



Building on the Foundations: The Need for a Specialist Neuromuscular Service Across England

A report presented to the All Party
Parliamentary Group for Muscular Dystrophy

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The report has received endorsement from a number of organisations, including:

- Charcot-Marie-Tooth Disease UK
- Duchenne Family Support Group
- FSH Muscular Dystrophy Support Group
- Myasthenia Gravis Association
- The Association for Glycogen Storage Disease (UK)

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Foreword

In this hard hitting report, a number of my medical colleagues set out a compelling case in my view for national commissioning of services for patients with neuromuscular disease.

Although there are some centres of excellence in Newcastle, London, Oxford and Oswestry, in other parts of the country the picture is regrettably very different indeed, since services for patients with neuromuscular disease and particularly for Duchenne muscular dystrophy are extremely limited.



The result of this situation is that patients with Duchenne muscular dystrophy are having far fewer years of life if they happen to live in a poorly-served region than if they are fortunate to live in the North East, for example, where they can access the excellent services provided in Newcastle.

I have a personal interest in this field since I began research into neuromuscular disease, and in particular the muscular dystrophies, in the early 1950s, and established a major research centre in Newcastle upon Tyne. Subsequently for many years I directed the neuromuscular services and its research arm in Newcastle before I moved to Oxford as Warden of Green College in 1989. I am delighted that in both of those cities outstanding work on neuromuscular disease, in research but also in patient care, has continued to burgeon over the years.

I think the case made in this document is a very powerful one, and I urge policy makers, parliamentarians and professionals to take note of its findings and do all they can to improve patient care for people – both children and adults - living with neuromuscular conditions. A commitment to develop a national neuromuscular service supported by specialist commissioning is urgently needed.

A handwritten signature in black ink that reads "John Walton" with "(Walton of Detchant)" written below it in a smaller, cursive hand.

Lord Walton of Detchant, Kt TD MA MD DSc FRCP Fmed Sci

Biographical note

Lord Walton was a founder of the Muscular Dystrophy Campaign, its Chair from 1971-95 and is now Life President. He had a distinguished medical career and his outstanding contribution to research and clinical development has been widely recognised through his holding of the offices of President of the General Medical Council, President of the British Medical Association, President of the Royal Society of Medicine, President of the Association of British Neurologists and President of the World Federation of Neurology. He was made an Honorary Freeman of Newcastle upon Tyne in 1980 and became a crossbencher, independent life peer in 1989.

Introduction

“Our ambition should be nothing less than the creation of a world class NHS that prevents ill health, saves lives and improves the quality of people’s lives.”¹

Professor Lord Darzi, Health Minister

The current care for people with neuromuscular conditions in England fails to meet the ambition set out for the NHS by Health Minister, Professor Lord Darzi.

It is simply unacceptable that many patients are not receiving good levels of care, people’s survival is limited according to the area in which they happen to live and many are not enjoying the best possible quality of life.

These problems and shortcomings in NHS services in many parts of the country are avoidable – we know the improvements that need to be made and the steps required are described in this paper.

The current situation is leading to premature deaths and poor standards of care in many parts of the country with services at the specialist centres vulnerable and not firmly embedded. We have specialist neuromuscular centres in four regions of England and the NHS should now commit to the planning and development of a national neuromuscular service.

Comprehensive neuromuscular services should be designated within the Specialised Services National Definition Set, an audit of current services should be undertaken and the Department of Health should work with the emerging British Myology Society (the professional body for neuromuscular specialists) in approving a Standard of Diagnosis and Care for Neuromuscular Conditions.

The muscular dystrophies and related neuromuscular conditions

There are more than 60 different types of muscular dystrophy and related neuromuscular conditions. Over 1,000 children and adults for every 1 million of the population are affected by muscle wasting neuromuscular diseases in England. These include muscle diseases such as muscular dystrophy and peripheral nerve diseases such as Charcot-Marie-Tooth disease. Inherited neuromuscular disease includes a number of rare disorders which frequently result in progressive muscle wasting and weakness, orthopaedic deformity, cardiac and respiratory compromise.

Some of these disorders, for example myotonic dystrophy, affect multiple systems. The definition of an inherited neuromuscular disorder includes: the muscular dystrophies, the spinal muscular atrophies, the congenital neuropathies, metabolic myopathies, inherited myasthenic syndromes, channelopathies, the myotonias and the inherited neuropathies.

These disorders cause progressive muscle wasting and weakness and result in premature death or life-long disability. They often start in childhood or young adult life. Most neuromuscular diseases are genetic but some are acquired.

