GEPHE SUMMARY

Gephebase Gene
MC1R (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^MC1R^#gephebase-summary-title)

GepheID
GP00001466

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

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

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

Trait State in Taxon A
Philomachus pugnax - independent morph - brown

Trait State in Taxon B
Philomachus pugnax - satellite morph - white ruff and white tufts - dominant allele (supergene includes effects on testis size; body size; and behavior)

Ancestral State
Taxon A

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

Taxon A

Latin Name
Calidris pugnax (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Calidris pugnax^#gephebase-summary-title)

Common Name
ruff

Synonyms
Machetes pugnax; Pavoncella pugnax; Philomachus pugnax; ruff; Calidris pugnax (Linnaeus, 1758)

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Charadriiformes; Scolopacidae; Calidris

Parent
Calidris () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8918)

NCBI Taxonomy ID
198806 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 198806)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Calidris pugnax (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Calidris pugnax^#gephebase-summary-title)

Common Name
ruff

Synonyms
Machetes pugnax; Pavoncella pugnax; Philomachus pugnax; ruff; Calidris pugnax (Linnaeus, 1758)

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Charadriiformes; Scolopacidae; Calidris

Parent
Calidris () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8918)

NCBI Taxonomy ID
198806 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 198806)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
MC1R

Synonyms
CMM5; MSH-R; SHEP2; MSHR

String
9606.ENSP00000451605 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000451605)

Sequence Similarities
Belongs to the G-protein coupled receptor 1 family.

GO - Molecular Function
GO:0008528 : G protein-coupled peptide receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008528)
GO:0004977 : melanocortin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004977)

UniProtKB Homo sapiens
Q01726 (http://www.uniprot.org/uniprot/Q01726)

GenebankID or UniProtKB
106888292 (https://www.ncbi.nlm.nih.gov/nuccore/106888292)

GO:0004980 : melanocyte-stimulating hormone receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004980)
GO:0031625 : ubiquitin protein ligase binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0031625)

GO - Biological Process

GO:0007275 : multicellular organism development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007275)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0042438 : melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042438)
GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0007186 : G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007186)
GO:0051897 : positive regulation of protein kinase B signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0051897)
GO:0019233 : sensory perception of pain
(https://www.ebi.ac.uk/QuickGO/term/GO:0019233)
GO:0007189 : adenylate cyclase-activating G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007189)
GO:0035556 : intracellular signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0035556)
GO:0007187 : G protein-coupled receptor signaling pathway, coupled to cyclic nucleotide
second messenger (https://www.ebi.ac.uk/QuickGO/term/GO:0007187)
GO:0032720 : negative regulation of tumor necrosis factor production
(https://www.ebi.ac.uk/QuickGO/term/GO:0032720)
GO:0010739 : positive regulation of protein kinase A signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0010739)
GO:0090037 : positive regulation of protein kinase C signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0090037)
GO:0009650 : UV protection (https://www.ebi.ac.uk/QuickGO/term/GO:0009650)
GO:0070914 : UV-damage excision repair
(https://www.ebi.ac.uk/QuickGO/term/GO:0070914)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)

Presumptive Null

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

Nonsynonymous

Molecular Details of the Mutation

Several amino acid changes. The His207Arg substitution most likely has functional consequences because the same variant is associated with light color in the red-footed booby.

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	-	-	-

Main Reference

Structural genomic changes underlie alternative reproductive strategies in the ruff (Philomachus pugnax). (2016) (https://pubmed.ncbi.nlm.nih.gov/26569123)

Authors

Lamichhaney S; Fan G; Widemo F; Gunnarsson U; Thalmann DS; Hoeppner MP; Kerje S; Gustafson U; Shi C; Zhang H; Chen W; Liang X; Huang L; Wang J; Liang E; Wu Q; Lee SM; Xu X; HÃ¶glund J; Liu X; Andersson L

Abstract

The ruff is a Palearctic wader with a spectacular lekking behavior where highly ornamented males compete for females. This bird has one of the most remarkable mating systems in the animal kingdom, comprising three different male morphs (independents, satellites and faeders) that differ in behavior, plumage color and body size. Remarkably, the satellite and faeder morphs are controlled by dominant alleles. Here we have used whole-genome sequencing and resolved the enigma of how such complex phenotypic differences can have a simple genetic basis. The Satellite and Faeder alleles are both associated with a 4.5-Mb inversion that occurred about 3.8 million years ago. We propose an evolutionary scenario where the Satellite chromosome arose by a rare recombination event about 500,000 years ago. The ruff mating system is the result of an evolutionary process in which multiple genetic changes contributing to phenotypic differences between morphs have accumulated within the inverted region.

Additional References

A supergene determines highly divergent male reproductive morphs in the ruff. (2016) (https://pubmed.ncbi.nlm.nih.gov/26569125)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Supergene @Pleiotropy @SexualTrait @HeterozygoteAdvantate @Dominance @BalancingSelection. The mutation is within a 45.5-Mb-inverted region at position 10.2 Mb. This inverted region is associated with the distinction between "satellite"; "independent" and "faeder" morphs. The inversion disrupts the essential gene CENP-N is homozygous lethal.