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Current Position

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Current Project Members

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Keywords

Late onset Alzheimer's disease (LOAD) – Rare variants - Metabolomics

Science

Alzheimer's disease (AD), the most common form of dementia, is a neuro-degenerative disease that affects over 5% of individuals older than 65 years of age. Pathologically AD is characterized. In early-onset AD by extra-cellular plaques of β -amyloid ($A\beta$) and neurofibrillary tangles consisting of hyper phosphorylated tau protein brain changes are the primary cause of AD. In contrast to early-onset AD, the more common Late-Onset AD (LOAD) is a complex disease with considerable environmental and genetic components (heritability ~60-80%). New pathways identified in genetic (immune response, cholesterol transport, regulation of endocytosis, and proteasome-ubiquitin activity) and clinical epidemiological AD research (vascular and metabolic pathology) may guide discovery of new systemic biomarkers and may guide future (preventive) interventions. Various metabolic processes at the core of AD dictate a search for biomarker profiles to characterize subgroups that reflect the interaction between genes, proteins and environment. In the metabolomics projects I am involved in we aim to identify blood biomarkers to predict cognitive decline in an early stage.

The apolipoprotein E (APOE) is the gene most strongly associated with common LOAD. Genome-wide association studies (GWAS) have identified genetic variants for LOAD at multiple loci Variants identified through GWAS are typically common low-penetrant mutations that explain a limited percent of the population attributable risk and contribute little, beyond APOE, to the prediction of LOAD. Recently new rare variants have been implicated in LOAD, one in the known APP gene and two in new genes, TREM2 and PLD3. In my research I aim to identify other rare high risk variants, using exome chip and exome sequencing data. To improve risk prediction and help pave the way towards more personalized approaches to prevention.

Current Research Projects

Association of the variants on the Exome chip to AD.
Association of AD genes to metabolomic data.