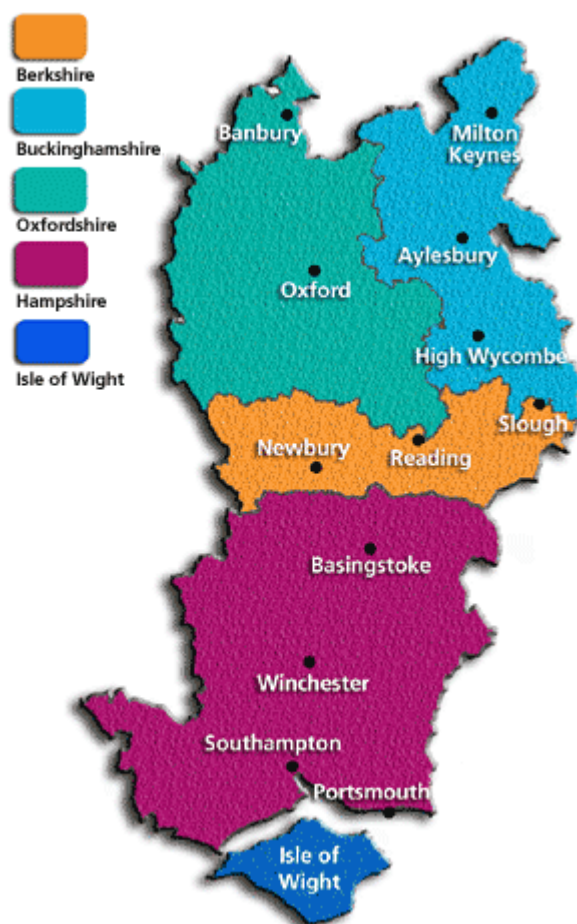


Muscular
Dystrophy
Campaign

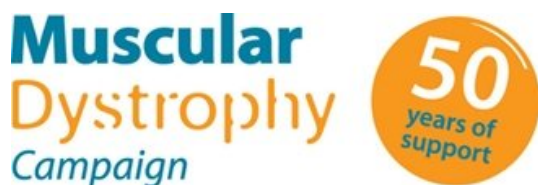


Building on the Foundations: The Need for a Neuromuscular Service Serving Patients in the NHS South Central Region

May 2009



A report by the Muscular Dystrophy Campaign with contributions from and endorsed by the leading neuromuscular clinicians in the South Central region.



Building on the Foundations: The Need for a Neuromuscular Service Serving Patients in the NHS South Central Region May 2009

Executive summary:

This report by the Muscular Dystrophy Campaign draws on the views and experience of leading specialists, patients and their families from across the South Central Strategic Health Authority region to set out a number of serious concerns regarding the provision of specialist clinical services in South Central for patients with muscular dystrophy and related neuromuscular conditions.

We are calling for a major shift in the way services are commissioned in the South Central region in line with the Department of Health's guidance that services for patients with this group of rare conditions should be regarded as specialised and therefore subject to collaborative commissioning arrangements.

The specialised commissioning of these services would be an effective way of delivering care for rare and high cost treatments. The arrangements would provide best value for money and long-term savings for Primary Care Trusts and would ensure fair access to clinically effective, first class, specialised services right across the South Central region.

Action needed:

- **A short life working group should be established to carry out an in-depth review of current service provision and its vulnerability in South Central. This review would involve families, clinicians, PCTs and the SCG, and would bring forward proposals in autumn 2009 to secure and develop the comprehensive, multi-disciplinary service for children and adults, including transition services for young people.**
- **A neuromuscular network should be established on the model of a managed clinical network. This will ensure coordination between the existing specialist centres at Oxford and Southampton and ensure that expertise is shared with all clinicians and AHPs in each PCT in South Central. The network should be supported by a Network Coordinator. This model has been agreed in the South West and has been successful in Scotland with the Scottish Muscle Network. It also reflects the model set out in the National Definition for specialist neuromuscular services.**

- **Three additional full-time Regional Care Advisors with expertise in muscular dystrophy and related neuromuscular conditions should be established and embedded in the NHS to serve the 4,000 people in the area living with these conditions.**
- **Ongoing physiotherapy should be provided to all adults and children with a neuromuscular condition in each PCT, supported and developed in each PCT area by enhanced specialist physiotherapy support from the specialist clinics. The existing specialist neuromuscular physiotherapist role should be funded by and embedded in the NHS. There is currently no paediatric clinic physio input**
- **A new post of adult neuromuscular nurse specialist should be based at the Southampton centre to provide where necessary clinical support for late onset adult conditions that can have a devastating impact on quality of life for many patients.**
- **Resources to be allocated to ensure that vital respiratory and cardiac support is provided for all neuromuscular patients who require it. These are key factors in extending life.**
- **Psychological support should be provided as part of a multi-disciplinary approach to care for individuals and family members living with a neuromuscular condition in each PCT area across South Central.**

Our key findings include:

- **Specialist neuromuscular services are uncoordinated, and vulnerable due to their reliance on charitable funding.**
 - **Specialist clinics at Oxford and Southampton do not have the capacity to serve the 4,000 people living with muscle disease in South Central.**
 - **Services are reliant on one lead consultant in each centre with no cover for holidays, illness or succession planning.**
 - **A number of cases of misdiagnosis of myasthenia gravis were reported, usually where diagnosis was made by a non-specialist. The diagnosis was only corrected when the patient received specialist care at the Oxford Muscle and Nerve Centre**
 - **Almost one in five patients currently have no access to a neuromuscular specialist and receive no specialist care.**
- **There is only one Regional Care Advisor¹ serving the region – funded by the Muscular Dystrophy Campaign. Four full-time Regional Care Advisors are needed to serve the 4,000 people in the area with a neuromuscular condition – 75% of whom currently do not have access to the service.**

- A widespread deficit in provision of services by allied health professionals (AHPs) was also identified with very limited access in particular to ongoing physiotherapy.² Specialist physiotherapists are required to support outreach clinics and provide training and professional development for community physiotherapists. Further, the specialist physiotherapy post at the Oxford Muscle and Nerve Centre only covers adults and is vulnerable due to its dependence on charitable funding.
- There is no dedicated psychology service for neuromuscular patients despite its importance as part of multi-disciplinary care for this patient group with rare and very rare progressive conditions, often genetic in origin and with no known cures and only limited treatments available.
- Families face considerable waits for muscle biopsies to confirm suspected diagnoses at the Southampton paediatric clinic as there is no dedicated neuromuscular theatre time.
- Greater support at transition from paediatric to adult services is needed given the evidence of services being removed or greatly reduced when people leave paediatric services even though needs may well increase given the progressive nature of many conditions.
- There are lengthy waits for patients at Oxford to receive genetic counselling and there are no joint clinics between the genetic and muscle teams.

