PAH Mutation Analysis Consortium Database: 1997. Pro mutation databases

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Citation Report

#	Article	IF	Citations
1	Alterations in protein aggregation and degradation due to mild and severe missense mutations (A104D,) Tj ETQq0 0 0 rgBT /Overlock 10	2.5	29
2	A European Multicenter Study of Phenylalanine Hydroxylase Deficiency: Classification of 105 Mutations and a General System for Genotype-Based Prediction of Metabolic Phenotype. American Journal of Human Genetics, 1998, 63, 71-79.	6.2	310
3	A Second Locus for Familial High Myopia Maps to Chromosome 12q. American Journal of Human Genetics, 1998, 63, 1419-1424.	6.2	243
4	Missense mutations in the phenylalanine hydroxylase gene (PAH) can cause accelerated proteolytic turnover of PAH enzyme: A mechanism underlying phenylketonuria. Journal of Inherited Metabolic Disease, 1999, 22, 208-212.	3.6	32
5	Genomics, mutations and the Internet: The naming and use of parts. Journal of Inherited Metabolic Disease, 1999, 22, 519-530.	3.6	8
6	Monogenic traits are not simple: lessons from phenylketonuria. Trends in Genetics, 1999, 15, 267-272.	6.7	408
7	Molecular genetics and outcome in PKU. Mental Retardation and Developmental Disabilities Research Reviews, 1999, 5, 113-116.	3.6	3
8	Structural Insight into the Aromatic Amino Acid Hydroxylases and Their Disease-Related Mutant Forms. Chemical Reviews, 1999, 99, 2137-2160.	47.7	167
9	The Structural Basis of Phenylketonuria. Molecular Genetics and Metabolism, 1999, 68, 103-125.	1.1	132
10	PAHdb: A locus-specific knowledgebase. Human Mutation, 2000, 15, 99-104.	2.5	87
11	Phenylketonuria and hyperphenylalaninemia in eastern Germany: A characteristic molecular profile and 15 novel mutations. Human Mutation, 2000, 15, 254-260.	2.5	19
12	Structural interpretation of mutations in phenylalanine hydroxylase protein aids in identifying genotypeâ€“phenotype correlations in phenylketonuria. European Journal of Human Genetics, 2000, 8, 683-696.	2.8	40
13	Expression Analysis of Phenylketonuria Mutations. Journal of Biological Chemistry, 2000, 275, 29737-29742.	3.4	79
14	Characterization of the Mouse Phenylalanine Hydroxylase Mutation Pahenu3. Molecular Genetics and Metabolism, 2001, 72, 27-30.	1.1	12
15	Impaired Arachidonic (20:4n-6) and Docosahexaenoic (22:6n-3) Acid Synthesis by Phenylalanine Metabolites as Etiological Factors in the Neuropathology of Phenylketonuria. Molecular Genetics and Metabolism, 2001, 72, 185-198.	1.1	42
16	PAH Gene Mutations in the Sicilian Population: Association with Minihaplotypes and Expression Analysis. Molecular Genetics and Metabolism, 2001, 74, 353-361.	1.1	16
17	A silent mutation induces exon skipping in the phenylalanine hydroxylase gene in phenylketonuria. Human Genetics, 2001, 108, 14-19.	3.8	67
18	Aspartame loading test in PKU heterozygous individuals bearing severe and moderate mutations. Clinical Genetics, 2001, 58, 86-88.	2.0	4

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19	Ten novel mutations in the phenylalanine hydroxylase gene (PAH) observed in Brazilian patients with phenylketonuria. Human Mutation, 2001, 17, 77-77.	2.5	6
20	Mutations of the phenylalanine hydroxylase (PAH) gene in Brazilian patients with phenylketonuria. Human Mutation, 2001, 17, 122-130.	2.5	27
21	Conjugation of phenylalanine hydroxylase with polyubiquitin chains catalysed by rat liver enzymes. BBA - Proteins and Proteomics, 2001, 1547, 379-386.	2.1	9
22	Congenital Heart Disease in Maternal Phenylketonuria: Report from the Maternal PKU Collaborative Study. Pediatric Research, 2001, 49, 636-642.	2.3	80
23	Genetic Heterogeneity in Five Italian Regions: Analysis of PAH Mutations and Minihaplotypes. Human Heredity, 2001, 52, 154-159.	0.8	20
24	Estimation of amino acid pairs sensitive to variants in human phenylalanine hydroxylase protein by means of a random approach. Peptides, 2002, 23, 2085-2090.	2.4	26
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29	Modeled ligand-protein complexes elucidate the origin of substrate specificity and provide insight into catalytic mechanisms of phenylalanine hydroxylase and tyrosine hydroxylase. FEBS Journal, 2003, 270, 1065-1075.	0.2	12
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32	In Vivo Studies of Phenylalanine Hydroxylase by Phenylalanine Breath Test: Diagnosis of Tetrahydrobiopterin-Responsive Phenylalanine Hydroxylase Deficiency. Pediatric Research, 2004, 56, 714-719.	2.3	13
34	Effects of tetrahydrobiopterin and phenylalanine on in vivo human phenylalanine hydroxylase by phenylalanine breath test. Molecular Genetics and Metabolism, 2007, 92, 308-314.	1.1	4
35	ThePAH gene, phenylketonuria, and a paradigm shift. Human Mutation, 2007, 28, 831-845.	2.5	288
36	Impact of Neonatal Protein Metabolism and Nutrition on Screening for Phenylketonuria. Journal of Pediatric Gastroenterology and Nutrition, 2008, 46, 561-569.	1.8	19
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