

## GEPHE SUMMARY

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

## PHENOTYPIC CHANGE

<p>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</a>)</p> <p>Coloration (coat) (<a +coloration+(coat)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration+(coat)^#gephebase-summary-title</a>)</p> <p>-</p> <p>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)</p> <p>Taxon A</p> <p>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</a>)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>-</p> <p>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)</p> <p>-</p> <p>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</a>)</p>	<p>-</p> <p>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)</p> <p>-</p> <p>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</a>)</p>
<p>Taxon A</p> <p>Mus musculus (<a +mus+musculus^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Mus+musculus^#gephebase-summary-title</a>)</p> <p>house mouse</p> <p>house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid</p> <p>species</p> <p>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</p> <p>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>)</p> <p>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>)</p> <p>No</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Infrasppecies?</p>	<p>Taxon B</p> <p>Mus musculus (<a +mus+musculus^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Mus+musculus^#gephebase-summary-title</a>)</p> <p>house mouse</p> <p>house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid</p> <p>species</p> <p>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</p> <p>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>)</p> <p>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>)</p> <p>No</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Infrasppecies?</p>

## GENOTYPIC CHANGE

<p>Asip</p> <p>As; ASP; A&lt;y&gt;; ASIP; a</p> <p>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>)</p> <p>-</p> <p>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>)</p> <p>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>)</p> <p>GO:0031782 : type 4 melanocortin receptor binding</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>Q03288 (<a href="http://www.uniprot.org/uniprot/Q03288">http://www.uniprot.org/uniprot/Q03288</a>)</p> <p>0</p> <p>-</p> <p>-</p>	<p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
--	--	--	--

(<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

No ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null+No))

Molecular Type

Cis-regulatory ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=Cis-regulatory))

Aberration Type

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

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

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#gpepbase-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

