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Genotype may influence the onset of axial signs in early-stage Parkinson's disease

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Background and aims: Heterogeneity exists regarding the onset of axial signs in Parkinson's disease (PD). Dysarthria, swallowing disturbance and respiratory muscles dysfunction can be observed in early-onset PD patients. Due to their impact on the outcome and quality of life, evidencing risk factors of these symptoms is essential to optimize the follow-up of our patients. The aim of our study was to assess the association between the genotype and the axial signs.

Methods: MAPT haplotypes and COMT polymorphism were tested in 31 PD patients (mean age= 61.4 years±6.5) of the Prodigy-Park 1 cohort with a mean disease duration of 1.1 years (±1.1). Neurological, swallowing and voice and pulmonary function testing evaluations were performed.

Results: A valine homozygous polymorphism (n=11) was associated with a significantly higher sniff nasal inspiratory pressure (SNIP) in comparison with methionine homozygous (n=7) and heterozygous polymorphism (n=13) (78%±14.2 vs. 60.9%±19.8 - p=0.02). Regarding MAPT gene, patients with a H1/H1 haplotype (n=21) had a significantly higher severity of their dysarthria assessed by a French adaptation of the Frenchay Dysarthria Assessment (4±2.7 vs. 1.4±2.2 - p=0.02).

Conclusion: In early-stage PD, the onset of dysarthria or inspiratory muscle weakness might be associated with the genotype. Dopamine might impact on the ventilatory function and MAPT H1 haplotype could lead to a pseudobulbar palsy. These preliminary results need to be confirmed in a larger cohort to assess the influence of MAPT haplotypes or COMT polymorphism in the other features of the axial signs (such as the swallowing disturbance).

Disclosure: Nothing to disclose

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ParkLink Bologna - an Italian record linkage system for Parkinson's disease: Ready, set, go!

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Background and aims: Record linkage systems (RLS), matching data across administrative health databases, help providing information on diseased populations. However, detailed clinical information is often missing. The aim of the project (ParkLink Bologna) is to create a RLS based on clinical diagnosis to perform clinical epidemiologic studies on Parkinson's disease (PD) and parkinsonism (Ps) in a population-based setting (Bologna Health District, Emilia-Romagna Region, Italy).

Methods: Since January 2016, we are inviting neurologists working in private practice or public health service to enroll patients with a clinical suspect of PD or Ps residing in Bologna Health District (870507 inhabitants). Clinical diagnosis, date and type of onset and level of disability of patients who gave consent are linked to different administrative databases.

Results: On December 2016 six databases were linked (drug prescriptions, ER access, hospital discharges, copayment exemption, medical home-care, mortality); 15 neurologists out of 42 joined ParkLink. About 25% (539) of expected prevalent patients were already included (no refusals); 466 had a final diagnosis of PD (73%) or Ps (27%). Compared to Ps, PD were younger, with longer disease duration, lower disability level (Table 1) and a slightly different treatment prescribing pattern (Table 2). Risk of ER access and hospital admission increased with disability level.