Curriculum Vitae

Professor Michael G Hanna

2014

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

Current and recent positions:

Director, UCL Institute of Neurology, Queen Square

Professor in Clinical Neurology
Department of Molecular Neuroscience
UCL Institute of Neurology

Director, UCL MRC Centre for Neuromuscular Diseases

Divisional Clinical Director, Queen Square Division, UCLH NHS FT (2007-2012)

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 in Neurology, University of Iowa, USA

Co-Director, Neurological Diseases Theme
UCL Partners Academic Health Sciences Centre (2009-2012)

Founding Chairman, British Myology Society

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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
- 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
- 2009-2012 Co-Director UCL Partners Neurological Diseases Theme
- 2011-present Honorary Professor in Clinical Neurology, Newcastle University
- 2012-present Director UCL Institute of Neurology

Academic Activity: basic discovery science and translational research

- 1995-2014 Published over 200 peer reviewed papers and reviews, over 250 published abstracts, 17 book chapters and 1 book. H-Index >40.
- 1998-2014 Obtained over £14 million external basic and translational research funding including MRC, Wellcome Trust plus £8million NHS NCG funding.
- 1998-2014 Supervised 18 clinical and non clinical PhD research fellows/students and 3 post doctoral research fellows
- 2007-2017 Successfully lead joint initial application and renewal, across UCL and Newcastle and direct joint UCL Newcastle MRC Centre for Neuromuscular Diseases->£10m

Editorships and other esteem

- 2003-present 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

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 ~£475 000 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-present 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 4th edition of well known undergraduate test "Tutorials in Differential Diagnosis" sold 12000 copies world-wide, translated into 6 languages.

Private Fundraising and Philanthropy

- 2007-2008 raised £1.7m which combined with successful CBRC 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 entre
- 2007-present £350k Kennedy's Disease Fund
- 2009-worked with National Brain Appeal to raise £1m for new ULCH NHNN Brain tumour unit.
- 2011 £250,000 Chandris Foundation towards Neuromuscular research

Clinical Management and Neuromuscular Clinical Services

- 2007-present 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.
- 16 years front line consultant general neurologist - I have broad front-line experience in all major general neurological conditions.

Full Curriculum Vitae

2. Personal Details

Michael G. Hanna 04.10.63 (51 years)

3. Education/Qualifications

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

4. Current Post

My current consultant position includes

- Research- Director UCL MRC Centre for Neuromuscular Diseases-since 2008
- Clinical work and patient care-Consultant Neurologist providing a specialist muscle service including two Nationally commissioned NCG services-since 1998
- Senior Management-Divisional Clinical Director, Queen Square-since 2007

4a. Research Programmes-Discovery and Translational

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. As part of the MRC Centre I have developed wide-spread collaborations and an interest in the cross cutting theme of muscle degeneration.

Since 2000 I have obtained in excess of **~£10** million pounds sterling in external peer reviewed funding to support my research programmes into mitochondrial disease and human neurological channelopathies. These include project grants, cooperative component grants, training fellowships and programme grants from many sources including the MRC, Wellcome Trust, Action Research, NIHR and NIH-USA.

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 **£8** million pounds sterling 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 have 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 the underlying 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 have also defined previously unrecognized neonatal ion channel diseases including intermittent hyoptonia and stridor–knowledge of which has lead to improved and safer care of affected patients in the neonatal period.

- We identified we that 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.

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, welcome Trust 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 have 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 2008-2012

Eight 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 am the Centre Director (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.***

This new centre which I direct is entirely in line with the 2010-2015 mission and strategy of UCL School of Life and Medical Sciences.

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 for the five years 2008-2014

- 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.

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–present

- 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 have been Queen Square Divisional Clinical Director since 2007 and in collaboration with the Divisional Manager-Jackie Sullivan I have responsibility for Divisional financial performance and activity, quality, workforce, governance, estates and clinical strategy.

- The management team I lead produced the new 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 am jointly responsible with the National Hospital Divisional manager for all clinical, staffing financial and governance issues in the Hospital. The current budget revenue is £143m pa and the workforce is 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 patients 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 gives 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 since January 2004. Involves 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
- North American Muscle Interest Group member and Scientific Advisor 2003-present
- On various trials steering group for example "Immunospuression in myositis" MRC trial 2007-present

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 for: The Wellcome Trust, MRC, Telethon (Italia), Action Research, Wolfson Foundation.