A report by the Muscular Dystrophy Campaign with contributions from and endorsed by the leading neuromuscular clinicians in the South Central region:

- Dr Simon Hammans, Specialist Neuromuscular Consultant, Southampton General Hospital, Southampton University Hospital Trust
- Dr David Hilton-Jones Specialist Neuromuscular Consultant, Oxford Muscle and Nerve Centre, Oxford Radcliffe Hospitals Trust
- Dr Sandeep Jayawant, Consultant Paediatric Neurologist(Neuromuscular lead),Oxford Children's Hospital, Oxford Radcliffe Hospitals Trust
- Jane Stein Regional Care Advisor, Oxford Muscle and Nerve Centre, Oxford Radcliffe Hospitals Trust

1. Background:

There are over 200 muscular dystrophies and related neuromuscular conditions. They are multi-system disorders, which require complex long-term surveillance and care.

Without specialist multi-disciplinary care most patients and their families experience a further reduction in quality of life. Improved genetic counselling is likely to cause a small reduction in the overall incidence of these conditions but improved survival will increase their prevalence in the adult population.

2. Demographics:

There are some 4,000 people in the South Central region affected by a form of muscular dystrophy or a related neuromuscular condition.

The South Central region has a population of approximately four million people, a figure which is expected to increase by 5% over the next ten years.³ The region's main population centres are Reading and Southampton (populations of 230,000 plus) and Milton Keynes, Oxford, Portsmouth and Slough (populations between 125,000-200,000).⁴ Adult patients from Milton Keynes, Buckinghamshire, Berkshire East and West and Oxfordshire PCTs generally visit Oxford for specialist care, with those from Hampshire, Southampton City and Portsmouth City tending to travel to the specialist clinics in Southampton. Child patients from across the region often travel to London for the specialist care at Great Ormond Street Hospital (this clinic was previously located at Hammersmith Hospital). However, increasingly more children and families are accessing the paediatric neuromuscular clinic in Oxford.

Specialised care in the region is commissioned by the South Central Specialist Commissioning Group - a joint sub-committee of the nine PCTs in the region (Berkshire East, Berkshire West, Buckinghamshire, Hampshire, Isle of Wight, Milton Keynes, Portsmouth and Southampton). It is managed by Hampshire PCT and has a commissioning budget for 2008/09 in excess of £300 million.⁵

3. Current level of essential, specialist provision in South Central:

People living with severe disabling and/or life limiting neuromuscular conditions need access to the appropriate interventions and support as their condition progresses. Essential, specialist services should be delivered by a range of professionals from local, regional and national service providers. A neuromuscular Regional Care Advisor is essential to provide vital specialist care, support and advice for each individual and family living with one of these conditions.

Specialist multi-disciplinary care can improve quality of life and extend life expectancy. For example, without treatment, the mean age of death in Duchenne Muscular Dystrophy is 19 years.⁶ With specialist care and home ventilation life expectancy is raised to almost 30 years.⁷

Coordinated and comprehensive multi-disciplinary specialist care should include a neuromuscular specialist consultant and, dependent on medical need, may also include specialist cardiac, respiratory and orthopaedic care.⁸ Genetic counselling and psychological services should also be offered, together with locality based dietetic, occupational therapy, physiotherapy and speech therapy provision which can both improve the quality of these patients' lives and increase their life spans. Boys with Duchenne muscular dystrophy who are still ambulant should be offered the opportunity to discuss treatment with steroids such as deflazacort which studies have shown can stabilise muscle strength and delay the loss of ambulation and may also delay the onset of breathing complications (see appendix 4). For a number of neuromuscular conditions, regular check ups are required irrespective of symptomatology, because deterioration can advance rapidly over the course of months.⁹

o **Specialist muscle clinics:**

Patients currently face an unacceptable regional variation in the provision of specialist neuromuscular care, caused by the inadequate provision of specialist muscle clinics. Only a third of the Primary Care Trusts in the region fund a muscle clinic for children or adults: (children: Southampton, Hampshire and Oxfordshire, adults: Hampshire, Milton Keynes and Oxfordshire).

Patients can have round trips of up to 160 miles to access specialist care within the region. Slightly over a third (34%) of respondents to the Muscular Dystrophy Campaign (MDC) 2008 Patient Survey travel to John Radcliffe Hospital in Oxford, with 8% attending the muscle clinic at Southampton, and 3% at Milton Keynes. However 26% travel outside the South Central region to access care in London - primarily children travelling to Great Ormond Street Hospital for specialist treatment. This information is backed up by the evidence provided by the Muscular Dystrophy Campaign's Regional Care Advisor at GOSH, who sees 54 families from the region, with the majority from Berkshire. Patients also travel to the specialist centres at Oswestry and Newcastle.

In addition, almost one-fifth of respondents to the MDC Patient Survey do not receive any specialist care: two thirds of these are people over the age of 60, with the remaining third aged over 30, who have generally either not received a clear diagnosis, or were diagnosed several years ago, and told that there was no treatment or care available for their condition. Many elderly or severely disabled patients have difficulties travelling to clinics.

a. **Oxford Muscle and Nerve Centre:**

The Muscular Dystrophy Campaign provides funding for the Oxford Muscle and Nerve Centre hosted at John Radcliffe Hospital. It provides a comprehensive clinical service for patients with neuromuscular conditions and exemplifies the value of linking such a service to research. Patients provide the data and material for clinical and laboratory research, while research advances rapidly benefit patients. 1000 adults and 200 children currently attend clinics at Oxford, a figure which is increasing annually. For example, the increase in paediatric patients has led to a quadrupling in the frequency of clinics from once a month in 2002 to four or five clinics a month currently. The Oxford Centre sees patients South Central region, and also Northamptonshire, and Wiltshire. Some families also travel from Warwickshire, Hampshire and Gloucestershire, and parts of the South West to Oxford for treatment.

