

## GEPHE SUMMARY

	Gephebase Gene	GephelD
tryptophan phenylalanine hydroxylase ( <a href="https://www.gephebase.org/search-criteria?/and+Gene Gephebase^tryptophan phenylalanine hydroxylase^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene Gephebase^tryptophan phenylalanine hydroxylase^#gephebase-summary-title</a> )	GP00002003	
	Entry Status	Main curator
Published	Courtier	

## PHENOTYPIC CHANGE

	Trait Category
Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait Category^Physiology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait Category^Physiology^#gephebase-summary-title</a> )	
Enzymatic activity ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Enzymatic activity^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Enzymatic activity^#gephebase-summary-title</a> )	Trait
Drosophila melanogaster - wild-type	Trait State in Taxon A
Drosophila melanogaster - reduced activity	Trait State in Taxon B
Taxon A	Ancestral State
Intraspecific ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic Status^Intraspecific^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic Status^Intraspecific^#gephebase-summary-title</a> )	Taxonomic Status

Taxon A	Latin Name	Taxon B	Latin Name
Drosophila melanogaster ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms^Drosophila melanogaster^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms^Drosophila melanogaster^#gephebase-summary-title</a> )		Drosophila melanogaster ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms^Drosophila melanogaster^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms^Drosophila melanogaster^#gephebase-summary-title</a> )	
fruit fly	Common Name	fruit fly	Common Name
Sophophora melanogaster; fruit fly; Drosophila melanogaster Meigen, 1830; Sophophora melanogaster (Meigen, 1830); Drosophila melangaster	Synonyms	Sophophora melanogaster; fruit fly; Drosophila melanogaster Meigen, 1830; Sophophora melanogaster (Meigen, 1830); Drosophila melangaster	Synonyms
species	Lineage	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; Ephydrioidea; Drosophilidae; Drosophilinae; Drosophilini; Drosophila; Sophophora; melanogaster group; melanogaster subgroup		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; Ephydrioidea; Drosophilidae; Drosophilinae; Drosophilini; Drosophila; Sophophora; melanogaster group; melanogaster subgroup	
melanogaster subgroup () - (Rank: species subgroup) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32351">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32351</a> )	Parent	melanogaster subgroup () - (Rank: species subgroup) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32351">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32351</a> )	Parent
7227 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7227">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7227</a> )	NCBI Taxonomy ID	7227 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7227">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7227</a> )	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

## GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Drosophila melanogaster
Hn		
bu; CG7399; Dmel\CG7399; DTPH; DTPHu; pah; Pah; PAH; Tph; TpH; TPH; Trh; TRH	Synonyms	
7227.FBpp0076523 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 7227.FBpp0076523">http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 7227.FBpp0076523</a> )	String	
Belongs to the bipterin-dependent aromatic amino acid hydroxylase family.	Sequence Similarities	
GO - Molecular Function		
GO:0005506 : iron ion binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005506">https://www.ebi.ac.uk/QuickGO/term/GO:0005506</a> )		
GO:0004505 : phenylalanine 4-monoxygenase activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004505">https://www.ebi.ac.uk/QuickGO/term/GO:0004505</a> )		

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.gepheebase.org/search-criteria?/and+Presumptive+Null=^No^#gepheebase-summary-title>)

Molecular Type

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

Aberration Type

Insertion (<https://www.gepheebase.org/search-criteria?/and+Aberration+Type=^Insertion^#gepheebase-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.gepheebase.org/search-criteria?/and+Experimental+Evidence=^Candidate+Gene^#gepheebase-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

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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.gepheebase.org/search-criteria?/or+TaxonID=^7227^/and+Trait=Enzymatic+activity/and+groupHaplotypes=true#gepheebase-summary-title>)

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

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