Institute, University, and Hospital Committees

- *Institute of Neurology*: Academic Board, Education subcommittee 2000-2007
- *National Hospital*: Medical Committee (Secretary 2000-2003),
- *National Hospital* Chair Neurology directorate committee (200-2007)
- *University College Hospital Trust*: UCLH-trust special trustees research award committee(2001-2007)
- *University College London*: Neurosciences new curriculum undergraduate planning committee (2003-2007)
- *University College London NHS Trust:* member use medicines committee (02-05).

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.

9. Grants

Previous Grant Funding:
1992
Medical Research Council Training Fellowship-1992-1994

The study of the molecular pathogenesis of primary defects of mtDNA using a human myoblast culture system
MG Hanna-supervisor Prof AE Harding
£60,000

1996
Joint Research Advisory Committee Institute of Neurology-1996-1997
Mechanisms of phenotypic diversity associated with the mitochondrial A3243G point mutation
MG Hanna Principal applicant and Collaborator with Dr IP Nelson
£35,000

1997
Brain Research Trust-1997-1999
Clinical and Genetic studies of human neurological channelopathies
MG Hanna Principal applicant
£80,000

Brain Research Trust -1997-1999
Expression studies of human neurological channelopathies
MG Hanna co-applicant with Professor DM Kullmann
£90,000

1998
Wellcome entry level fellowship for medical research fellow-1998-1999
Studies of mtDNA in human Neurological disease
MG Hanna sponsor and supervisor for Dr Siddique.
£57,000

1999
Wellcome entry level fellowship for medical research fellow-1999-2000
Molecular genetic studies of human skeletal muscle channelopathies
MG Hanna sponsor and supervisor for Dr N Davis.
£59,000

Epilepsy Research Foundation-1999-2001
Clinical, Genetic and Expression studies of the voltage gated calcium channel (CACNA1A) in complex human epilepsy phenotypes
MG Hanna, Principal applicant
£50,000

Brain Research Trust-1999-2001
Molecular basis of phenotypic heterogeneity associated with mtDNA mutations
MG Hanna principal applicant
£55,000

2000
Wellcome training fellowship-2000-2003
MG Hanna, sponsor and supervisor for N Davies

Molecular genetic and expression studies on muscle chloride channelopathies
£120 000

MRC mitochondrial cooperative: mitochondria in health and disease
Cooperative status awarded 1999-2004
MG Hanna co-principal applicant with Prof JB Clarke, Prof M Duchen, Prof Wood, Prof Rich, Prof Crompton and Prof Moncado-UCL

MRC mitochondrial cooperative –component grant-2000-2003
MG Hanna-principal applicant one component grant
Identifying nuclear genes which influence the expression of mtDNA mutations
£180 000

Special Trustees of the UCLH NHS trust 2000-2002
MG Hanna principal applicant
Clinical, genetic and expression studies of the voltage gated calcium channel CACNA1A in neurological disease
£69000

2001
National Institutes of Health [Bethesda USA] 2001
Research proposal planning grant
An international trial of treatment in skeletal muscle channelopathies
USA PI-Professor RC Griggs and UK Co PI-MG Hanna
$35 000

2002
Competitive NHS-service related grants 2002-recurrent
National Specialist Commissioning Agency [NCG]
Principal applicant.
Recognised by the Department of Health as a National Centre for DNA-diagnosis in muscle channel disease. Permanent funding of £450,000 per annum to provide this service. Funding for DNA-scientist, consultant sessions and DNA equipment.

2003
Kennedy's disease research fund grant 2003-2007
Co-PI Dr MG Hanna Linda Greensmith. Clinical research fellow undertook molecular genetic research into Kennedy's disease.
£75,000

MRC Cooperative-Epilepsy-2003-2005
Component grant £375,000
Commenced July 2003-2006
Awarded to DM Kullmann, NW Wood and MG Hanna
Ion channels in epilepsy

National Institutes of Health Bethesda [USA]-2004-2008
Clinical Investigations into Neurological Channelopathies "CINCH"
UK Centre Co PI on International multicentre study in neurological channelopathies

Funded one clinical research fellow salary per year for 5 years-£250,000

Wellcome Integrative Physiology Ion Channel Programme grant to Oxford University
OXION 2003-2007 £3.4 million
Co-Investigator on grant

