Curriculum Vitae

Professor Michael G Hanna

2018

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

Current positions:

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

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

Director, UCL MRC Centre for Neuromuscular Diseases

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

Honorary Consultant Neurologist, Great Ormond Street Hospital NHS Trust.

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

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

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

Recent positions

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

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

Founding Chairman, British Myology Society (2008-2016)

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

Curriculum Vitae Contents

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

Leadership Roles

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

Academic Activity: basic discovery science and translational research

- 1995-2017 Published over 275 peer reviewed papers and reviews, over 150 published abstracts, 17 book chapters and 1 book. H-Index 52.
- 1998-2017 Obtained over £22 million external basic and translational research funding including MRC, Wellcome Trust plus £8million NHS NCG funding.
- 1998-2017 Supervised 35 clinical and non-clinical PhD research fellows/students and 3 post doctoral research fellows
- 2007-2020 Successfully led joint initial application and renewal, across UCL and Newcastle and direct joint UCL Newcastle MRC Centre for Neuromuscular Diseases->£10m
- 2019-2025 Lead PI on MRC strategic award to establish an international centre for genomic medicine in neuromuscular diseases £3.66m from MRC and £2m host universities

Editorships and other esteem

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

National Neuromuscular Clinical Services

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

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

Undergraduate Neurology Teaching

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

Private Fundraising and Philanthropy

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

Clinical Management and Neuromuscular Clinical Services

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

Specialist Clinical Interests

- I have an international profile in clinical neuromuscular muscle wasting neurological diseases; especially mitochondrial diseases and channelopathies and also muscular dystrophy, peripheral neuropathy, motor neuron diseases, myasthenia gravis, and all genetic neuromuscular diseases.
- 20 years front line consultant general neurologist
- 20 years medicolegal practice: includes personal injury and negligence; 60% plaintiff

Full Curriculum Vitae

2. Personal Details

Michael G. Hanna 04.10.63

3. Education/Qualifications

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

4. Current Post

Director UCL Institute of Neurology 2012-present
I am Director of UCL institute of Neurology (IoN) which is the largest Neurology research Institute in the UK and one of the largest in the world. It has over 900 academic staff and currently has £258m in active grants. I have driven a new academic strategic plan to increase translation of science into patient benefit. This has included successful award of £20m from the Wolfson Foundation for a new experimental neurology centre at Queen Square resulting in a 500% increase in experimental neurology trials and multiple new industry partnerships and investments. In our successful REF bid 2014 IoN was number 1 in the UK for Neurology research strength. I have supported research embedded education development which now includes 500 MSc and PhD students, with very high student rating/experience scores. I successfully lead the Athena Swan Silver award to the Institute in 2015. I have successfully recruited many senior academics to the Institute including the first Nobel prize winner to join the Institute, Jim Rothman 2014. I have worked closely with UCL partners academic health sciences centre (as Co-Director of neurological disease theme) to develop population level impact especially in stroke and dementia. I recently lead for IoN in capital fundraising programme that has successfully raised £240m to rebuild the main IoN wet lab science building; building work will commence 2019. Under my leadership I want to continue to maximize the opportunities for science to benefit all patients with neurological diseases. I was recently central to the successful award of the MRC UK Dementia Research Institute to UCL at IoN and I lead a successful capital bid to HEFCE for £29m for capital towards new IoN building.

My current consultant position also includes:

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

(£1.3m pa NHS funding).

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

My research has focused on improved understanding of the molecular genetics basis and molecular mechanisms of neurological diseases caused by mitochondrial dysfunction and by ion channel dysfunction. Many of these diseases particularly affect the neuromuscular system and my clinical specialist interest has focused on developing better services for patients with neuromuscular neurological diseases. Wherever possible, I have aimed to translate my research findings into clinical practice. I successfully established an MRC Centre for translational research in neuromuscular diseases in 2008 which is now a national UK platform and resource that has had major impact in several areas including clinical and no n clinical PhD training, new gene discovery, biobanking, patient cohorts and clinical trials in neuromuscular diseases.

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

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

Ion Channel Research Programme-Summary and Impact

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

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

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

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

- We have shown how genetic dysfunction of muscle sodium, potassium, calcium or chloride channels can relate to specific episodic muscle conditions including periodic paralysis, muscle stiffness syndromes and episodic cardiac arrhythmias.

- We have shown that the genetic architecture of the commonest form of periodic paralysis predicts the presence of an abnormal gating pore current in muscle sodium or calcium channels supporting the presence of a brand new mechanism for disease causation and target.

- We defined previously unrecognized neonatal ion channel diseases including intermittent hyoptonia and stridor–knowledge of which has lead to improved and safer care of affected patients in the neonatal period.

- We identified mutations in important presynaptic neuronal potassium and calcium channels can lead to episodic ataxia- a disorder characterized by profound disabling attacks of unpredictable unsteadiness. Furthermore, we have shown that such patients are 17 times more likely to develop epilepsy compared to the background population risk.

- We provided evidence that muscle sodium channel dysfunction is a risk factor for sudden infant death syndrome.

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

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

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

Mitochondrial Research Programme-Summary and Impact

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

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

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

The UCL MRC Centre for Neuromuscular Diseases 2008-2018

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

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

The main programmes of research within the centre build on existing funded themes currently attracting in excess of £18m of grant income, are developing new cross cutting

collaborations and has facilitated and is now capitalizing on the recruitment of world class senior academic personnel to UCL. All the main programmes of research impact upon and benefit from the following key areas that are being newly developed in the centre. [The underdevelopment of these key areas is a current "block" to effective UK translational research in neuromuscular disease];

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

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

Strategy and objectives 2008-2020
We are building on existing world class basic and clinical scientists at the Institute of Neurology, the Institute of Child Health and across UCL and have recruited three new world class senior clinical and basic research staff to establish an unrivalled critical mass of expertise in the UK.

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

- We are add value to existing excellence in education and training of basic and clinical neuromuscular scientists of the future thereby building the UK's future capacity
- We will take capitalize on the clinical resource at the co-located Neuromuscular centres of Great Ormond Street NHS Trust and National Hospital for Neurology, especially within two Department of Health (National Specialist Commissioning Group) funded centres for children and adults with rare neuromuscular diseases
- We are collaborating widely in the UK and internationally to form patient and scientist networks.

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

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

4b. Clinical Services

General Neurology Service

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

Specialist Clinical Service-The National Hospital Centre for Neuromuscular Disease

- I have established a centre for neuromuscular disease with colleagues at the National Hospital in order to enhance services to patients with muscle and peripheral nerve diseases. The center now comprises over 60 clinical and research staff. It represents a critical mass of clinical neurologists with expertise in muscle and nerve disease, neurophysiologists, specialist nursing staff and a state of the art computerized myometry system as well as support staff. We moved into new state of the art facilities in 2009 financed by a £2m charity fund raising project which I lead.

- I run the bi-weekly muscle-genetic clinic which has a regional and national referral base for patients with acquired and genetic muscle disease.

- I obtained funding from the Department of Health [National Specialist Commissioning Group] to support the muscle channel and mitochondrial components of this muscle service. This now amounts to £1.4m per annum on a recurring basis. This NCG funding allows me to provide a comprehensive, clinical electrophysiological and genetic service to this group of patients from anywhere in

the UK without cost implications to the local referring centre. The NCG services I offer are part of a National consortium for the diagnosis of rare neuromuscular diseases which I established with colleagues in Oxford, Newcastle and London.

- Neurological trainees as well as consultant colleagues from the UK and internationally frequently attend my clinic.

- I run a joint inflammatory muscle disease clinic with Professors Isenberg and Schapira for complex cases six times per year.

- I am an honorary consultant at Great Ormond Street and undertake joint transition neuromuscular clinics with Professor Muntoni.

4c Clinical Management from 2002–2012

- I was Associate Divisional Clinical Director from 2002 to 2007 and worked closely with Alan Thompson to produce the NHNN clinical service strategy document 2005-2008.

- I was Queen Square Divisional Clinical Director since 2007-12 and in collaboration with the Divisional Manager-Jackie Sullivan I had responsibility for Divisional financial performance and activity, quality, workforce, governance, estates and clinical strategy.

- The management team I lead produced the NHNN strategy document 2009-2014 in conjunction with key partners in the context of UCLP (see appendix).

- I have an intimate knowledge of all clinical and research services at NHNN and strong links UCLH Trust-wide.

- I work closely and was jointly responsible with the National Hospital Divisional manager for all clinical, staffing financial and governance issues in the Hospital. The budget revenue was £143m pa and the workforce 1245 whole time equivalent including 160 consultant staff.

- I am proud of the achievements delivered with the team under my leadership over the past four years including:

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

  Progressive improvement in absolute year-end financial position

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

2008 Hyper Acute Stroke Unit-successful application to Health Care for London and delivery of a single unified HASU in the North Central sector now managed by Queen Square. Centralization of all sector consultants into one team and already resulted in a reduction in stroke 28 day in-patient mortality. High throughput of fully phenotyped patients is a major opportunity for many types of stroke clinical research.

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

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

5. Professional History

Aug 88-July 89
Preregistration Positions
Professorial House Physician General Medicine and Neurology
Professor D Neary & Professor S Tomlinson, Manchester Royal Infirmary
Professorial House Surgeon
Professor Sir Miles Irving, Hope University Hospital, Manchester

Aug 89-Jan 92
Senior House Officer Positions
Newcastle-upon-Tyne central teaching medical rotation.
General medicine & endocrinology. Dr WMG Tunbridge, Newcastle General Hospital
General medicine & gastroenterology. Dr C Record, Royal Victoria Infirmary
Cardiology Professor F Campbell, Freeman Hospital
Chest medicine Professor J Gibson & Professor P Corris, Freeman Hospital
Neurology Dr P Hudgson & Dr J Foster, Newcastle General Hospital
Renal medicine Professor R Wilkinson, Freeman Hospital

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

Feb 92-July 92
Registrar/Clinical Lecturer
General medicine & geratology, Nuffield Department of Clinical Medicine, Oxford University
Professor J Grimley-Evans, John Radcliffe Hospital & Radcliffe Infirmary.

Aug 92-July 94

MRC Research Fellow
Medical Research Council clinical training fellow,
Mitochondrial DNA in human mitochondrial disease
University Department of Clinical Neurology, Institute of Neurology, Queen Square, London;
Supervisor *the late* Professor Anita E Harding

Aug 94-April 96
Registrar in Clinical Neurology
University & NHS Departments of Clinical Neurology, Radcliffe Infirmary, Oxford
Drs R Greenhall, M Donaghy, D Hilton-Jones, N Hymen, J Oxbury, C Fursden-Davis, G Wright, P Davis, P Matthews & Professor Newsom-Davis.

May 96-Nov 96
Registrar in Clinical Neurology
The National Hospital for Neurology and Neurosurgery, Queen Square, London
Professors Shorvon, Duncan, Fish, Marsden, Frackowiak, Schapira Quinn, Wood and Goadsby

Nov 96-January 98
Senior Registrar in Neurology and Clinical Lecturer in Neurogenetics
The Institute of Neurology and the National Hospital for Neurology and Neurosurgery
Queen Square and St Mary's Hospital, Paddington, London
Professors Rossor and Wood and Drs Farmer, Thomas and Ball

6. Teaching Education Role

Undergraduate medical student clinical neurology teaching course 1998-2007

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

- My work in undergraduate teaching was recognized by the award of "Top clinical teacher" in 1999-2000 voted for by the clinical medical students. In 2002 Queen Square voted best undergraduate teaching site –UCL medical students.
- I co-authored the fourth edition of the well known textbook "Tutorials in Differential Diagnosis" now reprinted in several languages and with sales > 10,000

Postgraduate education roles

- I give lectures on the IoN MSc courses
- The MRC Centre has a major high profile postgraduate education role which I organise with colleagues in the Centre and includes:

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

7. Other Appointments and Affiliations

Deputy Editor, Journal of Neurology, Neurosurgery and Psychiatry 2004-2015
Involved daily running of Journal and handling 2600 manuscripts per year with the Editor and three associate editors. Progressive rise in Impact factor from 3.1 to 4.791.

Membership of Societies-advisory panels

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

I regularly review papers and books for the following journals: Brain; Human Molecular Genetics; Journal of Neurology; Journal of Neurology, Neurosurgery and Psychiatry; Journal of Medical Genetics; Movement Disorders; European Journal of Neurology; Genomics, Annals of Human Genetics, Annals of Neurology, European Journal of Human

Genetics, Journal of Neurological Sciences, Neuroscience, American Journal of Human Genetics, Neuropediatrics, American Journal of Medical Genetics, Journal of Clinical Investigation and Nature Genetics.

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

8. Prizes, Awards and other Honours

Mabel Harriet Florence Smith travel scholarship 1987
Awarded by Leeds City Council, to best medical student elective, on the basis of a written submission and an oral presentation.

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

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

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

Guarantors of Brain 2009
Elected Guarantor of Brain 2009

American Neurology Association 2010
Elected corresponding member 2010

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

9. Grants

Previous Grant Funding:

1992-1994	Medical Research Council: Training Fellowship The study of the molecular pathogenesis of primary defects of mtDNA using a human myoblast culture system £60,000 MG Hanna - Supervisor Prof AE Harding
1996-1997	Joint Research Advisory Committee Institute of Neurology: Mechanisms of phenotypic diversity associated with the mitochondrial A3243G point mutation PI Hanna £35,000
1997-1999	Brain Research Trust: Expression studies of human neurological channelopathies

	Co-I Hanna £90,000
1998-1999	Wellcome Trust: entry level fellowship for medical research fellow Studies of mtDNA in human Neurological disease Hanna sponsor and supervisor for Dr Siddique £57,000
1999-2000	Wellcome Trust: entry level fellowship for medical research fellow Molecular genetic studies of human skeletal muscle channelopathies Hanna sponsor and supervisor for Dr N Davis £59,000
1999-2001	Epilepsy Research Foundation: Clinical, Genetic and Expression studies of the voltage gated calcium channel (CACNA1A) in complex human epilepsy phenotypes Hanna Principal Applicant £50,000
1999-2001	Brain Research Trust: Molecular basis of phenotypic heterogeneity associated with mtDNA mutations Hanna principal applicant £55,000
1999-2004	MRC: Mitochondrial cooperative - mitochondria in health and disease Cooperative status Hanna co-principal applicant with Prof JB Clarke, Prof M Duchen, Prof Wood, Prof Rich, Prof Crompton and Prof Moncado-UCL
2000-2003	MRC: Mitochondrial cooperative – component grant Identifying nuclear genes which influence the expression of mtDNA mutations Hanna-principal applicant £180,000
2000-2003	Wellcome Trust: Training fellowship: Molecular genetic and expression studies on muscle chloride channelopathies Hanna sponsor and supervisor for N Davies £120,000
2000-2002	Special Trustees of the UCLH NHS Trust: Clinical, genetic and expression studies of the voltage gated calcium channel CACNA1A in neurological disease Hanna principal applicant £69,000
2001	National Institutes of Health [USA]: Research proposal planning grant - An international trial of treatment in skeletal muscle channelopathies USA PI- Professor RC Griggs and UK Co PI - Hanna US$35,000
2003-2007	Kennedy's disease research fund grant: Clinical research fellow undertook molecular genetic research into Kennedy's disease Co-PI Hanna £75,000
2003-2005	MRC Cooperative: Epilepsy. Ion channels in epilepsy Component grant DM Kullmann, NW Wood and MG Hanna £375,000
2004-2008	National Institutes of Health [USA]: Clinical Investigations into Neurological Channelopathies "CINCH" UK Centre International multicentre study in neurological channelopathies Funded one clinical research fellow salary per year for 5 years Hanna CoPI £250,000
2003-2007	Wellcome Trust: Integrative Physiology Ion Channel Programme grant to Oxford University OXION. Hanna Co-I £3.4million.
2004-2009	National Institutes of Health [USA]: An international trial of pharmacological intervention in periodic paralysis. USA PI and Coordinator Professor RC Griggs. Only UK centre Co-PI Hanna. US$3.1million
2004-2005	Guarantors of Brain: Research fellowship for Dr Tracey Graves. Supervisor Hanna. £55,000

2004	UCL Special Trustees: Fast track Award for Post Doc "Nuclear genes and mitochondrial disease". Hanna principal applicant and supervisor £29,000
2005-2007	Medical Research Council: Clinical Research Training Fellowship 'Molecular studies of mouse fetal motor neurons in Kennedy's disease' Dr N Niranamathan. Hanna and Greensmith joint supervisors £135,000
2005-2007	Action Research: Clinical Training Fellowship 'Brain calcium channel and neurological disease' Dr T Graves. Hanna sponsor and supervisor £113,000
2005-2008	Medical Research Council: Component Grant 'Nuclear genes and mitochondrial disease' Hanna Co-I with Dr S Rahman. £80,000.
2005-2006	UCLH CRDC: Clinical Training Fellowship 'A molecular study of renal dysfunction in human mitochondrial disease' Dr Andrew Hall. Supervisors Hanna, Prof Unwin and Prof Duchen. £75,000
2007-2009	Wellcome Trust: Training fellowship 'The effect of alterations in the P/Q-type calcium channel in ataxia' Dr Sanjeev Rajakulendran. Supervisor Hanna. £111,812
2008-2011	Muscular Dystrophy Campaign: Joint Centre Grant. PI Muntoni Co-PI Hanna and Reilly £450,000
2008-2009	UK Brain Research Trust: Investigation of human neurological ion channel disorders PI Hanna £26,091
2008-2009	British Medical Association: Vera Down Award, Project Grant: Investigation of human neurological ion channels PI Hanna £50,000
2008–2009	Charities Aid Foundation: Patrick Berthoud Clinical Research Fellowship to Dr S Tomlinson 'Investigation of human neurological ion nerve excitability' Supervisor MG Hanna £115,000
2008–2010	Action Medical Research: Project Grant 'Investigation of human neurological ion nerve excitability testing protocols' Co-PI-MG Hanna with DM Kullmann £150,000
2008–2010	Senexis (SME): Project Grant 'Inclusion body Myositis UCL-Senexis collaboration' Co-PI MG Hanna with David Satelle and Linda Greensmith £91,149
2008 – 2009	British Medical Association: Vera Down Fellowship Clinical and electrophysiological studies in muscle channelopathies Supervisor Hanna £50,000
2008-2013	OXION-Wellcome: Integrative Physiology Ion Channel programme grant to Oxford University. Hanna Co-Investigator £4.4 million- Programme renewal.
2009-2011	Medical Research Council: Mitochondrial Disease Patient Cohort PI Turnbull Co-PI Hanna Chinnery McFarland £969,400
2009- 2010	Medical Research Council: Technology Project Grant, 'New therapeutic approaches to IBM Co-applicant with Linda Greensmith PI David Satelle £154,658
2009	Rare Diseases Research Network-NIH: Project grant, 'Genetic studies on voltage sensors in periodic paralysis'

	PI Hanna US$89,000
2010 – 2014	Medical Research Council: Clinical Research Training Fellowship 'Whole exome analysis in muscle channelopathies' Fellow Dr Dipa Rayan Supervisor MG Hanna £180,627
2010 – 2011	Arthritis Research Campaign: 'Arimoclomol trial to upregulate Heat Shock proteins in Inclusion Body Myositis' PI Hanna £133,413
2010-2011	UCLH Fast Track Grant: 'Gene expression profiles in muscle with deleted mitochondrial DNA ' PI Hanna £32,000
2010-2012	National Institute of Health Research: 'Clinical trial of carbonic anhydrase in inhibitors in periodic paralysis' Clinical Local Research Network Contingency Funding PI Hanna £67,540
2010 - 2012	NIHR: Clinical Research Network contingency funding Arimoclomol for Sporadic Inclusion Body Myositis (IBM) PI Hanna £66,641
2010-2013	UCL Impact Studentship with private donor: Heat shock protein upregulation therapy in the Kennedys Disease mouse model. PhD studentship Anna Gray Primary Supervisor Greensmith, Secondary supervisor Hanna £60,372
2010-2013	UCL Impact Phd Studentship: 'Mitochondrial dysfunction, impaired axonal transport and inherited neuropathy' Ellen Cottenie. Joint Primary Supervisor Hanna and MM Reilly £60,372
2010-2014	UCL Impact PhD Studentship with National Hospital Development Foundation: 'Mechanisms of phenotype diversity in muscle channelopathies' Siobhan Durran Primary supervisor Hanna, Secondary supervisor Houlden £60,372
2011 – 2012	UCL/UCLH Comprehensive Biomedical Research Centre: 'Regulation of mitochondrial proliferation in mitochondrial disease' Co-PI with Michael Duchen £46,785
2011 – 2014	Medical Research Council: Project Grant 'Whole exome analysis in families with neuromuscular diseases' PI Houlden Co-PI Hanna & Reilly £320,000
2011 – 2016	Wellcome Trust: Equipment Grant 'Solid Next Generation Sequencing platform ' PI Houlden Co-PI Hanna, Reilly, Wood, Hardy £661,363
2011-2016	UCL/UCLH CBRC: Capital Bid 'Next generation Sequencing bioinformatics platform' PI Houlden Co-PI Hanna , Wood, Hardy, Reilly £339,000
2011–2015	Muscular Dystrophy Campaign: four year PhD studentship 'Whole exome analysis and molecular expression studies in periodic paralysis' Alice Gardiner Primary Supervisor Hanna Secondary Supervisor Houlden £112,000
2012-2017	FP7 Neuromics consortium: Work package - Whole exome analysis and molecular expression in muscle channelopathies PI Hanna £186,000
2012-2017	European Union: FP7 Neuromics consortium: Work package - Whole

	exome analysis and molecular expression in muscle channelopathies £186,000 PI Hanna
2013-2015	NIHR Rare Disease Translational Research Collaboration – IBM PI Hanna £250,000
2013-2016	Action Medical Research: Mitochondrial quality control pathways as therapeutic targets in genetic mitochondrial disease PI Duchen Co-PI Hanna £192,243
2013 – 2017	Medical Research Council: Project Grant 'Whole exome analysis in Inclusion body myositis' PI-Houlden Co-PI Hanna £420,000
2013 – 2017	UCLH BRC: Fast Track Grant 'A randomized, double-blind, placebo controlled phase IIa experimental pilot trial assessing efficacy of a single dose or repurposed bumetanide in genetically defined hypokalaemic periodic paralysis assessed using the electyrophysiological McManus protocol' Co-I Hanna £40,000
2014-2016	UCL Impact PhD Studentship with National Hospital Development Foundation: 'Aberrant cellular calcium handling and muscle degeneration in sodium channel disease' Neta Amior Primary supervisor Duchen secondary supervisor Hanna £60,372
2014-2017	NIHR Rare Disease Translational Research Collaboration: Postdoctoral Clinical Fellowship – IBM – Dr Pedro Machado PI Hanna £401,333
2014-2017	NIHR Rare Disease Translational Research Collaboration Postdoctoral Clinical Fellowship – Channelopathies – Dr Emma Matthews PI Hanna £363,060
2014-2017	Medical Research Council: Project Grant 'Periodic paralysis: from molecules to mice' PI Hanna £464,146
2014-2017	National Institutes of Health: Clinical Investigation in Channelopathies $1.4m PI Griggs Co-PI Hanna (US$385,000 to Hanna)
2015-2017	Novartis trial: Arimoclomol for Inclusion Body Myositis PI Hanna £345,324
2015-2018	The Lily Foundation: Clinical Research Fellowship Dr Olivia Poole £124,000
2016-2017	Medical Research Council: MYOPROSP Consortium PI Hanna £66,301

