Genetic risk for atypical heart attack in women identified

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New research published by teams from Leicester, UK and Paris, France in collaboration with international partners from the US and Australia, has found a common genetic factor that confers a significant risk of

atypical heart attacks in women.

The genetic factor, located on chromosome 6, increases the risk of developing spontaneous coronary artery dissection (SCAD). This type of heart attack almost exclusively affects young to middle-aged women, including in some patients around the time of pregnancy.

Spontaneous coronary artery dissection (SCAD) is a peculiar and atypical form of heart attack that appears to affect women in apparent good health. The underlying mechanisms of the disease remain poorly understood but affected patients develop a bleed or bruise within the wall of a coronary artery. This leads to external compression of the artery preventing blood flowing normally to the heart muscle leading to a heart attack.

Dr. David Adlam, interventional cardiologist at Leicester's Hospitals and Associate Professor at the University of Leicester, jointly lead the work. He said: "It has been shown that a significant proportion of people with a SCAD-type heart attack also have abnormalities in other arteries elsewhere in the body. Recent studies have identified a genetic factor on chromosome 6 as being associated with an increased risk of fibromuscular dysplasia, a type of arterial abnormality common in SCAD patients. In this study we investigated whether this factor was also associated with SCAD. And the answer is yes, this genetic factor—the A allele of the genetic variant rs9349379 located in the PHACTR1 gene (which stands for Phosphatase and actin regulator 1) - is also a risk factor for SCAD.

"Interestingly, it also appears to give a small protective effect from a classic heart attack, for which another allelic form of the same genetic variant (G) is already known to increase the risk in both men and women."

Dr. Nabila Bouatia-Naji, Director of research and team leader at the Paris Cardiovascular Research Center at INSERM, Paris, France said: "Another important feature of the A allele is that it is very common in the general population and it confers a moderate risk increase of around 70 per cent, indicating that many other genetic factors must also contribute to genetic susceptibility to SCAD.

"As with any disease that has many genetic and environmental factors that contribute to its development, SCAD needs to be investigated with a more holistic approach to better understand it and be able to improve the management of patients affected. Cardiovascular disease is now the leading cause of death among women after breast cancer, so better understanding these atypical heart attacks by the medical and scientific profession would contribute to a better grasp of women's cardiovascular health in general."

Although it is considered to be rare, recent studies indicate that it may actually be responsible for up to one-third of acute heart attacks in women under 60 years of age. The rate of these types of heart attack is clearly underestimated because the patients may not present with classical risk factors. Furthermore, a correct diagnosis is complex; it requires sophisticated imaging techniques and clinical expertise.

The research teams in Leicester and Paris have a common ambition to work with international colleagues to establish an exhaustive genetic map of SCAD and to highlight the biological consequences of the associated genetic factors. The ultimate goal is to be able to better understand the cause and best treatment for this condition.

More information: Association of the PHACTR1/EDN1 Genetic Locus With Spontaneous Coronary Artery Dissection. *Journal of the American College of Cardiology*. DOI: 10.1016/j.jacc.2018.09.085

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