GEPHE SUMMARY

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

GepheID
GP00000650

Entry Status
Published

Main curator
Martin

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
Coturnix japonica

Trait State in Taxon B
Coturnix japonica - lavender

Ancestral State
Data not curated

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

Taxon A

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

Common Name
Japanese quail

Synonyms
Coturnix coturnix Japonicus; Coturnix coturnix japanica; Coturnix coturnix japonica; Coturnix japonica japonica; Japanese quail; Coturnix japonica Temminck & Schlegel, 1849

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; Galloanserae; Galliformes; Phasianidae; Perdicinae; Coturnix

Parent
Coturnix () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9090)

NCBI Taxonomy ID
93934 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 93934)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
Japanese quail

Synonyms
Coturnix coturnix Japonicus; Coturnix coturnix japanica; Coturnix coturnix japonica; Coturnix japonica japonica; Japanese quail; Coturnix japonica Temminck & Schlegel, 1849

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; Galloanserae; Galliformes; Phasianidae; Perdicinae; Coturnix

Parent
Coturnix () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9090)

NCBI Taxonomy ID
93934 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 93934)

is Taxon B an Infraspecies?
Yes

Taxon B Description
Coturnix japonica - lavender

GENOTYPIC CHANGE

Generic Gene Name
Mlph

Synonyms
In; I1Rk3; Slac-2a; AW228792; D1Wsu84e; l(1)-3Rk; 2210418F23Rik; 5031433I09Rik; Ln; Slac2a

String
10090.ENSMUSP00000027528 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000027528)

Sequence Similarities
-

GO - Molecular Function
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0017137 : Rab GTPase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0017137)

UniProtKB Mus musculus
Q91V27 (http://www.uniprot.org/uniprot/Q91V27)

GenebankID or UniProtKB
AFR46004 (https://www.ncbi.nlm.nih.gov/nuccore/AFR46004)

GO:0003779 : actin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003779)
GO:0030674 : protein binding, bridging
(https://www.ebi.ac.uk/QuickGO/term/GO:0030674)
GO:0051010 : microtubule plus-end binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0051010)
GO:0017022 : myosin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0017022)
GO:0031489 : myosin V binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031489)

GO - Biological Process

GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0032400 : melanosome localization
(https://www.ebi.ac.uk/QuickGO/term/GO:0032400)
GO:0006605 : protein targeting (https://www.ebi.ac.uk/QuickGO/term/GO:0006605)

GO - Cellular Component

GO:0015629 : actin cytoskeleton (https://www.ebi.ac.uk/QuickGO/term/GO:0015629)
GO:0030425 : dendrite (https://www.ebi.ac.uk/QuickGO/term/GO:0030425)
GO:0048471 : perinuclear region of cytoplasm
(https://www.ebi.ac.uk/QuickGO/term/GO:0048471)
GO:0005815 : microtubule organizing center
(https://www.ebi.ac.uk/QuickGO/term/GO:0005815)
GO:0030864 : cortical actin cytoskeleton
(https://www.ebi.ac.uk/QuickGO/term/GO:0030864)
GO:0042470 : melanosome (https://www.ebi.ac.uk/QuickGO/term/GO:0042470)
GO:0001725 : stress fiber (https://www.ebi.ac.uk/QuickGO/term/GO:0001725)
GO:0016461 : unconventional myosin complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0016461)

Presumptive Null

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

Large deletions and rearrangement with pseudogenization of MLPH and loss of two other genes

Experimental Evidence

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

Main Reference

Plumage colour mutations and melanins in the feathers of the Japanese quail: a first comparison. (2009) (https://pubmed.ncbi.nlm.nih.gov/19496774)

Authors

Minvielle F; Cecchi T; Passamonti P; Gourichon D; Renieri C

Abstract

The absorbance of melanin content from dorsal feathers was compared between wild-type Japanese quail and nine other quail plumage colours determined by single mutations in one of seven genes: extended brown (MC1R), yellow (ASIP), silver (MITF), lavender (MLPH), roux (TYRP1), imperfect albinism (SLC45A2) and rusty. As compared with wild-type quail, all mutations but extended brown decreased total melanins. The largest decrease was observed in quail with one of the dilution mutations at TYRP1, MLPH or SLCA45A2. No difference in eumelanins was found between the 10 plumage colours. Despite visible colour differences, homozygous and heterozygous mutants at MITF, or the two imperfect albino (white) and cinnamon (pale yellow) alleles at SLC45A2, could not be differentiated on the basis of melanins. In contrast, the two white phenotypes caused by mutations at MITF and SLC45A2, or the two reddish plumage colours caused by the roux and rusty non-allelic mutations had different total melanin contents. The results showed that rusty was not likely to be a dilution mutation.

Additional References

The lavender plumage colour in Japanese quail is associated with a complex mutation in the region of MLPH that is related to differences in growth, feed consumption and body temperature. (2012) (https://pubmed.ncbi.nlm.nih.gov/22937744)

RELATED GEPHE

Related Genes

6 (Agouti, Endothelin receptor B, MC1R, Microphtalmia-associated transcription factor, SLC45A2=MATP, tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^93934^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Parallelism https://omia.org/OMIA001445/93934/