Executive Summary

The Problem

- Survival - Patient survival is significantly reduced for some conditions in those regions of the country where comprehensive neuromuscular services are not provided (as data for patients with Duchenne muscular dystrophy shows);
- Variable - Evidence from a Freedom of Information Request to Primary Care Trusts in October 2007, specialist centre records and patients' experience shows that neuromuscular services are variable across the country and where patients live often adversely affects the care they receive;
- Vulnerable - Where excellent services are in place in just four regions, they are vulnerable given their dependence on a handful of leading clinicians who may in time move on or retire. Funding received by the centres is vulnerable and dependent on charitable subsidy;
- Advances in recent years have provided clear evidence that specialist care provided by a multidisciplinary team prolongs life, reduces morbidity and helps prevent transmission of the genetic disease in patients with neuromuscular diseases. Many patients in England do not receive such multi-disciplinary specialist care and outcomes are adversely affected.

Key Asks

We are calling on the Government and the NHS to:

- Recognise that current service provision for neuromuscular conditions is failing many patients and acknowledge that all patients with neuromuscular conditions require access to specialist diagnosis, treatment and on-going care;
- Designate specialised neuromuscular services within the Department of Health's Specialised Services Definition Set so that Specialised Commissioning Groups (SCGs) strategically plan for these services across England;
- Press SCGs to ensure that specialised neuromuscular services are firmly embedded in their region of England in order to overcome current fragmentation, weakness and vulnerability;
- Pay particular attention to those regions highlighted in this report where patient survival is reduced and services are failing;
- Recognise that services should be delivered through specialised neuromuscular regional centres which also give support and clinical leadership to a network of local clinics;
- Ensure that these neuromuscular centres and clinics have specialised multi-disciplinary teams to provide all patients with a comprehensive level of care.

Issues to be Addressed

- Specialised Neuromuscular Services are not recognised on the Department of Health's Specialised Services Definition Set;
- Many patients in England with neuromuscular diseases do not receive specialist multi-disciplinary care;
- Inequality of access to specialist care across different regions in England leads to a 'postcode lottery' for patients;
- Life expectancy for muscular dystrophy varies in different regions in the UK;
- Ventilation support prolongs life (for example in DMD as part of multi-disciplinary management from a mean of 19 years in the 1970s to nearly 30 years now, Eagle et al, 2007) but is not provided in a systematic way across all regions of England;
- Many patients with neuromuscular disease do not receive an accurate genetic diagnosis or accurate advice regarding prognosis and transmission risks
- Many patients with muscular dystrophies such as DMD are not offered 'best standard of care' treatments such as corticosteroids, and the optimal follow up to avoid serious complications
- Those with treatable conditions, such as myasthenia, frequently do not receive optimum treatment and suffer morbidity, and occasionally mortality;
- Current PCT commissioning arrangements are failing patients with neuromuscular diseases. Adequately resourced clinics are needed to care effectively for patients with neuromuscular diseases;
- Current PCT commissioning arrangements for neuromuscular conditions do not result in effective collaboration of services in certain regions;
- PCT commissioning arrangements mean that in some places core services are only provided by fundraising efforts of charities (such as the Muscular Dystrophy Campaign) and patients themselves.

1. Specialised neuromuscular services are not designated on the Department of Health's Specialised Services Definition Set

Although rare neuromuscular services are linked to other services designated within the Specialised Services Definition Set², a comprehensive neuromuscular service is not included within the Set. It is clinically recognised that neuromuscular diseases require a multi-disciplinary specialist approach to care. It is clear that services currently fall short of what is needed in several regions with a consequent impact on patient outcomes including survival rates.

The neuromuscular conditions are low prevalence conditions and specialist services are not consistently commissioned by Primary Care Trusts (PCTs) or by Specialised Commissioning Groups (SCGs). Therefore, NHS commissioning arrangements currently fail to support services that meet the needs of many children and adults with neuromuscular conditions.

Further, the designation of comprehensive neuromuscular services within the National Definition Set would require SCGs to establish a broad base-line position, make initial comparisons on activity and spend and provide an important basis for service reviews and strategic planning in addition to the benefits for patients.

