

AHCDO John Lloyd Clinical Excellence Grant

Andrea Johannessen

The Australian Haemophilia Centre Directors' Organisation (AHCDO) Executive Committee are pleased to announce that the successful application for the AHCDO John Lloyd Clinical Excellence Grant 2022 round is Dr Radha Ramanan for the project titled **Phenotypic characterisation and molecular profiling of congenital fibrinogen disorders: the Australian experience.**



Dr Radha Ramanan

Dr Radha Ramanan is a Haematology Fellow at Alfred Health in Melbourne and is also undertaking a PhD at Monash University, having completed her advanced training in clinical and laboratory haematology in 2021 at Alfred Health. Radha is also the current AHCDO Research Fellow. Radha has an interest in bleeding disorders and molecular pathology and seeks to broaden our understanding of these fields through research funded by AHCDO.

Congenital fibrinogen disorders are rare disorders affecting one of the major clotting proteins of the blood, fibrinogen. These disorders are inherited which means these patients are born with the problem. This disorder causes different problems in different patients, for example, one person may experience blood clots while another may have problems with bleeding, while someone else again may have no noticeable problems at all.

Given the rarity of this disorder, we are still trying to understand out why some patients behave differently

to others. Some of this difference may relate to the various gene mutations that can be seen with this condition. Genes are the genetic material which code for all the proteins in the body, including the fibrinogen clotting factor.

This project will look closely at the types of gene mutations seen in various patients with a fibrinogen disorder by genetic testing. We will also record the types of bleeding or clotting problems these patients have experienced over their lifetime, and if any of those problems arose during pregnancy or surgeries. We will assess types of treatment these patients have received in these different settings.

This project aims to provide an analysis of real-world data concerning the cohort of people with congenital fibrinogen disorders across Australia. The results obtained will inform how successful current therapeutic approaches are in the prevention of bleeding/thrombotic events. It will also broaden our understanding of the genotype-phenotype correlation present within this disorder and guide whether molecular typing at diagnosis may help us better characterise the clinical phenotype, navigate future treatment decisions, particularly regarding prophylaxis, and ascribe inheritance risk in this complex cohort of patients.

AHCDO is grateful to the following organisations for their financial support of the John Lloyd Clinical Excellence Fund - Roche, Sanofi Genzyme, Pfizer and Novo Nordisk for their support.

More information about the John Lloyd Clinical Education Fund is available from the AHCDO website - www.ahcdo.org.au.

.....
Dr Andrea Johannessen is AHCDO
Executive Officer
.....