

Curriculum Vitae

Professor Michael G Hanna

2024

Professor Michael G Hanna  
MBChB(Hons) BSc(Hons) ECFMG (USA) FRCP(UK) MD FMedSci

***Current positions:***

Director, UCL Queen Square Institute of Neurology, turnover £120m

Professor in Clinical Neurology  
Department of Neuromuscular Diseases  
UCL Queen Square Institute of Neurology

Director, UCL Centre for Neuromuscular Diseases

Director, UCL International Centre for Genomic Medicine in Neuromuscular Diseases

Consultant Neurologist, UCLH National Hospital for Neurology and Neurosurgery  
Queen Square

Honorary Consultant Neurologist, Great Ormond Street Hospital NHS Trust.

Honorary Professor in Clinical Neurology, University of Newcastle-upon-Tyne

Adjunct Professor of Neurology, Department of Neurology, Iowa University, USA

Chairman, Board of Trustees of Muscular Dystrophy UK charity- (turnover £7m)

NIHR Senior Investigator from 2023

***Recent positions***

Divisional Clinical Director, Queen Square Division, UCLH NHS FT (2007-2012)  
turnover £180m

Co-Director, Neurological Diseases Theme  
UCL Partners Academic Health Sciences Centre (2010-2014)

Founding Chairman, British Myology Society (2008-2016)

National Theme lead; NIHR Neuromuscular Translational Research Collaboration (14-16)

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## 1. CV Summary and key achievements - Professor Michael G Hanna

### Leadership Roles

- 1998-present Consultant Neurologist National Hospital, Queen Square
- 1998-2007 Consultant Neurologist Middlesex and UCLH
- 1998-present Consultant in Neurogenetics
- 2007-present Honorary Consultant Neurologist Great Ormond Street
- 2000-present Lead muscle clinician Centre for Neuromuscular Diseases
- 2006-present Professor in Clinical Neurology, UCL Institute of Neurology
- 2003-2007 Associate Clinical Director, Queen Square Division
- 2007-2012 Divisional Clinical Director, Queen Square Division
- 2007-present Director UCL MRC Centre for Neuromuscular Diseases
- 2009-2019 Co-Director UCL Partners Neurological Diseases Theme
- 2011-present Honorary Professor in Clinical Neurology, Newcastle University
- 2012-present Director UCL Queen Square Institute of Neurology
- 2016-present Chairman of the Board, Muscular Dystrophy UK charity
- 2017-2020 MRC Fellowship and training board- full board member
- 2024- Treasurer, Council officer and trustee of Academy Medical Sciences

### Academic Activity: basic discovery science and translational research

- 1995-2024 Published over 395 peer reviewed papers and reviews, over >250 published abstracts, 18 book chapters and 1 book. H-Index 87
- 1998-2024 Obtained over £22 million external basic and translational research funding including MRC, Wellcome Trust plus £8million NHS NCG funding.
- 1998-2024 Supervised 33 clinical and non-clinical PhD research fellows/students and 3 post doctoral research fellows
- 2007-2020 Successfully led joint initial application and renewal, across UCL and Newcastle and direct joint UCL Newcastle MRC Centre for Neuromuscular Diseases->£10m
- 2019-2024 Lead PI on MRC strategic award to establish an international centre for genomic medicine in neuromuscular diseases £3.66m from MRC and £2m host universities

### Editorships and other esteem

- 2003-2015 Deputy Editor of the Journal of Neurology, Neurosurgery and Psychiatry (2600 manuscripts per annum)
- 2003 Goulstonian Lectureship to the Royal College of Physicians London
- 2009 Corresponding member of the American Neurology Association
- 2009 Guarantor of Brain
- 2011 Ian MacDonald Lectureship, Australian & NZ Neurology Association
- 2019 Inaugural JS Chopra Oration, Indian Academy of Neurology

### National Neuromuscular Clinical Services

- 2001-Established the UK diagnostic service for neurological channelopathies. Obtained recurring funding from DoH National Specialist Commissioning Group-NCG

- to provide this service ~£375 000 per annum to UCLH
- 2007-with Newcastle jointly established NCG funded national diagnostic and treatment service for mitochondrial diseases. Recurring funding £1.1m pa to UCLH
- 2008-2016 Founding chairman of the British Myology Society

#### Undergraduate Neurology Teaching

- 1998-2004-Established and ran new undergraduate clinical neurology teaching course in 1998 based for the first time at the National Hospital.
- 2000 Awarded UCL top undergraduate teacher by undergraduate medical students
- 2003 Co-edited 4<sup>th</sup> edition of well known undergraduate text “Tutorials in Differential Diagnosis” sold 12000 copies world-wide, translated into 6 languages.

#### Private Fundraising and Philanthropy

- 2007-2017 raised £1.7m which combined with successful BRC application for £0.5m provided capital for brand new UCL-UCLH translational research centre for neuromuscular diseases opened May 2009 and accommodates a team of over 50 clinical and research staff.
- 2007-£250k Clore Foundation capital for new Neuromuscular centre
- 2007-present £350k Kennedy’s Disease Fund
- 2009-worked with National Brain Appeal to raise £1m for new UCLH NHNN Brain tumour unit
- 2011 £250,000 Chandris Foundation towards Neuromuscular research
- 2018 £234,000 Clore Foundation clinical research fellowship
- 2018 £2m Wolfson Foundation to support new QS IoN build

#### Clinical Management and Neuromuscular Clinical Services

- 2007-2012 Divisional Clinical Director Neurosciences Queen Square Division, managing £143m budget and >1,200 staff, working in and leading a team of 15 clinical and non clinical managers
- 2000 Established the National Hospital virtual Centre for Neuromuscular disease
- 2000-2009 built clinical team now 7 consultants, 4 CNSs, 2 physios
- 2001-2008 built NCG genetic diagnostic team now 5 DNA clinical scientists
- National referral patient base 8-10% growth referrals pa now -4000 pa
- 2009 New physical centre for neuromuscular disease opened by MRC CEO

#### Specialist Clinical Interests

- I have an international profile in clinical neuromuscular muscle wasting neurological diseases; especially mitochondrial diseases and channelopathies and also muscular dystrophy, peripheral neuropathy, motor neuron diseases, myasthenia gravis, and all genetic neuromuscular diseases
- Over 25 years front line consultant general neurologist

## Full Curriculum Vitae

### 2. Personal Details

Michael G. Hanna 04.10.63

### 3. Education/Qualifications

4 A'levels, Lawnswood Comprehensive School, Leeds	Aug	1982
BSc Biochemistry Hons 2.1(Manchester)	July	1986
MB ChB Honours (Manchester)	July	1988
ECFMG I&II (USA)	July	1990
MRCP (UK)	March	1991
MD (Manch)	February	1996
FRCP (UK)	May	2002
FMedSci (UK)		2020

### 4. Current Post

#### Director UCL Institute of Neurology 2012-present

I am Director of UCL Queen Square Institute of Neurology (IoN), which is the largest Neurology research Institute in the UK and one of the largest in the world. It has over 900 academic staff and currently has £258m in active grants. I have driven a new academic strategic plan to increase translation of science into patient benefit. This has included successful award of £20m from the Wolfson Foundation for a new experimental neurology centre at Queen Square resulting in a 500% increase in experimental neurology trials and multiple new industry partnerships and investments. In our successful REF bid 2014 IoN was number 1 in the UK for Neurology research strength. I have supported research embedded education development which now includes 500 MSc and PhD students, with very high student rating/experience scores. I successfully lead the Athena Swan Silver award to the Institute in 2015. I have successfully recruited many senior academics to the Institute including the first Nobel prize winner to join the Institute, Jim Rothman 2014. I have worked closely with UCL partners academic health sciences centre (as Co-Director of neurological disease theme) to develop population level impact especially in stroke and dementia. I recently lead for IoN in capital fundraising programme that has successfully raised £240m to rebuild the main IoN wet lab science building; building work will commence 2019. Under my leadership I want to continue to maximize the opportunities for science to benefit all patients with neurological diseases. I was recently central to the successful award of the MRC UK Dementia Research Institute to UCL at IoN and I lead a successful capital bid to HEFCE for £29m for capital towards new IoN building.

My current consultant position also includes:

- Research - Director UCL MRC Centre for Neuromuscular Diseases-since 2008
- Clinical work and patient care - I am a Consultant Neurologist providing a specialist muscle service including two Nationally commissioned highly specialised clinical services-for patients with mitochondrial disease and channelopathies since 1998 (£1.3m pa NHS funding).

#### 4a. Research Programmes-Discovery, Translational and Global initiative

My research has focused on improved understanding of the molecular genetics basis and molecular mechanisms of neurological diseases caused by mitochondrial dysfunction and by ion channel dysfunction. Many of these diseases particularly affect the neuromuscular system and my clinical specialist interest has focused on developing better services for patients with neuromuscular neurological diseases. Wherever possible, I have aimed to translate my research findings into clinical practice. I successfully established an MRC Centre for translational research in neuromuscular diseases in 2008 which is now a national UK platform and resource that has had major impact in several areas including clinical and non clinical PhD training, new gene discovery, biobanking, patient cohorts and clinical trials in neuromuscular diseases.

Since 2000 I have obtained in excess of ~£22 million pounds sterling in external peer reviewed funding from many funders including MRC, Wellcome and NIH, to support my research programmes into mitochondrial disease and human neurological channelopathies.

In addition, I have been successful in translating my research into nationally commissioned diagnostic service through the competitive NHS national commissioning rounds. Since 2001 I have secured over £12 million pounds sterling in NHS contracts to deliver these services for patients.

##### **Ion Channel Research Programme-Summary and Impact**

Many important neurological diseases are episodic causing patients to experience attacks of unpredictable severe neurological dysfunction separated by periods of apparent normality. The commonest episodic neurological disorders are epilepsy and migraine but their precise molecular pathophysiology is an important unsolved neuroscience challenge. In addition, there are many severe disabling disorders of episodic muscle dysfunction such as episodic total muscle periodic paralysis and intermittent and severe disabling muscle myotonic stiffness.

I have established a collaborative interdisciplinary clinical, genetic and cellular electrophysiological research programme in the Institute of Neurology that has progressed fundamental understanding of the pathophysiology of episodic neurological diseases, resulted in new diagnostic tests speeding diagnosis and which has improved patient outcomes.

We collated the world's largest cohort of over 1400 families with inherited channelopathies and have identified 100's of unique mutations in specific genes responsible for the diseases. The molecular electrophysiological consequences on single ion channel function have been studied using detailed cellular expression techniques allowing a more precise understanding of pathophysiology.

Many key observations have been made that have resulted in improved fundamental knowledge. For example:

- We have shown how genetic dysfunction of muscle sodium, potassium, calcium or chloride channels can relate to specific episodic muscle conditions including periodic paralysis, muscle stiffness syndromes and episodic cardiac arrhythmias.
- We have shown that the genetic architecture of the commonest form of periodic paralysis predicts the presence of an abnormal gating pore current in muscle sodium or calcium channels supporting the presence of a brand new mechanism for disease causation and target.
- We defined previously unrecognized neonatal ion channel diseases including intermittent hypotonia and stridor—knowledge of which has led to improved and safer care of affected patients in the neonatal period.
- We identified mutations in important presynaptic neuronal potassium and calcium channels can lead to episodic ataxia- a disorder characterized by profound disabling attacks of unpredictable unsteadiness. Furthermore, we have shown that such patients are 17 times more likely to develop epilepsy compared to the background population risk.
- We provided evidence that muscle sodium channel dysfunction is a risk factor for sudden infant death syndrome.

This research has had direct patient benefit and has been translated into the only nationally commissioned clinical service for the entire UK. Patients from all over the UK are referred to the Institute of Neurology-National Hospital clinic and now achieve a more rapid and precise genetic diagnosis and appropriate treatment selection.

We have shown that therapy response relates to genotype and this has directed more effective therapy selection with clear positive patient impact. We have defined the exact genetic architecture and devised a national DNA based diagnostic service. Highly specialised electrophysiological techniques that we apply to patients have been developed and we have shown this allows us to predict the likely genotype and direct genetic testing. Drug selection is now based on genotype and this has improved patient outcomes.

