

PhD Position: Program INPhINIT Fundació La Caixa

“Development of new tools to evaluate cognition for Huntington’s Disease (HD)”

(Dr. Jaime Kulisevsky)

CENTRE: IIB SANT PAU - Fundació Institut de Recerca de l’Hospital de la Santa Creu i Sant Pau

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CENTRE DESCRIPTION: The Research Institute of the Hospital de la Santa Creu i Sant Pau (HSCSP-IR) was created on 4 June 1992 as a private scientific foundation. Its mission is to promote basic, clinical, epidemiological and healthcare research in the health science and biomedical fields, with the ultimate aim of improving the health of the population. On 10 December 2003, the Autonomous Government of Catalonia approved affiliation of the HSCSP-IR as a University Research Institute attached to the Autonomous University of Barcelona (UAB).

The HSCSP-IR has as its mission to improve the health and quality of life of the population through the production and dissemination of scientific knowledge, the training of researchers to an international standard, innovation in health and the incorporation of medical advances in clinical practice and in healthcare policies.

The HSCSP-IR is currently one of the most active research centres in Catalonia, especially in relation to translational research and the application of new discoveries to clinical practice. Since 2011 it has been part of the Catalan System of Research Centres (CERCA).

On 17 May 2009, the HSCSP-IR and nine other organizations created the Sant Pau Biomedical Research Institute (IIB Sant Pau), with the aim of strengthening collaborative translational research and bridging the gap between basic research and clinical practice so as to ultimately improve patient care.

AREA OF KNOWLEDGE: Life Sciences Panel

GROUP OF DISCIPLINES: Medicine, Public Health, Sports Science, Nutrition, Clinical Psychology, Healthcare Management

GROUP LEADER: Dr. Jaime Kulisevsky - jkulisevsky@santpau.cat

RESEARCH PROJECT/RESEARCH GROUP

Website of the movement disorders unit that Dr. Kulisevsky leads

<http://www.santpau.cat/en/web/public/neulo-recerca-unitat-de-parkinson-i-transtorns-de-moviment>

POSITION DESCRIPTION

-Research Project / Research Group Description:

Huntington's disease (HD) is a neurodegenerative disease with an autosomal dominant inheritance, caused by an abnormal expansion of CAG trinucleotide in the short arm of the HTT gene on chromosome 4, which encodes the protein huntingtin (Htt). People carrying more than 40 CAG repeats, will inevitably develop the disease throughout middle adulthood as a triad of progressive motor, cognitive and behavioral disturbances. Although new To date, there are no treatments that stop or slow the course of the disease.

Certain changes in specific cognitive domains can be measurable up to 15 years before clinical diagnosis of the disease, leading to cognitive decline in pre-motor manifest gene expansion carriers, which gradually worsens in late stages of HD.

Cognitive decline in HD was conceptualized to mainly produce frontal-executive deficits, and the most used cognitive tools are focused on tasks measuring frontal-subcortical alterations. It is presently known that cognitive impairment in HD follows a heterogeneous course between individuals with other affected domains that can be prevalent since early stages of the disease. However, there is lack of a formal definition of dementia and mild cognitive impairment in HD and there are no validated or specific instruments to reliably screen for global cognitive function in HD population useful to differentiate cognitive subgroups of patients more prone to rapidly deteriorate. This is urgent at the light of the upcoming therapeutic trials. Thus, a recent review of the Movement Disorders Society Committee on Rating Scales Development concluded that that there is a need to further validate currently available cognitive scales in HD.

Based on the group experience on the development and validation of specific cognitive instruments for Parkinson's disease, our present objective is twofold: a) to validate previous global instruments; and b) to develop a new HD-specific and brief scale which includes assessment of all cognitive domains.

-Job position description:

The position is to cover the neuropsychologist who will be responsible for the execution of the project. That includes: development of scale, organization of visits, comply with all inclusion and exclusion patient criteria, review board of project with ethic committee, give information to participants, answering any questions during study performance, signature of written informed consent and statics analysis. Additionally, the same person will be the responsible to undertake the project, which includes the administration of the new scale and functional questionnaire to all participants.

OTHER RELEVANT WEBSITES

Website of the network for biomedical research in Neurodegeneratives diseases (CIBERNED). The research group forms part of CIBERNED, which is the only research center in Spain, integrated into the international network of Centres of Excellence in Neurodegeneration.

<https://ciberned.es/grupo-kulisevsky.html>

Website of the research institute of Sant Pau: <http://www.recercasantpau.cat/en/>

