

# GEPHE SUMMARY

MNR2 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase="MNR2">#gephebase-summary-title)	Gephebase Gene GP00000675	GephelD
Published	Entry Status Martin	Main curator

## PHENOTYPIC CHANGE

Trait #1	Trait Category
Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category="Morphology">#gephebase-summary-title)	Trait
Bird head comb ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Bird+head+comb">#gephebase-summary-title)</a>	Trait State in Taxon A
Gallus gallus	Trait State in Taxon B
Gallus gallus - Rose Comb	

Trait #2	Trait Category
Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category="Physiology">#gephebase-summary-title)	Trait
Fertility (sperm motility) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Fertility+(sperm+motility)">https://www.gephebase.org/search-criteria?/and+Trait=^Fertility+(sperm+motility)</a> #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	

Taxon A	Ancestral State	Taxonomic Status
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status="Domesticated">#gephebase-summary-title)		
Taxon A	Latin Name	Taxon B
Gallus gallus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Gallus+gallus">#gephebase-summary-title)</a>	Gallus gallus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Gallus+gallus">#gephebase-summary-title)</a>	Latin Name
chicken	Common Name	Common Name
Gallus gallus domesticus; chicken; bantam; chickens	Synonyms	Synonyms
species	Rank	Rank
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	Lineage	Lineage
Gallus () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030</a> )	Parent	Parent
9031 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031</a> )	NCBI Taxonomy ID	NCBI Taxonomy ID
is Taxon A an Infraspecies?		is Taxon B an Infraspecies?
No	Yes	Taxon B Description
		Gallus gallus - Rose Comb

## GENOTYPIC CHANGE

MNR2	Generic Gene Name	UniProtKB Gallus gallus
-	Synonyms	GenebankID or UniProtKB
-	String	
-	Sequence Similarities	
-	GO - Molecular Function	
GO:0043565 : sequence-specific DNA binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043565">https://www.ebi.ac.uk/QuickGO/term/GO:0043565</a> )	GO - Biological Process	
GO:0006355 : regulation of transcription, DNA-templated ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0006355">https://www.ebi.ac.uk/QuickGO/term/GO:0006355</a> )	GO - Cellular Component	
GO:0005634 : nucleus ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005634">https://www.ebi.ac.uk/QuickGO/term/GO:0005634</a> )		Presumptive Null
No ( <a href="https://www.gephbase.org/search-criteria?/and+Presumptive Null=^No^#gephbase-summary-title">https://www.gephbase.org/search-criteria?/and+Presumptive Null=^No^#gephbase-summary-title</a> )		Molecular Type
Gene Amplification ( <a href="https://www.gephbase.org/search-criteria?/and+Molecular Type=^Gene Amplification^#gephbase-summary-title">https://www.gephbase.org/search-criteria?/and+Molecular Type=^Gene Amplification^#gephbase-summary-title</a> )		Aberration Type
Inversion ( <a href="https://www.gephbase.org/search-criteria?/and+Aberration Type=^Inversion^#gephbase-summary-title">https://www.gephbase.org/search-criteria?/and+Aberration Type=^Inversion^#gephbase-summary-title</a> )		Inversion Size
>1 Mb		
7.4Mb inversion (and secondary structural re-arrangement in some individuals)		Molecular Details of the Mutation
Linkage Mapping ( <a href="https://www.gephbase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephbase-summary-title">https://www.gephbase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephbase-summary-title</a> )		Experimental Evidence
The Rose-comb mutation in chickens constitutes a structural rearrangement causing both altered comb morphology and defective sperm motility. (2012) ( <a href="https://pubmed.ncbi.nlm.nih.gov/22761584">https://pubmed.ncbi.nlm.nih.gov/22761584</a> )		Main Reference
Imsland F; Feng C; Boije H; Bed'hom B; Fillon V; Dorhorst B; Rubin CJ; Liu R; Gao Y; Gu X; Wang Y; Gourichon D; Zody MC; Zecchin W; Vieaud A; Tixier-Boichard M; Hu X; Hallbäck F; Li N; Andersson L		Authors
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 relocation 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.	Abstract	
		Additional References

## RELATED GEPHE

4 (BMP2, HAO1, EOMES (eomesodermin), SOX5) ( <a href="https://www.gephbase.org/search-criteria?/or+Taxon ID=^9031^/and+Trait=Bird head comb/or+Taxon ID=^9031^/and+Trait=Fertility/and+groupHaplotypes=true#gephbase-summary-title">https://www.gephbase.org/search-criteria?/or+Taxon ID=^9031^/and+Trait=Bird head comb/or+Taxon ID=^9031^/and+Trait=Fertility/and+groupHaplotypes=true#gephbase-summary-title</a> )	Related Genes
No matches found.	Related Haplotypes

## 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

