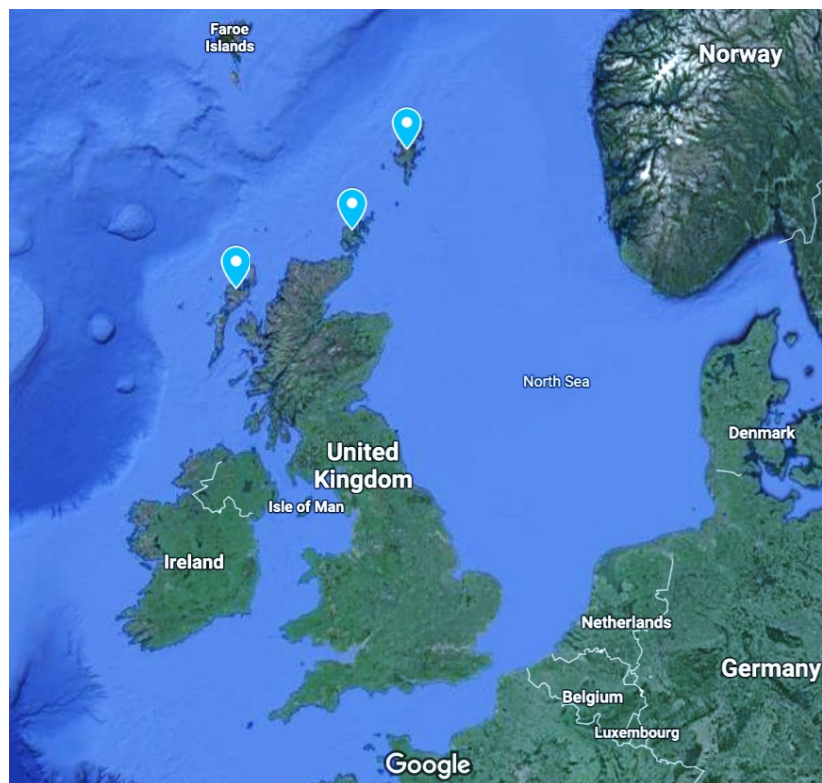


GENOMIC MEDICINE FOR THE SCOTTISH ISLANDS

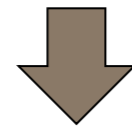
Benjamin S. Fletcher (b.s.fletcher@sms.ed.ac.uk) and Prof James F. Wilson (jim.wilson@ed.ac.uk)

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VIKING GENES



Geographic isolation of Orkney, Shetland, and the Hebrides



Some genetic variants increased in frequency, some decreased

Actionable genetic findings – harmful rare genetic variants for which treatment or disease prevention exists

DATA

~10K worldwide volunteers with two or more grandparents from Orkney, Shetland, or the Hebrides

ORCADES



2,090

VIKING I



2,108

VIKING II & III



5,244

DNA & exome sequence from blood/saliva 2005-2023



ACTIONABLE FINDINGS

2.4% of participants have at least one actionable finding

Overall rate similar to mainland Scotland however:

In Scottish islands, **some rare variants are much more common, while others have disappeared**



Breast and ovarian cancer *BRCA1, BRCA2*
Haemochromatosis *HFE*
Hypercholesterolaemia *LDLR*
Long QT syndrome *KCNH2*
Cardiomyopathy *TTN*

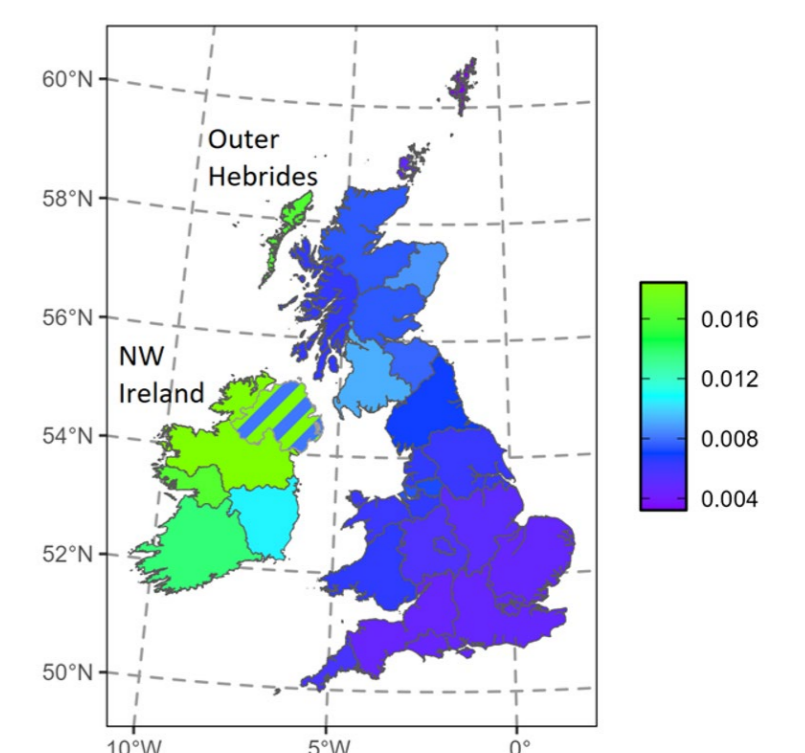
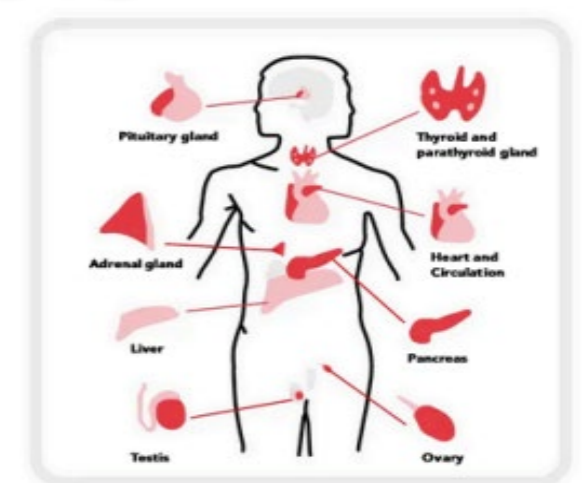


