The Genealogical Society of Ireland and the Royal College of Surgeons in Ireland have launched an important All-Ireland project to create a collection of DNA samples from individuals of Irish origin, which will be used to explore human genetic variation in the Irish population.

Over the past decade or so genealogists from around the world have become increasingly intrigued by the possibilities afforded through the advances in genetic genealogy to augment or confirm our traditional record based research. This new project is aimed at promoting an awareness, appreciation and knowledge of genetic genealogy. Operationally the project has two strands, genealogy and genetics.

The Society appointed its Director of Archival Services, Séamus O’Reilly, FGSi, to coordinate the collection of the genealogical data. Dr. Gianpiero Cavalleri of the Royal College of Surgeons in Ireland (RCSI) will direct all the scientific aspects of this new and exciting project.

For over 200 years the RCSI has played a major role in medical education and training in Ireland. Founded in 1784 to train surgeons, today the College provides extensive education and training in the healthcare professions at undergraduate and postgraduate level.

The RCSI is committed to performing high levels of research activity, and helping to drive the Irish economy through the commercialising of intellectual property arising from its research, and the development of collaborative links with industry, educational and research institutions both nationally and internationally.

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This joint project will compile an Irish DNA Atlas through the collection of birth briefs and DNA samples to investigate the diversity of the Irish genome, which is a valuable, yet largely unexplored, resource of the Irish nation. As an island population on the edge of Europe, Ireland has a rich cultural heritage that is the product of ancient migrations from the neighbouring island and from mainland Europe.

Understanding and preserving this history enriches our culture. Although historical records and archaeological studies have uncovered many wonderful aspects of Irish history, there are many questions left unanswered.

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However, this project is not just concerned with the movements and interrelationships of population groups, it also has an immensely valuable contribution to the study of the health of the people of Ireland. Participation in this project is certainly science for the benefit of mankind.

The aims of the *Irish DNA Atlas* Project are twofold, genetic genealogy and health research. A greater understanding human genetic variation in the Irish population is required for two principal purposes: (1) to further our knowledge of the population history of Ireland and its connections with other populations in Europe and (2) to help us understand how genes influence health in Ireland through the creation of a resource for use as ‘healthy’ controls in researching how genes influence common diseases in Ireland, including (though, not confined to) diabetes, heart disease and cancer. Through the collection and scientific analysis of this type of data it may be possible to identify genetic risk factors for disease and with this information, improve the nature of future treatments, including drug design or indeed lifestyle decisions on how to prevent the development of disease in the first place. As the Royal College of Surgeons is one of Ireland’s foremost research institutions, this project will have both a national and an international dimension involving researchers in a number of different fields.
What is DNA?

As we are all aware, the human body is made up of millions of cells, most consist of a complete set of our genes. These genes contain the blueprint for the development and functioning of our bodies. They also contribute to our physical characteristics such as height, eye, hair and skin colours, blood type etc all inherited, at least in part, from our parents and ultimately from our ancestors.

DNA is the chemical substance that makes up these genes and therefore, DNA can be used to explore the genetic history of an individual or a population. Three broad approaches are taken to genetic ancestry studies and these differ based on the section of DNA that is being studied. They are (1) Y chromosome, (2) mitochondrial DNA (mtDNA) and (3) whole genome. These three general approaches are complementary and have strengths and weaknesses.

Chromosomes are thread-like structures which contain the genes. The usual number of chromosomes in humans is forty-six with one set of twenty-three inherited from the mother and the other from the father.

Men carry a Y chromosome which is inherited from their father and their father’s father before them through the generations very much like an inherited surname.

There are many different types of Y chromosomes and just like a surname, they can be typical of a region or associated with particular historical groups which have different origins, e.g. Norse, Pictish, Anglo-Saxon, Irish, etc. Reading the Y chromosome code of an individual can tell us about the ancestry of that person or the region from which they come.

