



Queen Square Centre for Neuromuscular Diseases

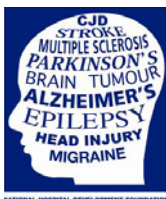
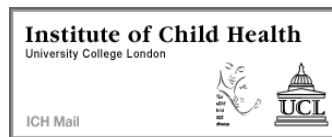


Centre Information

www.cnmd.ac.uk



Queen Square Centre for Neuromuscular Diseases Partners



**NHS National Commissioning Group
For Highly Specialised Services**



MRC

Centre for
Neuromuscular Diseases

Contents

1. Overview and mission of the Centre	4
2. Clinical services in the Centre	7
3. Research and the MRC Centre	19
4. Neuromuscular clinical trials unit	24
5. Current clinical trials	25
6. The Cochrane neuromuscular unit	36
7. Photo gallery	37
8. Who's who in the Centre	41
9. Running the Centre	52

1. The new Centre for Neuromuscular Diseases, Queen Square - overview and mission

The new Centre for Neuromuscular Diseases at the National Hospital for Neurology and Neurosurgery Queen Square, UCLH-Bloomsbury, is a unique development in UK translational medicine for patients with disabling muscle wasting neuromuscular diseases. The brand new centre facilities which officially opened in December 2009 enables full integration of the highest quality NHS clinical care with cutting edge translational research to benefit patients

Nearly 5000 adult patients with muscle wasting neuromuscular diseases are assessed and treated at the National Hospital Queen Square each year. Many neuromuscular diseases develop in childhood and the Centre works closely with the Victor Dubowitz neuromuscular unit at Great Ormond Street to ensure effective transition to adult care. Neuromuscular diseases include genetic and acquired disorders that often cause major disability and premature death. Examples include muscular dystrophies, myositides, mitochondrial diseases, motor neuron diseases, peripheral neuropathies and disorders of the neuromuscular junction such as myasthenia gravis. Although there have been many important genetic and molecular advances in these diseases there are still hardly any effective treatments that benefit patients. The new Centre combines excellent clinical care, with research and clinical trials aimed at developing new treatments for patients.

The Mission of the Queen Centre for Neuromuscular Diseases:

- Deliver the highest quality patient care and outcomes through cohesive iterative multidisciplinary team working
- Translate research findings into clinical trials and new treatments for NHS patients with neuromuscular diseases
- Act as a national and international centre of excellence for clinical/research training and education in neuromuscular diseases

The new Centre for Neuromuscular Diseases, Queen Square

The key components of the centre which will facilitate excellent clinical care alongside cutting edge translational research include:

- Multidisciplinary clinical teams for diagnosis, treatment and care of patients with all genetic and acquired neuromuscular diseases including myositis, muscular dystrophy, mitochondrial diseases, muscle channelopathies, peripheral neuropathies, myasthenia gravis and motor neuron disease.
- Department of Health nationally commissioning group (NCG) neuromuscular services for patients with muscle channelopathies and mitochondrial diseases
- Transition care support in collaboration with GOS Victor Dubowitz unit and centre education/transition pod
- The MRC Centre for translational research in neuromuscular diseases
- The Cochrane neuromuscular unit
- State of the art neuromuscular clinical trials facility

What happens in the Centre for Neuromuscular Diseases?

A fully integrated NHS clinical care and translational research programme is made possible by the co-location of over fifty key staff members including:

- NHS Clinical teams
- MRC Centre research staff
- UCL Institute of Neurology research staff
- Nationally Commissioned NHS teams
- Cochrane unit team
- Active neuromuscular clinical trials unit

2. Clinical Services

Muscle clinical team and services

Muscle service team

Consultants

Professor Michael Hanna

Dr Matt Parton

Dr Chris Turner

Dr Shamima Rahman

Professor Tony Schapira

Dr Janice Holton- muscle pathology

Clinical Nurse Specialist

Catherine Parry

Clinical Support Nurse

Georgie Mewing

Physiotherapist

Liz Dewar

Office Manager & NCG Mitochondrial and Channel Coordinator

Vina Pswarayi

PA to Professor Hanna

Anne Grayson

Secretary to Dr Parton

Marcia Forde

Secretary to Dr Turner

Penny Schafer

Muscle Clinical Research Fellows

Dr Adrian Miller

Dr Emma Matthews

Dr James Burge

Dr Robert Pitceathly

Dr Simona Portaro

Dr Dipa Raja Rayan

Dr Jasper Morrow (joint with nerve)

Typical referral pathway for specialist muscle disease specific clinics

Professor Michael G Hanna, Dr Matt Parton, Dr Shamima Rahman, Dr Chris Turner and Georgie Mewing and Cath Parry Clinical Nurse Specialists

Referrals to the specialist muscle service are accepted from any part of the United Kingdom with either a standard referral letter or completed NCG pro-forma for either of the UK national NCG supported services in Muscle ion channel and Mitochondrial disease



Following receipt of referral, an appointment will be made via the hospitals partial bookings team and a letter confirming the appointment will be sent to the patient



For the first consultation the patient will be seen in our unselected referral Specialist Muscle Clinic (URSMC) for an initial assessment by Prof Hanna, Dr Parton, Dr Turner, Georgie Mewing and Cath Parry (CNS)



The URSMC does provide a “one stop” appointment for clinical assessment neurophysiology, genetic testing, genetic counselling and routine blood tests



If further investigations such as MRI, muscle biopsy and lumbar puncture are required, an admission to our investigation day case unit will be arranged



Follow-up for review of selected investigations, diagnosis and treatment initiation will be in our URSMC



Ongoing care:

1. Follow-up in one of the Specialist Muscle Multidisciplinary Disease Specific Clinics
2. Follow-up in the URSMC
3. Local follow-up



Specialist Muscle Multidisciplinary Disease Specific Clinics

1. National Muscle ion channel clinic supported by NCG
2. National Mitochondrial disease clinic supported by NCG
3. Myotonic Dystrophy clinic
4. Inflammatory disease muscle clinic
5. Inclusion body myopathy clinic
6. Nurse-led immunosuppression treatment clinic

Day case, 5 and 7 day inpatient service

Diagnostic investigations and intravenous treatments for adult muscle patients are provided in the National Hospital Queen Square. This is most commonly provided within the Day Care Unit in the National Hospital for Neurology and Neurosurgery with the patients going home each evening or being put up by the hospital in a local hotel. For those with more severe mobility problems they are admitted to either the five or seven day unit in hospital for this type of treatment.

This specialised muscle service also offers an inpatient service for those patients who require more detailed investigations including muscle biopsies, lumbar puncture and specialised scanning, and more complex treatment. This is particularly used by other neurological centres around the country requiring tertiary or quaternary opinions. As well as offering a muscle biopsy service we also offer a service to give a second opinion on muscle biopsies done elsewhere nationally.

Nationally commissioned neuromuscular services in the Centre for Neuromuscular Diseases

Two nationally commissioned clinical services are provided from the Centre for Neuromuscular Diseases and are lead by Professor Hanna.

The Queen Square National Specialist Service for Mitochondrial Diseases [established in 2007] works closely with centres in Oxford and Newcastle to provide the national comprehensive specialist clinical and diagnostic service for patients with complex mitochondrial diseases. The service includes clinical assessment and comprehensive diagnostic work up including genetics, muscle biopsy and biochemistry.

The Queen Square National Diagnostic service for muscle channelopathies [established in 2001] provides a national clinical and diagnostic service for patients with muscle channelopathies and includes clinical assessment allied to specialist electrophysiology and genetic evaluation. This service is part of the consortium for the diagnosis of rare neuromuscular diseases in collaboration with colleagues in the Dubowitz Neuromuscular Unit GOS, Oxford and Newcastle.

Peripheral Nerve service team and service

Consultants

Dr Mary Reilly

Dr Hadi Manji

Dr Michael Lunn

Clinical Nurse Specialist

Karen Bull

Physiotherapists

Liz Dewar

Alex Pollard

PA to Dr Reilly

Carol Brown

Secretary to Dr Reilly

Michelle Bovell

Secretary to Dr Manji and Dr Lunn

Stephanie Grisdale

Clinical Research Fellows

Elsbeth Hutton

Matilde Laurá

Sinead Murphy

Massimo Russo

Jasper Morrow (joint with muscle)

Peripheral nerve clinical service in the centre for neuromuscular diseases

The Peripheral Nerve Service in the Centre for Neuromuscular Diseases offers a comprehensive service for all peripheral nerve diseases. The Service is led by Dr Mary Reilly and the senior members are two other consultants, Dr Hadi Manji and Dr Mike Lunn, one clinical nurse specialist, Karen Bull and one neuromuscular physiotherapist, Liz Dewar. Dr Ginsberg provides specialist peripheral nerve clinics at the Royal Free Hospital.

