LRRK2 MUTATIONS IN PARKINSON DISEASE - A PORTUGUESE STUDY

A. Morgadinho¹, F. Moreira², M.R. Almeida³, P. Coutinho⁴, C. Januário²

¹Neurology Department, Coimbra Central Hospital E.P.E and Centro Hospitalar entre Douro e Vouga, E.P.E; ²Neurology Department, Coimbra University Hospital E.P.E.; ³CNC, Coimbra University, Coimbra; ⁴Neurology Department, Centro Hospitalar entre Douro e Vouga, E.P.E, Santa Maria da Feira, Portugal

Introduction: Mutations in LRRK2 gene are the most commonly identified monogenic etiology of Parkinson disease (PD). Over-represented in the Ashkenazi Jewish population, the prevalence of p.G2019S mutation is also very high in some Southern European countries.

Aims: To identify the p.G2019S mutation in a series of Portuguese PD patients and to make a clinical characterization of these patients.

Methods: We determined to test for G2019S mutation in a clinic case series of PD patients followed in the Movement Disorders Clinics in the centre of Portugal. After obtaining informed consent, a blood sample was taken and DNA extracted by standard procedures. Exon 41 was PCR amplified and sequenced on capillary automated sequencer. All patients were evaluated by a standard clinical protocol.

Results: 17 patients had the G2019S mutation. Three cases were sporadic, the others were familial cases of PD. The age at onset of PD varies between 38 and 76 and the duration of the disease between 4 and 34 years. The first PD symptom was bradykinesia in five, tremor in eleven, dystonia in one. The UPDRS (in on) varies between 22 and 57 and Hoehn and Yahr between 2.5 and 4. Three patients have dementia.

Conclusions: Our results confirm that G2019S mutation is a frequent cause of PD in Portugal, with relevant implications for genetic counselling. The phenotype of the patients reported is similar to late onset PD although in some patients the progression of the disease seems to be slower and milder than usually seen in idiopathic PD.