Mitochondrial DNA is a section of DNA carried by both men and women but inherited from their mother and their mother’s mother before them. Just like a Y chromosome there are many different types of mtDNA and they can be typical of a region or historical group. Again, just like the Y chromosome they can tell us about the ancestry of a person or a particular population. The difference is that the mtDNA explores the historical maternal line as opposed to the paternal line with the Y chromosome.

Whole genome approaches look at the nature of the code across all of our DNA. This approach is very good for providing a broad picture of ancestry, for example your DNA looks 100% “Irish” or you look 50% Irish, 25% French, 25% Italian. It does not however, provide the resolution that the Y chromosome or mtDNA does for particular lineages.

The Project team will apply all or a combination of the above approaches to explore the broad history of Ireland, particular regions within Ireland and/or ethnic groups resident in Ireland.

The Collection of the Data

The Irish DNA Atlas will create a DNA collection that allows genetic analysis of population structure within Ireland, and ethnic groups across the island. Analysis of such a collection will reveal ancient demographic movements and inform on the ancestry of specific regions and ethnic groups within Ireland. The Irish DNA Atlas will also create a DNA collection to act as controls in population based studies of health in Ireland. The Project aims to recruit individuals representing each of the 32 counties of Ireland. Participants may opt to take part in either the historical or medical components of the study or both. The Project will provide each participant with a saliva collection kit (saliva is a rich source of DNA!) and a brief questionnaire to gather some general health information and a pedigree chart (Birth Brief) to include all eight great grandparents. Each sample will be processed, stored and used by the RCSI Biobanking Facility in Dublin. During processing the Project will assign an unique code to each individual sample. However, the names and contact details of each participant will be kept on record by the RCSI as Dr. Cavalleri or members of his team may wish to contact participants by telephone, letter or e-mail in the future. The Project will use each DNA sample to determine the genetic profile of each participant. In order to generate this profile the RCSI may on occasion be required to send your DNA to another institute, possibly overseas, with which the RCSI has a formal collaborative agreement. Should the RCSI wish to send any DNA sample to another laboratory or collaborating centre it will be sent in coded format under the terms of a formal agreement and under equivalent data protection measures. Each participant will receive an IDA Project Pack which includes the DNA sample kit, Birth Brief, a short health questionnaire for completion and a participation consent form which must be signed by each participant.

Eight Great Grandparents & NASC

Participants will be asked to supply a completed Birth Brief showing the genealogical details of their immediate ancestors back to the eight great grandparents and to provide a DNA sample which is a simple saliva swab for analysis. All of the eight great grandparents should come from the same general area so that their DNA represents that particular region of Ireland. The Project team had originally considered using the county boundaries as the research areas, however, this proved to be problematic from many perspectives including historical, social and cultural. Therefore, having looked at all existing or former administrative boundaries it was considered that a new formula—NASC—was required. NASC stands for ‘natural areas of social cohesion’ and takes account for communities that straddle county boundaries. For example communities living in south Co. Wicklow and north Co. Wexford are like to have more in common with each other than with the rest of their respective counties. This situation exists throughout Ireland and, in many cases, such close connections predate the formation of the existing county boundaries. As to what constitutes a NASC depends entirely on the information supplied by the participant. Did the eight great grandparents live, for example, within a 30 kilometre radius of the participant’s ancestral homestead? This would easily represent a NASC. The Project team will monitor the entries received to ensure that each of the 32 counties of Ireland is covered by the research and therefore, the Project may concentrate on certain areas over time.

A collaboration between the Genealogical Society of Ireland and the Royal College of Surgeons in Ireland
The Project’s Two Strands

The project’s aims are both historical and medical and it has essentially two strands - one dealing with the collection of the genealogical data and the other dealing with the scientific analysis and interpretation of the DNA samples. Participants may opt to take part in either the historical or medical components of the study or both and supply information accordingly.

