The International Endogene Study and Genes Behind Endometriosis Study  UPDATE  2003

We are sorry that we didn't get a newsletter sent out to study participants in 2002. We did mail around 900 gynaecologists who had helped the study, but our pre-Christmas plans were overtaken by the need to prepare for a videoconference with our University of Oxford collaborators in December. Like most people, we had a very busy year and we would like to tell you now what has been happening with the study. We ask if you would kindly pass this newsletter on to members of your family, as unfortunately we do not have the budget this year to mail everybody who helped us. Firstly, we hope that the last year brought you better health, peace and happiness.

What is happening with the study now?
Recruitment for just the Australian component of the study was a huge exercise. Throughout Australia we achieved the largest collection of sibling pairs (sisters in our case) for a complex disease that has ever been achieved anywhere in the world! In total, we had around 4,000 with surgically confirmed endometriosis participate in our study. Adding parents and other family members brought the total to around 10,000 people across Australia! We had a wonderful team of research assistants between 1995 and 2002 who helped us in our contact with you (Shona, Rebeakah, Alana, Carmel, Stephanie, Lynette, Cassie, Kim, Jan, Linda, Sue, and Heather). Now they are working on other projects, because our focus is now "in the lab" and "on our computers" for statistical analyses. In the lab we still have Leanne helping with setting up samples, and we have David and Olivia helping with computing.

In June 2002 we stopped accepting new participants and families into the study. So no more blood samples came into the lab for processing. 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?
In December 2002 we completed our "genome screen" of the 931 families we found where 2 or more sisters both had endometriosis. We were 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 2 chromosomal regions. 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 has been found to be causative. Our method has at least focused us on 2 regions, but genes are tiny sequences and there are hundred 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!). Still, adding all the extra families since we wrote to you last time has helped a lot, because it narrowed the search down from 5 to 2 regions. We are exactly where we planned to be.

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). We have an Honours student from QUT, Trudi Armitage, working on one possible gene. Trudi is on a mission to help us find answers.

The Gene CRC (Cooperative Research Centre for Discovery of Genes for Common Human Diseases) is a research organisation that brings together Australia’s leading genetics experts. It has funded our project since mid-1997, but the Gene CRC will wind up in mid 2004. We have had huge support from the CRC Director, Dr. Andrea Douglas, and in the past also from Dr. Matthijs Smith of Cerylid Biosciences Ltd. We already have to think about applying for funds to continue our gene search in 2004. Our collaboration with Oxford has helped, but if we are not lucky in the next year (and sometimes luck is involved), it may take another 3 or so years before we actually pinpoint the genes. I must always apologise for the slowness of this sort of research, but it's very painstaking work. If you would like a copy of our article on the study that appeared in the journal Fertility and Sterility in late 2002, please contact us. We can post or email it to you.

Both the environment and our genes influence most common diseases and disorders. For such complex diseases, the genetic and environmental factors contribute to the disease, and these factors also interact in often-unpredictable ways. Tanya Bell, our PhD student, is still working on her case-control study of risk factors for endometriosis through the questionnaire we have asked some women and their mothers to complete. Tanya hopes to complete her data analysis and writing up this year, so we should have some results to report to you before next year.

Last year we tried to get funding to investigate the role of dioxins (chemicals taken up in the food chain) in endometriosis, but unfortunately we were not successful. In early 2002 there was a lot of interest in genetic factors in endometriosis, and in environmental factors such as dioxins, at the World Congress on Endometriosis held in San Diego. Another small project we are starting up in collaboration with the University of Queensland is to explore what it means for participants to be part of a study like this one. However, in the end there is still a critical need to better understand endometriosis at a genetic level so that we can help the community manage their health, improve existing medical treatments and ultimately find a cure.

**Information for participants**

When we get more definite results they will be posted on our web site. We will also keep participants informed of overall study progress and results through the mail. 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:

1800 069 967 (Freecall within Victoria between 9am and 1pm weekdays.
(If calling from outside Victoria, please call 03 9879 2199).
OR
Endometriosis Association (Qld) Inc.
http://www.qendo.org.au (under construction)
or tel. 07 3836 3752 (pager service, please leave message)

Our very best wishes,

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**Our study web site**  http://genepi.qimr.edu.au/studies/en will eventually have information on our findings.