Before the development of this clinical service patients with neurological channelopathies were often misdiagnosed as psychogenic disorder or there was an unacceptably long delay in achieving a diagnosis. This service is unique world-wide and now receives large numbers of international referrals. The innovative investigative patient pathway combines clinical assessment and specialized electrophysiological analysis to direct DNA-testing with a linked diagnostic cellular molecular expression service to determine if newly identified mutations are pathogenic. New treatments are being tested in large multicentre NIH and MRC funded treatment trials now in progress in the UCL MRC Centre for Neuromuscular Disease. (All publications relating to channel research and discovery programme are listed in section 16).



### **Mitochondrial Research Programme-Summary and Impact**

I undertook my MD thesis research as an MRC training fellow in 1992 under the supervision of late Professor Anita Harding and have maintained an active funded programme of research into mitochondrial diseases every since with funding from a variety of sources including MRC, Wellcome and Action research.

- I have been particularly interested in the link between mitochondrial DNA mutations and human disease. I have defined a large number of human pathological mtDNA mutations and have elucidated their molecular pathogenesis through various expression systems including human primary muscle cell cultures.
- We established that approximately 70% of human adult mitochondrial disease is caused by primary mtDNA mutations.
- More recently we have used whole exome genetic approaches in the remaining 30% of adult case and have recently been successful in identifying new nuclear genes that encode proteins that are targeted to the mitochondria and control respiratory chain assembly and mitochondrial fission.
- I have translated my mitochondrial research into a nationally commissioned joint national service with colleagues from Newcastle and Oxford. This provides >£1m per annum to Queen Square to provide a comprehensive clinical and genetic diagnostic service for patients with mitochondrial diseases

**Publications:** I have consistently published my ion channel and mitochondrial discovery research in top tier high impact journals including New England Journal of Medicine, Lancet, American Journal of Human Genetics, Human Molecular Genetics, Annals of Neurology, Neurology and Brain (see section 16).

### **The UCL MRC Centre for Neuromuscular Diseases 2008-2018**

Ten years ago I made a decision to add to my discovery science activity by specifically focusing on developing a strategy to lead on tackling the increasing “translational gap” between discovery and patient benefit in neuromuscular diseases. Although we at UCL and other colleagues across the UK had been world leading in discovering new genes and disease mechanisms, we had been poor at finding ways of translating this knowledge into patient diagnostic and/or therapeutic benefit that has real impact.

In 2006 I brought together world-class colleagues from across UCL and the University of Newcastle-upon-Tyne to form a translation research centre specifically focused upon identifying and overcoming important gaps in translation. In 2008 I was successful in leading a bid to the MRC that resulted in an MRC Centre (£3.4m) award to UCL. I lead the a successful renewal and the Centre will continue to 2020. I am the Centre Director ([www.cnmd.ac.uk](http://www.cnmd.ac.uk)). The interdisciplinary Centre formally links the UCL Institutes of Neurology and Child Health with other departments in the UCL Faculty of Life Sciences and with major groups in two separate faculties at Newcastle University. ***The Mission of the Centre is- To translate basic science findings into clinical trials and new treatments for children and adults with disabling neuromuscular diseases.***

The main programmes of research within the centre build on existing funded themes currently attracting in excess of £18m of grant income, are developing new cross cutting collaborations and has facilitated and is now capitalizing on the recruitment of world class

senior academic personnel to UCL. All the main programmes of research impact upon and benefit from the following key areas that are being newly developed in the centre. [The underdevelopment of these key areas is a current “block” to effective UK translational research in neuromuscular disease];

- Developing a range of specific clinical assessment tools to facilitate future clinical trials in neuromuscular disease in the UK
- Establishing a North and South of England neuromuscular clinical trials centre
- Establishing new cutting edge MRI of nerve and muscle disease in animals and humans as a biomarker of disease activity
- Establishing a unique biobank of human neuromuscular patients tissues and cells
- Establishing a network and resource for elucidating the pathogenesis of neuromuscular conditions in mutant mice
- Attracting and training a new generation of basic and clinical neuromuscular scientists to build future “capacity” in the UK

### **The main programmes of research cover major diseases of Muscle and Nerve**

Molecular mechanism in muscular dystrophy [Bushby, Straub, Brown, Duchen, Holden, Muntoni, Sewry], Mitochondrial DNA neuromuscular disease [Turnbull, Chinnery, Duchen, Hanna, Wood], Ion channel neuromuscular disease [Bostock, Hanna, Wood, Koltzenburg, Kullmann], Muscle stem cells [Morgan, Muntoni; Yousry], Genetic neuropathies [Brandner, Fisher, Greensmith, Houlden, Jessen, Reilly], Spinal muscular atrophy [Duchen, Fisher, Greensmith, Muntoni], Generation of neuromuscular disease mutant mice [Brown, Fisher], MRI of nerve and muscles in animals and humans [Koltzenburg, Muntoni, Hanna, Reilly, Yousry]. Trials & outcomes in neuromuscular disease [Goldblatt, Hanna, Muntoni, Reilly, Thompson].

### **Strategy and objectives 2008-2020**

We are building on existing world class basic and clinical scientists at the Institute of Neurology, the Institute of Child Health and across UCL and have recruited three new world class senior clinical and basic research staff to establish an unrivalled critical mass of expertise in the UK.

- We are addressing unresolved aspects of the pathophysiology of common neuromuscular disorders, notably the effects of identified mutations of structural proteins on signalling mechanisms in nerve and muscle
- We are adding value to existing funded basic neuromuscular research themes at UCL by forging new collaborations with animal/human MRI, a ‘bio-bank’ and the UCL outcomes unit
- We are building on existing funded natural history studies and phase I/ II and III clinical trials and develop new clinical assessment methodology to monitor natural history and treatment effects in disabling neuromuscular disease
- We are using a number of different animal models of disorders, many of which have previously been identified by members of our group, in order to design and refine cutting edge therapies
- We are investigating novel therapeutic interventions using preclinical models
- We are add value to existing excellence in education and training of basic and clinical neuromuscular scientists of the future thereby building the UK’s future capacity

- We will take capitalize on the clinical resource at the co-located Neuromuscular centres of Great Ormond Street NHS Trust and National Hospital for Neurology, especially within two Department of Health (National Specialist Commissioning Group) funded centres for children and adults with rare neuromuscular diseases
- We are collaborating widely in the UK and internationally to form patient and scientist networks.

**MRC strategic award to establish an international centre for genomic medicine in neuromuscular diseases; to commence 2019 £3.66m**

I led a recent successful bid to the Medical Research Council which will establish a consortium across five continents to build patient cohorts and define the genetic architecture of neuromuscular diseases across continents.

#### **4b. Clinical Services**

##### **General Neurology Service**

From 1998-2007 I provided a comprehensive general neurology service to the Middlesex Hospital and the Heart Hospital, part of the UCLH NHS-trust. This involved an on-site neurology outpatient clinic and a same day neurology ward liaison service to any ward in the hospital. This liaison service allowed any inpatient at the Middlesex rapid access to a neurological opinion. Acute neurology care pathways were developed. I developed and delivered a new Trust-wide acute Neurology service centered on the new UCH hospital. I now undertake one general neurology clinics at NHNN. I do regular on call for general neurology.

##### **Specialist Clinical Service-The National Hospital Centre for Neuromuscular Disease**

- I have established a centre for neuromuscular disease with colleagues at the National Hospital in order to enhance services to patients with muscle and peripheral nerve diseases. The center now comprises over 60 clinical and research staff. It represents a critical mass of clinical neurologists with expertise in muscle and nerve disease, neurophysiologists, specialist nursing staff and a state of the art computerized myometry system as well as support staff. We moved into new state of the art facilities in 2009 financed by a £2m charity fund raising project which I lead.
- I run the bi-weekly muscle-genetic clinic which has a regional and national referral base for patients with acquired and genetic muscle disease.
- I obtained funding from the Department of Health [National Specialist Commissioning Group] to support the muscle channel and mitochondrial components of this muscle service. This now amounts to £1.4m per annum on a recurring basis. This NCG funding allows me to provide a comprehensive, clinical electrophysiological and genetic service to this group of patients from anywhere in the UK without cost implications to the local referring centre. The NCG services I offer are part of a National consortium for the diagnosis of rare neuromuscular diseases which I established with colleagues in Oxford, Newcastle and London.

- Neurological trainees as well as consultant colleagues from the UK and internationally frequently attend my clinic.
- I run a joint inflammatory muscle disease clinic with Professors Isenberg and Schapira for complex cases six times per year.
- I am an honorary consultant at Great Ormond Street and undertake joint transition neuromuscular clinics with Professor Muntoni.

#### **4c Clinical Management from 2002–2012**

- I was Associate Divisional Clinical Director from 2002 to 2007 and worked closely with Alan Thompson to produce the NHNN clinical service strategy document 2005-2008.
- I was Queen Square Divisional Clinical Director since 2007-12 and in collaboration with the Divisional Manager-Jackie Sullivan I had responsibility for Divisional financial performance and activity, quality, workforce, governance, estates and clinical strategy.
- The management team I lead produced the NHNN strategy document 2009-2014 in conjunction with key partners in the context of UCLP (see appendix).
- I have an intimate knowledge of all clinical and research services at NHNN and strong links UCLH Trust-wide.
- I work closely and was jointly responsible with the National Hospital Divisional manager for all clinical, staffing financial and governance issues in the Hospital. The budget revenue was £143m pa and the workforce 1245 whole time equivalent including 160 consultant staff.
- I am proud of the achievements delivered with the team under my leadership over the past four years including:

Marked increase in all patient activity 60,000 pa 2006/7 130,000pa 2010/11

Progressive improvement in absolute year-end financial position

- Successful delivery of new joint Institute-Hospital initiatives that benefit patient now and produce huge opportunities for experimental research leading to impact in the near-medium and term, three examples:

**2008 Hyper Acute Stroke Unit**-successful application to Health Care for London and delivery of a single unified HASU in the North Central sector now managed by Queen Square. Centralization of all sector consultants into one team and already resulted in a reduction in stroke 28 day in-patient

mortality. High throughput of fully phenotyped patients is a major opportunity for many types of stroke clinical research.

**2009 Neuromuscular Centre-** Successful MRC Centre grant (£3.4m) and successful philanthropic and BRC fund raising (£2m) to open a state of the art translational Neuromuscular Centre at Queen Square with effective genuine partnership links across UCL (Faculty of Life Sciences and Population Sciences-Institute of Child Health) and Newcastle University.

**2010 Brain Tumour Unit-**Successful collaborative philanthropic fundraising and opening of a new dedicated £2m Brain Tumour unit at Queen Square. Key research links to the UCL Cancer Institute and Institute of Neurology.

## 5. Professional History

### **Aug 88-July 89**

#### Preregistration Positions

Professorial House Physician General Medicine and Neurology

Professor D Neary & Professor S Tomlinson, Manchester Royal Infirmary

Professorial House Surgeon

Professor Sir Miles Irving, Hope University Hospital, Manchester

### **Aug 89-Jan 92**

#### Senior House Officer Positions

Newcastle-upon-Tyne central teaching medical rotation.

General medicine & endocrinology. Dr WMG Tunbridge, Newcastle General Hospital

General medicine & gastroenterology. Dr C Record, Royal Victoria Infirmary

Cardiology Professor F Campbell, Freeman Hospital

Chest medicine Professor J Gibson & Professor P Corris, Freeman Hospital

Neurology Dr P Hudgson & Dr J Foster, Newcastle General Hospital

Renal medicine Professor R Wilkinson, Freeman Hospital

Oxford Department of Clinical Neurology

Neurology Professor Newsom-Davis, Drs R Greenhall, D Hilton-Jones, C Fursden-Davis, G Wright, J Oxbury, N Hyman and M Donaghy, Radcliffe Infirmary, Oxford

### **Feb 92-July 92**

#### Registrar/Clinical Lecturer

General medicine & geratology, Nuffield Department of Clinical Medicine, Oxford University

Professor J Grimley-Evans, John Radcliffe Hospital & Radcliffe Infirmary.

### **Aug 92-July 94**

#### MRC Research Fellow

Medical Research Council clinical training fellow,

Mitochondrial DNA in human mitochondrial disease

University Department of Clinical Neurology, Institute of Neurology, Queen Square, London;

Supervisor *the late* Professor Anita E Harding

**Aug 94-April 96**

Registrar in Clinical Neurology

University & NHS Departments of Clinical Neurology, Radcliffe Infirmary, Oxford  
Drs R Greenhall, M Donaghy, D Hilton-Jones, N Hymen, J Oxbury, C Fursden-Davis, G Wright, P Davis, P Matthews & Professor Newsom-Davis.