Current grant income and competitive NHS funding:

2001 – ongoing	National Specialist Commissioning Agency (NCG) / Highly Specialised Service (HSS): Specialised diagnostic and advisory Service for muscle channel diseases. £466,706 per annum (Total £4,667,080 to date) Clinical Lead MG Hanna
2006 – ongoing	National Specialist Commissioning Agency (NCG) / Highly Specialised Service (HSS); Specialised Mitochondrial Disease Service. £1,050,589 to Queen Square per annum (Total £5,150,000 to date)

	Clinical lead MG Hanna
2008 – 2020	Medical Research Council Centre Grant: MRC Centre for Neuromuscular Diseases renewed Total >£6M £3,071,144 MRC plus £3,321,456 joint BRC and host support. PI and Centre Director MG Hanna
2013 - 2017	GOS NIHR BRC: Matched funding for MRC Centre for Neuromuscular Diseases Hanna PI £282,714
2013 – 2018	UCLH NIHR BRC: Matched funding for MRC Centre for Neuromuscular Diseases. Hanna PI £493,106
2015-2018	MRC: Clinical Training Fellowship Dr Karen Suetterlin £185,000
2015-2020	Wellcome Trust Strategic Award: Synaptopathies: genetics, biophysics and circuit mechanisms of paroxysmal neurological disorders £4,194,451 PI Kullmann Co-PI Hanna
2016-2019	MRC: Clinical Training Fellowship Dr Helen Devine Hanna Sponsor £176,000
2017-2018	The Lily Foundation: Lily-Stoneygate Research Awards Programme – treatments for mitochondrial disease Hanna PI £83,686
2017-2019	Muscular Dystrophy UK: An experimental medicine study to assess the safety and efficacy of 2-deoxyglucose in patients with m.3243G mutant mitochondrial DNA Hanna PI £146,520
2017-2021	UCLH/NIHR BRC: Neuromuscular Disease Theme Hanna PI £398,514
2018-2022	FDA USA: Phase II/III Study of Arimoclomol in Inclusion Body Myositis (IBM) PI Hanna US$214,235
2018-2022	European Union: SOLVE-RD Co-PI Hanna €20,000
2018-2020	Wellcome Trust: Clinical Research Career Development Fellowship 'Skeletal muscle channelopathies: severe infantile phenotypes and sudden infant death syndrome' Dr Emma Matthews Hanna Sponsor £283,711
2019-2024	Medical Research Council: MRC Strategic Award to establish an International Centre for Genomic Medicine in Neuromuscular Diseases PI Hanna £3,139,610
2018-2021	The Clore Duffield Foundation: Clinical Research Fellowship Hanna PI £234,000
2019-2024	Medical Research Council: Clinician Scientist Fellowship; Lipid metabolism in mitochondrial diseases, Dr R Pitceathly Hanna Sponsor £1,001,993

10. Selected Invited National and International Lectures

1995

1. April 1995: Oxford University Department of biochemistry invited seminars
   "New mitochondria DNA mutations and myoblast studies on their molecular pathogenesis"

2. Sept 1995: Oxford University, Nuffield Department of Pathology, invited seminar
   "The emerging role of mitochondria in human disease"

1997

3. April 1997: Department of Clinical Neurology, University of Queensland, Brisbane, Australia "*Channelopathies: A new name for old diseases*"

4. May 1997: Institute of Neurology short course in Neurogenetics
   "Channelopathies"

5. Nov 1997*:* Recent advances in Epilepsy, Glaxo Wellcome, Paris
   "Modern Genetics of Epilepsy"

6. Nov 1997: Institute of Child Health, UCL- research seminar
   "*Ion channel disorders of skeletal muscle"*

7. Dec 1997: UCL centre for human genetics annual meeting
   "Ion channels and epilepsy"

1998

8. Jan 1998: Department of Neurology, Kings College Hospital research seminar
   "Mitochondrial genetics: focus on myopathies"

9. March 1998: Glaxo Wellcome centre for neuromuscular disease annual meeting, Liverpool
   "Recent advances in the genetics of neuromuscular diseases"

10. March 1998: Annual Neuromuscular Symposium, Walton Centre for Neurology, Liverpool
    "*Skeletal muscle channelopathies"*

11. Sept 1998: Annual meeting of the European Federation of Neurology, Seville, Spain, Scientific symposium
    "*Channelopathies: a new field"*

12. Sept 1998: Association of British Neurologists autumn meeting, scientific symposium
    "*Ion channels and neurological disease"*

13. Oct 1998: International league against epilepsy, Annual Scientific Meeting, Oxford
    "Ion channels and paroxysmal disorders"

14. Oct 1998: Oxford clinical genetics seminars
"Developing a skeletal muscle ion channel DNA diagnostic service"

15. Nov 1998: Department of Neurology, Leeds University
"Skeletal muscle ion channel disorders"

1999

16. Feb 1999: Annual Salpetriere Queen Square Neurology meeting, Paris
"New mutational mechanisms in mitochondrial diseases"

17. March 1999: Dept Neurology, Addenbrooke's Hospital, Cambridge
"*Neurological channelopathies-the future"*

18. March 1999: Neurology for Neuroscientists, Magdalen College Oxford
"Ion channels and Neurological Disease"

19. April 1999: Annual Clinical Genetics Day, Institute of Child Health, Great Ormond Street *"Mitochondrial Deafness"*

20. April 1999: International League against Epilepsy, British branch, annual meeting, Birmingham, *Ion Channels and Epilepsy-the future"*

21. June 1999: Muscle Course, Hammersmith Hospital, London
"Molecular genetics of inherited myotonic disorders"

22. June 1999: Dept Neurology, Western General Infirmary, Edinburgh
"Neurological Channelopathies"

2000

23. Jan 2000: British Paediatric Neurologists Association annual meeting, Glasgow
"Ion channel dysfunction-a common problem in epilepsy"

24. Jan 2000: Institute of Neurology-UCL-Annual Glaxo Lecture
"Ion channels and neurological disease"

25. March 2000: University of Sheffield, Dept of Neurology
"*Neurological channelopathies"*

26. April 2000: National neurology meeting Glaxo Wellcome-Stratford
"*Molecular mechanisms in paroxysmal neurological disease"*

27. May 2000: University of Birmingham, Dept of Neurology
"*Ion channels and Neurological diseases"*

28. June 2000: University of Birmingham, Dept Clinical Genetics
"The genetic ataxias"

29. Sept 2000: Salpetriere Hospital, Dept Neurology, Paris
"Molecular mechanisms in mitochondrial disease"

30. October 2000: University of Bologna, Dept of Neurology, Italy
"Brain potassium channel dysfunction and neurological disease"

2001

31. Feb 2001: Institute of Child Health, Dept metabolic medicine, UCL
"Paediatric channelopathies"

32. April 2001: Advanced Neuromuscular course, Hammersmith Hospital
"Skeletal muscle channelopathies"

33. July 2001: University of Sheffield, Channelopathies 2001
"*Human neurological disease and ion channel dysfunction"*

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

2002

35. April 2002: University of Milan, Dept Neurology
"*Calcium channels and epilepsy"*

36. July 2002, International Congress on Neuromuscular disease, Vancouver Canada workshop-
"*Painful muscle channelopathies"*

37. September 2002, Birmingham Heartlands Hospital NSCAG meeting
"*The National diagnostic service for muscle channelopathies"*

38. October 2002, Atkinson's Morley's Hospital, Dept of Neurology,
"*An Update on Brain Channelopathies*"

2003

39. March 2003, Royal Society Medicine, London
"Episodic Ataxias-mechanisms in human cerebellar channelopathies"

40. March 2003 University of Oxford Wellcome Trust ion channel initiative
"*Human brain channelopathies*"

41. November 2003 Bath Advanced Neurology course
"Neurological channelopathies"

42. November 2003 Goulstonian Lectureship

Royal College of Physicians, London
"Neurological channelopathies"

43. March 2003 UCL annual rheumatology course
"Inflammatory myopathies"

44. May 2003 Dept Neurology University Nottingham
"Muscle channelopathies"

2004

45. June 2004 Dept Neurology, University of Bonn
"*Nuclear Mitochondrial disease"*

46. September 2004 EFNS Paris
Invited to organize workshop
"Neurological channelopathies"

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

2005

48. April 2005 Dept Neurology, University of Bristol
"Neurological channelopathies"

49. April 2005, Dept of Neurology, University of Newcastle
"Brain paroxysmal disorders"

50. May 2005, Dept Paediatrics, Alder Hay Hospital, Liverpool
"Paediatric channelopathies"

51. May 2005 Short course on Muscle Disease, Institute of Neurology, London
"Inflammatory myopathies"

2006

52. Feb 2006 NIH-CINCH collaborators' symposium UCLA-SanDiego USA
"Molecular mechanisms in Episodic ataxia"

53. Jan 2006-Irish National Annual Neuromuscular Symposium-Cork, Ireland
Guest Lecture "*Human skeletal muscle channelopathies - mechanisms and treatment*"

54. June 2006-Dept of Neurology, St George's Hospital, London
"*Neuronal channelopathies*"

55. April 2004 Neurology for Neuroscientists, Guarantors of Brain-Oxford
Neurological channelopathies

56. Sept 2006 - International Society of Neurophysiology annual conference, Edinburgh
Invited lecture - "*Genetic neurological channelopathies*"
Workshop organiser: "*Channelopathies*"

2007

57. January - Oxford Grand rounds
"*Neurological channelopathies*"

58. July-University of Kansas USA invited seminar
"*Molecular mechanisms in myotonia*"

2008

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

2010

60. June - Centre Hospitalier Universitaire Vaudois, Lausanne, France
Clinical Neurosciences Grand Round – "*muscle channelopathies*"

61. September - North American Muscle Study Group, Rochester, NY, USA
"*European perspective for recruitment*"

62. October – Centre for Neuroscience, Department of Medicine, Imperial College London "*Muscle channelopathies*"

2011

63. March – Royal Society of Medicine Muscle Symposium, London
"*Acquired muscle diseases*"

64. May – Cardiff Neurosciences Centre, University of Cardiff
"*Muscle channelopathies*"

65. May – Australian and New Zealand Association of Neurologists Annual Scientific Meeting, Hobart, Australia
W Ian McDonald Lecture "*Translational Research in Muscle Channelopathies - Genetics, Disease Mechanisms and Treatment Trials*"

66. May – Australian and New Zealand Association of Neurologists Annual Scientific Meeting, Hobart, Australia
Muscle symposium: "*Neuronal Channel Dysfunction and Episodic Ataxia - An Oxford-Australia-London Collaboration*"

67. Twenty-first meeting of the European Neurological Society, Lisbon, Portugal **"***How to evaluate the patient with a suspected channelopathy*" & Chair, Muscle Basic Course

68. July 11 - University of Newcastle Medical School, Department of Neurology Grand Round-"*Neurological channelopathies*".

2012

69-76 tbc

2013

77. October – Association of British Neurologists, Annual Meeting, Royal College of Physicians, London
"*Inclusion body myositis: bench to bedside*"

2014

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

2015

79. Neurology 2015 Institute of Neurology
"*Masterclass in muscle diseases*"

80. Milan Neurology Grand round
"*Channelopathies from genes to treatments*"

2016

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

2017

82. March - UK Kennedy's Disease Day, Derby
'*Introduction to Kennedy's Disease'*

83. March – *" Periodic Paralysis"* Danish Muscular Dystrophy Association, Copenhagen

84. October - Oslo University Hospital, Norway
'*The periodic paralyses – an overview'*, and '*The periodic paralyses – treatment and recent research*.

2018

85. April - 4th Annual Neuromuscular Disorders Seminar, Houston,
'*Update on genetic testing in Neuromuscular Diseases*'

86. May – ABN annual meeting, Birmingham
'Common muscle conditions - clinical-genetic diagnosis and management'

87. May – Update in Neuromuscular Disorders, Queen Square
'Muscle channelopathies update'

11. Academic Supervision and Muscle Disease Clinical Attachments

All primary supervisor unless otherwise indicated.

1997-2000
Ms Louise Eunson BSc
3 years funding BRT and ERF (1997-2000)
"Molecular genetic studies on brain channelopathies"
Queen Square essay prize 2000 [the enigma of paroxysmal disorders in neurology-the role of ion channels"]
Graduated PhD August 2001. Currently Genetics Lecturer in Biology University of Colchester"

1999-2000
Dr A Siddique 1999-2000 medical research fellow
Wellcome entry level fellowship-mitochondrial disease 1999-2000
Transferred to MRC funded "Genetics of epilepsy project" [Prof Wood]
Currently locum consultant Neurologist Kings College.

1998-2001
Dr T Pulkes MD
3 years funding BRT 1998-2001
"Molecular mechanisms in mitochondrial disease"
PhD graduated University of London 2004
Currently Consultant Neurologist and associate Chair neurology, Bangkok, Thailand

1999-2001
Ms Ruth Rea BSc
3 years funding Wellcome Trust
Co-supervised with Prof Kullmann
"Molecular expression in neurological channelopathies"
Graduated PhD University of London 2003
Currently Post-doc Stanford USA.

1999-2002
Dr NP Davies MBBS
Wellcome research training fellow commenced 1999 for 4 years [PhD]
"Skeletal muscle channelopathies"

Currently Consultant Neurologist, Birmingham.

2001-2004
Dr D Liolitsa PhD
Postdoctoral research assistant commenced 2001 for 4 years
Funding MRC coop component grant
"Nuclear genes and mitochondrial disease"
Danae became fascinated by Neurology and mitochondrial diseases and transferred to medicine UCL and graduated in medicine 2011.

2002-2004
Dr P Imbrici PhD
Post doctoral research assistant
Special trustees grant commence 2002 2 years
"Functional expression studies of calcium channel mutations causing human neurological disease"
Currently post-doc in ion channel research Rome.

2004-2007
Dr Tracey Graves medical research fellow 2004-2007
Brain Research Trust-Calcium channelopathies
NIH –USA-CINCH Fellow
Action Research Clinical training fellowship
Registered for PhD-UCL
Graduated PhD 2010
Currently SpR Neurology Oxford

2004-2007
Dr Doreen Fialho medical research fellow 2004-2007
NIH –USA-Muscle channelopathies
NSCAG Molecular expression scientist grant
Registered for PhD-UCL
Graduated PhD 2010
Currently Consultant Neurophysiologist, Queen Square.

2004-2007
Dr Niranjanan 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
Graduated PhD 2016
"Whole exome analysis to identify new genes in muscle channelopathies"

2009–2014
MRC Centre for Neuromuscular Diseases Clinical Research Fellow
Dr Jasper Morrow
Graduated PhD 2017
Jointly supervised with Mary Reilly Tarek Yousry
"Developing MRI as an outcome measure in neuromuscular disease"

2010-2013
Action Rheumatology Clinical Research Fellow
Dr Pedro Machado (Portuguese trainee)
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

2013-2017
MRC Centre Non-Clinical PhD Student – Charlotte Spicer
Investigating the effects of pharmacological up-regulation of the heat shock response in models of inclusion body myopathy
Primary Supervisor L Greensmith Secondary Supervisor MG Hanna

2013-2017
MRC Centre Non-Clinical PhD Student – Louise King
Mitophagy deficiencies in mitochondrial DNA disease
Primary Supervisor H Plun-Favreau Secondary Supervisors MG Hanna, M Sweeney

2013-2017
MRC Centre Non-Clinical PhD Student – Andreea Manole

Genetic and Functional Investigation of an Inherited Neuropathy and a Channelopathy
Primary Supervisor H Houlden Secondary Supervisor D Kullmann, MG Hanna

2013- 2019 (paused twice for maternity leave)
MRC Clinical Training Fellow - Helen Devine
The Pathogenesis of Spinal Bulbar Muscular Atrophy
Primary Supervisor MG Hanna Secondary Supervisors L Greensmith, R Patani

2013- 2019 (paused twice for maternity leave)
MRC Clinical Training Fellow – Karen Suetterlin
A Molecular Pathophysiological Study of the Skeletal Muscle Channelopathies
Primary Supervisor MG Hanna Secondary Supervisors E Matthews, R Mannikko

2015-2018
Lily Foundation Clinical Research Fellow
Dr Olivia Poole
Mitochondrial disease: clinical studies and molecular mechanisms
Primary Supervisor MH Hanna, Secondary Supervisor R Pitceathly

2016-2019
MRC Centre for Neuromuscular Diseases Clinical Research Fellow
Dr Enrico Bugiardini
Clinical, functional and genetic characterization of mitochondrial diseases
Primary Supervisor MG Hanna Secondary Supervisor H Houlden

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

2001
Prof Patrick Chinnery, now Professor of Neurology, University of Cambridge

2002
Dr Kevin Talbot, now Neuromuscular Consultant neurologist Oxford
2003
Dr Paul Maddison, now Neuromuscular Consultant Neurologist, Nottingham,

2004
Dr John Cole, now Muscle Disease - Consultant Neurologist Manchester
Dr Charlotte Brierley, now Muscle Disease - Consultant Neurologist Cambridge

2005
Dr Veronica Tan, now Consultant Neurophysiologist London
Dr Aisling Ryan, now Consultant Neurologist, Cork, Ireland

2006
Dr Singh- now consultant genetics Manchester

Dr Andrew Smith-now Consultant Neurology Bristol

2007
Dr Andrea Merrison- Consultant Neurologist Bristol

2008
Dr Chris Everett - now Consultant Neurologist at the Royal London

2009
Dr Susie Tomlinson – now Consultant Neurologist Sydney

2010
Dr Jasper Morrow – now Consultant Neurologist Lister Stevenage & NHNN

2011
Dr Tracey Graves Senior - now SpR Oxford

12. Teaching Activity-PhD Examiner

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

1997
1. Supervisor Professor DM Turnbull
"Mitochondrial heteroplasmy and 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 (see publications).
- I have published original channel research widely in international journals including Lancet, Annals of Neurology, Brain, Journal of Physiology, and Neurology. I have published several reviews in this area, most recently I have been invited to provide a review for Nature Neurology in Clinical Practice.

Clinical Aspects of Human Muscle Diseases

- As part of the MRC Centre I am actively involved in developing better ways to measure disease activity, disease damage and response to treatment in patients with inflammatory muscle disease. I published new scales to analyse these parameters with colleagues from UCL and NIH -Bethesda in the Journal Rheumatology 2004/11. I am adapting these scales to use in treatment of certain dystrophies with the intention of using new agents such as myostatin antibodies [through collaboration with Wyeth].

The ability of the neurogenetics group to translate research into clinical practice rapidly, particularly in terms of preclinical and prenatal diagnosis and thus improved genetic counseling, was enhanced by the establishment of a DNA service laboratory (initially staffed by three clinical scientists but which has rapidly expanded over the last 10 years I have been involved). This forms part of the National Hospitals' Clinical and Molecular Neurogenetics Unit, which is unique in simultaneously providing three components of

services related to inherited neurological disorders in adults: clinical diagnosis, DNA services and genetic counseling. In the DNA laboratory, we offer gene carrier prediction, prenatal diagnosis and mutation detection (where appropriate) for many inherited neurological diseases on a regional and national basis, including Huntington's disease, myotonic dystrophy, mitochondrial encephalomyopathies, Leber's hereditary optic neuropathy, hereditary motor and sensory neuropathy, dominant ataxias, prion diseases, familial amyloidoses, HMSN I, Friedreich's ataxia, primary generalized dystonia and X-linked bulbospinal neuronopathy. The provision of this service enhances recruitment of material suitable for study and over the past 10 years the unit has collected over 20,000 DNA samples.

