

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
23rd
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 Francisco

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
24th
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)
- 12.30** • **Flash Talks:** How can we better align mouse with
human phenotyping?
- 13:00** • **Open Discussion:** aimed at aligning mouse and human terms
(speakers as panel members) - all
- 13.30** • **Light lunch and departure**



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