Specialist peripheral nerve multidisciplinary clinics available

The Peripheral Nerve Service offers a range of peripheral nerve clinics as listed below:

1. General peripheral nerve clinic
2. Genetic peripheral nerve clinic
3. Inflammatory neuropathy/immunosuppression peripheral nerve clinic
4. HIV neuropathy clinic
5. Nurse-led immunosuppression treatment clinic
6. Nurse-led genetic counselling clinic
7. There are also three nurse-led telephone clinics weekly, dealing with all peripheral nerve problems, but particularly immunosuppression

Mechanism of Referral

Referrals to the specialist peripheral nerve service are accepted from any part of the United Kingdom either as tertiary referrals from other consultants or as second referrals from GPs. Referrals are booked into the appropriate clinic. Most referrals are seen in the general peripheral nerve clinic except for specialised referrals to Dr Reilly's genetic clinic which are directly booked into the genetic clinic. In all peripheral nerve clinics, all new patients are seen by consultants.

All the follow up patients are also seen by consultants but some of these may be seen by Specialist Registrars initially. This is in keeping with our motto of delivering a consultant-led service.

Description of typical clinic attendance for patients

All patients are pre-warned that clinic may take up to half a day. All patients are seen and examined and have either neurophysiology done in the clinic or

go to the neurophysiology department for the neurophysiology to be done. In some of the clinics, including Dr Reilly's Thursday general peripheral nerve and Thursday genetic clinic neurophysiological assessment in clinic is done by Dr Julian Blake who has a particular interest in neuropathies.

During the clinic appointment, patients are offered as is necessary a session with the neuromuscular specialist nurse, Karen Bull, and/or a session with the neuromuscular physiotherapist, Liz Dewar.

At the end of the clinic the patient is again seen by the consultant to go over all of the results from clinic and to plan further investigations or treatments.

Copies of all clinic letters from clinic are sent to the patient.

Day case, 5 and 7 day inpatient service

Many peripheral nerve patients who have an inflammatory neuropathy require frequent infusions of immunomodulatory treatments including intravenous immunoglobulin and Cyclophosphamide. This is most commonly provided within the Day Care Unit in the National Hospital for Neurology and Neurosurgery with the patients going home each evening or being put up by the hospital in a local hotel. For those with more severe mobility problems they are admitted to either the five or seven day unit in hospital for this type of treatment.

This specialised peripheral nerve service also offers an inpatient service for those patients who require more detailed investigations including nerve biopsies, lumbar puncture and specialised scanning, and more complex treatment. This is particularly used by other neurological centres around the country requiring tertiary or quaternary opinions. As well as offering a peripheral nerve biopsy service by which we mean we do the biopsies and read the biopsies we also offer a service to give a second opinion on nerve biopsies done elsewhere nationally and routinely request the nerve biopsies of any patient referred to us for a second opinion who has already had a nerve biopsy so that we can review them ourselves.

Ongoing Care

Following a patient's appointment in clinic or an inpatient episode a decision is made that they will either followed up in one of the specialist peripheral nerve multidisciplinary disease specific clinics or the general peripheral nerve clinic, the nurse led clinics, the nurse led telephone clinics or have local follow up with either the referring physician or general practitioner.

Motor Neuron/Myasthenia clinical services

Consultants

Professor Dimitri Kullmann- myasthenia

Dr Robin Howard-myasthenia and motor neuron disease

Dr Katie Sidle- motor neuron disease

Dr Richard Orrell- motor neuron disease

Dr Nick Hirsch- respiratory support service

Clinical Nurse Specialists

Jan Clarke

Natalie James

Helen Eddlestone

PA to Dr Howard

Sarah Mazdon

Clinical Research Fellow

Dr Jennifer Spillane - John Newsom-Davis Myasthenia Fellow

Motor neuron disease service

The NHNN is the MND Association regional care centre for patients with MND and the clinical team are responsible for all aspects of the care of patients with MND in the weekly MND clinic.

Myasthenia Gravis service

A comprehensive outpatient and inpatient service is provided. This includes access to ventilatory support and state of the art neurological medical intensive care facility at Queen Square.

NHS Clinical activities supported by the Centre

Mondays

- 08:00 – 09:00 Peripheral nerve neuropathology meeting
Dr Reilly/Dr Lunn/ Dr Manji
Seminar Room
- 13:00 – 14:00 Peripheral nerve pre-clinic meeting
Dr Reilly/Karen Bull/Clinical Research Fellows
Seminar Room
- 14:00 – 15:00 NCG genetics/biochemistry meeting
Dr Rahman/Prof Hanna/Dr Heales
Seminar Room
- 14:00 – 17:00 Myasthenia Clinic
Prof Kullmann
Basil Samuel OPD
- 14:00 – 17:00 Immunosuppression and complex peripheral nerve
clinic
Dr Lunn
Basil Samuel OPD

Tuesdays

- 08:00 – 09:00 Neuromuscular journal club
Dr Reilly
Seminar Room
- 09:00 Peripheral nerve general neuropathy clinic
Dr Reilly
Basil Samuel OPD
- 09:45 Neuromuscular Radiology meeting
Prof Hanna
- 10:15 Muscle ward round
Prof Hanna/Dr Parton/Dr Turner

- 11.30 Muscle pathology meeting
Prof Hanna/Dr Parton/Dr Turner/Dr Holton
Seminar room
- 13:30 Muscle clinic and NCG channelopathy clinic
Prof Hanna/Dr Turner/Dr Parton
Catherine Parry/Georgie Mewing/Liz Dewar
33 Queen Square OPD
- 13:30 Peripheral nerve/general neurology clinic
Dr Lunn
Basil Samuel OPD
- 14:00 Respiratory support clinic
Dr Hirsch
Basil Samuel OPD
- PM Peripheral nerve CNS-led telephone clinic
Karen Bull

Alternate weeks

- 14:00 Muscle clinic
Prof Schapira
Basil Samuel OPD

Monthly

- 16:30 MRC Centre invited speaker trainee tutorial
Seminar room
- 17:30 MRC Centre invited seminar
33 Queen Square lecture theatre

Alternate Months

- 14:00 Joint inflammatory muscle disease clinic
Prof Hanna/Prof Schapira/Prof Isenberg
Basil Samuel OPD

Wednesdays

09:00 – 13:00

Teaching ward round
Dr Reilly
Basil Samuel OPD

09:00 – 13:00

Peripheral nerve CNS-led telephone clinic
Karen Bull

Monthly

09:00

IBM clinic
Prof Hanna/Dr Parton
33 Queen Square OPD

Thursdays

09:00

Muscle clinic
Dr Parton
Basil Samuel OPD

09:00

General peripheral nerve/genetic (alternate weeks)
Dr Reilly

09:00

General peripheral nerve clinic
Dr Lunn

09:00

General peripheral nerve clinic
Dr Manji

09:00

Motor neuron disease and myasthenia clinics
Dr Howard/Dr Sidle/Dr Orrell/Natalie James

12.30

Muscle team MDT
Seminar room

14:00

NCG mitochondrial clinic
Prof Hanna/Dr Rahman
RLHH

- 14:00 Myotonic dystrophy clinic
Dr Turner
- 14:00 Peripheral nerve/pain clinic
Prof Koltzenburg
Basil Samuel OPD
- 14:00 Gower's Grand Round
(Neuromuscular round 4 times per year)
Wolfson Lecture Theatre
- 17:00 Peripheral nerve CPC
- Monthly**
PM Transition muscle clinic
Prof Hanna/Prof Muntoni/Dr Manzur/Dr Robb
GOS
- Fridays**
Research and trials
Clinical trials room
- 14:00 Ward round
Dr Reilly
- Monthly**
15:30 Joint ION/GOS neuromuscular MRI meeting
Prof Yousry/Dr Rosendahl
Radiology seminar rooms, 8-11 QS/GOS

3. Research activities in the Centre

The MRC Centre for Neuromuscular diseases is one of the MRC's translational research centres and opened in 2008. It is directed by Professor Hanna and is a partnership between the UCL Institutes of Neurology and Child Health, and the University of Newcastle upon Tyne.

