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Exome analyses in subfamily trios from large family tree in the south-eastern Moravia (Czech Republic) population with high incidence of parkinsonism

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There has been previously described higher prevalence of Parkinsonism in small isolated region from the South-Eastern Moravia. We used NGS Ion AmpliSeq Exome method (IonTorrent) for two (A and B) subfamily trios. Each trio comprised of two affected and one healthy person. DNA exome libraries were sequenced on IonPI chips. Variants were predicted using Torrent Suite and Ion Reporter softwares. Aligned reads (BAM files) were than analyzed using Ion Reporter Whole Exome Trio workflow. Final filtering was done with respect to population frequency, variant effects and with respect to the presence of variants in Parkinsonism disease responsible genes. Last filter was done with respect to the segregation of the disease. Almost whole exome was sequenced with coverage 1-20 and 90% of exome was covered more than 20x in all the samples. Together more than 70,000 variants with average base coverage depth 75 were analyzable in both trios before filtering. After filtering there were found 99 and 96 variants in trio A and B respectively. The most potentially associating variants with parkinsonism are as given in tabulated form as follows:

Trio A:	Trio B:
SLC18A2:p.Gly195Ser;c.583G>A	TENM4:p.Asn965Ser;c.2894A>G
DRD1:p.Ala353Val;c.1058C>T	MON2: p.Gln531Arg;c.1592A>G
AP2A2:p.Asn401Ser;c.1202A>G	MTCL1:p.Ala482Val;c.1445C>T
CCDC88C:p.Leu1696Pro;c.5087T>C	NEPRO:p.Val297Ile;c.889G>A
ZFHX3:p.Met2102Thr;c.6305T>C	FAM131A:p.Leu280Val;c.838C>G
ARAP2:p.Pro159Ser;c.475C>T	ADH1C:p.Arg48His;c.143G>A
CYP4F11:p.Trp29Ser;c.86G>C	SYNE1:p.Lys3729Asn;c.11187G>T
MRPS15:p.Thr252Ile;c.755C>T	RXFP2:p.Thr222Pro;c.664A>C
MRPS28:p.Arg48Pro;c.143G>C	AKAP11:p.Ile183Met;c.549A>G
PRELID2:p.Val62Met;c.184G>A	ZNF19:p.Pro216Ser;c.646C>T
FAM171A1:p.Ser844Leu;c.2531C>T	LRRK2:p.Arg1514Gln;c.4541G>A
CAPRIN2:p.Arg373His;c.1118G>A	OSBPL1A:c.115_116insAATT
FAM186B:p.Ala727Val;c.2180C>T	SACS:p.Met1359Thr;c.4076T>C
CROT:p.Glu118Asp;c.354A>C	ZFHX3:p.Met2102Thr;c.6305T>C
MPDZ:p.Cys119Ser;c.356G>C	COL18A1:p.Ala1381Thr;c.4141G>A

Detailed whole exome analyses in genetic isolated parkinsonism patients could contribute to further understanding of molecular-genetic mechanism and background of the disease.

Biography

Radek Vodicka has completed his PhD study of Medical Genetics at the Faculty of Medicine and Dentistry, Palacky University Olomouc, Czech Republic in 2003. In 2015 he was appointed as an Associate Professor in the same field. Since 2001, he has been working in the DNA Diagnostics Laboratory at the Institute of Medical Genetics, University Hospital Olomouc, Czech Republic. He is also working as an Associate Professor at the Faculty of Medicine and Dentistry, Palacky University Olomouc. He has published more than 35 papers.

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