**May 96-Nov 96**

Registrar in Clinical Neurology

The National Hospital for Neurology and Neurosurgery, Queen Square, London  
Professors Shorvon, Duncan, Fish, Marsden, Frackowiak, Schapira Quinn, Wood and Goadsby

**Nov 96-January 98**

Senior Registrar in Neurology and Clinical Lecturer in Neurogenetics

The Institute of Neurology and the National Hospital for Neurology and Neurosurgery  
Queen Square and St Mary's Hospital, Paddington, London  
Professors Rossor and Wood and Drs Farmer, Thomas and Ball

## 6. Teaching Education Role

### ***Undergraduate medical student clinical neurology teaching course 1998-2007***

- In 1998 I established a new two week medical student neurology programme. Prior to this although special module neurology teaching occurred for those students who selected it (Prof AJ Lees) no formal neurology by teaching by neurologists existed for UCL medical students.
- The course I established initially took place on the Middlesex Hospital site but subsequently I was able to transfer it to the Queen Square site, which had traditionally only taken postgraduate neurology trainees. This proved very successful. I extensively revised this course to make it five weeks long and incorporated all the requirements for the "New Curriculum" for the training of UK doctors.
- The course is summative towards the final MB and therefore involves organizing and coordinating a written exam and an OSCE exam each term. I was a member of the medical school neuroscience undergraduate planning committee. I also acted as an academic advisee for four undergraduate medical students at any one time. This involved regular meetings offering various forms of support throughout their clinical three year course.
- My work in undergraduate teaching was recognized by the award of "Top clinical teacher" in 1999-2000 voted for by the clinical medical students. In 2002 Queen Square voted best undergraduate teaching site –UCL medical students.
- I co-authored the fourth edition of the well known textbook "Tutorials in Differential Diagnosis" now reprinted in several languages and with sales > 10,000

### ***Postgraduate education roles***

- I give lectures on the IoN MSc courses

- The MRC Centre has a major high profile postgraduate education role which I organise with colleagues in the Centre and includes:
  - A monthly invited speaker seminar programme with a dedicated pre lecture session for the PhD students to meet the speaker
  - An annual one week MRC Centre neuromuscular clinical update course
  - An annual MRC Centre UK Neuromuscular translational research scientific conference now in its fourth year and attracting >300 delegates each time. I have rotated this around the UK London, Newcastle and Oxford to date.

## **7. Other Appointments and Affiliations**

### **Deputy Editor, Journal of Neurology, Neurosurgery and Psychiatry 2004-2015**

Involved daily running of Journal and handling 2600 manuscripts per year with the Editor and three associate editors. Progressive rise in Impact factor from 3.1 to 4.791.

### **Membership of Societies-advisory panels**

- Association of British Neurologists
- World Muscle Society
- European Neurological Society
- Centre lead-National Specialist Commissioning Agency funded National muscle channelopathy and mitochondrial service, National Hospital Neurology, UCLH NHS Trust 2002-present
- Oxford University Wellcome Trust Initiative in Channelopathies-co-investigator and collaborator 2003-present
- Co-chair North American Muscle Interest Group member and Scientific Advisor 2003-present
- Founding chairman of the British Myology Society 2008-2018
- Chairman of the Board of Trustees Muscular Dystrophy UK (UK largest NMD charity-£8m annual turnover)
- Trustee National Brain Appeal from 2016- present
- Governor on the board of governors for UCLH NHS Trust

I regularly review papers and books for the following journals: Brain; Human Molecular Genetics; Journal of Neurology; Journal of Neurology, Neurosurgery and Psychiatry; Journal of Medical Genetics; Movement Disorders; European Journal of Neurology; Genomics, Annals of Human Genetics, Annals of Neurology, European Journal of Human Genetics, Journal of Neurological Sciences, Neuroscience, American Journal of Human Genetics, Neuropediatrics, American Journal of Medical Genetics, Journal of Clinical Investigation and Nature Genetics.

Grant reviewer: Wellcome, MRC, Telethon (Italia), Action Research, Wolfson Foundatio

## **8. Prizes, Awards and other Honours**

**Mabel Harriet Florence Smith travel scholarship 1987**

Awarded by Leeds City Council, to best medical student elective, on the basis of a written submission and an oral presentation.

**The Dorothy Clarke Memorial Prize University of Manchester 1998** Awarded to the best medical student in neurology based on an additional final year clinical examination in Neurology

**UCL Top teacher award 2000**, University College London Medical school. Voted best clinical teacher by clinical medical students year 2000

**Goulstonian Lectureship to the Royal College of Physicians 2003**

Awarded to the youngest elected fellow after assessment of achievements by the senior college officers. Lecture delivered Nov 2003 at Royal College of Physicians, Regents Park, London "Neurological Channelopathies: a new field"

**Guarantors of Brain 2009**

Elected Guarantor of Brain 2009

**American Neurology Association 2010**

Elected corresponding member 2010

**Ian MacDonald Lecture 2011**

Invited guest named Lecture to the Association of Neurologists of Australia and New Zealand at the annual Scientific meeting, Hobart Australia, May 2011

**Inaugural JS Chopra Oration 2019**

Invited guest named Lecture to the Indian Academy of Neurology at the annual conference (IANCON), Hyderabad India, October 2019

## 9. Grants

### Previous Grant Funding:

1992-1994	Medical Research Council: Training Fellowship The study of the molecular pathogenesis of primary defects of mtDNA using a human myoblast culture system £60,000 MG Hanna - Supervisor Prof AE Harding
1996-1997	Joint Research Advisory Committee Institute of Neurology: Mechanisms of phenotypic diversity associated with the mitochondrial A3243G point mutation PI Hanna £35,000
1997-1999	Brain Research Trust: Expression studies of human neurological channelopathies Co-I Hanna £90,000
1998-1999	Wellcome Trust: entry level fellowship for medical research fellow Studies of mtDNA in human Neurological disease Hanna sponsor and supervisor for Dr Siddique £57,000
1999-2000	Wellcome Trust: entry level fellowship for medical research fellow



	Molecular genetic studies of human skeletal muscle channelopathies Hanna sponsor and supervisor for Dr N Davis £59,000
1999-2001	Epilepsy Research Foundation: Clinical, Genetic and Expression studies of the voltage gated calcium channel (CACNA1A) in complex human epilepsy phenotypes Hanna Principal Applicant £50,000
1999-2001	Brain Research Trust: Molecular basis of phenotypic heterogeneity associated with mtDNA mutations Hanna principal applicant £55,000
1999-2004	MRC: Mitochondrial cooperative - mitochondria in health and disease Cooperative status Hanna co-principal applicant with Prof JB Clarke, Prof M Duchen, Prof Wood, Prof Rich, Prof Crompton and Prof Moncado-UCL
2000-2003	MRC: Mitochondrial cooperative – component grant Identifying nuclear genes which influence the expression of mtDNA mutations Hanna-principal applicant £180,000
2000-2003	Wellcome Trust: Training fellowship: Molecular genetic and expression studies on muscle chloride channelopathies Hanna sponsor and supervisor for N Davies £120,000
2000-2002	Special Trustees of the UCLH NHS Trust: Clinical, genetic and expression studies of the voltage gated calcium channel CACNA1A in neurological disease Hanna principal applicant £69,000
2001	National Institutes of Health [USA]: Research proposal planning grant - An international trial of treatment in skeletal muscle channelopathies USA PI- Professor RC Griggs and UK Co PI - Hanna US\$35,000
2003-2007	Kennedy's disease research fund grant: Clinical research fellow undertook molecular genetic research into Kennedy's disease Co-PI Hanna £75,000
2003-2005	MRC Cooperative: Epilepsy. Ion channels in epilepsy Component grant DM Kullmann, NW Wood and MG Hanna £375,000
2004-2008	National Institutes of Health [USA]: Clinical Investigations into Neurological Channelopathies "CINCH" UK Centre International multicentre study in neurological channelopathies Funded one clinical research fellow salary per year for 5 years Hanna CoPI £250,000
2003-2007	Wellcome Trust: Integrative Physiology Ion Channel Programme grant to Oxford University OXION. Hanna Co-I £3.4million.
2004-2009	National Institutes of Health [USA]: An international trial of pharmacological intervention in periodic paralysis. USA PI and Coordinator Professor RC Griggs. Only UK centre Co-PI Hanna. US\$3.1million
2004-2005	Guarantors of Brain: Research fellowship for Dr Tracey Graves. Supervisor Hanna. £55,000
2004	UCL Special Trustees: Fast track Award for Post Doc "Nuclear genes and mitochondrial disease". Hanna principal applicant and supervisor £29,000
2005-2007	Medical Research Council: Clinical Research Training Fellowship 'Molecular studies of mouse fetal motor neurons in Kennedy's

	disease' Dr N Niranamathan. Hanna and Greensmith joint supervisors £135,000
2005-2007	Action Research: Clinical Training Fellowship 'Brain calcium channel and neurological disease' Dr T Graves. Hanna sponsor and supervisor £113,000
2005-2008	Medical Research Council: Component Grant 'Nuclear genes and mitochondrial disease' Hanna Co-I with Dr S Rahman. £80,000.
2005-2006	UCLH CRDC: Clinical Training Fellowship 'A molecular study of renal dysfunction in human mitochondrial disease' Dr Andrew Hall. Supervisors Hanna, Prof Unwin and Prof Duchen. £75,000
2007-2009	Wellcome Trust: Training fellowship 'The effect of alterations in the P/Q-type calcium channel in ataxia' Dr Sanjeev Rajakulendran. Supervisor Hanna. £111,812
2008-2011	Muscular Dystrophy Campaign: Joint Centre Grant. PI Muntoni Co-PI Hanna and Reilly £450,000
2008-2009	UK Brain Research Trust: Investigation of human neurological ion channel disorders PI Hanna £26,091
2008-2009	British Medical Association: Vera Down Award, Project Grant: Investigation of human neurological ion channels PI Hanna £50,000
2008-2009	Charities Aid Foundation: Patrick Berthoud Clinical Research Fellowship to Dr S Tomlinson 'Investigation of human neurological ion nerve excitability' Supervisor MG Hanna £115,000
2008-2010	Action Medical Research: Project Grant 'Investigation of human neurological ion nerve excitability testing protocols' Co-PI-MG Hanna with DM Kullmann £150,000
2008-2010	Senexis (SME): Project Grant 'Inclusion body Myositis UCL-Senexis collaboration' Co-PI MG Hanna with David Satelle and Linda Greensmith £91,149
2008 - 2009	British Medical Association: Vera Down Fellowship Clinical and electrophysiological studies in muscle channelopathies Supervisor Hanna £50,000
2008-2013	OXION-Wellcome: Integrative Physiology Ion Channel programme grant to Oxford University. Hanna Co-Investigator £4.4 million- Programme renewal.
2009-2011	Medical Research Council: Mitochondrial Disease Patient Cohort PI Turnbull Co-PI Hanna Chinnery McFarland £969,400
2009- 2010	Medical Research Council: Technology Project Grant, 'New therapeutic approaches to IBM Co-applicant with Linda Greensmith PI David Satelle £154,658
2009	Rare Diseases Research Network-NIH: Project grant, 'Genetic studies on voltage sensors in periodic paralysis' PI Hanna US\$89,000
2010 - 2014	Medical Research Council: Clinical Research Training Fellowship 'Whole exome analysis in muscle channelopathies' Fellow Dr Dipa Rayan Supervisor MG Hanna £180,627

