Project Title: The Undiagnosed Genetic Heart Disease Program	**Code: CENT3**
Host School / Institute: Centenary Institute	**Address:** Building 93, RPAH Grounds, Missenden Road, Camperdown NSW
Certificates & Clearances required: No	
Primary Supervisor: A/Prof Jodie Ingles	
Phone: 0409 307 387	**Email:** j.ingles@centenary.org.au
Co-Supervisor/team: The research team includes clinicians, genetic counsellors, students and research assistants.	
Project Type: Clinical	
Project Category: Cardiovascular; Genetics	
Skills / Attributes of a successful student: Clinical skills (i.e. medical student) would be desirable as the project will require a lot of interpretation of medical information.	
Project Keywords: Undiagnosed diseases; Cardiomyopathies; Genomics; Arrhythmia syndromes; Phenotyping	

Project Description: A proportion of patients attending the Genetic Heart Disease clinics at Royal Prince Alfred Hospital do not meet any formal diagnostic criteria. It is hypothesised these patients have either (1) a more diverse and unrecognised spectrum of a known disease, or (2) a rare and as yet unclassified disease. Rare diseases are considered those with a prevalence of less than 1 in 2000, the large majority are genetic and affect young people.

The patients with undiagnosed diseases in our clinic are likewise often younger with more severe presentations, with a very low diagnostic yield when sequencing known disease genes. We propose to adopt a similar model to the Undiagnosed Disease Network in the USA, which uses a multidisciplinary clinical and research approach to solve medical mysteries. No such program exists that is specifically focused on primary cardiac diseases. We aim to develop an Undiagnosed Genetic Heart Disease Program, which will make a diagnosis in our undiagnosed patients where current knowledge fails to do so, and to evaluate the effectiveness of the program.

This project will involve the student to help interpret medical information and work on entering Human Phenotype Ontology (terms) in to a specific database. The student will also assist with re-analysis of exome or genome data, specifically working through candidate variants to determine whether they are likely candidates.