THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS

Guest Lectures

11.03.2025

35 Years
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DR CHRIS TURNER

Dr Chris Turner graduated from Oxford University and undertook his clinical training in Oxford, Hammersmith Hospital, the Whittington, St George's, Chelsea and Westminster and the National Hospital for Neurology and Neurosurgery.

He was awarded a Wellcome Clinical Research training fellowship in 2000 and completed a PhD in the molecular pathogenesis of Huntington's disease.

In 2007 he became a consultant at the National Hospital for Neurology and Neurosurgery at the MRC Centre for Neuromuscular Diseases. Dr Turner runs neuromuscular clinics at Queen Square.

Dr Turner has a specialist interest in myotonic dystrophy and leads the myotonic dystrophy clinic at Queen Square in conjunction with a clinical nurse specialist, Susan MacDonald.
Dr Turner is involved in over 10 clinical trials in myotonic dystrophy and publishes regularly in DM1.

Dr Turner was appointed as Divisional Clinical Director of Queen Square division in June 2018.

Lecture

Advanced therapeutics for Myotonic Dystrophy type 1

Abstract:
Over the past 10 years there has been huge advances in developing advanced therapeutics for DM1. Several of these are in the later phases of trial development and will potentially enable therapeutic delivery to whole populations of DM1 patients over the next 5 years. The necessary infrastructure and expertise will need to be developed to deliver thee treatments.

PROF. HANNS LOCHMULLER

Prof. Hanns Lochmuller is a neurologist and clinical academic specializing in genetic neuromuscular disorders and rare disease. He is Senior Scientist at the Children's Hospital of Eastern Ontario (CHEO) Research Institute. He also holds appointments as Professor of Neurology in the University of Ottawa Faculty of Medicine and the Department of Medicine, Division of Neurology at The Ottawa Hospital. He is affiliated with the University of Ottawa Brain and Mind Research Institute and Department of Cellular and Molecular Medicine and with the Ottawa Centre for Neuromuscular Disease.

Prof. Lochmuller trained as a neurologist in Munich, Germany and in Montreal, Canada. From 2007 to 2017, he held the chair of experimental myology at the Institute of Genetic Medicine at Newcastle University in the UK. He continues to hold a scientific appointment at the Department of Neuropediatrics and Muscle Disorders of the Medical Center – University of Freiburg in Germany and as visiting scientist at the Centro Nacional de Análisis Genómico (CNAG), Barcelona in Spain.

His research interests include molecular therapies of neuromuscular disorders; molecular pathogenesis of muscle and neuromuscular junction disorders; neurogenetics and translational research; data sharing and -omics in neuromuscular and rare diseases; and genomics and systems medicine. In addition to his scientific and clinical research interests, he is internationally active in rare disease science policy and research collaborations. He chaired the Interdisciplinary Scientific Committee of the International Rare Diseases Research Consortium (IRDiRC) and the Executive Committee of the TREAT-NMD Alliance.

He initiated and coordinated the highly successful "RD-Connect" international infrastructure for rare disease data and biosample sharing and analysis, is co-founder and former coordinator of the German muscular dystrophy network (MD-NET), and former scientific coordinator of EuroBioBank, a European (and Canadian) network of biobanks for rare disorders.

PROF. HANNS LOCHMULLER

Prof. Lochmuller's clinical activities focus on clinical research and care of patients with rare neuromuscular disorders, including myotonic dystrophy (DM1), spinal muscular atrophy (SMA), muscular dystrophy and congenital myasthenic syndromes (CMS). He has a strong commitment to working with patients and patient organizations in Canada, as he has with organizations in Europe for many years.

Lecture

Congenital myasthenic syndromes – inherited neuromuscular transmission disorders – from basic research to translation into patient benefit

Abstract:

Congenital myasthenic syndromes (CMS) are a heterogeneous group of rare inherited neuromuscular disorders characterized by fatigable weakness owing to compromised function of the neuromuscular junction. There is substantial clinical heterogeneity among the CMS subtypes, from childhood onset with episodic apnea and intellectual disability, to later onset with limb-girdle distribution of weakness. Severity and course of disease are highly variable, ranging from minor symptoms to progressive disabling weakness. Many patients can be treated with oral "off label" medications including esterase inhibitors, salbutamol and 3,4-DAP. Treatments are targeted towards the underlying molecular defect caused by mutations in 35 different genes. Recent advances in model systems including zebrafish and mice have opened the door to more effective and safe molecular treatments for CMS.

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