2010 – 2011	Arthritis Research Campaign: 'Arimoclomol trial to upregulate Heat Shock proteins in Inclusion Body Myositis' PI Hanna £133,413
2010-2011	UCLH Fast Track Grant: 'Gene expression profiles in muscle with deleted mitochondrial DNA ' PI Hanna £32,000
2010-2012	National Institute of Health Research: 'Clinical trial of carbonic anhydrase inhibitors in periodic paralysis' Clinical Local Research Network Contingency Funding PI Hanna £67,540
2010 - 2012	NIHR: Clinical Research Network contingency funding Arimoclomol for Sporadic Inclusion Body Myositis (IBM) PI Hanna £66,641
2010-2013	UCL Impact Studentship with private donor: Heat shock protein upregulation therapy in the Kennedys Disease mouse model. PhD studentship Anna Gray Primary Supervisor Greensmith, Secondary supervisor Hanna £60,372
2010-2013	UCL Impact Phd Studentship: 'Mitochondrial dysfunction, impaired axonal transport and inherited neuropathy' Ellen Cottenie. Joint Primary Supervisor Hanna and MM Reilly £60,372
2010-2014	UCL Impact PhD Studentship with National Hospital Development Foundation: 'Mechanisms of phenotype diversity in muscle channelopathies' Siobhan Durran Primary supervisor Hanna, Secondary supervisor Houlden £60,372
2011 – 2012	UCL/UCLH Comprehensive Biomedical Research Centre: 'Regulation of mitochondrial proliferation in mitochondrial disease' Co-PI with Michael Duchen £46,785
2011 – 2014	Medical Research Council: Project Grant 'Whole exome analysis in families with neuromuscular diseases' PI Houlden Co-PI Hanna & Reilly £320,000
2011 – 2016	Wellcome Trust: Equipment Grant 'Solid Next Generation Sequencing platform ' PI Houlden Co-PI Hanna, Reilly, Wood, Hardy £661,363
2011-2016	UCL/UCLH CBRC: Capital Bid 'Next generation Sequencing bioinformatics platform' PI Houlden Co-PI Hanna , Wood, Hardy, Reilly £339,000
2011–2015	Muscular Dystrophy Campaign: four year PhD studentship 'Whole exome analysis and molecular expression studies in periodic paralysis' Alice Gardiner Primary Supervisor Hanna Secondary Supervisor Houlden £112,000
2012-2017	FP7 Neuromics consortium: Work package - Whole exome analysis and molecular expression in muscle channelopathies PI Hanna £186,000
2012-2017	European Union: FP7 Neuromics consortium: Work package - Whole exome analysis and molecular expression in muscle channelopathies £186,000 PI Hanna
2013-2015	NIHR Rare Disease Translational Research Collaboration – IBM PI Hanna £250,000
2013-2016	Action Medical Research: Mitochondrial quality control pathways as

	therapeutic targets in genetic mitochondrial disease PI Duchen Co-PI Hanna £192,243
2013 – 2017	Medical Research Council: Project Grant ‘Whole exome analysis in Inclusion body myositis’ PI-Houlden Co-PI Hanna £420,000
2013 – 2017	UCLH BRC: Fast Track Grant ‘A randomized, double-blind, placebo controlled phase IIa experimental pilot trial assessing efficacy of a single dose or repurposed bumetanide in genetically defined hypokalaemic periodic paralysis assessed using the electrophysiological McManus protocol’ Co-I Hanna £40,000
2014-2016	UCL Impact PhD Studentship with National Hospital Development Foundation: ‘Aberrant cellular calcium handling and muscle degeneration in sodium channel disease’ Neta Amior Primary supervisor Duchen secondary supervisor Hanna £60,372
2014-2017	NIHR Rare Disease Translational Research Collaboration: Postdoctoral Clinical Fellowship – IBM – Dr Pedro Machado PI Hanna £401,333
2014-2017	NIHR Rare Disease Translational Research Collaboration Postdoctoral Clinical Fellowship – Channelopathies – Dr Emma Matthews PI Hanna £363,060
2014-2017	Medical Research Council: Project Grant ‘Periodic paralysis: from molecules to mice’ PI Hanna £464,146
2014-2017	National Institutes of Health: Clinical Investigation in Channelopathies \$1.4m PI Griggs Co-PI Hanna (US\$385,000 to Hanna)
2015-2017	Novartis trial: Arimoclomol for Inclusion Body Myositis PI Hanna £345,324
2013 - 2017	GOS NIHR BRC: Matched funding for MRC Centre for Neuromuscular Diseases Hanna PI £282,714
2013 – 2018	UCLH NIHR BRC: Matched funding for MRC Centre for Neuromuscular Diseases. Hanna PI £493,106
2017-2018	The Lily Foundation: Lily-Stoneygate Research Awards Programme – treatments for mitochondrial disease Hanna PI £83,686
2015-2018	The Lily Foundation: Clinical Research Fellowship Dr Olivia Poole £124,000
2015-2018	MRC: Clinical Training Fellowship Dr Karen Suetterlin £185,000
2016-2019	MRC: Clinical Training Fellowship Dr Helen Devine Hanna Sponsor £176,000
2017-2019	Muscular Dystrophy UK: An experimental medicine study to assess the safety and efficacy of 2-deoxyglucose in patients with m.3243G mutant mitochondrial DNA Hanna PI £146,520

2018-2020	Wellcome Trust: Clinical Research Career Development Fellowship 'Skeletal muscle channelopathies: severe infantile phenotypes and sudden infant death syndrome' Dr Emma Matthews Hanna Sponsor £283,711
2019 – 2020	National Brain Appeal: Neuromuscular Clinical Trial Coordinator PI Hanna £56,294
2016-2020	Medical Research Council: MYOPROSP Consortium PI Hanna £66,301
2017-2021	UCLH/NIHR BRC: Neuromuscular Disease Theme Hanna PI £398,514
2019 – 2021	Orphazyme: MRI sub-study Co-I Hanna £453,251
2019 – 2021	Charities Aid Foundation: Studying a rare mitochondrial disease to better understand a common eye disease Co-I Hanna £200,000
2018-2022	European Union: SOLVE-RD Co-PI Hanna €20,000
2018-2022	FDA USA: Phase II/III Study of Arimoclomol in Inclusion Body Myositis (IBM) PI Hanna USD\$214,235
2020 – 2022	UCLH/NIHR BRC: Neuromuscular Disease Theme 3 Hanna PI £257,408

**Current grant income and competitive NHS funding:**

2001 – ongoing	National Specialist Commissioning Agency (NCG) / Highly Specialised Service (HSS): Specialised diagnostic and advisory Service for muscle channel diseases. £466,706 per annum (Total £4,667,080 to date) Clinical Lead MG Hanna
2006 – ongoing	National Specialist Commissioning Agency (NCG) / Highly Specialised Service (HSS); Specialised Mitochondrial Disease Service. £1,050,589 to Queen Square per annum (Total £5,150,000 to date) Clinical lead MG Hanna
2019-2024	Medical Research Council: MRC Strategic Award to establish an International Centre for Genomic Medicine in Neuromuscular Diseases PI and Centre Director MG Hanna £3,139,610
2019-2024	UCL/UCLH BRC; matched funding for MRC Strategic Award to establish an International Centre for Genomic Medicine in Neuromuscular Diseases PI and Centre Director MG Hanna £1,000,000
2008 – 2020	Medical Research Council Centre Grant: MRC Centre for Neuromuscular Diseases renewed Total >£6M £3,071,144 MRC plus £3,321,456 joint BRC and host support. PI and Centre Director MG Hanna
2015-2021	Wellcome Trust Strategic Award: Synaptopathies: genetics, biophysics and circuit mechanisms of paroxysmal neurological disorders PI Kullmann Co-PI Hanna £4,194,451

2018-2024	The Clore Duffield Foundation: Clinical Research Fellowship Dr Chiara Pizzamiglio Hanna PI £234,000
2018-2023	European Union: SOLVE-RD Co-PI Hanna €20,000
2019 – 2024	National Brain Appeal: Neuromuscular Research Manager Hanna PI £200,312
2019 – 2023	UCLH: Clinical Research Fellow Dr Vino Vivekanandam Hanna PI £275,000
2019-2024	Medical Research Council: Clinician Scientist Fellowship; Lipid metabolism in mitochondrial diseases, Dr R Pitceathly Hanna Sponsor £1,001,993
2020 – 2024	J P Moulton Charitable Foundation: First head-to-head trial in non-dystrophic myotonias. PI Hanna £50,877
2022 – 2028	Medical Research Council: MitoCluster Consortium: An Integrated Phenotyping And Mouse Model Generation Platform For Mitochondrial Disease And Dysfunction Hanna PI Pitceathly Co-I £2,933,088 (£550,500 to UCL)
2022 – 2025	Medical Research Council: TransNAT: Transforming delivery, safety, and efficacy of nucleic acid therapeutics; from intracellular uptake to targeting brain, heart, and muscle Hanna PI £8,000,000 (£1,777,000 to UCL)
2022 – 2023	Medical Research Council: GCRF and Newton Consolidated Impact Funding for International Centre for Genomic Medicine in Neuromuscular Disease Hanna PI £115,716
2022 – 2024	UCLH/NIHR BRC: Centre for Neuromuscular Disease (BRC4) Hanna PI £210,663
2023 – 2027	NIHR Senior Investigator Award Hanna PI £80,000

## 10. Selected Invited National and International Lectures

### 1995

1. April 1995: Oxford University Department of biochemistry invited seminars  
*“New mitochondria DNA mutations and myoblast studies on their molecular pathogenesis”*
2. Sept 1995: Oxford University, Nuffield Department of Pathology, invited seminar  
*“The emerging role of mitochondria in human disease”*

### 1997

3. April 1997: Department of Clinical Neurology, University of Queensland, Brisbane, Australia  
*“Channelopathies: A new name for old diseases”*
4. May 1997: Institute of Neurology short course in Neurogenetics  
*“Channelopathies”*
5. Nov 1997: Recent advances in Epilepsy, Glaxo Wellcome, Paris

*"Modern Genetics of Epilepsy"*

6. Nov 1997: Institute of Child Health, UCL- research seminar  
*"Ion channel disorders of skeletal muscle"*
7. Dec 1997: UCL centre for human genetics annual meeting  
*"Ion channels and epilepsy"*

**1998**

8. Jan 1998: Department of Neurology, Kings College Hospital research seminar  
*"Mitochondrial genetics: focus on myopathies"*
9. March 1998: Glaxo Wellcome centre for neuromuscular disease annual meeting, Liverpool  
*"Recent advances in the genetics of neuromuscular diseases"*
10. March 1998: Annual Neuromuscular Symposium, Walton Centre for Neurology, Liverpool  
*"Skeletal muscle channelopathies"*
11. Sept 1998: Annual meeting of the European Federation of Neurology, Seville, Spain, Scientific symposium  
*"Channelopathies: a new field"*
12. Sept 1998: Association of British Neurologists autumn meeting, scientific symposium  
*"Ion channels and neurological disease"*
13. Oct 1998: International league against epilepsy, Annual Scientific Meeting, Oxford  
*"Ion channels and paroxysmal disorders"*
14. Oct 1998: Oxford clinical genetics seminars  
*"Developing a skeletal muscle ion channel DNA diagnostic service"*
15. Nov 1998: Department of Neurology, Leeds University  
*"Skeletal muscle ion channel disorders"*

**1999**

16. Feb 1999: Annual Salpetriere Queen Square Neurology meeting, Paris  
*"New mutational mechanisms in mitochondrial diseases"*
17. March 1999: Dept Neurology, Addenbrooke's Hospital, Cambridge  
*"Neurological channelopathies-the future"*
18. March 1999: Neurology for Neuroscientists, Magdalen College Oxford  
*"Ion channels and Neurological Disease"*

19. April 1999: Annual Clinical Genetics Day, Institute of Child Health, Great Ormond Street *"Mitochondrial Deafness"*
20. April 1999: International League against Epilepsy, British branch, annual meeting, Birmingham, *Ion Channels and Epilepsy-the future"*
21. June 1999: Muscle Course, Hammersmith Hospital, London  
*"Molecular genetics of inherited myotonic disorders"*
22. June 1999: Dept Neurology, Western General Infirmary, Edinburgh  
*"Neurological Channelopathies"*

## **2000**

23. Jan 2000: British Paediatric Neurologists Association annual meeting, Glasgow  
*"Ion channel dysfunction-a common problem in epilepsy"*
24. Jan 2000: Institute of Neurology-UCL-Annual Glaxo Lecture  
*"Ion channels and neurological disease"*
25. March 2000: University of Sheffield, Dept of Neurology  
*"Neurological channelopathies"*
26. April 2000: National neurology meeting Glaxo Wellcome-Stratford  
*"Molecular mechanisms in paroxysmal neurological disease"*
27. May 2000: University of Birmingham, Dept of Neurology  
*"Ion channels and Neurological diseases"*
28. June 2000: University of Birmingham, Dept Clinical Genetics  
*"The genetic ataxias"*
29. Sept 2000: Salpetriere Hospital, Dept Neurology, Paris  
*"Molecular mechanisms in mitochondrial disease"*
30. October 2000: University of Bologna, Dept of Neurology, Italy  
*"Brain potassium channel dysfunction and neurological disease"*