2. Many patients in England with neuromuscular diseases do not receive specialist multi-disciplinary care

Specialist multi-disciplinary care has been developed by leading clinicians as the best model for delivering effective care for such complex, multi-system diseases. The provision of expert physiotherapy, orthotics, early cardiac monitoring and intervention and corticosteroids has been shown to improve muscle function and maintain independent mobility.³ The judicious use of spinal surgery and expert respiratory services (including non invasive positive pressure ventilation) helps to improve quality of life, delay the onset of respiratory failure and prolong the life of these patients.⁴

The specialists that deliver this care vary from neurologists, to clinical geneticists, to paediatricians and paediatric neurologists, to rehabilitation physicians. It is unusual to find any region or centre in the UK that can offer a comprehensive service which is able to address all the requirements for patients with genetic and acquired muscle and peripheral nerve diseases. Furthermore, while patients in some areas may see a specialist in terms of a neurologist or paediatric neurologist, these "specialists" in practice often have little or no expertise of neuromuscular conditions.

The evolution of services for patients with neuromuscular diseases in England has been inconsistent and, indeed, heavily dependent on the research interests of dedicated individuals who have developed a clinical interest in a neuromuscular disease. There is usually no strategy in place for succession planning, leaving the services fragile and vulnerable in view of their heavy dependence on the lead clinicians.

The Muscular Dystrophy Campaign (MDC) undertook a survey of 152 PCTs in England in October 2007 in order to build a picture of access to healthcare services for people with neuromuscular conditions. The MDC asked the PCTs to provide information regarding services commissioned locally for people with neuromuscular disease. (Appendix 3)

Out of the 114 PCTs that responded (a response rate of 75%), the following picture has emerged:

- **74%** of PCTs who responded do not support a muscle clinic that offers a service to adults with neuromuscular conditions;
- **69%** of PCTs who responded do not support a muscle clinic that offers a service to children with neuromuscular conditions;
- **65%** of PCTs who responded do not support any adult or child muscle clinics within their area

3. Inequality of access to specialist care across different regions in England

Recognised specialist neuromuscular centres, which are highly dependent on charity subsidy, are currently only located in London, Newcastle, Oxford and Oswestry. It is important that the resources necessary are provided to ensure that these centres become embedded in a properly resourced, long term service.

While patients who live relatively close to one of the neuromuscular centres may be referred there and receive excellent care, most patients live outside these regions and receive inferior services or, indeed, may receive no services at all (unless they are referred out of area to one of the centres).

Failure to invest in the development of specialist neuromuscular services will in time lead to a growing increase in inequality for patients in terms of both service provision and long term clinical outcomes. Preliminary evidence from a study in progress at the Newcastle Centre (highlighted below) has established the benefits for patients of the specialised multi-disciplinary care model.

Benefits of specialised multi-disciplinary care - preliminary evidence from a qualitative research study:

The care provided to the families attending the paediatric muscle clinic at Newcastle has recently been assessed by a PhD student who studied the availability of and satisfaction with Home and Community Based Services for Children with Neuromuscular Disorders.

The results of this study offer the first concrete evidence of a positive benefit to the care model of a Muscle Centre with multidisciplinary input in improving the experience of patients with chronic disability. These parents were not experiencing the same level of difficulty described in most previous research about the support needs of disabled children and families. Part of the reason for this was felt to be the support provided by the Muscle Team.

This qualitative study, supervised by Professor John Carpenter, initially of the University of Durham but now in Bristol, aimed to explore children and young people with neuromuscular impairments and their parents' experiences with education, health and social care services. The study used the accounts of children and young people themselves and those of their parents or carers, exploring their perceptions of education, health and social care services.

Existing specialist centres

Lead clinicians	Specialist centres
Professor Michael Hanna, Consultant Neurologist Professor Francesco Muntoni, Consultant Paediatric Neurologist Dr Mary Reilly Consultant Neurologist	MRC Centre for Neuromuscular Diseases, London Dubowitz Neuromuscular Centre, London
Professor Katie Bushby Professor Volker Straub Professor Hanns Lochmuller, Consultant Clinical Geneticists Professor Doug Turnbull Professor Patrick Chinnery Dr James Miller, Consultant Neurologists	MRC Centre for Life, Newcastle
Dr David Hilton-Jones, Consultant Neurologist	MDC Oxford Muscle and Nerve Centre
Dr Ros Quinlivan, Consultant Paediatric Neurologist Dr Martin Smith, Paediatric Neurologist Dr Debbie Short, Consultant in Rehabilitation Mr Nigel Kiely, Children's Orthopaedic Surgeon Professor Caroline Sewry, Clinical Scientist & Muscle Pathologist	The Wolfson Centre for Inherited Neuromuscular Disease, Oswestry

Specialist staff at a Neuromuscular Centre

The lead clinicians at the existing specialist centres have identified and developed the specialist staff team needed to provide a comprehensive specialist neuromuscular service. (In Appendix 1, the staff specialisms at the Newcastle centre are highlighted and the specialist services provided at the Hammersmith Hospital in London are set out in Appendix 2.)