15. Publications

15a Some example key papers

Andreu AL, **Hanna MG**, Reichman H, Bruno C, Penn AS, Tanji K, Palloti F, Iwata S, Shanske S, Bonilla E, Morgan-Hughes JA, DiMauro S (1999) Aches, pains and cramps: Exercise intolerance due to mutations in the cytochrome b gene of mitochondrial DNA.
New England Journal of Medicine 341: 1037-1078
Cited 256 times [Google scholar] Impact Factor 34.833

This work published in the New England Journal of Medicine was the first demonstration that somatic mutations in the protein encoding gene cytochrome b in human mitochondrial DNA could associate with a commonly encountered clinical phenotype of exercise intolerance and myalgia. This was a collaborative research project with colleagues in USA and Germany. We identified a group of patients in which the defect in cytochrome b in mtDNA was only detectable in muscle [not in blood]. Prior to these findings point mutations in human mtDNA which were only confined to single somatic tissue had not been described. This suggested that the mutational event causing this form of human mitochondrial disease must have occurred in a cell line already destined to differentiate into muscle. Such late mutational events [ie not germline] had not been recognized previously for point mutations. We undertook detailed biochemical experiments to confirm a specific defect in complex III of the respiratory chain. This work served to initiate the area of somatic mtDNA point mutations in human disease. Subsequently my group and others have identified further somatic mutations in other mtDNA encoded genes in patients with a similar phenotypes confirming this is a new category of mtDNA mitochondrial disease [see ref 56 and 68]. From a clinical viewpoint this work emphasized the need to not rely on analysis of mtDNA point mutations in blood when assessing such patients. This is in contrast to germline mtDNA point mutations which are always detectable in blood. There was an accompanying Editorial Review in the New England Journal of Medicine by Professor RC Griggs (Head of Department of Neurology, University of Rochester New York and Professor George 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. PMID: 17932099. (Cited 44 times Google Scholar)

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

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

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

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

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

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

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

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

New Insights into fundamental mechanisms of ion channel function

- Expression of mutations in the muscle chloride channel mutations in oocyte expression system show dominant negative interactions are mediated at the interface linking the two monomers that make up the function channel

- Loss of positive charge mutations in the S4 segments of both sodium and calcium muscle channels link to the phenotype of hypokalaemic periodic paralysis

- Genetic evidence supports the view that loss of charge mutations induce a gating pore current

New insights into genotype phenotype correlations

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

- Dominant mutations found more commonly in chloride channel than previous reports with important genetic counseling implications

- Neonatal hypotonia recognised as a new phenotype for sodium channel disease altering guidance for neonatal care of children born to affected parents

- Previously unexplained severe neonatal stridor/laryngospasm shown to be a new sodium channel disease phenotype with implications for diagnosis and management of neonatal stridor

Delivery of human randomized clinical trials in a rare disease

- I am the senior author on a recently published trial reprofiling an agent to target human muscle sodium channels to treat an inherited myotonic disorder. This 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. PMID: 23032552

15b. Books/Thesis

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

15c. Chapters

1. Rudel R, Hanna MG, Lehmann-Horn F (1999). Muscle ion channelopathies: malignant hyperthermia, periodic paralyses, paramyotonia congenita and myotonia. Muscle Diseases. Eds: Schapira AHV, Griggs R. Butterworth Heinmann, *pp 134-154.*

2. Hanna MG (2000). Cases from the National Hospital. Eds: CD Marsden, A Wills. Martin Dunitz, London, two cases, *pp23-26 & pp56-59.*

3. Davies NP Hanna MG (2001). Genetic neurological channelopathies. New Treatments in Neurology. Ed: Scolding N. Butterworth Heinemann, London, *pp234-247.*

4. Kinton L, Hanna MG, Wood NW (2001). Genetics of epilepsy and paroxysmal movement disorders. Epilepsy and Movement Disorders. Eds: Guerinni, Aicardi, Andermann, Hallet. Cambridge University Press, *pp67-79.*

5. Davies NP, Hanna MG (2003). Myopathies and neurorehabilitation. Textbook of Neurorehabilitation. Ed: Greenwood R. *pp256-274.*

6. Pulkes T, Hanna MG (2003). Mitochondrial diseases. Mitochondrial DNA and Human Disease. Ed: Holt IJ. Butterworth Heinemann, *pp57-75.*
7. Liolitsa D, Hanna MG (2003). Cell models in mitochondrial disease. Mitochondrial Function and Dysfunction in Human Disease. Eds: Schapira AHV, DeLauro S. Butterworth Heinemann, London, *pp89-110.*

8. Hanna MG (2003). Peripheral neuropathy and mitochondrial diseases. Peripheral Neuropathy. Eds: Dyck D, Thomas PK. Saunders, *pp1937-1949.*

9. Hanna MG (2004). Disorders of voluntary muscle. Oxford Textbook of Medicine, 4th Ed*.* Oxford, *pp1344 -1346.*

10. Fontaine B, Hanna MG (2009). Muscle ion channelopathies and related disorders. Disorders of Voluntary Muscle, 8th Ed. Eds: George Karpati, David Hilton-Jones, Kate Bushby, Robert C. Griggs. Cambridge University Press, *pp409-426.*

11. Hanna MG, Parton M (2009). Muscle diseases. Neurology: A Queen Square Textbook. Eds: Clarke C, Shorvon S, Howard R, Rossor M. 1st Ed. Wiley-Blackwell, Chichester, *pp.337-410*.

12. Raja Rayan D, Hanna MG (2010). Ion channels and human disorders. Encyclopaedia of Life Sciences. John Wiley & Sons, *pp1-17.*

13. Fialho D, Hanna MG (2010). Periodic paralysis. Handbook of Neurology, Eds: Hilton Jones DA, Laing D. Heinemann, *pp 92-118.*
14. *B*urge J, Hanna MG (2011). Muscle channelopathies. Neurology in Practice: Neuromuscular Disorders. Eds: Rabi N. Tawil, Shannon Venance. Wiley-Blackwell.

15. Hanna MG, Kullmann DM (2012). Channelopathies. Neurogenetics: A Guide for Clinicians. Ed: Nicholas W. Wood. Cambridge University Press, *pp121-135.*

16. Matthews E, Hanna MG (2013). Cav1.1 hypokalaemic periodic paralysis. Pathologies of Calcium Channels. Eds: Norbert Weiss, Alexandra Koschak. Springer.

17. Hanna MG (2013). Muscle Disease: Pathology and Genetics. 2nd Ed. Eds: Hans H. Goebel, Caroline A. Sewry, Roy O. Weller. Wiley-Blackwell.

18. Hanna MG (2014). Diseases of the muscles. Scully's Medical Problems in Dentistry. 7th Ed. Ed: Scully C. Elsevier, *chapter 16*.

19. Matthews E, Hanna MG (2014). Skeletal Muscle Channelopathies. Oxford Textbook of Neuromuscular Disorders. Oxford Textbooks in Clinical Neurology. Eds: David Hilton-Jones, Martin R. Turner. Oxford University Press, *pp 316-325.*

20. Suetterlin K, Hanna MG (2016). Muscle Channelopathies. International Neurology: A Clinical Approach. 2nd Ed. Wiley-Blackwell. *Part 13, ch. 116.*

21. Pitceathly R, Hanna MG, Reilly MM (2018). Neurology – the Peripheral Nervous System. Clinical Mitochondrial Medicine. Cambridge University Press, *section 2, ch 8*.

22. Hanna MG (in press). Neurogenetics. The Oxford Handbook of Molecular Medicine Ed: Lo. Oxford University Press.

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. 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 mitochondrial DNA 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. 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. 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. 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

2001

35. Zuberi SM, **Hanna MG**. Ion channels and neurology. Arch Dis Child. 2001 Mar;84(3):277-80. Review. PMID: 11207185

36. Pulkes T, **Hanna MG**. Human mitochondrial DNA diseases. Adv Drug Deliv Rev. 2001 Jul 2;49(1-2):27-43. Review. PMID: 11377801

37. Davies NP, **Hanna MG**. The skeletal muscle channelopathies: basic science, clinical genetics and treatment. Curr Opin Neurol. 2001 Oct;14(5):539-51. Review. PMID: 11562564

38. Jouvenceau A, Eunson LH, Spauschus A, Ramesh V, Zuberi SM, Kullmann DM, Hanna MG. Human epilepsy associated with dysfunction of the brain P/Q-type calcium channel. Lancet. 2001 Sep 8;358(9284):801-7. PMID: 11564488

39. Davies NP, Eunson LH, Samuel M, **Hanna MG**. Sodium channel gene mutations in hypokalemic periodic paralysis: an uncommon cause in the UK. Neurology. 2001 Oct 9;57(7):1323-5. PMID: 11591859

40. Davies NP, **Hanna MG**. Inherited muscle and brain channelopathies. Expert Rev Neurother. 2001 Nov;1(2):247-65. PMID: 19811036

2002

41. Rea R, Spauschus A, Eunson LH, **Hanna MG**, Kullmann DM. Variable K(+) channel subunit dysfunction in inherited mutations of KCNA1. J Physiol. 2002 Jan 1;538(Pt 1):5-23. PMID: 11773313

42. McCabe DJ, Wood NW, Ryan F, **Hanna MG**, Connolly S, Moore DP, Redmond J, Barton DE, Murphy RP. Intrafamilial phenotypic variability in Friedreich ataxia associated with a G130V mutation in the FRDA gene. Arch Neurol. 2002 Feb;59(2):296-300. PMID: 11843702

43. Littleton ET, Man WD, Holton JL, Landon DN, **Hanna MG**, Polkey MI, Taylor GP. Human T cell leukaemia virus type I associated neuromuscular disease causing respiratory failure. J Neurol Neurosurg Psychiatry. 2002 May;72(5):650-2. PMID: 11971056

44. Garrard P, Blake J, Stinton V, **Hanna MG**, Reilly MM, Holton JL, Landon DN, Honan WP. Distal myopathy with tubular aggregates: a new phenotype associated with multiple deletions in mitochondrial DNA? J Neurol Neurosurg Psychiatry. 2002 Aug;73(2):207-8. PMID: 12122190

45. Jacques TS, Holton J, Watts PM, Wills AJ, Smith SE, **Hanna MG**. Tubular aggregate myopathy with abnormal pupils and skeletal deformities. J Neurol Neurosurg Psychiatry. 2002 Sep;73(3):324-6. PMID: 12185172

46. Chinnery PF, Walls TJ, **Hanna MG**, Bates D, Fawcett PR. Normokalemic periodic paralysis revisited: does it exist? Ann Neurol. 2002 Aug;52(2):251-2. PMID: 12210802

47. Liolitsa D, Rahman S, Benton S, Carr LJ, **Hanna MG**. Is the mitochondrial complex I ND5 gene a hot-spot for MELAS causing mutations? Ann Neurol. 2003 Jan;53(1):128-32. PMID: 12509858

48. Liolitsa D, **Hanna MG**. Models of mitochondrial disease. Int Rev Neurobiol. 2002;53:429-66. Review. PMID: 12512349

49. **Hanna MG**, Wood NW. Running a neurogenetic clinic. J Neurol Neurosurg Psychiatry. 2002 Dec;73 Suppl 2:II2-4. PMID: 12536152

50. Reilly MM, **Hanna MG**. Genetic neuromuscular disease. J Neurol Neurosurg Psychiatry. 2002 Dec;73 Suppl 2:II12-21. Review. PMID: 12536154

51. Kullmann DM, **Hanna MG**. Neurological disorders caused by inherited ion-channel mutations. Lancet Neurol. 2002 Jul;1(3):157-66. Review. PMID: 12849484

2003

52. Isenberg DA, Allen E, Farewell V, Ehrenstein MR, **Hanna MG**, Lundberg IE, Oddis C, Pilkington C, Plotz P, Scott D, Vencovsky J, Cooper R, Rider L, Miller F; International Myositis and Clinical Studies Group (IMACS). International consensus outcome measures for patients with idiopathic inflammatory myopathies. Development and initial validation of myositis activity and damage indices in patients with adult onset disease. Rheumatology (Oxford). 2004 Jan;43(1):49-54. Review. PMID: 12867580

53. Miller RF, Shahmonesh M, **Hanna MG**, Unwin RJ, Schapira AH, Weller IV. Polyphenotypic expression of mitochondrial toxicity caused by nucleoside reverse transcriptase inhibitors. Antivir Ther. 2003 Jun;8(3):253-7. PMID: 12924544

54. Sorajja P, Sweeney MG, Chalmers R, Sachdev B, Syrris P, **Hanna M**, Wood ND, McKenna WJ, Elliott PM. Cardiac abnormalities in patients with Leber's hereditary optic neuropathy. Heart. 2003 Jul;89(7):791-2. PMID: 12807863

55. Brancati F, Valente EM, Davies NP, Sarkozy A, Sweeney MG, LoMonaco M, Pizzuti A, **Hanna MG**, Dallapiccola B. Severe infantile hyperkalaemic periodic paralysis and paramyotonia congenita: broadening the clinical spectrum associated with the T704M mutation in SCN4A. J Neurol Neurosurg Psychiatry. 2003 Sep;74(9):1339-41. PMID: 12933953

56. Mundy HR, Jones SJ, Hobart JC, **Hanna MG**, Lee PJ. A randomized controlled study of modified cobratoxin in adrenomyeloneuropathy. Neurology. 2003 Aug 26;61(4):528-30. PMID: 12939429

57. Davies NP, **Hanna MG**. The skeletal muscle channelopathies: distinct entities and overlapping syndromes. Curr Opin Neurol. 2003 Oct;16(5):559-68. Review. PMID: 14501839

58. Hargreaves IP, Heales SJ, Briddon A, Lee PJ, **Hanna MG**, Land JM. Primary pyruvate dehydrogenase E3 binding protein deficiency with mild hyperlactataemia and hyperalaninaemia. J Inherit Metab Dis. 2003;26(5):505-6. PMID: 14518830

59. Pulkes T, Liolitsa D, Nelson IP, **Hanna MG**. Classical mitochondrial phenotypes without mtDNA mutations: the possible role of nuclear genes. Neurology. 2003 Oct 28;61(8):1144-7. PMID:14581685

2004

60. McKenzie M, Liolitsa D, **Hanna MG**. Mitochondrial disease: mutations and mechanisms. Neurochem Res. 2004 Mar;29(3):589-600. Review. PMID: 15038606

61. Roedling S, Pearl D, Manji H, **Hanna MG**, Holton JL, Miller RF. Unusual muscle disease in HIV infected patients. Sex Transm Infect. 2004 Aug;80(4):315-7. Review. PMID: 15295133

62. Kinali M, Jungbluth H, Eunson LH, Sewry CA, Manzur AY, Mercuri E, **Hanna MG**, Muntoni F. Expanding the phenotype of potassium channelopathy: severe neuromyotonia and skeletal deformities without prominent Episodic Ataxia. Neuromuscul Disord. 2004 Oct;14(10):689-93. PMID: 15351427

63. Imbrici P, Jaffe SL, Eunson LH, Davies NP, Herd C, Robertson R, Kullmann DM, **Hanna MG**. Dysfunction of the brain calcium channel CaV2.1 in absence epilepsy and episodic ataxia. Brain. 2004 Dec;127(Pt 12):2682-92. PMID: 15483044

64. Nirmalananthan N, Holton JL, **Hanna MG**. Is it really myositis? A consideration of the differential diagnosis. Curr Opin Rheumatol. 2004 Nov;16(6):684-91. Review. PMID: 15577605

2005

65. Graves TD, **Hanna MG**. Neurological channelopathies. Postgrad Med J. 2005 Jan;81(951):20-32. Review. PMID: 15640425

66. Murphy RT, Mogensen J, McGarry K, Bahl A, Evans A, Osman E, Syrris P, Gorman G, Farrell M, Holton JL, **Hanna MG,** Hughes S, Elliott PM, Macrae CA, McKenna WJ.Adenosine monophosphate-activated protein kinase disease mimicks hypertrophic cardiomyopathy and Wolff-Parkinson-White syndrome: natural history. J Am Coll Cardiol. 2005 Mar 15;45(6):922-30. PMID: 15766830

67. Pulkes T, Liolitsa D, Wills AJ, Hargreaves I, Heales S, **Hanna MG**. Nonsense mutations in mitochondrial DNA associated with myalgia and exercise intolerance. Neurology. 2005 Mar 22;64(6):1091-2. PMID: 15781840

68. Pulkes T, Liolitsa D, Eunson LH, Rose M, Nelson IP, Rahman S, Poulton J, Marchington DR, Landon DN, Debono AG, Morgan-Hughes JA, **Hanna MG**. New phenotypic diversity associated with the mitochondrial tRNA(SerUCN) gene mutation. Neuromuscul Disord. 2005 May;15(5):364-71. PMID: 15833431

69. Eunson LH, Graves TD, **Hanna MG**. New calcium channel mutations predict aberrant RNA splicing in episodic ataxia. Neurology. 2005 Jul 26;65(2):308-10. PMID: 16043807

70. Imbrici P, Eunson LH, Graves TD, Bhatia KP, Wadia NH, Kullmann DM, **Hanna MG**. Late-onset episodic ataxia type 2 due to an in-frame insertion in CACNA1A. Neurology. 2005 Sep 7;65(6):944-6. PMID: 16186543

71. Venance SL, Cannon SC, Fialho D, Fontaine B, **Hanna MG**, Ptacek LJ, Tristani-Firouzi M, Tawil R, Griggs RC; CINCH investigators. The primary periodic paralyses: diagnosis, pathogenesis and treatment. Brain. 2006 Jan;129(Pt 1):8-17. Review. PMID: 16195244

72. Davies NP, Imbrici P, Fialho D, Herd C, Bilsland LG, Weber A, Mueller R, Hilton-Jones D, Ealing J, Boothman BR, Giunti P, Parsons LM, Thomas M, Manzur AY, Jurkat-Rott K, Lehmann-Horn F, Chinnery PF, Rose M, Kullmann DM, **Hanna MG**. Andersen-Tawil syndrome: new potassium channel mutations and possible phenotypic variation. Neurology. 2005 Oct 11;65(7):1083-9. PMID: 16217063

73. Gioltzoglou T, Cordivari C, Lee PJ, **Hanna MG**, Lees AJ. Problems with botulinum toxin treatment in mitochondrial cytopathy: case report and review of the literature. J Neurol Neurosurg Psychiatry. 2005 Nov;76(11):1594-6. Review. PMID: 16227561

2006

74. Chegounchi M, **Hanna MG**, Neild GH. Progressive neurological disease induced by tacrolimus in a renal transplant recipient: case presentation. BMC Nephrol. 2006 Mar 31;7:7. PMID: 16573841

75. Hudson G, Deschauer M, Taylor RW, **Hanna MG**, Fialho D, Schaefer AM, He LP, Blakely E, Turnbull DM, Chinnery PF. POLG1, C10ORF2, and ANT1 mutations are uncommon in sporadic progressive external ophthalmoplegia with multiple mitochondrial DNA deletions. Neurology. 2006 May 9;66(9):1439-41. PMID: 16682683

76. **Hanna MG**. Genetic neurological channelopathies. Nat Clin Pract Neurol. 2006 May;2(5):252-63. Review. PMID: 16932562

77. Horvath R, Hudson G, Ferrari G, Fütterer N, Ahola S, Lamantea E, Prokisch H, Lochmüller H, McFarland R, Ramesh V, Klopstock T, Freisinger P, Salvi F, Mayr JA, Santer R, Tesarova M, Zeman J, Udd B, Taylor RW, Turnbull D, **Hanna M**, Fialho D, Suomalainen A, Zeviani M, Chinnery PF. Phenotypic spectrum associated with mutations of the mitochondrial polymerase gamma gene. Brain. 2006 Jul;129(Pt 7):1674-84. PMID: 16621917

78. Nirmalananthan N, **Hanna MG**. Predicting steroid response in muscle disease. J Neurol Neurosurg Psychiatry. 2006 Oct;77(10):1104-5. PMID: 16980654

79. Abou-Sleiman PM, **Hanna MG**, Wood NW. Genetic association studies of complex neurological diseases. J Neurol Neurosurg Psychiatry. 2006 Dec;77(12):1302-4. Review. PMID: 17110744

2007

80. Rajakulendran S, Schorge S, Kullmann DM, **Hanna MG**. Episodic ataxia type 1: a neuronal potassium channelopathy. Neurotherapeutics. 2007 Apr;4(2):258-66. Review. PMID: 17395136

81. Sisodiya S, Cross JH, Blümcke I, Chadwick D, Craig J, Crino PB, Debenham P, Delanty N, Elmslie F, Gardiner M, Golden J, Goldstein D, Greenberg DA, Guerrini R, **Hanna M**, Harris J, Harrison P, Johnson MR, Kirov G, Kullman DM, Makoff A, Marini C, Nabbout R, Nashef L, Noebels JL, Ottman R, Pirmohamed M, Pitkänen A, Scheffer I, Shorvon S, Sills G, Wood N, Zuberi S. Genetics of epilepsy: epilepsy research foundation workshop report. Epileptic Disord. 2007 Jun;9(2):194-236. PMID: 17525034

82. Jen JC, Graves TD, Hess EJ, **Hanna MG**, Griggs RC, Baloh RW; CINCH investigators. Primary episodic ataxias: diagnosis, pathogenesis and treatment. Brain. 2007 Oct;130(Pt 10):2484-93. Review. PMID: 17575281

83. Tomlinson SE, **Hanna MG**. Acute neurology. Clin Med. 2007 Jun;7(3):272-7. Review. PMID: 17633950

84. Ryan AM, Matthews E, **Hanna MG**. Skeletal-muscle channelopathies: periodic paralysis and nondystrophic myotonias. Curr Opin Neurol. 2007 Oct;20(5):558-63. Review. PMID: 17885445

85. Cavalleri GL, Weale ME, Shianna KV, Singh R, Lynch JM, Grinton B, Szoeke C, Murphy K, Kinirons P, O'Rourke D, Ge D, Depondt C, Claeys KG, Pandolfo M, Gumbs C, Walley N, McNamara J, Mulley JC, Linney KN, Sheffield LJ, Radtke RA, Tate SK, Chissoe SL, Gibson RA, Hosford D, Stanton A, Graves TD, **Hanna MG**, et al. Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. Lancet Neurol. 2007 Nov;6(11):970-80. PMID: 17913586

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

87. McKenzie M, Liolitsa D, Akinshina N, Campanella M, Sisodiya S, Hargreaves I, Nirmalananthan N, Sweeney MG, Abou-Sleiman PM, Wood NW, **Hanna MG**, Duchen MR. Mitochondrial ND5 gene variation associated with encephalomyopathy and mitochondrial ATP consumption. J Biol Chem. 2007 Dec 21;282(51):36845-52. PMID: 17940288

