

GEPHE SUMMARY

| | | | |
|---|----------------|------------|--------------|
| TMEM263 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^TMEM263^#gephebase-summary-title) | Gephebase Gene | GP00002161 | GepheID |
| Published | Entry Status | Martin | Main curator |

PHENOTYPIC CHANGE

| | | | |
|--|------------------------|--|--|
| Physiology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Physiology^#gephebase-summary-title) | Trait Category | | |
| Body size (dwarfism) (https://www.gephebase.org/search-criteria?/and+Trait=^Body size (dwarfism)^#gephebase-summary-title) | Trait | | |
| White Leghorn ; normal size | Trait State in Taxon A | | |
| Dwarf chickens adw from Cornell K-strain of White Leghorns ; autosomal recessive | Trait State in Taxon B | | |
| | Ancestral State | | |
| Taxon A | Taxonomic Status | | |
| Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=^Domesticated^#gephebase-summary-title) | | | |

| Taxon A | Latin Name | Taxon B | Latin Name |
|--|---|--|---|
| Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Gallus gallus^#gephebase-summary-title) | Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Gallus gallus^#gephebase-summary-title) | Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Gallus gallus^#gephebase-summary-title) | Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Gallus gallus^#gephebase-summary-title) |
| chicken | Common Name | chicken | Common Name |
| Gallus gallus domesticus; chicken; bantam; chickens | Synonyms | Gallus gallus domesticus; chicken; bantam; chickens | Synonyms |
| species | Rank | species | 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 | 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 |
| Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030) | Parent | Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030) | Parent |
| 9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031) | NCBI Taxonomy ID | 9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031) | NCBI Taxonomy ID |
| No | is Taxon A an Intraspecies? | No | is Taxon B an Intraspecies? |

GENOTYPIC CHANGE

| | | | |
|---|-------------------------|--|-------------------------|
| - | Generic Gene Name | 0 | UniProtKB |
| - | Synonyms | Q8WUH6 (https://www.ncbi.nlm.nih.gov/nucore/Q8WUH6) | GenebankID or UniProtKB |
| - | String | | |
| - | Sequence Similarities | | |
| - | GO - Molecular Function | | |
| - | GO - Biological Process | | |
| - | GO - Cellular Component | | |
| - | | | 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

g.53688583C>T c.433G>A p.Trp59â—

Experimental Evidence

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

| | Taxon A | Taxon B | Position |
|------------|---------|---------|----------|
| Codon | - | - | - |
| Amino-acid | Trp | STP | 59 |

A Novel Loss-of-Function Variant in Transmembrane Protein 263 (TMEM263) of Autosomal Dwarfism in Chicken. (2018) (<https://pubmed.ncbi.nlm.nih.gov/29930570>)

Main Reference

Wu Z; Derks MFL; Dibbits B; Megens HJ; Groenen MAM; Crooijmans RPMA

Authors

Autosomal dwarfism (adw) in chickens is a growth deficiency caused by a recessive mutation. Characteristic for adw is an approximately 30% growth reduction with short shank. The adw variant was first recognized in the Cornell K-strain of White Leghorns, but the genetic causal variant remained unknown. To identify the causal variant underlying the adw phenotype, fine mapping was conducted on chromosome 1, within 52-56 Mb. This region was known to harbor the causal variant from previous linkage studies. We compared whole-genome sequence data of this region from normal-sized and adw chickens in order to find the unique causal variant. We identified a novel nonsense mutation NP_001006244.1:p.(Trp59), in the transmembrane protein 263 gene (TMEM263), completely associated with adw. The nonsense mutation truncates the transmembrane protein within the membrane-spanning domain, expected to cause a dysfunctional protein. TMEM263 is reported to be associated with bone mineral deposition in humans, and the protein shows interaction with growth hormone 1 (GH1). Our study presents molecular genetic evidence for a novel loss-of-function variant, which likely alters body growth and development in autosomal dwarf chicken.

Abstract

Additional References

RELATED GEPHE

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

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS