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Next generation sequencing data analysis evaluation in patients with Parkinsonism from genetically isolated population

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Parkinson's disease (PD) can be caused by genetic changes in a lot of genes. The effect of these changes is determined by the nature of the mutation and ranges from weak associations to pathogenic mutation which leads to loss of protein function. Our study is based on epidemiological data which show significantly increased prevalence of PD (2.9%) in an isolated population of south-eastern Moravia in the Czech Republic. We compared two different Next Generation Sequencing (NGS) data analysis approaches in DNA from 28 PD patients in the genes responsible for Parkinsonism (*ADH1C, ATP13A2, EIF4G1, FBXO7, GBA + GBAP1, GIGYF2, HTRA2, LRRK2, MAPT, PARK2, PARK7, PINK1, PLA2G6, SNCA, UCHL1* and *VPS35*) using: 1) Already described missense rare variants or pathogenic mutations and 2) Twelve control DNA samples from the same isolated population. Ion Torrent NGS data processing and trimming from Fastaq through "bam" to "vcf" files was done parallel by Torrent Suite/Ion Reporter and NextGene software. Variants were than filtered using following parameters: AQ>20; Read coverage >20; MAF<0,01; SIFT: 0 - 0,05 and/or PolyPhen-2: 0,2 -1. After filtering out, three missense mutations were found in *LRRK2* gene: rs33995883 in 6/0 patients/control (p/c), rs33958906 in 1/1p/c, rs781737269 in 3/0p/c, one missense mutation in *MAPT* gene rs63750072 in 6/1p/c and one mutation in *HTRA2* gene rs72470545 in 3/1p/c. Both the results from NextGene with Ion Torrent adaptation and from Ion Reporter significantly correlated in variant calling. Our study may contribute to further explanation of genetic background of Parkinsonism.

Biography

Radek Vodicka obtained his degree in Biology from the Faculty of Science, Masaryk University, Brno, Czech Republic, specializing in Genetics and Molecular Biology. He joined the DNA laboratory of the Department of Medical Genetics and Foetal Medicine, the University Hospital, Olomouc, where he obtained a post-graduate qualification in the Laboratory Method of Medical Genetics (2002). He defended his PhD thesis 'Y-chromosomal sequences in Turner syndrome patients' in 2003 and habilitated in 2013. His main research and clinical interest is a quantitative analysis of Human DNA sequences in relation to genetic diseases, infertility and cancerogensis.

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