88. van de Warrenburg BP, Cordivari C, Ryan AM, Phadke R, Holton JL, Bhatia KP, **Hanna MG**, Quinn NP. The phenomenon of disproportionate antecollis in Parkinson's disease and multiple system atrophy. Mov Disord. 2007 Dec;22(16):2325-31. PMID: 17960814

89. Everett CM, Graves TD, Lad S, Jäger HR, Thom M, Isenberg DA, **Hanna MG**. Aggressive CNS lupus vasculitis in the absence of systemic disease activity. Rheumatology (Oxford). 2008 Jan;47(1):107-9. PMID: 18037608

90. Craig K, Elliott HR, Keers SM, Lambert C, Pyle A, Graves TD, Woodward C, Sweeney MG, Davis MB, **Hanna MG**, Chinnery PF. Episodic ataxia and hemiplegia caused by the 8993T->C mitochondrial DNA mutation. J Med Genet. 2007 Dec;44(12):797-9. PMID: 18055910

2008

91. 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

92. Graves TD, **Hanna MG**. Channelling into the epilepsies. Epilepsy Curr. 2008 Mar-Apr;8(2):37-8. PMID: 18330464

93. Healy DG, Goadsby PJ, Kitchen ND, Yousry T, **Hanna MG**. Neurological picture.Spontaneous intracranial hypotension, hygromata and haematomata. J Neurol Neurosurg Psychiatry. 2008 Apr;79(4):442. PMID: 18344394

94. Graves TD, **Hanna MG**. Episodic ataxia: SLC1A3 and CACNB4 do not explain the apparent genetic heterogeneity. J Neurol. 2008 Jul;255(7):1097-9. PMID: 18446307

95. Hall AM, Unwin RJ, **Hanna MG**, Duchen MR. Renal function and mitochondrial cytopathy (MC): more questions than answers? QJM. 2008 Oct;101(10):755-66. Review. PMID: 18487272

96. Graves TD, Imbrici P, Kors EE, Terwindt GM, Eunson LH, Frants RR, Haan J, Ferrari MD, Goadsby PJ, **Hanna MG**, van den Maagdenberg AM, Kullmann DM. Premature stop codons in a facilitating EF-hand splice variant of CaV2.1 cause episodic ataxia type 2. Neurobiol Dis. 2008 Oct;32(1):10-5. PMID: 18606230

97. Fialho D, **Hanna MG**. Periodic paralysis. Handb Clin Neurol. 2007;86:77-106. PMID: 18808996

98. Fialho D, Kullmann DM, **Hanna MG**, Schorge S. Non-genomic effects of sex hormones on CLC-1 may contribute to gender differences in myotonia congenita. Neuromuscul Disord. 2008 Nov;18(11):869-72. PMID: 18815035

99. 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

2009

100. Healy DG, Goadsby PJ, Kitchen ND, Yousry T, **Hanna MG**. Spontaneous intracranial hypotension, hygromata and haematomata. BMJ Case Rep.2009: PMID: 21687310

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

102. Merrison AF, **Hanna MG**. The bare essentials: muscle disease. Pract Neurol. 2009 Feb;9(1):54-65. PMID: 19151243

103. Stewart JD, Tennant S, Powell H, Pyle A, Blakely EL, He L, Hudson G, Roberts M, du Plessis D, Gow D, Mewasingh LD, **Hanna MG**, Omer S, Morris AA, Roxburgh R, Livingston JH, McFarland R, Turnbull DM, Chinnery PF, Taylor RW. Novel POLG1 mutations associated with neuromuscular and liver phenotypes in adults and children. J Med Genet. 2009 Mar;46(3):209-14. PMID: 19251978

104. Machado P, Miller A, Holton J, **Hanna M**. Sporadic inclusion body myositis: an unsolved mystery. Acta Reumatol Port. 2009 Apr-Jun;34(2A):161-82. Review. PMID: 19474772

105. Meola G, **Hanna MG**, Fontaine B. Diagnosis and new treatment in muscle channelopathies. J Neurol Neurosurg Psychiatry. 2009 Apr;80(4):360-5. Review. PMID: 19289476

106. Trender-Gerhard I, Sweeney MG, Schwingenschuh P, Mir P, Edwards MJ, Gerhard A, Polke JM, **Hanna MG**, Davis MB, Wood NW, Bhatia KP. Autosomal-dominant GTPCH1-deficient DRD: clinical characteristics and long-term outcome of 34 patients. J Neurol Neurosurg Psychiatry. 2009 Aug;80(8):839-45. PMID: 19332422

107. Labrum RW, Rajakulendran S, Graves TD, Eunson LH, Bevan R, Sweeney MG, Hammans SR, Tubridy N, Britton T, Carr LJ, Ostergaard JR, Kennedy CR, Al-Memar A, Kullmann DM, Schorge S, Temple K, Davis MB, **Hanna MG**. Large scale calcium channel gene rearrangements in episodic ataxia and hemiplegic migraine: implications for diagnostic testing. J Med Genet. 2009 Nov;46(11):786-91. PMID: 19586927

108. Rahman S, **Hanna MG**. Diagnosis and therapy in neuromuscular disorders: diagnosis and new treatments in mitochondrial diseases. J Neurol Neurosurg Psychiatry. 2009 Sep;80(9):943-53. Review. PMID: 19684231

109. Tomlinson SE, **Hanna MG**, Kullmann DM, Tan SV, Burke D. Clinical neurophysiology of the episodic ataxias: insights into ion channel dysfunction in vivo. Clin Neurophysiol. 2009 Oct;120(10):1768-76. Review. PMID: 19734086

110. Rajakulendran S, Tan SV, Matthews E, Tomlinson SE, Labrum R, Sud R, Kullmann DM, Schorge S, **Hanna MG**. A patient with episodic ataxia and paramyotonia congenita due to mutations in KCNA1 and SCN4A. Neurology. 2009 Sep 22;73(12):993-5. PMID: 19770477

111. Heeroma JH, Henneberger C, Rajakulendran S, **Hanna MG**, Schorge S, Kullmann DM. Episodic ataxia type 1 mutations differentially affect neuronal excitability and transmitter release. Dis Model Mech. 2009 Nov-Dec;2(11-12):612-9. PMID: 19779067

2010

112. Matthews E, Fialho D, Tan SV, Venance SL, Cannon SC, Sternberg D, Fontaine B, Amato AA, Barohn RJ, Griggs RC, **Hanna MG**; CINCH Investigators. The non-dystrophic myotonias: molecular pathogenesis, diagnosis and treatment. Brain. 2010 Jan;133(Pt 1):9-22. Review. PMID: 19917643

113. Hilton-Jones D, Miller A, Parton M, Holton J, Sewry C, **Hanna MG**. Inclusion body myositis: MRC Centre for Neuromuscular Diseases, IBM workshop, London, 13June 2008. Neuromuscul Disord. 2010 Feb;20(2):142-7. PMID: 20074951

114. Tomlinson S, Burke D, **Hanna M**, Koltzenburg M, Bostock H. In vivo assessment of HCN channel current (I(h)) in human motor axons. Muscle Nerve. 2010 Feb;41(2):247-56. PMID: 19813191

115. Limongelli G, Tome-Esteban M, Dejthevaporn C, Rahman S, **Hanna MG**, Elliott PM. Prevalence and natural history of heart disease in adults with primary mitochondrial respiratory chain disease. Eur J Heart Fail. 2010 Feb;12(2):114-21. PMID: 20083621

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

117. Rajakulendran S, Graves TD, Labrum RW, Kotzadimitriou D, Eunson L, Davis MB, Davies R, Wood NW, Kullmann DM, **Hanna MG**, Schorge S. Genetic and functional characterization of the P/Q calcium channel in episodic ataxia with epilepsy. J Physiol. 2010 Jun 1;588(Pt 11):1905-13. PMID: 20156848

118. Zhou H, Lillis S, Loy RE, Ghassemi F, Rose MR, Norwood F, Mills K, Al-Sarraj S, Lane RJ, Feng L, Matthews E, Sewry CA, Abbs S, Buk S, **Hanna MG**, Treves S, Dirksen RT, Meissner G, Muntoni F, Jungbluth H. Multi-minicore disease and

atypical periodic paralysis associated with novel mutations in the skeletal muscle ryanodine receptor (RYR1) gene. Neuromuscul Disord. 2010 Mar;20(3):166-73. PMID: 20080402

119. Rajakulendran S, Kuntzer T, Dunand M, Yau SC, Ashton EJ, Storey H, McCauley J, Abbs S, Thonney F, Leturcq F, Lobrinus JA, Yousry T, Farmer S, Holton JL, **Hanna MG**. Marked hemiatrophy in carriers of Duchenne muscular dystrophy. Arch Neurol. 2010 Apr;67(4):497-500. PMID: 20385919

120. Fratter C, Gorman GS, Stewart JD, Buddles M, Smith C, Evans J, Seller A, Poulton J, Roberts M, **Hanna MG**, Rahman S, Omer SE, Klopstock T, Schoser B, Kornblum C, Czermin B, Lecky B, Blakely EL, Craig K, Chinnery PF, Turnbull DM, Horvath R, Taylor RW. The clinical, histochemical, and molecular spectrum of PEO1 (Twinkle)-linked adPEO. Neurology. 2010 May 18;74(20):1619-26. PMID: 20479361

121. Raja Rayan DL, **Hanna MG**. Skeletal muscle channelopathies: nondystrophic myotonias and periodic paralysis. Curr Opin Neurol. 2010 Oct;23(5):466-76. Review. PMID: 20634695

122. Rajakulendran S, Tan SV, **Hanna MG**. Muscle weakness, palpitations and a small chin: the Andersen-Tawil syndrome. Pract Neurol. 2010 Aug;10(4):227-31. Review. PMID: 20647529

123. Graves TD, Rajakulendran S, Zuberi SM, Morris HR, Schorge S, **Hanna MG**, Kullmann DM. Nongenetic factors influence severity of episodic ataxia type 1 in monozygotic twins. Neurology. 2010 Jul 27;75(4):367-72. PMID: 20660867

124. Rajakulendran S, Schorge S, Kullmann DM, **Hanna MG**. Dysfunction of the Ca(V)2.1 calcium channel in cerebellar ataxias. F1000 Biol Rep. 2010 Jan 18;2. PMID: 20948794

125. Sinclair CD, Miranda MA, Cowley P, Morrow JM, Davagnanam I, Mehta H, Hanna MG, Koltzenburg M, Reilly MM, Yousry TA, Thornton JS. MRI shows increased sciatic nerve cross sectional area in inherited and inflammatory neuropathies. J Neurol Neurosurg Psychiatry. 2010 Oct 22. PMID: 20971754

126. Tomlinson SE, Tan SV, Kullmann DM, Griggs RC, Burke D, **Hanna MG**, Bostock H. Nerve excitability studies characterize Kv1.1 fast potassium channel dysfunction in patients with episodic ataxia type 1. Brain. 2010 Dec;133(Pt 12):3530-40. PMID: 21106501

2011

127. Forrest KM, Al-Sarraj S, Sewry C, Buk S, Tan SV, Pitt M, Durward A, McDougall M, Irving M, **Hanna MG**, Matthews E, Sarkozy A, Hudson J, Barresi R, Bushby K, Jungbluth H, Wraige E. Infantile onset myofibrillar myopathy due to recessive CRYAB mutations. Neuromuscul Disord. 2011 Jan;21(1):37-40. PMID: 21130652

128. 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

129. Malik B, Nirmalananthan N, Bilsland LG, La Spada AR, **Hanna MG**, Schiavo G, Gallo JM, Greensmith L. Absence of disturbed axonal transport in spinal and bulbar muscular atrophy. Hum Mol Genet. 2011 May 1;20(9):1776-86. PMID: 21317158

130. Becker EB, Fogel BL, Rajakulendran S, Dulneva A, **Hanna MG**, Perlman SL, Geschwind DH, Davies KE. Candidate screening of the TRPC3 gene in cerebellar ataxia. Cerebellum. 2011 Jun;10(2):296-9. PMID: 21321808

131. Pitceathly RD, Fassone E, Taanman JW, Sadowski M, Fratter C, Mudanohwo EE, Woodward CE, Sweeney MG, Holton JL, **Hanna MG**, Rahman S. Kearns-Sayre syndrome caused by defective R1/p53R2 assembly. J Med Genet. 2011 Mar 22. PMID: 21378381

132. 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

133. Sinclair CD, Morrow JM, Miranda MA, Davagnanam I, Cowley PC, Mehta H, **Hanna MG**, Koltzenburg M, Yousry TA, Reilly MM, Thornton JS. Skeletal muscle MRI magnetisation transfer ratio reflects clinical severity in peripheral neuropathies. J Neurol Neurosurg Psychiatry. 2011 May 25. PMID: 21613652

134. Sultan SM, Allen E, Cooper RG, Agarwal S, Kiely P, Oddis CV, Vencovsky J, Lundberg IE, Dastmalchi M, **Hanna MG**, Isenberg DA. Interrater reliability and aspects of validity of the myositis damage index. Ann Rheum Dis. 2011 Jul;70(7):1272-6. PMID: 21622773

135. Fratter C, Raman P, Alston CL, Blakely EL, Craig K, Smith C, Evans J, Seller A, Czermin B, **Hanna MG**, Poulton J, Brierley C, Staunton TG, Turnpenny PD, Schaefer AM, Chinnery PF, Horvath R, Turnbull DM, Gorman GS, Taylor RW. RRM2B mutations are frequent in familial PEO with multiple mtDNA deletions. Neurology. 2011 Jun 7;76(23):2032-4. PMID: 21646632

136. Scoto M, Cirak S, Mein R, Feng L, Manzur AY, Robb S, Childs AM, Quinlivan RM, Roper H, Jones DH, Longman C, Chow G, Pane M, Main M, **Hanna MG**, Bushby K, Sewry C, Abbs S, Mercuri E, Muntoni F. SEPN1-related myopathies: clinical course in a large cohort of patients. Neurology. 2011 Jun 14;76(24):2073-8. PMID: 21670436

137. Matthews E, Miller JA, Macleod MR, Ironside J, Ambler G, Labrum R, Sud R, Holton JL, **Hanna MG**. Sodium and chloride channelopathies with myositis:

Coincidence or connection? Muscle Nerve. 2011 Aug;44(2):283-8. PMID: 21698652

138. Statland JM, Wang Y, Richesson R, Bundy B, Herbelin L, Gomes J, Trivedi J, Venance S, Amato A, **Hanna MG**, Griggs R, Barohn RJ; Cinch Consortium. An interactive voice response diary for patients with non-dystrophic myotonia. Muscle Nerve. 2011 Jul;44(1):30-5. PMID: 21674518

139. Sinclair CD, Morrow JM, **Hanna MG**, Reilly MM, Yousry TA, Golay X, Thornton JS. Correcting radiofrequency inhomogeneity effects in skeletal muscle magnetisation transfer maps. NMR Biomed. 2011 Jul 27. PMID: 21796708

140. Rajakulendran S, Parton M, Holton JL, **Hanna MG.** Clinical and pathological heterogeneity in late-onset partial merosin deficiency. Muscle Nerve. 2011 Oct;44(4):590-3. PMID: 21922472

141. Guergueltcheva V, Müller JS, Dusl M, Senderek J, Oldfors A, Lindbergh C, Maxwell S, Colomer J, Hanna MG, Lochmüller H et al. Congenital myasthenia syndrome with tubular aggregates caused by GFTP-1 mutations. J Neurol. 2011 Oct 6. PMID: 21975507

142. Rajakulendran S, Kaski D, **Hanna MG**. Neuronal P/Q-type calcium channel dysfunction in inherited disorders of the CNS.Nat Rev Neurol. 2012 Jan 17;8(2):86-96. Review. PMID: 22249839

143. Matthews E, Portaro S, Ke Q, Sud R, Haworth A, Davis MB, Griggs RC, **Hanna MG**. Acetazolamide efficacy in hypokalemic periodic paralysis and the predictive role of genotype. Neurology. 2011 Nov 29;77(22):1960-4. PMID: 22094484

2012

144. Lee HY, Huang Y, Bruneau N, Roll P, Roberson ED, Hermann M, Quinn E, Maas J, Edwards R, Ashizawa T, Baykan B, Bhatia K, Bressman S, Bruno MK, Brunt ER, Caraballo R, Echenne B, Fejerman N, Frucht S, Gurnett CA, Hirsch E, Houlden H, Jankovic J, Lee WL, Lynch DR, Mohammed S, Müller U, Nespeca MP, Renner D, Rochette J, Rudolf G, Saiki S, Soong BW, Swoboda KJ, Tucker S, Wood N, **Hanna M**, Bowcock AM, Szepetowski P, Fu YH, Ptáček LJ. Mutations in the gene PRRT2 cause paroxysmal kinesigenic dyskinesia with infantile convulsions. Cell Rep. 2012 Jan 26;1(1):2-12. PMID: 22832103

145. Burge JA, **Hanna MG**. Novel insights into the pathomechanisms of skeletal muscle channelopathies. Curr Neurol Neurosci Rep. 2012 Feb;12(1):62-9. Review. PMID: 22083238

146. Matthews E, Plotz PH, Portaro S, Parton M, Elliott P, Humbel RL, Holton JL, Keegan BM, **Hanna MG**. A case of necrotizing myopathy with proximal

weakness and cardiomyopathy. Neurology. 2012 May 8;78(19):1527-32. PMID: 22565568

147. Raja Rayan DL, Haworth A, Sud R, Matthews E, Fialho D, Burge J, Portaro S, Schorge S, Tuin K, Lunt P, McEntagart M, Toscano A, Davis MB, **Hanna MG**. A new explanation for recessive myotonia congenita: exon deletions and duplications in CLCN1. Neurology. 2012 Jun 12;78(24):1953-8. PMID: 22649220

148. Pitceathly RD, Smith C, Fratter C, Alston CL, He L, Craig K, Blakely EL, Evans JC, Taylor J, Shabbir Z, Deschauer M, Pohl U, Roberts ME, Jackson MC, Halfpenny CA, Turnpenny PD, Lunt PW, **Hanna MG** et al. Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain. 2012 Nov;135(Pt 11):3392-403. PMID: 23107649

149. Fratta P, Malik B, Gray A, La Spada AR, **Hanna MG**, Fisher EM, Greensmith L.FUS is not dysregulated by the spinal bulbar muscular atrophy androgen receptor polyglutamine repeat expansion. Neurobiol Aging. 2012 Oct 9. PMID: 23062703

150. Statland JM, Bundy BN, Wang Y, Rayan DR, Trivedi JR, Sansone VA, Salajegheh MK, Venance SL, Ciafaloni E, Matthews E, Meola G, Herbelin L, Griggs RC, Barohn RJ, **Hanna MG**; Consortium for Clinical Investigation of Neurologic Channelopathies. Mexiletine for symptoms and signs of myotonia in nondystrophic myotonia: a randomized controlled trial. JAMA. 2012 Oct 3;308(13):1357-65. PMID: 23032552

151. Statland JM, Bundy BN, Wang Y, Trivedi JR, Raja Rayan D, Herbelin L, Donlan M, McLin R, Eichinger KJ, Findlater K, Dewar L, Pandya S, Martens WB, Venance SL, Matthews E, Amato AA, **Hanna MG**, Griggs RC, Barohn RJ; CINCH Consortium. A quantitative measure of handgrip myotonia in non-dystrophic myotonia. Muscle Nerve. 2012 Oct;46(4):482-9. PMID: 22987687

152. Pitceathly RD, Tomlinson SE, Hargreaves I, Bhardwaj N, Holton JL, Morrow JM, Evans J, Smith C, Fratter C, Woodward CE, Sweeney MG, Rahman S, **Hanna MG**. Distal myopathy with cachexia: an unrecognised phenotype caused by dominantly-inherited mitochondrial polymerase γ mutations. J Neurol Neurosurg Psychiatry. 2013 Jan;84(1):107-10. PMID: 22933815

153. Pitceathly RD, Murphy SM, Cottenie E, Chalasani A, Sweeney MG, Woodward C, Mudanohwo EE, Hargreaves I, Heales S, Land J, Holton JL, Houlden H, Blake J, Champion M, Flinter F, Robb SA, Page R, Rose M, Palace J, Crowe C, Longman C, Lunn MP, Rahman S, Reilly MM, **Hanna MG**. Genetic dysfunction of MT-ATP6 causes axonal Charcot-Marie-Tooth disease. Neurology. 2012 Sep 11;79(11):1145-54. PMID: 22933740

154. Portaro S, Musumeci O, Rizzo V, Rodolico C, Sweeney MG, Buccafusca M, **Hanna MG**, Toscano A. Stiffness as a presenting symptom of an odd clinical

condition caused by multiple sclerosis and myotonia congenita. Neuromuscul Disord. 2012 Aug 23. PMID: 22921319

155. Tan SV, Z'graggen WJ, Boërio D, Rayan DL, Howard R, **Hanna MG**, Bostock H. Membrane dysfunction in Andersen-Tawil syndrome assessed by velocity recovery cycles. Muscle Nerve. 2012 Aug;46(2):193-203. PMID: 22806368

156. Pitceathly RD, Rahman S, **Hanna MG**. Single deletions in mitochondrial DNA--molecular mechanisms and disease phenotypes in clinical practice. Neuromuscul Disord. 2012 Jul;22(7):577-86. PMID: 22578526

157. Matthews E, Plotz PH, Portaro S, Parton M, Elliott P, Humbel RL, Holton JL, Keegan BM, **Hanna MG**. A case of necrotizing myopathy with proximal weakness and cardiomyopathy. Neurology. 2012 May 8;78(19):1527-32. PMID: 22565568

158. Pulkes T, Dejthevaporn C, Apiwattanakul M, Papsing C, **Hanna MG**. Paroxysmal neuromyotonia: a new sporadic channelopathy.Neuromuscul Disord. 2012 Jun;22(6):479-82. PMID: 22305523

