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Endometriosis Research at QIMR

In this update we aim to provide you with information on our progress in finding causes of endometriosis since our last communication in early 2005. We have written it with study participants and families in mind, but it may be of interest to others. We continue to be extremely appreciative of the part played by all our study participants and their family members in our research. Your information and samples continue to be useful. In some cases we might need to come back and ask more questions, so we hope you will consider ongoing involvement.

In summary, we have made substantial progress, but cannot yet identify an individual gene(s) responsible for increasing women’s risk of developing endometriosis.

Genes behind endometriosis… what have we found so far?

In September 2005 we published the first major paper of results from the study in collaboration with Stephen Kennedy’s group in Oxford. In the paper we showed evidence, based on genetic information from families with sisters diagnosed with endometriosis, that important genes for endometriosis lie in a region on chromosome 10, and quite probably also in a region on chromosome 20. This has narrowed down the search area, but we’re still “combing” the region and testing for evidence in specific genes. This scientific paper is available at http://www.pubmedcentral.nih.gov/articlerender.fcgi?tool=pubmed&pubmedid=16080113.

We also published a paper showing results of testing for variants in a gene related to progesterone, a female hormone receptor gene, which other small studies had suggested might influence endometriosis. Our results showed no evidence for association, and the search continues.

Another method we have used is to pool fractions of DNA from women with endometriosis and compare the frequencies of genetic variants in this pool with those in pooled DNA from a group of women who are unlikely to have had endometriosis. Of course, we can’t be 100% certain that all women in the latter “control” group have never had endometriosis because only surgery can tell this. Results show DNA pooling is a promising strategy.

What is happening with the genetic study now?

The extracted DNA samples from the nearly 10,000 people in our study are now stored in our freezers, and we have been using them for different tests of association with endometriosis. The work is now focused on genotyping in the lab and statistical analysis of the genetic data. We were awarded a 3-year grant by the National Health and Medical Research Council for 2005-2007 to make progress in finding genes using information and DNA from the Australian participants.
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We are hopeful of success with an application for funding from the US National Institutes of Health to continue our genetic work with the Oxford group, but we do not yet know the final outcome.

Our team is now being led by Dr. Grant Montgomery, who runs the Molecular Epidemiology laboratory here at QIMR. As well as collaborating with Dr. Stephen Kennedy’s team from the University of Oxford, we plan to collaborate with A/Prof Peter Rogers from Monash University in Melbourne for laboratory work on gene expression in relevant tissue samples, such as from the lining of the uterus (the endometrium) and endometriotic lesions themselves.

Risk Factors for Endometriosis: Findings from a Case-Control Study

Tanya Bell’s study, “Risk factors for endometriosis”, also conducted at the Queensland Institute of Medical Research, is now complete. Her study involved the participation of 268 women with endometriosis who were compared with 244 women without endometriosis. All of the women in the study completed a brief, self-administered questionnaire that covered topics such as menstrual cycle characteristics, personal habits such as smoking and talc use, as well as physical characteristics such as weight.

Analysis of the collected data has provided a number of interesting results. One of the strongest findings showed that women with endometriosis were more than 4 times likely to experience shorter menstrual cycles (less than 24 days). Women with endometriosis were also over 1.5 times more likely to report experiencing heavy bleeding during menstruation and 2.5 times more likely to report having pain during menstruation than women without endometriosis. Interestingly, women who reported being overweight at age 10 were more than 2.5 times more likely to have endometriosis than those who were of average weight at this age.

In contrast to observations from other studies, tampon use did not appear to increase the risk of endometriosis and factors such as exercise and smoking did not appear to decrease the risk of endometriosis.

The results from this study highlight the need for awareness of menstrual characteristics such as short cycle length, heavy menstrual bleeding and pain during menstruation that may indicate the early onset of endometriosis. Hopefully, an increasing awareness of signs of endometriosis will decrease the long delays between symptom onset and diagnosis (averaging 7.4 years) that was observed in this study.

Study on breast density in women with endometriosis

This study, funded by the National Health and Medical Research Council to the end of 2006, is being conducted jointly by the University of Melbourne's Centre for Genetic Epidemiology (Prof. John Hopper and others) and our group at QIMR. The study has been inviting participants in the Genes Behind Endometriosis study who are in the families with sisters diagnosed with endometriosis and are aged 40-70 years old, for permission to scan a recent mammogram.

