

## **Richard Hughes Travel Bursary May 2023**

I am very grateful to the BPNS for their support towards my attendance at the joint meeting of the Association of British Neurologists (ABN) and the Irish Neurological Association (INA) meeting in Belfast.

As many members of my family are originally from Belfast, it was a personal and unique opportunity to travel back to a beautiful part of Ireland and experience the delights the city has to offer and mingle with fellow colleagues. I had the honour of delivering a platform presentation entitled, 'Skin biopsy as a diagnostic tool for ATTRv amyloid neuropathy in the UK.' This is a hugely exciting time in the field of inherited neuropathies as gene silencing therapy for ATTRv now represents the first treatment for adult onset inherited neuropathy. Importantly this study described how skin biopsy has allowed a substantial number of patients with the T60A and V122I TTR variants early access to gene silencing therapy by diagnosing amyloid neuropathy at an earlier stage, often when neurophysiology is normal. My talk was particularly fitting given the T60A founder population was described back in 1991 by my supervisor, Professor Mary Reilly. How remarkable it is, that now, over 32 years later, we are not just describing the disease but now treating it and enhancing the lives of patients. It is a privilege to be part of this journey that continues onwards to this day.

Continuing with the theme of ATTRv, I also presented a poster entitled, 'Spinal MRI findings in UK patients with hereditary ATTRv amyloidosis.' This was particularly interesting given that ATTRv is a multisystemic disease, affecting not just different organs but also various tissue types, for example the ligamentum flavum, where it can result in spinal stenosis. This poster presentation allowed me to showcase our findings in relation to ATTRv patients with spinal stenosis and how it can affect patients with multiple different TTR variants. The feedback and interest in this was very positive and of course educational, as it highlights the importance of considering spinal stenosis when ATTRv patients present with neuropathic symptoms.

I was also able to showcase my research with another poster entitled, 'Validating MRI biomarkers for clinical trials in Charcot-Marie-Tooth Disease 1A using automated segmentation.' Lower limb muscle MRI is rapidly becoming more recognised as a highly responsive biomarker in slowly progressive diseases like CMT1A. It also has huge potential as an outcome measure in clinical trials and much of this research is dedicated to providing further evidence for this. However the segmentation process of lower limb muscle MRI images can be time consuming. Our team at UCL have developed an AI-enabled automated segmentation method which dramatically reduces processing times for these images. The practical advantage of this, is that it will potentially increase the efficiency of clinical trials for CMT patients in the future where MRI is being used as an outcome measure.

The combined meeting of the ABN and INA was unique as it represented a huge opportunity to interact with colleagues from multiple disciplines and sub-specialties. My supervisor, Professor Mary Reilly delivered the ABN medallist lecture. This was very inspirational and gave us all a great insight into what has been achieved in the field of peripheral neuropathy research and the challenges that lie ahead. As many past and fellow fellows can attest to, being part of this team at Queen Square and contributing to this field of research is something we all cherish.

Another brilliant lecture was delivered by Professor Christopher McGuigan on clinically isolated syndrome and how we can identify important characteristics in these patients that signify a higher likelihood of transforming into multiple sclerosis. This was particularly informative as it highlighted that these higher risk patient groups need to be followed up more closely.

Overall, I would like to thank the BPNS again for supporting me in presenting my work. It offered me the chance to catch up with colleagues including those I have previously worked with and it inspired me further, to continue the research journey to improve the lives of the patients we treat.