

Title: Locus-Specific Mutation Database Solutions for Neurogenetics

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Short one paragraph description: To standardize data submission and data access the use of dedicated database software is strongly recommended. Neurogenetics databases focusing on storing gene variants can use the Leiden Open-source Variation Database software (LOVD, freely available at www.LOVD.nl). For an example of a single gene database see <http://www.LOVD.nl/SMC1A>. For an example of storing genome wide studies see <http://www.LOVD.nl/MR>. The Gen2Phen project (see <http://www.gen2phen.org>) aims to establish and unify sequence variation databases (LSDBs). Within the setting of this EU-project we are able to offer any help you might need establishing fully web-based databases (this includes free server space and back-ups) to store genotype-phenotype information at the level of individual genes, a.o. serving clinical diagnosis and thus those affected and their families. These LSDBs will serve as a resource and collection point of all new variants identified world-wide by labs screening these genes for sequence variants. The web-based submission and curation supports easy maintenance and updates of the data, providing direct insight in which variants have been found in both patients and controls, facilitating the diagnostic conclusion whether specific variants should be considered to have pathogenic consequences or not. Please note that the LOVD software follows current recommendations of the Human Genome Variation Society (HGVS) and of the Human Variome Project (HVP). LOVD has the option to export the data to be shared with central repositories like dbSNP (NCBI) and the UCSC browser (PhenCode track). These can then link directly to each specific entry into the LOVD database(s). LOVD also has links to a Wiki-environment; you can allow it to have your data indexed by Wiki-Professional. When you have a list of variants (published and unpublished) in electronic format (e.g. as a spreadsheet) we can establish draft databases for you within a few days.

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