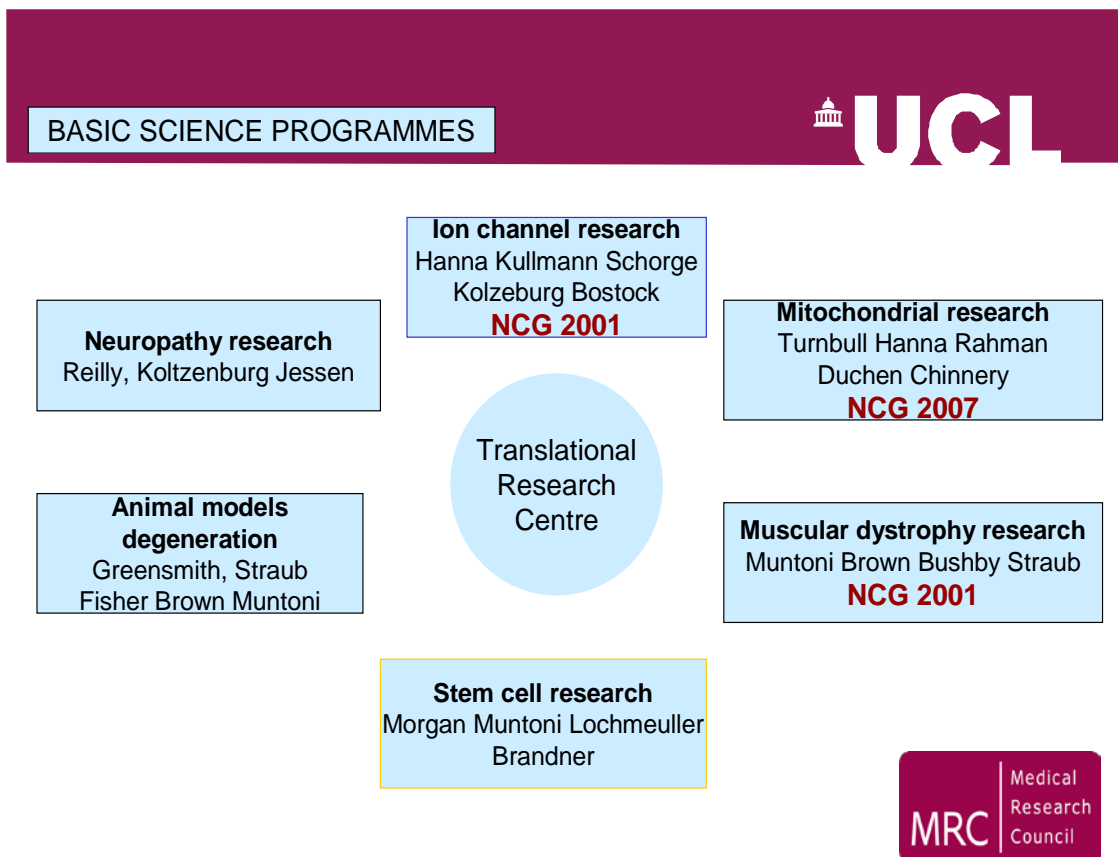
Main programmes of research

The main programmes of research within the centre have built on existing funded themes currently attracting in excess of £20m of grant income across PI groups in UCL and Newcastle, and have developed new cross cutting collaborations which have capitalized on the recruitment of world class senior academic personnel to UCL and Newcastle.

The main programmes of basic research across the MRC Centre cover major diseases of muscle and nerve

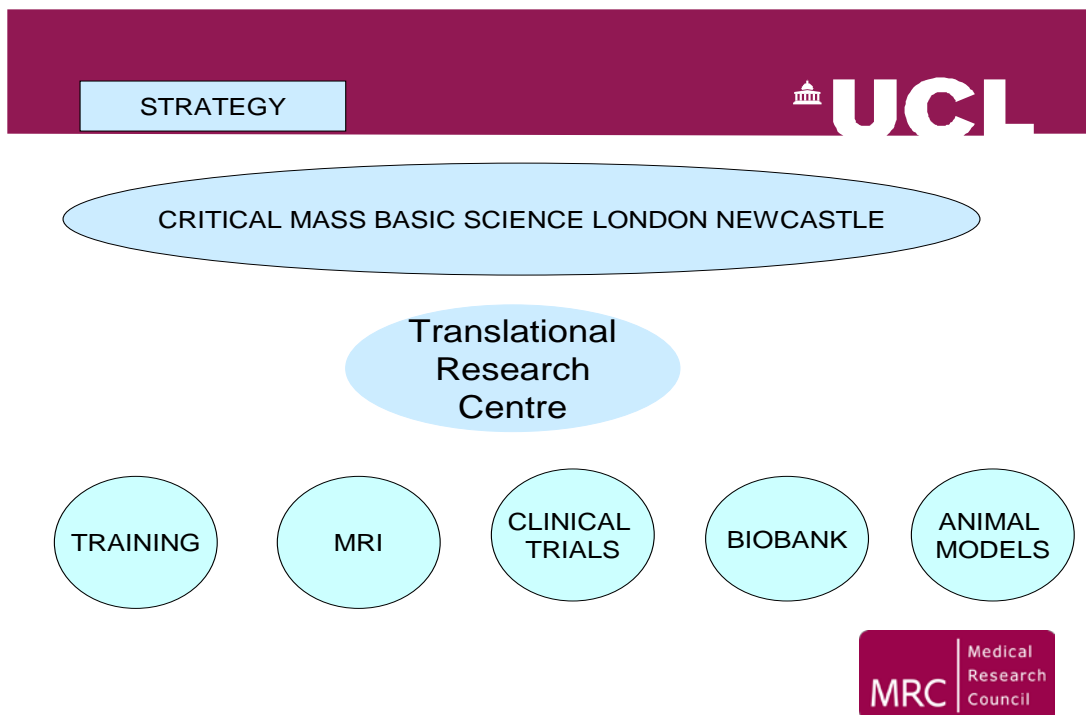
- Molecular mechanisms in muscular dystrophy [Brown, Duchen, Holden, Muntoni, Sewry]
- Mitochondrial DNA neuromuscular disease [Duchen, Hanna]
- Ion channel neuromuscular disease [Hanna, Koltzenburg, Tann, Bostock, Kullmann]
- Muscle stem cell biology [Morgan, Muntoni]
- Genetic neuropathies [Brandner, 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 [Hanna, Koltzenburg, Muntoni, Reilly, Yousry]
- Trials & outcomes in neuromuscular disease [Hanna, Muntoni, Reilly, Thompson]

*Key science programmes and representative investigators.
NCG-National Commissioning Group-refers to NHS funded national diagnostic and advisory services run by MRC Centre PI's for muscle channelopathies (London-Hanna), mitochondrial disease (joint: Newcastle-Turnbull and London-Hanna joint), congenital muscular dystrophies (London- Muntoni) and Limb-girdle dystrophies (Newcastle-Bushby)*



All the main programmes of basic research impact upon and benefit from the following five key areas that have been newly developed in the centre (the underdevelopment of these key areas is a current “block” to effective UK translational research in neuromuscular disease).

- The MRC Centre is initiating and running new clinical trials and is developing a range of specific clinical assessment tools to facilitate future clinical trials in neuromuscular disease in the UK
- The MRC Centre is establishing new cutting edge MRI of nerve and muscle disease in animals and humans
- The MRC Centre is establishing a unique UK biobank of human neuromuscular patients tissues and cells
- The MRC Centre is establishing a network and resource for elucidating the pathogenesis of neuromuscular conditions in mutant mice
- The MRC Centre is attracting and training a new generation of basic and clinical neuromuscular scientists to build future “capacity” in the UK



MRC Centre for translational research in neuromuscular diseases - staff list

Centre Director

Professor Michael G Hanna

Centre Deputy Director London

Professor Martin Koltzenburg

Professor Francesco Muntoni

Centre Deputy Director Newcastle

Professor Katie D Bushby

Centre Steering Committee

Professor Michael G Hanna

Professor Francesco Muntoni

Professor Martin Koltzenburg

Dr Mary Reilly

Professor Dimitri Kullmann

Professor Tarek Yousry

Professor Katie Bushby

Professor Doug Turnbull

Centre Principal Investigators-London Centre Principal Investigators-Newcastle

Professor Sebastian Brandner

Dr Sue Brown

Professor Michael Duchen

Professor Elizabeth Fisher

Professor Linda Greensmith

Professor John Hardy

Professor David Isenberg

Professor Kristjan Jessen

Professor Dimitri Kullmann

Dr Jenny Morgan

Professor Francesco Muntoni

Dr Mary Reilly

Professor Anthony Schapira

Professor Alan Thompson

Professor Nicholas Wood

Professor Tarek Yousry

Professor Patrick Chinnery

Dr Elaine McColl

Professor Hanns Lochmüller

Professor Volker Straub

Professor Douglass Turnbull

Professor Katie Bushby

London Full time staff appointed following commencement of the centre

Non-clinical translational PhD studentships - four years each

Mhoriam Ahmed

Alex Clark

Amy Innes

Phil McGoldrick

Alice Neal

Clinical translational PhD studentships - three years each

Dr Adrian Miller

Dr Jasper Morrow

Centre administrator - five years

Zoë Scott

Clinical trial coordinator - five years

Gisela Barreto

Biobank technician - five years

Diana Johnson

MRI physicist - three years

Dr Chris Sinclair

Newcastle full time staff appointed following commencement of the Centre

Non clinical translational PhD studentships - four years each

Sally Spendiff

Alasdair Wood

Clinical trials coordinator - five years

Geoff Bell

Biobank technician - five years

Mojgan Reza

4. Clinical trials unit Centre for Neuromuscular Diseases

A major aim of the centre is to make clinical trials happen by providing a state of the art trials facility which includes a trial consultation room, neurophysiology equipment and a range of clinical trials, myometry, and exercise equipment including:

- Treadmill with un-weighting system
- Codamotion 3D movement analysis
- Cybex system
- Exercise ergometer
- Elliptical trainer
- Cortex metalyzer metabolic system – to measure aerobic capacity

The equipment in the trials unit has been specifically designed to harmonise with the Newcastle MRC Centre trials gym so that synchronous clinical trials in the North of England (Newcastle) and the South of England (Queen Square) can be undertaken throughout the MRC Centre, thereby enhancing collaboration and recruitment opportunities.