The centre has a multi-disciplinary approach led by:

- Clinical Director and Consultant Neurologist (Adults) Dr David Hilton-Jones
- Consultant Paediatric Neurologist Dr Sandeep Jayawant,
- Regional Care Advisor, Jane Stein
- Specialist Neuromuscular Physiotherapist, Jane Freebody.

Three adult neuromuscular clinics are held at Oxford each week with a paediatric neuromuscular clinic four to five times a month. Transition services are in place for the move from paediatrics to adult care. Respiratory and cardiac services are available on site for paediatrics and a mile away for adult patients. Spinal and general paediatric orthopaedic care is provided at the Nuffield Orthopaedic Centre (NOC). Joint clinics are held twice a year for children with orthopaedic problems that need monitoring or treatment. There is also access available to orthotist services from the NOC. For myasthenia gravis patients there is a specialist nurse available who is funded jointly between the Muscular Dystrophy Campaign and the Myasthenia Gravis Association. A specialist NSCAG clinic is also located in Oxford which is the designated national referral centre for Congenital Myasthenia.

There is no specialist neuromuscular paediatric physiotherapist, and only one part-time physiotherapist for adults. As the physiotherapist is only part-time, there is not the capacity to meet the needs of the adults with these conditions and currently she is unable to even attend all of the neuromuscular clinics provided. There is no clinical psychologist available, and no occupational therapist is present at the clinics. There is some orthotic support to the paediatric clinic but not all clinics are covered due to inadequate resources.

The absence of a paediatric physiotherapist in Oxford has meant that there is no tertiary centre physiotherapy assessment and advice to families and referral centre teams. It has also precluded Oxford from joining in with nationally coordinated standardized assessments for children with neuromuscular problems such as the North Star and SMART projects. In the future as clinical trials progress, this will severely compromise the ability to participate in trials and offer standardized follow-up and assessment.

The Oxford centre previously hosted joint clinics with the genetics team. This arrangement worked well, and was valued by patients, but ended when the genetics consultant post changed, with no succession planning. Families are now forced to wait several weeks to obtain an appointment for genetic counselling.

Dr Hilton-Jones also travels to Milton Keynes to host a monthly muscle clinic for adults in the region. This clinic is also supported by the Regional Care Advisor and the specialist adult neuromuscular physiotherapist.

The clinics at Oxford are vulnerable due to the reliance on funding by the Muscular Dystrophy Campaign. For example, the posts of regional care advisor and specialist adult neuromuscular physiotherapist are funded entirely by the Muscular Dystrophy Campaign. To ensure the service is continued and developed in the long-term, these services should be embedded in the NHS. There is an urgent need to fund a paediatric physiotherapy post.

b. Southampton and Hampshire:

Paediatric:

A paediatric muscle clinic was established in 1993 by Dr Thomas at Southampton General Hospital, hosted alternatively by Consultant Paediatric Neurologists, Dr Thomas and Dr Goodwin, and attended by a specialist neurology nurse. The clinic is a regional muscle unit which takes referrals from across Hampshire, Surrey, Dorset, and Wiltshire and in each monthly session there two slots for new patients, and five follow-up slots. There are good links with the paediatric cardiac clinic on the same site, which patients can be referred on to and with orthopaedic, spinal, and genetic services.

As only one clinic is held a month, waiting lists are considerable – for example families are currently forced to wait four months for an urgent appointment to discuss steroid treatment. Due to the limited capacity of the clinic a further 25% of neuromuscular patients are seen in regional outreach clinics, and then brought to Southampton for specific aspects of care during the course of their condition. However, the outreach clinics are general neurology clinics, and not specialist neuromuscular clinics. Patients therefore do not have multi-disciplinary care, and may not always be seen by a neuromuscular consultant.

There can also be lengthy delays in securing a confirmed diagnosis. Muscle biopsies are carried out on the same hospital site, but require general anaesthetic. The neuromuscular clinic is not allocated theatre time to carry out these biopsies and instead has to borrow capacity from the paediatric oncology department. However, this is vulnerable to the workload of the oncology department and clearly unsustainable. To arrange a muscle biopsy it is necessary to obtain an available theatre slot with the oncology department which also coordinates with the diaries of the anaesthetist and the neuromuscular consultant Dr Thomas (who is the only clinician at the clinic able to perform the procedure). This is exacerbated by the fact that Dr Thomas' neuromuscular role is only part-time, and that there is no cover provided when he takes medical or holiday leave. Once the biopsy has taken place there is a six week wait for the results to be returned from the neuro-pathology department. A further delay is then caused by the previously mentioned high demand for clinic slots in which to discuss the results with the parents.

There is no dedicated psychological provision for neuromuscular patients, and the team have proposed a 0.5 whole time equivalent (WTE) psychologist to assist patients with the difficulties of coming to terms with diagnosis, transition and end of life issues.

There is also no provision for non-invasive ventilation on the Southampton site which means that patients requiring this treatment currently are forced to travel to the Royal Brompton hospital in London. While a business case to provide non-invasive ventilation at Southampton has been proposed by the respiratory department, and agreed, it has not yet been implemented.

There is an urgent need for extra physiotherapy support for the clinic. At present the team report that many physiotherapy appointments are double-booked (as they are for siblings). To resolve this, two physiotherapists are required to attend each clinic. Additional physiotherapy time is also necessary for the three to six patients annually admitted for intensive rehabilitation, when going into knee-ankle-foot orthoses. In

addition, there are two or three ad hoc assessments between clinics per month. To achieve this, a further 0.5 WTE equivalent senior physiotherapist is required.

Occupational therapy input is currently provided at the clinic, but requires a further 0.5 WTE specialist neuromuscular occupational therapist to provide advice to patients and their families at clinic and to liaise with schools and the local medical teams.

