

## GEPHE SUMMARY

	Gephebase Gene	GephelD
Agouti (ASIP) ( <a href="https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Agouti (ASIP)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Agouti (ASIP)^#gephebase-summary-title</a> )	GP00002107	Main curator
Published	Entry Status	Courtier

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title</a> )	Trait Category
Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^#gephebase-summary-title</a> )	Trait
	Trait State in Taxon A
-	Trait State in Taxon B
heterozygote A(vy)/a: yellow fur; obesity; diabetes and increased susceptibility to tumours -- isogenic A(vy): coats that vary in a continuous spectrum from full yellow; through variegated yellow/agouti; to full agouti (pseudoagouti)	Ancestral State

Taxon A	Taxonomic Status
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title</a> )	

Taxon A	Latin Name	Taxon B	Latin Name
Mus musculus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Mus musculus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Mus musculus^#gephebase-summary-title</a> )		Mus musculus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Mus musculus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Mus musculus^#gephebase-summary-title</a> )	
house mouse	Common Name	house mouse	Common Name
house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid	Synonyms	house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid	Synonyms
species	Rank	species	Rank
	Lineage		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; Mus; Mus		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; Mus; Mus	
Mus () - (Rank: subgenus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507</a> )	Parent	Mus () - (Rank: subgenus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507</a> )	Parent
10090 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090</a> )	NCBI Taxonomy ID	10090 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090</a> )	NCBI Taxonomy ID
	is Taxon A an Infraspecies?		is Taxon B an Infraspecies?
No		No	

## GENOTYPIC CHANGE

Asip	Generic Gene Name	UniProtKB Mus musculus
As; ASP; A<y>; ASIP; a	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000029123 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123</a> )	String	0
	Sequence Similarities	
GO:0031779 : melanocortin receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031779">https://www.ebi.ac.uk/QuickGO/term/GO:0031779</a> )	GO - Molecular Function	
GO:0031781 : type 3 melanocortin receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031781">https://www.ebi.ac.uk/QuickGO/term/GO:0031781</a> )		
GO:0031782 : type 4 melanocortin receptor binding		

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

No (<https://www.gephebase.org/search-criteria?/and+Presumptive+Null=^No^#gephebase-summary-title>)

Molecular Type

Cis-regulatory (<https://www.gephebase.org/search-criteria?/and+Molecular+Type=^Cis-regulatory^#gephebase-summary-title>)

Aberration Type

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

Insertion Size

1-10 kb

Molecular Details of the Mutation

insertion of an intra-cisternal A particle (IAP) retrotransposon upstream of the agouti gene (A). This activates transcription and causes ectopic expression of agouti protein; resulting in yellow fur; obesity; diabetes and increased susceptibility to tumours. The pleiotropic effects of ectopic agouti expression are presumably due to effects of the paracrine signal on other tissues.

Experimental Evidence

Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=^Candidate+Gene^#gephebase-summary-title>)

Main Reference

Epigenetic inheritance at the agouti locus in the mouse. (1999) (<https://pubmed.ncbi.nlm.nih.gov/10545949>)

Authors

Morgan HD; Sutherland HG; Martin DI; Whitelaw E

Abstract

Epigenetic modifications have effects on phenotype, but they are generally considered to be cleared on passage through the germ line in mammals, so that only genetic traits are inherited.

Here we describe the inheritance of an epigenetic modification at the agouti locus in mice. In viable yellow (A<sup>vy</sup>/a) mice, transcription originating in an intra-cisternal A particle (IAP) retrotransposon inserted upstream of the agouti gene (A) causes ectopic expression of agouti protein, resulting in yellow fur, obesity, diabetes and increased susceptibility to tumours. The pleiotropic effects of ectopic agouti expression are presumably due to effects of the paracrine signal on other tissues. Avy mice display variable expressivity because they are epigenetic mosaics for activity of the retrotransposon: isogenic Avy mice have coats that vary in a continuous spectrum from full yellow, through variegated yellow/agouti, to full agouti (pseudoagouti). The distribution of phenotypes among offspring is related to the phenotype of the dam; when an A<sup>vy</sup> dam has the agouti phenotype, her offspring are more likely to be agouti. We demonstrate here that this maternal epigenetic effect is not the result of a maternally contributed environment. Rather, our data show that it results from incomplete erasure of an epigenetic modification when a silenced Avy allele is passed through the female germ line, with consequent inheritance of the epigenetic modification. Because retrotransposons are abundant in mammalian genomes, this type of inheritance may be common.

Additional References

## RELATED GEPHE

Related Genes

4 (Agouti, MC1R, PMEL17, SLC45A2=MATP) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=^10090^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

@TE - Maternal @Epigenetics effect resulting from incomplete erasure of an epigenetic modification when a silenced Avy allele is passed through the female germ line; with consequent inheritance of the epigenetic modification

