

## WHG/UKSFN Project: Researcher Profiles

*The following eight profiles contain information about researchers at the Wellcome Centre for Human Genetics (WHG), who have signed up to take part in this project. These profiles include names, proposed topics, any relevant format preferences or audience preferences, and whether they have any previous public engagement experience. In advance of completing your Expression of Interest (EOI) form, please review these profiles as you will be asked to state your preferences for the researchers with whom you would most like to be matched.*

*These proposed topics are intended to give you an idea of the research focuses of the researchers, enable you to think about your preferences, and to act as a starting point for collaboration between festivals and the researchers going forward, where you will work together to develop the event content.*

### Researcher 1: Gabrielle Samuel

#### Proposed Topic

In the current climate emergency, in which society is paying increasing attention to carbon emissions and other environmental impacts, it is vital that all innovations – whether or not they meet sustainable development goals—are developed using a tri-pillar approach that considers environmental, social, and economic aspects. Little attention has focused on the adverse environmental and health impacts of genomics. This is problematic because there appears to be an internal contradiction between, on the one hand, the goal of improving health conditions through the use of genomics, and on the other hand, its environmental and health risks. In fact, those using genomics technologies (or at least those funding and governing them) have a special interest in addressing its adverse environmental and health impacts, not only as a matter of international priority, but also as a commitment to health. Genomics is associated with a range of adverse environmental and health risks. During wet-lab research, emissions are associated with running laboratories and storing samples at ultra-low temperatures, as well as waste generated from solvents and plastics. Impacts associated with dry lab research are less explored. Nevertheless, increasingly large volumes of genomic data are stored and analysed. During this process, heavy carbon dioxide emissions result from the energy required to generate and process large amounts of data, and running algorithms in genome-wide association studies can be particularly energy hungry. Genomics practices also contribute to the global demand for mineral consumption associated with developing digital infrastructures, as well as the massive amount of e-waste that contains hazardous materials such as lead, cadmium, mercury and nickel. Both practices are associated with a wide range of health and well-being harms. I hope that my event can highlight these issues.

#### Preferred Event Format

No preference

#### Preferred Audiences

No preference

#### Public Engagement Experience

Very little. I have given a couple of presentations to members of the public and once to school kids (yr 11)

## Researcher 2: Megan Payne

### Proposed Topic

Finding new drugs from nature - my DPhil project is based around using proteins found in the saliva of ticks to target cardiovascular diseases. I'm also happy to be involved in another topic or event.

### Preferred Event Format

No preference

### Preferred Audiences

No preference

### Public Engagement Experience

N/a

## Researcher 3: Juan Shen

### Proposed Topic

How we look in situ protein structures using electron cryotomography (CryoET).

### Preferred Event Format

Interactive event (e.g. drop-in activities)

### Preferred Audiences

Young people and rural audiences

### Public Engagement Experience

N/a

## Researcher 4: Serena Vales

### Proposed Topic

My DPhil is about discovering peptides derived salivary proteins of ticks to overcome the chemokine network in inflammation, specifically in cardiovascular diseases.  
Perhaps, how we can utilise nature to discover life-changing therapeutics/treatments

### Preferred Event Format

No preference

### Preferred Audiences

No preference

### Public Engagement Experience

Science Day at Primary school

## Researcher 5: Jacqueline Siu

### Proposed Topic

Our research (LEGACY project) aims to understand the immune response upon flu vaccination not only in human blood but also in lymph nodes where a lot of the immune reaction originates from. We are able to investigate the in vivo lymph node response by using ultrasound guided fine-needle aspirates. Our topic will primarily be about how understanding the immune response in lymph nodes can help improve future vaccines, and how fine-needle aspirates work using an ultrasound machine activity. In addition, our study focuses on participants with self-declared Black or Asian ancestry where we are aiming to help build a diverse human cell atlas. We can also discuss how single cell technologies such as single cell RNA sequencing have helped further our immunology understanding, but how incorporating ancestral diversity is still a work in progress.

### Preferred Event Format

No

### Preferred Audiences

Interested in engaging with audiences from Black, Asian and Minority Ethnic backgrounds where possible

### Public Engagement Experience

I have personally worked with Cambridge Hands on Science (CHaOS), and also been a workshop facilitator for Genome BC in Canada where I travelled to rural schools / festivals. Our LEGACY project team has also presented at the Imperial Great Exhibition Road Science festival in July 2022.