HAEMOCHROMATOSIS

Iron overload disorder caused by variants in the *HFE* gene

Gradual, widespread organ damage when left untreated – but treatment is straightforward *once diagnosed*

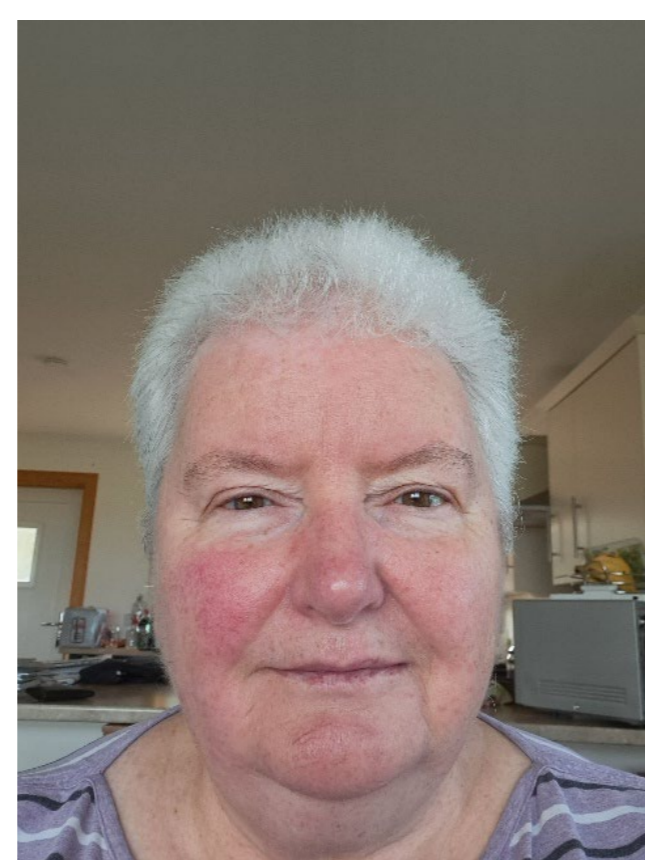
Outer Hebrides has highest frequency in the UK



MAIRI'S STORY

Mairi is a VIKING III volunteer from the Hebrides.

After receiving a letter from Viking Genes, Mairi underwent blood tests with her GP to confirm she had Hereditary Haemochromatosis.



"I am grateful to Viking Genes for highlighting this risk, allowing me to confirm the existence of the condition, and to have appropriate treatment at a point where I was still asymptomatic, as it seems likely that I would have experienced some effects of the condition in due course."

More: <https://viking.ed.ac.uk/about-us/volunteer-stories>

TAILORING GENOMIC MEDICINE

Isolated populations have unique risk profiles for heritable disease

Equitable preventative medicine thus requires a tailored approach

Because of increased risk, NHS England offers *BRCA* gene testing to anyone with Jewish grandparents

Our goal: identify variants relevant to **bespoke screening panels** for Orkney, Shetland, the Hebrides

Shetland Community Screening Project

- raise £1 million to screen 5000 Shetlanders for ~50 disease-causing variants

WHAT NEXT?

- Publish and communicate findings to volunteers & scientific community
- Analyse recessive variants for reproductive carrier screening e.g. cystic fibrosis
- Assess utility of polygenic risk scores in Orkney, Shetland, the Hebrides

Learn more, get involved, and donate at: <https://viking.ed.ac.uk/>



MRC Human Genetics Unit



THE UNIVERSITY of EDINBURGH

