GEPHE SUMMARY

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

GepheID
GP00002250

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

Trait State in Taxon A
Domestic pigeon with bar plumage

Trait State in Taxon B
Barless ; autosomal recessive ; homozygotes have a high incidence of vision defects

Ancestral State
Taxon A

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

Taxon A

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

Common Name
rock pigeon

Synonyms
Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789

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; Columbiformes; Columbidae; Columba

Parent
Columba () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8931)

NCBI Taxonomy ID
8932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8932)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
rock pigeon

Synonyms
Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789

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; Columbiformes; Columbidae; Columba

Parent
Columba () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8931)

NCBI Taxonomy ID
8932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8932)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Ndp

Synonyms
ND; Ndph

String
10090.ENSMUSP00000046692 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000046692)

Sequence Similarities
-

GO - Molecular Function
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0005125 : cytokine activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005125)
GO:0005109 : frizzled binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005109)

GO - Biological Process

UniProtKB Mus musculus
P48744 (http://www.uniprot.org/uniprot/P48744)

GenebankID or UniProtKB
()

GO:0045893 : positive regulation of transcription, DNA-templated
(https://www.ebi.ac.uk/QuickGO/term/GO:0045893)
GO:0051091 : positive regulation of DNA-binding transcription factor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0051091)
GO:0016055 : Wnt signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0016055)
GO:0001890 : placenta development
(https://www.ebi.ac.uk/QuickGO/term/GO:0001890)
GO:0035426 : extracellular matrix-cell signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0035426)
GO:0110135 : Norrin signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0110135)
GO:0061299 : retina vasculature morphogenesis in camera-type eye
(https://www.ebi.ac.uk/QuickGO/term/GO:0061299)

GO - Cellular Component

GO:0062023 : collagen-containing extracellular matrix
(https://www.ebi.ac.uk/QuickGO/term/GO:0062023)
GO:0031012 : extracellular matrix (https://www.ebi.ac.uk/QuickGO/term/GO:0031012)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)
GO:0009986 : cell surface (https://www.ebi.ac.uk/QuickGO/term/GO:0009986)

Presumptive Null

Yes (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)

Aberration Type

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

SNP Coding Change

Nonsense

Molecular Details of the Mutation

Start-codon change predicted to truncate the amino terminus of the NDP protein by 11 amino acids thereby disrupting the 24-amino acid signal peptide sequence

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	Met	Thr	1

Main Reference

Introgression of regulatory alleles and a missense coding mutation drive plumage pattern diversity in the rock pigeon. (2018) (https://pubmed.ncbi.nlm.nih.gov/30014848)

Authors

Vickrey AI; Bruders R; Kronenberg Z; Mackey E; Bohlender RJ; Maclary ET; Maynez R; Osborne EJ; Johnson KP; Huff CD; Yandell M; Shapiro MD

Abstract

Birds and other vertebrates display stunning variation in pigmentation patterning, yet the genes controlling this diversity remain largely unknown. Rock pigeons (Columba livia) are fundamentally one of four color pattern phenotypes, in decreasing order of melanism: T-check, checker, bar (ancestral), or barless. Using whole-genome scans, we identified NDP as a candidate gene for this variation. Allele-specific expression differences in NDP indicate cis-regulatory divergence between ancestral and melanistic alleles. Sequence comparisons suggest that derived alleles originated in the speckled pigeon (Columba guinea), providing a striking example of introgression. In contrast, barless rock pigeons have an increased incidence of vision defects and, like human families with hereditary blindness, carry start-codon mutations in NDP. In summary, we find that both coding and regulatory variation in the same gene drives wing pattern diversity, and post-domestication introgression supplied potentially advantageous melanistic alleles to feral populations of this ubiquitous urban bird.

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Additional References

RELATED GEPHE

Related Genes

6 (MC1R, Mlana, slc2a11b, SLC45A2=MATP, SOX10, tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^8932^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^ndp (norrin)^/and+Taxon ID=^8932^/or+Gene Gephebase=^ndp (norrin)^/and+Taxon ID=^8932^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@Parallelism @Pleiotropy (deleterious pleiotropy with foggy vision as in human Norrie disease)