Many trials and interventions are planned or underway including:

- Collation of natural history information re physical disease progression
- Exercise intervention trials in muscle (mito) and nerve (CMT)
- Balance studies
- Gait and orthotic studies
- Fatigue studies
- Exercise physiology studies

Full details of active clinical trials mapping to the MRC Centre are listed in the next chapter.

5. Clinical trials supported by the Centre

Clinical trials linked to the MRC Centre and supported by different funding agencies including the Medical Research Council, Muscular Dystrophy Campaign, UK Department of Health, National Institutes of Health (USA), Food and Drug Administration (USA), AVI Biopharma and PTC Therapeutics.

Open Trials

RANDOMISED DOUBLE-BLIND PLACEBO CONTROLLED TRIAL OF LONG-TERM ASCORBIC ACID TREATMENT IN CHARCOT-MARIE-TOOTH DISEASE TYPE 1A

Status: Follow-up phase. Closed to recruitment

Sponsor: University College London

Funder: Muscular Dystrophy Campaign (MDC)

PI: Dr. Reilly

Charcot-Marie-Tooth disease 1A (CMT1A) is associated with a duplication of the peripheral myelin protein 22 (PMP22) gene. To date there is no pharmacological treatment for CMT1A patients. Treatments and therapy for CMT is restricted to symptomatic treatments such as physiotherapy and surgery for skeletal deformities.

Recently, treatment with ascorbic acid (AA) has been shown to be effective for transgenic mice over-expressing PMP22, a model of the human disease. Treated animals had much less severe neuropathy as compared to untreated controls as shown by clinical and histological findings. Some clinical parameters even improved during treatment.

This is a phase III prospective, multi-centre, randomised, double-blind, placebo-controlled study aiming to evaluate the efficacy of AA treatment in CMT1A.

The study has been running now almost for two years and it is now in the follow-up phase. Fifty participants were enrolled in the UK site at the National Hospital for Neurology and Neurosurgery.

For information about the study please contact Dr. Matilde Laura at m.laura@ion.ucl.ac.uk.

A PHASE IIb EFFICACY AND SAFETY STUDY OF PTC124 IN SUBJECTS WITH NONSENSE MUTATION-MEDIATED DUCHENNE AND BECKER MUSCULAR DYSTROPHY

Status: Closed to recruitment

Sponsor: PTC Therapeutics

Funder: PTC Therapeutics

PIs: Prof. Muntoni, Prof. Bushby

Duchenne muscular dystrophy (DMD) is an X-linked genetic disorder affecting young boys. The condition is disabling and life-threatening. A small subset of boys are classified as having Becker muscular dystrophy (BMD), a phenotypically milder form of the dystrophic muscle disease.

In approximately 10 to 15% of boys with DMD and BMD the causative defect is the presence of a nonsense mutation in the dystrophin gene that truncates dystrophin protein production by introducing a premature stop codon into the dystrophin messenger ribonucleic acid (mRNA).

PTC124 is a novel, orally bioavailable, small-molecule drug that promotes ribosomal read-through of mRNA containing a premature stop codon. Through this mechanism of action, PTC124 has the potential to overcome the genetic defect in boys for whom a nonsense mutation causes DMD/BMD.

In vitro studies in cell lines with dystrophin nonsense mutations have shown that PTC124 can restore production of the missing dystrophin gene.

This is an international, multi-centre, randomised, double-blind, placebo-controlled, dose-ranging, efficacy and safety study.

The study primary aim is to evaluate the effect of PTC124 on ambulation as assessed by the distance walked during a 6-minute walk test (6MWT).

The double-blind arm of the study randomised 174 participants worldwide which are to be followed for a period of 12 months. At the completion of the blinded treatment, all compliant participants will be eligible to receive open-label PTC124 in a separate extension study.

(Ataluren is now the non-proprietary generic name for PTC124).

ANTI SENSE OLIGONUCLEOTIDE INDUCED EXON SKIPPING IN DUCHENNE MUSCULAR DYSTROPHY

This initiative is led by the MDEX consortium (The MDEX consortium led by Professor Muntoni, is a multidisciplinary enterprise to promote translational research into muscular dystrophies, and is formed by the clinical groups of Professor Francesco Muntoni (UCL Institute of Child Health) and Professor Kate Bushby and Professor Volker Straub (Newcastle University), and scientists from Imperial College London (Professor Dominic Wells), UCL Institute of Child Health (Dr Jennifer Morgan), Royal Holloway University of London (Professor George Dickson and Dr Ian Graham), Oxford University (Dr Matthew Wood) and University of Western Australia (Prof Steve Wilton). In addition, the charities Muscular Dystrophy Campaign (MDC), Action Duchenne and Duchenne Family Support Group also participate in the Consortium, www.mdex.org.uk).

The current two trials led by the consortium are mentioned below.

RESTORING DYSTROPHIN EXPRESSION IN DUCHENNE MUSCULAR DYSTROPHY: A PHASE I/II CLINICAL TRIAL USING AVI-4658

Status: completed. Closed to recruitment

Sponsor: Imperial College London

Funder: Department of Health (DoH)

PIs: Prof. Muntoni

The primary scope of the trial is to assess efficacy (dystrophin production) and safety of intramuscular administered morpholino oligomer directed against exon 51 (AVI – 4658 PMO).

Antisense therapy with the use of antisense oligomers has the potential to restore effectively the production of dystrophin, the defective protein, in >70% of DMD. This could result in increased life expectancy through improved muscle survival and function. Recent scientific research has demonstrated the potential of this technique to skip mutated dystrophin exons, restore the reading frame and generate functional dystrophin protein. Having demonstrated proof-of-principle in human cell culture and animal model studies, we now intend to determine efficacy and safety of this approach to induce dystrophin exon skipping in children with DMD. This study is aimed at children with Duchenne muscular dystrophy above the age of 10 years with mutations than can be rescued by the skipping of exon 51 [45-50; 47-50; 48-50; 49-50; 50; 52; 52-63].

DOSE-RANGING STUDY OF AVI-4658 TO INDUCE DYSTROPHIN EXPRESSION IN SELECTED DUCHENNE MUSCULAR DYSTROPHY (DMD) PATIENTS – (Systemic study)

Status: Open to recruitment

Sponsor: AVI Biopharma

Funder: Medical Research Council (MRC) and AVI Biopharma

PIs: Prof. Muntoni

This is a safety study of AVI-4658 (a 30-base phosphorodiamidate Morpholino oligomer [PMO]), to skip exon 51 of the dystrophin gene in relevant subjects with DMD.

This is an open-label, two-centre, dose-ranging comparative clinical study of duration twelve weeks.

The objectives of the study are to assess safety and to select the optimum dose that elicits at least 10% de novo dystrophin-positive fibres and dystrophin in a sentinel muscle group after an intravenous AVI-4658 dosing regimen.

A total of up to 16 subjects (ambulatory paediatric males, aged ≥ 5 and ≤ 15 years of age) will be enrolled in this study, consisting of four treatment cohorts and four subjects per cohort. It is expected that there will be four treatment arms ranging from 0.5 mg/kg to 4 mg/kg. All subjects will receive 12 weekly intravenous infusions of AVI-4658.

Precedent studies have demonstrate that AVI-4658 might have therapeutic relevance in managing DMD for boys whose frame-shifted dystrophin gene lesion could be restored after excision of exon 51 if sufficient drug is translocated into the nucleus of the afflicted muscle cell.

This trial is being conducted in London and Newcastle.

