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)

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
## Part 2:

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

### Friday 24th Nov.

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Speaker(s)</th>
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</thead>
<tbody>
<tr>
<td>09.00</td>
<td>Preliminary considerations</td>
<td><strong>Deb Henderson</strong> (Newcastle and NMGN)</td>
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<tr>
<td>09.30</td>
<td>Large-scale mouse cardiovascular phenotyping</td>
<td><strong>Nadine Spielmann</strong> (German Mouse Clinic, Munich)</td>
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<tr>
<td>10.00</td>
<td>Imaging phenotypes of CHD mouse models</td>
<td><strong>John Sled</strong> (Centre for Phenogenomics, SickKids, Toronto)</td>
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<tr>
<td>10.30</td>
<td>Coffee</td>
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<tr>
<td>11.00</td>
<td>Mouse and human cardiac atlases</td>
<td><strong>Steve Lisgo, Janet Kerwin, &amp; Lorraine Eley</strong> (Newcastle)</td>
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<td>11.30</td>
<td>Mouse mutant design and analysis</td>
<td><strong>Lydia Teboul</strong> (Mary Lyon Centre, Harwell)</td>
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<td>12.00</td>
<td>Understanding the underlying cardiovascular mechanism in your mouse model</td>
<td><strong>Stephane Zaffran</strong> (Marseille)</td>
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<td>12.30</td>
<td><strong>Flash Talks:</strong> How can we better align mouse with human phenotyping?</td>
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<tr>
<td>13.00</td>
<td><strong>Open Discussion:</strong> aimed at aligning mouse and human terms (speakers as panel members)</td>
<td><strong>all</strong></td>
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<tr>
<td>13.30</td>
<td>Light lunch and departure</td>
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