

Phenylalanine hydroxylase (PAH) mutation map

revised October 2003 by:
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462 mutations identified to date

- Missense mutation
 - Nonsense mutation
 - Silent mutation
 - △ Deletion of codon
 - ▽ Splice junction mutation
 - fs Frame shift (See database for nucleotide numbers, eg. P81fs as the result of c.266_267insC)
 - ★ CpG site
 - ☒ de novo mutation
 - del Deletion
 - ins Insertion
 - [H] Haplotype
 - + New RE site
 - # Unclassified haplotype
 - M1V etc. Met. to Val. substitution in codon 1
- Note** the exons represented as lines (L3 exons in total) separating the numbered introns.

