Resource Summary Report

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Lafora Progressive Myoclonus Epilepsy Mutation and Polymorphism Database

RRID:SCR_002919
Type: Tool

Proper Citation

Lafora Progressive Myoclonus Epilepsy Mutation and Polymorphism Database (RRID:SCR_002919)

Resource Information

URL: http://projects.tcag.ca/lafora/

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Description: The Lafora database is a repository of information related to progressive myoclonus epilepsy mutation and polymorphism data. Users may view all mutations in the database(Mutations of EPM2A and NHLRC1(EPM2B)), click on individual exons for mutations, or search the database by keyword. Nucleotide and amino acids positions were assigned based on the GenBank reference sequence NM_005670 for EPM2A and NM_198586 for EPM2B. The data can be viewed using the XRT Table Browser, and where possible, links to external sources such as NCBI, PubMed are provided. At this time, the database is under development. The BioXRT (Cross-Referenced Tables) Table Browser is a highly configurable tool for viewing complex, table based information. The tables can be displayed using pre-set options, or customized to view arbitrary subsets of rows, columns or other features.

Abbreviations: Lafora Database

Synonyms: Lafora Progressive Myoclonus Epilepsy Mutation Polymorphism Database, The Lafora Progressive Myoclonus Epilepsy Mutation and Polymorphism Database, The Lafora Progressive Myoclonus Epilepsy Mutation Polymorphism Database

Resource Type: database, data repository, storage service resource, data or information resource, service resource

Keywords: epilepsy, epm2a, genetics, laforin, malin, mutation, myoclonus, nhlrc1(epm2b), polymorphism

Resource Name: Lafora Progressive Myoclonus Epilepsy Mutation and Polymorphism Database

Resource ID: SCR_002919

Alternate IDs: nif-0000-00157

Record Creation Time: 20220129T080216+0000

Record Last Update: 20260815T182221+0000

Ratings and Alerts

No rating or validation information has been found for Lafora Progressive Myoclonus Epilepsy Mutation and Polymorphism Database.

No alerts have been found for Lafora Progressive Myoclonus Epilepsy Mutation and Polymorphism Database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 11 mentions in open access literature.

Listed below are recent publications. The full list is available at RRID .

Kumarasinghe L, et al. (2021) TRIM32 and Malin in Neurological and Neuromuscular Rare Diseases. Cells, 10(4).

Brewer MK, et al. (2021) An empirical pipeline for personalized diagnosis of Lafora disease mutations. iScience, 24(11), 103276.

Brenner D, et al. (2019) Genotypes and phenotypes of patients with Lafora disease living in Germany. Neurological research and practice, 1.

Tee SK, et al. (2019) Lafora disease in a Malaysian with a rare mutation in the EPM2A gene. Seizure, 67, 78.

García-Gimeno MA, et al. (2018) Lafora Disease: A Ubiquitination-Related Pathology. Cells, 7(8).

Romá-Mateo C, et al. (2015) Oxidative stress, a new hallmark in the pathophysiology of Lafora progressive myoclonus epilepsy. Free radical biology & medicine, 88(Pt A), 30.

Brewer MK, et al. (2014) Expression, purification and characterization of soluble red rooster laforin as a fusion protein in Escherichia coli. BMC biochemistry, 15, 8.

Jara-Prado A, et al. (2014) Late onset Lafora disease and novel EPM2A mutations: breaking paradigms. Epilepsy research, 108(9), 1501.

Salar S, et al. (2012) Four novel and two recurrent NHLRC1 (EPM2B) and EPM2A gene mutations leading to Lafora disease in six Turkish families. Epilepsy research, 98(2-3), 273.

Traoré M, et al. (2009) Novel mutation in the NHLRC1 gene in a Malian family with a severe phenotype of Lafora disease. Neurogenetics, 10(4), 319.

Galperin MY, et al. (2005) The Molecular Biology Database Collection: 2005 update. Nucleic acids research, 33(Database issue), D5.