Center for Neuroscience and Cell Biology

Neurogenetics Laboratory

University of Coimbra

Request Form for Genetic testing of Neurodegenerative Diseases

Per patient, two EDTA blood 3 ml tubes should be taken and each tube should be labelled with surname, forename and date of birth. To measure the progranulin concentration in serum, a clotted blood tube should also be sent to the laboratory. Store blood at room temperature (do not freeze).

This filled out request Form must accompany the samples.

Referring Clinician (person to whom result will be sent)

Forename: _____ Surname: _____

Hospital/Institute: _____ Department: _____

Request Date: _____ Signature:_____ Email: _____

Patient Data

Forename: _____ Surname: _____

Date of birth: ___/___/___ Gender: _____ Ref. Number _____

Invoice Address: _____

_____

Clinical Information

Clinical Diagnosis: _____ known mutation? No ☐ Yes ☐ which?_____

Patient (*index* case) ☐ Patient (*familiar* case) ☐ Presymptomatic* ☐ Carrier* ☐

* The Laboratory will not proceed with predictive testing without a copy of the signed informed consent and genetic counselling

Aditional Information (Please include any relevant clinical information such as symptoms, family history and pedigree if appropriate)

Tests Required (please tick the correct choice)

Alzheimer Disease

- ☐ All coding exons of presenilin 1 gene (*PSEN1*)
- ☐ All coding exons of presenilin 2 gene (*PSEN2*)
- ☐ Exons 16 and 17 of the amyloid precursor protein gene (*APP*)
- ☐ Apolipoprotein E (*ApoE)* genotype

Parkinson Disease

- ☐ G2019S mutation in leucine-rich repeat kinase 2 gene (*LRRK2*)
- ☐ All coding exons of parkin gene (*PARK2*)
- ☐ Exons 9 and 10 of the glucocerebrosidase gene (*GBA*)

Frontotemporal Lobar Degeneration(FTLD)

- ☐ Exons 1, 9-13 of microtubule-associated protein tau gene (*MAPT*)
- ☐ All coding exons of Progranulin gene (*PGRN*)
- ☐ Hexanucleotide Repeat Expansion in *C9ORF72* gene
- ☐ Determination of progranulin serum concentration
- ☐ All coding exons of sequestosome 1 gene (*SQSTM1*)

Fatal familial insomnia

- ☐ D178N mutation in the prion protein gene (*PRNP*)
- ☐ M129V polymorphism genotype in *PRNP* gene

Familial British dementia / Familial Danish dementia

- ☐ Exon 6 of the *BRI2* gene

Test for known mutation

- ☐ Please give details of mutation _____

and proband's name _____

- ☐ Othet Test_____

To be filled in by laboratory

Sample No. _____ Arrival Date: ___/___/___

Neurogenetics laboratory | University Hospitals of Coimbra - Praceta Prof. Mota Pinto 3000-075 Coimbra |
Tel.: +351 239 400 400 – Ext.: 12145 | Email: mralmeida2008@gmail.com| mribeiro@fmed.uc.pt