We are interested in finding out if, for hormonal reasons, breast density differs in women with endometriosis compared with women who have not had endometriosis, and therefore if there is any joint influences of particular genes.
We have been very pleased indeed with the response from participants in our Endometriosis study and have even found some additional sisters, who were not previously needed for the Endometriosis study, who have been willing to participate. We thank everyone who has participated for their time and information, and for their consent to us accessing information from their mammogram. Although recruitment for the study will be completed by the end of this year it will take some time to retrieve all the mammograms, so we might need to be in touch again regarding this, particularly in cases where the mammograms were done privately rather than by BreastScreen. If you have any questions at all regarding your participation, please contact our Project Coordinator, Clare Redfern, on 07 3362 0248.

Pilot project on why people participate in studies like our genetic study of endometriosis

During the long course of finding women and families interested in participating in our genetic study of endometriosis, the question arose in my mind as to “why” people volunteer for such studies. A very small pilot project we conducted showed that as well as wanting to find answers and help others avoid the pain and problems they had experienced, there were features of endometriosis itself that were important in some people’s motivation. These included the fact that it had been for too long an unrecognized and “hidden” disease, and people were pleased that it was out in the open and being studied and discussed.

9th World Congress on Endometriosis, September 2005

2005 was a very productive year for advancing understanding about endometriosis, in terms of both patient-doctor understanding and of genetic knowledge. The 9th World Congress on Endometriosis was held in Maastricht, The Netherlands, September 14-17th, and had as its theme “The Patient as Partner”. Many women from endometriosis support groups were able to be present along with doctors, nurses and researchers from many fields including nursing, gynaecology, epidemiology, genetics and the social sciences. The meeting will certainly lead to better communication between all groups and improved information about endometriosis. It is certainly encouraging that there are so many research groups and doctors researching the causes of endometriosis, and trying to find ways of improving the diagnosis and treatments available. Dr Grant Montgomery and Dr. Susan Treloar from QIMR both attended the World Congress and gave papers on results from our study.

10th World Congress on Endometriosis, March 2008, Melbourne

The next World Congress will be held in Melbourne from March 11-14 2008 (see http://www.wce2008.com/invitation.htm) under the chairmanship of Professor David Healy, Chair of Monash University’s Department of Obstetrics and Gynaecology. We hope by then we will have some specific results on particular genes that will lead to greater understanding of why some women rather than others are susceptible to endometriosis. QIMR’s work on endometriosis was also presented at the recent International Congress on Human Genetics held in Brisbane and at other international meetings.

Asia Pacific Endometriosis Endometriosis Alliance http://www.endometriosis.org/apea.html

This new alliance including support groups, gynaecologists and researchers in the Asia Pacific region was formed at the 2005 World Congress. The effort to form the Alliance was driven by Dr. Susan Evans from Adelaide, author of “Endometriosis and Other Pelvic Pain” (http://www.drsusanevans.com/) and Deborah Bush, CEO of the New Zealand Endometriosis Foundation.
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(https://www.nzendo.co.nz/index_home.htm). The Alliance continues the theme of the Congress – let’s all work together. It is very encouraging that there is such a strong body of people all working towards the same ultimate aim of improving the diagnosis and treatments for endometriosis and the lives of people affected by the condition.

Vale Kim Leanne Goodwin
We note with great sadness the very recent passing (2nd August 2006) of Kim Goodwin, immediate past President of the Endometriosis Association (QLD) Inc. Kim died tragically at the young age of 38. She was an inspiration to all who knew her in terms of warmth, tenacity and promotion of better information about endometriosis. Kim was a very strong advocate for more research on endometriosis and to this end, with her family, was a wonderful supporter of our endometriosis research program at QIMR. We will miss Kim greatly, as will her many friends and colleagues in endometriosis support groups and professional circles.

Endometriosis Associations
We greatly value the work done by the Endometriosis Associations in each State. They do an extraordinary job as volunteers, providing information and support in relation to endometriosis. As researchers we take very seriously our accountability to our “community”, especially women diagnosed with endometriosis and their families, as well as to our study participants. We are very honoured that Dr Susan Treloar has been offered the position of Patron of the Endometriosis Association (QLD) Inc.

For information and support for endometriosis, the following associations can be contacted:

Endometriosis Association (Qld) Inc.
http://www.qendo.org.au (e-mail via website)
Telephone 07 3321 4408 (messagebank service, please leave message).

Endometriosis Association (Victoria) Inc.
E-mail:info@endometriosis.org.au
Helpline and office: 03-9457 2933

Best wishes,

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Our study web site is http://genepi.qimr.edu.au/studies/en