Genome-Wide association identifies susceptibility loci for intracranial aneurysm

OCR Number: OCR 5066

Description:

Research estimates that 2-5% of the general population has an undetected intracranial aneurysm (IA), which is balloon-like dilation of blood vessels in the brain. Approximately 1 to 2% of IA patients suffer from aneurysmal rupture, which account for 500,000 cases of hemorrhagic stroke annually. Aneurysmal hemorrhage is often the first sign of the disease and carries a very poor prognosis with half of the patients not surviving this initial hemorrhage. The causes for IA formation or rupture are not well understood. People suspected to be at risk for IA can undergo diagnosis using CTA scan and MRA. However there are no widely accepted guidelines for identifying high-risk populations.

We identified 8 genetic loci for IA susceptibility. These common variants can be utilized in the development of cost-effective and easily applicable genetic screening tests. In particular, the genotype of the patients at a particular locus, such as EDNRA, can be used not only for risk prediction but also for treatment guidance, including but not limited to the decision whether a patient is likely to respond to a specific medication.

Application: The significant associations of these loci (with IA susceptibility have implications for pre-morbid diagnosis of individuals with IA. This is the first study linking endothelins to brain aneurysm.

Advantages: We have identified 8 high risk Loci for intracranial aneurysms. Susceptibility assessment based on genotyping for these risk alleles can be used as diagnostic markers for the pre-morbid diagnosis of IA patients.

Publications:


Licensing Contact: David Lewin
david.lewin@yale.edu