159. Gardiner AR, Bhatia KP, Stamelou M, Dale RC, Kurian MA, Schneider SA, Wali GM, Counihan T, Schapira AH, Spacey SD, Valente EM, Silveira-Moriyama L, Teive HA, Raskin S, Sander JW, Lees A, Warner T, Kullmann DM, Wood NW, **Hanna MG**, Houlden H. PRRT2 gene mutations: from paroxysmal dyskinesia to episodic ataxia and hemiplegic migraine. Neurology. 2012 Nov 20;79(21):2115-21. PMID: 23077024

160. Raheem O, Penttilä S, Suominen T, Kaakinen M, Burge J, Haworth A, Sud R, Schorge S, Haapasalo H, Sandell S, Metsikkö K, **Hanna M**, Udd B. New immunohistochemical method for improved myotonia and chloride channel mutation diagnostics.Neurology. 2012 Nov 27;79(22):2194-2200. PMID: 23152584

161.Tomlinson SE, Bostock H, Grinton B, **Hanna MG**, Kullmann DM, Kiernan MC, Scheffer IE, Berkovic SF, Burke D. In vivo loss of slow potassium channel activity in individuals with benign familial neonatal epilepsy in remission. Brain. 2012 Oct;135(Pt 10):3144-52. PMID: 23065794

2013

162. Tomlinson SE, Rajakulendran S, Tan SV, Graves TD, Bamiou DE, Labrum RW, Burke D, Sue CM, Giunti P, Schorge S, Kullmann DM, **Hanna MG**. Clinical, genetic, neurophysiological and functional study of new mutations in episodic ataxia type 1. J Neurol Neurosurg Psychiatry. 2013 Jan 24. PMID: 23349320

163. Burge JA, **Hanna MG**, Schorge S. Non-genomic actions of progesterone and 17β-estradiol on the chloride conductance of skeletal muscle. Muscle Nerve. 2013 Apr 26. PMID: 23625574

164. Thorne T, Fratta P, **Hanna MG**, Cortese A, Plagnol V, Fisher EM, Stumpf MP.

Graphical modelling of molecular networks underlying sporadic inclusion body myositis. Mol Biosyst. 2013 Apr 17. PMID: 23595110

165. Horga A, Raja Rayan DL, Matthews E, Sud R, Fialho D, Durran SC, Burge JA, Portaro S, Davis MB, Haworth A, **Hanna MG**. Prevalence study of genetically defined skeletal muscle channelopathies in England. Neurology. 2013 Apr 16;80(16):1472-5. PMID: 23516313

166. Cortese A, Machado P, Morrow J, Dewar L, Hiscock A, Miller A, Brady S, Hilton-Jones D, Parton M, **Hanna MG**. Longitudinal observational study of sporadic inclusion body myositis: Implications for clinical trials. Neuromuscul Disord. 2013 May;23(5):404-12. PMID: 23489664

167. Rajakulendran S, Roberts J, Koltzenburg M, **Hanna MG**, Stewart H. Deletion of chromosome 12q21 affecting KCNC2 and ATXN7L3B in a family with neurodevelopmental delay and ataxia. J Neurol Neurosurg Psychiatry. 2013 Mar 9. PMID: 23475819

168. Malik B, Nirmalananthan N, Gray AL, La Spada AR, **Hanna MG**, Greensmith L. Co-induction of the heat shock response ameliorates disease progression in a mouse model of human spinal and bulbar muscular atrophy: implications for therapy. Brain. 2013 Mar;136(Pt 3):926-43. PMID: 23393146

169. Smith MD, Seth JH, **Hanna MG**, Panicker JN. Detrusor overactivity in Becker muscular dystrophy. Muscle Nerve. 2013 Mar;47(3):464-5. PMID: 23382079

170. Nesbitt V, Pitceathly RD, Turnbull DM, Taylor RW, Sweeney MG, Mudanohwo EE, Rahman S, **Hanna MG**, McFarland R. The UK MRC Mitochondrial Disease Patient Cohort Study: clinical phenotypes associated with the m.3243A>G mutation—implications for diagnosis and management. J Neurol Neurosurg Psychiatry. 2013 Jan 25. PMID: 23355809

171. Sarkozy A, Hicks D, Hudson J, Laval SH, Barresi R, Hilton-Jones D, et al. ANO5 Gene Analysis in a Large Cohort of Patients with Anoctaminopathy: Confirmation of Male Prevalence and High Occurrence of the Common Exon 5 Gene Mutation. Hum Mutat. 2013 Apr 18. PMID: 23606453

172. Robert D.S. Pitceathly, Shamima Rahman, Yehani Wedatilake, James M. Polke, Sebahattin Cirak, A. Reghan Foley, Anna Sailer, Matthew E. Hurles, Jim Stalker, Iain Hargreaves, Cathy E. Woodward, Mary G. Sweeney, Francesco Muntoni, Henry Houlden, UK10K Consortium, Jan-Willem Taanman, **Michael G. Hanna**. NDUFA4 Mutations Underlie Dysfunction of a Cytochrome c Oxidase Subunit Linked to Human Neurological Disease, Cell Reports, 06 June 2013 PMID: 23746447

173. Willis TA, HollingsworthK, Coombs A, Sveen M, Andersen S, Stojkovic T, **Hanna MG** et al. Quantitative muscle MRI as an assessment tool for monitoring disease

progression in LGMD2I: a multicentre longitudinal study, PLoS One, 2013 Aug 14;8(8). PMID: 23967145

174. Ke Q, **Hanna MG** et al. Rare disease centers for periodic paralysis: China vs the US and UK. Muscle Nerve. 2013 July 28. PMID: 23893386

175. Pfeffer G, Horvath R, Klopstock T, Mootha VK, Suomalainen A, Koene S, **Hanna MG** et al. New treatments for mitochondrial disease – no time to drop our standards. Nature Reviews Neurology, July 2013, PMID: 23817350

176. Morrow JM, Matthews E, Raja Rayan D, Fischmann A, Sinclair CDJ, Reilly MM, Thornton JS, **Hanna MG**, Yousry T. Muscle MRI reveals distinct abnormalities genetically proven non-dystrophic myotonias. Neuromuscular Disorders. 2013. PMID: 23810313

177. Pitceathly RDS, Tomlinson SE, Holton JL, Morrow JM, Rahman S, **Hanna MG** et al. Distal myopathy with cachexia: an unrecognized phenotype caused by dominantly-inherited mitochondrial polymerase y mutations. JNNP 84(1):107-110. Jan 2013. PMID: 22933815

178. Pitceathly RD, Rahman S, Wedatilake Y, Polke JM, Cirak S, Foley AR, Sailer A, Hurles ME, Stalker J, Hargreaves I, Woodward CE, Sweeney MG, Muntoni F, Houlden H, Taanman JW, **Hanna MG**; UK10K Consortium. NDUFA4 mutations underlie dysfunction of a cytochrome c oxidase subunit linked to human neurological disease. Cell Rep. 2013 Jun 27;3(6):1795-805. PMID: 23746447

179. Trivedi JR, Bundy B, Statland J, Salajegheh M, Rayan DR, Venance SL, Wang Y, Fialho D, Matthews E, Cleland J, Gorham N, Herbelin L, Cannon S, Amato A, Griggs RC, **Hanna MG**, Barohn RJ; CINCH Consortium. Non-dystrophic myotonia: prospective study of objective and patient reported outcomes. Brain. 2013 Jul;136(Pt 7):2189-200. PMID: 23771340

180. Fratta P, **Hanna MG**, Fisher EM, Sidle K. An unusual presentation for SOD1-ALS:Isolated facial diplegia. Muscle Nerve. 2013 Jul 19. PMID: 23873540

181. Morrow JM, Reilly MM, **Hanna MG**. Reliability and accuracy of skeletal muscle imaging in limb-girdle muscular dystrophies. Neurology. 2013 Jun 11;80(24):2276. PMID: 23905174

182. Tan SV, Z'graggen WJ, Boërio D, Rayan DR, Norwood F, Ruddy D, Howard R, **Hanna MG**, Bostock H. Chloride channels in myotonia congenita assessed by velocity recovery cycles. Muscle Nerve. 2013 Sep 4. PMID: 24037712

183. Fratta P, Collins T, Pemble S, Nethisinghe S, Devoy A, Giunti P, Sweeney MG, **Hanna MG**, Fisher EM. Sequencing analysis of the spinal bulbar muscular atrophy CAG expansion reveals absence of repeat interruptions. Neurobiol Aging. 2013 Sep 13. PMID: 24041967

184. Turner C, Hilton-Jones D, Lochmüller H, **Hanna MG**. MRC Centre for Neuromuscular Diseases 1st (1st December 2010), and 2nd (2nd May 2012) myotonic dystrophy workshops, London, UK and the myotonic dystrophy standards of care and national registry meeting, Newcastle, UK July 2011. Neuromuscul Disord. 2013 Sep 18. PMID: 24054840

185. Spillane J, Fialho D, Hanna MG. Diagnosis of skeletal muscle channelopathies. Expert Opin Med Diagn. 2013 Sep 26. PMID: 24066928

186. Machado P, Brady S, Hanna MG. Update in inclusion body myositis. Curr Opin Rheumatol. 2013 Nov;25(6):763-71. PMID: 24067381

187. Pitceathly RD, Taanman JW, Rahman S, Meunier B, Sadowski M, Cirak S, Hargreaves I, Land JM, Nanji T, Polke JM, Woodward CE, Sweeney MG, Solanki S, Foley AR, Hurles ME, Stalker J, Blake J, Holton JL, Phadke R, Muntoni F, Reilly MM, **Hanna MG**; for the UK10K Consortium. COX10 Mutations Resulting in Complex Multisystem Mitochondrial Disease That Remains Stable Into Adulthood. JAMA Neurol. 2013 Oct 7. PMID: 24100867

188. Jaffer F, Reilly MM, Quinlivan R, Muntoni F, Turner C, Parton M, Lunn M, Hilton-Jones D, Korkodilos M, **Hanna MG**. Emergency neuromuscular admissions are avoidable: a regional audit of unplanned hospital admissions of neuromuscular patients 2009-2011: final results and recommendations. J Neurol Neurosurg Psychiatry. 2013 Nov;84(11):e2. PMID: 24109054

189. Fischmann A, Morrow JM, Sinclair CD, Reilly MM, **Hanna MG**, Yousry T, Thornton JS. Improved anatomical reproducibility in quantitative lower-limb muscle MRI. J Magn Reson Imaging. 2013 Oct 7. PMID: 24123788

190. Graham CD, Weinman J, Sadjadi R, Chalder T, Petty R, **Hanna MG**, Turner C, Parton M, Maddison P, Radunovic A, Longman C, Robb Y, Bushby K, Hilton-Jones D, Rose MR. A multicentre postal survey investigating the contribution of illness perceptions, coping and optimism to quality of life and mood in adults with muscle disease. Clin Rehabil. 2013 Nov 15. PMID: 24240060

191. Cortese A, Plagnol V, Brady S, Simone R, Lashley T, Acevedo-Arozena A, de Silva R, Greensmith L, Holton J, **Hanna MG,** Fisher EM, Fratta P. Widespread RNA metabolism impairment in sporadic inclusion body myositis TDP43-proteinopathy. Neurobiol Aging. 2013 Dec 30. PMID: 24462217

192. Rose MR, et al. ENMC International Workshop: Inclusion Body Myositis, 2-4 December 2011, Naarden, The Netherlands. Neuromuscular Disorders. 2013 Dec; 23(12):1044:55. PMID: 24268584

2014

193. Graves TD, Cha YH, Hahn AF, Barohn R, Salajegheh MK, Griggs RC, Bundy BN, Jen JC, Baloh RW, **Hanna MG**; CINCH Investigators. Episodic ataxia type 1:

clinical characterization, quality of life and genotype-phenotype correlation. Brain. 2014 Apr;137(Pt 4):1009-18. PMID: 24578548

194. Willis TA, Hollingsworth KG, Coombs A, Sveen ML, Andersen S, Stojkovic T, Eagle M, Mayhew A, de Sousa PL, Dewar L, Morrow JM, Sinclair CD, Thornton JS, Bushby K, Lochmuller H, **Hanna MG**, Hogrel JY, Carlier PG, Vissing J, Straub V. Quantitative magnetic resonance imaging in limb-girdle muscular dystrophy 2I: a multinational cross-sectional study. PLoS One. 2014 Feb 28;9(2) PMID: 24587344

195. Foley AR, Pitceathly RD, He J, Kim J, Pearson NM, Muntoni F, **Hanna MG**. Whole-genome sequencing and the clinician: a tale of two cities. J Neurol Neurosurg Psychiatry. 2014 Apr 4. PMID: 24706943

196. Morrow JM, Sinclair CD, Fischmann A, Reilly MM, **Hanna MG**, Yousry TA, Thornton JS. Reproducibility, and age, body-weight and gender dependency of candidate skeletal muscle MRI outcome measures in healthy volunteers. Eur Radiol. 2014 Apr 20. PMID: 24748539

197. Birbeck GL, **Hanna MG**, Griggs RC. Global opportunities and challenges for clinical neuroscience. JAMA. 2014 Apr 23-30;311(16):1609-10. PMID: 24756506

198. Fratta P, Nirmalananthan N, Masset L, Skorupinska I, Collins T, Cortese A, Pemble S, Malaspina A, Fisher EM, Greensmith L, **Hanna MG**. Correlation of clinical and molecular features in spinal bulbar muscular atrophy. Neurology. 2014 May 9. PMID: 24814851

199. Montague K, Malik B, Gray AL, La Spada AR, **Hanna MG**, Szabadkai G, Greensmith L. Endoplasmic reticulum stress in spinal and bulbar muscular atrophy: a potential target for therapy. Brain. 2014 Jul;137(Pt 7):1894-906. PMID: 24898351

200. Horga A, Pitceathly RDS, Blake JC, Woodward CE, Zapater P, Fratter C, Mudanohwo EE, Plant GT, Houlden H, Sweeney MG, **Hanna MG**, Reilly MM. Peripheral neuropathy predicts nuclear gene defect in patients with mitochondrial ophthalmoplegia. Brain. 2014 Dec;137(Pt 12):3200-12. PMID: 25281868

201. Matthews E, **Hanna MG**. Repurposing of sodium channel antagonists as potential new anti-myotonic drugs. Exp Neurol. 2014 Nov;261:812-5. PMID: 25218042

202. Singh RR, Tan SV, **Hanna MG**, Robb SA, Clarke A, Jungbluth H. Mutations in SCN4A: a rare but treatable cause of recurrent life-threatening laryngospasm. Pediatrics. 2014 Nov;134(5):e1447-50. PMID: 25311598

203. Corrochano S, Männikkö R, Joyce PI, McGoldrick P, Wettstein J, Lassi G, Raja Rayan DL, Blanco G, Quinn C, Liavas A, Lionikas A, Amior N, Dick J, Healy EG, Stewart M, Carter S, Hutchinson M, Bentley L, Fratta P, Cortese A, Cox R, Brown SD, Tucci V, Wackerhage H, Amato AA, Greensmith L, Koltzenburg M, **Hanna MG**, Acevedo-Arozena A. Novel mutations in human and mouse SCN4A implicate

AMPK in myotonia and periodic paralysis. Brain. 2014 Dec;137(Pt 12):3171-85. PMID: 25348630

204. Gang Q, Bettencourt C, Machado P, **Hanna MG**, Houlden H. Sporadic inclusion body myositis: the genetic contributions to the pathogenesis. Orphanet J Rare Dis. 2014 Jun 19;9:88. PMID: 24948216

205. Suetterlin K, Männikkö R, **Hanna MG**. Muscle channelopathies: recent advances in genetics, pathophysiology and therapy. Curr Opin Neurol. 2014 Oct;27(5):583-90. PMID: 25188014

206. Quinlivan R, Matthews E, **Hanna MG**. Innovative care model for patients with complex muscle diseases. Curr Opin Neurol. 2014 Oct;27(5):607-13. PMID: 25188015

2015

207. Fratta P, **Hanna MG**. Neuromuscular diseases: progress in gene discovery drives diagnostics and therapeutics. Lancet Neurology. 2015 Jan;14(1):13-4. PMID: 25496885

208. Gang, Q., Bettencourt, C., Machado, P.M., Fox, Z., Brady, S., Healy, E., Parton, M., Holton, J.L., Hilton-Jones, D., Shieh, P.B., Zanoteli, E., De Paepe, B., De Bleecker, J., Shaibani, A., Ripolone, M., Violano, R., Moggio, M., Barohn, R.J., Dimachkie, M.M., Mora, M., Mantegazza, R., Zanotti, S., **Hanna, M.G**., Houlden, H., The Muscle Study Group and The International IBM Genetics Consortium, The effects of an intronic polymorphism in TOMM40 and APOE genotypes in sporadic inclusion body myositis, Neurobiology of Aging. 2015, Jan. PMID: 25670332

209. Megan K Herbert, Judith Stammen-Vogelzangs, Marcel M Verbeek, Anke Rietveld, Ingrid E Lundberg, Hector Chinoy, Janine A Lamb, Robert G Cooper, Mark Roberts, Umesh A Badrising, Jan L De Bleecker, Pedro M Machado, **Michael G Hanna**, Lenka Plestilova, Jiri Vencovsky, Baziel van Engelen, Ger J M Pruijn. Disease specificity of autoantibodies to cytosolic 5′-nucleotidase 1A in sporadic inclusion body myositis versus known autoimmune diseases. *Ann Rheum Dis.* 2015 Feb 24. PMID: 25714931

210. Sewry CA, Holton JL, Dick DJ, Muntoni F, **Hanna MG**. Zebra body myopathy is caused by a mutation in the skeletal muscle actin gene (ACTA1). *Neuromuscul Disord*. 2015 Feb 14. PMID: 25747004

211. Hall AM, Vilasi A, Garcia-Perez I, Lapsley M, Alston CL, Pitceathly RD, McFarland R, Schaefer AM, Turnbull DM, Beaumont NJ, Hsuan JJ, Cutillas PR, Lindon JC, Holmes E, Unwin RJ, Taylor RW, Gorman GS, Rahman S, **Hanna MG**. The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. Kidney Int. 2015 Mar;87(3):610-22. PMID: 25207879

212. Broomfield A, Sweeney MG, Woodward CE, Fratter C, Morris AM, Leonard JV, Abulhoul L, Grunewald S, Clayton PT, **Hanna MG**, Poulton J, Rahman S. Paediatric single mitochondrial DNA deletion disorders: an overlapping spectrum of disease. J Inherit Metab Dis. 2015 May;38(3):445-57. PMID: 25352051

213. Pitceathly RD, Morrow JM, Sinclair CD, Woodward C, Sweeney MG, Rahman S, Plant GT, Ali N, Bremner F, Davagnanam I, Yousry TA, **Hanna MG**, Thornton JS. Extra-ocular muscle MRI in genetically-defined mitochondrial disease. Eur Radiol. 2015 May 21. PMID: 25994195

214. Figueroa-Bonaparte S, Hudson J, Barresi R, Polvikoski T, Williams T, Töpf A, Harris E, Hilton-Jones D, Petty R, Willis TA, Longman C, Dougan CF, Parton MJ, **Hanna MG**, Quinlivan R, Farrugia ME, Guglieri M, Bushby K, Straub V, Lochmüller H, Evangelista T. padMutational spectrum and phenotypic variability of VCP-related neurological disease in the UK. J Neurol Neurosurg Psychiatry. 2015 Jun 23. PMID: 26105173

215. Evangelista T, **Hanna MG**, Lochmüller H. Congenital Myasthenic Syndromes with Predominant Limb Girdle Weakness. Journal of neuromuscular diseases 2(Suppl 2):S21-S29 Jul 2015

216. Jaffer F, Avbersek A, Vavassori R, Fons C, Campistol J, Stagnaro M, De Grandis E, Veneselli E, Rosewich H, Gianotta M, Zucca C, Ragona F, Granata T, Nardocci N, Mikati M, Helseth AR, Boelman C, Minassian BA, Johns S, Garry SI, Scheffer IE, Gourfinkel-An I, Carrilho I, Aylett SE, Parton M, **Hanna MG**, Houlden H, Neville B, Kurian MA, Novy J, Sander JW, Lambiase PD, Behr ER, Schyns T, Arzimanoglou A, Cross JH, Kaski JP, Sisodiya SM. Faulty cardiac repolarization reserve in alternating hemiplegia of childhood broadens the phenotype. Brain. 2015 Oct;138(Pt 10):2859-74. PMID: 26297560

217. Habbout K, Poulin H, Rivier F, Giuliano S, Sternberg D, Fontaine B, Eymard B,Morales RJ, Echenne B, King L, **Hanna MG**, Männikkö R, Chahine M, Nicole S,Bendahhou S. A recessive Nav1.4 mutation underlies congenital myasthenic syndrome with periodic paralysis. Neurology. 2016 Jan 12;86(2):161-9. Dec 11. PMID: 26659129

218. Suetterlin KJ, Bugiardini E, Kaski JP, **Morrow JM**, Matthews E, **Hanna MG**, Fialho D. Long-term Safety and Efficacy of Mexiletine for Patients With Skeletal Muscle Channelopathies. JAMA Neurol. 2015 Dec;72(12):1531-3. PMID: 26658970

219. Poole OV, **Hanna MG**, Pitceathly RD. Mitochondrial disorders: disease mechanisms and therapeutic approaches. Discov Med. 2015 Nov;20(111):325-31. PMID: 26645904

220. Gang Q, Bettencourt C, **Houlden H**, **Hanna MG**, Machado PM. Genetic advances in sporadic inclusion body myositis. Curr Opin Rheumatol. 2015 Nov;27(6):586-94. Review. PMID: 26335925

221. Gardiner AR, Jaffer F, Dale RC, Labrum R, Erro R, Meyer E, Xiromerisiou G, Stamelou M, Walker M, Kullmann D, **Hanna MG** et al. The clinical and genetic heterogeneity of paroxysmal dyskinesias. Brain. 138:3567-3580. 1 Dec 2015. PMID: 26598494

