



WSU School of Medicine Researchers Participate in a Landmark Multicenter Study on Genetics of Vascular Diseases

A study titled ‘The same sequence variant on 9p21 associates with myocardial infarction, abdominal aortic aneurysm and intracranial aneurysm’ and published today in the online edition of *Nature Genetics* (www.nature.com/ng) reports that a genetic variant called a SNP (pronounced “snip” and referring to a single nucleotide polymorphism) on chromosome 9p21 previously associated with an increased risk of heart attack is also associated with up to 70% increase in risk of abdominal aortic aneurysm (AAA) and intracranial aneurysm (IA). This variant is the first to be described that affects the risk of AAA and IA in many populations.

AAA and IA are the most common aneurysms – balloon-like protrusions on arteries in the abdomen and head, respectively – and are potentially lethal if they are left untreated and burst. Wayne State University School of Medicine researchers Helena Kuivaniemi and Gerard Tromp have been studying these two diseases for more than 15 years and have co-authored several publications on the genetics of AAA and IA. The Wayne State University School of Medicine researchers were supported through grants from the National Institutes of Health’s National Heart, Lung, and Blood Institute and the National Institute of Neurological Disorders and Stroke as well as the American Heart Association. Their extensive patient collections obtained through active collaborations with multiple clinical sites in the U.S., Belgium, Canada and Finland, were included in the study. Three members of the Kuivaniemi-Tromp research group at the Center for Molecular Medicine and Genetics are also co-authors in this study. Guy Lenk and Shantel Weinsheimer were WSU graduate students and Yoshiki Kyo was a post-doctoral fellow.

Six months ago, deCODE reported that this SNP confers significantly increased risk of coronary artery disease (CAD) and accounts for roughly one-third of early-onset heart attacks. This discovery has now been replicated in studies of ten populations. The study published today was aimed in part at investigating how this risk is conferred. To do so, the deCODE team – in collaboration with academic researchers and thousands of their patients from Iceland, New Zealand, the Netherlands, Finland, the U.S., Belgium, Sweden, Italy, Denmark and the U.K. – analyzed the association of the variant with several other cardiovascular conditions. The results show that the ‘G’ allele is suggestively but not definitively associated with conditions involving the buildup of atherosclerotic plaque, including peripheral artery disease and large artery stroke. However,

those with one copy of this version of the SNP had a more than 30% greater risk of AAA or IA than non-carriers. Those with two copies – that is, who inherited the risk variant from both parents – were at more than 70% greater risk of AAA or IA than were non-carriers. What heart attack and these types of aneurysm have in common is that all three can result from an abnormal or deficient remodeling or repair of vessel walls.

About deCODE

deCODE is a biopharmaceutical company applying its discoveries in human genetics to the development of drugs and diagnostics for common diseases. deCODE is a global leader in gene discovery — its population approach and resources have enabled it to isolate key genes contributing to major public health challenges from cardiovascular disease to cancer, genes that are providing it with drug targets rooted in the basic biology of disease. deCODE is also leveraging its expertise in human genetics and integrated drug discovery and development capabilities to offer innovative products and services in DNA-based diagnostics, bioinformatics, genotyping, structural biology, drug discovery and clinical development. deCODE is delivering on the promise of the new genetics.SM Visit deCODE on the web at www.decode.com.

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