The International Endogene Study and Genes Behind Endometriosis Study

Firstly, we hope that the last year brought you better health and happiness. Here is a brief update on our endometriosis research being conducted from the Queensland Institute of Medical Research in Brisbane, Australia.

What is happening with the study now? Recruitment for just the Australian component of the study was a huge exercise. In June 2002 we stopped accepting new participants and families into the study. Throughout Australia we achieved the largest collection of sibling pairs (sisters in our case) for a complex disease that has never been achieved anywhere in the world! In total, we had around 4,000 women with surgically confirmed endometriosis participate in our study. Adding parents and other family members brought the total to around 10,000 people across Australia! All the extracted DNA samples are now stored in our freezers, and we have been using them for different tests of association with endometriosis.

What have we found so far? We have completed our "genome screen" of the 931 families we found where 2 or more sisters both had endometriosis. We have been looking to find on which part of which chromosomes the sisters were sharing more genetic markers than we would normally expect. This has narrowed our search down to two chromosomal regions. We hope to publish these results in a top medical / scientific journal early in 2005.

We are now looking in these regions to see if there are any likely known genes that could be relevant. Other researchers have come up with a lot of genes that might be relevant, but none of these so far have been found to be causative. Genes are tiny sequences and there are hundreds of genes in the areas we are looking in (that’s a tiny fraction of all the genes that we have – around 30,000 is one estimate!). We have started testing some possible genes for their association with endometriosis, using samples from all the women we found who didn't have a sister with endometriosis (and their parents).

Who is funding our research? We have been awarded a 3-year grant from the National Health and Medical Research Council for 2005-2007 to make progress in finding genes. The Gene CRC (Cooperative Research Centre for Discovery of Genes for Common Human Diseases) that previously funded our project wound up in mid 2004. We have put in an application for more funding from the US National Institutes of Health, but it will be some months before we hear news of this application. Our team is now being led by Dr. Grant Montgomery, who runs the Molecular Epidemiology laboratory here at the...
Institute. 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 lab work on gene expression.

**New Australian study on breast density in women with endometriosis.**
This new 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 will be 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. Endometriosis is a hormonally-influenced condition and women often take hormonal type medications for endometriosis. We know that breast density is a risk factor for breast cancer. A Scandinavian study some years ago showed a small increase in risk of breast cancer in women diagnosed with endometriosis. Therefore we think it’s important to investigate whether breast density differs in women diagnosed with endometriosis compared with other women, and whether breast density is influenced by genes, as suggested by results of an earlier twin study run by Prof. Hopper and his colleagues. The density of a woman’s breasts can only be assessed from a mammogram, not by feel. We really hope that women approached will be interested in helping this new study, even by having a mammogram if they previously haven’t had one.

**Information and support on endometriosis in Australia.** For information and support in relation to endometriosis itself, we really support the work done by the Endometriosis Associations in each State, and suggest that they are the best places to get information. There are some good websites now where you can get information and links to other sites. For example:
Endometriosis Association (Qld) Inc. http://www.qendo.org.au or telephone 07 3836 3752 (pager service, please leave message). OR Endometriosis Association (Victoria) Inc. http://www.endometriosis.org.au 1800 069 967 (Freecall within Victoria between 9am and 1pm weekdays. If calling from outside Victoria, please call 03 9879 2199).

Our very best wishes,
Susan Treloar, PhD

If you have any queries please do not hesitate to contact the project co-ordinator Clare Redfern on 07-3362 0248 or E-mail: clare.redfern@qimr.edu.au or free call tel. 1800 632 576 (from anywhere in Australia)
Our study web site http://genepi.qimr.edu.au/studies/en