PREVALENCE OF LRRK2 MUTATIONS IN A POPULATION STUDY IN TWO SELECTED RURAL BASQUE VILLAGES (SPAIN)

J. Ruiz-Martínez¹, I. Soria², A. Gorostidi³, A. Estanga⁴, A. Bergareche⁴, M. Etxart⁵, J. Emparanza⁶, A. Lopez De Munain⁴, J.F. Martí Massó¹

¹Neurology, Hospital Donostia, San Sebastián, ²Zarautz Health Center, Zarautz, ³Neurogenetics Laboratory, ⁴Neurology, ⁵Biodonostia, ⁶Epidemiology, Hospital Donostia, San Sebastian, Spain

Introduction: The most common LRRK2 mutation, G2019S, has been observed throughout the world. The LRRK2 R1441G mutation is responsible for 46% of familial PD and for 2.5% of sporadic PD patients with Basque origin, with a penetrance of 80% at 80 years old.

Aims: The aim of this study is to determine the prevalence of R1441G and G2019S mutations in a randomly population sample selected from two villages in the province of Gipuzkoa (Spain).

Methods: A random sample of 576 subjects of 65 years or older, from a total population of 4301 subjects in this age group in two rural villages (Azpeitia and Azkoitia) was selected according to the public health service (OSABIDE). A brief questionnaire and a physical examination were administrated, and all individuals were invited to participate in the genetic screening for these mutations.

Results: 219 subjects were screened, and we obtained only clinical information in the other 357. The prevalence of R1441G mutation was 1,82% (0,5-4,61), due to 4 subjects carriers (2 PD patients and 2 unaffected relatives), with no case of G2019S mutation carriers. The prevalence of PD, and unaffected relatives of PD was 3,19% (0,63-5,75), and 9,58% (5,46-13,77) respectively. In the total sample (576 subjects) 3 of the 10 PD patients screened for these mutations were R1441G carriers (30%, 6,67-65,25), and 5 were familial PD (50%, 18,70-81,29).

Discussion: R1441G mutation prevalence, and high percentage of familial PD in the studied population, could explain this higher prevalence of PD than those found in other studies.