2004
National Institutes of Health [Bethesda USA] awarded end 2004
An international trial of pharmacological intervention in periodic paralysis
$3.1million US USA PI and Coordinator-Professor RC Griggs-
only UK centre Co-PI MG Hanna

Guarantors of Brain 2004-2005
MG Hanna Supervisor for Dr Tracey Graves
Research fellowship
£55,000

UCL Special Trustees-Fast track grant application 2004
MG Hanna Principal applicant and supervisor [1 post doc]
"Nuclear genes and mitochondrial disease"
£29000

2005
Medical Research Council Clinical Research training fellowship 2005-2007
Molecular studies of mouse fetal motor neurons in Kennedy's disease
MG Hanna and L Greensmith joint co-supervisors
Dr N Niranamathan
£135,000

Action Research Clinical Training fellowship 2005-2007
MG Hanna sponsor and supervisor
Brain calcium channel and neurological disease
Dr T Graves
£113,000

MRC component grant 2005-2008
MG Hanna Co investigator With Dr S Rahman
Nuclear genes and mitochondrial disease
£80,000

UCLH CRDC clinical training fellowship 2005-2006
Supervisors Prof MG Hanna, Prof Unwin and Prof Duchen
For Dr Andrew Hall "A molecular study of renal dysfunction in human mitochondrial disease"
£75000

2007
Wellcome Trust training fellowship Grant 2007–2009
The effect of alterations in the P/Q-type calcium channel in ataxia

Supervisor MG Hanna –Dr Rajakulendran
£111,812

2008
Medical Research Council Centre Grant
MRC Centre for Neuromuscular Diseases 2008-2013
PI and Centre Director MG Hanna
£3,471, 144
Co-applicants Muntoni, Turnbull, Bushby, Thompson, Reilly, Wood, Koltzenburg, Kullmann

Muscular Dystrophy Campaign Joint Centre Grant
2008-2011
£450,000
PI Muntoni Co-PI Hanna and Reilly

UK Brain Research Trust 2008-2009
Investigation of human neurological ion channel disorders
PI MG Hanna
£26,091

British Medical Association-Vera Down Award
Project Grant 2008–2009
Investigation of human neurological ion channels
PI MG Hanna
£50,000

Charities Aid Foundation Patrick Berthoud Clinical Research Fellowship 2008–2009
Investigation of human neurological ion nerve excitability
Supervisor MG Hanna to Susie Tomlinson
£115,000

Action Medical Research Project Grant 2008–2010
Investigation of human neurological ion nerve excitability testing protocols
Co-PI-MG Hanna with DM Kullmann
£150,000

Senexis (SME) Project Grant 2008–2010
Inclusion body Myositis UCL-Senexis collaboration
Co-PI MG Hanna with David Satelle and Linda Greensmith
£91,149

British Medical Association Vera Down Fellowship 2008 – 2009
Clinical and electrophysiological studies in muscle channelopathies
Supervisor MG Hanna
£50,000

2009
Medical Research Council MRC, Mitochondrial Disease Patient Cohort
2009-2011

£969,400
PI Turnbull
Co-PI Hanna Chinnery McFarland
(£340,000 to Hanna)

Medical Research Council Technology Project Grant 2009- 2010
New therapeutic approaches to IBM
Co-applicant with Linda Greensmith PI David Satelle
£154,658

Rare Diseases Research Network-NIH
Project grant
Genetic studies on voltage sensors in periodic paralysis
PI-Hanna
$89,000

2010
Medical Research Council Clinical Research Training Fellowship
Whole exome analysis in muscle channelopathies
2010 – 2014
£180,627
Supervisor MG Hanna
Fellow Dr Dipa Ryan

Arthritis Research Campaign
Arimoclomol trial to upregulate Heat Shock proteins in Inclusion Body Myositis
2010 – 2011
£133,413
PI MG Hanna

UCLH Fast Track Grant
Gene expression profiles in muscle with deleted mitochondrial DNA
2010-2011
£32,000
PI MG Hanna

National Institute of Health Research
Clinical trial of carbonic anhydrase in inhibitors in periodic paralysis
Clinical Local Research Network Contingency Funding
2010-2012
£67,540
PI MG Hanna

