



Follow-up Study, Spring 2009

The current study of the Boyne Research Institute is the Follow-Up Study of Irish Families with Neural Tube Defects. This is our on-going study, started in 1995. The Follow-Up component started in summer 2008 and will continue into April, 2009. Sharon McGinty, nurse/researcher is the lead interviewer. Sharon is interviewing relatives who have participated in the past. She will also identify and interview relatives who have not previously been interviewed. Many of the recipients of this newsletter will have been interviewed and we thank you most sincerely for your participation.

Why are we doing this study? This follow-up study is designed to see if we can confirm previously unexpected results. Previously, when we interviewed relatives whose children had been born most recently (1996-2002), we found that mothers who took folic acid before and during early pregnancy (as recommended by the government to prevent neural tube defects) actually had fewer children with birth defects. That is, birth defects of all kinds considered together. This finding is included in a paper that has been submitted for publication.

So far: Sharon has completed 367 interviews with relatives between the ages of 18 and 44 years, not only on the island of Ireland, but also from Canada and the United States, Europe and Australia, among other countries. We hope to complete the study, clean the data and do the data analysis by the end of the summer.

2009 Summer Student Programme, July-August, 2008.

Each year the Boyne Research Institute offers a two-month summer training programme to two Leaving Cert students in Drogheda-area schools. Following an application and interview, two students are selected (usually one male and one female). Their work includes helping with our research projects, including data entry, and completing their own research project. Each student identifies a different topic, carries out the research, and prepares a formal powerpoint presentation. They present their work at a reception for an invited audience, including the Mayor of Drogheda and board members.



Patrick Sullivan came to us from St. Joseph's Christian Brothers School, and chose to investigate the way that the protein called RFC (reduced folate carrier) enables folate to enter the cell after digestive processing in the intestine.

Ellen Mathews was recruited from Our Lady's College, Greenhills. Ellen was the lead on preparing the family trees using a computer program called CYRILLIC.



Eimear Kelleher was in her final year of her degree in Human Nutrition and Dietetics from Dublin Institute of Technology. She completed three months internship with the Boyne Research Institute. Eimear's training and interests in nutrition fit in well with our current studies into the role of folic acid in the diet and level of folate in blood. She investigated the way that alcohol intake modified level of blood folate.

Mission The Boyne Research Institute is a community-based research facility. Our studies are directed towards a better understanding of the origins and prevention of birth defects and the long term consequences of cancer during childhood. In 2008 we were supported by the Jones Foundation of Washington, DC, by the Drogheda and District Charity Chest, by corporate and private donations; we received no government funding.

<p>2009 Staff of the Boyne Research Institute</p> <p>Rebecca Scott, administrator/researcher.</p> <p>Sharon McGinty, research nurse and study manager.</p> <p>Julianne Byrne, epidemiologist, director of the Boyne Research Institute.</p>	<p>Your donations to assist the work of the Boyne Research Institute are much appreciated</p> <p>In Ireland: Boyne Research Institute Duke House, Duke Street, Drogheda, IRELAND Tel: +353 (0)41-9836041 Email: admin@boyneresearch.ie An Irish-registered charity (no. 10275)</p> <p>In the USA: Boyne Research Foundation 1656 Newton Street, NW, Washington, DC 20010, USA Tel: +1 202-234-8719; Email: admin@boyneresearch.ie A 501(c)3 organization.</p>
--	--



Genotyping study, 2009

Background. We formed a collaboration with the Centers for Disease Control and Prevention (CDC) in Atlanta, Georgia, USA. The purpose was to evaluate some of the genetic components in blood samples from relatives. Blood samples were obtained during 2007 as part of our Pre-Fortification Study. They were frozen and shipped to Atlanta for genotyping. CDC evaluated 5 SNPs (single nucleotide polymorphisms, pronounced 'snips'), that is areas of the genome that control the metabolism of folic acid. The data analysis was chiefly aimed at seeing if the maternal inheritance patterns were related to any of these SNPs. You can find out more about CDC at www.cdc.gov.

Preliminary Results. Four of the five SNPs were not related to maternal inheritance, but one was. This SNP is called RFC (reduced folate carrier). One form of RFC occurs more often among relatives related through the mother. We also looked for an association between any of the SNPs and the level of blood folate. Here, we found that one form of another SNP was related to low levels of serum

folate. In the coming weeks we will be seeing if these patterns affect some families more than others. Once we are finished with the analyses, the results will be prepared for publication.

What's next? We have established another collaboration with scientists from Columbia University in New York. This collaboration also supplies genetic expertise to complement the information obtained by interview. Here, we hope to evaluate the genetic mechanisms that might explain the patterns of maternal inheritance that we are seeing in our families. Take a look at our website for further developments.

(www.boyneresearch.ie).

Annual Reports

Annual Reports for 2007 and 2008 are available at http://www.boyneresearch.ie/resources/Ann_Report_07_4Sept08a1.pdf

Recommendation

One 400 microgram tablet of folic acid taken every day by all women who are capable of becoming pregnant will prevent many cases of neural tube defects.

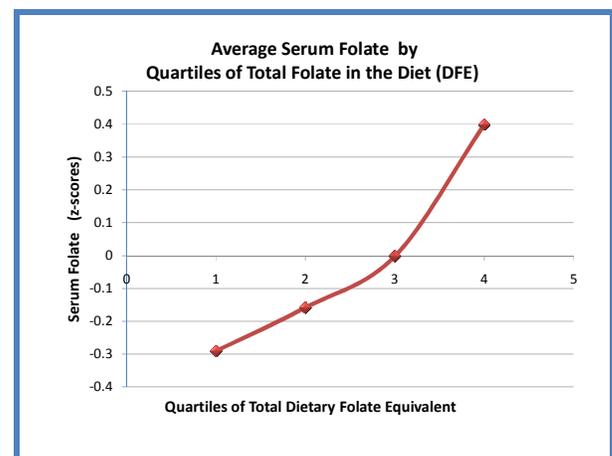
Results from the Pre-Fortification Study of 2007

Background: The Irish Government announced plans to fortify flour with folic acid in order to prevent neural tube defects and improve the chances that babies will be born healthy. We wanted to be sure that relatives in Irish families with neural tube defects would benefit from the government's programme.

Methods: We measured blood (serum) folate levels in relatives during the summer of 2007. Once fortification has been implemented, we plan to return to the relatives and ask for another blood sample. Participants also completed two questionnaires -- one about their lifestyles and health and one about food habits. From the second questionnaire we were able to measure the amount of folate in each participant's diet.

Results: The graph shows a strong relationship between the amount of total folate in the diet (including tablets, fortified foods and naturally-occurring folate in food) and the level of folate in the blood (measured in z-scores). A paper describing these results has been submitted for publication. Extra tubes of blood were stored (frozen) for future genetic tests.

Some results from these tests are described above (Genotyping study, 2009).



What's next: Additional genetic tests are planned with the samples provided in 2007.