There is no care advisor serving the clinic – nor at the adult clinic (below). Two WTE Regional Care Advisors are required to serve the approximately 1800 children and adults with a neuromuscular condition in Southampton, Portsmouth, Hampshire and Isle of Wight PCTs.

The paediatric team recommends that to meet current demand, and to bring all paediatric neuromuscular patients into a specialist clinic setting, a three times monthly clinic would be necessary. The bulk of these clinics would be provided at Southampton, but one option to consider would be the provision of specialist clinics locally, for example, two or three a year at Portsmouth and two at Poole, with the multi-disciplinary team travelling to deliver this clinic. A greater amount of consultant time is required to meet this need, with a 0.5 WTE consultant proposed.

Adults:

There is also a weekly adult muscle clinic held in Southampton by Dr Simon Hamman, a Consultant Neurologist who sees 6 patients per week (2-3 new, 3-4 old). The clinic was established in 1994 and sees patients from across Hampshire and the Isle of Wight, and also a large proportion from the Channel Islands, and some from the South West SHA region. The service also requires an increase in capacity to meet current demand.

A joint clinic is held monthly with the genetics team, and there are muscle pathology services available. There is a link to the respiratory support service provided by Dr Jane Wilkinson also at Southampton General Hospital for patients requiring non-invasive ventilation.

The need for a Regional Care Advisor to support the clinic, particularly at diagnosis for adult onset patients has been identified. In addition to further smooth the diagnosis pathway, greater administrative support is required to ensure that patients receive as speedy a diagnosis as possible.

Further resources are also needed to ensure multi-disciplinary care with provision of specialist physiotherapists, speech and language therapists, and specialist nurses. There are strong links with Caroline Hutchings, Consultant in Rehabilitation Medicine in Southampton, but greater support is required in this area in other parts of the region.

o Regional Care Advisors

Regional Care Advisors play an essential role in supporting individuals with muscular dystrophy and related conditions. They successfully coordinate their health and social care needs, provide support and information to families and help to ensure a seamless transition from child to adult services.

The region has only one Regional Care Advisor, Jane Stein, who is based at the John Radcliffe Hospital, Oxford. The service is highly valued with 88% of the people who have access to the Regional Care Advisor describing the service as excellent or good. However **three-quarters of patients** in the region do not receive the care and support of a Regional Care Advisor. An extra three WTE posts (each with a workload of 1,000 patients) are needed to serve the estimated 4,000 people with muscular dystrophy and related conditions in the region.

The South Central SHA identifies the provision of a named care advisor for people with long-term conditions as one of twelve key ambitions in their ten year healthcare vision document Towards a Healthier Future, published in May 2008.¹⁰

“Service provision is complex for patients; it can be difficult to access and work through the care system, leaving patients feeling confused and unsupported. Continuity of care for patients is important, particularly for those with long-term conditions. A named clinician should be a central point of contact.”

“**Ambition 8:** We will ensure that patients have access to services through a single point and that they are not left alone to manage their care; they will have a key professional assigned to coordinate their care and to help them navigate seamlessly through the care system.”

The provision of a named Regional Care Advisor is also stated as an aim by Health Minister Lord Darzi in his final report High Quality Care for All which set out how the Government intends to provide this more personalised level of care for people with long-term conditions.¹¹ In addition, the need for a Regional Care Advisor was highlighted in the Parker *et al* study of Duchenne patients at the Lane Fox Unit (2005) which noted: “Most patients received full provision of disability allowances, but full access to social services provision was inadequate, and often depended on the input of the muscular dystrophy key worker.”¹²

The results of the Muscular Dystrophy Campaign’s Patient Survey highlighted the need for an increase in Regional Care Advisors with over half of patients reporting that they are not satisfied with the level of emotional support available to their families and to themselves. Furthermore, only a third of patients are satisfied with the amount and clarity of information available to them.

Case study:

- *Mr C from Buckinghamshire had to give up work due to his FSH Muscular Dystrophy which severely reduced his income. However he is pleased with the care he receives, saying: “both my consultant and Care Advisor are available at all times. I find their care to be of great comfort.”*

o **Diagnosis experience:**

Two-thirds of patients in the area describe their experience of the diagnosis process as either poor or very poor, with many calling for greater information and support to be given to parents and families after diagnosis.¹³ This reflects the lack of a Regional Care Advisor for patients and families.

The lack of local specialists in the region can cause delays in starting treatment, and incorrect diagnosis, which can then lead to the patient being given dangerously incorrect advice regarding the management of their condition. This can be due to a lack of knowledge among GPs of these rare conditions, suggesting a need for greater education of the early symptoms of neuromuscular conditions.¹⁴

Case studies:

- *The mum of a baby boy from Buckinghamshire who was diagnosed with Ullrich Congenital Muscular Dystrophy in March 2008 describes the care and service her family has received during and since her son's diagnosis as "appalling". She says: "This has been the worst thing that has ever happened to us and we have been left alone to deal with it by ourselves. We feel incredibly let down. I have had to phone and literally fight for appointments and information."*
- *Mrs L, from Hampshire has a young son who was diagnosed with Becker Muscular Dystrophy in the last five years. She says: "More information is definitely needed. When my son was diagnosed we went home with no information or help. It was up to us to look into his condition and find out how we could help him."*
- *Mr R from Hampshire felt that his young son's diagnosis was poor: "We were told that my son had Duchenne Muscular Dystrophy – 'see you in 6 months and here's a leaflet on steroids'. All this after waiting 9 weeks for muscle biopsy results."*

The Oxford clinic has reported a number of cases where patients have been mistakenly diagnosed locally with Myasthenia Gravis, a rare auto-immune disorder. Once referred to the specialist clinic at Oxford it is found that they do not have the condition. Incorrect diagnoses such as this can cause huge unnecessary stress on families, and strain already stretched resources.