## **2001**

31. Feb 2001: Institute of Child Health, Dept metabolic medicine, UCL  
*"Paediatric channelopathies"*
32. April 2001: Advanced Neuromuscular course, Hammersmith Hospital  
*"Skeletal muscle channelopathies"*
33. July 2001: University of Sheffield, Channelopathies 2001



*"Human neurological disease and ion channel dysfunction"*

34. Oct 2001: Dept Clinical Neurology, University of Oxford  
*"Molecular Mechanisms in neuronal calcium channelopathies"*

## **2002**

35. April 2002: University of Milan, Dept Neurology  
*"Calcium channels and epilepsy"*
36. July 2002, International Congress on Neuromuscular disease, Vancouver Canada workshop-  
*"Painful muscle channelopathies"*
37. September 2002, Birmingham Heartlands Hospital NSCAG meeting  
*"The National diagnostic service for muscle channelopathies"*
38. October 2002, Atkinson's Morley's Hospital, Dept of Neurology,  
*"An Update on Brain Channelopathies"*

## **2003**

39. March 2003, Royal Society Medicine, London  
*"Episodic Ataxias-mechanisms in human cerebellar channelopathies"*
40. March 2003 University of Oxford Wellcome Trust ion channel initiative  
*"Human brain channelopathies"*
41. November 2003 Bath Advanced Neurology course  
*"Neurological channelopathies"*
42. November 2003 Goulstonian Lectureship  
Royal College of Physicians, London  
*"Neurological channelopathies"*
43. March 2003 UCL annual rheumatology course  
*"Inflammatory myopathies"*
44. May 2003 Dept Neurology University Nottingham  
*"Muscle channelopathies"*

## **2004**

45. June 2004 Dept Neurology, University of Bonn  
*"Nuclear Mitochondrial disease"*
46. September 2004 EFNS Paris  
Invited to organize workshop

*“Neurological channelopathies”*

47. October 2004 NIH-CINCH-grant collaborators’ symposium  
University of Rochester Medical School, Rochester, New York  
*“Molecular genetic basis of Periodic Paralysis in the UK”*

**2005**

48. April 2005 Dept Neurology, University of Bristol  
*“Neurological channelopathies”*
49. April 2005, Dept of Neurology, University of Newcastle  
*“Brain paroxysmal disorders”*
50. May 2005, Dept Paediatrics, Alder Hay Hospital, Liverpool  
*“Paediatric channelopathies”*
51. May 2005 Short course on Muscle Disease, Institute of Neurology, London  
*“Inflammatory myopathies”*

**2006**

52. Feb 2006 NIH-CINCH collaborators’ symposium UCLA-SanDiego USA  
*“Molecular mechanisms in Episodic ataxia”*
53. Jan 2006-Irish National Annual Neuromuscular Symposium-Cork, Ireland  
Guest Lecture *“Human skeletal muscle channelopathies - mechanisms and treatment”*
54. June 2006-Dept of Neurology, St George’s Hospital, London  
*“Neuronal channelopathies”*
55. April 2004 Neurology for Neuroscientists, Guarantors of Brain-Oxford  
*Neurological channelopathies*
56. Sept 2006 - International Society of Neurophysiology annual conference, Edinburgh  
Invited lecture - *“Genetic neurological channelopathies”*  
Workshop organiser: *“Channelopathies”*

**2007**

57. January - Oxford Grand rounds  
*“Neurological channelopathies”*
58. July-University of Kansas USA invited seminar  
*“Molecular mechanisms in myotonia”*

**2008**

59. September - North American Muscle Study Group, Rochester, NY, USA  
*"Muscle Channelopathies"*

**2010**

60. June - Centre Hospitalier Universitaire Vaudois, Lausanne, France  
Clinical Neurosciences Grand Round – *"muscle channelopathies"*
61. September - North American Muscle Study Group, Rochester, NY, USA  
*"European perspective for recruitment"*
62. October – Centre for Neuroscience, Department of Medicine, Imperial College  
London *"Muscle channelopathies"*

**2011**

63. March – Royal Society of Medicine Muscle Symposium, London  
*"Acquired muscle diseases"*
64. May – Cardiff Neurosciences Centre, University of Cardiff  
*"Muscle channelopathies"*
65. May – Australian and New Zealand Association of Neurologists Annual Scientific  
Meeting, Hobart, Australia  
W Ian McDonald Lecture *"Translational Research in Muscle Channelopathies -  
Genetics, Disease Mechanisms and Treatment Trials"*
66. May – Australian and New Zealand Association of Neurologists Annual Scientific  
Meeting, Hobart, Australia  
Muscle symposium: *"Neuronal Channel Dysfunction and Episodic Ataxia - An  
Oxford-Australia-London Collaboration"*
67. Twenty-first meeting of the European Neurological Society, Lisbon, Portugal  
*"How to evaluate the patient with a suspected channelopathy"* & Chair, Muscle  
Basic Course
68. July 11 - University of Newcastle Medical School, Department of Neurology  
Grand Round-*"Neurological channelopathies"*.

**2012**

69-76 tbc

**2013**

77. October – Association of British Neurologists, Annual Meeting, Royal College of  
Physicians, London

*“Inclusion body myositis: bench to bedside”*

**2014**

78. Newcastle Institute of Genetic Medicine July 2014  
*“Paroxysmal channelopathies”*

**2015**

79. Neurology 2015 Institute of Neurology  
*“Masterclass in muscle diseases”*
80. Milan Neurology Grand round  
*“Channelopathies from genes to treatments”*

**2016**

81. October - World Muscle Society invited Plenary Lecture Granada, Spain  
*“Developing new treatments in IBM”*

**2017**

82. March - UK Kennedy’s Disease Day, Derby  
*‘Introduction to Kennedy’s Disease’*
83. March – *“ Periodic Paralysis”* Danish Muscular Dystrophy Association, Copenhagen
84. October - Oslo University Hospital, Norway  
*‘The periodic paralyses – an overview’, and ‘The periodic paralyses – treatment and recent research.*

**2018**

85. April - 4th Annual Neuromuscular Disorders Seminar, Houston  
*‘Update on genetic testing in Neuromuscular Diseases’*
86. May – ABN annual meeting, Birmingham  
*‘Common muscle conditions - clinical-genetic diagnosis and management’*
87. May – Update in Neuromuscular Disorders, Queen Square  
*‘Muscle channelopathies update’*

**2019**

88. October - 27th Annual Conference of the Indian Academy of Neurology, Hyderabad  
Inaugural JS Chopra Oration - *‘From understanding molecular mechanisms to delivering treatments in neuromuscular diseases’*

## 2021

89. January - 25th Foundation Day Lecture, DBT-Centre for DNA Fingerprinting and Diagnostics, Hyderabad  
'Genomic Medicine in Neuromuscular Diseases: from Molecular Genetics to Diagnosis and Treatment'

## 11. Academic Supervision and Muscle Disease Clinical Attachments

All primary supervisor unless otherwise indicated.

### 1997-2000

Ms Louise Eunson BSc

3 years funding BRT and ERF (1997-2000)

"Molecular genetic studies on brain channelopathies"

*Queen Square essay prize 2000 [the enigma of paroxysmal disorders in neurology-the role of ion channels]"*

Graduated PhD August 2001. Currently Genetics Lecturer in Biology University of Colchester"

### 1999-2000

Dr A Siddique 1999-2000 medical research fellow

Wellcome entry level fellowship-mitochondrial disease 1999-2000

Transferred to MRC funded "Genetics of epilepsy project" [Prof Wood]

Currently locum consultant Neurologist Kings College.

### 1998-2001

Dr T Pulkes MD

3 years funding BRT 1998-2001

"Molecular mechanisms in mitochondrial disease"

PhD graduated University of London 2004

Currently Consultant Neurologist and associate Chair neurology, Bangkok, Thailand

### 1999-2001

Ms Ruth Rea BSc

3 years funding Wellcome Trust

Co-supervised with Prof Kullmann

"Molecular expression in neurological channelopathies"

Graduated PhD University of London 2003

Currently Post-doc Stanford USA.

### 1999-2002

Dr NP Davies MBBS

Wellcome research training fellow commenced 1999 for 4 years [PhD]

"Skeletal muscle channelopathies"

Currently Consultant Neurologist, Birmingham.

**2001-2004**

Dr D Liolitsa PhD

Postdoctoral research assistant commenced 2001 for 4 years

Funding MRC coop component grant

“Nuclear genes and mitochondrial disease”

Danae became fascinated by Neurology and mitochondrial diseases and transferred to medicine UCL and graduated in medicine 2011.

**2002-2004**

Dr P Imbrici PhD

Post doctoral research assistant

Special trustees grant commence 2002 2 years

“Functional expression studies of calcium channel mutations causing human neurological disease”

Currently post-doc in ion channel research Rome.

**2004-2007**

Dr Tracey Graves medical research fellow 2004-2007

Brain Research Trust-Calcium channelopathies

NIH –USA-CINCH Fellow

Action Research Clinical training fellowship

Registered for PhD-UCL

Graduated PhD 2010

Currently SpR Neurology Oxford

**2004-2007**

Dr Doreen Fialho medical research fellow 2004-2007

NIH –USA-Muscle channelopathies

NSCAG Molecular expression scientist grant

Registered for PhD-UCL

Graduated PhD 2010

Currently Consultant Neurophysiologist, Queen Square.

**2004-2007**

Dr Niranjanan Nirmalanathan

Medical research fellow 2004-2007

Kennedy’s research donation-Molecular aspects of Kennedy’s

Medical Research Council Clinical Training Fellowship 2005-2007

Registered for PhD-UCL

Graduated PhD 2011

Currently SpR pan-Thames neurology rotation.

**2004-2006**

Dr Charungthai Dejthevaporn 2004-2006

Neurology National Scholarship Thailand

Clinical studies on the natural history of mitochondrial disease

Currently Consultant Neurophysiologist, Bangkok Thailand.

**2006–2009**

BMA Vera Down Fellowship and Patrick Berthould Fellowship  
Clinical Research Fellow  
Dr Susan Tomlinson  
In vivo electrophysiology in episodic ataxia  
Registered PhD University of Sydney  
Graduated PhD 2013  
Currently staff Consultant Neurologist, King Alfred's Hospital, Sydney.

**2006-2010**

NIH CINCH Clinical Research Fellow  
& Wellcome Trust clinical training fellow  
Dr Sanjeev Rajakulendran  
Registered PhD UCL 2007  
Graduated Phd 2011  
Currently neurology SpR pan-Thames neurology rotation

**2006-2010**

NIH-CINCH and NIH RDCRN Clinical Research Fellow  
Dr Emma Matthews  
Molecular genetic studies in muscle channelopathies  
Registered PhD 2007  
Graduated PhD 2014  
Currently NIHR Senior Clinical Research Fellow, MRC Centre for Neuromuscular Diseases,  
and Honorary Consultant Neurologist, NHNN

**2008-2011**

MRC Centre for Neuromuscular Diseases Clinical Research Fellow  
Dr Adrian Miller  
Co-supervised with Linda Greensmith  
Graduated PhD 2012  
"Effect of heat shock protein upregulation on a cell model of IBM"  
Currently neurology SpR on pan-Thames neurology rotation

**2009-2010**

MRC Centre for Neuromuscular Diseases Visiting Research Fellow  
Funding Sicilian Research Fellow travel grant  
Dr Simona Portaro- clinical fellow  
"Analysis of the relationship between genotype and treatment response in periodic  
paralysis"  
Currently Consultant Neurologist, Sicily

**2008-2012**

NCG Clinical Research Fellow  
Dr James Burge  
Graduated PhD 2013  
"Developing an expression system to study chloride channel genetic variation in muscle  
disease"

**2009-2013**

MRC Mitochondrial Cohort Clinical Research Fellow

Dr Robert Pitceathly

“Establishing a UK mitochondrial cohort”

“Next generation sequencing to identify new nuclear mitochondrial disease genes”.