2016

222. Spillane J, Kullmann DM, **Hanna MG**. Genetic neurological channelopathies: molecular genetics and clinical phenotypes. J Neurol Neurosurg Psychiatry. 2016 Jan;87(1):37-48. Review. PMID: 26558925

223. Morrow JM, Sinclair CD, Fischmann A, Machado PM, Reilly MM, Yousry TA,Thornton JS, **Hanna MG**. MRI biomarker assessment of neuromuscular disease progression: a prospective observational cohort study. Lancet Neurol. 2016 Jan;15(1):65-77. PMID: 26549782

224. Rothwell S, Cooper RG, Lundberg IE, Miller FW, Gregersen PK, Bowes J,Vencovsky J, Danko K, Limaye V, Selva-O'Callaghan A, **Hanna MG**, Machado PM, Myositis Genetics Consortium et al. Dense genotyping of immune-related loci in idiopathic inflammatory myopathies confirms HLA alleles as the strongest genetic risk factor and suggests different genetic background for major clinical subgroups. Ann Rheum Dis. 2016 Aug;75(8):1558-66. PMID: 26362759

225. Gang Q, Bettencourt C, Machado PM, Brady S, Holton JL, Pittman AM, Hughes D, Healy E, Parton M, Hilton-Jones D, Shieh PB, Needham M, **Hanna MG**, Houlden H; Muscle Study Group, The International IBM Genetics Consortium et al. Rare variants in SQSTM1 and VCP genes and risk of sporadic inclusion body myositis. Neurobiol Aging. 2016 Aug 8. PMID: 27594680

226. Akman G, Desai R, Bailey LJ, Yasukawa T, Dalla Rosa I, Durigon R, Holmes JB, Moss CF, Mennuni M, Houlden H, Crouch RJ, **Hanna MG**, Pitceathly RD, Spinazzola A, Holt IJ. Pathological ribonuclease H1 causes R-loop depletion and aberrant DNA segregation in mitochondria. Proc Natl Acad Sci U S A. 2016 Jul 8. PMID: 27402764

227. Mestre TA, Manole A, MacDonald H, Riazi S, Kraeva N, **Hanna MG**, Lang AE, Männikkö R, Yoon G. A novel KCNA1 mutation in a family with episodic ataxia and malignant hyperthermia. Neurogenetics. 2016 Jun 8. PMID: 27271339

227. Bertolin C, Querin G, Da Re E, Sagnelli A, Bello L, Cao M, Muscas M, Pennuto M, Ermani M, Pegoraro E, Mariotti C, Gellera C, **Hanna MG**, Pareyson D, Fratta P,Sorarù G. No effect of AR polyG polymorphism on spinal and bulbar muscular atrophy phenotype. Eur J Neurol. 2016 Jun;23(6):1134-6. PMID: 27141859

229. Scalco RS, Gardiner AR, Pitceathly RD, Hilton-Jones D, Schapira AH, Turner C, Parton M, Desikan M, Barresi R, Marsh J, Manzur AY, Childs AM, Feng L, Murphy E, Lamont PJ, Ravenscroft G, Wallefeld W, Davis MR, Laing NG, Holton JL, Fialho D, Bushby K, **Hanna MG** et al. CAV3 mutations causing exercise intolerance,

myalgia and rhabdomyolysis: Expanding the phenotypic spectrum of caveolinopathies. Neuromuscul Disord. 2016 May 11. PMID: 27312022

230. Tan SV, Z'graggen WJ, Boërio D, Turner C, **Hanna MG**, Bostock H. In vivo assessment of muscle membrane properties in myotonic dystrophy. Muscle Nerve. 2016 Aug;54(2):249-57. PMID: 26789642

231. Rebelo AP, Abrams AJ, Cottenie E, Horga A, Gonzalez M, Bis DM, Sanchez-Mejias A, Pinto M, Buglo E, Markel K, Prince J, Laura M, Houlden H, Blake J, Woodward C, Sweeney MG, Holton JL, **Hanna M**, Dallman JE, Auer-Grumbach M, Reilly MM, Zuchner S. Cryptic Amyloidogenic Elements in the 3' UTRs of Neurofilament Genes Trigger Axonal Neuropathy. Am J Hum Genet. 2016 Apr 7;98(4):597-614. PMID: 27040688

232. Ahmed M, Machado PM, Miller A, Spicer C, Herbelin L, He J, Noel J, Wang Y, McVey AL, Pasnoor M, Gallagher P, Statland J, Lu CH, Kalmar B, Brady S, Sethi H, Samandouras G, Parton M, Holton JL, Weston A, Collinson L, Taylor JP, Schiavo G, **Hanna MG**, Barohn RJ, Dimachkie MM, Greensmith L. Targeting protein homeostasis in sporadic inclusion body myositis. Sci Transl Med. 2016 Mar 23;8(331):331ra41. PMID: 27009270

233. Tomlinson SE, Tan SV, Burke D, Labrum RW, Haworth A, Gibbons VS, Sweeney MG, Griggs RC, Kullmann DM, Bostock H, **Hanna MG**. In vivo impact of presynaptic calcium channel dysfunction on motor axons in episodic ataxia type 2. Brain. 2016 Feb;139(Pt 2):380-91. PMID: 26912519

234. Sansone VA, Burge J, McDermott MP, Smith PC, Herr B, Tawil R, Pandya S, Kissel J, Ciafaloni E, Shieh P, Ralph JW, Amato A, Cannon SC, Trivedi J, Barohn R, Crum B, Mitsumoto H, Pestronk A, Meola G, Conwit R, **Hanna MG**, Griggs RC; Muscle Study Group. Randomized, placebo-controlled trials of dichlorphenamide in periodic paralysis. Neurology. 2016 Apr 12;86(15):1408-16. PMID: 26865514

235. Rajakulendran S, Pitceathly RD, Taanman JW, Costello H, Sweeney MG, Woodward CE, Jaunmuktane Z, Holton JL, Jacques TS, Harding BN, Fratter C, **Hanna MG**, Rahman S. A Clinical, Neuropathological and Genetic Study of Homozygous A467T POLG-Related Mitochondrial Disease. PLoS One. 2016 Jan 6;11(1):e0145500. PMID: 26735972

236. Rajakulendran S, **Hanna MG**. The Role of Calcium Channels in Epilepsy. ColdSpring Harb Perspect Med. 2016 Jan 4;6(1):a022723. Review. PMID: 26729757

237. Zaharieva IT, Thor MG, Oates EC, van Karnebeek C, Hendson G, Blom E, Witting N, Rasmussen M, Gabbett MT, Ravenscroft G, Sframeli M, Suetterlin K, Sarkozy A, **Hanna MG** et al. Loss-of- function mutations in SCN4A cause severe foetal hypokinesia or 'classical' congenital myopathy. Brain. 2016 Mar;139(Pt 3):674-91. PMID: 26700687

238. Gilbert JR, Symmonds M, **Hanna MG**, Dolan RJ, Friston KJ, Moran RJ. Profiling neuronal ion channelopathies with non-invasive brain imaging and dynamic causal models: Case studies of single gene mutations. Neuroimage. 2016 Jan 1;124(Pt A):43-53. PMID: 26342528

239. **Hanna MG**. Lord Walton of Detchant - Obituary. Eur J Neurol. 2016 Jul;23(7):1137-8. PMID: 27272108

240. Parkes JE, Rothwell S, Day PJ, McHugh NJ, Betteridge ZE, Cooper RG, Ollier WE, Chinoy H, Lamb JA; Myositis Genetics Consortium (MYOGEN). Systematic protein-protein interaction and pathway analyses in the idiopathic inflammatory myopathies. Arthritis Research and Therapy. 2016 Jul 7;18(1):156. PMID: 27388770

241. Horga A, Tomaselli PJ, Gonzalez MA, Laurà M, Muntoni F, Manzur AY, **Hanna MG**, Blake JC, Houlden H, Züchner S, Reilly MM. SIGMAR1 mutation associated with autosomal recessive Silver-like syndrome. Neurology. 2016 Oct 11;87(15):1607-1612. PMID: 27629094

242. Ricotti V, Evans MR, Sinclair CD, Butler JW, Ridout DA, Hogrel JY, Emira A, Morrow JM, Reilly MM, **Hanna MG**, Janiczek RL, Matthews PM, Yousry TA, Muntoni F,Thornton JS. Upper Limb Evaluation in Duchenne Muscular Dystrophy: Fat-Water Quantification by MRI, Muscle Force and Function Define Endpoints for Clinical Trials. PLoS One. 2016 Sep 20;11(9):e0162542. PMID: 27649492

243. Scalco RS, Skorupinska I, Blochet C, Habib M, Matthews E, Morrow J, **Hanna MG**, Fialho D. Bumetanide in hypokalaemic periodic paralysis: a randomised, double-blind, placebo controlled phase II clinical trial with a crossover design. Neuromuscular Disorders 26 (Supplement):S197-26:S197. 09 Sep 2016

244. Sinclair CD, Morrow JM, Janiczek RL, Evans MR, Rawah E, Shah S, **Hanna MG**, Reilly MM, Yousry TA, Thornton JS. Stability and sensitivity of water T(2) obtained with IDEAL-CPMG in healthy and fat-infiltrated skeletal muscle. NMR Biomed. 2016 Dec;29(12):1800-1812. PMID: 27809381

245. Brady S, Healy EG, Gang Q, Parton M, Quinlivan R, Jacob S, Curtis E, Al-Sarraj S, Sewry CA, **Hanna MG**, Houlden H, Beeson D, Holton JL. Tubular Aggregates and Cylindrical Spirals Have Distinct Immunohistochemical Signatures. J Neuropathol Exp Neurol. 2016 Dec;75(12):1171-1178. PMID: 27941137

246. Rothwell S, Cooper RG, Lundberg IE, Gregersen PK, **Hanna MG**, Machado PM, Myositis Genetics Consortium et al. Immune-Array Analysis in Sporadic Inclusion Body Myositis Reveals HLA-DRB1 Amino Acid Heterogeneity Across the Myositis Spectrum. Arthritis Rheumatol. 2017 Jan 13. PMID: 28086002

247. Manole A, Männikkö R, **Hanna MG**; SYNAPS study group., Kullmann DM, Houlden H. De novo KCNA2 mutations cause hereditary spastic paraplegia. Ann Neurol. 2017 Feb;81(2):326-328. PMID: 28032718

248. Gonorazky HD, Marshall CR, Al-Murshed M, Hazrati LN, Thor MG, **Hanna MG**, Männikkö R, Ray PN, Yoon G. Congenital myopathy with "corona" fibres, selective muscle atrophy, and craniosynostosis associated with novel recessive mutations in SCN4A. Neuromuscul Disord. 2017 Feb 8. PMID: 28262468

249. Jaffer F, Fawcett K, Sims D, Heger A, Houlden H, **Hanna MG**, Kingston H, Sisodiya SM. Familial childhood-onset progressive cerebellar syndrome associated with the ATP1A3 mutation. Neurol Genet. 2017 Mar 27;3(2):e145 PMID: 28382329

250. Zollo M, Ahmed M, Ferrucci V, Salpietro V, Asadzadeh F, Carotenuto M, Maroofian R, Al-Amri A, Singh R, Scognamiglio I, Mojarrad M, Musella L, Duilio A, Di Somma A, Karaca E, Rajab A, Al-Khayat A, Mohan Mohapatra T, Eslahi A, **Hanna MG** et al. PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain. 2017 Apr 1;140(4):940-952. PMID: 28334956

251. Palmio J, Sandell S, **Hanna MG**, Männikkö R, Penttilä S, Udd B. Predominantly myalgic phenotype caused by the c.3466G>A p.A1156T mutation in SCN4A gene. Neurology. 2017 Apr 18;88(16):1520-1527. PMID: 28330959

252. Lilleker JB, Rietveld A, Pye SR, Mariampillai K, Benveniste O, Peeters MT, Miller JA, **Hanna MG**, Machado PM, Parton MJ, Gheorghe KR, Badrising UA, Lundberg IE, Sacconi S, Herbert MK, McHugh NJ, Lecky BR, Brierley C, Hilton-Jones D, Lamb JA, Roberts ME, Cooper RG, Saris CG, Pruijn GJ, Chinoy H, van Engelen BG; all UKMYONET contributors. Cytosolic 5'-nucleotidase 1A autoantibody profile and clinical characteristics in inclusion body myositis. Ann Rheum Dis. 2017 May;76(5):862-868. PMID: 28122761

253. Matthews E, **Hanna MG**. Dichlorphenamide efficacy in the primary periodic paralyses. Expert Opinion on Orphan Drugs. Vol 5, 2017, issue 3, 285-290

254. Bugiardini E, Poole OV, Manole A, Pittman AM, Horga A, Hargreaves I, Woodward CE, Sweeney MG, Holton JL, Taanman JW, Plant GT, Poulton J, Zeviani M, Ghezzi D, Taylor J, Smith C, Fratter C, Kanikannan MA, Paramasivam A, Thangaraj K, Spinazzola A, Holt IJ, Houlden H, **Hanna MG**, Pitceathly RDS. Clinicopathologic and molecular spectrum of RNASEH1-related mitochondrial disease. Neurol Genet. 2017 May 2;3(3):e149. PMID: 28508084

255. Horga A, Laurà M, Jaunmuktane Z, Jerath NU, Gonzalez MA, Polke JM, Poh R, Blake JC, Liu YT, Wiethoff S, Bettencourt C, Lunn MP, Manji H, **Hanna MG**, Houlden H, Brandner S, Züchner S, Shy M, Reilly MM. Genetic and clinical characteristics of NEFL-related Charcot-Marie-Tooth disease. J Neurol Neurosurg Psychiatry. 2017 May 13. PMID: 28501821

256. Matthews E, Silwal A, Sud R, **Hanna MG**, Manzur AY, Muntoni F, Munot P. Skeletal Muscle Channelopathies: Rare Disorders with Common Pediatric Symptoms. J Pediatr. 2017 Jun 26. PMID: 28662944

257. Bugiardini E, Rossor AM, Lynch DS, Swash M, Pittman AM, Blake JC, **Hanna MG**, Houlden H, Holton JL, Reilly MM et al. Homozygous mutation in HSPB1 causing distal vacuolar myopathy and motor neuropathy. Neurology. Genetics 3(4):e168 Aug 2017. PMID: 28702508

258. Reza M, Cox D, Phillips L, Johnson D, Manoharan V, Grieves M, Davis B, Roos A, Morgan J, **Hanna MG**, Muntoni F, Lochmüller H. MRC Centre Neuromuscular Biobank (Newcastle and London): Supporting and facilitating rare and neuromuscular disease research worldwide. Neuromuscul Disord. 2017 Jul 10. PMID: 28864117

259. Manole A, Jaunmuktane Z, Hargreaves I, Ludtmann MHR, Salpietro V, Bello OD, Pope S, Pandraud A, Horga A, Scalco RS, Li A, Ashokkumar B, Lourenço CM, Heales S, Horvath R, Chinnery PF, Toro C, Singleton AB, Jacques TS, Abramov AY, Muntoni F, **Hanna MG**, Reilly MM, Revesz T, Kullmann DM, Jepson JEC, Houlden H. Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain. 2017 Nov 1;140(11):2820-2837. PMID: 29053833

260. Mariot V, Joubert R, Hourdé C, Féasson L, **Hanna MG**, Muntoni F, Maisonobe T, Servais L, Bogni C, Panse R, Benvensite O, Stojkovic T, Machado PM, Voit T, Buj-Bello A, Dumonceaux J. Reversible downregulation of myostatin pathway in neuromuscular diseases explains challenges of antimyostatin therapeutic approaches. Nature Communications. 2017 Nov 30;8 (1): 1859 PMID: 29192144

261. Männikkö R, Shenkarev ZO, Thor MG, Berkut AA, Myshkin MY, Paramonov AS, Kulbatskii DS, Kuzmin DA, Sampedro Castañeda M, King L, Wilson ER, Lyukmanova EN, Kirpichnikov MP, Schorge S, Bosmans F, H**anna MG**, Kullmann DM, Vassilevski AA. Spider toxin inhibits gating pore currents underlying periodic paralysis. Proc Natl Acad Sci USA. 2018 Apr 10. PMID: 29636418

262. Männikkö R, Wong L, Tester DJ, Thor MG, Sud R, Kullmann DM, Sweeney MG, Leu C, Sisodiya SM, FitzPatrick DR, Evans MJ, Jeffrey IJM, Tfelt-Hansen J, Cohen MC, Fleming PJ, Jaye A, Simpson MA, Ackerman MJ, **Hanna MG**, Behr ER, Matthews E. Dysfunction of NaV1.4, a skeletal muscle voltage-gated sodium channel, in sudden infant death syndrome: a case-control study. Lancet. 2018 Mar 28. PMID: 29605429

263. Tan SV, Z;Graggen WJ, **Hanna MG**, Bostock H. In vivo assessment of muscle membrane properties in the sodium channel myotonias. Muscle Nerve. 2018, Apr; 57(4):586-59417. PMID 28877545

264. Statland JM, Fontaine B, **Hanna MG**, Johnson NE, Kissel JT, Sansone VA, Shieh PB, Tawil RN, Trivedi J, Cannon SC, Griggs RC. Review of the Diagnosis and Treatment of Periodic Paralysis. Muscle Nerve, 2018 Apr; 57(4):522-530. PMID: 29125635

265. Matthews E, Neuwirth C, Jaffer F, Scalco RS, Fialho D, Parton M, Raja Rayan D, Suetterlin K, Sud R, Spiegel R, Mein R, Houlden H, Schaefer A, Healy E, Palace J,

Quinlivan R, Treves S, Holton JL, Jungbluth H, **Hanna MG**. Atypical periodic paralysis and myalgia: A novel RYR1 phenotype. Neurology. 2018 Jan 30;90(5):e412-e418. PMID: 29298851

266. Keddie S, Jaunmuktane Z, Brandner S, Shah S, Maddison P, Rees JH, **Hanna MG**, Lunn MPT, Reilly MM, Rossor AM, Carr AS. A diagnostic conundrum. Pract Neurol. 2018 Apr;18(2):137-142. PMID: 29363556

267. Bishop CA, Ricotti V, Sinclair CDJ, Evans MRB, Butler JW, Morrow JM, **Hanna MG**, Matthews PM, Yousry TA, Muntoni F, Thornton JS, Newbould RD, Janiczek RL. Semi-Automated Analysis of Diaphragmatic Motion with Dynamic Magnetic Resonance Imaging in Healthy Controls and Non-Ambulant Subjects with Duchenne Muscular Dystrophy. Front Neurol. 2018 Jan 26;9:9. PMID: 29434565

268. O'Connor E, Vandrovcova J, Bugiardini E, Chelban V, Manole A, Davagnanam I, Wiethoff S, Pittman A, Lynch DS, Efthymiou S, Marino S, Manzur AY, Roberts M, **Hanna MG**, Houlden H, Matthews E, Wood NW. Mutations in XRCC1 cause cerebellar ataxia and peripheral neuropathy. J Neurol Neurosurg Psychiatry. 2018 Feb 22. PMID: 29472272

269. Ng YS, Lax NZ, Maddison P, Alston CL, Blakely EL, Hepplewhite PD, Riordan G, Meldau S, Chinnery PF, Pierre G, **Hanna MG** et al. MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine. 2018 Apr;30:86-93. PMID: 29506874

270. Matthews E, Hartley L, Sud R, **Hanna MG**, Muntoni F, Munot P. Acetazolamide can improve symptoms and signs in ion channel-related congenital myopathy. J Neurol Neurosurg Psychiatry. 2018 May 16. PMID: 29769250

271. Altamura C, Lucchiari S, Sahbani D, Ulzi G, Comi GP, D'Ambrosio P, Petillo R, Politano L, Vercelli L, Mongini T, Dotti MT, Cardani R, Meola G, Lo Monaco M, Matthews E, **Hanna MG**, Carratù MR, Conte D, Imbrici P, Desaphy JF. The analysis of myotonia congenita mutations discloses functional clusters of amino acids within CBS2 domain and C-terminal peptide of ClC-1 channel. Hum Mutat. 2018 Jun 23. PMID: 29935101

272. Luo S, Sampedro Castañeda M, Matthews E, Sud R, **Hanna MG**, Sun J, Song J, Lu J, Qiao K, Zhao C, Männikkö R. Hypokalaemic periodic paralysis and myotonia in a patient with homozygous mutation p.R1451L in NaV1.4. Sci Rep. 2018 Jun 26;8(1):9714. PMID: 29946067

273. Morrow JM, Evans MRB, Grider T, Sinclair CDJ, Thedens D, Shah S, Yousry TA, **Hanna MG**, Nopoulos P, Thornton JS, Shy ME, Reilly MM. Validation of MRC Centre MRI calfu muscle fat fraction protocol as an outcome measure in CMT1A. Neurology. 2018 Aug 17. PMID: 30120135

274. Ghosh SG, Becker K, Huang H, Salazar TD, Chai G, Salpietro V, Al-Gazali L, Waisfisz Q, Wang H, Vaux KK, Stanley V, Manole A, Akpulat U, Weiss MM,

Efthymiou S, **Hanna MG**, et al. Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. Am J Hum Genet. 2018 Jul 25. PMID: 30100084

275. Bugiardini E, Morrow JM, Shah S, Wood CL, Lynch DS, Pitmann AM, Reilly MM, Houlden H, Matthews E, Parton M, **Hanna MG**, Straub V, Yousry TA. The Diagnostic Value of MRI Pattern Recognition in Distal Myopathies. Front Neurol. 2018 Jun 26;9:456. PMID: 29997562

Other example publications selected abstracts, posters and oral presentations 1994-2017

1988

1 **Hanna MG**, Selam JL, Kayshap M, Albertii KGMM, Lozano J, Turner D, Jeandidier N, Chan E, Charles MA (1988). The effect of chronic intraperitoneal insulin administration on blood lipid, hormone and metabolite levels in type I diabetes. *Poster presentation to the Annual Conference of the European Association for the study of Diabetes. Paris. Diabetologia 4: 233*

1994

2 **Hanna MG**, Nelson I, Morgan-Hughes JA, Harding AE (1994). Studies on mitochondrial transcription and translation in human myoblasts harbouring the mitochondrial DNA tRNA lysine 8344 MERRF mutation. *Poster presentation: Gordon Conference on extrachromasomal elements, Volterra Italy.Published in proceedings*

3 **Hanna MG**, Nelson I, Morgan-Hughes JA, Harding AE (1994). A new mitochondrial DNA tRNA point mutation associated with an unusual mitochondrial encephalomyopathy phenotype comprising dementia and chorea. *Oral presentation at the European Neurology Society (ENS) Conference Barcelona. Journal of Neurology supplement I 241:S94*

4 **Hanna MG**, Nelson I, Morgan-Hughes JA, Harding AE (1994). Impaired mitochondrial translation in myoblasts harbouring the mitochondrial DNA tRNA lysine MERRF mutation.*Oral presentation at ENS 1994 Journal of Neurology supplement I 241: S94*

5 **Hanna MG**, Nelson IP, Sweeney MG, Morgan-Hughes JA, Harding AE (1994). New disease associated mitochondrial DNA mutations. *Poster presentation at the Bari Conference on Mitochondrial Pathology. Bari Italy & published in proceedings.*

6 **Hanna MG**, Nelson IP, Morgan-Hughes JA, Harding AE (1994) New mitochondrial DNA mutations associated with neurological disease. *Poster presentation at the American Society of Human Genetics Annual Conference, Montreal, Canada. American Journal of Human Genetics supplement 55 1020*

7 Nelson I, **Hanna MG**, Morgan-Hughes JA, Harding AE (1994). Studies on mitochondrial transcription and translation in human myoblasts harbouring the MERRF transfer RNA lysine 8344 mitochondrial DNA mutation. *Poster presentation at the American Society of Human Genetics Annual Conference Montreal, Canada. American Journal of Human Genetics supplement 55: 1019*

1995

8 **Hanna MG**, Nelson I, Sweeney MG, Morgan-Hughes JA, Harding AE (1995). Analysis of all mitochondrial transfer RNA genes in 20 patients with mitochondrial encephalomyopathies: identification of a new MELAS mutation and relationship of tRNA mutations to mitochondrial proliferation *Oral presentation at the ENS Munich. Journal of Neurology supplement 2 242;S42*

9 **Hanna MG**, Nelson IP, Morgan-Hughes JA, Harding AE (1995) Identification of a new MELAS mutation. *Poster presentation at the 3rd international meeting on human mitochondrial pathology (EUROMIT-III), Paris and published in proceedings.*

10 Nelson IP, **Hanna MG**, Harding AE. (1995) Comparison of □□ cybrid clones from two patients with the 3243 mtDNA mutation. *Poster presentation at EUROMIT-III and published in proceedings.*

11 Brockington M, Sweeney MG, **Hanna MG**, Woodard C, Morgan-Hughes JA, Harding AE (1995) Mitochondrial DNA rearrangements in human disease: genetic and clinical correlations.
Poster presentation at EUROMIT-III and published in proceedings.

