GEPHE SUMMARY

Gephebase Gene
Agouti (ASIP)

GephelD
GP00002107

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat)

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

Taxon A

Latin Name
Mus musculus

Common Name
house mouse

Synonyms
house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid

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; Mus; Mus

Parent
Mus () - (Rank: subgenus)

NCBI Taxonomy ID
10090

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Mus musculus

Common Name
house mouse

Synonyms
house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid

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; Mus; Mus

Parent
Mus () - (Rank: subgenus)

NCBI Taxonomy ID
10090

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Asip

Synonyms
As; ASP; A<y>; ASIP; a

String
10090.ENSMUSP00000029123

Sequence Similarities
-

GO - Molecular Function
GO:0031779 : melanocortin receptor binding
GO:0031781 : type 3 melanocortin receptor binding
GO:0031782 : type 4 melanocortin receptor binding

GO - Biological Process
GO:0008343 : adult feeding behavior
GO:0006091 : generation of precursor metabolites and energy
GO:0071514 : genetic imprinting
GO:0009755 : hormone-mediated signaling pathway

UniProtKB Mus musculus
Q03288

GenebankID or UniProtKB

GO:0042438 : melanin biosynthetic process
GO:0032438 : melanosome organization
GO:0032402 : melanosome transport
GO:0043473 : pigmentation
GO:0048023 : positive regulation of melanin biosynthetic process
GO:0040030 : regulation of molecular function, epigenetic

GO - Cellular Component
GO:0005576 : extracellular region
GO:0005623 : cell

Presumptive Null
No

Molecular Type
Cis-regulatory

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

Main Reference
Epigenetic inheritance at the agouti locus in the mouse. (1999)

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(vy)/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(vy) 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
3 (MC1R, PMEL17, SLC45A2=MATP)
Related Haplotypes
No matches found.

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