

Building on the Foundations: the need for a neuromuscular service across the West Midlands

April 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 West Midlands Strategic Health Authority region to set out a number of serious concerns regarding the provision of specialist clinical services in the West Midlands for patients with muscular dystrophy and related neuromuscular conditions.

Services for the 5,000 patients affected by muscular dystrophy and related neuromuscular conditions in the region are patchy and vulnerable due to their reliance on charitable funding. We have uncovered evidence of errors in diagnosis, poor patient outcomes and major gaps in the provision of physiotherapy and speech and language therapy, as well as diagnostic services (including pathology).

We are calling for a major shift in the way services are commissioned in the West Midlands 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 will ensure fair access to clinically effective, first class, specialised services right across the West Midlands.

This report contains evidence from Freedom of Information requests to all Primary Care Trusts and Acute Trusts in the West Midlands, as well as information from the latest clinical research papers on the impact of specialist services on those affected by muscular dystrophy and related neuromuscular conditions.

The report also contains data from the responses to the largest nationwide survey undertaken of people affected by muscular dystrophy and related neuromuscular conditions, published in September 2008 by the Muscular Dystrophy Campaign.¹

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

- Dr Martin Allen Consultant in Respiratory Medicine, University Hospital of North Staffordshire
- Dr Dev Banerjee Consultant Respiratory Physician, Birmingham Heartlands Hospital
- Dr Ros Quinlivan Consultant Paediatrician with a special interest in neuromuscular disorders, The Robert Jones and Agnes Hunt Orthopaedic and District Hospital, Oswestry
- Dr Helen Roper, Consultant Neurologist specialising in muscle, Birmingham Children's Hospital
- Dr Martin Samuels Consultant Paediatrician, University Hospital of North Staffordshire
- Dr Martina Walsh Consultant in Rehabilitation Medicine, West Midlands Neuromuscular Specialist Clinic
- Dr John Winer Consultant Neurologist specialising in muscle, University Hospital Birmingham

Action needed

- We are calling on the West Midlands SCG and all West Midlands PCTs to carry out a comprehensive review of the provision of specialist, multi-disciplinary care for people with neuromuscular conditions living in the region. A similar review has been undertaken by the South West SCG and PCTs and we call on the West Midlands SCG to follow their lead.
- Establish a West Midlands Neuromuscular Network to be led by a Network Manager/Coordinator to ensure that services are developed and improved across the 17 PCTs in the region.
- Recognise services for patients with muscular dystrophy and related neuromuscular conditions as specialised and subject to collaborative commissioning arrangements at a regional and local level, in line with the Department of Health's Specialised Services National Definition Set.
 - Ensure that every individual affected by muscular dystrophy is supported by a care advisor with an understanding of muscular dystrophy and related neuromuscular conditions. Five full-time care advisors are needed to serve the 5,000 people in the area with a neuromuscular condition – 90% of whom currently do not have access to the service.
- Ongoing physiotherapy to be provided to all adults and children with a neuromuscular condition in each PCT. While there are areas of good service provision for paediatric physiotherapy, this is not uniform across the region, with particular issues relating to outpatient respiratory physiotherapy for children living in the north of the region. Across the whole of the West Midlands the physiotherapy needs of adults are not currently met effectively.
- There is an urgent need to strengthen the input to clinical care from speech and language therapy and dietetics to tackle eating and swallowing difficulties that can cause major medical issues if not addressed. For example studies have reported malnourishment for some older patients with Duchenne muscular dystrophy. Problems such as this are avoidable with correct advice and treatment as conditions progress.²
- Psychological support is a vital element of a comprehensive service that should be provided for patients and families living with a neuromuscular condition in the West Midlands. These are in the main genetic conditions and families need psychological support throughout the lifetime of the condition and particularly during key times such as diagnosis, genetic counselling, when becoming wheelchair dependent, at transition stage, prior to spinal surgery and when end of life issues need to be addressed.
- There is an urgent need for preventative investment in cardiology services for individuals with neuromuscular conditions and also in interventions which can be predicted as conditions progress, such as spinal surgery.
- Wheelchairs to be included in the review to ensure that they are commissioned as specialised.