SCIENTIFIC ANALYSIS

The scientific aspects of the Irish DNA Atlas project will be directed by Dr. Gianpiero L. Cavalleri of the Royal College of Surgeon in Ireland. Dr. Cavalleri is a Senior Scientist, of Italian parentage but born and raised in Ireland, is a population geneticist who trained with Prof. Dan Bradley at Trinity College, Dublin before going on to work at Stanford University with Prof. Luca Cavalli-Sforza and Dr Peter Underhill. Dr. Cavalleri completed a PhD at University College London under Prof. David Goldstein studying the genetics of epilepsy predisposition and treatment. He is currently researching the genetics and pharmacogenetics of epilepsy at the Royal College of Surgeons, Dublin, Ireland. Dr. Cavalleri was the scientific consultant on the very popular television documentaries ‘The Blood of the Irish’ and ‘The Blood of the Travellers’.

GENEALOGICAL DATA

The promotion of the project and the collection of the genealogical data will be coordinated by Séamus O’Reilly, FGSi, Director of Archival Services. Séamus was appointed to the position of Archivist by the Board of the Society in 2005. Since taking over the position he has concentrated on the computerization and sorting of the collections including the sizeable manuscript collection. Séamus planned and directed the successful relocation of the archival collections to their current location at the Carlisle Pier in Dún Laoghaire in 2010. He instituted procedures for the secure storage and the speedy retrieval of items for research. As the Archive continues to grow, Séamus has embarked on an ambitious plan to scan and upload to the website all the articles published in the Society’s Journals and to have an on-line catalogue of all the collections. The collection of the genealogical data and the collation of the information will be undertaken by Séamus in the Society’s Archive & Research Centre—An Daoncharlann—at the Carlisle Pier in Dún Laoghaire, Co. Dublin. Séamus will examine the genealogical data submitted to ensure that it meets the criteria for the scientific study to be undertaken by Dr. Cavalleri. Séamus can be contacted on Irish.DNA@familyhistory.ie

Health and Lifestyle Issues

Normally the participant will not receive any results from this study unless the Project team identifies something it thinks is of importance to the participant’s own health. Participants should fully appreciate that, as with any other health investigation, this research has the potential to identify a genetic change that could be important in your lifestyle decisions. Should the Project team identify such a genetic signature that it deems to be of extreme clinical relevance and of import to lifestyle decisions, the Project team at the RCSI will contact the participant concerned offering the result together with genetic counselling. Genetic counselling provides information and support for people who are concerned about a medical condition that may have a genetic basis. Although participating in this study will not be of direct benefit to each participant, it is important scientifically. Information arising from this study has the potential to improve our understanding of common diseases such as cancer and cardiovascular disease and, of course, provide valuable new information of the origin and movement of populations on the island of Ireland. Information from a participant’s sample could be used for research involving the commercial sector including pharmaceutical companies. Through the collection and scientific analysis of this type of data it may be possible to predict diseases and help improve the nature of future treatments, including drug design or lifestyle decisions. It should be fully understood that any involvement of the commercial sector will be through collaboration or partnership with the RCSI involving formal assurances that equivalent data protection measures will be upheld by the commercial partner. The Project has been approved by the Ethics Committee of the Royal College of Surgeons in Ireland.

Collaboration and Partnerships

As with all scientific or academic research in Ireland, the development of collaborative ventures is essential. The Project team at the RCSI is keenly aware that it needs to be are the forefront of genetic research in Ireland and indeed, in the world. As an institution, the RCSI is totally committed to performing high levels of research activity and to commercialising intellectual property arising from its research through the development of collaborative links with industry, educational and research institutions at home and abroad. The commercialising of the intellectual property arising from research at the RCSI brings the benefits of that research to the public by way of medical devices, procedures, treatments or drugs. Ireland needs to be proactive in this regard to attract the best graduates into such research and indeed, to secure the research funding required for the development of new medicines or technologies.

