O-003 - EVALUATION OF THE FREQUENCY OF NON-MOTOR SYMPTOMS OF PARKINSON'S DISEASE IN ADULT PATIENTS WITH GAUCHER DISEASE TYPE 1

Wilke MVMB [1,2], Dornelles AD [2], Schuh AS [1,3], Vairo FP [2,4,5], Basgalupp SP [1,10], Siebert M [6,7,10], Nalin T [2], Piltcher OB [8], Schwartz IVD [1, 2, 9, 10]

1) Graduate Program in Medical Sciences, Faculdade de Medicina, Universidade Federal do Rio Grande do Sul, Porto Alegre, RS, Brazil; 2) Medical Genetics Service, Hospital de Clínicas de Porto Alegre, Porto Alegre, RS, Brazil; 3) Department of Pharmacology, Universidade Federal do Rio Grande do Sul, Porto Alegre, Brazil; 4) Center for Individualized Medicine, Mayo Clinic, Rochester, MN, USA; 5) Department of Clinical Genomics, Mayo Clinic, Rochester, MN, USA; 6) Graduate Program in Sciences of Gastroenterology and Hepatology, Universidade Federal do Rio Grande do Sul (UFRGS), Porto Alegre, RS, Brazil; 7) Laboratory Research Unit, Experimental Research Center, Hospital de Clínicas de Porto Alegre (HCPA), Porto Alegre, RS, Brazil; 8) Department of Ophthalmology and Otorhinolaryngology, Hospital de Clínicas de Porto Alegre, Porto Alegre, RS, Brazil; 9) Department of Genetics, Universidade Federal do Rio Grande do Sul, Porto Alegre, Brazil; 10) BRAIN Laboratory, Hospital de Clínicas de Porto Alegre, Porto Alegre, RS, Brazil.

INTRODUCTION: Gaucher disease (GD) is an inborn error of metabolism caused by deficiency of beta-glucocerebrosidase (GCase). More than 400 mutations of the GCase gene (GBA1) have been described. GD is conventionally classified into three clinical forms, on the basis of neurological involvement: type 1 is considered the non-neuropathic form, whereas types 2 and 3 affect the nervous system. Parkinson's disease (PD) is the second most common neurodegenerative condition. The classic motor symptoms of PD may be preceded by many non-motor symptoms (NMS), which include hyposmia, REM sleep behavior disorder, constipation, cognitive impairment, and depression. Population studies have identified mutations in GBA1 as the main risk factor for idiopathic PD. The present study sought to evaluate the prevalence of NMS in a cohort of patients with GD type 1 from Southern Brazil. ***METHODOLOGY:*** This is an observational, cross-sectional study, with a convenience sampling strategy. Cognition was evaluated by the Montreal Cognitive assessment (MoCa), daytime sleepiness by the Epworth Scale, depression by the Beck Inventory, constipation by the Unified Multiple System Atrophy Rating Scale, and REM sleep behavior disorder by the Single-Question Screen; hyposmia by the Sniffin' Sticks. Motor symptoms were assessed with part III of the Unified Parkinson's Disease Rating Scale. Patients were also genotypes for a GBA1 3′-UTR SNP (rs708606). ***RESULTS:*** Twenty-three patients (female=13; on enzyme replacement therapy=21, substrate reduction therapy=2) with a mean age of 41.45±15.3 years (range, 22-67) were included. Eight patients were heterozygous for the 3′-UTR SNP (rs708606). Fourteen patients (8 over age 40 years) presented at least one NMS; daytime sleepiness was the most frequent (n=10). Two patients (aged 63 and 64, respectively) also presented motor symptoms, probably drug-related. ***CONCLUSIONS:*** NMS were prevalent in this

cohort. We highlight the importance of a multidisciplinary follow-up focusing on earlier diagnosis of PD, especially for patients with GD type 1 over the age of 40.