

GEPHE SUMMARY

	Gephebase Gene	GephelD
FGF20 (https://www.gephebase.org/search-criteria/?and+Gene Gephebase=^FGF20^#gephebase-summary-title)	GP00002171	Main curator
Published	Entry Status	Martin

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria/?and+Trait Category=^Morphology^#gephebase-summary-title)	Trait		
Organ loss (feathers ; scales) (https://www.gephebase.org/search-criteria/?and+Trait=^Organ+loss+(feathers+;+scales)^#gephebase-summary-title)	Trait State in Taxon A		
WT	Trait State in Taxon B		
almost complete absence of feathers and scales (recessive sc mutation)	Ancestral State		
Taxon A	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria/?and+Taxonomic Status=^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)	
chicken	Common Name		Common Name
Gallus gallus domesticus; chicken; bantam; chickens	Synonyms		Synonyms
species	Rank		Rank
	Lineage		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		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	
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 Infraspecies?	Yes	is Taxon B an Infraspecies?
			Taxon B Description
			Israeli experimental line ; Storrs Connecticut (low line) ; UC Davis lines

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Homo sapiens
FGF20		
RHDA2; FGF-20	Synonyms	GenebankID or UniProtKB
9606.ENSP00000180166 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000180166)	String	
Belongs to the heparin-binding growth factors family.	Sequence Similarities	
	GO - Molecular Function	
GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)		
GO:0005102 : signaling receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005102)		
GO:0005104 : fibroblast growth factor receptor binding		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0005104>)
 GO:0043395 : heparan sulfate proteoglycan binding
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0043395>)
 GO:0090722 : receptor-receptor interaction
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0090722>)

GO - Biological Process

GO:0007165 : signal transduction (<https://www.ebi.ac.uk/QuickGO/term/GO:0007165>)
 GO:0043524 : negative regulation of neuron apoptotic process
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0043524>)
 GO:0070374 : positive regulation of ERK1 and ERK2 cascade
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0070374>)
 GO:0000165 : MAPK cascade (<https://www.ebi.ac.uk/QuickGO/term/GO:0000165>)
 GO:0008284 : positive regulation of cell proliferation
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0008284>)
 GO:0051897 : positive regulation of protein kinase B signaling
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0051897>)
 GO:0007267 : cell-cell signaling (<https://www.ebi.ac.uk/QuickGO/term/GO:0007267>)
 GO:0008543 : fibroblast growth factor receptor signaling pathway
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0008543>)
 GO:0060113 : inner ear receptor cell differentiation
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0060113>)
 GO:1904340 : positive regulation of dopaminergic neuron differentiation
 (<https://www.ebi.ac.uk/QuickGO/term/GO:1904340>)
 GO:0060043 : regulation of cardiac muscle cell proliferation
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0060043>)
 GO:0014059 : regulation of dopamine secretion
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0014059>)

GO - Cellular Component

GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)

Presumptive Null

Yes ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=%27Yes%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive%20Null=%27Yes%27#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=%27Coding%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular%20Type=%27Coding%27#gephebase-summary-title))

Aberration Type

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type=%27SNP%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration%20Type=%27SNP%27#gephebase-summary-title))

SNP Coding Change

Nonsense

Molecular Details of the Mutation

g.62878803A>T c.535A>T p.R179*

Experimental Evidence

Association Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=%27Association Mapping%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental%20Evidence=%27Association%20Mapping%27#gephebase-summary-title))

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	STP	179

Main Reference

Genome-wide SNP scan of pooled DNA reveals nonsense mutation in FGF20 in the scaleless line of featherless chickens. (2012) (<https://pubmed.ncbi.nlm.nih.gov/22712610>)

Authors

Wells KL; Hadad Y; Ben-Avraham D; Hillel J; Cahaner A; Headon DJ

Abstract

Scaleless (*sc/sc*) chickens carry a single recessive mutation that causes a lack of almost all body feathers, as well as foot scales and spurs, due to a failure of skin patterning during embryogenesis. This spontaneous mutant line, first described in the 1950s, has been used extensively to explore the tissue interactions involved in ectodermal appendage formation in embryonic skin. Moreover, the trait is potentially useful in tropical agriculture due to the ability of featherless chickens to tolerate heat, which is at present a major constraint to efficient poultry meat production in hot climates. In the interests of enhancing our understanding of feather placode development, and to provide the poultry industry with a strategy to breed heat-tolerant meat-type chickens (broilers), we mapped and identified the *sc* mutation.

Through a cost-effective and labour-efficient SNP array mapping approach using DNA from *sc/sc* and *sc/+* blood sample pools, we map the *sc* trait to chromosome 4 and show that a nonsense mutation in FGF20 is completely associated with the *sc/sc* phenotype. This mutation, common to all *sc/sc* individuals and absent from wild type, is predicted to lead to loss of a highly conserved region of the FGF20 protein important for FGF signalling. *In situ* hybridisation and quantitative RT-PCR studies reveal that FGF20 is epidermally expressed during the early stages of feather placode patterning. In addition, we describe a dCAPS genotyping assay based on the mutation, developed to facilitate discrimination between wild type and *sc* alleles.

This work represents the first loss of function genetic evidence supporting a role for FGF ligand signalling in feather development, and suggests FGF20 as a novel central player in the development of vertebrate skin appendages, including hair follicles and exocrine glands. In addition, this is to our knowledge the first report describing the use of the chicken SNP array to map genes based on genotyping of DNA samples from pooled whole blood. The identification of the *sc* mutation has important implications for the future breeding of this potentially useful trait for the poultry industry, and our genotyping assay can facilitate its rapid introgression into production lines.

Additional References

RELATED GEPHE

No matches found.

Related Genes

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA000889/9031/>