For information on the status of recruitment please contact Gisela Barreto, Trials Coordinator (MRC centre London site) at Gisela.barreto@uclh.nhs.uk or Geoff Bell, Trials Coordinator (MRC centre Newcastle site) at geoff.bell@nuth.nhs.uk.

HYP HOP: DICHLORPHENAMIDE vs. PLACEBO FOR PERIODIC PARALYSIS

Status: Set-up Phase

Sponsor: University College London (UCL)

Funder: National Institutes of Health (NIH - USA)

PI: Prof. Hanna

This is a phase III trial into Periodic Paralysis planned to start in 2009. This proposal involves a multi-centre, double-blind, placebo-controlled parallel group, nine-week studies comparing the effects of dichlorphenamide(DCP) vs placebo in patients with period paralysis (Hyper, Hypokalemic periodic paralysis). The 9-week studies will investigate the prevention of attacks of weakness and it will be followed by 1-year extensions without placebo to compare the long term effects of DCP on the course of the diseases and on inter-attack weakness. Approximately 40 participants will be recruited from the United Kingdom.

For information on the status of recruitment please contact Dr. James Burge at James.burge@uclh.nhs.uk or Gisela Barreto, Trials Coordinator at Gisela.barreto@uclh.nhs.uk.

THERAPEUTIC TRIAL OF MEXILETINE IN NON-DYSTROPHIC MYOTONIA

Full Title: A Phase II Randomised, Double-Blind, Placebo controlled, Cross-Over Study to Investigate the Efficacy of Mexiletine in Patients with Non-Dystrophic Myotonia

Status: REC review is now pending

Sponsor: University College London (UCL)

Planned Start date: June 2009

Funder: Food and Drug Administration (FDA – USA)

PI: Prof. Hanna

The non-dystrophic myotonia (NDM) is a group of rare neuromuscular disorders that causes episodes of muscle stiffness (known as myotonias) and paralysis. Predominantly the muscles of the face, hands and legs are affected. In addition to these episodes a permanent and debilitating muscle weakness can develop. The optimal treatment for these disorders is unknown.

Non-dystrophic myotonias are due to abnormalities of ion channels present in skeletal muscle membranes. There is experimental evidence that drugs like mexiletine which block the abnormal function of these ion channels allow the muscle to perform normally.

The study aims to test the efficacy of mexiletine in the treatment of the non-dystrophic myotonias.

This proposal involves a multi-centre, double-blind, placebo-controlled cross over trial of total duration nine weeks.

Approximately fifteen participants will be enrolled in the UK at the National Hospital for Neurology and Neurosurgery.

For information on the status of recruitment please contact Dr. Emma Matthews at e.matthews@ion.ucl.ac.uk or Gisela Barreto, Trials Coordinator at Gisela.barreto@uclh.nhs.uk

ARIMOCLOMOL FOR SPORADIC INCLUSION BODY MYOSITIS (IBM)

Full Title: A Randomised, Double-blinded, Placebo-controlled Pilot Study Assessing the Safety and Tolerability of Arimoclomol in Adult Patients with Sporadic Inclusion Body Myositis

Status: REC Review is now pending

Sponsor: University College London (UCL)

Planned start date: June 2009

Funder: Medical Research Council (MRC)

PI: Prof. Hanna

Sporadic Inclusion Body Myositis (IBM) is the commonest acquired disease of muscle affecting people aged 50 years and over. This is a progressive and debilitating disease with both muscle weakness and wasting, characteristically of the quadriceps and finger flexors. Over time the condition can lead to severe disability, falls and swallowing impairment. Affected muscle tissue demonstrates inflammation and degeneration.

Arimoclomol is a new compound which acts by enhancing a normal, inbuilt protective cell reaction to stresses. The products of this response are 'Heat Shock Proteins (HSPs) which counteract processes that end up leading to abnormal protein deposition and to damage mediated by inflammation.

This proposal involves a multi-centre, double-blind, placebo-controlled parallel study of total duration twelve weeks.

This study proposal aims to assess the safety and tolerability of Arimocloamol (100 mg TDS) as compared with placebo over 4 months of treatment in patients with IBM.

Recruitment will take place at the National Hospital for Neurology and Neurosurgery and twelve patients will be enrolled.

For information on the status of recruitment please contact Dr. Adrian Miller at a.miller@ion.ucl.ac.uk or Gisela Barreto, Trials Coordinator at Gisela.barreto@uclh.nhs.uk.

TAPP: THERAPEUTIC TRIAL OF POTASSIUM AND ACETAZOLAMIDE IN ANDERSEN-TAWIL SYNDROME

Status: Set-up Phase

Sponsor: University College London (UCL)

Funder: National Institutes of Health (NIH – USA)

PI: Prof Hanna

Andersen-Tawil Syndrome (ATS) is a rare form of periodic paralysis that is associated with serious heart-rhythm abnormalities. ATS is characterized by a triad of episodic muscle weakness, long-QT syndrome with potentially fatal cardiac dysrhythmias and skeletal developmental anomalies. The underlying cause of this potentially fatal condition is only partly understood and there are no established treatments. Mutations in the KCNJ2 gene encoding Kir2.1, an inward-rectifying potassium channel account for approximately 60% of ATS cases (termed ATS1), the remaining 40% are presumed to have an as yet undetermined gene lesion and are designated ATS2. ATS1 and ATS2 are phenotypically indistinguishable.

The treatment of ATS has been largely anecdotal and empirical.

This proposal involves a multi-centre, placebo-controlled 'n of 1' study design of total duration 45 weeks. The expected total enrolment for this multi-centre study is 16 participants.

The aim of this study is to determine whether potassium supplements and/or acetazolamide alter the duration of muscle weakness and potentially life-threatening heart rhythm abnormalities in patients with ATS.

For information on the status of recruitment please contact Dr. James Burge at James.burge@uclh.nhs.uk or Gisela Barreto, Trials Coordinator at Gisela.barreto@uclh.nhs.uk.

Natural History – Longitudinal Studies

NON-DYSTROPHIC MYOTONIAS: GENOTYPE AND PHENOTYPE CORRELATION AND LONGITUDINAL STUDIES

Status: Closed to recruitment

Sponsor: University College London

Funder: National Institutes of Health (NIH – USA)

PI: Prof. Hanna

This multi-centre project involves a prospective, cross-sectional and longitudinal natural history in non-dystrophic myotonias (NDM).

The aim is to collect standardized data from NDM patients, to include clinical symptoms, exam findings, as well as the results of strength, functional, and electrophysiological testing. Genetic testing will permit precise identification of individual NDM subtype. This information will allow for the identification and implementation of appropriate endpoints in studies of potential treatments.

This is a NIH funded study. Twenty patients were enrolled at the National Hospital for Neurology and Neurosurgery.

For more information about the study please contact Dr. Emma Matthews at e.matthews@ion.ucl.ac.uk.

ANDERSEN-TAWIL SYNDROME: GENOTYPE AND PHENOTYPE CORRELATION AND LONGITUDINAL STUDY

Status: Open to recruitment

Sponsor: University College London

Funder: National Institutes of Health (NIH – USA)

PI: Prof. Hanna

Andersen-Tawil syndrome is a neuromuscular disorder caused by a mutation in the KCNJ2 gene which codes for the inwardly rectifying potassium channel Kir2.1. A number of different mutations in this gene have already been identified in affected individuals. This disorder is characterised by the triad of

periodic paralysis, developmental abnormalities and cardiac arrhythmias.

This project is a natural history trial into Andersen-Tawil Syndrome. The aim of the trial is to study the relationship between the genetic abnormalities underlying the disorder and the diverse clinical features.

Eight patients have been enrolled so far at the National Hospital for Neurology and Neurosurgery.

For information on the status of recruitment please contact Dr. Sanjeev Rajakulendran at s.rajakulendran@ion.ucl.ac.uk.

Exercise Studies

STRENGTHENING HIP MUSCLES TO IMPROVE WALKING DISTANCE IN PEOPLE WITH CHARCOT- MARIE-TOOTH DISEASE

Status: REC Approved. Open to recruitment

Sponsor: University College London Hospitals

Funder: Muscular Dystrophy Campaign (MDC)

PI: Dr. Reilly

Charcot-Marie-Tooth (CMT) disease is a form of hereditary peripheral neuropathy.

People with CMT present with weakness, wasting and sensory loss as a result of degeneration of the long peripheral nerves supplying the distal muscles.

The aim of this study will be to investigate the efficacy of a 16 week home based programme of training to increase hip flexor muscle strength and walking endurance. Additional measures of gait speed, exertion, fatigue, disability and general activity will also be recorded. Baseline impairment measures will be obtained to ascertain predictors of strength gains.

This study will use a single blinded, randomised cross over design to investigate if training the hip flexor muscles will strengthen the hip flexor muscle and improve walking endurance in people with all types of CMT.

