Oxford Cambridge and RSA

Thursday 13 June 2019 – Morning

A Level Biology B (Advancing Biology)

H422/02 Scientific literacy in biology

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Learning from Iceland's model for genetic research

A few years ago, a group of scientists at the International Myeloma Foundation (IMF) identified a problem. Some people with monoclonal gammopathy of undetermined significance (MGUS), which is a benign condition, would later develop the malignant disease multiple myeloma. The risk was estimated to be as high as 40% over a lifetime.

Most doctors do not routinely screen for MGUS. As a result, most cases are never diagnosed unless patients develop multiple myeloma.

The chairman of IMF, Ian Durie, thought it might be possible to cure or prevent multiple myeloma. To do this, it would be necessary to intervene at an early stage and identify those patients with MGUS who later develop multiple myeloma. As only about 1% of people with MGUS go on to develop multiple myeloma each year, the IMF needed to have a clear way to identify the patients at risk.

In 2015, after discussions with other scientists working on myeloma, Durie proposed a potential solution and decided Iceland was a good place to test their ideas.

Iceland has a small population – only 320,000 people in 2013. About 20 new cases of multiple myeloma are reported in the country each year. It also has the most detailed genetic and medical databases of any country in the world. Iceland therefore offers a unique experimental setting. The IMF is able to detect and document all the people with MGUS. By studying MGUS patients, the scientists can work out which ones go on to develop myeloma. They will then have the opportunity to intervene, early on, and provide a cure.

The IMF launched a nationwide MGUS screening study in November 2016. The team of scientists has been encouraging all Icelanders older than 40 years of age to take part. More than 75,000 people had registered by April 2017, and the researchers had collected nearly 20,000 blood samples. This made the project the largest myeloma study ever conducted.

Celtic and Norwegian explorers settled in Iceland during the 9th century. The country's population has remained small ever since, owing to geographic isolation, its harsh environment, and several dramatic events. Two waves of plague hit Iceland during the 15th century, followed by a spate of smallpox in the early 1700s and a large volcanic eruption in 1783. As a result, the island has one of the most genetically homogeneous populations in Europe. This is important to the IMF's study because it is easier to pinpoint an unusual, disease-causing genetic variant in populations with little genetic diversity.

Iceland has a long tradition of recording family trees. In the 12th century, Icelandic priest Ari the Wise traced the country's history in a book called Íslendingabók – the book of Icelanders. Since then, church records and censuses have kept track of births, deaths and marriages, building a detailed picture of Icelandic ancestry.

This tradition was updated and modernised in 1996. Icelandic geneticist Kári Stefánsson founded the company deCODE Genetics and collected all available genealogical information in one online database, also called Íslendingabók.

The research power of this genealogical data has been boosted by integrating it with two more databases. One of them details genetic information collected from biological samples of more than 100,000 citizens; the other stores medical records of people participating in related research projects. Every single cancer that is diagnosed in Iceland is centrally registered, as are all medical procedures, clinical diagnoses and prescriptions.

Researchers at the IMF store thousands of blood samples from Icelanders. They test the samples with automated blood analysers which rely on flow cytometry. These expensive tests allow the identification of genetic variants that might be linked to the risk of developing multiple myeloma.

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The results from these Icelandic studies will assist research into human genomic variation around the world. Although it might be difficult to replicate the IMF studies elsewhere, the information from Iceland can be extrapolated and applied globally.

Source: http://www.the-scientist.com/?articles.view/articleNo/49439/title/Learning-from-Iceland-s-Model-for-Genetic-Research/

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