GEPHE SUMMARY

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

Entry Status
Published

GepheID
GP00002194

Main curator
Martin

PHENOTYPIC CHANGE

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

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

Trait State in Taxon A
WT chickens

Trait State in Taxon B
Short-legged chicken (heterozygous ; mutation is embryonic lethal in the homozygous state)

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?
No

GENOTYPIC CHANGE

Generic Gene Name
IHH

Synonyms
-

String
9031.ENSGALP00000018493 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9031.ENSGALP00000018493)

Sequence Similarities
Belongs to the hedgehog family.

GO - Molecular Function
GO:0005509 : calcium ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005509)
GO:0005113 : patched binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005113)
GO:0008233 : peptidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008233)

GO - Biological Process
GO:0045944 : positive regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0045944)

UniProtKB Gallus gallus
Q98938 (http://www.uniprot.org/uniprot/Q98938)

GenebankID or UniProtKB
()

GO:0030166 : proteoglycan biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0030166)
GO:0007267 : cell-cell signaling (https://www.ebi.ac.uk/QuickGO/term/GO:0007267)
GO:0003382 : epithelial cell morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0003382)
GO:0035264 : multicellular organism growth
(https://www.ebi.ac.uk/QuickGO/term/GO:0035264)
GO:0010628 : positive regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010628)
GO:0051216 : cartilage development (https://www.ebi.ac.uk/QuickGO/term/GO:0051216)
GO:0001649 : osteoblast differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0001649)
GO:0001947 : heart looping (https://www.ebi.ac.uk/QuickGO/term/GO:0001947)
GO:0045669 : positive regulation of osteoblast differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0045669)
GO:0010468 : regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010468)
GO:0001708 : cell fate specification (https://www.ebi.ac.uk/QuickGO/term/GO:0001708)
GO:0009880 : embryonic pattern specification
(https://www.ebi.ac.uk/QuickGO/term/GO:0009880)
GO:0048469 : cell maturation (https://www.ebi.ac.uk/QuickGO/term/GO:0048469)
GO:0060591 : chondroblast differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0060591)
GO:0048557 : embryonic digestive tract morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0048557)
GO:0002053 : positive regulation of mesenchymal cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0002053)
GO:0048745 : smooth muscle tissue development
(https://www.ebi.ac.uk/QuickGO/term/GO:0048745)
GO:0032331 : negative regulation of chondrocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0032331)
GO:0045880 : positive regulation of smoothened signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0045880)
GO:0035988 : chondrocyte proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0035988)
GO:0007224 : smoothened signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007224)
GO:0016540 : protein autoprocessing
(https://www.ebi.ac.uk/QuickGO/term/GO:0016540)
GO:0003413 : chondrocyte differentiation involved in endochondral bone morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0003413)
GO:0048666 : neuron development (https://www.ebi.ac.uk/QuickGO/term/GO:0048666)
GO:0061053 : somite development (https://www.ebi.ac.uk/QuickGO/term/GO:0061053)
GO:0042733 : embryonic digit morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0042733)
GO:0046638 : positive regulation of alpha-beta T cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0046638)
GO:0033089 : positive regulation of T cell differentiation in thymus
(https://www.ebi.ac.uk/QuickGO/term/GO:0033089)
GO:0031016 : pancreas development (https://www.ebi.ac.uk/QuickGO/term/GO:0031016)
GO:0001569 : branching involved in blood vessel morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0001569)
GO:0048596 : embryonic camera-type eye morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0048596)
GO:0045453 : bone resorption (https://www.ebi.ac.uk/QuickGO/term/GO:0045453)
GO:0060220 : camera-type eye photoreceptor cell fate commitment
(https://www.ebi.ac.uk/QuickGO/term/GO:0060220)
GO:0072498 : embryonic skeletal joint development
(https://www.ebi.ac.uk/QuickGO/term/GO:0072498)
GO:0090136 : epithelial cell-cell adhesion
(https://www.ebi.ac.uk/QuickGO/term/GO:0090136)
GO:0060323 : head morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0060323)
GO:0046639 : negative regulation of alpha-beta T cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0046639)
GO:1903042 : negative regulation of chondrocyte hypertrophy
(https://www.ebi.ac.uk/QuickGO/term/GO:1903042)
GO:0048074 : negative regulation of eye pigmentation
(https://www.ebi.ac.uk/QuickGO/term/GO:0048074)
GO:0033088 : negative regulation of immature T cell proliferation in thymus
(https://www.ebi.ac.uk/QuickGO/term/GO:0033088)
GO:0010694 : positive regulation of alkaline phosphatase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0010694)
GO:0010770 : positive regulation of cell morphogenesis involved in differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0010770)
GO:0032967 : positive regulation of collagen biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0032967)
GO:0007228 : positive regulation of hh target transcription factor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0007228)

GO:1902738 : regulation of chondrocyte differentiation involved in endochondral bone morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:1902738)
GO:1902733 : regulation of growth plate cartilage chondrocyte differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:1902733)
GO:0003420 : regulation of growth plate cartilage chondrocyte proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0003420)
GO:0003406 : retinal pigment epithelium development (https://www.ebi.ac.uk/QuickGO/term/GO:0003406)
GO:0030704 : vitelline membrane formation (https://www.ebi.ac.uk/QuickGO/term/GO:0030704)

GO - Cellular Component

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

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Deletion Size

10-100 kb

Molecular Details of the Mutation

11,896 bp large deletion region (chr7: 21,798,705-21,810,600) covering the entire Indian hedgehog (IHH) gene

Experimental Evidence

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

Main Reference

Deletion of Indian hedgehog gene causes dominant semi-lethal Creeper trait in chicken. (2016) (https://pubmed.ncbi.nlm.nih.gov/27439785)

Authors

Jin S; Zhu F; Wang Y; Yi G; Li J; Lian L; Zheng J; Xu G; Jiao R; Gong Y; Hou Z; Yang N

Abstract

The Creeper trait, a classical monogenic phenotype of chicken, is controlled by a dominant semi-lethal gene. This trait has been widely cited in the genetics and molecular biology textbooks for illustrating autosomal dominant semi-lethal inheritance over decades. However, the genetic basis of the Creeper trait remains unknown. Here we have utilized ultra-deep sequencing and extensive analysis for targeting causative mutation controlling the Creeper trait. Our results indicated that the deletion of Indian hedgehog (IHH) gene was only found in the whole-genome sequencing data of lethal embryos and Creeper chickens. Large scale segregation analysis demonstrated that the deletion of IHH was fully linked with early embryonic death and the Creeper trait. Expression analysis showed a much lower expression of IHH in Creeper than wild-type chickens. We therefore suggest the deletion of IHH to be the causative mutation for the Creeper trait in chicken. Our findings unravel the genetic basis of the longstanding Creeper phenotype mystery in chicken as the same gene also underlies bone dysplasia in human and mouse, and thus highlight the significance of IHH in animal development and human haploinsufficiency disorders.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage https://omia.org/OMIA000006/9031/