Genome-wide association study identifies variations in the DNA of women that predispose them to developing endometriosis

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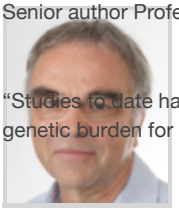
This is the first genome-wide association study to give robust evidence of variations in the DNA of women with endometriosis, showing that moderate-to-severe endometriosis is significantly more genetically driven than minimal-to-mild disease.

Researchers in the UK, Australia and USA have identified two regions on chromosomes 7 and 1 associated with endometriosis, a disease which can cause severe pelvic pain and infertility in women. The study, published today in *Nature Genetics* [1], gives the first robust evidence of variations in the DNA of women with endometriosis of European ancestry that predispose them to developing the condition, which affects an estimated 176 million women worldwide during their reproductive years.


Professor Grant Montgomery

Senior author Professor Grant Montgomery, Queensland Institute of Medical Research in Brisbane, explains:

"Studies to date have established that endometriosis is heritable, but have not addressed genetic burden for different disease stages. The International Endogene Consortium (IEC) has been able to conclude that moderate-to-severe endometriosis is significantly more genetically driven than minimal-to-mild disease, which has implications for how we research the condition further", said Montgomery.

The International Endogene Consortium (IEC) conducted the largest genome-wide associated study to date, and the first in women of European ancestry, comparing more than 500,000 DNA variants between a total of 3,194 endometriosis cases from Australia and the UK, and 7,060 controls. In the second step, involving analysis of a further 2,392 cases and 1,646 controls from the USA, a common DNA variant on chromosome 7 was confirmed to carry an approximately 20% increased risk of endometriosis.

The variant appeared to carry a greater increased risk (40%) of moderate-to-severe disease.

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The IEC study also confirmed suggestive evidence of a DNA variant on chromosome 1, previously reported to be associated with endometriosis in Japanese women [2]. This variant, which increases endometriosis risk by an estimated 16% in the IEC study, lies close to the *WNT4* gene, which is important for hormone metabolism and the development of the female reproductive tract, especially the ovaries, making it an important biological candidate for involvement in endometriosis.

Dr Krina Zondervan

Senior author Dr Krina Zondervan, Wellcome Trust Fellow at the University of Oxford, comments: “Further research to understand the effect of these variations on cells and molecules in the body will need to be conducted. We are confident that our findings will ultimately help to improve methods of diagnosis and treatment of this devastating disease, which affects millions of women during the prime years of their lives”, said Zondervan.

Dr Stacey Missmer

Senior author Dr Stacey Missmer, Harvard Medical School and Brigham & Women’s Hospital in Boston, adds: “It is our goal to continue to uncover the genetic and molecular aetiology of endometriosis through ever expanding large-scale collaborative research. Only through international collaboration are we able to maximise the opportunity to improve our understanding of the causes of endometriosis”.

The International Endogene Consortium (IEC) was established in 2008 and builds on over 15 years of research into endometriosis by the respective groups. The present study was funded by a grant from the Wellcome Trust, with additional funding provided by the Australian National Health and Medical Research Council.

References

1. Painter JL, et al. Genome-wide association study identifies a locus at 7p15.2 associated with endometriosis. Nature Genetics doi: 10.1038/ng.731
2. Uno S, et al. A genome-wide association study identifies genetic variants in the DKHN2BAS locus associated with endometriosis in Japanese. Nature Genetics 2010;42(8):707-10 .

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