The trial will include people, aged between 18 and 70 years, who have been diagnosed with CMT on the basis of genetic tests (where possible), family history and neurophysiology testing. Each subject will be involved with the study for a 40 week period.

For information about recruitment contact Alex Pollard, Research Physiotherapist at a.pollard@ion.ucl.ac.uk.

EXERCISE TRAINING IN PATIENTS WITH MITOCHONDRIAL DISEASE: ASSESSING THE BENEFITS

Status: Recruiting

Sponsor: University Newcastle

Funder: Muscular Dystrophy Campaign (MDC)

PI: Prof. Turnbull

Mitochondrial myopathies are a very important group of muscle diseases associated with weakness, pain and fatigue. At present, treatment options are very limited.

Exercise therapy has been found to have some benefit in this group of patients and we wish to explore this further in terms of both strength and endurance.

The aim of this study is to demonstrate that strength exercise training is an effective approach to therapy in certain patients with mitochondrial myopathy, specifically those with sporadic mutations in mitochondrial DNA. Based on our previous research studies, we believe that such training will improve muscle strength, mitochondrial function, exercise tolerance and overall quality of life.

The main objectives will be:

To confirm that endurance training in patients with mitochondrial abnormalities improves quality of life, exercise tolerance and oxidative capacity.

To determine the ability of resistance muscle strength training to improve skeletal muscle strength and oxidative capacity by incorporation of satellite cells into mature myofibres.

Participants are expected to commit to an exercise training and testing over a period of 4 to 8 months.

The study will include patients between the ages of 18 and 65 years who have had a previous muscle biopsy showing a defect in skeletal muscle mitochondrial DNA that is either in the form of a sporadic point mutation or single large-scale deletion. Patients who have this type of mutation and do not have any family members that are affected and have no major cardiac involvement, hypertension, pulmonary or peripheral vascular disease that may complicate findings.

For information about recruitment contact Geoff Bell at geoff.bell@nuth.nhs.uk or Caroline Hodgson at c.hodgson@ncl.ac.uk.

6. The Cochrane Neuromuscular Disease Group

The Cochrane Neuromuscular Disease Group (CNDG) is part of the Cochrane collaboration which produces and disseminates systematic reviews of healthcare interventions, and promotes the search for evidence in the form of clinical trials and other studies of interventions.

Registered with the Cochrane Collaboration in 1998 with its editorial base in King's College London, the Group moved to the MRC Centre in September 2008.

Aims

The Group aims to provide systematic reviews of all interventions for all neuromuscular diseases, including amyotrophic lateral sclerosis/motor neuron disease, peripheral nerve disorders, myasthenia gravis and neuromuscular function disorders, and muscle diseases.

Activity

In 10 years the CNDG has published 73 systematic reviews and 40 protocols (pre-reviews). The conditions most prevalent, lethal or seriously disabling have each been topics for multiple reviews with different sets of interventions. Developments include projects on reviews of Diagnostic Test Accuracy and now Overviews of Reviews which will pull together the multiple interventions for one condition under a single umbrella to increase accessibility to the information for healthcare professionals, policy makers and patients.

Further information about the work of the Group can be found on its website <http://www.neuromuscular.cochrane.org>

CNDG Staff

Professor Richard Hughes

Dr Michael Lunn

Jane Batchelor

Kate Jewitt

Angela Gunn

Rachel Barton

Co-ordinating Editor

Co-ordinating Editor

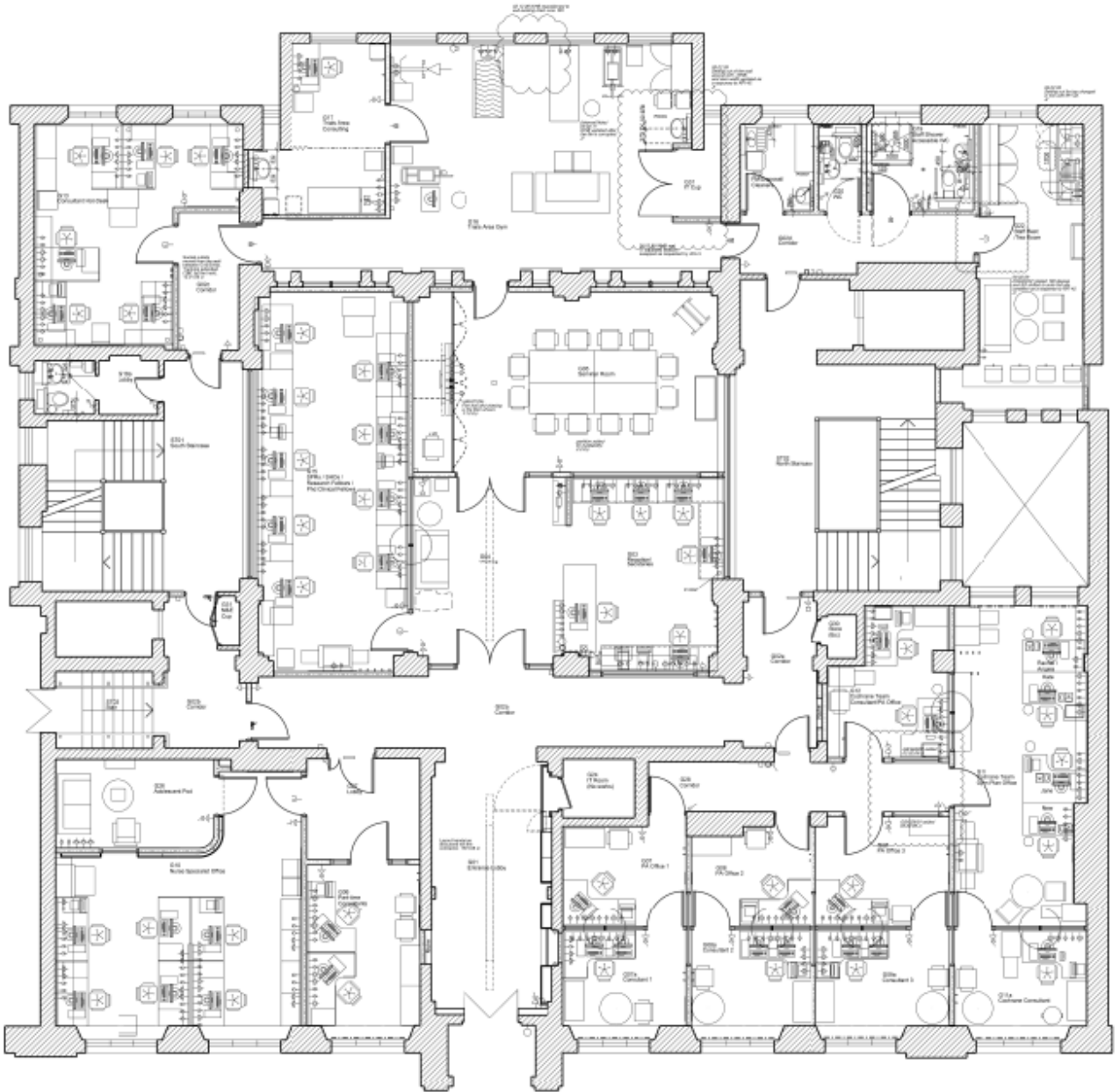
Secretary

Managing Editor

Trials Search Coordinator

Assistant Trials Search Coordinator

7. Floor plan and photo gallery









Who's who in the Centre for Neuromuscular Diseases



Gisela Barreto

Gisela Barreto has worked as a Trials Coordinator in the MRC Centre for Neuromuscular Diseases since February 2008. Her portfolio includes both commercial and non-commercial trials. She has a MEng in Medical Engineering from Queen Mary & Westfield College, University of London. Previously she has worked in cancer trials at the Institute of Cancer Research and also in phase I trials at Quintiles (CRO).

gisela.barreto@uclh.nhs.uk



Rachel Barton

Rachel Barton is one of two Trial Search Co-ordinators who support reviewers in designing and running search strategies on bibliographic databases such as MEDLINE. She moved with the Cochrane Group to Queen Square in 2008, and has previously worked as an NHS librarian.

r.barton@ion.ucl.ac.uk



Jane Batchelor

Jane Batchelor is Secretary to the Neuromuscular Disease Group which produces and disseminates systematic reviews of healthcare interventions. She joined the Group in 2006 when its editorial base was at King's College London. In September 2008 the base moved to UCLH in order to become part of the MRC Centre for Neuromuscular Diseases.

j.batchelor@ion.ucl.ac.uk



Michelle Bovell

Michelle Bovell is Medical Secretary to Dr Mary M Reilly. MRC Centre for Neuromuscular Diseases, and Medical Secretary to Karen Bull – Clinical Nurse Specialist. She works closely with Carol Brown, PA to Dr Reilly.