Our key findings include:

- Existing specialist clinical posts are vulnerable as they are reliant on charitable funding and not embedded within the system. The Muscular Dystrophy Campaign provides direct financial support for the only Care Advisor/Coordinator post in the West Midlands and the only clinical psychologist for patients with muscular dystrophy and related neuromuscular conditions. There is currently no NHS planning in place to ensure future stability and continuity of these posts.
- There is only one neuromuscular specialist rehabilitation clinic in the West Midlands. The service is led by a Consultant in Rehabilitation Medicine and is regarded as extremely successful but current levels of funding do not meet the huge patient demand for this service.
- Some patients have historically had errors in their diagnosis due to a lack of accessibility to specialist expertise with potentially alarming implications. Inappropriate liver biopsies have also been reported and none of these patients had been treated by a specialist in the field.
- There is no funding for transition services to support young people moving from paediatric services to adult services, despite the increase in young men with Duchenne muscular dystrophy surviving into adulthood.
- There are long waits in the West Midlands – well in excess of the 18 weeks target – for the provision of wheelchairs which are essential for mobility and independence.
- There is only one specialist muscle pathologist in the West Midlands. Furthermore she is due to retire and there is no succession planning in place for her replacement. This will severely jeopardise diagnostic services for these rare conditions.
- There is no dedicated psychology service despite its importance as part of multi-disciplinary care for this patient group with rare and very rare conditions, often genetic in origin and with no known cures and only limited treatments available.
- Respiratory services are commissioned locally but not on a region-wide basis. Some respiratory services in the region are overwhelmed by demand while others report serious concerns about referral practices.
- There is only one Care Advisor serving the region – funded in part by the Muscular Dystrophy Campaign.
- A widespread deficit in provision of services by allied health professionals was also identified with very limited access in particular to ongoing physiotherapy – over a third of patients do not see a physiotherapist and half feel that they do not receive adequate physiotherapy support.³ Specialist physiotherapists are required to support outreach clinics and provide training and professional development for community physiotherapists.

1. Background:

There are over 60 different muscular dystrophies and related neuromuscular conditions. They are multi-system disorders, which require complex long-term surveillance and care.

Without multi-disciplinary care most patients and their families experience a further reduction in quality of life and for some conditions, shortened life expectancy. 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 (see Appendix 1)

It is estimated that there are about 1,000 people for every 1 million of the population who are affected by neuromuscular diseases. Many of these are low-incidence conditions and some are ultra-orphan affecting less than 100 people in the whole of the UK.

Improved genetic counselling is likely to cause a small reduction in the overall incidence of these conditions but improved survival has increased their prevalence in the adult population. For example, the number of boys with Duchenne muscular dystrophy living into adulthood is increasing each year due to well documented management interventions.

There are some 5,000 people in the West Midlands affected by muscular dystrophy or a related neuromuscular condition.

3. Current Commissioning arrangements



The West Midlands Specialised Commissioning Team is responsible for overseeing the commissioning arrangements for specialised services on behalf of the 17 West Midlands Primary Care Trusts for the 5.5million population. (see Appendix 1)

The West Midlands SCG is hosted by Birmingham East and North PCT and provides support to Local Collaborative Commissioning Boards in the commissioning of specialised services at a local level. The West Midlands Strategic Commissioning Group is responsible for overseeing these commissioning arrangements.

4. Commissioning specialised services – the Specialised Definition Set:

The National Specialised Commissioning Group has designated muscular dystrophy and related neuromuscular conditions within the third edition of the Specialised Neurosciences Services definition.

Specialist wheelchairs and equipment are designated within Definition No 5: The Assessment and Provision of Equipment for People with Complex Physical Disabilities (all ages) and also within Definition No. 23: Specialised services for children (see appendix 2).

5. Multi-disciplinary care for muscular dystrophy and related conditions:

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 primary, secondary and tertiary providers working within a managed clinical network. Care Advisors/Coordinators are essential to provide vital specialist care, support and advice for each individual and family affected by one of these conditions.

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.⁴ Genetic counselling and psychological services should also be offered, together with locality based specialist dietetic, occupational therapy, physiotherapy and speech therapy provision which can both improve the quality of these patients' lives and increase their lifespan. For a number of neuromuscular conditions, regular check-ups are required irrespective of symptomatology as deterioration can advance rapidly over a very short period of time.⁵

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

Indeed life expectancy is increasing now to the late 30s for some patients with Duchenne muscular dystrophy and cases are reported of patients aged 40 and above.

6. Current level of essential, specialist provision in the West Midlands

Access to specialist multidisciplinary care is not available to all patients throughout the region.

One in four respondents to the Muscular Dystrophy Campaign 2008 patient survey reported that they did not receive any specialist care for their condition.

