LACK OF *CHCHD2* MUTATONS IN PARKINSON'S DISEASE IN A SOUTHERN ITALY POPULIATION

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INTRODUCTION

Parkinson's Disease (PD) is the most common form of degenerative parkinsonism with a prevalence of 1% of those older than 65 years. To date 5 genes are known to be causative to autosomal-recessive (*PARK2, PINK1, and DJ1*) or autosomal dominant (*SNCA* and *LRRK2*) forms of PD. Recently, *Funayama et* al. found a missense mutation (p.Thr61Ile) and three rare variants (p.Arg145Gln, c.300+5G>A, p.Pro2Leu) in CHCHD2 gene in familial Parkinson's disease. The aim of this study was to evaluate the presence of CHCHD2 mutations in a southern Italy cohort with clinically diagnosed PD.

MATERIALS AND METHODS

- ❖CHCHD2 screening in a cohort of 165 familial patients with clinically diagnosed PD and 200 control subjects from South Italy
- ❖Genomic DNA extraction from peripheral blood by standard methods
- ❖ Exclusion of mutations in other PD –related genes by sequencing analysis
- ❖Purified PCR products analysis on 3500 Genetic Analyzer (Applied Biosystems)

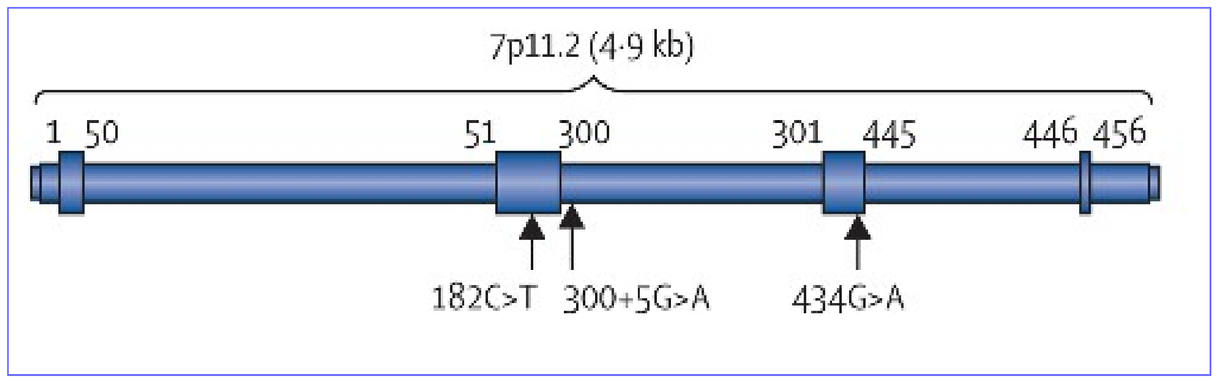

Figure 1. Funayama et al. 2015

RESULTS

No CHCHD2 mutations were identified by sequencing the entire coding region of this gene in our PD population.
The mutational screening of CHCHD2 gene has only highlighted the presence of intronic variants (rs816408, rs10043, rs816407, rs8406) with the same frequency in both PD and control cohort.

CONCLUSIONS

CHCHD2 gene contains 4 exons (Figure 1) and encodes for a mitochondrial protein may be linked to cytochrome c oxidase activity, further supporting the role of mitochondria dysfunction in the pathogenesis of PD. Recently, only in one Chinese family with autosomal dominant parkinsonism was identified the p.Thr61Ile mutation (*Shi et al., 2016*) and a nonsense mutation (p.Gln126X) was identified in a German patient with PD (*Koschmidder et al.,2016*) in CHCHD2 gene.
We did not observe significant associations between identified genetic variants (*Funayama et al. 2015; Koschmidder et al.,2016*) and risk of PD in our population. Our data suggest that genetic variants of CHCHD2 do not play a major role in our familial PD population.

REFERENCE

Funayama, M., Ohe, K., Amo, T., Furuya, N., Yamaguchi, J., Saiki, S., Li, Y., Ogaki, K., Ando, M., Yoshino, H., Tomiyama, H., Nishioka, K., Hasegawa, K., Saiki, H., Satake, W., Mogushi, K., Sasaki, R., Kokubo, Y., Kuzuhara, S., Toda, T., Mizuno, Y., Uchiyama, Y., Ohno, K., Hattori, N., 2015. CHCHD2 mutations in autosomal dominant late-onset Parkinson's disease: a genome-wide linkage and sequencing study. Lancet Neurol. 14, 274-282.

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