

# Gene science closes in on endometriosis

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Endometriosis affects one in 10 women and can cause severe pelvic pain. Credit: University of Queensland

In the world's largest study into the genetic causes of endometriosis, University of Queensland researchers have helped identify five new gene regions linked to the disease.

The genome-wide study analysed data from more than 200,000 women to help uncover genetic causes of the disease – a discovery that will help pave the way for future treatments.

Endometriosis affects one in 10 women and can cause severe pelvic pain, fertility problems, and bowel or bladder problems. The condition causes tissue similar to the uterus lining abnormally growing outside the uterus.

The research was jointly led by UQ Institute for Molecular Bioscience's Professor Grant Montgomery, and QUT Institute of Health and Biomedical Innovation's Associate Professor Dale Nyholt.

Professor Montgomery (pictured) said the findings were a step towards identifying the currently unknown causes of endometriosis.

"Although [genes](#) are not the only deciding factor

for whether or not a woman will develop endometriosis, it's important to identify genes increasing a woman's risk so we can understand the factors causing this disease," he said.

"We know genetic factors account for 50 per cent of a woman's risk of developing endometriosis; however, it is not one gene mutation that causes this disease, but multiple genetic variations that contribute to incrementally increasing a person's risk."

Professor Montgomery said the study confirmed nine of the 11 previously reported gene regions associated with endometriosis, as well as uncovering five new regions.

"Identifying the [genomic regions](#) is a critical first step, but in each of those regions there are a number of genes that could contribute to causing the disease," he said.

"Interestingly, the [genetic regions](#) we identified in this study suggest specific genes involved in the regulation and response to oestrogen are linked to endometriosis."

Professor Montgomery said the discovery gave scientists the opportunity to conduct more targeted research into the [genetic causes](#) of endometriosis.

"Although there is still a long road ahead before [endometriosis](#) can be diagnosed without laparoscopic surgery, or new treatments are created to better treat the [disease](#), we are making significant progress towards achieving this goal."

**More information:** Yadav Sapkota et al. Meta-analysis identifies five novel loci associated with endometriosis highlighting key genes involved in hormone metabolism, *Nature Communications* (2017). [DOI: 10.1038/ncomms15539](https://doi.org/10.1038/ncomms15539)

Provided by University of Queensland

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