The specialist staff team involved with patients with neuromuscular diseases at the Oxford Muscle and Nerve Centre includes the following specialisms:

- Adult neurologist with specialist interest in NMD (including myasthenia)
- Adult neurologist with specialist interest in neuropathies
- Adult neurologist with specialist interest in congenital myasthenia
- Paediatric neurologist with specialist interest in NMD
- Respiratory physician who runs the non-invasive ventilation service
- Cardiologist with specialist interest in NMD
- Genetic counsellor
- Muscle physiotherapist
- Regional Care Advisor
- Neuromuscular nurse specialist

In addition, there is a research capacity and administrative support at the Oxford centre provided through a neuromuscular research fellow, a laboratory scientist and an administrator.

A regional 'hub and spoke' model

The most effective way to deliver patient care for people living with a neuromuscular condition follows a regionally based 'hub and spoke' model with a network of specialist regional centres providing care to patients at the hub while also giving clinical leadership, supervising and sharing care with local hospitals and primary care teams. Indeed, the Newcastle Muscle Centre has for more than 15 years been providing "outreach" clinics to several sites across the Northern Region thereby providing specialist services closer to people's homes and allowing the better involvement of local staff.

Evidence from published studies, feedback from MDC Regional Care Advisors and the MDC survey of PCTs clearly show **the Southwest region of England is one of the most poorly served areas for adult neuromuscular services**. It also appears that relatively few adult patients are referred out of area as the Oxford Muscle and Nerve Centre sees fewer than 10 adult patients a year from the Southwest region.

An audit of existing services should be undertaken to identify other areas with particularly weak services but areas of immediate concern are the South West and Eastern regions of England.

Unfortunately, this does not mean that paediatric and adult services in other areas and regions are comprehensive. Indeed, there are obviously many areas where the clinical teams working in both child and adult clinics do not currently include some of the key specialisms such as neuromuscular physiotherapists or specialised speech and language and dietician specialists. Patients face long, tiring and expensive journeys if they want to access specialist support such as that provided at the MDC's NeuroMuscular Centre based in Winsford, Cheshire:

Patient Case No. 1

DK is 21 years old, has Becker muscular dystrophy and lives in Ripley in Derbyshire. In his own words: *"I really need regular physiotherapy to keep me flexible and mobile. This is simply not available to me in my home area. It takes me around 1 hour 45 minutes to travel to the NeuroMuscular Centre (NMC) in Cheshire for treatment and, of course, the same to get home (all at my own cost) but for an hour and half of specialist, knowledgeable physiotherapy it is really worth all that."*

A second example is identified in the Eastern region where, due to funding restraints at PCT level, the region no longer provides a Regional Care Advisor and also does not provide a specialist psychologist for patients with neuromuscular conditions as the psychology team is currently restricted to working with brain injury patients.

Patient Case No. 2

One family in Suffolk is forced to travel from Suffolk to London, a journey of up to five hours each way, in order to receive specialist healthcare for their severely disabled seven-year-old son. The mother says the long journey affects the treatment of the little boy, who is often exhausted and uncooperative by the time he sees the doctors.

The following conditions need to be covered by a specialist multi-disciplinary service but may not necessarily be provided by a single centre in most regions:

- All genetic muscle diseases
- All genetic peripheral nerve diseases
- All acquired muscle diseases
- All acquired nerve diseases
- All genetic myasthenia
- All acquired autoimmune myasthenia

4. Life expectancy for muscular dystrophy varies in different regions in the UK

It is unacceptable that patients are currently receiving relatively weak services in some regions and this leads to significant variations in survival rates for patients with Duchenne muscular dystrophy (DMD).

An audit of 40 sequential DMD deaths over 10 years in the Southwest region showed a median age of death of 18 years. This compares with a mean of age of death of almost 30 years in patients with DMD receiving home ventilation and specialist multi-disciplinary care reported by the Newcastle group in the most recent study by Eagle et al (2007).⁵

In addition, the extent of the feeding difficulties experienced by patients with neuromuscular disorders, and the implication for long term management and survival are only starting to be appreciated. Patients with neuromuscular conditions have presented with substantially faltering weight, and malnutrition may lead to an increased rate of chest infection. Lack of specialist dieticians and speech and language therapists has in some areas impacted on the morbidity of children with neuromuscular conditions.⁶

These variations in survival and clinical outcomes must be addressed and specialist neuromuscular services introduced where they are needed. In addition to the necessary investment in services, agreed standards and protocols should be established and promulgated widely.

