Both rare and common ABCA7 variants are associated with AD

In order to test whether a previously identified genetic variant is associated with AD, we performed a study involving a small cohort of patients with AD. The results of this study showed that both rare and common variants in the ABCA7 gene were significantly associated with AD.

A rare PLCG2 missense variant is associated with AD in the ADSP cohort

The same cohort of patients was also tested for associations with AD using the PLCG2 gene. The results showed that a rare missense variant in PLCG2 was associated with AD in the ADSP cohort.

PRIORITY OF LOAD GENETIC VARIANTS

Prioritization was based on:

- Replication across multiple studies
- Pathogenicity of SNPs
- Allele frequency
- Conservation of mouse to human gene
- Expression in the brain
- Relevance of gene pathway to Alzheimer’s disease
- Association of variants in distinct pathways, as shown below

PHENOTYPING OF NOVEL MODELS

1. Assess impact of novel variants on AD outcomes
2. Prioritize new models for deep phenotyping

DEEP PHENOTYPING GOALS:

- 1. Compare mouse models using translatonally relevant measures
- 2. Stage models for prescreening testing

FURTHER INFORMATION

- MODEL-AD: www.modelad.org
- AMP-AD Knowledge Portal: http://www.synapse.org/ampad
- Jax AD models: https://www.jax.org/alzheimers
- A2Forum research models: http://www.a2forum.org/search-models

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