GEPHE SUMMARY

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

GepheID
GP00002189

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

Trait State in Taxon A
Chicken with regular size

Trait State in Taxon B
Small-sized dwdw Connecticut females ; used for generation of heterozygotes of reduced size and feed consumption

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
GHR

Synonyms
GHBP; GHIP

String
9606.ENSP00000230882 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000230882)

Sequence Similarities
Belongs to the type I cytokine receptor family. Type 1 subfamily.

GO - Molecular Function
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0019901 : protein kinase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019901)

UniProtKB Homo sapiens
P10912 (http://www.uniprot.org/uniprot/P10912)

GenebankID or UniProtKB
()

GO:0017046 : peptide hormone binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0017046)
GO:0019955 : cytokine binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019955)
GO:0004896 : cytokine receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004896)
GO:0019838 : growth factor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019838)
GO:0004903 : growth hormone receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004903)
GO:0070064 : proline-rich region binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0070064)

GO - Biological Process

GO:0050731 : positive regulation of peptidyl-tyrosine phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0050731)
GO:0032870 : cellular response to hormone stimulus
(https://www.ebi.ac.uk/QuickGO/term/GO:0032870)
GO:0032355 : response to estradiol (https://www.ebi.ac.uk/QuickGO/term/GO:0032355)
GO:0040014 : regulation of multicellular organism growth
(https://www.ebi.ac.uk/QuickGO/term/GO:0040014)
GO:0006897 : endocytosis (https://www.ebi.ac.uk/QuickGO/term/GO:0006897)
GO:0048009 : insulin-like growth factor receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0048009)
GO:0006631 : fatty acid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006631)
GO:0040018 : positive regulation of multicellular organism growth
(https://www.ebi.ac.uk/QuickGO/term/GO:0040018)
GO:0006103 : 2-oxoglutarate metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006103)
GO:0042976 : activation of Janus kinase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0042976)
GO:0000187 : activation of MAPK activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0000187)
GO:0000255 : allantoin metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0000255)
GO:0006101 : citrate metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006101)
GO:0006600 : creatine metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006600)
GO:0046449 : creatinine metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0046449)
GO:0060396 : growth hormone receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0060396)
GO:0042445 : hormone metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042445)
GO:0006549 : isoleucine metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006549)
GO:0007259 : JAK-STAT cascade (https://www.ebi.ac.uk/QuickGO/term/GO:0007259)
GO:0060397 : JAK-STAT cascade involved in growth hormone signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0060397)
GO:0006107 : oxaloacetate metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006107)
GO:0046427 : positive regulation of JAK-STAT cascade
(https://www.ebi.ac.uk/QuickGO/term/GO:0046427)
GO:0042531 : positive regulation of tyrosine phosphorylation of STAT protein
(https://www.ebi.ac.uk/QuickGO/term/GO:0042531)
GO:0031623 : receptor internalization (https://www.ebi.ac.uk/QuickGO/term/GO:0031623)
GO:0046898 : response to cycloheximide
(https://www.ebi.ac.uk/QuickGO/term/GO:0046898)
GO:0006105 : succinate metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006105)
GO:0019530 : taurine metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0019530)
GO:0006573 : valine metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006573)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0043235 : receptor complex (https://www.ebi.ac.uk/QuickGO/term/GO:0043235)
GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
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)
GO:0036464 : cytoplasmic ribonucleoprotein granule
(https://www.ebi.ac.uk/QuickGO/term/GO:0036464)
GO:0009897 : external side of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0009897)

GO:0070195 : growth hormone receptor complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0070195)

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

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

Deletion Size

1-10 kb

Molecular Details of the Mutation

deletion of 1773 bp in the 3′ end of the coding region

Experimental Evidence

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

Main Reference

Dysfunctional growth hormone receptor in a strain of sex-linked dwarf chicken: evidence for a mutation in the intracellular domain. (1994) (https://pubmed.ncbi.nlm.nih.gov/7964293)

Authors

Agarwal SK; Cogburn LA; Burnside J

Abstract

The sex-linked dwarf (dwdw) chicken represents a valuable animal model for studying GH insensitivity and the consequence of mutations in the GH receptor (GHR) gene. We have recently reported undetectable hepatic GH-binding activity and an aberrantly sized transcript in a strain of dwdw chickens obtained from Arbor Acre Farms, Inc. (Glastonbury, CT, USA). Southern blot analysis of the chicken GHR (cGHR) gene revealed a restriction-fragment length polymorphism in HindIII and EcoRI digests of genomic DNA in this strain of dwdw chicken. In order to localize the molecular mutation, we analysed the gene structure and determined the complete sequence of the 3′ untranslated region (3′ UTR) of the normal cGHR. With the use of this information, we located a large deletion in the 3′ end of the cGHR gene of the Connecticut (CT) strain of dwdw chicken. This deletion (1773 bp) contained 27 highly conserved amino acids of the 3′ end of the coding region, the in-frame stop codon, a less frequently used poly(A) signal that is normally found 445 bp downstream of the stop codon, and a large portion of the 3′ UTR. Because of this deletion, 27 novel amino acids were substituted and the open reading frame was extended for an additional 26 amino acids before reaching the transcriptional termination site. The predicted amino acid sequence of the novel carboxyl-terminus of the dwdw cGHR is largely hydrophobic with a polylysine tail, whereas the carboxyl-terminus of the wild-type (DwDw) cGHR is composed of hydrophilic amino acids.(ABSTRACT TRUNCATED AT 250 WORDS)

Additional References

RELATED GEPHE

Related Genes

3 (miR-15a-16, TMEM263, RB1) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9031^/and+Trait=Body size/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA000309/9031/