

Dear ESHG,

I wish to thank the ESHG educational committee for selecting me for the 2024 mentorship programme. I am a neurology trainee with a subspecialty interest in neurometabolic and primary mitochondrial disease, and prior to medical studies I completed a basic science PhD in neurodegenerative disease and neuroinflammation. It is my goal to ultimately develop a specialised multidisciplinary service for primary mitochondrial disease in Ireland, something that is currently not available to these patients.

For my mentorship, I spent one week in Newcastle upon Tyne, in the Wellcome Centre for Mitochondrial Research in Newcastle University and the NHS Highly Specialised Services for rare mitochondrial disorders in adults and children. Under the mentorship of Prof Gráinne Gorman and her team, I was exposed to all aspects of the mitochondrial service, including direct patient care in the specialist clinic, the reproductive advice service, research study planning, and the diagnostic laboratory service.

Patients attending the weekly mitochondrial clinic are reviewed by a team comprising neurologists, specialist nurses, physiotherapy, speech therapists, social workers, thus ensuring all aspects of the patient's needs are addressed at the one appointment. There are also close relationships with specialties including cardiology, ophthalmology and endocrinology, to ensure non-neurological aspects are dealt with promptly. My attendance at this clinic provided inspiration for the type of service I would aspire to develop in Ireland.

During my week I met with multiple clinicians in the service, alongside the nursing and physiotherapy team, in order to gain insight into what the essential requirements would be to run an Irish mitochondrial service. Of particular benefit, I met with senior diagnostic laboratory scientists, who offered their time to discuss the complexity of some challenging cases, and provide detailed teaching on variant interpretation, which as a neurologist with no formal genetics training was particularly beneficial.

Throughout my time in Newcastle, I felt incredibly supported from the entire team, and was reassured that they were happy to provide any assistance I might require as I progress in my career. I was also reassured that the team would always be willing to provide an expert discussion forum for difficult clinical cases, and suggested we may ultimately formalise a regular case meeting over the coming years.

I hope to return to Newcastle for a period of post-training clinical fellowship, to gain a more immersive exposure to mitochondrial disease and consequently arm me with the necessary skills to diagnose and manage this complex patient group for the remainder of my career. As such, I am extremely grateful for the ESHG mentorship as I feel it has provided an important starting point upon my path to deliver a centralised first-in-Ireland service for mitochondrial disease.

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