

# Data linkage to examine equity of genetic testing in Wales

## Participant Information Sheet

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## Summary

This notification document is intended to inform the public about our study investigating factors affecting the equity of genetic testing in Wales.

It explains what we are planning to do, how we are going to do it, and how those who do not wish for their genetic test result to be included in the study can opt-out of this research project.

- Genetic testing is used to find changes in a person's genetic code that may cause illness or disease. As well as being used to find the correct diagnosis it is also used to find out whether a person is at risk of developing a genetic condition or whether they could pass a genetic condition on to their children.
- In Wales, genetic testing is available within the NHS for a number of diseases. The tests can be requested by healthcare professionals and are provided by the All Wales Medical Genomics Service (AWMGS).
- AWMGS has been performing genetic testing for over 30 years and during that time has built a large archive of genetic test results from the Welsh population. This archive can provide valuable data for health research that could benefit healthcare services for people in Wales.
- The Secure Anonymised Information Linkage (SAIL) Databank is a research databank containing anonymised health and administrative records for people in Wales and is used for health research.
- In our study we will select test results for a range of genetic conditions from the AWMGS archive. As the results contain Personal Identifiable Data we will first anonymise them, before linking them with other anonymised data within the SAIL Databank.
- We will study the linked records to find out if access to genetic testing is affected by socio-economic factors (e.g. income, employment, education, housing), Welsh language proficiency, and geographic factors such as living in more rural areas or living further away from a specialised hospital.
- The tests results will also be used to estimate how often particular genetic changes (mutations) happen in the Welsh population.
- We hope this study will help us assess whether over the last 30 years genetic testing in Wales has been truly accessible and inclusive.
- The study will also help us plan important future healthcare developments, including the Genomics Delivery Plan for Wales 2022-2025.

- We are not obtaining research consent from individuals whose genetic test data will be used in this study. This would be too difficult with the resources that we have. However, no new information will be collected, and all data will be securely anonymised (meaning we cannot identify individual participants).
- If you have had a genetic test in Wales for the following conditions, then it is very likely your test result will be included in our research project.
  - Huntingdon's Disease, Lynch Syndrome, Haemochromatosis, Fragile X Syndrome, Neurofibromatosis type 1, Hereditary breast cancer (BRCA1 and BRCA2), Epilepsy
- If you do not want your data to be used in this study you can submit an **opt-out request** to [genetic\\_testing\\_study@swansea.ac.uk](mailto:genetic_testing_study@swansea.ac.uk), write to Dr Owen Pickrell, Room 329, ILS 1, Swansea University Medical School, Singleton Park, Swansea, SA2 8PP or phone us on 01792 295134.

# Explainer

## What is our study?

We have received funding from [Health and Care Research Wales](#) to carry out research that will look at the equity of genetic testing for common conditions in Wales using linkage of health and genetic data.

## Why are we doing a study on this topic?

Genetics and genomics in Wales is being developed through the [Genomics Delivery Plan for Wales 2022-2025](#). As part of this it is important to understand where there could be limitations in access to existing testing so that these can be improved for the future. Also we want to develop better tools and methods for similar studies using anonymised routine testing data in the future. These studies will allow public health researchers to answer important health-related questions.

## What are the potential benefits?

We hope to find out important data about the historical and current barriers to genetic testing for common genetic conditions in Wales. This will be used to generate recommendations to improve genetic testing services in Wales. *We also hope that this study will provide important information that will help similar studies in the future answer important question about health in Wales.*

## What data will we use?

Figure 1 summarises the flow of data in our project. The [SAIL Databank](#) is a Secure eResearch Platform (SeRP) that holds routinely collected health and administrative data, collected by NHS