Case study:

- *A pensioner with Limb Girdle Muscular Dystrophy has not seen a specialist since 1993 and currently receives no medical care for her condition. Despite being fully reliant on a wheelchair, she receives no physiotherapy and was told that it would not benefit her.*

In the West Midlands clinical neuromuscular services are currently offered at the Robert Jones and Agnes Hunt Hospital (adults and children), Birmingham Heartlands Hospital (children), Birmingham Children's Hospital (children) and University Hospital Birmingham (adults). A neuromuscular specialist clinic for adults is offered at the West Midlands Rehabilitation Centre in Selly Oak.

o **Diagnosis:**

Some patients have historically had errors in their diagnosis due to a lack of accessibility to specialist expertise with potentially alarming implications;

Patients are not always routinely referred to muscle experts in a timely manner and as a result may subsequently receive an incorrect diagnosis.

Case study:

- *Two patients with Duchenne muscular dystrophy were misdiagnosed as having learning difficulties. This had catastrophic implications for the young boys and their families especially as Duchenne muscular dystrophy is in fact a life limiting condition. Due to the misdiagnosis and subsequent delay in referral to a specialised centre both boys missed the 'window of opportunity' to benefit from steroid therapy and became wheelchair dependent from the age of 9 years.*

Early diagnosis of Duchenne muscular dystrophy can allow the family to make informed choices about their family planning; in addition, any future therapeutic interventions are likely to make a real difference to prognosis only if they can be instituted early.⁸

Case studies:

- *There has been one case of a patient with congenital myasthenia incorrectly diagnosed with a mitochondrial disorder. The patient was formerly wheelchair dependent. But with correct diagnosis and assessment the patient was treated with pyridostigmine and is now mobile.*
- *There have been three reported cases of patients with limb girdle muscular dystrophy who were incorrectly diagnosed with polymyositis. They were not seen by muscle specialists who would have diagnosed correctly. For ten years they were treated for polymyositis with corticosteroids and immune-suppression drugs. The side effects of these drugs can be severe, including steroid myopathy, osteoporosis, gastric ulceration, hypertension, diabetes and cataracts. Immune-suppression can cause liver impairment, opportunistic infections and an increased risk of malignancy.*
- *Two patients with Becker muscular dystrophy were incorrectly diagnosed as having McArdle disease. This wrong diagnosis led to incorrect management, no cardiac surveillance and incorrect genetic counselling.*

Diagnostic and Pathology Services are patchy with no succession planning in place to replace the only dedicated muscle pathologist in the West Midlands

Currently muscle biopsies are reported at Birmingham Children's Hospital, Queen Elizabeth University Hospital Birmingham and the Robert Jones and Agnes Hunt Orthopaedic Hospital in Oswestry. This is covered by three clinical histopathologists, two clinical scientists and one clinical scientist who specialises in electron microscopy. Two of the clinical histopathologists are based at the Queen Elizabeth Hospital and one at Birmingham Children's Hospital. All have heavy responsibilities in other clinical areas.

One highly experienced clinical scientist specialising in muscle disease in the West Midlands is retiring in two months time and will not be replaced (the post is currently funded through Birmingham Children's Hospital). In addition the most experienced UK clinical scientist in muscle pathology at Oswestry is due to retire shortly, again with no succession planning in place.

The clinical scientists have made important contributions to research and development of muscle pathology at a regional and national level. It is essential for the West Midlands that a level of service is continued in order to improve the diagnosis of patients with these conditions.

Funding constraints caused by the withdrawal of Cullyer funding have also compromised the future of this research. The Department of Health is currently examining the training and career structure for all healthcare scientists (Modernising Scientific Careers), but no funding or training programmes have been put in place. Meanwhile valuable expertise will be lost as the current professionals retire, which will have a very detrimental affect on the diagnosis of patients in the West Midlands.

○ **Respiratory services:**

Some respiratory services in the region are overwhelmed by demand while others report serious concerns about referral practices

Respiratory monitoring and support is an essential part of multi-disciplinary specialist neuromuscular care - roughly 90% of deaths among untreated Duchenne muscular dystrophy are attributable to respiratory causes⁹. Studies have shown that non-invasive ventilation and other technological respiratory treatments can extend life expectancy and improve quality of life.¹⁰

An audit of 40 sequential Duchenne muscular dystrophy 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 Duchenne muscular dystrophy receiving home ventilation and specialist multi-disciplinary care reported by the Newcastle group in the most recent study by Eagle *et al* (2007).¹¹

One of the keys to good health is to anticipate respiratory needs and not to wait for a crisis to occur. Regular respiratory assessment of patients with the most severe neuromuscular conditions is especially necessary given that patients and their families become accustomed to the deficits of chronic illness and they rarely report or complain about respiratory symptoms spontaneously.¹²

▪ **Paediatric:**

North Staffordshire

The University Hospital of North Staffordshire NHS Trust does not provide an out-patient respiratory physiotherapy service for children with muscular dystrophy and related neuromuscular conditions.