michelle.bovell@uclh.nhs.uk



Carol Brown

Carol Brown joined the Peripheral Nerve Team in September 2004 as PA to Dr Mary Reilly. She has seen the team evolve into the MRC Centre for Neuromuscular Diseases (the joining of the Peripheral Nerve Team and the Muscle Team) and supports Dr Reilly in her clinical work, and also some of her research work. She enjoys the diversity of the Centre's work and also the interaction between clinical and academic staff.

carol.brown@uclh.nhs.uk



Karen Bull

Karen Bull Joined the team as a Clinical Nurse Specialist for neuromuscular disorders three years ago. In this time she has developed a specialist interest in the care of the peripheral nerve patients, offering support education and monitoring of the client group. Prior to joining the team she has had a varied career which has included general medical and paediatric experience. She developed an interest in neurology, and prior to joining the team spent 14 years in general neurology, the last six of which were spent as the ward manager for neuro-medical patients and stroke rehabilitation.

karen.bull@uclh.nhs.uk



James Burge

Dr James Burge is a Specialist Registrar in Clinical Neurophysiology at the National Hospital for Neurology and Neurosurgery, and started a PhD under the supervision of Prof. Hanna in September 2008. He is studying mutations in the chloride channel, CIC1, that cause Myotonia Congenita, and developing a skeletal muscle expression system that will facilitate pre-clinical testing of novel therapies for the disease.

j.burge@ion.ucl.ac.uk



Jan Clarke

Jan Clarke is the motor neurone disease clinical nurse specialist and works closely with Robin Howard, Katie Sidle and Richard Orrell.

jan.clarke@uclh.nhs.uk



Liz Dewar

Liz is a senior physiotherapist working in the MRC Centre for NMD. She has worked in the NHNN since 2001 and specifically within the Neuromuscular team since 2007. She is involved in providing specialist physiotherapy assessment and advice for people with Neuromuscular conditions and also assists in some of the clinical research trials run by the Centre.

liz.dewar@uclh.nhs.uk



Helen Eddlestone

Helen Eddelstone is the respiratory clinical nurse specialist and works closely with Dr Hirsch.

helen.eddlestone@uclh.nhs.uk



Marcia Forde

Marcia Forde is Secretary to Dr Matt Parton, Consultant Neurologist in the muscle team at the Centre for Neuromuscular Diseases.

marcia.forde@uclh.nhs.uk



Dr Lionel Ginsberg

Dr Lionel Ginsberg is a consultant neurologist at the Royal Free Hospital and National Hospital for Neurology and Neurosurgery. He has a special interest in neuropathy and metabolic neuromuscular diseases.

Lionel.Ginsberg@royalfree.nhs.uk



Anne Grayson

Anne Grayson has been PA to Professor Hanna for the last ten years.

anne.grayson@uclh.nhs.uk



Stephanie Grisdale

Stephanie Grisdale is PA to Dr Lunn and Dr Manji in the Centre.

stephanie.grisdale@uclh.nhs.uk



Angela Gunn

Angela Gunn is one of two Trial Search Co-ordinators who support reviewers in designing and running search strategies on bibliographic databases such as MEDLINE. She moved with the Cochrane Group to Queen Square in 2008, and has previously worked as an NHS librarian.

a.gunn@ion.ucl.ac.uk



Mike Hanna

Professor Hanna is a Consultant Neurologist and Director of the MRC Centre. He runs the specialist muscle services and the Nationally Commissioned services for mitochondrial diseases and Muscle channelopathies in partnership with the muscle clinical team Dr Chris Turner, Dr Matt Parton, Dr Shamima Rahman, Dr Janice Holton, Liz Dewar, Cath Parry and Georgie Mewing. He undertakes clinical and genetic research in muscle diseases. He is the Queen Square Divisional Clinical Director.

mhanna@ion.ucl.ac.uk



Nick Hirsch

Dr Nick Hirsch is Consultant Anaesthetist and Head of the medical neurological ITU. He specialises in respiratory failure in neuromuscular and neurological diseases and runs the myasthenia service in partnership with Dr Robin Howard and Professor Dimitri Kullmann. He leads the Queen Square home ventilation support service.

nicholas.hirsch@uclh.nhs.uk



Robin Howard

Dr Robin Howard is a consultant neurologist and runs the specialist motor neurone disease service in partnership with Dr Katie Sidle. He is a specialist in neurological intensive care and has published widely on many aspects of neuromuscular diseases.

robin.howard@uclh.nhs.uk



Richard Hughes

Professor Richard Hughes has been Visiting Professor of Neurology in the MRC Neuromuscular Disease Centre since November 2007. With Michael Lunn, he is joint co-ordinating editor of the Cochrane Neuromuscular Disease Review Group until April 2010 after which he will remain an editor. His main research interests are treatment for inflammatory neuropathy and systematic reviews of neuromuscular disease.

r.hughes@ion.ucl.ac.uk



Elsbeth Hutton

Dr Elspeth Hutton is an Australian Neurologist, who moved to Queen Square in 2007 to take up a post as the Australian and New Zealand Neurology Fellow. A Clinical Research Fellow with an interest in neuropathic pain, she is currently studying potential cutaneous neuroimmune mechanisms in neuropathic pain with Dr Michael Lunn and Prof Martin Koltzenburg.

e.hutton@ion.ucl.ac.uk



Martin Koltzenburg

Professor Martin Koltzenburg is Head of Clinical neurophysiology and undertakes research into small fibre neuropathy, pain and neuromuscular channelopathies. He is Deputy Director of the MRC Centre for Neuromuscular Diseases.

m.koltzenburg@ich.ucl.ac.uk



Dimitri Kullmann

Professor Dimitri Kullmann's main research interests are neurological channelopathies, myasthenia gravis, neurocritical care and basic science of neuronal signalling. He runs the clinical myasthenia gravis service in partnership with Dr Nick Hirsch and Dr Robin Howard.

d.kullmann@ion.ucl.ac.uk



Matilde Laurá

Dr Matilde Laurá has worked as a Clinical Research Fellow since 2006. Her interests are peripheral neuropathies and in particular inherited neuropathies. Together with Dr Mary Reilly she carried out the Ascorbic Acid trial for patients with Charcot-Marie-Tooth disease type 1A.

m.laura@ion.ucl.ac.uk



Michael Lunn

Dr Michael Lunn was appointed as consultant neurologist in 2005. He has an interest in inflammatory neuropathies and his research has concerned Guillain-Barre syndrome, CIDP and paraproteinaemic neuropathies. He is also clinical lead in neuroimmunology and Coordinating Editor of the Cochrane Neuromuscular Disease Group, now based in the MRC Centre.

michael.lunn@uclh.nhs.uk



Hadi Manji

Dr Hadi Manji is a Consultant Neurologist and Senior Lecturer. He qualified in 1982, Trinity Hall Cambridge and Middlesex Hospital, and trained in Neurology at National Hospital, Queen Square and L'Hopital Kremlin Bicetre, Paris. He was appointed Consultant in 1997. His main interests are in infectious and inflammatory peripheral nerve disorders. Other interests include neurological infections including HIV. He was Chief author and editor of the Oxford Handbook of Neurology published in 2007.

hadi.manji@uclh.nhs.uk



Emma Matthews

Dr Emma Matthews is a clinical PhD research fellow working on the genetics of muscle channelopathies with Professor Hanna.

e.matthews@ion.ucl.ac.uk



Sarah Mazdon

Sarah Mazdon is PA to Dr Robin Howard.

sarah.mazdon@uclh.nhs.uk



Georgie Mewing

Georgie Mewing joined the Centre 10 months ago in the new post of Clinical Support Nurse. She is originally from Australia and has experience in many different specialities of nursing, however has taken a special interest in neuromuscular diseases.

georgie.mewing@uclh.nhs.uk



Adrian Miller

Dr Adrian Miller is a Clinical Research Fellow and Neurology Specialty Trainee. With Professor Linda Greensmith, he uses *in vitro* disease models to research potential therapeutic mechanisms for Inclusion Body Myositis (IBM). He is also involved in several national and international clinical studies of IBM, with Professor Mike Hanna, and in the MRC Centre IBM Research Clinic.

a.miller@ion.ucl.ac.uk



Jasper Morrow

Dr Jasper Morrow is a Clinical Research Fellow in Neuromuscular MRI. His research involves investigating new MRI techniques to better describe and quantify inherited and acquired neuromuscular diseases. He came to Queen Square from New Zealand in August 2008, and has been working at the centre for neuromuscular diseases since August 2009.