○ **Respiratory clinics:**

Breathing disorders are recognised as the leading cause of mortality in neuromuscular disease.¹⁵ Respiratory muscle weakness is relatively common in most neuromuscular conditions and is almost inevitable in the late stages of Duchenne muscular dystrophy.¹⁶ However treatment, including ventilation, has been shown to improve both quality and length of life.¹⁷

An audit of 40 sequential DMD deaths over 10 years in the South West 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).¹⁸

Regular comprehensive check ups are required with clinicians being instructed to go through a full checklist of signs and symptoms. A study published in 2002 highlighted patients can become too accustomed to their chronic illness and therefore rarely raise complaints about respiratory distress spontaneously.¹⁹

Evidence from a 2003 study highlighted that it is more cost-effective to manage respiratory issues through check ups and home ventilation than through unplanned critical hospital admissions.²⁰

o **Cardiac clinics:**

As a number of neuromuscular conditions also affect the heart, cardiac monitoring should be part of a multi-disciplinary approach to care. The heart is affected in different ways – people affected by myotonic dystrophy and Emery-Dreifuss dystrophy are prone to abnormal heart rhythms, while cardiomyopathy is more likely for people affected by Duchenne or Becker muscular dystrophy.

Regular cardiac screenings are crucial even for conditions which appear to cause less severe weakening of the muscles, as “the severity of cardiomyopathy may be out of proportion to that of skeletal muscle involvement.”²¹ As an example of the frequency required for cardiac screenings, best practice guidelines for Duchenne muscular dystrophy recommend that they should take place before any surgery, every two years up to the age of 10 and annually after age 10.²² Without screening, cardiomyopathy can progress almost entirely without symptoms until signs of heart failure emerge, when all cardiac reserve has been eroded.²³

Cardiac screening should also be offered to women who are carriers of mutations in the dystrophin gene, who are at increased risk of cardiomyopathy, even if they experience no symptoms. Currently this only occurs on an ad hoc basis, if at all.

o **Physiotherapy:**

It is accepted that all patients with a neuromuscular condition will at some point during the course of their condition require access to ongoing and timely physiotherapy.²⁴ Physiotherapy is the physical treatment and management of a condition which enables people with neuromuscular conditions to reach their maximum physical potential by maintaining mobility, independence and improving quality of life. This should be provided by a specialist physiotherapist, who has skills in both neurological and musculoskeletal physiotherapy, experience in treating muscle conditions and the confidence to treat patients with rare disorders.²⁵ Specialist physiotherapy can delay the progression of the condition, reduce pain and minimise emergency hospital admissions.

In April 2008, the Muscular Dystrophy Campaign carried out a Freedom of Information request to all NHS Trusts and Primary Care Trusts across England about the provision of physiotherapy services. All nine PCTs and ten NHS Trusts in South Central responded – key findings include:

- Half of the PCTs and NHS Trusts in South Central do not provide ongoing physiotherapy for patients with muscular dystrophy and related conditions where required;
- Half of all Trusts and a third of PCTs in South Central do not have physiotherapists available to children or adults with specific training in muscular dystrophy and related neuromuscular conditions;

This correlates with the results of the Muscular Dystrophy Campaign Patient Survey in which 44% of respondents reported that they had no access to physiotherapy. More

children than adults received physiotherapy, often due to provision at their special school. However, this provision is then removed when the child leaves school or moves from paediatrics to adult services (see Transition section below).

Case study:

- *Mr Y from Berkshire has Becker Muscular Dystrophy, and has received no physiotherapy since turning eighteen. He says “as a child there were more facilities available. When I became an adult, there weren’t any!”*

The provision of physiotherapy in short blocks of sessions is problematic for patients and indicates a clinical focus on conditions in which quantifiable improvement can be measured, rather than the maintenance of chronic and progressive conditions. For example, Milton Keynes NHS General Hospital Trust provides physio in six week blocks with patients to be referred back for more treatment.

Case study:

- *Mrs W from Buckinghamshire: Mrs W receives weekly physiotherapy sessions for her Limb Girdle Muscular Dystrophy, but has to stop after 12 weeks, for a 12 week break. She says: “I struggle when I return to physio after a break. I just start to cope with the exercises when I have to have a break again.”*

o **Orthopaedic care**

Spinal deformity, such as scoliosis is common in many neuromuscular conditions, with 90% of people affected by Duchenne Muscular Dystrophy for example, likely to develop a clinically significant scoliosis.²⁶

Surgery to correct spinal deformity can prolong life and improve posture and comfort. It is imperative that the development of scoliosis is monitored by the specialist muscle clinic as success rates are likely to be highest and complication rates lowest if surgery is performed when the spine is still mobile at a Cobb angle of 20-40°.²⁷ As it is a major operative procedure, a multi-disciplinary approach, involving the paediatrician/paediatric neurologists and orthopaedic surgeons is essential in the approach to surgery.²⁸

As an example, the best practice guidelines for patients with Spinal Muscular Atrophy state that evaluation should take place every 3-6 months, and more frequently in clinically unstable non-sitters. The evaluation should include, depending on clinical need: inspection of the spine, chest x-rays and radiographic evaluations of scoliosis, swallow studies, pulse oximetry and polysomnography.²⁹

o **Rehabilitation and equipment:**

Specialist neuromuscular rehabilitation clinics aim to help maintain independence or in adapt to changes which affect social and domestic life and can include a number of services including physiotherapy, access to communication and controls, occupational therapy, speech and language therapy, wheelchair services and orthotics. Rehabilitation care can improve quality of life and delay progression of the condition. For example, poorly fitting knee-ankle-foot orthoses can severely compromise mobility and successful care. To avoid this orthotists with specific experience in neuromuscular disorders should be used to measure and supply orthotics.³⁰

A number of children and adults with neuromuscular conditions are considered to have profound disabilities where the assessment process requires greater knowledge and expertise than is often available in local wheelchair services. The Muscular Dystrophy Campaign September 2008 Patient Survey revealed that a number of people are not being properly assessed or being offered appropriate equipment.

Currently, as PCTs do not collaborate to provide specialist wheelchair services, children and adults affected by these rare and progressive conditions are competing for equipment with patients who have acute episodes, for example a leg fracture, and are often being forced to wait for long periods for essential equipment. For example, Freedom of Information requests have shown that disabled children in South Central wait on average 16 weeks to receive a powered chair, with waits of up to 26 weeks in Berkshire and 32 weeks in the Isle of Wight (see appendix 5). According to government targets, patients should have a maximum wait of 18 weeks from referral for equipment provision.

o **Psychologists:**

Psychology support has been identified as an important aspect of multi-disciplinary care, and as a key part of rehabilitation services.³¹ There is pressing need to develop clinical and educational psychology input and support for this patient group.

