People of the *British Isles*

Newsletter Issue 3 - August 2009

Welcome

Welcome to the third newsletter of the "People of the British Isles". Since we last updated you about the progress of the study we have been extremely busy trying to collect samples from other parts of the country to reach our target of 3,500 samples.

A busy evening in Canterbury

We now have over 4,000 samples and thanks must go to our collaborators throughout the country who played an important part in collecting them. On top of that, a large number of the samples was collected by those of us in Oxford, with Tammy Day organising all the collection events. We would like to thank each one of you for participating in our project.

Volunteer showing his family coat of arms

We would also like to take the opportunity once again to thank the media around the country for helping us by publicising the study. The various editors of newspapers and journals and the BBC all played their part as did voluntary societies such as family history and genealogy societies, agricultural shows, local libraries, Tourist Offices, the clergy, GPs and professors of other universities, not to mention close friends and families.

Where do we go from here?

Now we have the samples, the next stage is to do the major laboratory work and get the data on genetic markers in the PoBI samples. We have done some initial work, which we plan to publish very soon. We have also become involved in two major projects (the Wellcome Trust Case Control Consortium and the 1,000 Genomes Project), which will generate a lot of data. There is a some more information on these two projects at the end of the newsletter.

The biggest task we now have is to analyse the data that are coming in and to get them published. We will have a huge amount of data from the Wellcome Trust Case Control Consortium project in the near future and it will take some months to go through it all and submit it for publication. We aim to have something by the end of the year and will let you know when the first set of results are out. In the meantime, we are in the process of writing an initial paper on the PoBI samples, which will be referenced whenever data from the study is published. This will mark the completion of the initial project and the samples will be available as a resource for future research.

Explaining the project to volunteers in Canterbury

There is also more exciting news. We have received further funding from the Wellcome Trust and this will allow us to build on the current project. The main aim of this is to try and identify genes involved in facial features. There is a great deal of interest in the genetics of facial features and, in addition, the frequency of genetic variants for facial features may well differ significantly between different parts of the UK. We will also be collecting data on a variety of other normal features including height, hair and skin colour, handedness, milk tolerance, musical preferences and perfect pitch, taste and smell preferences and features of the hand.

Sir Walter and Tammy tracing the journey from Orkney to Ireland

The Balmoral Show, N. Ireland

This will be a collaboration with Professor Tim Spector (St Thomas' Hospital), who has a large collection of twins that we can use alongside samples we collect and Professor Kittler (University of Surrey), who is an expert in the analysis of 3D facial images. The plan is to take 3D photographs of some of the twins for whom there is a lot of genetic data already available, as well as from further volunteers collected as part of PoBI. From these photos, it will be possible to measure particular features and see how they vary throughout the UK. We should then be able to look for the genes that underlie these particular features. We will, obviously, be very careful with these photographs.

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Only researchers directly involved with the project will have access to the photographs and no recognisable photograph of a PoBI volunteer will be published. The photographs will be anonymised so that a direct link cannot be made between them and personal details such as addresses.

You may be interested to know that this phase of the project will not be restricted to new volunteers. Over the next few years, we will be revisiting some of the areas we have already been to. We will let you know if/when are in your area, probably using a mini-newsletter and we would be very interested in seeing many of you again at these events. Obviously, you won't need to supply us with more blood but we will need a cheek swab so that we can match the new data with the old data using genetics. This means that the samples will remain anonymous.

Sir Walter Bodmer

We will also collect further samples from surrounding countries in Europe, which will place PoBI in a European context so that we can investigate in detail the genetic impact these countries have had on the UK. In addition, we will collaborate with Professor Paul Longley (UCL) who works on the geographic distribution of surnames (eg. www.nationaltrustnames.org.uk) to see if genetic patterns differ between people with local surnames and those with surnames found throughout the UK.

Thank you once again for your contribution to the People of the British Isles.

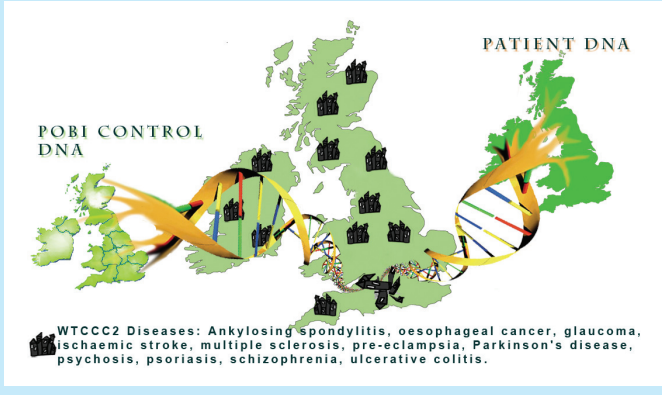
Until the next newsletter…

What are the WTCCC and 1,000 Genomes Projects?

The Wellcome Trust Case Control Consortium (www.wtccc.org.uk) is a large-scale effort to find genes involved in diseases. The first publication in 2007 was based on seven diseases (type 1 and, type 2 diabetes, coronary heart disease, hypertension, bipolar disorder, rheumatoid arthritis and Crohn's disease), with 2,000 patients from each disease and 3,000 controls. The idea behind these studies is to look for genetic differences between cases and controls. If a genetic marker is found more often in patients, it is likely to be associated with the disease. This project looked at 500,000 markers throughout the genome and came up with 39 hits, of which 25 were previously unknown. This is known as a Genome Wide Association Study (GWAS) and since then there have been a large number of further studies.

The WTCCC has now expanded to WTCCC2 and this time they are looking at one million markers in 12 diseases with a total of about 120,000 samples. To be able to publish results from these studies, they have to be reproduced in a separate study and this is where we come in. Three thousand of our samples are being genotyped as controls for these repeat studies, which immediately makes PoBI an important resource for the medical research world. We will also then have a lot of data that we can look at ourselves to see how genetic variation changes across the UK.

Another project we have become involved in is the 1,000 Genomes Project (www.1000genomes.org). This is a project that has become feasible with recent advances in the latest sequencing technology. It is a worldwide collaboration to sequence the genome of 1,000 samples. We are contributing 100 of our samples to this project and it will give us an unprecedented view of human genetic variation. The advances in sequencing technology have been breathtaking. The initial sequencing of the human genome took 10 years and cost $3 billion. At the beginning of last year, James Watson (of DNA double helix fame) had his genome sequenced in 2 months for $2 million and already things have improved considerably in the last year.



It should be pointed out that we had to update our ethical permission for the sequencing so we could only choose samples from those people who volunteered after September 2008.

Please do let us have your comments on, and questions about, this newsletter. You can contact us directly through the main website or by using the details below. Also, please don't forget to tell any friends who have not yet volunteered but fit our criteria of having four grandparents from the same rural area in which they live to get in touch with us either through the website or at:

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