



## RACP Foundation Research Awards

### FINAL REPORT

<b>Project / Program Title</b>		Identifying cardiac risk in Fabry disease
<b>Name</b>		Dr Rebecca Kozor
<b>Award Received</b>		2018 Sir Roy McCaughey Fellowship (Research Establishment)
<b>Report Date</b>		10 December 2019
<b>Chief Investigator / Supervisor</b>		Dr Rebecca Kozor, Professor Gemma Figtree
<b>Administering Institution</b>		University of Sydney
<b>Funding Period</b>	Start Date:	24 September 2018
	Finish Date:	23 September 2019

#### PROJECT SUMMARY

Fabry disease (FD) is a rare genetic disorder that can affect the heart, and can cause heart failure, rhythm problems and sudden death. Information on the frequency of proven rhythm problems (arrhythmias) in FD is limited, as is the ability of the electrocardiogram (ECG) to predict heart issues. Also, early cardiac involvement is difficult to detect but state-of-the-art technology – cardiovascular magnetic resonance (CMR) with T1 mapping and advanced electrocardiography (A-ECG) – offer diagnostic potential. We hypothesized that A-ECG can detect early cardiac involvement of FD as defined by CMR.

#### PROJECT AIMS / OBJECTIVES

This study aimed to assess the ability of advanced ECG to predict early cardiac involvement in FD using CMR as the reference standard.

#### SIGNIFICANCE AND OUTCOMES

**Results:** Two vectorcardiographic A-ECG variables derived from the 12-lead ECG predicted cardiac involvement (low native T1): (1) the direction of the azimuth of the T loop in the horizontal plane, (2) the QRS magnitude (mV) in the left sagittal plane. These 2 ECG variables were combined to make an A-ECG score that detected low native T1 with an AUC of 0.88 [95% CI (0.77-0.96)], specificity of 95% [95% CI (77-100%)] and sensitivity of 79% [95% CI (59-97%)].

**Conclusions:** A-ECG can predict cardiac involvement in FD with good sensitivity and high specificity using two variables derived from the 12-lead ECG. This supports the use of A-ECG as

a screening tool for cardiac involvement in patients diagnosed with FD. The ECG is a simple cheap investigation and thus has the potential for widespread adoption.

Further research with a larger sample size is required and ongoing. We have collaborated with 2 international sites to increase participant numbers, plus extend the study to analysing ECGs in those FD patients who are undergoing implantation of a loop recorder (heart rhythm recording device).

#### **PUBLICATIONS / PRESENTATIONS**

Student presentations:

1) Abstract – University of Sydney Cardiovascular Initiative Precision Medicine Symposium, Nov 2018, University of Sydney.

2) Abstract – Poster presentation at Society of Cardiovascular Magnetic Resonance Scientific Sessions, Feb 2019, Seattle USA.

Manuscript – in progress