

The Alzheimer Disease & Frontotemporal Dementia Mutation Database

Marc Cruts*, Nathalie Brouwers, Christine Van Broeckhoven

Neurodegenerative Brain Diseases Group, Department of Molecular Genetics, VIB, Antwerpen, Belgium;; Laboratory of Neurogenetics, Institute Born-Bunge, Antwerpen, Belgium; and University of Antwerp, Antwerpen, Belgium

The Alzheimer Disease & Frontotemporal Dementia Mutation Database (<http://www.molgen.ua.ac.be/ADMutations>, <http://www.molgen.ua.ac.be/FTDMutations>) is an online reference database of mutations in genes associated with inherited forms of Alzheimer disease, frontotemporal lobar degeneration and related neurodegenerative diseases. The database includes clinical mutations and non-clinical coding DNA variants. Mutations in eight genes are collected, i.e. the AD-related genes *APP*, *PSEN1* and *PSEN2*, and the FTLN-related genes *GRN*, *MAPT*, *VCP*, *CHMP2B*. Also, mutations in *TARDBP* are included and *FUS* mutations will be included soon. The latter two genes are associated with amyotrophic lateral sclerosis which can co-occur with frontotemporal dementia. The mutation database is updated continuously based on data collected from scientific literature, meeting presentations and direct submissions. To date, the database contains information on 505 DNA variants of which 382 are clinical mutations. Detailed genetic data are collected together with basic clinical and functional data and references to literature are provided. Mutations are presented in tabular listings and on schematic 2D and/or 3D protein models. Further, a custom annotation track for the UCSC human genome browser can be downloaded. Based on the expertise acquired from the Alzheimer Disease & Frontotemporal Dementia Mutation Database, a Parkinson Disease Mutation Database is being set up, which will become available soon.