# RIG-1 SIGNALING VARIANTS ARE ASSOCIATED WITH ALZHEIMER'S DISEASE

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**Aim**: Alzheimer's disease (AD) is an irreversible, progressive brain disorder and the most common cause of dementia among older adults. It is supposed that immune system's dysregulation is a key factor in the AD pathogenesis, particularly microglial immune cells. Microglial cells are hindered by the amyloid to perform their normal function – to clear debris and toxic materials from the brain. On the other side, neuroinflammation is regulated by pattern recognition receptors (PRRs) and their signaling pathways, like RIG-1 signaling. The aim of our study was to determine the presence and significance of variants in microglial and antigen-presenting genes in our cohort of AD patients.

**Materials and methods**: We selected 73 single nucleotide variants from literature data with an evidence to be linked with the immune involvement in AD. We tracked them in our exome data of 66 individuals with AD which consists of 162159 variants in protein-coding genes and control group of 100 individuals.

**Results:** Only 15 out of 73 immune associated variants were detected in our AD patients. Two variants connected with RIG-1 signaling were statistically significant: rs1801274 in *FCGR2A* (Fc fragment of IgG receptor), p-value=0,000021 and rs1736090 in *CTSB* (cathepsin B), p-value=0,0265. Neither of the variants connected with the microglia reached statistical significance.

**Conclusion**: In spite of the role of the microglial genes in amyloid plaque accumulation in AD brain, none of the microglia-associated variants in our data was statistically significant. The role of RIG-1 signaling in AD pathogenesis was confirmed.

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