NIHR Clinical Research Network contingency funding
Arimoclomol for Sporadic Inclusion Body Myositis (IBM)
2010 - 2012
£66,641 Contingency Funding
PI MG Hanna

UCL Impact Studentship with private donor
2010-2013
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

UCL Impact Studentship
2010-2013
Mitochondrial dysfunction, impaired axonal transport and inherited neuropathy
£60,372 PhD studentship – Ellen Cottenie
Joint Primary Supervisor MG Hanna and MM Reilly

2011
UCL/UCLH Comprehensive Biomedical Research Centre
Regulation of mitochondrial proliferation in mitochondrial disease
2011 – 2012
£46,785
Co-PI with Michael Duchen

Current grant income and competitive NHS funding

1. National Specialist Commissioning Agency
   (NCG) Specialised Diagnostic and advisory Service for muscle channel diseases
   Commenced 2001-ongoing
   £466,706 per annum
   (Total £4,667,080 to date)
   Clinical Lead MG Hanna

2. National Specialist Commissioning Agency (NCG)
   Specialised Mitochondrial Disease Service
   Commenced 2006-ongoing
   £1,050,589 to Queen Square pa
   (Total -£5,150,000 to date)
   Clinical lead MG Hanna

3. Medical Research Council Centre Grant, MRC Centre for Neuromuscular Diseases
   2013-2017 renewed TOTAL >£6M
   £3,071, 144 MRC plus £3,321,456 joint BRC and host support
   PI and Centre Director MG Hanna
   Co-applicants Muntoni, Turnbull, Bushby, Thompson, Reilly, Wood, Koltzenburg, Kullmann

4. National Institutes of Health Clinical Investigation in Channelopathies
   2005 -2011 ongoing
   $1.4m
   PI Griggs Co-PI Hanna ($385,000 to Hanna)

5. National Institutes of Health
   Periodic Paralysis (Hyp-Hop) trial

2008–ongoing
$3.4m
Only UK Co-PI Hanna

6. Wellcome Trust Equipment Grant
2011 – 2016
Solid Next Generation Sequencing platform
£661,363
PI Houlden Co-PI Hanna, Reilly, Wood, Hardy

7. Medical Research Council Project Grant
Whole exome analysis in families with neuromuscular diseases
2011 – 2014
£320,000
PI-Houlden Co-PI Hanna and Reilly

8. OXION- Wellcome Integrative Physiology Ion Channel programme grant to Oxford University 2008-2013 £4.4 million- Programme renewal.
Co-Investigator on grant

9. Medical Research Council Project Grant
Whole exome analysis in Inclusion body myositis
2013 – 2017
£420,000
PI-Houlden Co-PI Hanna

10. FP7 Neuromics consortium
Work package: Whole exome analysis and molecular expression in muscle channelopathies
2012-2017
£186,000
PI Hanna

11. Action Medical Research
Mitochondrial quality control pathways as therapeutic targets in genetic mitochondrial disease
2013-2016
£192,243
PI Duchen Co-PI Hanna

12. UCL/UCLH CBRC Capital Bid
Next generation Sequencing bioinformatics platform
2011-2016
£339,000
PI Houlden Co-PI Hanna , Wood, Hardy, Reilly

13. UCL Impact Studentship with National Hospital Development Foundation
Aberrant cellular calcium handling and muscle degeneration in sodium channel disease.

2010-2014
£60,372 PhD studentship –Neta Amior
Primary supervisor Duchen secondary supervisor MG Hanna

14. UCL Impact Studentship with National Hospital Development Foundation
Mechanisms of phenotype diversity in muscle channelopathies
2010-2014
£60,372 PhD studentship-Siobhan Durran
Primary Supervisor Hanna Secondary supervisor Houlden

15. Muscular Dystrophy Campaign four year prize studentship
Whole exome analysis and molecular expression studies in periodic paralysis
2011–2015
£112,000 PA PhD studentship Alice Gardiner
Primary Supervisor MG Hanna Secondary Supervisor H Houlden

16. UCL Impact Studentship with Senexis
Heat Shock protein manipulation in the VCP mouse model of IBM
2011-2014
£60,372 PhD studentship – to be appointed
Primary Supervisor Linda Greensmith Secondary Supervisor MG Hanna