## Researcher 6: Faranak Hardcastle

### Proposed Topic

Sonic representation of genomic data

### Preferred Event Format

Exhibition/interactive installation

### Preferred Audiences

Open to all

### Public Engagement Experience

None

## Researcher 7: Marsha Wallace

### Proposed Topic

Diabetes in Doggos (and what makes a Labrador a Labrador). Diabetes affects 1 in 300 dogs and is fatal without insulin therapy. As an unintentional result of selective breeding, diabetes risk is variable among breeds e.g. high risk Samoyed, moderate risk Labrador retriever, low risk Boxer. We undertook breed-comparative whole genome sequencing (WGS) to discover genetic variants contributing to differential diabetes risk in these breeds, using samples from the UK Canine Diabetes Database and Archive. We identified genetic changes specific to each breed, learning what makes each unique (what makes a Labrador a Labrador). This included genetic changes in genes that impact coat color, learning and behavior, and other breed characteristics. We developed an evidence-based bioinformatics workflow to rank and prioritize candidate diabetes risk variants based on within-breed and across-breed association statistics, allele frequencies in canine databases, and data from human studies (GWAS, UK Biobank data) to weight plausible roles in diabetes pathogenesis. Then, we performed targeted follow-up sequencing on the prioritized variants in the same breeds, plus 7 additional breeds (Cavalier King Charles Spaniels, Chihuahuas, Greyhounds, Jack Russell Terriers, Miniature Schnauzers, Shih Tzus, and Yorkshire Terriers). Notably, many diabetes-associated variants reside in genes involved in pancreatic beta-cell function or immune response and are distinct between breeds, confirming clinical observations that canine diabetes arises from multiple heterogeneous etiologies. Of particular importance, we found a Labrador-specific homozygous missense mutation in the gene *KCNJ11*, which plays a pivotal role regulating insulin secretion, strongly associated with diabetes. Mutations in *KCNJ11* are one of the most common causes of neonatal diabetes in humans, which can often be successfully treated with oral sulphonylureas instead of insulin injections. Of a total 1,231 diabetic dogs, 9 homozygotes for the mutation were identified, 8 of which were diabetic, implying a causal role in diabetes and potential for therapeutic management with oral sulphonylureas. This study demonstrates how utilization of WGS to investigate complex traits such as canine diabetes will help to inform a precision medicine approach to these conditions. The focus could be either what makes a dog breed unique, or on *KCNJ11* mutation. This topic has broad appeal, especially since during the pandemic, many households became dog owners for the first time. Labrador Retrievers are one of the most common dog breeds, and together with the other breeds included in our study, this topic will be of particular interest to a lot of people. causing diabetes in Labradors.

### Preferred Event Format

No preference

### Preferred Audiences

No Preference

### Public Engagement Experience

Outreach Award – Presented by University of Oxford for contribution to science outreach activities (2014). Educational Activities – Relay for Life, ORCRB & Wellcome Trust Taster Days, Cancer Research Centre Open Days. Career Mentoring – Highcrest Academy Career Fair, Job Shadow Mentor. Public Communications – Oxford University / Ludwig Cancer Institute Podcasts, Guest Scientist for the Nuffield Department of Medicine Twitter Feed, Ada Lovelace Day interview to inspire girls into STEM careers.

## Researcher 7: Katherine Bull and Jessica Kepple

### Proposed Topic

We build cellular pictures of kidney disease. Diabetic kidney disease is the leading cause of kidney failure worldwide. To devise better treatments we need to understand what happens to the kidneys in diabetes, and so our group studies kidney biopsy samples from people with diabetes, and other kidney diseases, at the level of individual cells. We can measure which genes are switched on in the cells, and now we are using new techniques to map exactly the location of each cell in the kidney. This information is crucial to help us build up a picture of the pathways and cellular cross-talk that is altered in kidney disease, which will change how we diagnose disease and provide us with new targets for future therapies.

### Preferred Event Format

Flexible. Could give a talk, but workshop format might be more stimulating. Could do something art related to link with the pictorial/ spatial nature of the work?

### Preferred Audiences

Ethically diverse. any

### Public Engagement Experience

Some experience with talks to schools and patient advocate groups.