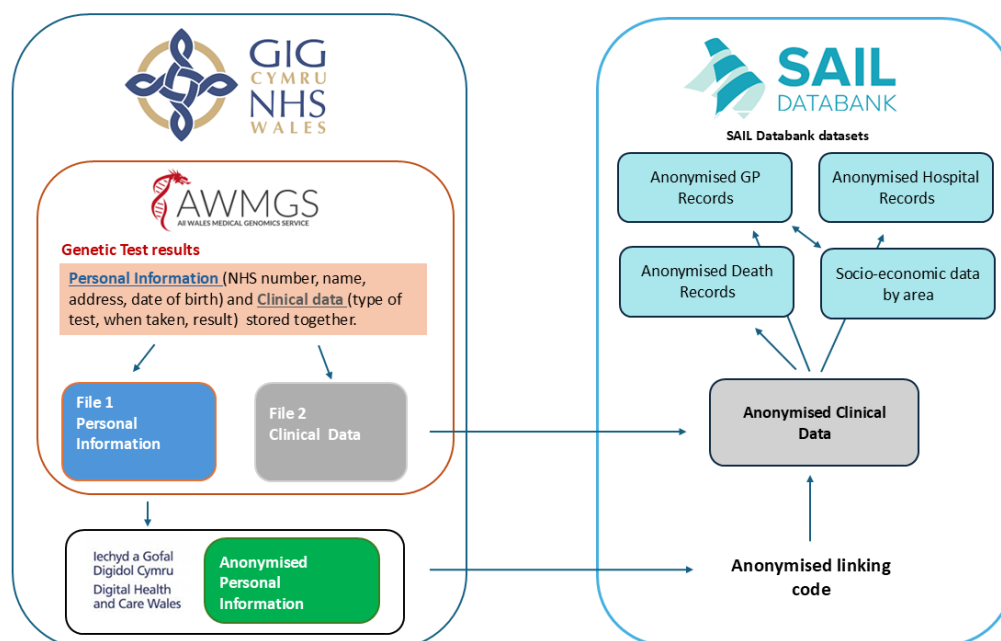


Figure 1. Summary of dataflow. Raw test records within AWMGS system will be split into Identifiable Personal Information and anonymised Clinical Data. Personal Information will be transferred to Digital Health and Care Wales for anonymisation and Clinical data will be uploadd directly to SAIL. Within SAIL, the anonymised datasets will be reunited and linked to other public datasets, such as GP and hospital records.

Wales and the Welsh Government, about the Welsh population (records of around 5 million people) in an anonymous format.

We will import anonymised test results for individuals tested for genetic conditions from the [All Wales Medical Genomics Service](#) into the SAIL Databank and anonymously link these genetic test results with other datasets, including GP and Hospital records, and data from the Census. This will allow us to see if there is any variation in genetic testing with social, geographic and health factors.

#### *Why are we not seeking explicit research consent?*

In our study we will be using only anonymised, routinely collected data, however, identifiable information will be accessed prior to the data linkage process. When genetic tests were performed, patients were not asked for their consent for their data to be used in research. We do not have the resources to trace every patient from the last 30 years, with potentially incomplete or out of date contact details, to seek explicit research consent. We have written to the [Confidentiality Advisory Group](#) (25/CAG/0002) to request section 251 support to waive the legal need for explicit consent given the safeguards in place. The application was reviewed by the Confidentiality Advisory Group (CAG). CAG is an independent group of lay people and professionals which provides expert advice on the use of confidential patient information without consent. CAG recommended that our application should be supported and the Decision Maker within the Health Research Authority approved this.

#### *What are the risks and safeguards?*

Using data about real people always carries the risk that personal information (such as name, postcode, etc.) may be exposed. All data held in the SAIL Databank is securely anonymised so that even if information is exposed, the individual behind it cannot be easily identified. As part of this project we will move genetic data from the All Wales Medical Genomics Service to the SAIL Databank and any personal information will be replaced with anonymisation tags before it leaves NHS systems which are highly secure. As such, no personal information of study participants will be stored outside of NHS systems at any point – only anonymised information will enter the SAIL Databank.

#### *What have we done so far?*

The study along with the supporting documentation has been reviewed by CAG under section 251 of the NHS Act 2006, by the [Research Ethics Committee](#) (REC), and the SAIL Databank [Information Governance Review Panel](#) (IGRP). We have also asked public health stakeholders such as Genetic Alliance UK for their views and we will be undertaking public talks to inform the public and gather their views.

#### *What do you need to know?*

If you are happy with your anonymised data being included in our study, you do not need to do anything. If you have questions or concerns, you can get in touch with the research team using the contact details at the end of this document.

If you do not wish for your anonymised data to be used as part of this study you can opt out of the project by contacting the research team (as above) within two months from the date of this

notification. Please be aware that once the test results are anonymised and uploaded to the SAIL Databank your records cannot be identified as such by the research team or by SAIL.

For the Welsh Longitudinal General Practice dataset, please contact your registered GP surgery to request an opt-out.

## General Data Protection Regulation (GDPR)

Under GDPR, the legal basis for processing personal data for research purposes is that it is a task in the public interest. For special categories of data, processing is permitted when necessary for archiving in the public interest and for scientific research purposes. For more details, please [follow this link](#).

To find out your rights (including contact details for any queries you have) under the GDPR and UK Data Protection Laws please [follow this link](#).

Rights in relation to automated decision making and profiling are not relevant in this study (i.e., we do not use automated decision making).

## What will happen to the results of the research study?

When the study is finished, we will want to share our findings widely. Findings will be presented at relevant conferences and meetings, and written up for publication in academic and scientific journals. Lay summaries for the public will be made accessible through social media, our blog, and public talks. We will not identify any individual in any of our results outputs.

## Data Controllers

For the purposes of all data pertaining to genetic test results, Cardiff and Vale University Health Board is the Data Controller and the All Wales Medical Genomics Service is the Data Processor. The following NHS Sites are involved:

The All Wales Medical Genomics Service, Cardiff and Vale University Health Board

## End of study | Archiving

Any files containing essential study documents will be archived at an approved external storage facility for a minimum of 15 years.

## Complaints

If you wish to raise a complaint on how we have handled your personal data, you can contact Cardiff University's Data Protection Officer, [inforequest@cardiff.ac.uk](mailto:inforequest@cardiff.ac.uk), who will investigate the matter.)

If you are not satisfied with our response or believe we are processing your personal data in a way that is not lawful you can complain to the Information Commissioner's Office (ICO) by visiting <https://ico.org.uk/make-a-complaint/> or by calling their helpline on 0303 1231113.