GEPHE SUMMARY

Gephebase Gene

tryptophan phenylalanine hydroxylase (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^tryptophan phenylalanine hydroxylase^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00002003

Main curator

Courtier

PHENOTYPIC CHANGE

Trait Category

Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait

Enzymatic activity (https://www.gephebase.org/search-criteria?/and+Trait=^Enzymatic activity^#gephebase-summary-title)

Trait State in Taxon A

Drosophila melanogaster - wild-type

Trait State in Taxon B

Drosophila melanogaster - reduced activity

Ancestral State

Taxon A

Taxonomic Status

Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name

Drosophila melanogaster (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Drosophila melanogaster^#gephebase-summary-title)

Common Name

fruit fly

Synonyms

Sophophora melanogaster; fruit fly; Drosophila melanogaster Meigen, 1830; Sophophora melanogaster (Meigen, 1830); Drosophila melangaster

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Ecdysozoa; Panarthropoda; Arthropoda; Mandibulata; Pancrustacea; Hexapoda; Insecta; Dicondylia; Pterygota; Neoptera; Holometabola; Diptera; Brachycera; Muscomorpha; Eremoneura; Cyclorrhapha; Schizophora; Acalyptratae; Ephydroidea; Drosophilidae; Drosophilinae; Drosophilini; Drosophila; Sophophora; melanogaster group; melanogaster subgroup

Parent

melanogaster subgroup () - (Rank: species subgroup) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32351)

NCBI Taxonomy ID

7227 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7227)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Drosophila melanogaster (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Drosophila melanogaster^#gephebase-summary-title)

Common Name

fruit fly

Synonyms

Sophophora melanogaster; fruit fly; Drosophila melanogaster Meigen, 1830; Sophophora melanogaster (Meigen, 1830); Drosophila melangaster

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Ecdysozoa; Panarthropoda; Arthropoda; Mandibulata; Pancrustacea; Hexapoda; Insecta; Dicondylia; Pterygota; Neoptera; Holometabola; Diptera; Brachycera; Muscomorpha; Eremoneura; Cyclorrhapha; Schizophora; Acalyptratae; Ephydroidea; Drosophilidae; Drosophilinae; Drosophilini; Drosophila; Sophophora; melanogaster group; melanogaster subgroup

Parent

melanogaster subgroup () - (Rank: species subgroup) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32351)

NCBI Taxonomy ID

7227 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7227)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

Hn

Synonyms

bu; CG7399; Dmel\CG7399; DTPH; DTPHu; pah; Pah; PAH; Tph; TpH; TPH; Trh; TRH

String

7227.FBpp0076523 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 7227.FBpp0076523)

Sequence Similarities

Belongs to the biopterin-dependent aromatic amino acid hydroxylase family.

GO - Molecular Function

GO:0005506 : iron ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005506)
GO:0004505 : phenylalanine 4-monooxygenase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004505)

UniProtKB Drosophila melanogaster

P17276 (http://www.uniprot.org/uniprot/P17276)

GenebankID or UniProtKB

()

GO:0004510 : tryptophan 5-monooxygenase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004510)

GO - Biological Process

GO:0007616 : long-term memory (https://www.ebi.ac.uk/QuickGO/term/GO:0007616)
GO:0006726 : eye pigment biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006726)
GO:0006559 : L-phenylalanine catabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006559)
GO:0042427 : serotonin biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042427)
GO:0006571 : tyrosine biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006571)

GO - Cellular Component

-

Presumptive Null

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

Molecular Type

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#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 the transposable element B104/roo in the exon 3 of the Phenylalanine hydroxylase gene. Its presence alters the Phenylalanine hydroxylase splicing pattern; producing at least two aberrant mRNAs which contain part of the B104 sequence interrupting the coding region. This aberrant splicing is provoked by the use of a cryptic donor site encoded by the B104 3' long terminal repeat in combination with either the gene intron 3 acceptor site or a novel acceptor site generated by the target duplication caused by transposition. One of them; referred as mRNA type 1; encodes a truncated protein that could be predictably non-functional. In mRNA type 2; in spite of a 42 nt insertion; the Phenylalanine hydroxylase reading frame is not altered and it would encode for a protein with 14 extra amino acids which would be able to account for the low enzyme activity detected in this mutant.

Experimental Evidence

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

Main Reference

Aberrant splicing of the Drosophila melanogaster phenylalanine hydroxylase pre-mRNA caused by the insertion of a B104/roo transposable element in the Henna locus. (1999) (https://pubmed.ncbi.nlm.nih.gov/10333570)

Authors

Ruiz-VÃizquez P; Silva FJ

Abstract

We report the insertion of the transposable element B104 in the Phenylalanine hydroxylase gene of the Drosophila mutant Henna-recessive 3. Its presence alters the Phenylalanine hydroxylase splicing pattern, producing at least two aberrant mRNAs which contain part of the B104 sequence interrupting the coding region. This aberrant splicing is provoked by the use of a cryptic donor site encoded by the B104 3' long terminal repeat in combination with either the gene intron 3 acceptor site or a novel acceptor site generated by the target duplication caused by transposition. One of them, referred as mRNA type 1, encodes a truncated protein that could be predictably non-functional. In mRNA type 2, in spite of a 42 nt insertion, the Phenylalanine hydroxylase reading frame is not altered and it would encode for a protein with 14 extra amino acids which would be able to account for the low enzyme activity detected in this mutant. These results demonstrated that Henna locus encodes the enzyme phenylalanine hydroxylase providing direct evidence of its participation in pteridine synthesis. Moreover, it constitutes an example of the ability of transposable elements to generate protein variation in populations with the evolutionary consequences that this implies.

Additional References

RELATED GEPHE

Related Genes

3 (Dopa oxidase-3 (Dox-3), glycerol-3-phosphate dehydrogenase (Gpdh), prophenoloxidase 1 (PPO1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^7227^/and+Trait=Enzymatic activity/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@TE @Splicing - http://flybase.org/reports/FBal0101652