17. NIHR Rare Disease Translational Research Collaboration – IBM
2013-2015
£250,000
PI Hanna

18. NIHR Rare Disease Translational Research Collaboration
Postdoctoral Clinical Fellowship – IBM – Dr Pedro Machado
2014-2017
£401,333
PI Hanna

19. NIHR Rare Disease Translational Research Collaboration
Postdoctoral Clinical Fellowship – Channelopathies – Dr Emma Matthews
2014-2017
£363,060
PI Hanna

20. Medical Research Council Project Grant
Periodic paralysis: from molecules to mice
2014-2017
£464,146
PI Hanna

10. 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
Neurological channelopathies
Guarantors of Brain-Oxford

56. Sept 2006 - International Society of Neurophysiology annual conference, Edinburgh
Invited lecture - "Genetic neurological channelopathies"

Workshop organizer: “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. 24 October – Association of British Neurologists, Annual Meeting, Royal College of Physicians, London
"Inclusion body myositis: bench to bedside"

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 and doing 3 month sabbatical in my clinic NHNN

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 Nirmalananthan
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
Registered PhD UCL
"Whole exome analysis to identify new genes in muscle channelopathies"

2009–2014
MRC Centre for Neuromuscular Diseases Clinical Research Fellow
Dr Jasper Morrow
Registered PhD UCL
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)
Registered PhD
"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)
Registered PhD UCL
Jointly lead 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

2011-2014
UCL Impact Studentship with Senexis
Heat Shock protein manipulation in the VCP mouse model of IBM
£60,372 PhD studentship – to be appointed
Primary Supervisor Linda Greensmith Secondary Supervisor MG Hanna

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, Neurogenetics, University of Newcastle upon-Tyne

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 triplasmy"
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
"Syncolin 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 tow 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 Duchen 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 lead 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 additional 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 (manuscript under review).
- 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 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 Medicine341: 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 Karparti, (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. Epub 2007 Oct 11. 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. Epub 2008 Dec 31. 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. Epub 2010 Feb 1. 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 trials 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. doi: 10.1001/jama.2012.12607.

15b. Books/Thesis

1. **Hanna MG**, co-authored with Holdright D, Beck E, Souhami R-Tutorials in differential diagnosis 4th Edition Publishers Harcourt London 2003 pp245. [6000 copies sold worldwide]

2. **Hanna MG** MD thesis University of Manchester 1996, “Molecular genetic studies in human mitochondrial disease”. Supervisor *the late* Professor Anita Harding

15c. Chapters

1999

3. Rudel R, Hanna MG, Lehmann-Horn F (1999) Muscle ion channelopathies: Malignant Hyperthermia, Periodic Paralyses, Paramyotonia congenita and Myotonia. In muscle Diseases; Butterworth Heinmann Ed Schapira AHV and Griggs B*. pp 134-154*

2000

4. Hanna MG. 50 (2000) cases from the National Hospital. Ed; CD Marsden and A Wills. Martin Dunitz, London *(two cases pp23-26&pp56-59)*

2001

5. Davies NP Hanna MG (2001) Genetic neurological channelopathies. In; New treatments in Neurology Ed Scolding N Butterworth Heinemann London*. pp234-247*

6. Kinton L, Hanna MG, Wood NW (2001) Genetics of Epilepsy and Paroxysmal Movement Disorders. Epilepsy and Movement Disorders. Ed Guerinni, Aicardi, Andermann and Hallet. Cambridge University Press. *pp67-79*

2003

1. Davies NP Hanna MG (2003). Myopathies and Neurorehabilitation. In Textbook of neurorehabilitation Editor; Greenwood R *pp256-274*

2. Pulkes T Hanna MG. Mitochondrial diseases (2003). In ; Mitochondrial DNA and Human disease Ed Holt IJ Butterworth Heinemann *pp57-75*

3. Liolitsa D Hanna MG. Cell Models in mitochondrial disease. (2003) In Mitochondrial function and dysfunction in human disease. Ed Schapira AHV, DeLauro S; Butterworth Heinemann, London *pp89-110*

4. Hanna MG Peripheral neuropathy and mitochondrial diseases (2003) In Peripheral Neuropathy Eds Dyck D and Thomas PK; Saunders *pp1937-1949*

