

GEPHE SUMMARY

	Gephebase Gene	GephelD
Agouti (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Agouti^#gephebase-summary-title)	GP00000067	
	Entry Status	Main curator
Published	Martin	

PHENOTYPIC CHANGE

	Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)	Trait
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^#gephebase-summary-title)	Trait State in Taxon A
Vulpes vulpes domesticus	Trait State in Taxon B
Vulpes vulpes domesticus - black	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
Vulpes vulpes (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Vulpes+vulpes^#gephebase-summary-title)	Common Name	Vulpes vulpes (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Vulpes+vulpes^#gephebase-summary-title)	Common Name
red fox	Synonyms	red fox	Synonyms
Canis vulpes; red fox; silver fox	Rank	Canis vulpes; red fox; silver fox	Rank
species	Lineage	species	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; Vulpes	Parent	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; Vulpes	Parent
Vulpes () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9625)	NCBI Taxonomy ID	Vulpes () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9625)	NCBI Taxonomy ID
9627 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9627)		9627 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9627)	
No	is Taxon A an Infraspecies?		is Taxon B an Infraspecies?
-	No		

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Mus musculus
Asip		
As; ASP; Aγ; ASIP; a	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000029123 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)	String	CAB58365 (https://www.ncbi.nlm.nih.gov/nuccore/CAB58365)
-	Sequence Similarities	
GO:0031779 : melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031779)	GO - Molecular Function	
GO:0031781 : type 3 melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031781)		
GO:0031782 : type 4 melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031782)		
		GO - Biological Process

GO:0008343 : adult feeding behavior
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0008343>)
 GO:0006091 : generation of precursor metabolites and energy
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0006091>)
 GO:0071514 : genetic imprinting (<https://www.ebi.ac.uk/QuickGO/term/GO:0071514>)
 GO:0009755 : hormone-mediated signaling pathway
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0009755>)
 GO:0042438 : melanin biosynthetic process
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
 GO:0032438 : melanosome organization
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)
 GO:0032402 : melanosome transport
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0032402>)
 GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
 GO:0048023 : positive regulation of melanin biosynthetic process
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0048023>)
 GO:0040030 : regulation of molecular function, epigenetic
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0040030>)

GO - Cellular Component

GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
 GO:0005623 : cell (<https://www.ebi.ac.uk/QuickGO/term/GO:0005623>)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Deletion Size

100-999 bp

Molecular Details of the Mutation

166 bp deletion in exon 1

Experimental Evidence

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

Main Reference

A non-epistatic interaction of agouti and extension in the fox, *Vulpes vulpes*. (1997) (<https://pubmed.ncbi.nlm.nih.gov/9054949>)

Authors

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Abstract

Agouti and extension are two genes that control the production of yellow-red (phaeomelanin) and brown-black (eumelanin) pigments in the mammalian coat. Extension encodes the melanocyte-stimulating hormone receptor (MC1R) while agouti encodes a peptide antagonist of the receptor. In the mouse, extension is epistatic to agouti, hence dominant mutants of the MC1R encoding constitutively active receptors are not inhibited by the agouti antagonist, and animals with dominant alleles of both loci remain darkly pigmented. In the fox the proposed extension locus is not epistatic to the agouti locus. We have cloned and characterized the MC1R and the agouti gene in coat colour variants of the fox (*Vulpes vulpes*). A constitutively activating C125R mutation in the MC1R was found specifically in darkly pigmented animals carrying the Alaska Silver allele (EA). A deletion in the first coding exon of the agouti gene was found associated with the proposed recessive allele of agouti in the darkly pigmented Standard Silver fox (aa). Thus, as in the mouse, dark pigmentation can be caused by a constitutively active MC1R, or homozygous recessive status at the agouti locus. Our results, demonstrating the presence of dominant extension alleles in foxes with significant red coat colouration, suggest the ability of the fox agouti protein to counteract the signalling activity of a constitutively active fox MC1R.

Additional References

RELATED GEPHE

Related Genes

2 (Kit (type III receptor protein-tyrosine kinase), MC1R) (<https://www.gephbase.org/search-criteria?/or+TaxonID=%279627%27/and+Trait=Coloration/and+groupHaplotypes=true#gephbase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omnia.org/OMIA000201/9627/>