1996

12 **Hanna MG**, Silburn PA, Mills KR, The BT, Larsson C, Wright GDS (1996). Familial episodic ataxia without myokymia: A clinical, electrophysiological and genetic study. *Poster presentation at the 4th International congress of movement disorders Vienna, Austria. Movement Disorders 11: supplement I, P104*

13 **Hanna MG**, Mills KR, Newsom-Davis J (1996). Primary orthostatic tremor with prominent muscle hypertrophy. *Poster presentation at the Association of British Neurologists Annual meeting Norwich Journal of Neurology, Neurosurgery and Psychiatry 61:226.*

14 Silburn PA, **Hanna MG**, Vaughan J, Renowden S, Squire M, Davis P, Greenhall RG. Two unusual clinical presentations of the mitochondrial A3243G point mutations in adult neurological practice (1996) *Poster presentation at the Association of British Neurologists Annual meeting Norwich Journal of Neurology, Neurosurgery and Psychiatry 61:224*

15 Nelson IP, **Hanna MG**, Wood NW, Harding AE (1996) Depletion of untransformed fibroblasts with a nucleoside analogue: A model for the study of nuclear-mitochondrial interactions. *Poster presentation at European meeting on Biogenesis of Mitochondria and Nuclear-mitochondrial interactions. Aussois, France & published in meeting proceedings.*

1997

16 **Hanna MG**, Sweeney MG, Noursadeghi M, Ellis CJ, Wood NW, Marsden CD (1997). A case of genetically confirmed Friedreich's ataxia presenting with chorea. *Poster presentation at the Association of British Neurologists annual meeting Leicester Journal of Neurology, Neurosurgery and Psychiatry 63:268.*

17 **Hanna MG**, Nelson IP, Rahman S, Schapira AHV, Morgan-Hughes JA, Wood NW (1997). Cytochrome c Oxidase deficiency associated with a new mitochondrial DNA

stop codon mutation.*Poster presentation to the American Association of Neurologists 122 Annual meeting San Diego USA. Annals of Neurology 42: 412 M79*

18 **Hanna MG**, Nelson IP. Morgan-Hughes JA, Wood NW (1997) The first mitochondrial DNA stop codon mutation is associated with recurrent encephalopathy and cytochrome c oxidase deficiency. *Oral presentation to the Oxford annual muscle meeting and published in proceedings.*

19 **Hanna MG**, Brockington M, Sweeney MG, Morgan-Hughes JA, Wood NW (1997) Mitochondrial DNA and human disease: clinical and genetic correlations *Oral presentation at the Association of British Neurologists annual autumn meeting. Journal of Neurology, Neurosurgery and Psychiatry 68:233*

20 **Hanna MG**, Nelson IP, Rahmann S, Cooper M, Schapira AHV, Morgan-Hughes JA Wood NW (1997) A new mitochondrial DNA stop codon point mutation. *Poster presentation at the World Muscle Society annual meeting in Tunis. Neuromuscular Disorders 7:450:GP3.10*

1998

21 Rahman S, Lake BD, Taanman J-W, Cooper JM, **Hanna MG**, Leonard JV, Schapira AHV. Patterns of subunit loss in cytochrome oxidase deficiency (1998) *Poster presentation to the Society for the study of inborn errors of metabolism, Goteborg, Sweeden Neuropaediatrics 5:529;A27*

22 **Hanna MG**, Zuberi SM, Eunson L, De Silva R, Tolmie J, McWilliam RC, Stephenson JPB, Wood NW (1998). A new mutation in the human voltage gated potassium channel (kv1.1) associates with episodic ataxia type I with epilepsy. *Platform presentation ENS Nice 1998 Journal of Neurology 245:6/7:349*

23 **Hanna MG**, Eunson L, Panayiotopoulos CP, Youroukos S, Wood NW (1998). The first stop codon point mutation in the human voltage-gated potassium channel KV1.1 associates with drug resistant episodic ataxia type I. *Poster presentation ENS 1998 Journal of Neurology245:6/7:408*

24 **Hanna MG**, Nelson IP, Eunson L, Rose M Rahman S, Schapira ANV, Wood NW,Morgan-Hughes JA (1998). Marked phenotypic heterogeneity associated with the C7472 insertion mutation in the mitochondrial tRNA serine gene the possible role of an intragenic polymorphism *Poster presentation at the ENS Nice 1998 Journal of Neurology 245:6/7:422*

25 Rahman S, Leonard JV, **Hanna MG**, Nelson IP, Lake BD, Schapira AHV (1998) Cytochrome oxidase deficiency associated with a novel heteroplasmic mitochondrial DNA point mutation. *Platform presentation ABN September 1998 Journal of Neurology, Neurosurgery and Psychiatry*

26 Spauschus A, Eunson L, Wood NW, Kullmann DM, **Hanna MG** (1998) Novel mutations in KCNA1 cause deficits in patients with episodic ataxia type I through different mechanisms *Platform presentation American Neuroscience Association, California USA Neuroscience*

27 Pulkes T, Eunson L, Patterson V, Wood NW, Nelson IP, **Hanna MG** (1998). MELAS: A mitochondrial DNA complex I (ND5) gene subunit mutation in three families *Platform presentation, ABN September 1998 JNNP*

28 **Hanna MG**, Nelson IP, Eunson L, Schapira AHV, Cooper JM, Wood NW and Morgan-Hughes JA (1998). The role of an intragenic polymorphism ion the

expression of disease associated with the mtDNA serine (UCN) insertion. *Oral presentation Oxford annual Muscle meeting 1998.*

29 Rahman S, **Hanna MG**, Schapira AHV, Leonard JV (1998). A new COX II mutation associates with childhood benign COX deficiency *IX International Congress on Neuromuscular Disorders, Adelaide Australia*, *Muscle and Nerve supplement 7 p6w-21*

1999

30 Pulkes T, Suddiqui A, Nelson IP, Wood NW, Morgan-Hughes JA, **Hanna MG** (1999) Mitochondrial complex I deficiency associated with a novel mtDNA mutation *Poster presentation, ABN March 1999 I JNNP 34:324*

31 Eunson LH, Jouvenceau A, Ramesh V, Zuberi SM, Nairne A, Hyman N, Wood NW, Spauschus A, Kullmann DM and **Hanna MG** (1999). New mutations in the voltage-gated calcium channel gene CACNA1A in British Families with Episodic Ataxia type 2. *Poster Presentation, Annual meeting of the American Society of Human Genetics. San Francisco 19-23 October 1999.43;P324*

32 Eunson LH, Davis MB, Wood NW and **Hanna MG** (1999). The National DNA-based Diagnostic Service for Periodic Paralyses. *Poster Presentation, Annual meeting of The British Society of Human Genetics. York 27-29 September 1999.*

2000

33 Rea R, Spauschus A, Eunson LH, **Hanna MG**, Kullmann DM (2000). C-terminal truncation of the hKv1.1 subunit in episodic ataxia type 1 affects channel assembly and tracking. *Society for Neuroscience abstracts, New Orleans:614.9*

34 Rea R, Spauschus A, Eunson LH, **Hanna MG**, Kullmann DM (2000). Functional analysis of interactions between wild-type and mutant subunits in hKv1.1 in episodic ataxia type 1. *Journal of Physiology 525.P:76P*

35 Pulkes T, Andreu AL, Siddiqui A, Morgan-Hughes JA, DiMauro S, **Hanna MG** (2000) Exercise induced myalgia associated with five new somatic mutations in the mitochondrial cytochrome b gene: an important cause of muscle aches, pains and cramps*. Journal of Neurology, Neurosurgery and Psychiatry 2000;68:268*

36 Pulkes T, Wills AJ, Heales S, **Hanna MG** (2000). Exercise intolerance associated with a novel stop codon mutation in the mitochondrial ND2 gene. *Annals of Neurology 2000;48;469*

37 Pulkes T, Sweeney MG, **Hanna MG** (2000). A polymorphism in the mitochondrial tRNA leucine gene influences A3243G-associated phenotypes in humans. *Annals of Neurology 2000;48:438*

2001

38 **Hanna MG**, Jouvenceau A, Eunson LH, Ramesh V, Kullmann DM (2001). Human epilepsy-A possible role of the voltage-gated P/Q-type calcium channels.*Journal of the Neurological Sciences 187:S280*

39 Eunson LH, Davies NP, Davies MB, **Hanna MG** (2001). DNA-based Diagnosis of the Periodic Paralyses in the UK. *Journal of the Neurological Sciences 187:S324*

40 Pulkes T, Morgan-Hughes JA, **Hanna MG** (2001). Mitochondrial progressive external ophthalmoplegia-A new mutation and evidence of further genetic heterogeneity *Journal of the Neurological Sciences 187:S354*

41 Eunson LH, Davies NP, Nairne A, Hymen N, Clifford-Jones RE, Wood NW, **Hanna MG** (2001). New mutations in the voltage gated calcium channel gene CACNA1A in British Families with Episodic ataxia type 2 *Journal of the Neurological Sciences 187:S356*

42 Davies NP, Lee P, Elliot P, Holton J, Landon DN, **Hanna MG** (2001). Phenotypic and Pathological variability in Danon's disease (X-linked vacuolar myopathy, cardiomyopathy and mental retardation) *Journal of the Neurological Sciences 187:S58*

43 Davies NP, Eunson LH, **Hanna MG** (2001). Clinical, Genetic and Molecular Genetic studies of myotonia congenita in the UK *Journal of the Neurological Sciences 187:S506*

44 Eunson LH, Davies NP, Wadia NH, Bhatia KP, **Hanna MG** (2001). A new calcium channel mutation causes late onset progressive ataxia. *Journal of the Neurological Sciences 187:S520*

45 Spauschus A, Rea R, Eunson LH, Wood NW, **Hanna MG**, Kullmann DM (2001). Alterations in mutant potassium channel subunits reflect the clinical phenotype of patients in five families with episodic ataxia type 1. *Journal of the Neurological Sciences 187:S536*

46 Pulkes T, Liolitsa, D, Nelson IP, **Hanna MG** (2001) Mutation analysis of the mtDNA sequence in patients with mitochondrial encephalomyopathies; evidence for nuclear gene involvement. Poster at the 5th European meeting on mitochondrial pathology, Venice, Italy
Published in proceedings

2002

47 Davies NP, Eunson LH, **Hanna MG** (2002) Molecular genetic diagnosis of in myotonia congenital
Journal of Neurology, Neurosurgery and Psychiatry 73;345

48 Davies NP, Imbrici P, Kullmann DM, **Hanna MG** (2002) Potassium channel mutations underlie periodic paralysis with cardiac involvement-Andrsen's Syndrome. *Journal of Neurology, Neurosurgery and Psychiatry 73;346*

49 Rahman S, Liolitsa D, Benton S, Carr L, **Hanna MG** (2002) Two new mitochondrial ND5 mutations associate with MELAS without ragged red fibres: Is ND5 gene a hotspot for MELAS causing mutations. 2002 *J. Inherit. Metab. Dis* 25. 161

2003

50 **Hanna MG** Duchen M (2003) Homoplasmic mtDNA mutations-problems in ascribing pathogenicity Platform at the Oxford annual Muscle and Nerve meeting *Published in proceedings*

2004

51 Graves TD, Bhatia, KP, Imbrici P, Kullmann DM, **Hanna MG** (2004) Neuronal calcium channel dysfunction in late onset progressive ataxia *Journal of Neurology, Neurosurgery and Psychiatry P10; 74:344*

52 Mckenzie M, Liolitsa D, Duchen M, **Hanna MG** (2004) Mitochondrial ATP consumption -A new mechanism in mitochondrial disease? Platform presentation at 6th European meeting on human mitochondrial pathology
Published in proceedings

2005

53 Everitt C, Graves TD, Isenberg D, **Hanna MG** (2005) CNS lupus without systemic disease activity *Journal of the Neurological Science 2l L23*

54 Liolitsa D, McKenzie, Hargreaves I, Sisodiya S, Wood NW, Duchen, **Hanna MG**. MELAS, a young-onset stroke disorder associated with two homoplasmic ND5 mitochondrial DNA mutations. *Journal of Neurology 34;s34*

55 Eunson L. H., Graves T. D. and **Hanna M. G** (2005) New calcium channel mutations predict aberrant RNA splicing in episodic ataxia. *Neurology*.;65(2):308-10

2006

56. Nirmalananthan N, Liolitsa D, Woodward C, Groves M, Reilly MM, Coker R, **Hanna MG.** (2006) A novel phenotype associated with deficiency of mitochondrial complex IV activity. Fall Meeting of the Association of British Neurologists, London, ENGLAND, 04 Oct 2006 - 06 Oct 2006. *JNNP* 78: 211-211.

57. Venance S. L., Cannon S. C., Fialho D., Fontaine B., **Hanna M. G.,** Ptacek L. J., Tristani-Firouzi M., Tawil R. and Griggs R. C. (2006) The primary periodic paralyses: diagnosis, pathogenesis and treatment. *Brain*;129(Pt 1):8-17

2007

58 Nirmalananthan N, Dick JRT, Thomas PS, La Spada AR, Greensmith L, **Hanna MG.** (2007) Physiological analysis of the phenotype of a YAC transgenic mouse model of Kennedy's disease. Fall Meeting of the Association-of-British-Neurologists, London, ENGLAND, 04 Oct 2006 - 06 Oct 2006. *JOURNAL OF NEUROLOGY NEUROSURGERY AND PSYCHIATRY*, 78: 211-211

59 Graves TD, Schorge S, Morris H, Kullmann DM, **Hanna MG**. (2007) A novel voltage-sensing mutation in Kv1.1 is associated with episodic ataxia type 1. 17th Meeting of the European-Neurological-Society, Rhodes, GREECE, 16 Jun 2007 - 20 Jun 2007. *JOURNAL OF NEUROLOGY.* 254: 28-29.

60 Chinnery PF, Hudson G, Stewart J, Craig K, Taylor RW, Turnbull DM, Ramesh V, McFarland R, Burn DJ, **Hanna MG**, et al. (2007) When and how should neurologists test for mutations in POLG? Spring Scientific Meeting of the Association-of-British-Neurologists, Homerton Coll, Cambridge, ENGLAND, 11 Apr 2007 - 13 Apr 2007. *JNNP*. 78: 1014-1014

61 Jen JC, Graves TD, Hess EJ, **Hanna MG**, Griggs RC, Barohn RW, CINCH investigators. (2007) Primary episodic ataxias: diagnosis, pathogenesis and treatment. International Conference on Episodic Ataxia Syndromes, Santa Monica, CA, 05 Feb 2006 - 06 Feb 2006. *Brain*. 130: 2484-2493

62 Ryan, E. Matthews and **Hanna MG.** (2007) Skeletal-muscle channelopathies: periodic paralysis and nondystrophic myotonias. *Curr Opin Neurol*.;20(5):558-63

2008

63 Graves TD, Morris H, Zuberi S, **Hanna MG**, Kullmann DM, Schorge S. (2008) Episodic ataxia type 1 in twins: Genotype-phenotype correlation. Annual Meeting of the Association-of-British-Neurologists, London, ENGLAND, 14 Nov 2007 - 16 Nov 2007. *JNNP*. 79: 344-344

64 Graves TD, Zuberi SM, Morris H, Schorge S, Kullmann DM, **Hanna MG**. (2008) Episodic ataxia type 1 in identical twins: genotype-phenotype correlation. 18th Meeting of the European-Neurological-Society, Nice, FRANCE, 07 Jun 2008 - 11 Jun 2008. *Journal of Neurology* 255: 16-16

65 Duncan AJ, Sweeney MG, Stern E, Taylor RW, Woodward C, Davis MB, **Hanna MG**, Rahman S. (2008) Identification of novel and recurrent mitochondrial DNA mutations in paediatric onset mitochondrial disease using the mitochip resequencing array. *J. Inherited Metabolic Disorders*. 31: 61-61.

66 Taylor R, Fratter C, Sweeney MG, Poulton J, Brown GK, Rahman S, McFarland R, Seller A, Davis MB, **Hanna MG**, et al. (2008) The NCG Service for Rare Mitochondrial Disorders of Adults and Children: a clinical and molecular genetics perspective. British Human Genetics Conference, Univ York, York, ENGLAND, 15 Sep 2008 - 17 Sep 2008. *Journal of Medical Genetics*. 45: S83-S83

67 Woodward C, Sweeney MG, Duncan AJ, Stern E, Al-Dosary M, Taylor RW, **Hanna MG**, Davis MB, Rahman S. (2008) Comparative human mitochondrial genome analysis using the Affymetrix MitoChip and conventional cycle sequencing. British Human Genetics Conference, Univ York, York, ENGLAND, 15 Sep 2008 - 17 Sep 2008. *Journal of Medical Genetics*. 45: S78-S78.

68 Matthews E, Parton M, Humbel RL, Elliot P, Parry C, Holton J, **Hanna MG.** (2008) Severe necrotising myopathy and cardiomyopathy with anti-signal recognition peptide antibodies. 13th International Congress of the World-Muscle-Society, Newcastle upon Tyne, ENGLAND, 29 Sep 2008 - 02 Oct 2008. *Neuromuscular Disorders*.18: 771-772.