Graduated PhD 2014

Currently Neurology SpR on pan-Thames neurology rotation

**2009–2014**

MRC Clinical training Research Fellowship

Dr Dipa Raja Rayan

Graduated PhD 2016

“Whole exome analysis to identify new genes in muscle channelopathies”

**2009–2014**

MRC Centre for Neuromuscular Diseases Clinical Research Fellow

Dr Jasper Morrow

Graduated PhD 2017

Jointly supervised with Mary Reilly Tarek Yousry

“Developing MRI as an outcome measure in neuromuscular disease”

**2010-2013**

Action Rheumatology Clinical Research Fellow

Dr Pedro Machado (Portuguese trainee)

Graduated PhD 2013

“An experimental medicine study into heat shock protein upregulation in patients with Inclusion body myositis”

Currently NIHR Senior Clinical Research Fellow, MRC Centre for Neuromuscular Diseases, and Honorary Consultant Neurologist, NHNN

**2011-2014**

MRC Centre for Neuromuscular Diseases Clinical Research Fellow Joint with MRCV mitochondrial Biology unit Cambridge

Dr Alejandro Horga (Spanish trainee)

Graduated PhD UCL

Jointly led project Hanna Holt (MRC MBU) and Walker (MRC MBU)

“Genetic and cellular studies of Mitochondrial dysfunction and axonal neuropathy”

**2011–2012**

MRC Centre for Neuromuscular Diseases Visiting Research Fellow

Dr Andrea Cortese (Italian visiting fellow)

“Establishing a cohort to study the natural history of IBM”

**2011-2015**

UCL Impact Studentship with National Hospital Development Foundation

Aberrant cellular calcium handling and muscle degeneration in sodium channel



disease.

£60,372 PhD studentship – Neta Amior

Primary supervisor Duchen secondary supervisor MG Hanna

**2011-2014**

UCL Impact Studentship with National Hospital Development Foundation

Mechanisms of phenotype diversity in muscle channelopathies

£60,372 PhD studentship-Siobhan Durran

Primary Supervisor Hanna Secondary supervisor Houlden

**2011-2013**

UCL Impact Studentship

Mitochondrial dysfunction, impaired axonal transport and inherited neuropathy

£60,372 PhD studentship – Ellen Cottenie

Joint Primary Supervisor MG Hanna and MM Reilly

**2011-2013**

UCL Impact Studentship with private donor

Heat shock protein upregulation therapy in the Kennedys Disease mouse model

£60,372 PhD studentship Anna Gray

Primary Supervisor Linda Greensmith Secondary supervisor MG Hanna

**2011-2014**

Muscular Dystrophy Campaign four year prize studentship

Whole exome analysis and molecular expression studies in periodic paralysis

£112,000 PA PhD studentship Alice Gardiner

Primary Supervisor MG Hanna Secondary Supervisor H Houlden

**2013-2017**

MRC Centre Non-Clinical PhD Student – Charlotte Spicer

Investigating the effects of pharmacological up-regulation of the heat shock response in models of inclusion body myopathy

Primary Supervisor L Greensmith Secondary Supervisor MG Hanna

**2013-2017**

MRC Centre Non-Clinical PhD Student – Louise King

Mitophagy deficiencies in mitochondrial DNA disease

Primary Supervisor H Plun-Favreau Secondary Supervisors MG Hanna, M Sweeney

**2013-2017**

MRC Centre Non-Clinical PhD Student – Andreea Manole

Genetic and Functional Investigation of an Inherited Neuropathy and a Channelopathy

Primary Supervisor H Houlden Secondary Supervisor D Kullmann, MG Hanna

**2013- 2019 (paused twice for maternity leave)**

MRC Clinical Training Fellow - Helen Devine

The Pathogenesis of Spinal Bulbar Muscular Atrophy

Graduated PhD

Primary Supervisor MG Hanna Secondary Supervisors L Greensmith, R Patani

**2013- 2019 (paused twice for maternity leave)**

MRC Clinical Training Fellow – Karen Suetterlin

Graduated PhD

A Molecular Pathophysiological Study of the Skeletal Muscle Channelopathies

Primary Supervisor MG Hanna Secondary Supervisors E Matthews, R Mannikko

**2015-2018**

Lily Foundation Clinical Research Fellow

Dr Olivia Poole

Mitochondrial disease: clinical studies and molecular mechanisms

Graduated PhD

Primary Supervisor MH Hanna, Secondary Supervisor R Pitceathly

**2016-2019**

MRC Centre for Neuromuscular Diseases Clinical Research Fellow

Dr Enrico Bugiardini

Clinical, functional and genetic characterization of mitochondrial diseases

Graduated PhD

Primary Supervisor MG Hanna Secondary Supervisor H Houlden

**2019 - 2023**

Centre for Neuromuscular Diseases Clinical Research Fellow

Dr Vinoji Vivekanandam

Defining phenotypes and treatment responses in skeletal muscle channelopathies.

Primary Supervisor Hanna MG Secondary Supervisor Jayaseelan D

**2020 – 2023**

Clore Duffield Foundation Clinical Research Fellow

Dr Chiara Pizzamiglio

Clinical outcomes, Biomarkers, and Genetic Studies in Primary Mitochondrial Diseases

Primary supervisor Pitceathly R Secondary Supervisors Vandrovцова J, Hanna M

**2019 – 2023**

Dr William Macken

**Clinical Research attachments to my specialist muscle clinic**

The colleagues listed below received four to six month senior SpR neuromuscular training attachments with in my muscle service. They have all now been appointed to senior genetic/muscle consultant posts or are senior trainees as indicated.

**2001**

Prof Patrick Chinnery, now Professor of Neurology, University of Cambridge

**2002**

Dr Kevin Talbot, now Neuromuscular Consultant neurologist Oxford

**2003**

Dr Paul Maddison, now Neuromuscular Consultant Neurologist, Nottingham,

**2004**

Dr John Cole, now Muscle Disease - Consultant Neurologist Manchester

Dr Charlotte Brierley, now Muscle Disease - Consultant Neurologist Cambridge

**2005**

Dr Veronica Tan, now Consultant Neurophysiologist London

Dr Aisling Ryan, now Consultant Neurologist, Cork, Ireland

**2006**

Dr Singh- now consultant genetics Manchester

Dr Andrew Smith-now Consultant Neurology Bristol

**2007**

Dr Andrea Merrison- Consultant Neurologist Bristol

**2008**

Dr Chris Everett - now Consultant Neurologist at the Royal London

**2009**

Dr Susie Tomlinson – now Consultant Neurologist Sydney

**2010**

Dr Jasper Morrow – now Consultant Neurologist Lister Stevenage & NHNN

**2011**

Dr Tracey Graves Senior - now SpR Oxford

## **12. Teaching Activity-PhD Examiner**

### ***PhD Examiner***

I have been appointed as an examiner for eighteen PhD theses for the Universities of London, Oxford and Newcastle

**1997**

1. Supervisor Professor DM Turnbull  
“Mitochondrial heteroplasmy and triplasmmy”  
University of Newcastle-upon-Tyne

**2002**

2. Supervisors Professors Turnbull and Lightowlers  
“Ageing and mitochondria”  
University of Newcastle-upon-Tyne

**2003**

3. Supervisor Professor DM Turnbull

“Mitochondrial disease and neurodegeneration”  
University of Newcastle-upon –Tyne

4. Supervisor Professor K Bushby  
“Genetics of Limb girdle muscular dystrophy”  
University of Newcastle-upon-Tyne

**2004**

5. Supervisor Professor R Smith  
“Genetics and expression studies of ryanodine receptor mutations”  
University of London
6. Supervisor Professor F Muntoni  
“Clinical and genetic study of congenital muscular dystrophy”  
University of London
7. Supervisor Professor F Muntoni  
“Clinical and genetic study of minicore myopathy”  
University of London

**2005**

8. Supervisor Professor A Vincent  
“Genetics of congenital myasthenia gravis”  
University of Oxford
9. Supervisor Professor A Vincent  
“Musk antibodies and Myasthenia Gravis”  
University of Oxford

**2006**

10. Supervisor Professor J Poulton  
“Mitochondrial liver disease”  
University of Oxford
11. Supervisor Professor C Shaw  
“Respiratory chain dysfunction in ALS”  
University of London

**2007**

12. Supervisor Professor Kay Davis  
“Syncoilin a key protein in muscular dystrophy”  
University of Oxford

**2008**

13. Supervisor Professor Scolding  
“Developing stem cell transplant as a treatment for muscle diseases”  
University of Bristol

**2009**

14. Supervisor Professor Turnbull  
“Formation of mtDNA deletions”  
University of Newcastle upon Tyne

**2010**

15. Supervisor Professor Turnbull  
“Prevention of transmission of mtDNA mutations”  
University of Newcastle upon Tyne
16. Supervisor Professor Muntoni  
“Congenital muscular dystrophy”  
University of London

**2011**

17. Supervisor Professor Turnbull  
“Investigating the neuropathology of mitochondrial diseases”  
University of Newcastle upon Tyne

**2013**

18. Supervisor Professor Stephen D.R. Harridge  
King’s College London

### **13. Enabling Activity**

**Local**

The Institute of Neurology/UCL:

My research involves mentoring, supervising and overseeing the training and development of clinical and non-clinical PhD students and Post-doctoral research workers. I have supported the progression of all trainees who have left my group and have supported their career subsequently. All trainees have achieved good publication records and have made good career progress, many to consultant level, as outlined in section 12 above. In addition, I contribute to the neurogenetics unit and collaborate closely with Professors Wood, Houlden and Reilly. This has two main functions DNA service and research. I am responsible for coordinating the DNA based diagnostic work for muscle genetic disorders such as mitochondrial disorders and for channelopathies. I integrate this with the specific training needs of highly competent clinical scientists, many of whom will move back into clinical practice as neurologists with a primary interest in neurogenetics. I established the current UCL undergraduate neurology teaching course and moved the course to be based at Queen Square in 2000. I was awarded UCL top clinical teacher in 2000 by undergraduate medical students.

As **Clinical Director** I have worked closely with the Institute and delivered several major projects that have important research links including

- The brain tumour unit
- The Centre for Neuromuscular Diseases

- The Hyper-acute stroke unit
- I have initiated a comprehensive review of the neurogenetics service to achieve improved responsive service standards and ensure rapid translation for research into clinical practice.

**UCL Partners** is the UCL linked Academic Health Sciences system that has a tripartite mission of linking UCL with neighboring large Trusts to deliver improved patient health outcomes through research and education. As **Co-Director of the UCL Partners Neurological disease theme** I have worked with Alan Thompson and Tony Schapira to develop a clear strategy for neurological diseases within the partnership and we have focused on delivery of progress in the three key areas: 1. neuro-oncology 2 stroke disease and 3 neurodegenerative diseases.

### **National**

Over the past ten years I have obtained ~£8m in competitive NHS funding to run two nationally commissioned services.

In 2001 in open competition with other centres I obtained recurring funding [currently £450k per annum] from the Department of Health through National Specialist Commissioning to run a National Clinical and DNA-based diagnostic service for patients with skeletal muscle channelopathies. This funding now supports three clinical scientists [2 DNA 1 molecular expression], consultant sessions, a full time clinical nurse specialist, and consumables. In addition I have made several successful capital bids amounting to an additional £350K to purchase a HPLC wave DNA analyzer, a DNA handling robot for the neurogenetics lab and a new clinical neurophysiology work station based in the neurophysiology department.

This National service now in its tenth year has strengthened the position of the National Hospital and the Institute of Neurology as the clear leader in this area in the UK and recognized internationally. A unique database of brain and muscle channel patients has been established and has been a major resource facilitating both internal collaborations in the Institute eg Professor Kullmann and International collaborations eg grants from NIH [USA] in collaboration with Professor Griggs, University Rochester USA. The muscle clinics I run in the Centre for Neuromuscular disease have attracted trainees from all over the UK. I operate 4-6 month attachments. Many colleagues who have attended my clinics are now neuromuscular consultants all over the UK and beyond [see list of individuals].

In 2006 I obtained competitive NCG funding jointly with Newcastle and Oxford to establish the national specialist mitochondrial disease service. The Queen Square part of the service receives £1m per annum to provide a comprehensive clinical and genetic diagnostic service.

### **International**

I have developed an international reputation as an expert in neuromuscular diseases and particularly channelopathies and mitochondrial diseases. My work in establishing the UK national clinical and genetic centre for channelopathies has stimulated international research collaborations [see grants NIH USA in collaboration with Professor Griggs-University Rochester USA]. I am a scientific advisor for the North American Muscle Study Group. This group established by Professor Griggs involves many of the key international figures in muscle research and has an important goal of developing new treatments through basic research and clinical trials. I have organized workshops and been invited to lecture at numerous meetings in USA, Europe and Australia. I am Deputy Editor of the Journal of Neurology, Neurosurgery and Psychiatry, an internationally recognized Journal. I co-

authored the fourth edition of the well known undergraduate text book "Tutorials in differential diagnosis" published in 2003 and now on its third print run having sold over 12000 copies world-wide and translated into several languages.

