Project Plan

Viking Genes Shetland – Shetland Community Screening Project

Introduction

Over the past 20 years, the Viking Genes Study has provided remarkable insights into the ancestry, health and unique genetic make-up of Scotland's island populations. More than 250 academic papers have been published.

More recently, Viking Genes has focussed on improving the health and well-being of Scottish islanders, having discovered that they face a disproportionately high prevalence of genetic health risks. Many of these risks can be managed by early preventative medical intervention and over 150 individuals have so far benefited from being informed of their individual genetic health risk via Viking Genes.

The return of actionable health findings to Viking Genes participants from Shetland, such as the risk of cancer from BRCA2, is a leading example of the impact of genomic medicine. It is contributing to future health policy across the UK, as well as academic research and medical education.

The Shetland Community Screening Project plans to extend the delivery of health benefits in Shetland. This document sets out the proposal for a Shetland-specific project to further the Viking Genes study. As well as health and economic benefits from a community-wide screen of a higher risk population, the project is the first of its kind in Scotland that can serve as a practical example to healthcare providers nationally.

Shetland Community Screening Project - Aims

- To save lives in Shetland through identifying individuals at risk through free community-wide genetic screening and then providing a pathway for treatment.
- To improve the health and wellbeing of the Shetland community and in doing so reduce the terrible impact of inherited genetic health conditions on families.
- To remove disadvantages and ensure all those with higher genetic health risks have the information and evidence they need to be able to make decisions on accessing preventative healthcare pathways.
- To raise awareness with healthcare professionals caring for Shetlanders of the unusually high prevalence of rare disease-causing variants in the population.
- To educate the Shetland public on the risks of genetic variants affecting their health (and that of their children).
- To demonstrate the long-term health and cost advantages of community-wide genetic screening programmes as part of preventative medicine.
- To further the research evidence base supporting access to community-wide genetic screening through published peer-reviewed scientific papers.
- To influence public health policies in Scotland and the UK towards implementing preventative genetic health programmes.
- To advance genomic medicine as a preventative measure beneficial to the future direction of healthcare – to act as an exemplar for others to follow.

Background

Viking Genes www.viking.ed.ac.uk is a project led by Professor Jim Flett Wilson of the University of Edinburgh, that has studied the DNA of over 10,000 people in the Scottish islands and their descendants living across the globe. Around one-third of the early cohorts were Shetlanders. The

project has revealed a higher risk of a number of life-threatening diseases stemming from genetic variants in island communities compared to the general population. This is most obvious in Shetland.

Many of the genetic health risks facing the Shetland community can be managed through early medical interventions which stop disease progression and keep the individuals healthier. This is cost-effective for the NHS, as early intervention and prevention is a lower-cost and less time-consuming option than disease treatment, but only actionable if the patient is known to face an elevated genetic risk.

Unlike most other genetic studies, Viking Genes has developed the return of results from its studies. In recent years, Professor Wilson has been able to return such actionable health findings to the affected screened volunteers. The vast majority of volunteers have opted to receive their results.

High Risk Genetic variants in Shetland

The Viking Genes study has revealed a number of gene variants increasing the risk of specific diseases, many times higher than elsewhere in Scotland. Examples of the Shetland actionable variants that have been returned with treatment pathways include:

- BRCA2 variant linked to breast, ovarian and prostate cancer
- KCNH2 variant causing a heart condition which can lead to fatal cardiac arrest
- TTN variant causing a form of heart disease, which can lead to heart failure
- ATP7B variants causing Wilson disease, a disease of copper metabolism

All these variants are actionable, in the sense that individuals can take action to address their risks through preventative measures, e.g., medicine, more frequent screening or preventative surgery. The NHS recognises these health risks and has established care pathways for each.

Scotland's island communities also have a high prevalence of certain genetic "carrier" variants. Carrier variants only pose a health risk when a baby inherits two copies of them, one from each parent: this can have devastating consequences for the child, with a 25% chance of developing the inherited condition.

Carrier variants prevalent in Shetland include:

- Batten disease – a degenerative disease, usually fatal in childhood
- Myotonia – a muscle disease that causes great physical difficulty
- Cystic fibrosis – a lung disease that shortens lifespan

Currently, many of these diseases are incurable but for some there are risk-mitigating treatments available. For the future, Viking Genes aims to be able to move on to this area and within two years hopes to start returning information to carriers of these variants, particularly relevant to couples wishing to start a family.

