

RHO	Generic Gene Name	P08100 ( <a href="http://www.uniprot.org/uniprot/P08100">http://www.uniprot.org/uniprot/P08100</a> )	UniProtKB Homo sapiens
RP4; OPN2; CSNBAD1	Synonyms	AAO16237 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/AAO16237">https://www.ncbi.nlm.nih.gov/nuccore/AAO16237</a> )	GenebankID or UniProtKB
9606.ENSPO0000296271	String		
( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000296271">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000296271</a> )			
Belongs to the G-protein coupled receptor 1 family. Opsin subfamily.	Sequence Similarities		
GO:0046872 : metal ion binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046872">https://www.ebi.ac.uk/QuickGO/term/GO:0046872</a> )	GO - Molecular Function		
GO:0004930 : G protein-coupled receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004930">https://www.ebi.ac.uk/QuickGO/term/GO:0004930</a> )			
GO:0008020 : G protein-coupled photoreceptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008020">https://www.ebi.ac.uk/QuickGO/term/GO:0008020</a> )			
GO:0005502 : 11-cis retinal binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005502">https://www.ebi.ac.uk/QuickGO/term/GO:0005502</a> )	GO - Biological Process		

GO:0007186 : G protein-coupled receptor signaling pathway  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0007186)  
 GO:0001523 : retinoid metabolic process  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0001523)  
 GO:0006468 : protein phosphorylation  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0006468)  
 GO:0018298 : protein-chromophore linkage  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0018298)  
 GO:0007601 : visual perception (https://www.ebi.ac.uk/QuickGO/term/GO:0007601)  
 GO:0071482 : cellular response to light stimulus  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0071482)  
 GO:0007602 : phototransduction (https://www.ebi.ac.uk/QuickGO/term/GO:0007602)  
 GO:0016038 : absorption of visible light  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0016038)  
 GO:0045494 : photoreceptor cell maintenance  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0045494)  
 GO:0007603 : phototransduction, visible light  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0007603)  
 GO:0022400 : regulation of rhodopsin mediated signaling pathway  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0022400)  
 GO:0060041 : retina development in camera-type eye  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0060041)  
 GO:0016056 : rhodopsin mediated signaling pathway  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0016056)

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:0000139 : Golgi membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0000139)  
 GO:0005887 : integral component of plasma membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0005887)  
 GO:0005794 : Golgi apparatus (https://www.ebi.ac.uk/QuickGO/term/GO:0005794)  
 GO:0005911 : cell-cell junction (https://www.ebi.ac.uk/QuickGO/term/GO:0005911)  
 GO:0001750 : photoreceptor outer segment  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0001750)  
 GO:0097381 : photoreceptor disc membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0097381)  
 GO:0060170 : ciliary membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0060170)  
 GO:0030660 : Golgi-associated vesicle membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0030660)  
 GO:0001917 : photoreceptor inner segment  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0001917)  
 GO:0060342 : photoreceptor inner segment membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0060342)  
 GO:0042622 : photoreceptor outer segment membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0042622)

Mutation #1

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null="No"#gpebase-summary-title)

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type="Coding"#gpebase-summary-title)

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

Nonsynonymous

D83N + A292S both have an effect

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

Presumptive Null

Molecular Type

Aberration Type

SNP Coding Change

Molecular Details of the Mutation

Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Asp	Asn	83

Main Reference

Rod monochromacy and the coevolution of cetacean retinal opsins. (2013) (https://pubmed.ncbi.nlm.nih.gov/23637615)

Authors

Meredith RW; Gatesy J; Emerling CA; York VM; Springer MS

Abstract

Cetaceans have a long history of commitment to a fully aquatic lifestyle that extends back to the Eocene. Extant species have evolved a spectacular array of adaptations in conjunction with their deployment into a diverse array of aquatic habitats. Sensory systems are among those that have experienced radical transformations in the evolutionary history of this clade. In the case of vision, previous studies have demonstrated important changes in the genes encoding rod opsin (RH1), short-wavelength sensitive opsin 1 (SWS1), and long-wavelength sensitive opsin (LWS) in selected cetaceans, but have not examined the full complement of opsin genes across the complete range of cetacean families. Here, we report protein-coding sequences for RH1 and both color opsin genes (SWS1, LWS) from representatives of all extant cetacean families. We examine competing hypotheses pertaining to the timing of blue

