1. The DMDD programme

**WHAT**
The DMDD programme is a systematic study of mouse knockout genes that result in embryonic or perinatal death. Our online database of high-resolution embryo images and phenotypes is a unique and expanding resource for both developmental biologists and clinicians.

**WHY**
Our goal is to further understanding of the genes critical for embryo development and survival. The data has already suggested links between specific genes and abnormal embryonic heart structure, and has huge potential to shed further light on the aetiology of human developmental disorders.

**HOW**
Embryos are comprehensively examined using the highest-resolution 3D imaging methods. A team of expert anatomists then scores structural abnormalities in tissue organisation and organ structure to identify the likely causes of the failure of the embryo to develop properly.

**WHO**

2. Our database dmdd.org.uk

At the heart of the DMDD programme is the commitment to make our data freely available to the biomedical community. Our online database holds the complete DMDD image and phenotype dataset, and is continually updated with new results.

- **Over 5 million images**
- **Over 650 embryos**
- **Over 3,000 images per embryo**
- **Complete image stacks**
- **Standardised phenotypes**
- **Penetrance data**
- **Reference embryos**

3. Cardiovascular phenotypes are highly prevalent

78% of lines have at least one cardiovascular phenotype

4. Detailed images of phenotypes available

- Double outlet right ventricle
- Perimembranous ventricular septal defect
- Muscular ventricular septal defect
- Bicuspid aortic valve
- Common arterial trunk and VSD
- Normal placenta
- Abnormal placenta

5. Psat1 and Psph knockouts model Neu-Laxova Syndrome

Caused by disruption to the L-Serine pathway

Characterised by:
- severe intracranial growth restriction
- proptosis (bulging eyes)
- eyelid malformations
- nose malformations
- round and gaping mouth
- micromaxhia (small jaw)
- cleft lip or cleft palate
- limb malformations
- edema
- neural tube defects

6. Placental defects are important

Around a third of gene knockouts that cause placental phenotypes also cause a heart defect. A statistical study shows this is a genuine link.