Case study:

- *A young man with myopathy and rigid spine syndrome was waiting for over a year to be assessed for use of a Cough Assist device by a physiotherapist. In order to get him seen his consultant had to admit him as an inpatient.*

Birmingham

There are 15 children on non-invasive ventilation via the paediatric respiratory clinic at Birmingham Heartlands Hospital. Access to respiratory care at Birmingham Children's Hospital is more difficult with separate clinics and longer waiting times. There is no multi-disciplinary team apart from a Paediatric Consultant for the muscle clinic at Birmingham Children's hospital.

- **Adult**

North Staffordshire

Respiratory care for patients with muscular dystrophy and related neuromuscular conditions is provided at the University Hospital of North Staffordshire. Non-invasive ventilation for inpatients with severe respiratory problems is provided in a specialist six-bedded unit on a respiratory ward. The service covers a population of 1.4 million people across Staffordshire and Shropshire.

Currently 70 patients with muscular dystrophy and related neuromuscular conditions are seen by respiratory services. However only 17 young men with Duchenne muscular dystrophy (which occurs 1:3,500 live male births) are on non-invasive ventilation. **It is concerning that so few young men have been referred to this service.**

The non-invasive ventilation clinic provides specific physiotherapy advice but the ventilatory clinic does not always have physiotherapy attendance to support the commencement of assisted coughing.

Difficulties have been reported with the level of the tariff which has to cover the cost of the doctor, nurse, technical support, consumables, oximetry and blood gases with limited outstanding funds to support a physiotherapy service. Funds are reported to be insufficient to meet the cost of physiotherapy and speech and language support.

Birmingham

In September 2007 a new clinic opened at the Birmingham Heartlands Hospital; previously there was no such dedicated adult respiratory clinic for patients with muscular dystrophy in Birmingham. Since the clinic opened there has been a 50% increase in outpatient activity in the months September to December 2008 compared to September to December 2007.

The clinic receives referrals from across the West Midlands i.e. from Dudley to Coventry and there are currently 30 adult neuromuscular patients. The majority of these patients are affected by Duchenne muscular dystrophy; 14 patients are on non-invasive ventilation and one patient is tracheotomy ventilated.

The clinic is supported by a physiotherapist, but does not have a speech and language therapist. The Muscular Dystrophy Campaign is funding a clinical psychology post which will be particularly beneficial to patients, including those on non-invasive ventilation and those who are tracheotomy ventilated.

○ Cardiology services

As a number of neuromuscular conditions affect the heart, cardiac monitoring should be recognised as an essential part of a multi-disciplinary approach to care. People affected by Myotonic dystrophy and Emery-Dreifuss dystrophy are prone to abnormal heart rhythms, while people affected by Duchenne or Becker muscular dystrophy are more likely to experience cardiomyopathy.

Regular cardiac screenings are crucial even for conditions which may 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.”¹³ However, the current cardiology clinic is overwhelmed by the demand for this service.

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 10.¹⁴ Without screening, cardiomyopathy can progress almost entirely without symptoms until signs of heart failure emerge, when all cardiac reserve has been eroded.¹⁵

Further, women who are carriers of mutations in the dystrophin gene are at increased risk of cardiomyopathy. They should be offered cardiac screening, even if they are not experiencing symptoms, and this occurs only on an ad hoc basis, if at all, in the West Midlands.

○ Physiotherapy:

Many patients, particularly adults, do not receive continuous specialist physiotherapy or any ongoing physiotherapy at all. In the West Midlands half of all respondents to the Muscular Dystrophy Campaign 2008 patient survey reported that they did not have access to a physiotherapist.

