Resource Summary Report

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Phenylalanine Hydroxylase Locus Knowledgebase

RRID:SCR_013381
Type: Tool

Proper Citation

Phenylalanine Hydroxylase Locus Knowledgebase (RRID:SCR_013381)

Resource Information

URL: http://www.pahdb.mcgill.ca

Proper Citation: Phenylalanine Hydroxylase Locus Knowledgebase (RRID:SCR_013381)

Description: A database of centralized mutation data on the PAH gene. Searchable fields of the database available to users are: mutation name, polymorphic haplotype, population, geographic location, gene region, codon number, mutation type, substitution, phenotype, author's name and many more. The complete information provided for each mutation is regularly updated from both published data and personal communications.

Abbreviations: PAHdb

Synonyms: Phenylalanine Hydroxylase Database

Resource Type: data or information resource, database

Keywords: FASEB list

Funding:

Resource Name: Phenylalanine Hydroxylase Locus Knowledgebase

Resource ID: SCR_013381

Alternate IDs: nif-0000-03237

Record Creation Time: 20220129T080315+0000

Record Last Update: 20250426T060326+0000

Ratings and Alerts

No rating or validation information has been found for Phenylalanine Hydroxylase Locus Knowledgebase.

No alerts have been found for Phenylalanine Hydroxylase Locus Knowledgebase.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 56 mentions in open access literature.

Listed below are recent publications. The full list is available at FDI Lab - SciCrunch.org.

Zhou J, et al. (2022) Characterization of phenylalanine hydroxylase gene variants and analysis of genotype-phenotype correlation in patients with phenylalanine hydroxylase deficiency from Fujian Province, Southeastern China. Molecular biology reports, 49(11), 10409.

Ferreira F, et al. (2021) Phenylketonuria in Portugal: Genotype-phenotype correlations using molecular, biochemical, and haplotypic analyses. Molecular genetics & genomic medicine, 9(3), e1559.

Qiang R, et al. (2021) Development of a mutation hotspot detection kit for the phenylalanine hydroxylase gene by ARMS-PCR combined with fluorescent probe technology. Bioscience reports, 41(2).

Masi L, et al. (2021) Bone fragility in patients affected by congenital diseases non skeletal in origin. Orphanet journal of rare diseases, 16(1), 11.

Scala I, et al. (2020) Large Neutral Amino Acids (LNAAs) Supplementation Improves Neuropsychological Performances in Adult Patients with Phenylketonuria. Nutrients, 12(4).

Alibakhshi R, et al. (2019) The status of PAH gene-VNTR alleles and mini-haplotypes associations with PAH gene mutations in Iranian Kurdish PKU patients. Medical journal of the Islamic Republic of Iran, 33, 88.

Alibakhshi R, et al. (2018) Spectrum of Phenylalanine Hydroxylase Gene Mutations in Hamadan and Lorestan Provinces of Iran and Their Associations with Variable Number of Tandem Repeat Alleles. Iranian journal of medical sciences, 43(3), 318.

Rastegar Moghadam M, et al. (2018) Mutation analysis of Phenylalanine hydroxylase gene in Iranian patients with Phenylketonuria. Medical journal of the Islamic Republic of Iran, 32, 21.

Pampukha V, et al. (2017) Analysis of EX5del4232ins268 and EX5del955 PAH gene mutations in Ukrainian patients with phenylketonuria. Genes & diseases, 4(2), 108.

Ney DM, et al. (2017) Metabolomic changes demonstrate reduced bioavailability of tyrosine and altered metabolism of tryptophan via the kynurenine pathway with ingestion of medical foods in phenylketonuria. Molecular genetics and metabolism, 121(2), 96.

Stroup BM, et al. (2017) Dietary amino acid intakes associated with a low-phenylalanine diet combined with amino acid medical foods and glycomacropeptide medical foods and neuropsychological outcomes in subjects with phenylketonuria. Data in brief, 13, 377.

North TL, et al. (2016) A study of common Mendelian disease carriers across ageing British cohorts: meta-analyses reveal heterozygosity for alpha 1-antitrypsin deficiency increases respiratory capacity and height. Journal of medical genetics, 53(4), 280.

Ney DM, et al. (2016) Glycomacropeptide for nutritional management of phenylketonuria: a randomized, controlled, crossover trial. The American journal of clinical nutrition, 104(2), 334.

Masica DL, et al. (2016) Towards Increasing the Clinical Relevance of In Silico Methods to Predict Pathogenic Missense Variants. PLoS computational biology, 12(5), e1004725.

Biglari A, et al. (2015) Mutations of the phenylalanine hydroxylase gene in Iranian patients with phenylketonuria. SpringerPlus, 4, 542.

Scala I, et al. (2015) Long-term follow-up of patients with phenylketonuria treated with tetrahydrobiopterin: a seven years experience. Orphanet journal of rare diseases, 10, 14.

Gu Y, et al. (2014) Mutation spectrum of six genes in Chinese phenylketonuria patients obtained through next-generation sequencing. PloS one, 9(4), e94100.

Hozyasz KK, et al. (2014) Association of common variants in PAH and LAT1 with non-syndromic cleft lip with or without cleft palate (NSCL/P) in the Polish population. Archives of oral biology, 59(4), 363.

Gallego-Villar L, et al. (2014) A sensitive assay system to test antisense oligonucleotides for splice suppression therapy in the mouse liver. Molecular therapy. Nucleic acids, 3(9), e193.

Imperlini E, et al. (2014) Altered brain protein expression profiles are associated with molecular neurological dysfunction in the PKU mouse model. Journal of neurochemistry, 129(6), 1002.