PI profile

## Hugh Watkins

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| https://www.well.ox.ac.uk/people/hugh-watkins-1/@@haiku.profiles.portrait/173ba8816907423eb2e0eb830bfa1399/@@images/image/w1140?e9cf05d3-7610-4c5c-878d-fd7b21ac5172 | **Professor Hugh Watkins**  **Titles**: Radcliffe Professor of Medicine, Group Leader / PI and Unit Director  **Location**: Wellcome Centre for Human Genetics & West Wing, John Radcliffe Hospital  **Department**: Radcliffe Department of Medicine (RDM), Department of Cardiovascular Medicine  **Group**: BHF Molecular Cardiology Laboratory  **Webpage**: <https://www.well.ox.ac.uk/people/hugh-watkins-1>  **Email**: hugh.watkins@rdm.ox.ac.uk  **PA**: Tuula Itkonen <watkinspa@rdm.ox.ac.uk |

### GMS themes:

### Research Overview

The genetic contribution to heart disease is important across the allele frequency spectrum. Inherited, monogenic heart diseases are amongst the most common serious, yet treatable forms of inherited disease; coronary artery disease, a classic complex trait, is the single biggest killer worldwide. Our work in inherited cardiomyopathies has moved from gene discovery, through to mechanistic understanding in cell and rodent models to successful clinical trials of repurposed drugs in light of our new mechanistic understanding. We have shown that genetic testing is a cost-effective tool for saving lives. However, our recent large-scale data also indicate that improved, quantitative approaches are needed for reliable interpretation of pathogenicity for rare missense variants – either in gene panels or whole genome data (eg *Genetics in Medicine* 2017).

Our large-scale studies in genetic susceptibility to coronary artery disease (as part of the Cardiogram+C4D Consortium, *Nature Genetics* 2013, 2015 & 2017) have led to the identification of a large number of new risk loci for CAD whose effect is not mediated by known risk factors; thus a major focus now is on identification of new biological pathways and targets. This uses computational and high-throughput screening tools to prioritise loci and variants for functional studies.

Project areas: genetics, epigenetics & genomics, cardiovascular medicine, inherited heart disease.

### Specific project proposals:

Please contact directly for further information.

*These pages were reviewed/updated:* ***10/06/2022***

Project proposal

# **Title**: **[Project title here]**

Supervisors: [name and title of relevant individuals]

Wet/dry lab mix (approx): X% wet lab, X% dry lab

### Description:

[Write a ~ half-page page description of the project here].

### Training Opportunities:

[Write a brief description of the training opportunities the project will provide].

### Background reading / references:

Please include references as desired. Suggested format:

* [Surname] [Firstname], [other authors]… **(year in bold)** . [Title]. [Journal name], [other details]. Available at: [link]

Insert any additional project description(s) on subsequent pages if applicable. Please use the same template and use separate pages for each project.