2004

5. Hanna MG. Disorders of Voluntary muscle. Oxford Textbook of Medicine 2004 (4th Ed).*pp1344-1346*

2009

6. Fontaine B and Hanna MG. Muscle ion channelopathies and related disorders (2009) In Disorders of Voluntary Muscle, 8th edn., eds. George Karpati, David Hilton-Jones, Kate Bushby and Robert C. Griggs. Published by Cambridge University Press *pp409-426*

2010

7. L Raja Rayan D and Hanna MG. Ion Channels and Human Disorders (2010) In ENCYCLOPEDIA OF LIFE SCIENCES & 2010, John Wiley & Sons, Ltd *pp1-17*

8. Fialho D Hanna MG-Periodic Paralysis-in Handbook of Neurology, Editors Hilton Jones DA, Laing D. Heinemann 2010 *pp 92-118*

9. Hanna MG and Parton M-Muscle Diseases in Queen Square Textbook of Neurology-Editors Shorvon, Howard, Rossor. (1st ed. pp.337-410). Chichester: Wiley-Blackwell.

10. Matthews E and Hanna MG - Cav1.1 hypokalaemic periodic paralysis in Pathologies of Calcium Channels, eds. Norbert Weiss and Alexandra Koschak, Springer 2013.

11. Hanna MG Chapter in Muscle Disease: Pathology and Genetics, 2nd Edition, eds. Hans H. Goebel, Caroline A. Sewry, Roy O. Weller, Wiley-Blackwell 2013.

In press

12. Hanna MG Neurogenetics *in* The Oxford Handbook of Molecular medicine Editor

D Lo, Oxford University Press *in press*

13. Hanna MG Fialho D;-Muscle channelopathies-in Paediatric channelopathies; Butterworths, Editors Zuberi S *in press*

14. Graves T Hanna MG- Channelopathies causing movement disorders in Paediatric Channelopathies Butterworths, Editors Zuberi *in press*

15. Hanna MG and Kullmann DM- Neurological channelopathies- a clinicians guide in Neurogenetics. Cambridge University Press-Ed NW Wood *in press*

16. Hanna MG–Diseases of the muscles in Medical Problems in Dentistry. Elsevier. Eds Scully C and Cawson R. *in press*

17. Burge J and Hanna MG - Muscle Channelopathies Neuromuscular Disorders. Eds Venance and Tawil *in press*

18. Matthews E and Hanna MG – Oxford Textbook of Neuromuscular Disorders. Oxford University Press 2013 *In press*

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.

3. **Hanna MG**, Nelson I, Sweeney MG, Cooper JM, Watkins PJ, Morgan-Hughes JA, Harding AE. Congenital encephalomyopathy and adult-onset myopathy and diabetes mellitus: different phenotypic associations of a new heteroplasmic mtDNA tRNA glutamic acid mutation. Am J Hum Genet. 1995 May;56(5):1026-33. PMID:7726155.

4. Nelson I, **Hanna MG**, Wood NW, Harding AE. Depletion of mitochondrial DNA by ddC in untransformed human cell lines. Somat Cell Mol Genet. 1997 Jul;23(4):287-90. PMID: 9542530.**Hanna MG**, Bhatia KP. Movement disorders and mitochondrial dysfunction. Curr Opin Neurol. 1997 Aug;10(4):351-6. Review. PMID: 9266161

5. Hanna M, Mills K, Pazdera L, Newsom-Davis J.Primary orthostatic tremor with prominent muscle hypertrophy. Neurology. 1997 Sep;49(3):872-4. PMID:9305357

6. Hammans SR, Sweeney MG, **Hanna MG**, Brockington M, Morgan-Hughes JA, Harding AE. The mitochondrial DNA transfer RNALeu(UUR) A-->G(3243) mutation. A clinical and genetic study. Brain. 1995 Jun;118 (Pt 3):721-34. PMID: 7600089.

7. **Hanna MG**, Nelson IP, Morgan-Hughes JA, Harding AE. Impaired mitochondrial translation in human myoblasts harbouring the mitochondrial DNA tRNA lysine 8344 A-->G (MERRF) mutation: relationship to proportion of mutant mitochondrial DNA. J Neurol Sci. 1995 Jun;130(2):154-60. Erratum in: J Neurol 1995 Sep;132(1):95. PMID: 8586979.

8. **Hanna MG**. Vitamin E deficiency. BMJ. 1995 Jun 24;310(6995):1673. PMID: 7795478.

