Phenylalanine hydroxylase (PAH) mutation map

528 mutations identified to date:
- Missense mutation
- Nonsense mutation
- Silent mutation
- Deletion of codon
- Splice junction-mutation
- Frame shift (use database for suitable protein, e.g. PAH is the result of 3.2kb/351491136)
- Q85X
- de novo mutation
- In Deletion
- P1 Haplotype
- New RE site
- Unclassified haplotype

5'UTR 3'UTR

M1V etc.

Q20X

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Note

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