## **14. Research Activity**

The neurogenetics research group in the Department of Clinical Neurology was established in 1985 (by the late Professor AE Harding). Anita Harding supervised my original research higher degree and inspired my interest in studying the molecular basis of neuromuscular disease. Anita Harding taught me "patient centered" molecular genetic research. Since my consultant/Senior Lecturer appointment in January 1998 I have undertaken clinical/molecular genetic research into mitochondrial and ion channel neurological disorders. The Ion channel research has successfully translated into the UK national clinical and genetic diagnostic service for patients with muscle channelopathies, and also mitochondrial diseases funded directly by the Department of Health.

Major research areas:

### **Human Mitochondrial diseases**

- The expertise in mitochondrial diseases which was established under Prof AE Harding. My group continues to define new mutations in mtDNA associated with human neurological disease which permits new insights into pathogenetic mechanisms. I have spent several years defining new mitochondria DNA mutations and investigating the mechanisms of pathogenicity using a different primary and transformed cell systems. More recently I have studied the role of nuclear mitochondrial interactions as a disease mechanism in human mitochondrial disease. We developed a cell model which allows nuclear-mitochondrial genetic interactions to be studied, with the aim of identifying important nuclear genes. We have recently identified two new nuclear encoded genes in families with previously uncharacterized mitochondrial disease.
- I published in high impact journals such as New England Journal of Medicine, the Lancet and American Journal of Human Genetics. I have written invited reviews and book chapters in this area. Recently, I was principal applicant on a successful five year MRC cooperative application "Mitochondria in Health and Disease". I have received two components grants from this cooperative. Currently I am actively pursuing the role of nuclear genes in mitochondrial disease [the second component project grant] in collaboration with Dr Rahman in the Institute of Child Health. Recent collaborations with Prof Chinnery and Turnbull in Newcastle have allowed us to define new phenotypes associated with mitochondrial nuclear gene mutations. I now also collaborate closely with Professor Michael Duchon in Physiology UCL. Through a jointly funded post-doc position we have recently provided evidence for a nuclear role in the expression of a mitochondrial DNA mutation and obtained data that suggest mutant mitochondria not only fail to produce ATP but also consume cellular ATP thereby suggesting a new mechanism of cellular pathophysiology-published in Journal of Biological Chemistry. All my basic research work in mitochondrial disease is allied to a large clinical mitochondrial service based in the National Hospital. In my specialist muscle clinic I follow up a large cohort of mitochondrial patients and am now building a cohort Nationally in collaboration with Doug Turnbull following a successful joint MRC Cohort grant application. The mitochondrial work has lead to the Nationally commissioned mitochondrial disease service now £1m

per annum to Queen Square.

### **Human Neurological Channelopathies**

- There are an increasing number of neurological conditions that have now been shown to be due to inherited abnormalities of ion channel function. I established a National database for patients with neurological channelopathies in 1996 with the support of the Association of British Neurologists Surveillance Unit. This data-base is now one of the largest collections of such patients in the world and has been an invaluable asset in forming collaborations with colleagues locally [Professor Kullmann and Wood], nationally [as a co-investigator on the Oxford ion channel initiative funded by the Wellcome Trust-Professor Francis Ashcroft and Professor Kay Davies] and internationally [eg NIH grants with Professor RC Griggs North America]. The setting up of an interdisciplinary group in ION/UCL with cellular physiologists (Prof DM Kullmann Dr Schorge and Prof A Dolphin) and geneticists (Prof Wood) has allowed my group to investigate some of these patients at a molecular genetic and cellular electrophysiological level. We have extensively studied clinical and molecular aspects of muscle channelopathies and some neuronal channelopathies. We have defined the range of clinical phenotypes associated with neuronal potassium channel dysfunction and related this to the cellular electrophysiological phenotypes. We have shown that there is an increased risk of epilepsy in patients with neuronal potassium channel dysfunction [Annals of Neurology, Brain]. We have provided evidence that the brain P/Q-type calcium channel may have a role in human absence epilepsy [Lancet, Brain]. In collaboration with Prof Wood, Dr Sisodiya and Prof Goldstein [Duke USA] we are currently addressing the role of calcium channel variation as a risk factor in common forms of epilepsy. This study employs a population based linkage disequilibrium approach developed by Professors Wood and Goldstein. This ion channel work has led directly to the establishment of the UK national centre for diagnosis and treatment of neurological channelopathies funded directly by the UK department of health currently £450 000 per year. In addition I have obtained funding from the NIH-USA, MRC-UK, Action Research UK and the Wellcome Trust UK, to support basic science and clinical research into channelopathies. I am a co-investigator on the recent successful Oxford-based Wellcome funded integrative physiology programme grant on ion channels and human disease.
- It is probable that some other neurological disease areas (most notably the epilepsies, migraine and paroxysmal movement disorders) are also due to channels. This functional work will complement our positional cloning strategies once these genes are cloned. Just recently in collaboration with Louis Ptacek in USA we have identified a new gene for the paroxysmal movement disorder PKD (see publications).
- I have published original channel research widely in international journals including Lancet, Annals of Neurology, Brain, Journal of Physiology, and Neurology. I have published several reviews in this area, most recently I have been invited to provide a review for Nature Neurology in Clinical Practice.

### **Clinical Aspects of Human Muscle Diseases**

- As part of the MRC Centre I am actively involved in developing better ways to measure disease activity, disease damage and response to treatment in patients with inflammatory muscle disease. I published new scales to analyse these parameters with colleagues from UCL and NIH -Bethesda in the Journal Rheumatology 2004/11. I am adapting these



scales to use in treatment of certain dystrophies with the intention of using new agents such as myostatin antibodies [through collaboration with Wyeth].

The ability of the neurogenetics group to translate research into clinical practice rapidly, particularly in terms of preclinical and prenatal diagnosis and thus improved genetic counseling, was enhanced by the establishment of a DNA service laboratory (initially staffed by three clinical scientists but which has rapidly expanded over the last 10 years I have been involved). This forms part of the National Hospitals' Clinical and Molecular Neurogenetics Unit, which is unique in simultaneously providing three components of services related to inherited neurological disorders in adults: clinical diagnosis, DNA services and genetic counseling. In the DNA laboratory, we offer gene carrier prediction, prenatal diagnosis and mutation detection (where appropriate) for many inherited neurological diseases on a regional and national basis, including Huntington's disease, myotonic dystrophy, mitochondrial encephalomyopathies, Leber's hereditary optic neuropathy, hereditary motor and sensory neuropathy, dominant ataxias, prion diseases, familial amyloidoses, HMSN I, Friedreich's ataxia, primary generalized dystonia and X-linked bulbospinal neuronopathy. The provision of this service enhances recruitment of material suitable for study and over the past 10 years the unit has collected over 20,000 DNA samples.

## 15. Publications

### 15a Some example key papers

Andreu AL, **Hanna MG**, Reichman H, Bruno C, Penn AS, Tanji K, Palloti F, Iwata S, Shanske S, Bonilla E, Morgan-Hughes JA, DiMauro S (1999) Aches, pains and cramps: Exercise intolerance due to mutations in the cytochrome b gene of mitochondrial DNA.

*New England Journal of Medicine* 341: 1037-1078

Cited 256 times [Google scholar] Impact Factor 34.833

This work published in the New England Journal of Medicine was the first demonstration that somatic mutations in the protein encoding gene cytochrome b in human mitochondrial DNA could associate with a commonly encountered clinical phenotype of exercise intolerance and myalgia. This was a collaborative research project with colleagues in USA and Germany. We identified a group of patients in which the defect in cytochrome b in mtDNA was only detectable in muscle [not in blood]. Prior to these findings point mutations in human mtDNA which were only confined to single somatic tissue had not been described. This suggested that the mutational event causing this form of human mitochondrial disease must have occurred in a cell line already destined to differentiate into muscle. Such late mutational events [ie not germline] had not been recognized previously for point mutations. We undertook detailed biochemical experiments to confirm a specific defect in complex III of the respiratory chain. This work served to initiate the area of somatic mtDNA point mutations in human disease. Subsequently my group and others have identified further somatic mutations in other mtDNA encoded genes in patients with a similar phenotypes confirming this is a new category of mtDNA mitochondrial disease [see ref 56 and 68]. From a clinical viewpoint this work emphasized the need to not rely on analysis of mtDNA point mutations in blood when assessing such patients. This is in contrast to germline mtDNA point mutations which are always detectable in blood. There was an accompanying Editorial

Review in the New England Journal of Medicine by Professor RC Griggs (Head of Department of Neurology, University of Rochester New York and Professor George Karpati, (Head of the Montreal Institute of Neuromuscular disease).

In addition this work generated subsequent correspondence in the New England Journal of Medicine.

N Engl J Med. 1999 Sep 30;341(14):1077-8.

N Engl J Med. 2000 Feb 10;342(6):438-9; author reply 439-40.

**Hanna MG**, Nelson IP, Rahmann S, Schapira AHV, Morgan-Hughes JA Wood NW (1998) Cytochrome c Oxidase deficiency associated with the first stop codon mutation in human mitochondrial DNA *American Journal of Human Genetics* 63:29-36  
Cited 54 times [Google scholar] Impact Factor 11.602

This paper reports some of our findings resulting from a larger body of work studying mitochondrial DNA in human disease. At the time of this report although a number of pathogenic mutations in human mtDNA had been identified it was notable that mutations in protein coding genes that caused premature truncation of protein synthesis had never been reported despite being common in lower organisms. In this study I analyzed the entire human mitochondrial genome by direct DNA sequence analysis in a patient with a severe reduction in mitochondrial complex IV activity. I identified the first stop codon mutation described in human mitochondrial disease. The mutation occurred in subunit III of the cytochrome oxidase protein and resulted in loss of a large part of the C terminal region of the protein which is key for the catalytic activity of cytochrome c oxidase. Subsequently we and others identified additional similar mutations [see reference 68]

Pulkes T, Eunson L, Patterson V, Wood NW, Nelson IP, Morgan-Hughes JA, **Hanna MG** (1999) The mitochondrial DNA G13513A transition in ND5 is associated with a LHON/MELAS overlap syndrome and may be a frequent cause of MELAS. *Ann Neurol* 46:916-919.  
Cited 76 times [Google scholar] Impact Factor 8.71

Soon after I discovered the first stop codon mutation in human mitochondrial DNA I collaborated with Professor Turnbull's group in Newcastle upon Tyne on further cases with a similar myopathological phenotype of profound cytochrome c oxidase reduction. We successfully characterized the first initiation codon mutation in humans as a further genetic cause of this marked cytochrome c oxidase deficiency. Together the findings published in these two papers in the American Journal of Human Genetics were the first to show that defects in mitochondrial protein termination [ie premature termination] and initiation [ie failure of initiation] could cause human disease.