j.morrow@ion.ucl.ac.uk



Sinead Murphy

Dr Sinead Murphy started a neuromuscular fellowship from Dublin with Dr Mary Reilly in July 2009. She will be at Queen Square for one year to gain subspecialty experience in peripheral nerve disorders.

sinead.murphy@uclh.nhs.uk



Richard Orrell

Dr Richard Orrell has longstanding clinical and research interests in the wide range of neuromuscular disorders. This includes genetic, inflammatory, and degenerative conditions of muscle and nerve. He is particularly interested in motor neuron diseases, including amyotrophic lateral sclerosis. He is a consultant neurologist and runs the motor neurone disease service in partnership with Robin Howard and Katie Sidle.

r.orrell@medsch.ucl.ac.uk



Catherine Parry

Catherine Parry joined the team as a Clinical Nurse Specialist for neuromuscular disorders five years ago. In this time she has developed a specialist interest in the care of patients with muscle disease, in particular the areas of mitochondrial muscle disease, muscle channel disease and transition of care. Prior to joining the team she has had a varied career working in acute medicine and critical care settings. She developed an interest in neuromuscular disease when she spent 8 years previous to joining the team working on a long term ventilation unit, of which the last 18 months were spent as the Ward Manager. As well, she helped set up and develop a clinic specifically for patients with Duchenne Muscular Dystrophy and their families and worked on a Trust project for improving transition of care.

catherine.parry@uclh.nhs.uk



Matt Parton

Dr Matt Parton has been a consultant neurologist since 2006. Together with Professor Mike Hanna and Dr Chris Turner, he provides regular specialist muscle disease clinics. His particular interests in the service are in inclusion body myositis, the management of immune-mediated myositis and investigation of hereditary dystrophies. He is the clinical governance lead for the neuromuscular service. He also works as a general neurologist at Whipps Cross Hospital in East London and has wider interests in teaching and training.

matt.parson@uclh.nhs.uk



Dr Rob Pitceathly

Rob Pitceathly is a Clinical Research Fellow in Mitochondrial disease working for Professor Michael Hanna and Dr Shamima Rahman at the MRC Centre for Neuromuscular Diseases. He is currently recruiting patients with mitochondrial disease to a national database as part of a collaborative cohort study with colleagues at the MRC Centre for Neuromuscular Diseases in Newcastle.

r.pitceathly@ion.ucl.ac.uk



Alex Pollard

Alex Pollard is a Research Physiotherapist with an interest in peripheral neuropathies and in particular Charcot-Marie-Tooth Disease (CMT). He joined the centre in December 2009 and is currently working with Dr Reilly, Dr Lunn, Dr Ramdharry, Karen Bull and Liz Dewar on an exercise study for people with CMT.

a.pollard@ion.ucl.ac.uk



Simona Portaro

Dr Simona Portaro is a visiting research fellow from the University of Messina, working with Professor Hanna on muscle channelopathies.

simonaportaro@hotmail.it



Vina Pswarayi

Vina Pswarayi is the NCG mitochondrial and channel co-ordinator who answers and addresses all queries of an NCG nature

vina.pswarayi@uclh.nhs.uk



Shamima Rahman

Dr Shamima Rahman is DH/HEFCE Senior Lecturer at the UCL Institute of Child Health, Honorary Consultant in Mitochondrial Medicine at NHNN and Honorary Consultant in Paediatric Metabolic Medicine at Great Ormond Street Hospital (GOS). With Professor Mike Hanna, she runs the London National Commissioning Group (NCG)-funded clinical service for Rare Mitochondrial Diseases in Adults and Children (fortnightly adult mitochondrial clinic at NHNN and fortnightly paediatric mitochondrial clinic at GOS). Her research interests include molecular mechanisms underlying primary mitochondrial diseases (both mitochondrial DNA and nuclear gene-encoded); natural history of mitochondrial diseases; mitochondrial deafness and genetic susceptibility to aminoglycoside-mediated ototoxicity; and small molecule therapy of mitochondrial diseases.

s.rahman@ich.ucl.ac.uk



Dipa Raja Rayan

Dr Dipa Raja Rayan is a neurology trainee clinical research fellow undertaking a PhD in muscle channelopathies with Professor Hanna.

d.rajarayan@ion.ucl.ac.uk



Gita Ramdharry

Dr Gita Ramdharry has worked as a physiotherapist since 1995 and developed a special interest in neurology early on. She worked as a clinical physiotherapist at the NHNN on 2001 and moved into research at the ION in 2004. She completed a PhD in 2008 looking at walking patterns, endurance and orthotic interventions for people with Charcot-Marie-Tooth disease. She is now a part-time honorary researcher at the MRC centre; the rest of her time is spent as a lecturer at the School of Physiotherapy at St George's University of London.

g.Ramdharry@sgul.kingston.ac.uk



Mary Reilly

Dr Mary Reilly is a consultant neurologist and lead for the peripheral nerve service. She is interested clinically in all forms of neuropathies but has a particular interest in inherited neuropathies such as Charcot-Marie Tooth Disease. She is currently conducting genetic lab and clinical based research and clinical trials in this area.

m.reilly@ion.ucl.ac.uk



Massimo Russo

Dr Massimo Russo is completing a neuromuscular fellowship at the MRC Centre from Italy. His primary interest is neuropathy, particularly inherited.

massimorusso81@hotmail.it



Penny Schafer

Penny Schafer is secretary to Dr Chris Turner for the muscle service in the centre.

penny.schafer@uclh.nhs.uk



Anthony Schapira

Professor Schapira's research area in neuromuscular disease is within the mitochondrial myopathies, particularly in the area of mitochondrial DNA depletion syndromes and mitochondrial involvement in neurodegenerative disorders. He has been at Queen Square since 1984 and has been a Professor at Queen Square since 1990.

anthony.schapira@royalfree.nhs.uk



Zoë Scott

Zoë has been Centre Senior Administrator of the MRC Centre for Neuromuscular Diseases since March 2008, and works with Professor Hanna, Dr Reilly and other members of the Steering Committee in London and Newcastle, coordinating all aspects of the Centre's activities.

z.scott@ion.ucl.ac.uk



Katie Sidle

Dr Sidle is a consultant neurologist specializing in the field of Motor Neurone Disease (MND). The NHNN is the MND Association regional care centre for patients with MND and she is responsible for the care of patients with MND in the weekly MND clinic. She has a background in molecular genetics and is currently developing research interests in the field of MND with colleagues within the neighbouring Institute of Neurology.

katie.sidle@uclh.nhs.uk



Chris Sinclair

Dr Chris Sinclair, the centre MRI Physicist, came to Queen Square in 2008 to work on neuromuscular magnetic resonance imaging (MRI). He is principally involved in developing advanced quantitative MRI techniques for application in neuromuscular diseases, including several patient studies within the centre.

c.sinclair@ion.ucl.ac.uk



Jennifer Spillane

Dr Jennifer Spillane is the John Newsom Davis clinical research fellow in myasthenia gravis undertaking a PhD in the centre.

j.spillane@ion.ucl.ac.uk



Chris Turner

Dr Chris Turner has been working as a consultant at the MRC Neuromuscular Centre and UCLH since 2007. He has a specialist interest in muscle disease and runs a twice monthly myotonic dystrophy clinic at Queen Square

chris.turner@uclh.nhs.uk

9. Running the Centre

Consultant Leadership Team

A monthly consultant leadership team will meet to consider all aspects related to running the Centre. There will be an agenda which will include, but not be limited to , the following standing items:

- Allocation of space in the centre.
- Consideration of applications for trials using centre facilities
- Maintenance of the centre website
- Production of the annual centre brochure
- Links with the Cochrane unit
- Links with the MRC Centre
- Fundraising
- Staffing
- Maintenance of infrastructure including the trials equipment
- Centre database and coordinator
- Blue sky for the centre-phase II etc

The consultant leadership team will be in addition to the regular team and leadership meetings which include:

- MRC Centre steering committee
- NCG mitochondrial and Muscle channel MDT meetings
- Muscle team MDT
- Peripheral Nerve team MDT
- Peripheral Nerve genetic meeting
- Muscle pathology meeting
- Peripheral Nerve pathology meeting
- Neuroimmunology team meeting

Booking rooms in the centre

It is essential that all usage of seminar rooms and trials consultation room facilities are booked in advance.

Seminar room:	Anne Grayson	x713014
Trials room:	Carol Brown	x713457

For help using the audiovisual facilities in the seminar room please contact Anne Grayson or Zoë Scott.

Centre for Neuromuscular Diseases

Box 102

National Hospital for Neurology and Neurosurgery

Queen Square

London WC1N 3BG

UK

T: +44 845 155 5000 x723013

F: +44 20 7676 2079

admin-cnmd@ion.ucl.ac.uk