Service standards and protocols for the neuromuscular conditions

Progress is being made in developing agreement on standards of care for neuromuscular conditions through the major TREAT-NMD initiative which is co-ordinated from the Newcastle centre (www.treat-nmd.eu). TREAT-NMD brings together some of the world's leading neuromuscular specialists in a pan-European 'network of excellence' and engages leading specialists worldwide to create international consensus on care and management for patients suffering from neuromuscular diseases. Consensus has already been agreed by an international group of experts for the standard of care of patients with spinal muscular atrophy, and this will follow for other diseases as the network develops.

Further, it is planned to launch the British Myology Society (BMS) in early 2008. The BMS will ensure that standards of care and best practice guidelines are developed and introduced in the UK for all neuromuscular diseases and will also support and facilitate relevant aspects of the work of TREAT-NMD. The BMS will also take on the role of the professional body in the UK that provides the clinical lead to commissioners regarding specialist service provision.

The third example shows the lack of service provision for adults in the South West:

Patient Case No. 3

“My brother and I are adults and we have Becker muscular dystrophy. We have been waiting for over two years to see a specialist in our condition and in that time I have experienced a real deterioration in my mobility. Two years ago we were seen at Frenchay Hospital but not by a specialist and so the appointment was a waste of time.

“We were again referred by our GP to a consultant who we had been told specialised in muscular dystrophy. Our mother had to chase the appointment for months as we heard nothing. When we eventually had the appointment we saw another consultant, who said he did not know why he was seeing us as he did not have an expert knowledge of our condition. We have recently gone back to our GP who is very supportive and stunned by the poor treatment we have received to this date. She has now requested an urgent referral to the muscle centre in Oxford. However, there is no guarantee that this referral will be approved as it is out of area and while we are desperate to receive specialised services we are concerned about the distance we will have to travel. I have also been experiencing great difficulty seeing a specialist neuromuscular physiotherapist. The only centre I know of that provides such treatment is 170 miles away in Cheshire and this is no good as I really need regular treatment.”

5. Ventilatory support prolongs life but is not provided in a systematic way across all regions of England

Many patients living with Duchenne muscular dystrophy and their families are aware of the significant increase in survival rates that are being achieved in some countries and, indeed, in some parts of the UK.

However, the increase in survival rates is not being achieved uniformly in the regions of England (as reported in section 4 above) and it is unacceptable that patients' survival can depend largely upon the area in which they happen to live.

It should be noted that funding for ventilation and respiratory support for adult patients with neuromuscular conditions has to be negotiated locally each year at Queen Square from the tariff for specialist clinic attendance, in competition with other clinical needs. This is another example of the fragility of the service.

6. Many patients with neuromuscular disease do not receive an accurate diagnosis, a precise genetic diagnosis or accurate advice regarding prognosis and transmission risks

The importance of an accurate diagnosis cannot be overstated as the following case study illustrates:

Patient Case No. 4

Mr B was referred to the Muscle Centre in Oswestry. He had been diagnosed with McArdle's disease 10 years earlier. McArdle's disease is a rare disorder of muscle metabolism which causes difficulty with strenuous exercise with a risk of severe muscle damage (rhabdomyolysis) and acute renal failure.

The patient had been posting messages on a website for this condition, encouraging other patients to undertake weight lifting as a past-time as it made him feel better.

When the patient was seen in a specialist centre, it was clear that he did not have McArdle's disease but rather Becker muscular dystrophy, confirmed by both muscle biopsy and DNA analysis. The discovery that he had a progressive disorder leading to increasing physical disability and that his two daughters were carriers of the condition (having previously been informed that he could not transmit the disorder to his offspring) led to an acute depressive state leading to the loss of employment and breakdown of his marriage. Furthermore, the patient's website messages could potentially have resulted in collapse and acute renal failure in a definite McArdle's patient.

7. Those with treatable conditions, such as myasthenia, frequently do not receive optimum treatment and suffer morbidity, and occasionally mortality

There are a number of recognised treatments for myasthenia gravis which depend on a correct diagnosis being made. It is an autoimmune disease in which muscle contractions are weak following the loss of muscle receptors. Myasthenia is one of the adult onset conditions and diagnosis should be made by a neurologist with experience of the condition.

8. PCT commissioning arrangements do not result in sufficient service provision for patients with neuromuscular diseases

As the neuromuscular conditions are not currently designated in the National Definition Set, there is a complete lack of a coordinated strategy being followed either by SCGs at the regional level or by PCTs acting alone or in consortia at the local level.

