GEPHE SUMMARY

Gephebase Gene

SGK3 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SGK3^#gephebase-summary-title)

GepheID

GP00002263

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

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

Trait State in Taxon A

WT coat

Trait State in Taxon B

Hairless dog breed - Scottish Deerhound (recessive)

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name

Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus familiaris^#gephebase-summary-title)

Common Name

dog

Synonyms

Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758

Rank

subspecies

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Canis; Canis lupus

Parent

Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)

NCBI Taxonomy ID

9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus familiaris^#gephebase-summary-title)

Common Name

dog

Synonyms

Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758

Rank

subspecies

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Canis; Canis lupus

Parent

Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)

NCBI Taxonomy ID

9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

Sgk3

Synonyms

fy; fz; Cisk; 2510015P22Rik; A330005P07Rik; Sgkl

String

10090.ENSMUSP00000126861 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000126861)

Sequence Similarities

Belongs to the protein kinase superfamily. AGC Ser/Thr protein kinase family.

GO - Molecular Function

GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)
GO:0004674 : protein serine/threonine kinase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004674)
GO:0035091 : phosphatidylinositol binding (https://www.ebi.ac.uk/QuickGO/term/GO:0035091)
GO:0015459 : potassium channel regulator activity

UniProtKB Mus musculus

Q9ERE3 (http://www.uniprot.org/uniprot/Q9ERE3)

GenebankID or UniProtKB

()

(https://www.ebi.ac.uk/QuickGO/term/GO:0015459)

GO - Biological Process

GO:0018105 : peptidyl-serine phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0018105)
GO:0035556 : intracellular signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0035556)
GO:2001240 : negative regulation of extrinsic apoptotic signaling pathway in absence of
ligand (https://www.ebi.ac.uk/QuickGO/term/GO:2001240)

GO - Cellular Component

GO:0005654 : nucleoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005654)
GO:0005769 : early endosome (https://www.ebi.ac.uk/QuickGO/term/GO:0005769)
GO:0031410 : cytoplasmic vesicle (https://www.ebi.ac.uk/QuickGO/term/GO:0031410)
GO:0043231 : intracellular membrane-bounded organelle
(https://www.ebi.ac.uk/QuickGO/term/GO:0043231)
GO:0055037 : recycling endosome (https://www.ebi.ac.uk/QuickGO/term/GO:0055037)

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

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

Insertion Size

1-9 bp

Molecular Details of the Mutation

c.137_138insT p.Glu47GlyfsTer3

Experimental Evidence

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

Main Reference

A frameshift insertion in SGK3 leads to recessive hairlessness in Scottish Deerhounds: a candidate gene for human alopecia conditions. (2019) (https://pubmed.ncbi.nlm.nih.gov/30927068)

Authors

HytÃ¶nen MK; Lohi H

Abstract

Hairlessness is a breed-specific feature selected for in some dog breeds but a rare abnormality in some others such as Scottish Deerhounds (SD). In SDs, the affected puppies are born with sparse hair but lose it within the first 2Â months leaving the dogs completely hairless. The previous studies have implicated variants in FOXI3 and SGK3 in hairlessness; however, the known variants do not explain hairlessness in all breeds such as SDs. We investigated the genetic cause in 66 SDs, including a litter with two hairless dogs. We utilized a combined approach of genome-wide homozygosity mapping and whole-genome sequencing of a hairless SD followed by recessive filtering according to a recessive model against 340 control genomes. Only two homozygous-coding variants were discovered in the homozygosity regions, including a 1-bp insertion in exon 2 of SGK3. This results in a predicted frameshift and very early truncation (49/490 amino acids) of the SGK3 protein. Additional screening of the recessive variant demonstrated a full segregation with the hairlessness and a 12% carrier frequency in the SD breed. The variant was not found in the related Irish Wolfhound breed. This study identifies the second hairless variant in the SGK3 gene in dogs and further highlights its role as a candidate gene for androgen-independent hair loss or alopecia in human.

Additional References

Whole Genome Analysis of a Single Scottish Deerhound Dog Family Provides Independent Corroboration That a SGK3 Coding Variant Leads to Hairlessness. (2020) (https://pubmed.ncbi.nlm.nih.gov/31727632)

RELATED GEPHE

Related Genes

4 (FGF5, R-spondin-2 (RSPO2), FOXI3, KRT71) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9615^/and+Trait=Hair/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^SGK3^/and+Taxon ID=^9615^/or+Gene Gephebase=^SGK3^/and+Taxon ID=^9615^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@AllelicSeries https://omia.org/OMIA001279/9615/ GWAS-like analysis on pedigreed family in 2020