Embryology and cardiovascular genomics

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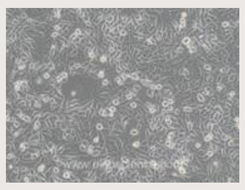
Victor Chang Cardiac Research Institute

Why are babies born with heart defects?

Here at the **Embryology laboratory at the Victor Chang Cardiac Research Institute**, we are trying to answer this question. **1 in 10 babies are born with a heart defect.** Current research tells us that gene mutations or environmental factors like diet, drugs or disease can cause these defects. But so far, we only have **an answer for 25% of the families** who are looking for answers. So, we've come up with a few ways to change the odds in our favour.

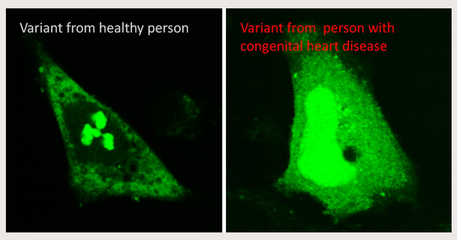
Whole genome sequencing and functional genomics

Whole genome sequencing is a revolutionary technique that lets us identify all variations in a baby's DNA. Currently, we have **sequenced the DNA of 156 families** affected by congenital heart defects. In these families, we have found mutations genes that could be the **cause of the heart defect**. In the lab, we are using **cell lines to test how these mutations could be affecting heart development**.



Transfection of cultured cells with DNA encoding variants

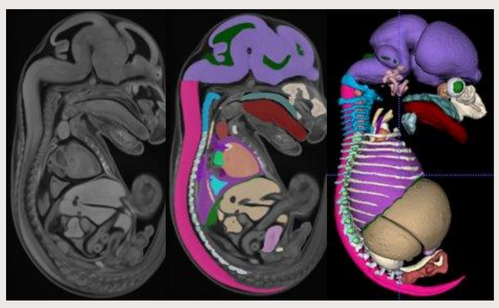
Automated imaging using Opera Phenix robot

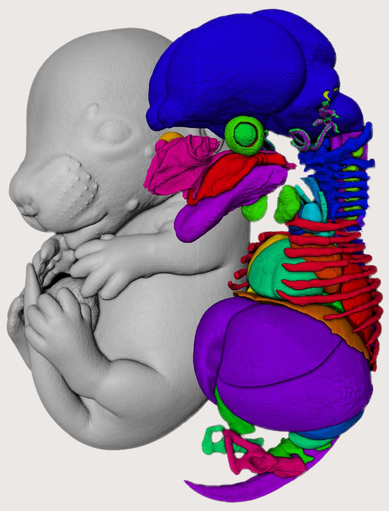


Imaging data analysis

Mouse models of congenital heart defects

Our lab **uses pioneering gene editing techniques** to create mouse models with heart defects and analyse them using **state of the art** imaging techniques (Micro-CT) together with an automated phenotyping pipeline. **Studying a human mutation in a mouse model** is an essential way to prove that the gene is crucial for **development and proper function of the heart.**





Where you come in

Our research has lead to the **discovery of new heart genes, new pathways needed for heart development, new ways to provide healthcare interventions for at-risk pregnant women**. But there is still so much to do and to discover. We need more people to join us. If you're excited about learning about how a single nucleotide change can stop the heart from forming, then we're the lab for you.

What you will walk away with:

Genome sequencing analysis and variant interpretation

Molecular biology techniques (cloning, cell culture, imaging etc)

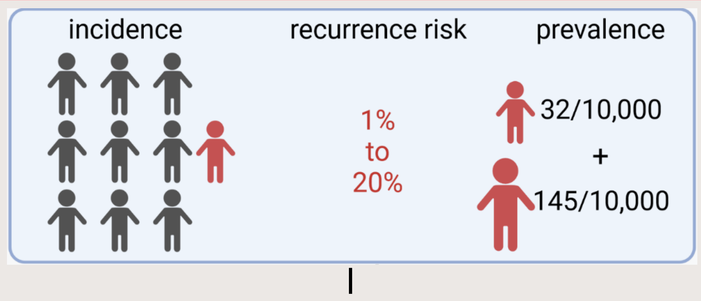
Micro-CT and automated embryo phenotyping

Animal handling

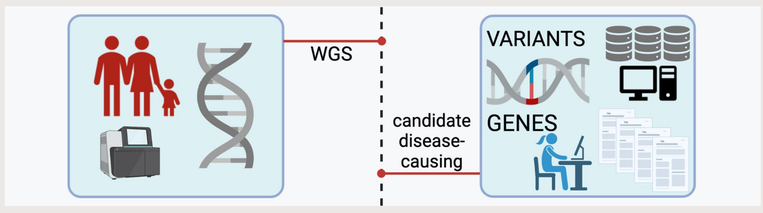
Understanding of cardiovascular genetics and functional genomics

Laboratory experience in one of the top cardiovascular genetics labs in the world

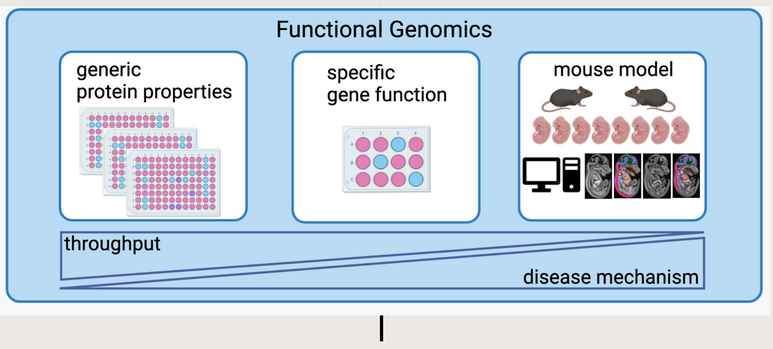
1. Recruiting patients and families with congenital heart defects



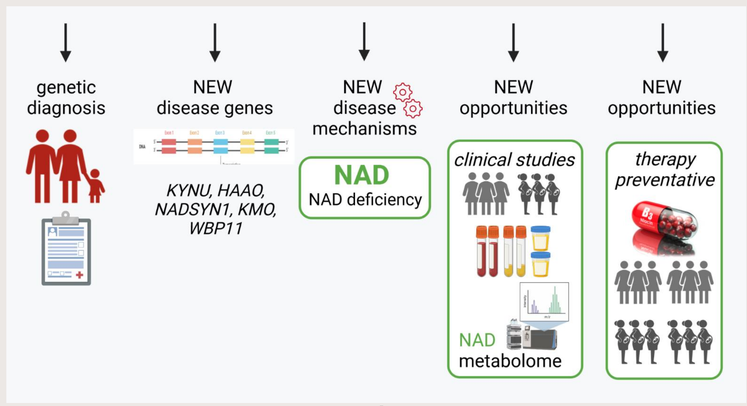
2. Genome sequencing analysis and variant identification



3. Functional genomics



4. Outcomes



5. Your honours project!

People involved

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