This was well illustrated in one of the PCT responses to the MDC survey which recognised that for a local PCT the group of people with neuromuscular conditions is too small to develop an efficient specialist service.

Without the adoption of an England-wide strategy for the planning and development of specialist neuromuscular provision on a 'hub and spoke' regional model, services have evolved on an ad hoc basis dependent on interested and committed clinicians leading to the inherent fragility of this approach (as emphasised in this report).

To ensure that the much needed improvements are made in service provision, work should be initiated without delay to develop a national Department of Health approved Standard of Diagnosis and Care for neuromuscular conditions, to be endorsed by the emergent BMS. This would provide SCGs and PCTs with a benchmark against which neuromuscular services can be regularly assessed and is essential in maintaining and driving up standards across the country.

9. PCT commissioning arrangements do not result in adequately resourced clinics to care for patients effectively

While neurologists usually operate out of neuroscience centres, it has to be remembered that not all neuroscience centres have neurologists who specialise in neuromuscular disease and some muscle centres do not operate out of Neuroscience Centres.

Similarly, there is no requirement within the commissioning structure to ensure that the staffing levels within paediatric neurology departments include a specialist in neuromuscular diseases and most departments rely on the individual interests of applicants for posts rather than recruiting directly a specialist with an interest in a neuromuscular disease.

In addition, the MDC survey of PCTs shows that where there are neuromuscular clinics these are not consistently able to provide a comprehensive multi-disciplinary approach to care. The survey findings support those reported by Hill and Phillips who identified that 32% of clinics did not routinely monitor respiratory function and 10% did not routinely perform ECGs. They further recorded that follow-up was variable; physiotherapists only offered long-term review of patients in seven centres, only five of which had a specialist muscle physiotherapist.⁷

10. PCT commissioning arrangements for neuromuscular conditions do not result in effective collaboration of services in certain regions

The Muscular Dystrophy Campaign survey of PCTs revealed a worrying lack of communication internally between PCTs over their collaboration and commissioning arrangements.

One PCT which leads a regional specialist commissioning consortium responded to the survey questions on behalf of all the 24 PCTs in its area. The lead PCT reported that a neuromuscular paediatric service

was commissioned in the 2007/08 planning round. Further, they commented that adult neuromuscular patients are referred to the comprehensive specialist centres in Newcastle, Oxford and London.

However, one of the PCTs within this regional consortium itself responded directly to our FOI request and this PCT informed us that it currently commissions an adult neuromuscular clinic which is actually located within its area. This reflects at the very least a lack of communication between commissioners within this regional commissioning consortium and it also reveals that the lead PCT is unaware that there is a closer centre in the region which adults could attend.

In planning service growth and development, it would be helpful to examine whether one of the existing adult neuromuscular clinics in a region can be developed to provide a comprehensive, regional specialist service for adult patients with a research and clinical leadership role.

11. PCT commissioning arrangements means that in some places core services are only provided by fund raising efforts of patients and charities

The Muscular Dystrophy Campaign is providing direct financial support in 2007/8 and 2008/9 to maintain some of the specialist multidisciplinary services at a number of the existing centres. These are mainstream NHS centres and it should be recognised that the charity is unable to give any long-term commitment regarding future funding. Further, in some areas, patients are also raising funds themselves to support existing services and this underlines the fragility of the current neuromuscular service.

Specialist neuromuscular services need to be embedded and resourced within the NHS and their long-term development and future secured. Designating specialised neuromuscular services within the Specialised Services Definition Set will be a crucial step in ensuring that this happens.

