



# Congenital Anomalies Cardiovascular Geneboree: From Cardiac Gene Variant to Mouse Model

## Aim:

- Create a framework for accurately modelling and phenotyping human congenital heart defects in mouse
- Link human and mouse phenotyping

## Audience:

- Clinicians, clinical geneticists and developmental biologists with an interest in finding new genes for congenital heart defects and validating them in mouse models

## When:

- Thursday 23rd-Friday 24th November, 2023

## Where/How:

- MRC Harwell hosted as a hybrid event
- Places are limited
- Free registration required via Eventbrite
- Scan the QR code to register





# Part 1:

## The Complexity of CHD Genetics and Phenotyping

Thursday  
23<sup>rd</sup>  
Nov.

- 12.30** • **Arrival & Lunch**
- 13.30** • Welcome and Introduction:  
**Deborah Henderson** and **Pleasantine Mill**
- 13.40** • Overview of how to phenotype a human heart – segmental analysis and importance of nomenclature.  
**Andrew Cook** (ICH-UCL)
- 14.10** • Phenotyping and coding of congenital heart defects in humans. **Marina Hughes** (Norfolk and Norwich University Hospitals NHS Foundation Trust)
- 14.40** • Human cardiovascular genetics – what can we/have we learnt from large scale screening of CHD patients?  
**Bernard Keavney** (Manchester, 100K Genomes CHD GeCIP lead)
- 15.10** • **Coffee**
- 15.40** • The complexities of modelling disease variants and creating mouse models of human CHD  
**Jose Luis de la Pompa** (CNIC – Madrid)
- 16.10** • The importance of appropriate CHD phenotyping, filtering and variant validation.  
**Bill Chaudhry** (Newcastle and Chair ESC WG-DAP)
- 16.40** • Can we link mouse phenotypes to “coded CHD” in people?  
**Lucile Houyel** (Necker, Paris)  
• **Flash talks** – 5 mins x 5-6 speakers about their variant/mouse
- 17.10** • **Short Break**
- 17.15** • **Virtual Presentation Keynote Speaker**  
**Benoit Bruneau** - Gladstone Institute of Cardiovascular Disease, San Fransisco
- 18.00** • **Finish and meal for all attendees**



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# Part 2:

## How should you validate your CHD VUS, phenotype your mouse mutant for CHD and understand the underpinning mechanism?

Friday  
**24<sup>th</sup>**  
Nov.

- 09.00** Preliminary considerations  
**Deb Henderson** (Newcastle and NMGN)
- 09.30** Large-scale mouse cardiovascular phenotyping  
**Nadine Spielmann** (German Mouse Clinic, Munich)
- 10.00** Imaging phenotypes of CHD mouse models  
**John Sled** (Centre for Phenogenomics, SickKids, Toronto)
- 10.30** **Coffee**
- 11.00** Mouse and human cardiac atlases  
**Steve Lisgo, Janet Kerwin, & Lorraine Eley** (Newcastle)
- 11.30** Mouse mutant design and analysis  
**Lydia Teboul** (Mary Lyon Centre, Harwell)
- 12.00** Understanding the underlying cardiovascular mechanism in your mouse model  
**Stephane Zaffran** (Marseille)  
- Or flash talks here
- 12.30** How can we better align mouse with human phenotyping?  
Open discussion aimed at aligning mouse and human terms  
(speakers as panel members) - all
- 13.15** **Light lunch and departure**



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