1997

9. **Hanna MG**, Vaughan JR, Silburn PA, Davis PT, Greenhall RC, Squier MV, Mills KR, Renowden S, Sellar A. Two unusual clinical presentations of the mitochondrialDNA A3243G point mutation in adult neurological practice. J Neurol Neurosurg Psychiatry. 1997 May;62(5):544-6. PMID: 9153625.

1998

10. **Hanna MG**, Davis MB, Sweeney MG, Noursadeghi M, Ellis CJ, Elliot P, Wood NW, Marsden CD. Generalised chorea in two patients harboring the Friedreich's ataxia gene trinucleotide repeat expansion. Mov Disord. 1998 Mar;13(2):339-40.PMID: 9539351.

11. Leff AP, McNabb AW, **Hanna MG**, Clarke CR, Larner AJ. Complex partial status epilepticus in late-onset MELAS. Epilepsia. 1998 Apr;39(4):438-41. PMID:9578035.

12. **Hanna MG**, Nelson IP, Rahman S, Lane RJ, Land J, Heales S, Cooper MJ, Schapira AH, Morgan-Hughes JA, Wood NW. Cytochrome c oxidase deficiency associated with the first stop-codon point mutation in human mtDNA. Am J Hum Genet. 1998 Jul;63(1):29-36. PMID: 9634511.

13. **Hanna MG**, Stewart J, Schapira AH, Wood NW, Morgan-Hughes JA, Murray NM. Salbutamol treatment in a patient with hyperkalaemic periodic paralysis due to a mutation in the skeletal muscle sodium channel gene (SCN4A). J Neurol Neurosurg Psychiatry. 1998 Aug;65(2):248-50. PMID: 9703181.

14. **Hanna MG**, Wood NW, Kullmann DM. Ion channels and neurological disease: DNA based diagnosis is now possible, and ion channels may be important in common paroxysmal disorders. J Neurol Neurosurg Psychiatry. 1998 Oct;65(4):427-31. Review. PMID: 9771758.

15. **Hanna MG**, Nelson IP, Morgan-Hughes JA, Wood NW. MELAS: a new disease associated mitochondrial DNA mutation and evidence for further genetic heterogeneity. J Neurol Neurosurg Psychiatry. 1998 Oct;65(4):512-7. PMID: 9771776.

1999

16. Morgan-Hughes JA, **Hanna MG**. Mitochondrial encephalomyopathies: the enigma of genotype versus phenotype. Biochim Biophys Acta. 1999 Feb 9;1410(2):125-45. Review. PMID: 10076022.

17. Elliott PM, **Hanna MG**, Ward SA, Chinnery PF, Turnbull DM, Wood NW, McKenna WJ. Diagnostic utility of metabolic exercise testing in a patient with cardiovascular disease. Heart. 1999 Apr;81(4):441-3. PMID: 10092577.

18. Clark KM, Taylor RW, Johnson MA, Chinnery PF, Chrzanowska-Lightowlers ZM, Andrews RM, Nelson IP, Wood NW, Lamont PJ, **Hanna MG**, Lightowlers RN, Turnbull DM. An mtDNA mutation in the initiation codon of the cytochrome C oxidase subunit II gene results in lower levels of the protein and a mitochondrial encephalomyopathy. Am J Hum Genet. 1999 May;64(5):1330-9. PMID: 10205264.

19. Zuberi SM, Eunson LH, Spauschus A, De Silva R, Tolmie J, Wood NW, McWilliam RC, Stephenson JB, Kullmann DM, **Hanna MG**. A novel mutation in the human voltage-gated potassium channel gene (Kv1.1) associates with episodic ataxia type 1 and sometimes with partial epilepsy. Brain. 1999 May;122 (Pt 5):817-25. Erratum in: Brain. 2007 Mar;130(Pt 3):879. Stephenson, J P [corrected to Stephenson, J B]. Brain. 2010 May;133(Pt 5):1569. PMID: 10355668.

20. **Hanna MG**, Nelson IP. Genetics and molecular pathogenesis of mitochondrial respiratory chain diseases. Cell Mol Life Sci. 1999 May;55(5):691-706. Review. PMID: 10379358.

21. Spauschus A, Eunson L, **Hanna MG**, Kullmann DM. Functional characterization of a novel mutation in KCNA1 in episodic ataxia type 1 associated with epilepsy. Ann NY Acad Sci. 1999 Apr 30;868:442-6. PMID: 10414318.