Children and adults with neuromuscular conditions, including Duchenne muscular dystrophy, myotonic dystrophy and congenital myotonic dystrophy, would particularly benefit from the input of a clinical psychologist to help families develop management strategies. Specific issues for patients with muscular dystrophy and related neuromuscular conditions include support at the time of diagnosis, chronic illness, loss of ambulation, transition to adulthood, times of crisis and bereavement.

Studies have shown that the incidence of autistic spectrum disorders, attention deficit hyperactivity disorders and obsessive compulsive disorders is higher in males affected by Duchenne muscular dystrophy.³² In addition behavioural changes have been shown to be an adverse side effect of treatment with corticosteroids – which is used to prolong ambulation and preserve muscle strength and respiratory function.³³ Early input from a clinical psychologist may help parents develop strategies with which to manage these behavioural difficulties and thus prevent the need to withdraw steroid treatment.

o **Transition**

Increasing numbers of young people with complex conditions are reaching transition and living longer because of improvements in therapies and medical care. For young people living with muscle disease, the period between mid and late teens is crucial and the transition from paediatric and adolescent care into adult-oriented healthcare services must be as smooth as possible.³⁴

However, despite the significance of this period for younger people with these progressive neuromuscular conditions, the majority do not have access to a Regional Care Advisor who can support their transition to adulthood.

The difficulties are shown by respondents to the MDC Patient Survey, a quarter of who describe the transition process as 'poor' or 'very poor' (only 14% rated the process as good or excellent, with the question not being applicable to the rest of respondents).

Case studies:

- *Mr P from Reading: "I was on child ward/outpatients until the age of 32."*
 - *Mr I from Hampshire: "I was abandoned by the neurologist for over 10 years on entry into adult services."*
 - *Mr D from Berkshire: "Once you leave school you seem to fall into a grey area where no body seems to know what is going to happen or does not seem to want to take responsibility for you."*
-

Appendix 1:

South Central Demographics:

South Central PCT populations

Population data taken from PCT websites/annual reports

PCT	Resident population	Prevalence of neuromuscular conditions	Prevalence of muscular dystrophy	2009-10 PCT Revenue Allocations (£ thousands)
Berkshire East	376500	377	188	560,009
Berkshire West	500000	500	250	627,760
Buckinghamshire	500000	500	250	685,650
Hampshire	1250000	1250	625	1,799,471
Isle of Wight	138500	139	69	245,882
Milton Keynes	230000	230	115	338,522
Oxfordshire	592329	592	296	873,673
Portsmouth	190000	190	95	328,095
Southampton	257000	257	129	388,555
<i>Totals</i>	4034329	4035	2017	5,847,617

Appendix 2:

Background to report:

The report contains:

- Evidence from the leading neuromuscular clinicians working in the South Central area.

- Information from the latest research papers on the impact of specialist services on those affected by muscular dystrophy and related neuromuscular conditions.

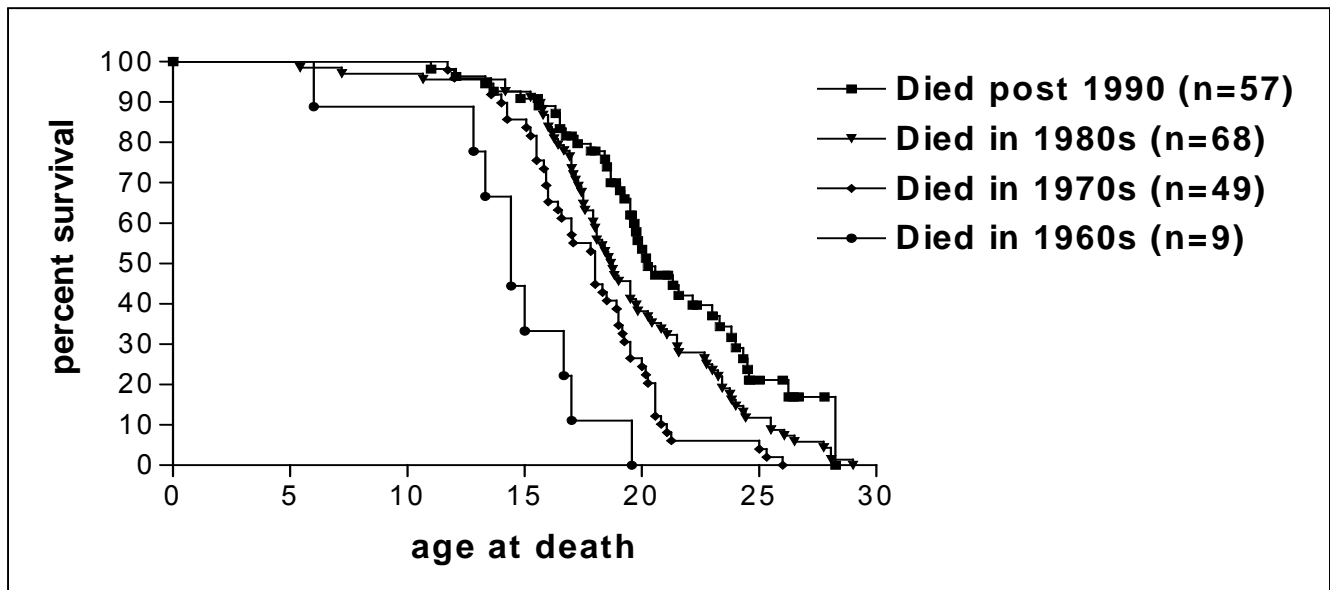
- Data from the responses to the largest nationwide survey of people affected by muscular dystrophy and related conditions, published in September 2008 by the Muscular Dystrophy Campaign. 850 people completed the survey from across the UK – including 62 families from South Central.

