GEPHE SUMMARY

Gephebase Gene
MNR2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^MNR2^#gephebase-summary-title)

GepheID
GP00000675

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait #1

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

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

Trait State in Taxon A
Gallus gallus

Trait State in Taxon B
Gallus gallus - Rose Comb

Trait #2

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

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

Trait State in Taxon A
Gallus gallus

Trait State in Taxon B
Gallus gallus - Rose Comb with decreased sperm motility in homozygous males

Ancestral State
Taxon A

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

Taxon A

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

Common Name
chicken

Synonyms
Gallus gallus domesticus; chicken; bantam; chickens

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; Phasianinae; Gallus

Parent
Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)

NCBI Taxonomy ID
9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
chicken

Synonyms
Gallus gallus domesticus; chicken; bantam; chickens

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; Phasianinae; Gallus

Parent
Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)

NCBI Taxonomy ID
9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)

is Taxon B an Infraspecies?
Yes

Taxon B Description
Gallus gallus - Rose Comb

GENOTYPIC CHANGE

Generic Gene Name

MNR2

Synonyms

-

String

-

Sequence Similarities

-

GO - Molecular Function

GO:0043565 : sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043565)

GO - Biological Process

GO:0006355 : regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0006355)

GO - Cellular Component

GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)

UniProtKB Gallus gallus

Q9YHY8 (http://www.uniprot.org/uniprot/Q9YHY8)

GenebankID or UniProtKB

AF066860 (https://www.ncbi.nlm.nih.gov/nuccore/AF066860)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Inversion Size

>1 Mb

Molecular Details of the Mutation

7.4Mb inversion (and secondary structural re-arrangement in some individuals)

Experimental Evidence

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

Main Reference

The Rose-comb mutation in chickens constitutes a structural rearrangement causing both altered comb morphology and defective sperm motility. (2012) (https://pubmed.ncbi.nlm.nih.gov/22761584)

Authors

Imsland F; Feng C; Boije H; Bed'hom B; Fillon V; Dorshorst B; Rubin CJ; Liu R; Gao Y; Gu X; Wang Y; Gourichon D; Zody MC; Zecchin W; Vieaud A; Tixier-Boichard M; Hu X; HallbÃ¶Ã¶k F; Li N; Andersson L

Abstract

Rose-comb, a classical monogenic trait of chickens, is characterized by a drastically altered comb morphology compared to the single-combed wild-type. Here we show that Rose-comb is caused by a 7.4 Mb inversion on chromosome 7 and that a second Rose-comb allele arose by unequal crossing over between a Rose-comb and wild-type chromosome. The comb phenotype is caused by the relocalization of the MNR2 homeodomain protein gene leading to transient ectopic expression of MNR2 during comb development. We also provide a molecular explanation for the first example of epistatic interaction reported by Bateson and Punnett 104 years ago, namely that walnut-comb is caused by the combined effects of the Rose-comb and Pea-comb alleles. Transient ectopic expression of MNR2 and SOX5 (causing the Pea-comb phenotype) occurs in the same population of mesenchymal cells and with at least partially overlapping expression in individual cells in the comb primordium. Rose-comb has pleiotropic effects, as homozygosity in males has been associated with poor sperm motility. We postulate that this is caused by the disruption of the CCDC108 gene located at one of the inversion breakpoints. CCDC108 is a poorly characterized protein, but it contains a MSP (major sperm protein) domain and is expressed in testis. The study illustrates several characteristic features of the genetic diversity present in domestic animals, including the evolution of alleles by two or more consecutive mutations and the fact that structural changes have contributed to fast phenotypic evolution.

Additional References

RELATED GEPHE

Related Genes

4 (BMP2, HAO1, EOMES (eomesodermin), SOX5) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9031^/and+Trait=Bird head comb/or+Taxon ID=^9031^/and+Trait=Fertility/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Pleiotropy @Epistasis @HeterozygoteAdvantage https://omia.org/OMIA000884/9031/ Sperm motility defect may be due to inversion breakpoint coinciding with a gene (CCDC108) whose encoded peptide includes a major sperm protein domain