69 Duncan AJ, Sweeney MG, Stern E, Taylor R, Woodward C, Davis MB, **Hanna MG**, Rahman S. (2008) Comparative human mitochondrial genome analysis using the affymetrix Mitochip v2 and conventional cycle sequencing. 13th International Congress of the World-Muscle-Society, Newcastle upon Tyne, ENGLAND, 29 Sep 2008 - 02 Oct 2008. *Neuromuscular Disorders*. 18: 755-756

70 Rajakulendran S, Farmer S, Yousry T, Ashton E, Abbs S, Holton J, **Hanna MG**, Matthews E. (2008) Marked hemi-atrophy in a manifesting carrier of Duchenne muscular dystrophy - Possible role of skewed X-inactivation 13th International Congress of the World-Muscle-Society, Newcastle upon Tyne, ENGLAND, 29 Sep 2008 - 02 Oct 2008. *Neuromuscular Disorders*. 18: 804-804

71 Nirmalananthan N, Dick JRT, La Spada AR, Greensmith L, **Hanna MG**. (2008) A co-inducer of the heat shock response ameliorates disease in a mouse model of SBMA.13th International Congress of the World-Muscle-Society, Newcastle upon Tyne, ENGLAND, 29 Sep 2008 - 02 Oct 2008. *Neuromuscular Disorders.* 18: 764-764

72 Graves TD, Zuberi SM, Morris H, Schorge S, Kullmann DM, **Hanna MG**. (2008) Episodic ataxia type 1 in identical twins. MG 13th International Congress of the World-Muscle-Society, Newcastle upon Tyne, ENGLAND, 29 Sep 2008 - 02 Oct 2008. *NeuromuscularDisorders.* 18: 746-746

73 Fialho D, Kullmann DM, **Hanna MG**, Schorge S. (2008) Non-genomic effects of sex hormones on CIC-1 may contribute to gender differences in myotonia congenital. 13th International Congress of the World-Muscle-Society, Newcastle upon Tyne, ENGLAND, 29 Sep 2008 - 02 Oct 2008. *Neuromuscular Disorders.*18: 745-745

74 Graves T. D. and **Hanna MG**. Episodic ataxia: SLC1A3 and CACNB4 do not explain the apparent genetic heterogeneity. *J Neurol*;255(7):1097-9 2008

2009

75 Tomlinson SE, Burke D, Howells J, Trevillion L, **Hanna MG**, Koltzenburg M, Bostock H (2009) Spring Scientific Meeting of the Association-of-British-Neurologists, Dublin, IRELAND, 26 Mar 2008 - 28 Jun 2008. Notch appearance in nerve excitability studies: identification and importance. *JNNP*. 80: 121-121

76 Tomlinson SE, Burke D, **Hanna MG**, Koltzenburg M, Bostock H. (2009) In vivo assessment of HCN channel function (IH) in human motor axons. Spring Scientific Meeting of the Association-of-British-Neurologists, Dublin, IRELAND, 26 Mar 2008 - 28 Jun 2008. *JNNP*. 80: 117-118

77 Tomlinson SE, Burke D, **Hanna MG**, Bostock H, Koltzenburg M. (2009) How reproducible are multiple nerve excitability parameters over time? Spring Scientific Meeting of the Association-of-British-Neurologists, Dublin, IRELAND, 26 Mar 2008 - 28 Jun 2008. *JNNP*. 80: 121-121

78 Tomlinson SE, **Hanna MG**, Holton JL, Rahman S (2009) A novel POLG1 mutation resulting in severe cachexia and muscle wasting 110th Annual Meeting of the British-Neuropathological-Society, Inst Child Hlth, London, England, 07 Jan 2009 - 09 Jan 2009. *Neuropathology and Applied Neurobiology.* 35: 16-16

79 Tomlinson SE, Tan SV, Kullmann DM, Burke D, **Hanna MG**, Bostock H. (2009) Axonal excitability changes in genetic neuronal ION channel disorders. Annual Meeting of the Peripheral-Nerve-Society, Wurzburg, Germany, 04 Jul 2009 - 08 Jul 2009. Journal of the Peripheral Nervous System.14: 144-145

80 Sweeney M, Woodward CE, Mudanohwo EE, Rahman S, **Hanna MG**, Davis MB. (2009) Urine epithelial cells as an alternative to muscle biopsies in the detection of mitochondrial DNA mutations. British Human Genetics Conference, Univ Warwick, York, England, 31 Aug 2009 - 02 Sep 2009. *Journal of Medical Genetics*. 46: S69-S69

81 Fratter C, Gorman G, Stewart JD, Buddles M, Smith C, Evans J, Seller A, Poulton J, Roberts M, **Hanna MG**, et al. (2009) Autosomal dominant Progressive External Ophthalmoplegia (adPEO) due to mutations in the PEO1 gene: A clinical, histochemical and molecular survey of 33 patients. 14th International Congress of the World-Muscle-Society, Geneva, Switzerland, 09 Sep 2009 - 12 Sep 2009. *Neuromuscular Disorders.*19: 562-562

82 Labrum R, Rajakulendran S, Sweeney MG, Bevan R, **Hanna MG**, Davis MB. (2009) Screening for mutations causing episodic ataxia type 1 (EA1) and 2 (EA2). British Human Genetics Conference, Univ Warwiphck, York, ENGLAND, 31 Aug 2009 - 02 Sep 2009. *Journal of Medical Genetics*. 46: S71-S71

83 Sewry CA, Holton J, Dick DJ, Jacques T, Muntoni F, **Hanna M**. (2009) Zebra body myopathy resolved. *Neuromuscular Disorders*. 19: 637-638

84 Rajakulendran S, Graves T, Kullmann D, Schorge S, **Hanna M**. (2009) Variation in CACNA1A associated with episodic ataxia and epilepsy. 13th Congress of the

European-Federation-of-Neurological-Societies, Florence, ITALY, 12 Sep 2009 - 15 Sep 2009. *European Journal of Neurology*. 16: 46-46.

85 Rajakulendran, S. Tan V., Matthews E., Tomlinson S. E., Labrum R., Sud R., Kullmann D. M., Schorge S. and **Hanna MG**. (2009) A patient with episodic ataxia and paramyotonia congenita due to mutations in KCNA1 and SCN4A. *Neurology*;73(12):993-5

86 Venance L, LaDonna KA, Matthews E, Wang Y, Bundy B and Barohn RJ. Nondystrophic myotonias: measuring quality of life in a longitudinal natural history study. *Neurology*;72(Suppl 3):A468 2009

2010

87 **Hanna M** & Barohn R, Nondystrophic Myotonias, Podcast 054, *American Association of Neuromuscular and Electrodiagnostic Medicine.*

88 **Hanna M.** Skeletal Muscle Channelopathies. XII International Congress on Neuromuscular Diseases, 17th-22nd July 2010, Naples, Italy

89 Portaro S, Matthews E, Tan SV, Sud R, Davis MB, **Hanna MG.** A new mutation in SCN4A drastically alters phenotype. XII International Congress on Neuromuscular Diseases, 17th-22nd July 2010, Naples, Italy

90 Pitceathly DS, Holton J, Woodward C, Sweeney MG, Fratter C, Rahman S, **Hanna MG**. Novel POLG mutation causing distal myopathy and cachexia. XII International Congress on Neuromuscular Diseases, 17th-22nd July 2010, Naples, Italy

91 Barohn R, Statland J, Want Y, Richesson R, Bundy B, Herbelin L, Gomes J, Trivedi J, Venance S, Amato A, **Hanna MG**, Griggs R, CINCH Consortium. Interactive voice response diary: a patient-reported outcome measure in non-dystrophic myotonias. XII International Congress on Neuromuscular Diseases, 17th-22nd July 2010, Naples, Italy

92 Morrow JM, Sinclair CDJ, Reilly MM, **Hanna MG**, Yousry TA, Golay X, Thornton JS Reproducibility and reliability of quantitative MRI measures for neuromuscular diseases. XII International Congress on Neuromuscular Diseases, 17th-22nd July 2010, Naples, Italy

93 Matthews E, Sud R, Labrum R, Strycharczuk L, Sinclair CDJ, Yousry TA, **Hanna MG**. Using MRI as a diagnostic tool in the skeletal muscle channelopathies. United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, ENGLAND, 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders .*20: S29-S30. Mar 2010

94 Sinclair CDJ, Miranda MA, Cowley P, Morrow JM, Davagnanam I, Mehta H, **Hanna MG**, Koltzenburg M, Reilly MM, Yousry TA, et al. Magnetic resonance imaging and sciatic nerve cross-sectional area in inherited and inflammatory neuropathies. United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, ENGLAND, 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders*. 20: S29-S29

95 Sinclair CDJ, Morrow JM, Yousry TA, Reilly MM, **Hanna MG**, Golay X, Thornton JS. Inter-scan reproducibility of quantitative neuromuscular MRI. United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, ENGLAND, 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders.* 20: S28-S28

96 Pitceathly RDS, Holton JL, Miller JAL, Quinlivan R, **Hanna MG.** The expanding histopathological profile of the myofibrillar myopathies. United Kingdom

Neuromuscular Translational Research Conference 2010, Oxford, England, 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders.* 20: S28-S28

97 Parton M, **Hanna MG**, Miller AD, Morrow JM. IBM-Net: a clinical database of inclusion body myositis patients United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, England, 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders.* 20: S27-S27

98 Ahmed M, Miller AD, **Hanna MG**, Greensmith L United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, England, 25 Mar 2010 - 26 Mar 2010. Heat shock protein induction as a therapeutic strategy for inclusion body myositis. *Neuromuscular Disorders.*20: S27-S27

99 Miller AD, Ahmed M, **Hanna MG**, Greensmith L. Augmentation of the heat shock response in an in vitro model of sporadic inclusion body myositis. United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, England 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders.*20: S26-S27

100 Pitceathly RDS, Woodward CE, Mudanohwo EE, Sweeney MG, Davis MB, **Hanna MG**, Rahman S. Non-invasive diagnosis of single deletion disorders in children with suspected mitochondrial disease United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, England, 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders.* 20: S24-S24

101 Pitceathly RDS, Nesbitt V, Rahman S, McFarland R, **Hanna MG**, Turnbull DM. MRC mitochondrial cohort study: development of a UK database. United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, ENGLAND, 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders.* 20: S23-S23

102 Dewar L, Matthews E, Bahlke G, **Hanna MG.** Quantification of grip myotonia using a novel accelerometer device: a pilot study. United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, ENGLAND, 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders.* 20: S17-S17

103 Rayan DR, Matthews E, Barreto G, Tan SV, Dewar L, Burge J, **Hanna MG.** Double-blind placebo controlled cross-over study to investigate the efficacy of mexiletine in patients with non-dystrophic myotonia in the UK. United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, England, 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders.* 20: S17-S17

104 Rajakulendran S, Matthews E, Tan SV, Dewar L, Griggs RC, **Hanna MG**, CINCH Group. The genetic skeletal muscle channelopathies: genotype-phenotype correlation and longitudinal studies. United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, ENGLAND, 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders.* 20: S16-S17

105 Portaro S, Matthews E, Sud R, Davis MB, **Hanna MG**. Acetazolamide response in patients affected by hypokalemic periodic paralysis. United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, ENGLAND, 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders.* 20: S16-S16

106 Rajakulendran S, Parton M, Holton JL, **Hanna MG**. Clinical and pathological heterogeneity in partial merosin deficiency. United Kingdom Neuromuscular Translational Research Conference 2010, Oxford, ENGLAND, 25 Mar 2010 - 26 Mar 2010. *Neuromuscular Disorders.*20: S11-S11

107 Pitceathly RDS, Holton JL, Hargreaves I, Heales S, Woodward C, Sweeney MG, Davis MB, Evans J, Smith C, Fratter C, Rahman S, **Hanna MG**. Novel POLG mutation causing distal myopathy and cachexia. British Human Genetics

Conference, Warwick, RI, 06 Sep 2010 - 08 Sep 2010. *Journal of Medical Genetics.* 47: S54-S54

108 Willis TA, Hollingsworth KG, Sveen ML, Morrow J, Vandenheede J, Mayhew A, Eagle M, Bushby K, Lochmuller H, **Hanna M**, et al. Assessing muscle pathology by MRI in LGMD2I. 15th International Congress of the World-Muscle-Society, Kumamoto, Japan, 12 Oct 2010 - 16 Oct 2010. *Neuromuscular Disorders*. 20: 666-666

109 Graves T, Phadke R, Holton JL, **Hanna MG**, Rahman S, Bhardwaj N. (2010) PONM21 Electron microscopy does not add to the diagnostic accuracy of muscle biopsy for suspected mitochondrial disease. *J.Neurol.Neurosurg.Psychiatry*. 81: e65

110 Pitceathly R, Rahman S, Maritz C, **Hanna MG**, Lachmann R, Murphy E (2010) England. PONM13 Fatty acid oxidation disorders in adults: a potentially treatable cause of muscle disease. *J Neurol Neurosurg Psychiatry*. 81: e63

111 Rajakulendran S, Graves TD, Labrum RW, Kotzadimitriou D, Eunson L, Davis MB, Davies R, Wood NW, Kullmann DM, **Hanna MG**. and Schorge S. (2010) Genetic and functional characterisation of the P/Q calcium channel in episodic ataxia with epilepsy. *J Physiol*;588(Pt 11):1905-13

112 Ryan DP, da Silva MR, Soong TW, Fontaine B, Donaldson MR, Kung AW., Jongjaroenprasert W, Liang MC, DH, Cheah JS, Ho SC, Bernstein HS, Maciel RM, Brown RH, Jr.and Ptacek LJ. (2010) Mutations in potassium channel Kir2.6 cause susceptibility to thyrotoxic hypokalemic periodic paralysis. *Cell*;140(1):88-98

113 Tomlinson SE, Tan SV, Kullmann DM, Griggs RC, Burke D, **Hanna MG**. and Bostock H.. (2010) Nerve excitability studies characterize Kv1.1 fast potassium channel dysfunction in patients with episodic ataxia type 1. *Brain*; 133(Pt 12):3530-40

2011

114 Nesbitt V, Pitceathly RDS, Rahman S, Poulton J, Turnbull DM, **Hanna MG**, McFarland R. The MRC Centre for Translational Research in Neuromuscular Disease: Mitochondrial Disease Patient Cohort Study UK United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S29-S30

115 Miller A, Machado P, Morrow JM, Hiscock A, Dewar L, Brady S, Hilton-Jones D, **Hanna MG**, Parton M. The natural history of sporadic inclusion body myositis: development of an electronic database IBMnet. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders* 21: S29-S29

116 Muller T, Cirak S, Parton M, Lunn M, **Hanna MG**, Muntoni F. An integrative database for clinical and research studies in neuromuscular diseases. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S29-S29.

117 Sinclair CDJ, Morrow JM, **Hanna MG**, Reilly MM, Yousry TA, Golay X, Thornton JS. Improved magnetization transfer MRI of skeletal muscle in myopathy and neuropathy. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S29-S29

118 Sinclair CDJ, Morrow JM, Fischmann A, **Hanna MG**, Reilly MM, Yousry TA, Golay X, Thornton JS. MRI shows increased tibial nerve size in CMT1A. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S28-S28.

119 Miller A, Ahmed M, **Hanna MG**, Greensmith L. The effects of arimoclomol on pathological outcome measures of inclusion body myositis in vitro.United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S27-S27.

120 Machado P, Miller A, Parton M, Dewar L, Holton JL, Dimachkie M, Herbelir L, Greensmith L, Barohn R, **Hanna MG**. A randomised, double-blinded, placebo-controlled pilot study assessing the safety and tolerability of Arimoclomol in sporadic inclusion body myositis (IBM). United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders.* 21: S27-S27

121 Brady S, Squier W, **Hanna MG**, Hilton-Jones D, Sewry C, Holton JL. Inclusion body myositis: a diagnostic challenge. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S27-S27

122 Ahmed M, Miller A, **Hanna MG**, Greensmith L. Investigating the effects of pharmacological up-regulation of the heat shock response on protein degradation pathways in an in-vitro model of sporadic inclusion body myositis. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S26-S27

123 Scoto M, Cirak S, Mein R, Feng L, Manzur AY, Robb S, Childs AM, Quinlivan RM, Roper H, Hilton-Jones D, Longman C, Chow G, Pane M, Main M, **Hanna MG**, Bushby K, Sewry C, Abbs S, Mercuri E, Muntoni F. SEPN1 related myopathies: Clinical course in a large cohort of patients. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders.* 21: S26-S26

124 Pitceathly RDS, He J, Foley AR, Muntoni F, Pearson N, **Hanna MG**. Whole genome analysis in a family with dominant muscle disease. United Kingdom Neuromuscular Translational Research Conference 2011, London, ENGLAND, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S25-S25

125 Fratter C, Raman P, Alston C, Blakely EL, Craig K, Smith C, Evans J, Seller A, Czermin B, **Hanna MG**, et al. Dominant and recessive RRM2B mutations cause familial PEO and multiple mtDNA deletions in muscle. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S23-S23

126 Nesbitt V, Pitceathly RDS, Rahman S, Hanna MG, McFarland R, Turnbull DM. A3243G-more than just MELAS! United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S21-S21

127 Spillane JE, **Hanna MG**, Kullmann DM. Synaptic mechanisms in P/Qdeficient neuromuscular Junctions. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders.* 21: S15-S15

128 Rayan DLR, Haworth A, Sud R, Burge J, Portaro S, Toscano A, **Hanna MG**. Large scale chloride channel gene DNA rearrangements are an important cause of

recessive myotonia congenita - implications for diagnostic screening. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S14-S15

129 Rayan DLR, Rajakulendran S, Barreto G, Tan SV, Dewar L, Griggs RC, **Hanna MG**, CINCH Grp. Genotype-phenotype correlation and longitudinal study of Andersen-Tawil Syndrome in the UK. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S14-S14

130 Rayan DLR, Matthews E, Barreto G, Tan SV, Dewar L, Burge J, Barohn R, **Hanna MG**, CINCH Grp. Assessing the efficacy of Mexiletine in UK patients with non-dystrophic myotonia. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders* 21: S14-S14

131 Rayan DLR, Matthews E, Rajakulendran S, Barreto G, Tan SV, Dewar L, Burge J, Griggs RC, Barohn R, **Hanna MG**, et al. Genotype-phenotype correlation and longitudinal three year natural history study in the non-dystrophic myotonias in the UK. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S14-S14

132 Burge J, Horga A, Griggs RC, **Hanna MG**, HYP HOP Investigators. Double-blind, placebo-controlled, parallel group, phase Ill study comparing dichlorphenamide vs. placebo for the treatment of periodic paralysis (HYP HOP trial). United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders* 21: S13-S13

133 Durran S, Matthews E, Rayan DLR, Sud R, Polke J, Haworth A, Holton JL, Sweeney MG, **Hanna MG**. Genetic heterogeneity and mechanisms of phenotypic variability in human skeletal muscle channelopathies - a new S4 mutation not associated with HypoPP. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders* 21: S13-S13

134 Gray A, Malik B, Montague K, Dick J, **Hanna MG**, Greensmith L. Investigating pathophysiology and therapeutic strategies in a mouse model of spinal and bulbar muscular atrophy (SBMA). United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders* 21: S10-S10

135 Morrow JM, Matthews E, Rayan DLR, Amer I, Fischmann A, Sinclair CDJ, Thornton JS, Reilly MM, Yousry TA, **Hanna MG**. Magnetic resonance imaging in the non-dystrophic myotonias. United Kingdom Neuromuscular Translational Research Conference 2011, London, ENGLAND, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S5-S5

136 Sinclair CDJ, Morrow JM, Fischmann A, **Hanna MG**, Reilly MM, Yousry TA, Golay X, Thornton JS. Skeletal muscle MRI-determined fat fraction and myometric strength in inclusion body myositis and Charcot-Marie-Tooth disease Type 1A. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders*. 21: S5-S5

137 Willis T, Hollingsworth KG, Sveen ML, Morrow JM, Sinclair CDJ, Thornton JS, Vandenheede J, Strojkovic T, Eagle M, Mayhew A, Bushby K, Lochmuller H, **Hanna MG**, Vissing J, Carliers P, Straub V. Assessing muscle pathology by MRI in

LGMD2I. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders* 21: S4-S5

138 Duchen MR, Heath K, Baruch NB, **Hanna MG**, Muntoni F. Dysregulation of calcium and mitochondrial function as potential therapeutic targets in muscle disease. United Kingdom Neuromuscular Translational Research Conference 2011, London, England, 29 Mar 2011 - 30 Mar 2011. *Neuromuscular Disorders.* 21: S1-S2

139 Mudanohwo EE, Sweeney MG, Woodward CE, Pitceathly RDS, Davis MB, Rahman S, **Hanna NG**. Detection of single large scale rearrangements of the mitochondrial genome in urine epithelial cells: an alternative to muscle biopsy analysis? Zaragoza, Spain Jun 20-23, 2011 *Euromit 8*

147 Ciafaloni E, Rajakulendran S, Sansone V, Trivedi J, Venance S, Tristani-Firouzi M, Bundy B, Hart K, Meola G, Griggs R, **Hanna MG**, CINCH. Preliminary results of Andersen-Tawil syndrome genotype-Phenotype longitudinal study from the consortium for clinical investigation of neurologic channelopathies (CINCH), World Congress of Neurology, Marrakesh, Morocco, November 12-17, 2011

148 Morrow JM, Dewar EL, Ramdharry GM, Laura M, Yousry TA, **Hanna MG**, Reilly MM. Isometric and isokinetic lower limb strength in Charcot-Marie-Tooth disease 1A. Peripheral Nerve Society, Biennial Meeting of the Peripheral Nerve Society, Maryland, Washington, USA June 25-29, 2011

149 Morrow JM, Sinclair CDJ, **Hanna MG**. MRI quantification of lower limb muscle fatty atrophy: a potential outcome measure in chronic neuromuscular disease. Association of British Neurologists annual meeting, Newcastle upon Tyne, October 2011.

150 Machado P, **Hanna MG**. Valosin Containing Protein (VCP) and Myofibrillar Myopathies (MFM) genes' mutations are not associated with sporadic Inclusion Body Myositis (sIBM). Association of British Neurologists annual meeting, Newcastle upon Tyne, October 2011.

151 Machado P, Cortese A, **Hanna MG**. Clinical features and clinical course of sporadic inclusion body myositis (IBM) - a prospective cohort study-IBM-net. Association of British Neurologists annual meeting, Newcastle upon Tyne, October 2011.

152 Horga A, **Hanna MG**. Association of British Neurologists annual meeting, Newcastle upon Tyne, October 2011.

153 Barohn RJ, Wang Y, Fairway B, Bundy L, Herbelin L, Trivdei J, **Hanna MG** et al. Phase II therapeutic trial of mexiletine in non-dystrophic myotonia. Neurology. 2011: 76 (Supplement 4: A645.

154 Statland J, Wang Y, Bundy B, Trivedi J, Herbelin L, Pandya S, Martens WB, Venance S, Amato AA, Hanna MG, Griggs RC, Barohn RJ and CINCH Consortium. A quantitative measure of handgrip myotonia in patients with non-dystrophic myotonia. Neurology. 2011; 76 (Supplement 4):A531.

155 Matthews E, Zanoteli E, Scalco RS, O'Callaghan B, Sud R, McCall S, **Hanna MG**, Castenada MS, Mannikko R, Poulson H. Hypokalaemic periodic paralysis due to a novel ATP1A2 mutation: a new periodic paralysis gene? 10th UK Neuromuscular Translational Research Conference. 23 Mar 2017