- The responses to Freedom of Information requests to all Primary Care Trusts and Acute Trusts regarding specialist services,

Appendix 3:

Duchenne Muscular Dystrophy Survival data 1960-1990

(Eagle et al *Survival in Duchenne muscular dystrophy: improvements in life expectancy since 1967 and the impact of home nocturnal ventilation*³⁵)



The authors reviewed the notes of 197 patients with Duchenne muscular dystrophy whose treatment was managed at the Newcastle muscle centre from 1967 to 2002, to determine whether survival has improved over the decades and whether the impact of nocturnal ventilation altered the pattern of survival.

Results:

1960s: Mean life expectancy: 14.4 years - No survivors beyond 19.29 years

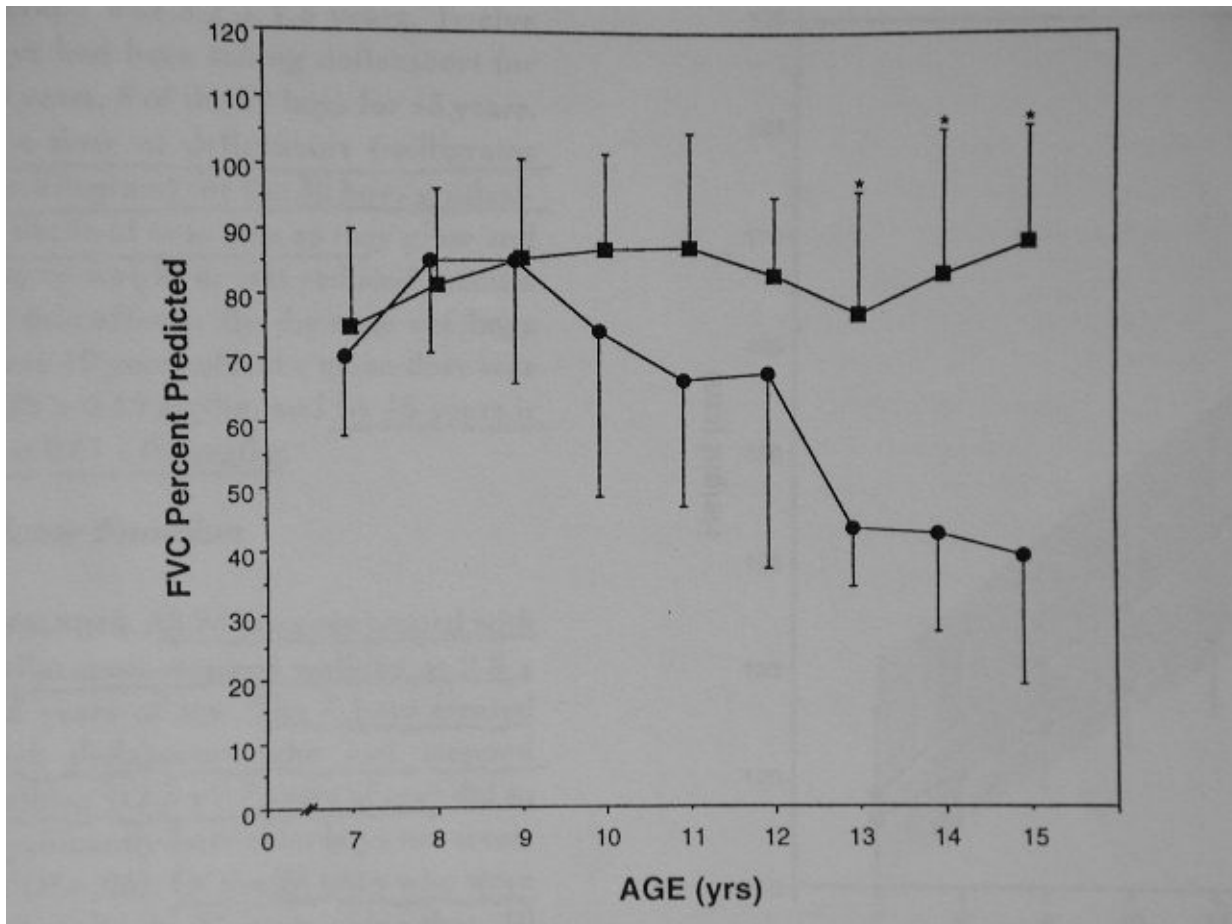
1990s: Mean life expectancy: 19.5 years

Improvement is due to multi-disciplinary care

Appendix 4:

Preserved lung function

(from Biggar WD, Harris VA, Eliasoph L, Alman B. Long-term benefits of deflazacort treatment for boys with Duchenne muscular dystrophy in their second decade. *Neuromuscular Disorders*)³⁶



The article compares the clinical course of 74 boys 10-18 years of age with Duchenne muscular dystrophy (DMD) treated (40) and not treated (34) with deflazacort.

Results for lung function:

- Deflazacort group: 88% (\pm 18%)
- No treatment Group 39% (\pm 20%)

References:

