RCPA Foundation Mike and Carole Ralston Travelling Fellowship Recipient 2016
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Final Report Summary

The 2016 Mike and Carole Ralston Travelling Fellowship supported my research activities undertaken at the Karolinska Institutet in Stockholm, Sweden as part of my PhD candidature, under the supervision of Professor Lennart Hammarström in the Division of Clinical Immunology, Department of Laboratory Medicine. My research focussed on strategies for newborn screening for primary immunodeficiency diseases.

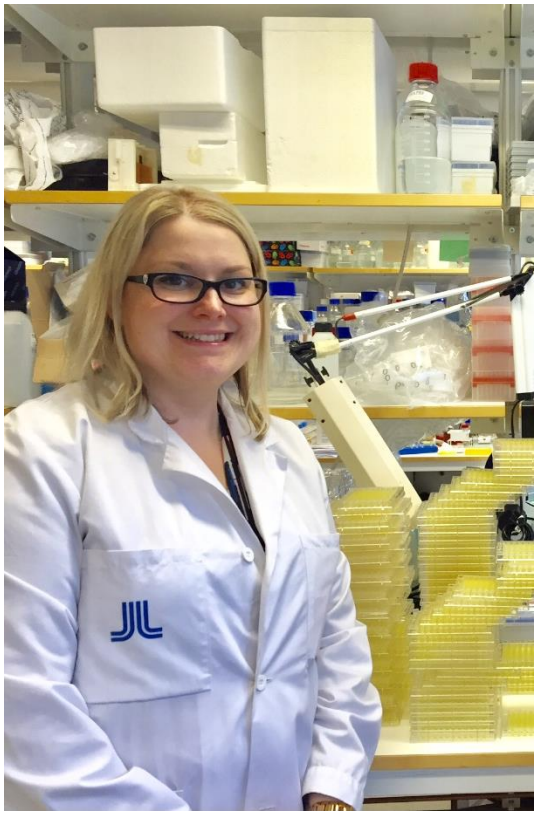
Primary immunodeficiency diseases (PID) are a heterogeneous group of over 320 different inborn errors of immunity which result in significant morbidity and mortality. Early diagnosis and treatment improve outcomes for affected patients. In particular, this has been demonstrated for babies with severe combined immunodeficiency (SCID), where survival and long-term outcomes are significantly improved if haematopoietic stem cell transplantation is performed prior to 3.5 months of age. Newborn screening provides an opportunity for early identification of affected infants within the first weeks of life.

The ideal strategy by which to screen infants for PID remains controversial, and this research examined the current status of newborn screening for PID internationally. We evaluated the utility of screening infants for severe forms of PID using a DNA-based assay performed on infant dried blood spots, demonstrating that a combined TREC/KREC (T cell receptor excision circle/kappa-deleting recombination excision circle) assay was able to successfully identify infants with PID manifested by T and/or B cell lymphopaenia. We further evaluated this assay in clinical contexts outside of the newborn screening period in older patients with suspected PID. We also reported other novel approaches by which to screen infants for PID. One strategy harnessed transcriptomics to identify infants with hypogammaglobulinaemia, the second utilised a genotyping assay to identify genetic polymorphisms conferring specific disease susceptibility.

These research findings have been published in peer-reviewed journals, and in a PhD Thesis entitled ‘Newborn Screening for Primary Immunodeficiency Diseases’. They were also presented at the 2018 RCPA Pathology Update meeting. I would like to thank Mike and Carole Ralston, and the RCPA Foundation, for their generous support which enabled me to undertake this research. I hope that this work will contribute to the evolving field of newborn screening for PID, and that ongoing advancements in genetic techniques will continue to improve the lives of children with PID and other genetic disorders.

Testimonial

The 2016 Mike and Carole Ralston Travelling Fellowship supported my research activities undertaken at the Karolinska Institutet in Stockholm, Sweden as part of my PhD candidature, under the supervision of Professor Lennart Hammarström in the Division of Clinical Immunology, Department of Laboratory Medicine. My research focussed on strategies for newborn screening for primary immunodeficiency diseases. I would like to thank Mike and Carole Ralston, and the RCPA Foundation, for their generous support which enabled me to undertake this research. I hope that this work will contribute to the evolving field of newborn screening for PID, and that ongoing advancements in genetic techniques will continue to improve the lives of children with PID and other genetic disorders.



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