



RACP Foundation Research Awards

FINAL REPORT

Project / Program Title	Investigating pathways important for mycobacterial immunity in patients with primary immunodeficiency	
Name	Dr Katrina Randall	
Award Received	2015 AstraZeneca Research Establishment Fellowship in Medical Research	
Report Date	16 June 2015	
Chief Investigator / Supervisor	Prof Matthew Cook	
Administering Institution	Australian National University	
Funding Period	Start Date:	1 January 2015
	Finish Date:	31 December 2015

PROJECT SUMMARY

Primary immunodeficiencies are diseases in which patients can't fight infection due to a genetic defect that affects a particular part of the immune system. We study primary immunodeficiencies not only to help the patients with these diseases but also to gain an insight into how a normal immune system works.

Recent advances in genetic sequencing have meant that in more cases we are able to find the genetic defect responsible for a particular primary immunodeficiency called common variable immunodeficiency (CVID) which encompasses a number of different diseases all characterised by low antibody levels.

This project looks at patients with CVID who also have mycobacterial infections (such as tuberculosis) which is an unusual infection to have in this condition and indicate that their genetic defect may differ from other patients with this condition.

PROJECT AIMS / OBJECTIVES

The aims of this project are to investigate a particular kindred with CVID and mycobacterial infection to find the predisposing genetic defect and to apply any assays set up to investigate this to other patients with the same condition.

SIGNIFICANCE AND OUTCOMES

Research is ongoing to confirm the likely genetic defect.