Zuberi S, Eunson L, Spauschus A, DeSilva R, Wood NW, McWilliam RC, Stephenson JPB, Tolmie J, Kullmann DM and **Hanna MG** (1999) A new potassium channel mutation associates with episodic ataxia type I and epilepsy *Brain* 122:817-825  
Cited 75 times (Google Scholar) Impact factor 9.16  
Jouvencaeu A, Eunson LH, Spauschus A, Ramesh V, Kullmann DM and **Hanna MG** (2001)

Human absence epilepsy associates with dysfunction of the brain P/Q type calcium channel CACNA1A *Lancet* 34:45-51

Cited 142 times (Google Scholar) Impact Factor 18.316

I established a UK National database of patients with suspected brain and muscle channelopathies at the end of 1996. This subsequently evolved into the current UK national clinical and diagnostic service funded directly by the UK department of Health [following my application in open competition]. The two papers cited above published in *Brain* and in the *Lancet* stemmed initially from clinical observations I made in patients with episodic ataxia. It was evident that some of these patients had epilepsy in addition to the ataxia phenotype. However, it was not clear if this was a chance association or if the epilepsy was generated by the dysfunctional brain ion channels I suspected these patients must have. In collaboration with Professor Dimitri Kullmann and Professor Nicholas Wood we studied such families in detail from a clinical, genetic and molecular expression view point. I undertook extensive DNA sequence analysis of candidate channel genes. New mutations in the neuronal potassium channel KCNA1 gene and the brain calcium channel CACNA1A gene were discovered. We went onto prove molecular mechanisms of pathogenicity of identified mutations through molecular expression experiments. In the paper published in *Brain* 1999 we provided strong evidence that in the patients with neuronal potassium channel mutations there is an increased risk of epilepsy as a direct result of the potassium channel dysfunction. This has subsequently been confirmed by others and is now generally accepted. In addition, we went on to define new phenotypes of human disease associated with the new KCNA1 mutations we identified. We showed that the range of phenotypes was from mild isolated neuromyotonia through to severe drug resistant episodic ataxia with epilepsy [See ref 31 published in *Annals of Neurology*]. The *Lancet* publication provides the first evidence for a link between human brain calcium channel dysfunction and epilepsy. My first Phd student Ms Louise Eunson obtained the Queen Square prize 2000 [graduated UCL PhD 2001] for this work published in *Brain* and the *Lancet*. We have recently described a significant body of evidence strengthening the link between calcium channel dysfunction and human absence epilepsy. Part of this evidence is now published in *Brain* 2004 see ref 64. Further evidence in the form of new CACNA1A mutations and an extensive review of all EA2 cases published is in preparation [see ref 86]. Recently, Professor Geoffrey Noebels who first identified spontaneous CACNA1A mutant mice with absence epilepsy in 1979 [*Science* 204;1334-1336] has published a detailed commentary on our *Brain* 2004 paper supporting the role of CACNA1A in human epilepsy see Noebels JL *Epilepsy Current* [Literature in Basic Science] 2005: 5; 95-7

### **Recent key muscle channelopathy publications:**

Fialho D, Schorge S, Pucovska U, Davies NP, Labrum R, Haworth A, Stanley E, Sud R, Wakeling W, Davis MB, Kullmann DM, **Hanna MG**. Chloride channel myotonia: exon 8 hot-spot for dominant-negative interactions. *Brain*. 2007 Dec;130(Pt 12):3265-74. PMID: 17932099. (Cited 44 times Google Scholar)

Matthews E, Tan SV, Fialho D, Sweeney MG, Sud R, Haworth A, Stanley E, Cea G, Davis MB, **Hanna MG**. What causes paramyotonia in the United Kingdom? Common and new SCN4A mutations revealed. *Neurology*. 2008 Jan 1;70(1):50-3. PMID: 18166706. (Cited 41 times Google Scholar)

Matthews E, Labrum R, Sweeney MG, Sud R, Haworth A, Chinnery PF, Meola G, Schorge S, Kullmann DM, Davis MB, **Hanna MG**. Voltage sensor charge loss accounts for most cases of hypokalemic periodic paralysis. *Neurology*. 2009 May 5;72(18):1544-7. PMID: 19118277. (Cited 39 times Google Scholar)

Editorial By Steve Cannon on this article in same edition of *Neurology* entitled "Getting a charge out of periodic paralysis" *Neurology* May 5 72 1432.

Matthews E, **Hanna MG**. Muscle channelopathies: does the predicted channel gating pore offer new treatment insights for hypokalaemic periodic paralysis? *J Physiol*. 2010 Jun 1;588(Pt 11):1879-86.. Review. PMID: 20123788.

Matthews E, Guet A, Mayer M, Vicart S, Pemble S, Sternberg D, Fontaine B, **Hanna MG**. Neonatal hypotonia can be a sodium channelopathy: recognition of a new phenotype. *Neurology*. 2008 Nov 18;71(21):1740-2. PMID: 19015492 (Google scholar cites 31 times)

Matthews E, Manzur AY, Sud R, Muntoni F, **Hanna MG**. Stridor as a neonatal presentation of skeletal muscle sodium channelopathy. *Arch Neurol*. 2011 Jan;68(1):127-9. PMID: 21220685.(Google Scholar cited 31 times)

Tan SV, Matthews E, Barber M, Burge JA, Rajakulendran S, Fialho D, Sud R, Haworth A, Koltzenburg M, **Hanna MG**. Refined exercise testing can aid DNA-based diagnosis in muscle channelopathies. *Ann Neurol*. 2011 Feb;69(2):328-40. PMID: 21387378. Cited 29 Times (Google Scholar cited 26 times)

The above muscle channel publications describe a series of studies which have made a number of important observations in relation to muscle channelopathies that provide both insights into fundamental mechanisms of ion channel function, as well as revealing new genotype phenotype correlations with clinical impact.

### ***New Insights into fundamental mechanisms of ion channel function***

- Expression of mutations in the muscle chloride channel mutations in oocyte expression system show dominant negative interactions are mediated at the interface linking the two monomers that make up the function channel
- Loss of positive charge mutations in the S4 segments of both sodium and calcium muscle channels link to the phenotype of hypokalaemic periodic paralysis
- Genetic evidence supports the view that loss of charge mutations induce a gating pore current

### ***New insights into genotype phenotype correlations***

- The full spectrum of chloride and sodium channel mutations associated with inherited myotonias in the UK population defined

- Dominant mutations found more commonly in chloride channel than previous reports with important genetic counseling implications
- Neonatal hypotonia recognised as a new phenotype for sodium channel disease altering guidance for neonatal care of children born to affected parents
- Previously unexplained severe neonatal stridor/laryngospasm shown to be a new sodium channel disease phenotype with implications for diagnosis and management of neonatal stridor

### ***Delivery of human randomized clinical trials in a rare disease***

- I am the senior author on a recently published trial reprofiling an agent to target human muscle sodium channels to treat an inherited myotonic disorder. This trial was coordinated across several countries and demonstrated that it is possible to deliver trials in rare neurological diseases by international coordination. It was published with an Editorial in **JAMA 2012**
- Statland JM, Bundy BN, Wang Y, Rayan DR, Trivedi JR, Sansone VA, Salajegheh MK, Venance SL, Ciafaloni E, Matthews E, Meola G, Herbelin L, Griggs RC, Barohn RJ, **Hanna MG**; Consortium for Clinical Investigation of Neurologic Channelopathies. Mexiletine for symptoms and signs of myotonia in nondystrophic myotonia: a randomized controlled trial. *JAMA*. 2012 Oct 3;308(13):1357-65. PMID: 23032552

### **15b. Books/Thesis**

1. **Hanna MG**, co-authored with Holdright D, Beck E, Souhami R. (2003). *Tutorials in differential diagnosis*, 4<sup>th</sup> Edition. Harcourt, London: p245. [6,000 copies sold worldwide].
2. **Hanna MG** (1996). MD thesis. University of Manchester. "Molecular genetic studies in human mitochondrial disease". Supervisor *the late* Professor Anita Harding.

### **15c. Chapters**

1. Rudel R, Hanna MG, Lehmann-Horn F (1999). Muscle ion channelopathies: malignant hyperthermia, periodic paralyses, paramyotonia congenita and myotonia. *Muscle Diseases*. Eds: Schapira AHV, Griggs R. Butterworth Heinemann, pp 134-154.
2. Hanna MG (2000). Cases from the National Hospital. Eds: CD Marsden, A Wills. Martin Dunitz, London, two cases, pp23-26 & pp56-59.
3. Davies NP Hanna MG (2001). Genetic neurological channelopathies. *New Treatments in Neurology*. Ed: Scolding N. Butterworth Heinemann, London, pp234-247.

4. Kinton L, Hanna MG, Wood NW (2001). Genetics of epilepsy and paroxysmal movement disorders. *Epilepsy and Movement Disorders*. Eds: Guerinni, Aicardi, Andermann, Hallet. Cambridge University Press, *pp67-79*.
5. Davies NP, Hanna MG (2003). Myopathies and neurorehabilitation. *Textbook of Neurorehabilitation*. Ed: Greenwood R. *pp256-274*.
6. Pulkes T, Hanna MG (2003). Mitochondrial diseases. *Mitochondrial DNA and Human Disease*. Ed: Holt IJ. Butterworth Heinemann, *pp57-75*.
7. Liolitsa D, Hanna MG (2003). Cell models in mitochondrial disease. *Mitochondrial Function and Dysfunction in Human Disease*. Eds: Schapira AHV, DeLauro S. Butterworth Heinemann, London, *pp89-110*.
8. Hanna MG (2003). Peripheral neuropathy and mitochondrial diseases. *Peripheral Neuropathy*. Eds: Dyck D, Thomas PK. Saunders, *pp1937-1949*.
9. Hanna MG (2004). Disorders of voluntary muscle. *Oxford Textbook of Medicine*, 4<sup>th</sup> Ed. Oxford, *pp1344 -1346*.
10. Fontaine B, Hanna MG (2009). Muscle ion channelopathies and related disorders. *Disorders of Voluntary Muscle*, 8th Ed. Eds: George Karpati, David Hilton-Jones, Kate Bushby, Robert C. Griggs. Cambridge University Press, *pp409-426*.
11. Hanna MG, Parton M (2009). Muscle diseases. *Neurology: A Queen Square Textbook*. Eds: Clarke C, Shorvon S, Howard R, Rossor M. 1st Ed. Wiley-Blackwell, Chichester, *pp.337-410*.
12. Raja Rayan D, Hanna MG (2010). Ion channels and human disorders. *Encyclopaedia of Life Sciences*. John Wiley & Sons, *pp1-17*.
13. Fialho D, Hanna MG (2010). Periodic paralysis. *Handbook of Neurology*, Eds: Hilton Jones DA, Laing D. Heinemann, *pp 92-118*.
14. Burge J, Hanna MG (2011). Muscle channelopathies. *Neurology in Practice: Neuromuscular Disorders*. Eds: Rabi N. Tawil, Shannon Venance. Wiley-Blackwell.
15. Hanna MG, Kullmann DM (2012). Channelopathies. *Neurogenetics: A Guide for Clinicians*. Ed: Nicholas W. Wood. Cambridge University Press, *pp121-135*.
16. Matthews E, Hanna MG (2013). Cav1.1 hypokalaemic periodic paralysis. *Pathologies of Calcium Channels*. Eds: Norbert Weiss, Alexandra Koschak. Springer.
17. Hanna MG (2013). Muscle Disease: Pathology and Genetics. 2<sup>nd</sup> Ed. Eds: Hans H. Goebel, Caroline A. Sewry, Roy O. Weller. Wiley-Blackwell.
18. Hanna MG (2014). Diseases of the muscles. *Scully's Medical Problems in Dentistry*. 7<sup>th</sup> Ed. Ed: Scully C. Elsevier, *chapter 16*.

19. Matthews E, Hanna MG (2014). Skeletal Muscle Channelopathies. Oxford Textbook of Neuromuscular Disorders. Oxford Textbooks in Clinical Neurology. Eds: David Hilton-Jones, Martin R. Turner. Oxford University Press, pp 316-325.
20. Suetterlin K, Hanna MG (2016). Muscle Channelopathies. International Neurology: A Clinical Approach. 2<sup>nd</sup> Ed. Wiley-Blackwell. Part 13, ch. 116.
21. Pitceathly R, Hanna MG, Reilly MM (2018). Neurology – the Peripheral Nervous System. Clinical Mitochondrial Medicine. Cambridge University Press, section 2, ch 8.
22. Hanna MG (in press). Neurogenetics. The Oxford Handbook of Molecular Medicine Ed: Lo. Oxford University Press.
23. Hanna MG (in press). Neurological Channelopathies. Ed. Hanna. Elsevier.
24. Pizzamiglio C, Pitceathly RDS, Hanna MG (in press). White Matter Disorders, Mitochondrial Disorders, ch 4. Eds. Lynch, Houlden. Elsevier. (Handbook of Clinical Neurology Series)

#### 15d Peer Reviewed Original Publications

##### 1989

1. Selam JL, Kashyap M, Alberti KG, Lozano J, **Hanna M**, Turner D, Jeandidier N, Chen E, Charles MA. Comparison of intraperitoneal and subcutaneous insulin administration on lipids, apolipoproteins, fuel metabolites, and hormones in type I diabetes mellitus. Metabolism. 1989 Sep;38(9):908-12. PMID 2671601

##### 1995

2. Nelson I, **Hanna MG**, Alsanjari N, Scaravilli F, Morgan-Hughes JA, Harding AE. A new mitochondrial DNA mutation associated with progressive dementia and chorea: a clinical, pathological, and molecular genetic study. Ann Neurol. 1995 Mar;37(3):400-3. PMID: 7695240
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