

Yes (<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>)

Aberration Type

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

Deletion Size

100-999 bp

Molecular Details of the Mutation

126-bp deletion overlapping with end of exon 4 and resulting in missplicing. Linkage analysis and genomic studies have shown that *Bombyx arylalkylamine-N-acetyl transferase*, the homologous gene (*Dat*) that converts dopamine into N-acetyl dopamine, encodes a precursor of N-acetyl dopamine, sclerotin in *Drosophila* and it is the gene responsible for *mln*.

Experimental Evidence

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

Main Reference

Disruption of an N-acetyltransferase gene in the silkworm reveals a novel role in pigmentation. (2010) (<https://pubmed.ncbi.nlm.nih.gov/21062865>)

Authors

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Abstract

The pigmentation of insects has served as an excellent model for the study of morphological trait evolution and developmental biology. The melanism (*mln*) mutant of the silkworm *Bombyx mori* is notable for its strong black coloration, phenotypic differences between larval and adult stages, and its widespread use in strain selection. Here, we report the genetic and molecular bases for the formation of the *mln* morphological trait. Fine mapping revealed that an arylalkylamine N-acetyltransferase (*AANAT*) gene co-segregates with the black coloration patterns. Coding sequence variations and expression profiles of *AANAT* are also associated with the melanic phenotypes. A 126 bp deletion in the *mln* genome causes two alternatively spliced transcripts with premature terminations. An enzymatic assay demonstrated the absolute loss of *AANAT* activity in the mutant proteins. We also performed RNA interference of *AANAT* in wild-type pupae and observed a significant proportion of adults with ectopic black coloration. These findings indicate that functional deletion of this *AANAT* gene accounts for the *mln* mutation in silkworm. *AANAT* is also involved in a parallel melanin synthesis pathway in which *ebony* plays a role, whereas no pigmentation defect has been reported in the *Drosophila* model or in other insects to date. To the best of our knowledge, the *mln* mutation is the first characterized mutant phenotype of insects with *AANAT*, and this result contributes to our understanding of dopamine metabolism and melanin pattern polymorphisms.

Additional References

Mutations of an arylalkylamine-N-acetyltransferase, *Bm-iAANAT*, are responsible for silkworm melanism mutant. (2010) (<https://pubmed.ncbi.nlm.nih.gov/20332088>)

RELATED GEPHE

Related Genes

9 (apontic-like, cardinal, cortex, SCARB1, SCRB15, Wnt1, Carotenoid-binding protein (CBP), Tyrosine hydroxylase, UGT86 (*Bm-UGT10286*)) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=~7091^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

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