ONE YEAR EXPERIENCE OF TESTING FOR PARKIN AND LRRK2 GENES IN PORTUGUESE PATIENTS WITH PARKINSON'S DISEASE

M.R. Almeida¹, A. Morgadinho², C. Januário³

¹The Centre of Neuroscience and Cell Biology, University of Coimbra, ²Neurology Department, Centro Hospitalar entre Douro e Vouga, E.P.E e Centro Hospitalar de Coimbra, E.P.E., ³Neurology Department, Coimbra University Hospital, Coimbra, Portugal

Introduction: The genetic heterogeneity of PD is well demonstrated by the identification of several genes in which mutations cause the disease in a mendelian trait. Since then, genotype-phenotype correlations have been reported. However, the outcome of such associations is far of being straightforward. Given the clinical features overlap amongst PD patients with different genes mutated and also the frequent variability of a single mutation, it is extremely difficult to decide which gene should be primarily tested for each patient and what are the implications for the individual and/or family of such genetic tests.

Aims: To determine the frequency of mutations in Parkin and LRRK2 genes and to discuss the molecular findings implications, in daily practice, since they represented the PD population-based sample referred to our Neurogenetics laboratory, during last year.

Methods: During the period considered, 87 consecutive patients recruited with regard to an early-onset of disease and/or family history, were studied. The entire coding region of the Parkin gene and exon 41 of LRRK2 were PCR amplified followed by direct sequenced on a capillary automated sequencer.

Results: Eight out of the 69 patients (12%) had G2019S mutation in LRRK2 gene while mutations in Parkin gene occurred in seven out of 57 patients tested (12%).

Conclusions: The similar frequency of mutations found in both genes (12%) emphasizes the importance of these genes in the etiology of PD in our population. To the ones with a mutation identified, practical issues have been raised concerning the appropriate genetic counselling.