This Irish DNA Atlas Project will contribute significantly to our knowledge of the genetic composition of the Irish population and provide invaluable research information on certain health issues. However, it is essential if the true potential of the research is to be achieved that collaborative endeavours and research partnerships are secured, especially with the scientific and medical industries. For further information please contact Dr. Gianpiero Cavalleri at the RCSI.
**PROJECT FAQs**

**Does participation in the Irish DNA Atlas Project cost anything?**

**Answer:** There is absolutely no cost other than the return postage of the Birth Brief and the sample.

**Does each participant receive an analysis of their own DNA?**

**Answer:** No. Participation is on a pro bono basis providing information for the analysis of the samples by the RCSI. The Project team can direct participants to institutions providing DNA services.

**Will the Irish DNA Atlas be published?**

**Answer:** The Project team at the RCSI will publish the findings of the research in the appropriate medical and scientific journals. Individual participants will not be identifiable within such publications.

**Is the DNA information supplied by the participant confidential?**

**Answer:** The DNA samples receive a specific code and all research conducted by the Project team at the RCSI will be based on the code and not the name of the participant.

**Is participation confined to existing members of the Genealogical Society of Ireland?**

**Answer:** No. This is an All-Ireland project open to anybody fitting the criteria, terms and conditions of participation.

**Is participation confined to people resident on the island of Ireland?**

**Answer:** No. If all of your eight great grandparents come from Ireland and meet the criteria, terms and conditions of participation that’s fine.

For further information please contact: Irish.DNA@familyhistory.ie

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**Essential Information for ALL Participants**

Participation in the Irish DNA Atlas Project is strictly on a pro bono basis for all participants. All genetic data will be studied in a specially designed coded format by which individual participants cannot be identified by name. This special code linking each participant to their DNA sample will be held securely by the main investigator Dr. Cavalleri. This information will not be used for any other purpose other than for this particular Project. The results of the study, including data generated, may be shared with other scientists and published at a later date, but the names of the participants will not appear in such publications. As with any study involving clinical or genetic information, the risk of data loss or misplacement is always present. The Project will take utmost precautions to avoid such complications. Participants have the absolute right to withdraw from the Project at any stage, for whatever reason. The sample will then be destroyed and the genealogical information provided will be returned to the former participant. Each participant opting to take part in the medical component of the research, will be presented with a confidential questionnaire on general health issues, containing for example, questions on whether you have ever been diagnosed with high blood pressure; experienced a heart attack or a stroke; suffer from diabetes (type 1 or type 2); ever been diagnosed with epilepsy, autism, bipolar disorder or schizophrenia; or ever been diagnosed with cancer. This information is strictly confidential and is provided in a sealed envelope along with your DNA saliva sample. This information will only be accessible to Dr. Cavalleri and his team at the RCSI. All participants will be required to sign a consent form, a copy of which will be returned for their own personal records. Participants are strongly advised to keep a copy of this issue of the Irish DNA Atlas Project Newsletter for future reference. It is the responsibility of each participant to ensure that he/she understands all the information presented by the Project team before agreeing to participate in this study. The Project team will endeavour to answer any queries that intending participants may have on the Project.

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**IRISH DNA ATLAS PROJECT NEWSLETTER**

This Newsletter is an occasional publication of the Irish DNA Atlas Project and is intended to provide information on the Project for participants, members of the Genealogical Society of Ireland and the general public. Further issues of this Newsletter will be published, as and when required, to provide an update on the Project or to recruit participants from specific areas of the island of Ireland. It is intended that the RCSI will determine the duration and scope of the Project depending on the levels and the spread of the participation achieved. Interim reports on the progress or findings of the Project may be published by the RCSI or the Project team in scientific, health or other academic journals. It is not intended to publish such reports or analyses of the research data in this Newsletter and therefore, all enquiries in respect of such should be forwarded directly to the Project’s Scientific Director, Dr. Gianpiero Cavalleri, by e-mail at gcavalleri@rcsi.ie or by mail to: Dr. Gianpiero Cavalleri, Molecular and Cellular Therapeutics, The Royal College of Surgeons, 123, St. Stephen’s Green, Dublin 2, Ireland.