

GEPHE SUMMARY

KRT71 (<a +krt71+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+KRT71+"#gephebase-summary-title)	Gephebase Gene	GP00001729	GepheID
Published	Entry Status	Courtier	Main curator

PHENOTYPIC CHANGE

Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title)	Trait Category		
Hair type (curly and hairless) (<a +hair+type+(curly+and+hairless)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Hair+type+(curly+and+hairless)+"#gephebase-summary-title)	Trait		
Rattus norvegicus - various breeds	Trait State in Taxon A		
Rattus norvegicus - Rex strain KFRS5A/Kyo - curly hair (heteroygous mutant) or hairless (homozygous mutant)	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (<a +domesticated+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated+"#gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Rattus norvegicus (<a +rattus+norvegicus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Rattus+norvegicus+"#gephebase-summary-title)	Latin Name	Rattus norvegicus (<a +rattus+norvegicus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Rattus+norvegicus+"#gephebase-summary-title)	Latin Name
Norway rat	Common Name	Norway rat	Common Name
rat; rats; Norway rat; brown rat; Rattus norvegicus8; Rattus norvegicus	Synonyms	rat; rats; Norway rat; brown rat; Rattus norvegicus8; Rattus norvegicus	Synonyms
species	Rank	species	Rank
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	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	Lineage
Rattus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10114)	Parent	Rattus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10114)	Parent
10116 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10116)	NCBI Taxonomy ID	10116 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10116)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

Krt71	Generic Gene Name	Q9R0H5 (http://www.uniprot.org/uniprot/Q9R0H5)	UniProtKB Mus musculus
Ca; Cu; Cal4; mK6irs; Krt2-6g; mK6irs1; AA589543; K6irs1; Kb34; Krt6g	Synonyms	()	GenebankID or UniProtKB
10090.ENSMUSP00000023710 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000023710)	String		
Belongs to the intermediate filament family.	Sequence Similarities		
GO:0005198 : structural molecule activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005198)	GO - Molecular Function		
GO:0031069 : hair follicle morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0031069)	GO - Biological Process		
GO:0045109 : intermediate filament organization			

(<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](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](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](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](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

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

@Splicing Acceptor Site