

## GEPHE SUMMARY

|  |   |                                    |
|--|---|------------------------------------|
| <p>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>)</p> <p>Published</p> | <p>Gephebase Gene</p> <p>GP00002003</p> <p>Courtier</p> <p>Entry Status</p> | <p>GepheID</p> <p>Main curator</p> |
|--|---|------------------------------------|

## PHENOTYPIC CHANGE

|  |   |  |  |
|--|---|--|--|
| <p>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>)</p> <p>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>)</p> <p>Drosophila melanogaster - wild-type</p> <p>Drosophila melanogaster - reduced activity</p> <p>Taxon A</p> <p>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>)</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>Taxon A</p> <p>Latin Name</p> <p>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>)</p> <p>Common Name</p> <p>fruit fly</p> <p>Synonyms</p> <p>Sophophora melanogaster; fruit fly; Drosophila melanogaster Meigen, 1830; Sophophora melanogaster (Meigen, 1830); Drosophila melangaster</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p>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>)</p> <p>NCBI Taxonomy ID</p> <p>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>)</p> <p>is Taxon A an Infrappecies?</p> <p>No</p> | <p>Taxon B</p> <p>Latin Name</p> <p>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>)</p> <p>Common Name</p> <p>fruit fly</p> <p>Synonyms</p> <p>Sophophora melanogaster; fruit fly; Drosophila melanogaster Meigen, 1830; Sophophora melanogaster (Meigen, 1830); Drosophila melangaster</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p>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>)</p> <p>NCBI Taxonomy ID</p> <p>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>)</p> <p>is Taxon B an Infrappecies?</p> <p>No</p> |
|--|---|--|--|

## GENOTYPIC CHANGE

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|--|--|
| <p>Hn</p> <p>bu; CG7399; Dmel\CG7399; DTPH; DTPHu; pah; Pah; PAH; Tph; TpH; TPH; Trh; TRH</p> <p>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>)</p> <p>Sequence Similarities</p> <p>Belongs to the bipterin-dependent aromatic amino acid hydroxylase family.</p> <p>GO - Molecular Function</p> <p>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>)</p> <p>GO:0004505 : phenylalanine 4-monooxygenase activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004505">https://www.ebi.ac.uk/QuickGO/term/GO:0004505</a>)</p> | <p>Generic Gene Name</p> <p>P17276 (<a href="http://www.uniprot.org/uniprot/P17276">http://www.uniprot.org/uniprot/P17276</a>)</p> <p>String</p> <p>UniProtKB Drosophila melanogaster</p> <p>GenebankID or UniProtKB</p> |
|--|--|

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

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Presumptive Null

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No^#gephebase-summary-title](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](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](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](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-Vázquez 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](https://www.gephebase.org/search-criteria?/or+TaxonID=~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>