22. Marques W Jr, **Hanna MG**, Marques SR, Sweeney MG, Thomas PK, Wood NW. Phenotypic variation of a new P0 mutation in genetically identical twins. J Neurol. 1999 Jul;246(7):596-9. PMID: 10463363.

23. Vergani L, Rossi R, Brierley CH, **Hanna M**, Holt IJ. Introduction of heteroplasmic mitochondrial DNA (mtDNA) from a patient with NARP into two human rho degrees cell lines is associated either with selection and maintenance of NARP mutant mtDNA or failure to maintain mtDNA. Hum Mol Genet. 1999 Sep;8(9):1751-5. PMID: 10441339.

24. Rahman S, Taanman JW, Cooper JM, Nelson I, Hargreaves I, Meunier B, Hanna MG, García JJ, Capaldi RA, Lake BD, Leonard JV, Schapira AH. A missense mutation of cytochrome oxidase subunit II causes defective assembly and

myopathy. Am J Hum Genet. 1999 Oct;65(4):1030-9. PMID: 10486321.

25. Andreu AL, **Hanna MG**, Reichmann H, Bruno C, Penn AS, Tanji K, Pallotti F, Iwata S, Bonilla E, Lach B, Morgan-Hughes J, DiMauro S. Exercise intolerance due to mutations in the cytochrome b gene of mitochondrial DNA. N Engl J Med. 1999 Sep 30;341(14):1037-44. PMID: 10502593.

26. Pulkes T, Eunson L, Patterson V, Siddiqui A, Wood NW, Nelson IP, Morgan-Hughes JA, **Hanna MG**. 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. 1999 Dec;46(6):916-9. Erratum in: Ann Neurol 2000 Jun;47(6):841. PMID: 10589546.

27. Davies NP, **Hanna MG**. Neurological channelopathies: diagnosis and therapy in the new millennium. Ann Med. 1999 Dec;31(6):406-20. Review. PMID: 10680855.

2000

28. Rahman S, Lake BD, Taanman JW, **Hanna MG**, Cooper JM, Schapira AH, Leonard JV. Cytochrome oxidase immunohistochemistry: clues for genetic mechanisms. Brain. 2000 Mar;123 Pt 3:591-600. PMID: 10686181.

29. Davies NP, Eunson LH, Gregory RP, Mills KR, Morrison PJ, **Hanna MG**. Clinical, electrophysiological, and molecular genetic studies in a new family with paramyotonia congenita. J Neurol Neurosurg Psychiatry. 2000 Apr;68(4):504-7. Erratum in: J Neurol Neurosurg Psychiatry 2000 Jul;69(1):139. PMID: 10727489.

30. Münchau A, Valente EM, Shahidi GA, Eunson LH, **Hanna MG**, Quinn NP, Schapira AH, Wood NW, Bhatia KP. A new family with paroxysmal exercise induced dystonia and migraine: a clinical and genetic study. J Neurol Neurosurg Psychiatry. 2000 May;68(5):609-14. PMID: 10766892.

31. Cottrell DA, Ince PG, Blakely EL, Johnson MA, Chinnery PF, **Hanna M**, Turnbull DM. Neuropathological and histochemical changes in a multiple mitochondrial DNA deletion disorder. J Neuropathol Exp Neurol. 2000 Jul;59(7):621-7. PMID: 10901234.

32. Eunson LH, Rea R, Zuberi SM, Youroukos S, Panayiotopoulos CP, Liguori R, Avoni P, McWilliam RC, Stephenson JB, **Hanna MG**, Kullmann DM, Spauschus A. Clinical, genetic, and expression studies of mutations in the potassium channel gene KCNA1 reveal new phenotypic variability. Ann Neurol. 2000 Oct;48(4):647-56. PMID: 11026449.

33. Pulkes T, Siddiqui A, Morgan-Hughes JA, **Hanna MG**. A novel mutation in the mitochondrial tRNA(TYr) gene associated with exercise intolerance. Neurology. 2000 Oct 24;55(8):1210-2. PMID: 11071502.

34. Pulkes T, Sweeney MG, **Hanna MG**. Increased risk of stroke in patients with the A12308G polymorphism in mitochondria. Lancet. 2000 Dec 16;356(9247):2068-9. PMID: 11145497.

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