



**Walter+Eliza Hall**

Institute of Medical Research

DISCOVERIES FOR HUMANITY

20 August 2019

RCPA Foundation Chair,  
The Royal College of Pathologists of Australasia,  
Durhan Hall,  
207 Albion Street,  
Surry Hills, NSW 2010,  
Australia.

Dear RCPA Foundation Chair,

**Re: RCPA Foundation Illumina Cancer Research Grant 2017 – Final Report.**

I am writing to provide the final report on the outcome of my project “Investigating the utility of next-generation sequencing technologies in predicting relapse in patients with core-binding factor acute myeloid leukaemia (CBF-AML)” that was supported by the RCPA Foundation Illumina Cancer Research Grant.

Since the grant was awarded at Pathology Update 2018, I have used the funds to analyse the transcriptomic profiles of 119 samples collected from 35 patients with CBF-AMLs who participated in the Australasian Leukaemia and Lymphoma Group (ALLG)’s AMLM13 trial. Working closely with WEHI’s bioinformaticians, Anna Quagliari and Dr Christoffer Flensburg, we showed that the two cytogenetic subtypes of CBF-AMLs (inversion(16) and translocation(8;21)) have distinctive gene expression profiles. We found that our results were consistent with other international cancer datasets, such as those generated as part of The Leucegene Project. We also demonstrated that the tissue source of the sample has an impact on gene expression and have developed approaches to account for this issue.

We developed a method to detect genetic mutations from the transcriptome sequencing data. We identified frequent mutations in the Ras signalling pathway, which was consistent with published results from The Cancer Genome Project and The Leucegene Project. We have gone on to characterise genetic mutations and expression profiles associated with relapsed AML. We plan to extend this work to a separate cohort of CBF-AML samples to see if the findings can be validated. We hope to present the results to ALLG in their next scientific meeting.

I would like to thank the RCPA Foundation for their support. We remain committed to better understanding this deadly haematologic disease that is in urgent need of new treatments.

Kind regards,

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