# ARMI SPECIAL SEMINAR 2023

## Human iPSCs as a disease modeling platform to explore congenital heart defects and cardiomyopathies in rare genetic disorders


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### Abstract

Noonan syndrome (NS) belongs to a group of genetic disorders called RASopathies that are caused by germ-line mutations that affect genes residing along the canonical RAS-MAPKinase signaling pathway. NS constitutes one of the most common causes of congenital heart defects (CHDs). Infants with NS present with cardiomyopathies and a variety of severe CHDs and no specific treatment exists for children with NS and exhibiting hypertrophic cardiomyopathy (HCM). Moreover, the molecular mechanisms underlying cardiogenesis defects in NS remain poorly understood and an important need lingers to uncover therapeutic strategies through the in-depth investigation of the molecular causes of CHDs and cardiomyopathies in NS. In this seminar, I will first present how we used human induced pluripotent stem cell (hiPSC)-derived cardiomyocytes to decipher the molecular pathways that underlie HCM in NS. I will next highlight our ongoing efforts to understand the function of RAF1 in early human cardiogenesis as well as the impact of NS *RAF1* mutations in this process. I will end by presenting my research program. I will discuss how we can leverage our hPSC-based platform to study cardiogenesis and cardiac function in genetic disorders as a unique opportunity to discover the molecular perturbations responsible for CHDs or cardiomyopathies.


### Bio

Fabrice Jaffré is an Assistant Professor of Cell and Developmental Biology at Weill Cornell Medical College in New York City. He obtained his PhD at the Sorbonne University in Paris, where he explored the function of serotonin receptors in pathological cardiac hypertrophy. Fabrice later joined Beth Israel Deaconess Medical Center, Harvard Medical School where he used hiPSCs to delineate the molecular mechanisms underlying hypertrophic cardiomyopathy in RASopathies. His work was supported by a Scientist Development Grant from the American Heart Association. His lab uses human iPSCs, cardiac directed differentiation, genome editing (CRISPR), multi-omics, 3D organoids and engineered heart tissues as developmental and disease models. Fabrice's work is currently supported by the Upenn Orphan disease Center, the UK Noonan syndrome Association and by the Additional Ventures Single Ventricle Research Fund Award.

## EVENT DETAILS

### DATE:

September 06th, 2023

### TIME:

10.00 – 11.00noon

### VENUE:

G19, Opposite Cinque Lire
15 Innovation Walk
Monash University
Clayton Campus

### HOST:

Prof. Peter Currie

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