GEPHE SUMMARY

Gephebase Gene

KRT71 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^KRT71^#gephebase-summary-title)

GepheID

GP00001729

Entry Status

Published

Main curator

Courtier

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

Rattus norvegicus - various breeds

Trait State in Taxon B

Rattus norvegicus - Rex strain KFRS5A/Kyo - curly hair (heteroygous mutant) or hairless (homozygous mutant)

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

Norway rat

Synonyms

rat; rats; Norway rat; brown rat; Rattus norvegicus8; Rattus norwegicus

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Rattus

Parent

Rattus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10114)

NCBI Taxonomy ID

10116 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10116)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

Norway rat

Synonyms

rat; rats; Norway rat; brown rat; Rattus norvegicus8; Rattus norwegicus

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Rattus

Parent

Rattus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10114)

NCBI Taxonomy ID

10116 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10116)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

Krt71

Synonyms

Ca; Cu; Cal4; mK6irs; Krt2-6g; mK6irs1; AA589543; K6irs1; Kb34; Krt6g

String

10090.ENSMUSP00000023710 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000023710)

Sequence Similarities

Belongs to the intermediate filament family.

GO - Molecular Function

GO:0005198 : structural molecule activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005198)

GO - Biological Process

GO:0031069 : hair follicle morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0031069)
GO:0045109 : intermediate filament organization

UniProtKB Mus musculus

Q9R0H5 (http://www.uniprot.org/uniprot/Q9R0H5)

GenebankID or UniProtKB

()

(https://www.ebi.ac.uk/QuickGO/term/GO:0045109)

GO - Cellular Component

GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)

GO:0045095 : keratin filament (https://www.ebi.ac.uk/QuickGO/term/GO:0045095)

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

1-9 bp

Molecular Details of the Mutation

a 7-bp deletion at the splicing acceptor site of intron 1. The deletion provoked a 6-amino acid in-frame deletion (p.Val149_Gln154del) in the alpha-helical rod domain of KRT71 protein

Experimental Evidence

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

Main Reference

Identification of the rat Rex mutation as a 7-bp deletion at splicing acceptor site of the Krt71 gene. (2010) (https://pubmed.ncbi.nlm.nih.gov/20179389)

Authors

Kuramoto T; Hirano R; Kuwamura M; Serikawa T

Abstract

The rat autosomal dominant Rex (Re) mutation on chromosome 7 causes curly hair in Re/+ and hair loss in Re/Re rats. Histopathologically, the Re/+ rat showed dilatation of the hair follicle and hairs with irregularly-coated cuticles, and the Re/Re rat showed more severe effects. We identified Re as a 7-bp deletion at the splicing acceptor site of intron 1 of the keratin 71 (Krt71) gene, which is located within the Re critical chromosomal region and plays an important role in hair formation. The deletion provoked a 6-amino acid in-frame deletion (p.Val149_Gln154del) in the alpha-helical rod domain of KRT71 protein. Identification of the Re mutation (Krt71(Re)) enables us to further understand the biological function of KRT71.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Splicing Acceptor Site