

GEPHE SUMMARY

TMEM263 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+TMEM263+gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+TMEM263+gephebase-summary-title</a> )	Gephebase Gene	GP00002161	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category+Physiology+gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category+Physiology+gephebase-summary-title</a> )	Trait Category		
Body size (dwarfism) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Body+size+(dwarfism)+gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Body+size+(dwarfism)+gephebase-summary-title</a> )	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 ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated+gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated+gephebase-summary-title</a> )			

Taxon A	Latin Name	Taxon B	Latin Name
Gallus gallus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Gallus+gallus+gephebase-summary-title">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">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">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">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Gallus+gallus+gephebase-summary-title</a> )
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) ( <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	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
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	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
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

-	Generic Gene Name	0	UniProtKB
-	Synonyms	Q8WUH6 ( <a href="https://www.ncbi.nlm.nih.gov/nucore/Q8WUH6">https://www.ncbi.nlm.nih.gov/nucore/Q8WUH6</a> )	GenebankID or UniProtKB
-	String		
-	Sequence Similarities		
-	GO - Molecular Function		
-	GO - Biological Process		
-	GO - Cellular Component		
-			Presumptive Null
Yes ( <a href="https://www.gephebase.org/search-criteria?/and+Presumptive+Null+Yes+gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Presumptive+Null+Yes+gephebase-summary-title</a> )			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; Dibbitts 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