Action Needed

1. Neuromuscular diseases require a multi-disciplinary specialist approach to care and comprehensive neuromuscular services should be designated within the Department of Health's Specialised Definition Set. This would provide an important basis for commissioning, service reviews and strategic planning and enable commissioners to establish a broad base-line position and make initial comparisons on activity and spend.
2. The care of all patients with a neuromuscular condition should be led from regional Specialist Neuromuscular Centres with specialist multi-disciplinary teams. These centres should be regionally based to ensure better access to care and will provide a comprehensive package of care to patients. Patient care should be delivered by a 'hub and spoke' arrangement with the specialist centre at the hub providing care and also sharing care provision with local clinics based in hospitals and primary care teams. While details of the service model may vary between patients and areas, the key is that specialist supervision supports and oversees local provision. **Further, the additional cost of introducing the national neuromuscular service based on a network of regional centres and local muscle clinics is offset by the fact that many specialist staff are already working in the NHS albeit not currently operating formally within a co-ordinated, national service.**
3. A national Department of Health approved Standard of Diagnosis and Care for the neuromuscular conditions should be developed and agreed. The British Myology Society, to be launched in 2008, will endorse such standards for diagnosis and care and it will also support the work of TREAT-NMD, an international initiative backed by the Department of Health, so that agreed standards of care and best practice guidelines are introduced in the UK for all neuromuscular diseases. These steps will provide standards against which the service can be assessed by commissioners and lead to consistent, high quality care with improved patient outcomes.
4. The agreed Standards of Diagnosis and Care should be disseminated throughout England, including to patients and patient groups. This would ensure equity of access to specialist multi-disciplinary services and provide assurance to patients of the level of care they can expect.
5. An audit of existing specialist neuromuscular services should be undertaken. This will assess the current levels of care in each SHA against the agreed standards and lead to action to address any weaknesses in regional and local provision.
6. Neuromuscular Centres must be adequately resourced in staffing terms to offer comprehensive care to patients. Some evidence is already available as to the cost effectiveness of specialist staff (including the neuromuscular nurse) in reducing in-patient days and ensuring better clinical management.
7. The needs of patients also require a renewed focus on workforce planning with, in particular, specialist posts to be designated and established within the specialist centres and clinics. Further, the issue of succession planning should be addressed urgently especially for the lead clinician posts which are vital to the specialist neuromuscular service.



Appendix 1

Expertise Needed at a Neuromuscular Centre - Example of the Centre for Life, Newcastle

Population in the area: 3 million.

There are some 1100 patients within the inherited categories (excluding peripheral nerve).

Clinic organisation purely for this inherited group each week: one new patient clinic, one adult clinic, one paediatric clinic. Clinics organized centrally as well as in peripheral hospitals. In addition, one orthotics clinic per week and one myotonic dystrophy clinic every two weeks.

Essential staff

Consultants: for a population of 3 million people, 2 WTE consultants are needed purely on the inherited side calculated as follows:

1.5 WTE clinical academic professor of genetics

Paediatric neurology: approx consultant Pas per week- 4 from paed neuro.

Adult neurology: approx 2 Pas per week in "our" part of the service: if the mitochondrial neuropathies and acquired areas are included estimated input of 1 WTE overall.

Every clinic needs to have input from: nurse specialist, physiotherapy, RCA. Preferably also psychology and dietetics. From this Centre's experience that there should be at least two full time nurses and two full time physiotherapists plus a physiotherapy assistant covering simply the paediatric and adult inherited service as defined above.

Newcastle covers genetics automatically: uses 0.5 FTE of genetic nurse time plus around 0.5 PA of a genetic consultant

Formal links to other specialties: cardiology, respiratory assessment and support, orthopaedics, GI etc

Plus access to diagnostics expertise

Example from Hammersmith Hospital

Multidisciplinary activities providing specialised neuromuscular clinical care in a paediatric neurology environment at the Hammersmith Hospital

1. Multidisciplinary activities of the core team include:

- Histopathology team (muscle biopsies; weekly combined muscle pathology meetings);
- Medical team;
- Specialist Physiotherapists;
- Speech and language therapists and dietician;
- Specialist muscle nurse;
- General paediatric nurse;
- Research physiotherapist;
- Psychologist;
- Family Care advisor;
- Clinical secretaries.

Each week, there are 2 multidisciplinary meetings; in addition there are at least monthly team activities (scientific presentations; audit; policy making meetings and so on).

2. Other Hammersmith Hospital Trust / Imperial College collaborators include:

- Cardiologists from the HHT - with whom there is a very active collaboration;
- Neurophysiologist and the neuroradiologist - regular clinic-academic meetings are held with these specialities.

3. Combined activities with colleagues from other disciplines and other hospitals include:

- Combined respiratory clinic;
- Neurogenetic clinic;
- Orthopaedic shoulder clinic;
- Orthopaedic spine clinic;
- Orthopaedic soft tissue clinic;
- Peripheral neuropathy clinic;
- Lax ligamens clinic;
- Meetings with the anaesthetists and the spinal surgery nurse;
- Sitting clinic OT.

Appendix 3

Survey of Primary Care Trusts in England

Introduction

The purpose of the survey was to identify which areas in England commission specialist muscle clinics for children and adults with neuromuscular conditions.

Method

In October 2007, the Muscular Dystrophy Campaign contacted by email 152 Primary Care Trusts in England and under the Freedom of Information Act asked the following questions:

- Does your PCT currently support a muscle clinic that offers a comprehensive service to (a) children and (b) adults with a neuromuscular condition?
- If you do support a muscle clinic for children and/or adults, where is the clinic located and who is the lead clinician/Head of service?
- If patients are referred out of the local area, I would be grateful if you could indicate this and provide details.

