Royal College of Physicians

Professor Mary M. Reilly Biography

Professor Mary M. Reilly graduated from University College Dublin in 1986, received her MD in 1996, FRCP in 2002 and her FRCPI in 2003. She was appointed a consultant neurologist at the National Hospital for Neurology and Neurosurgery, Queen Square in 1998 and a Professor of Clinical Neurology at UCL in 2010. She leads the peripheral nerve clinical service in the National Hospital for Neurology and Neurosurgery, leads a research group in the MRC centre for Neuromuscular Diseases in the Institute of Neurology (ION) and is head of the Division of Clinical Neurology in ION.

She has a longstanding interest in the clinical management and research in the inherited peripheral neuropathies and runs a research program encompassing gene identification, pathogenetic studies, natural history studies, development of outcome measures and conducting clinical trials in inherited neuropathies. Professor Reilly is a co-director of both the UCL MRC Centre for Neuromuscular Diseases and the NIH Inherited Neuropathy Consortium. She is the President of the Association of British Neurologist (ABN), a past President of the British Peripheral Nerve Society (BPNS), and immediate past President of the international Peripheral Nerve Society (PNS).

Presentation Summary

Topic: Rare Diseases – Global Impact

Identifying the genes responsible for inherited conditions and developing therapies for these conditions is an area of medicine where there has been major advances in the last decade.

This is particularly relevant for neurological diseases as there are both a vast number of rare inherited neurological diseases and increasingly recognition of the genetic influences on common neurological conditions such as Alzheimer's diseases, epilepsy and Parkinson's disease. It could be argued that every disease represents "personalised medicine" for the individual patient.

Cumulatively the rare inherited neurology conditions pose a major health and economic burden globally. The development of therapies for some of these conditions is now a reality with gene silencing and gene editing therapies already available for diseases like Spinal Muscle Atrophy and Familial Amyloid polyneuropathy and many others in phase 3 clinical trials.

These advances have brought both opportunities and challenges.

The opportunities include being able to understand these diseases and develop both preventative and disease modifying therapies. These rare genetic diseases can also provide a window into the understanding of the more common neurological diseases especially diseases of aging.

The major challenges are global access to diagnosis, care and new therapies. Although the costs of genetic diagnosis using next generation sequencing techniques is reducing all the time, the costs of the new therapies is challenging even for the wealthiest countries.

This talk will explore the issues above using some of the inherited neuropathies as examples.