shifts in RH1 relative to SWS1 inactivation in the early history of Cetacea, and we test the hypothesis that some cetaceans are rod monochromats. Molecular evolutionary analyses contradict the "coastal" hypothesis, wherein SWS1 was pseudogenized in the common ancestor of Cetacea, and instead suggest that RH1 was blue-shifted in the common ancestor of Cetacea before SWS1 was independently knocked out in baleen whales (Mysticeti) and in toothed whales (Odontoceti). Further, molecular evidence implies that LWS was inactivated convergently on at least five occasions in Cetacea: (1) Balaenidae (bowhead and right whales), (2) Balaenopteroidea (rorquals plus gray whale), (3) Mesoplodon bidens (Sowerby's beaked whale), (4) Physeter macrocephalus (giant sperm whale), and (5) Kogia breviceps (pygmy sperm whale). All of these cetaceans are known to dive to depths of at least 100 m where the underwater light field is dim and dominated by blue light. The knockout of both SWS1 and LWS in multiple cetacean lineages renders these taxa rod monochromats, a condition previously unknown among mammalian species.

Additional References

Mutation #2

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title>)

Presumptive Null

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

Molecular Type

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

Aberration Type

Nonsynonymous

SNP Coding Change

D83N + A292S both have an effect

Molecular Details of the Mutation

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

Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ala	Ser	292

Rod monochromacy and the coevolution of cetacean retinal opsins. (2013) (<https://pubmed.ncbi.nlm.nih.gov/23637615>)

Main Reference

Meredith RW; Gatesy J; Emerling CA; York VM; Springer MS

Authors

Cetaceans have a long history of commitment to a fully aquatic lifestyle that extends back to the Eocene. Extant species have evolved a spectacular array of adaptations in conjunction with their deployment into a diverse array of aquatic habitats. Sensory systems are among those that have experienced radical transformations in the evolutionary history of this clade. In the case of vision, previous studies have demonstrated important changes in the genes encoding rod opsin (RH1), short-wavelength sensitive opsin 1 (SWS1), and long-wavelength sensitive opsin (LWS) in selected cetaceans, but have not examined the full complement of opsin genes across the complete range of cetacean families. Here, we report protein-coding sequences for RH1 and both color opsin genes (SWS1, LWS) from representatives of all extant cetacean families. We examine competing hypotheses pertaining to the timing of blue shifts in RH1 relative to SWS1 inactivation in the early history of Cetacea, and we test the hypothesis that some cetaceans are rod monochromats. Molecular evolutionary analyses contradict the "coastal" hypothesis, wherein SWS1 was pseudogenized in the common ancestor of Cetacea, and instead suggest that RH1 was blue-shifted in the common ancestor of Cetacea before SWS1 was independently knocked out in baleen whales (Mysticeti) and in toothed whales (Odontoceti). Further, molecular evidence implies that LWS was inactivated convergently on at least five occasions in Cetacea: (1) Balaenidae (bowhead and right whales), (2) Balaenopteroidea (rorquals plus gray whale), (3) Mesoplodon bidens (Sowerby's beaked whale), (4) Physeter macrocephalus (giant sperm whale), and (5) Kogia breviceps (pygmy sperm whale). All of these cetaceans are known to dive to depths of at least 100 m where the underwater light field is dim and dominated by blue light. The knockout of both SWS1 and LWS in multiple cetacean lineages renders these taxa rod monochromats, a condition previously unknown among mammalian species.

Abstract

Additional References

## RELATED GEPHE

3 (opsin - (SWS1), opsin - rhodopsin (LWS), opsin - rhodopsin (MWS=duplicate of LWS)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^40674^/and+Trait=Color vision/or+Taxon ID=^9721^/and+Trait=Color vision/and+groupHaplotypes=true#gephebase-summary-title>)

Related Genes

6 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^opsin - rhodopsin1 \(RH1\)^/and+Taxon ID=^40674^/or+Gene Gephebase=^opsin - rhodopsin1 \(RH1\)^/and+Taxon ID=^9721^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^opsin - rhodopsin1 (RH1)^/and+Taxon ID=^40674^/or+Gene Gephebase=^opsin - rhodopsin1 (RH1)^/and+Taxon ID=^9721^#gephebase-summary-title))

Related Haplotypes

## EXTERNAL LINKS

## COMMENTS

@SeveralMutationsWithEffect