- ¹ Care Advisors may also be known as key workers or neuromuscular care coordinators. They carry out the role envisaged by Lord Darzi in his 2008 High Quality Care for All report.
- ² Muscular Dystrophy Campaign Patient Survey – State of the Nation, September 2008
- ³ South Central SHA Towards a Healthier Future (May 2008)
- ⁴ National Statistics <http://www.statistics.gov.uk/statbase/Product.asp?vlnk=15106> Accessed Jan '09.
- ⁵ South Central SCG http://www.nscteam.org.uk/3a06_south_central.htm Accessed Feb 2009.
- ⁶ Eagle M, Bourke J, Bullock R, Gibson M, Straub V and Bushby K. (2007) 'Managing Duchenne muscular dystrophy – The additive effect of spinal surgery and home nocturnal ventilation in improving survival' Neuromuscular Disorders Volume 17, Issue 6, p.470-475
- ⁷ Bushby, K. Bourke, J. Bullock, R. Eagle, M. Gibson, M. Quinby, J. 'The Multidisciplinary management of Duchenne muscular dystrophy' Current Paediatrics (2005) 15. 292-300. p. 294.
- ⁸ Bushby, K. Bourke, J. Bullock, R. Eagle, M. Gibson, M. Quinby, J. 'The Multidisciplinary management of Duchenne muscular dystrophy' Current Paediatrics (2005) 15. 292-300. p. 292-293.
- ⁹ A.E. Parker, S.A. Robb, J. Chambers, A.C. Davidson, K. Evans, J. O'Dowd, A.J. Williams and R.S. Howard. Analysis of an adult Duchenne muscular dystrophy population p. 730
- ¹⁰ South Central SHA Towards a Healthier Future (May 2008)
- ¹¹ Department of Health High Quality Care for All (June 2008): http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_085825
- ¹² A.E. Parker, S.A. Robb, J. Chambers, A.C. Davidson, K. Evans, J. O'Dowd, A.J. Williams and R.S. Howard. Analysis of an adult Duchenne muscular dystrophy population p. 734.
- ¹³ Muscular Dystrophy Campaign Patient Survey – State of the Nation, September 2008
- ¹⁴ Heap, R. Mander, M. Bond, J. Bushby, K. (1996) 'Management of Duchenne Muscular Dystrophy in the Community: Views of Physiotherapists, GPs and School Teachers' Physiotherapy Vol. 82. No. 4. p. 258-263. p. 262.
- ¹⁵ Benditt J O. Management of pulmonary complications in neuromuscular disease. Physical Medicine and Rehabilitation Clinics of North America 1998; 9(1): 167-185
- ¹⁶ A.E. Parker, S.A. Robb, J. Chambers, A.C. Davidson, K. Evans, J. O'Dowd, A.J. Williams and R.S. Howard. Analysis of an adult Duchenne muscular dystrophy population p. 734.
- ¹⁷ Bushby, K. Bourke, J. Bullock, R. Eagle, M. Gibson, M. Quinby, J. 'The Multidisciplinary management of Duchenne muscular dystrophy' Current Paediatrics (2005) 15. 292-300. p. 294.
- ¹⁸ Eagle M, Bourke J, Bullock R, Gibson M, Straub V and Bushby K. (2007) 'Managing Duchenne muscular dystrophy – The additive effect of spinal surgery and home nocturnal ventilation in improving survival' Neuromuscular Disorders Volume 17, Issue 6, p.470-475.,
- ¹⁹ Birnkrant , D.J. 'The Assessment and Management of the Respiratory Complications of Pediatric Neuromuscular Diseases' Clinical Pediatrics (2002) Vol. 41. No. 301 p. 302-303.
- ²⁰ Howard R.S and Davidson, C. Long Term Ventilation in Neurogenic Respiratory Failure J Neurol Neurosurg Psychiatry 2003;74(Suppl III):iii24–iii30 p. iii30.
- ²¹ Bushby, K. Muntoni, F. Bourke, J.P. Workshop Report: 107th ENMC International Workshop: the management of cardiac involvement in muscular dystrophy and myotonic dystrophy. 7th–9th June 2002, Naarden, the Netherlands Neuromuscular Disorders 13 (2003) 166–172 p. 170.
- ²² Bushby, K. Muntoni, F. Bourke, J.P. Workshop Report: 107th ENMC International Workshop: the management of cardiac involvement in muscular dystrophy and myotonic dystrophy. 7th–9th June 2002, Naarden, the Netherlands Neuromuscular Disorders 13 (2003) 166–172 p. 166.
- ²³ Bushby, K. Bourke, J. Bullock, R. Eagle, M. Gibson, M. Quinby, J. 'The Multidisciplinary management of Duchenne muscular dystrophy' Current Paediatrics (2005) 15. 292-300. p.298.
- ²⁵ Hill, M.E. Phillips, M.F. '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. p.110-111.
- ²⁶ Bushby, K. Bourke, J. Bullock, R. Eagle, M. Gibson, M. Quinby, J. 'The Multidisciplinary management of Duchenne muscular dystrophy' Current Paediatrics (2005) 15. 292-300. p. 295.

-
- ²⁷ Bushby, K. Bourke, J. Bullock, R. Eagle, M. Gibson, M. Quinby, J. 'The Multidisciplinary management of Duchenne muscular dystrophy' Current Paediatrics (2005) 15. 292-300. p.295.
- ²⁸ Muntoni, F. Bushby, K. Manzur, A. 'Workshop report: Muscular Dystrophy Campaign Funded Workshop on Management of Scoliosis in Duchenne Muscular Dystrophy 24 January 2005, London, UK' Neuromuscular Disorders 16 (2006) 210–219 p. 216.
- ²⁹ Ching H. Wang, et al. 'Consensus Statement for Standard of Care in Spinal Muscular Atrophy' Journal of Child Neurology Volume 22 Number 8 August 2007 1027-1049 p. 1033.
- ³⁰ Eagle, M. 'Report on the muscular Dystrophy Campaign workshop: Exercise in Neuromuscular diseases, Newcastle January 2002' Neuromuscular Disorders 12, (2002) 975-983 p. 978.
- ³¹ Nätterlund, B. and Ahlström, G. 'Activities Of Daily Living And Quality Of Life In Persons With Muscular Dystrophy' Rehab Med 2001; 33: 206–211.
- ³² Hendriksen, J.G.M. and Vles, J.S.H. 'Neuropsychiatric Disorders in Males With Duchenne Muscular Dystrophy: Frequency Rate of Attention-Deficit Hyperactivity Disorder (ADHD), Autism Spectrum Disorder, and Obsessive–Compulsive Disorder' Journal of Child Neurology 2008 May; 23(5): p. 477-81.
- ³³ Manzur, AY. Kuntzer, T. Pike, M. Swan, AV. Glucocorticoid corticosteroids for Duchenne muscular dystrophy (Cochrane Review) The Cochrane Library 2009, Issue 1.
- ³⁴ Hill, M.E. Phillips, M.F. '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. p.110.
- ³⁵ Eagle et al Survival in Duchenne muscular dystrophy: improvements in life expectancy since 1967 and the impact of home nocturnal ventilation Neuromuscular Disorders, Volume 12, Issue 10, p. 926.
- ³⁶ Biggar WD, Harris VA, Eliasoph L, Alman B. Long-term benefits of deflazacort treatment for boys with Duchenne muscular dystrophy in their second decade. Neuromuscul Disord 2006;16:249-5
-