Richard Hughes Travel Bursary July 2018 – Menelaos Pipis' report for attendance at the annual Peripheral Nerve Society Meeting in Baltimore, USA.

I am very grateful to the BPNS for their support towards my attendance at the annual Peripheral Nerve Society (PNS) meeting which was held in Baltimore, USA between 21st-25th July 2018. This meeting brings together neurologists, clinical scientists and allied health professionals with an expertise in peripheral nerve disease from around the world and is an excellent opportunity for an update on recent clinical and scientific advances as well as fostering professional relationships and collaborations.

As an Inherited Neuropathy Consortium (INC) fellow, I had the opportunity to attend a pre-arranged meeting of the INC on the first day of the conference where I met principal investigators (PIs) from other US and international centres as well as other INC fellows. During the meeting, we discussed the progress of the consortium's various projects including the natural history project and the whole exome sequencing project whose purpose is to identify genetic modifiers of CMT1A. As part of my PhD and under the supervision of Mary Reilly and Alex Rossor, I am leading the collective efforts of the consortium to better characterise the genotype-phenotype correlations in CMT2A using the cohort of 209 patients (158 kindreds) enrolled in the INC; this meeting also gave me the opportunity to discuss various aspects of the project with other PIs, such as Mike Shy from Iowa City, USA.

Delegates of the PNS meeting had the opportunity to attend plenary lectures and oral presentations delivered by distinguished speakers, specialist interest groups (hereditary and acquired neuropathies) that ran in parallel, as well as poster sessions. John Svaren delivered oral presentations on the recent success of the PMP22 antisense oligonucleotide (ASO) therapy in the CMT1A rodent model as well as the advancing genetic therapies in CMT and related disorders. These sessions provided an excellent update in the field and an insight into the challenges we might encounter as a scientific community in trying to push these therapies from the bench to the clinical trial phase. Claudia Sumner's and Arthur Burghes' lectures on the progress of gene therapy in spinal muscular atrophy exemplified the opportunities that open up with these therapies and challenges relating to dosing, delivering the ASO intracellularly and achieving and measuring appropriate target engagement. In a late breaking abstract, Andrea Cortese presented the work he has done with Mary Reilly and Henry Houlden in deciphering the recessively inherited genetic cause of CANVAS. This was an eye-opener, since it illustrates that late-onset recessive conditions may be more common than previously thought and clinicians should bear this in mind when considering genetic testing for late onset neuropathies which may be slowly progressive.

During the poster sessions, I had the opportunity to present my ongoing work on the genotype-phenotype correlations of CMT due to mutations in NEFH. In this context I also met Byung-Ok Choi and his team from the Samsung Medical Centre, Seoul, South Korea, with whom we are co-authoring an important phenotype paper on this CMT subtype and which includes their family with NEFH. Attending other poster presentations and discussing with other delegates the various new genetic causes and pathomechanisms of CMT was important and has given me take-home messages that I will use when subsequently analysing next-generation sequencing (NGS) data.

In the era of NGS and whole genome sequencing (WGS), international collaborations are important in carrying out enrichment analyses of WGS data and the PNS meeting gave me the opportunity to meet Pieter Van Doorn's and his team from the Netherlands and discuss a potential collaboration. They are collecting a significant cohort of more than 100 people with peripheral neuropathies in which they are carrying out WGS and we are already working together to try and analyse our data in parallel and learn from each other's bioinformatic approaches.

In conclusion, attending the PNS was a fantastic opportunity both from an educational as well as research point of view and once again I am grateful to the BPNS for their support.

Menelaos Pipis