Case histories

In Shetland, around 2.5% of the screened volunteers (one in 40) have been found to have a gene variant creating a risk of treatable disease. The Volunteer Stories section of Viking Genes website https://viking.ed.ac.uk/about-us/volunteer-stories provides examples of cases where the volunteers have been able to beneficially use their results to obtain preventative treatments. In addition to the screened volunteers, immediate family members of those with variants have been able to access NHS screening, multiplying the number of people picked up and able to benefit from preventative healthcare.

For the vast majority of people, they will have peace of mind that they are not affected.

The Shetland Community Screening Project

In 2024, Viking Genes commenced a three-year extension aimed at implementing a community-wide genetic health screen in Shetland. This extension provides the foundation for the proposed new screening project to commence. The new screen will be open to 5,000 residents of Shetland (irrespective of whether or not they have Shetland ancestors), aiming to capture both those with a family history of disease and others (50% of previously found carriers did not have a family history).

Based on the proportion of previously screened volunteers with actionable findings and taking into account some will already know; we predict up to 100 individuals being able to access important medical care that they might otherwise not have been aware of. In addition, over the lifetime of the new screening project, the awareness of carrier conditions has the potential to positively impact the health of future generations.

The Viking Genes Fund

To support this work, the Viking Genes Fund was launched in 2024. The fund is held within the University of Edinburgh Development Trust, a charity (SC004307) where the Objectives of the Trust are to accept, hold and apply funds for furthering the aims of the University. The Trust operates as a vehicle for receipt and disbursement of philanthropic funds. The Viking Genes Fund is a restricted fund where the donations can only be used in accordance with the aims of the Viking Genes study. The University's Development and Alumni department, on behalf of the Trust, is legally obliged when accepting restricted funds for a specific purpose, to spend the funds according to the donor's wishes and expectations. If the donor's wishes cannot be met, the funds may need to be returned to the donor.

Local engagement with Viking Genes Fund

Following a Viking Genes public presentation in Lerwick in September 2024, a committee of local volunteers, with the endorsement of Professor Wilson, is supporting the Shetland Community Screening Project by building awareness and helping to raise funds. The committee has a Constitution and an Agreement with the University of Edinburgh.

While respecting the opinion of those that do not want to know what lies before them, the committee recognises the desire for the option to be screened and make healthcare choices.

In addition, a number of other local volunteers have independently arranged their own fundraisers and donations to support Viking Genes Fund, realising over £45k to date (at May 2025). These funds are enabling the current extension project, employing a PhD student.

Shetland Community Screening Project Implementation

Using the experience of running Viking Genes and its current datasets, work is underway to create a bespoke screening panel covering up to 50 important genetic variants relevant to Shetland. These will be the variants the new screening project will check for. The cost and complexity of screening is reduced by focussing only on known variants and conditions.

Once sufficient funds are pledged to enable the Project to start, it is planned to recruit a project delivery team - a Project Manager, Database Manager, Research Assistant and part-time Communications Manager. It is envisaged that they will take up to one year to prepare the groundwork to start screening, which will then be a potential three-year programme to conduct the screening and returning of results.

Preparatory work will include gaining the ethical permissions required, ordering the spit and testing kits, contracting the laboratory to perform the tests, setting up the secure database at the University of Edinburgh for online consent of screening volunteers and tracking of saliva samples.

Recruitment of 5,000 adult Shetlanders (over 16 years of age) will commence and participants only need to provide a one-time saliva sample by post. It is hoped to also provide some community outreach days to maximise the reach. Newly proposed is an innovative digital delivery platform to make it easy for participants to access their results via a password-protected online dashboard, hopefully starting within three months of returning their saliva sample. The aim is to integrate into NHS systems and streamline the pathway provision to follow up for any treatment.

The 3,000+ Shetlanders previously screened in VIKING I and VIKING II campaigns do not need to be screened again, their data is securely held at the University of Edinburgh.

Cost

The overall cost for the Shetland Community Screening Project through to 2029 is £1.8M. The University of Edinburgh contribution is £800k (45%), leaving £1M to be raised.

A summary of the £1M expenditure:

- Spit tests and laboratory – 25%
- Salaries for 3.5 FTE project delivery team – 65%
- Misc expenses and NHS costs – 10%

Note: The University funds Professor Wilson and other establishment costs.
Screening 5,000 Shetlanders at a cost of £1M equates to £200 per screen.

Legacy

Undertaking the Shetland project will save lives and improve the health of the Shetland community. It will also serve as a pathfinder for preventative healthcare in Scotland and deliver a practical and ethical exemplar for other communities to emulate.

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