

Update of the Parkinson's Disease Mutation Database

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This Parkinson's Disease Mutation Database (<http://grenada.lumc.nl/LOVD2/TPI/home.php>) was created to capture the mutation spectrum and ultimately the mutation frequency of the genes involved in monogenic forms of Parkinson's disease (PD). To date, at least five different genes have been clearly related to PD: the *SNCA* and *LRRK2* gene exhibit an autosomal dominant mode of inheritance, whereas the genes for *Parkin*, *PINK1* and *DJ-1* show an autosomal recessive pattern of inheritance. In contrast to the PD Gene database (<http://www.pdgene.org/>) that describes polymorphisms in association studies, the focus of the Parkinson's Disease Mutation Database is predominantly on disease-causing pathogenic mutations.

We search the Medline database (via PubMed, online at <http://www.ncbi.nlm.nih.gov>) for publications for studies screening PD patients for mutations in PD genes. We use the search terms "[Gene name] AND mutation" and "[Gene name] gene". We select original reports of disease-causing PD mutations. We have implemented LOVD 2.0 that allows external registered users to submit mutations online, which will be curated and then made available to the public.

The database is organized by gene and the initial table includes: gene name, chromosomal location, locus, mode of inheritance, reference sequences used, synonyms for the gene, and the number of mutations listed. In the following pages with the mutation lists for each gene, the tables include: exon location the standardized DNA and protein change, mutation type. The references are linked to the abstracts on Medline. A standard nomenclature column was included to create coherence among all the reported variations.

This PD mutation database will provide a comprehensive tool for basic scientists as well as clinicians to quickly get an overview of the known and recurrent mutations and compare their findings and testing results with known reported mutations. In the long term, this database might even allow to test for genotype-phenotype correlations when clinical datasets can be linked or implemented.