Primary Care Trusts who responded

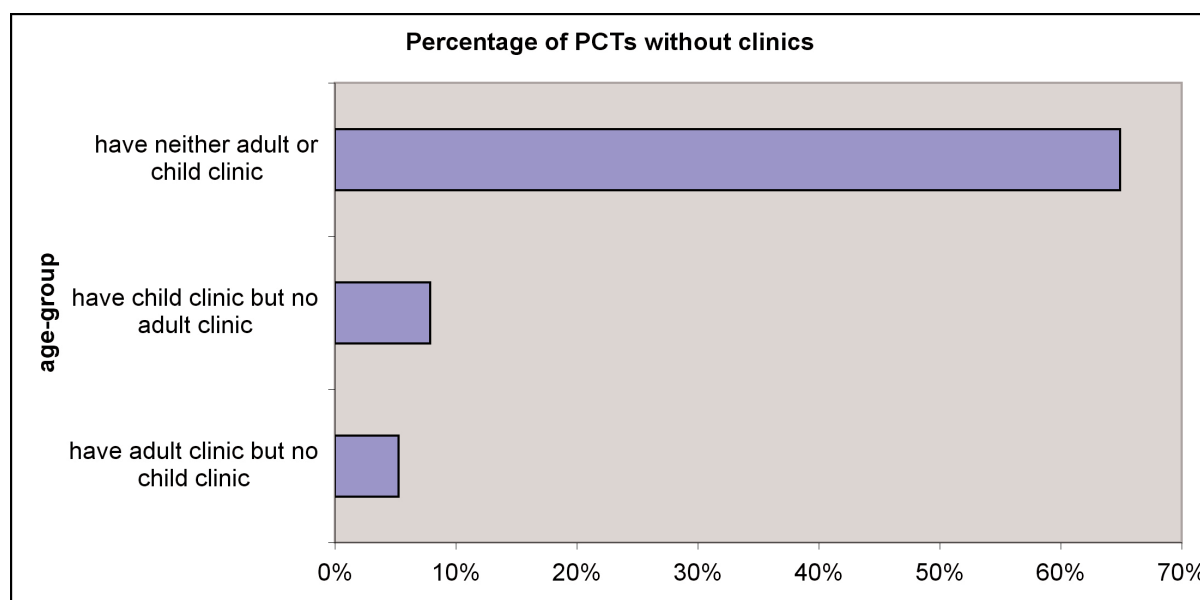
Out of the 114 PCTs that have so far responded, the following picture has emerged:

- 74% of PCTs who responded do not support a muscle clinic that offers a service to adults with neuromuscular conditions.
- 69% of PCTs who responded do not support a muscle clinic that offers a service to children with neuromuscular conditions.
- 65% of PCTs who responded do not support any adult or child muscle clinics within their area.

Services identified

The following table and graph set out the percentage of responding PCTs who commission a muscle clinic.

Age group	Percentage of PCTs without clinics
Have adult clinic but no child clinic	5%
Have child clinic but no adult clinic	8%
Have neither adult or child clinic	65%



It can be said from the evidence submitted by the PCTs that there are limited providers of muscle services around England, with the exception of adult services in the Southwest region where there is no service or lead clinician. However, the degree to which they provide a comprehensive service is highly variable. Further research should be undertaken to identify the clinical workforce at these clinics.

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- ² Specifically the following definitions:
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No. 6 - Specialised Spinal Services (all ages);
No. 8 – Specialised Neurosciences Services (adult)
No. 20 – Medical Genetic Services (all ages);
No. 23 – Specialised Services for Children;
No. 25 – Specialised Pathology Services (all ages);
No. 29 - Specialised Respiratory Services (adult).
- ³ Drachman DB, Toyka KV and Myer E. Prednisone in Duchenne muscular dystrophy. *Lancet* 1974; 2:1409–1412;
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Ramelli GP, Aloysius A, King C, Davis T, Muntoni F. Gastrostomy placement in paediatric patients with neuromuscular disorders: indications and outcome. *Dev Med Child Neurol*. 2007 May;49(5):367-71.
- ⁷ Marguerite E.Hill & Margaret F. Phillips, "Service provision for adults with long-term disability: A review of services for adults with chronic neuromuscular conditions in the United Kingdom," *Neuromuscular Disorders* 16 (2006) 107-112

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