Many patients only have access to short blocks of therapy, i.e. six to eight sessions on a yearly basis; others have no access at all except in acute situations or for chest physiotherapy when required. There is no standard referral practice for physiotherapy. Indeed, some patients are referred by their GP, others from their consultant neurologist and some have to make a self-referral.¹⁶

Case study:

- *“My daughter, who like me has muscular dystrophy, has been waiting over a year for an appointment with a physiotherapist. I have always felt that, as from the day of diagnosis, because no treatment for a cure could be offered it was a case of go home and do what you can do!”*

According to the Chartered Society of Physiotherapy: “Physiotherapy has a vital role to play throughout every stage in the treatment and management of the 60,000 people with neuromuscular conditions in the UK. Without it, mobility and independence can suffer and in some cases their condition can rapidly deteriorate. It is recognised that early and ongoing intervention of physiotherapy can help reduce unplanned hospital admissions.”

o **Speech and Language Therapy and Dietetics**

Speech and Language Therapy (SALT) and dietetics support for neuromuscular patients are necessary for managing nutrition and dealing with swallowing and chewing difficulties. In some areas a lack of specialist dietitians and speech and language therapists has impacted on the morbidity of children with neuromuscular conditions, and can have substantial impact on quality of life.¹⁷

Chewing and swallowing difficulties are frequent among neuromuscular patients. This has been related to increased weakness of the masticatory muscles, malocclusion or other abnormalities of the oropharyngeal phase. As a consequence of this, patients may be at risk of food aspiration and aspiration pneumonias. Chewing difficulties become increasingly present with age, associated with a progressive increase in the duration of meals.

The leading clinicians in the field recommend a multidisciplinary team approach, including input from speech therapist, dietician, neurologist, and respiratory clinicians to evaluate and managing feeding problems. A systematic evaluation of weight gain, feeding abilities and respiratory function should be part of the routine medical examination in order to identify early signs of failure to thrive and the best options of management. A speech and language therapist will take a feeding history and look at how the muscles of the tongue, lips and throat are working. In addition they will look at any other problems that may affect chewing, for example with teeth. The safety of swallowing and if there are any risks of food or drink going down the wrong way (aspiration) need to be assessed to enable safe management. The doctors and speech therapist will evaluate if this needs to be assessed in more detail. A videofluoroscopy may be done to look closely at how food is chewed and swallowed.

In the West Midlands access to specialist speech and language therapists and dieticians remains poor.

The provision of speech and language therapists as part of a multi-disciplinary team at neuromuscular clinics in the West Midlands is absent. Further, unlike other centres of excellence in England speech therapists in the West Midlands do not have specific expertise in the diagnosis and provision of advice regarding the problems associated with neuromuscular disorders.

The situation is compounded by the lack of patient knowledge about what the speech therapist has to offer and referring physicians lack insight into when a patient should be referred for speech and language therapy.

o **Care Advisor/Coordinator**

There is currently only one Care Advisor/Coordinator in the West Midlands which is not sufficient to provide support to all families in the region. Indeed in the West Midlands nine out of ten respondents to the Muscular Dystrophy Campaign 2008 patient survey reported that they had no access to a key worker or care co-ordinator.

The Care Advisor/Coordinator plays an essential role in providing a comprehensive package of care to children and adults with muscular dystrophy and related neuromuscular conditions. They successfully co-ordinate the health and social care needs of patients, provide support and information to families and other local professionals and ensure a seamless transition from child to adult services.

Further, the Care Advisor/Coordinator post is cost effective. Care Advisors reduce pressure on consultant's time through the provision of additional information and support in clinic, advise patients about their overall wellbeing and crucially recognise when a planned admission is needed thus reducing costly and stressful unplanned admissions and emergencies.

As early as 2005 the Department of Health recognised that: "Some people with more complex needs requiring skilled multi-disciplinary input from a number of different agencies will need an identified person who co-ordinates care. This role includes developing a comprehensive care plan involving a range of agencies and may involve arranging access to appropriate health and social services." (National Service Framework for Long-term Conditions.)

o Psychology support

Psychology support has been identified as an important aspect of multi-disciplinary care.¹⁸ Children and adults with neuromuscular conditions would benefit from the input of a clinical psychologist to help families develop management strategies.¹⁹ Specific issues include support at the time of diagnosis, chronic illness, loss of ambulation, transition to adulthood, times of crisis and bereavement.²⁰

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.²¹ 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.²² 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.

In the West Midlands, three out of five respondents to the Muscular Dystrophy Campaign 2008 patient survey reported that they were not satisfied with the level of emotional support available to their families and to themselves. There is a clear and pressing need to develop clinical and educational psychology input and support for this patient group.

o Young adults – transition services

Increasing numbers of young people with complex conditions are living longer and reaching transition because of improvements in therapies and medical care. For young people living with muscle disease, the period between mid and late teens is often a time of great change in their condition so any handover needs to be as smooth as possible, and must enable the patients, their families and paediatricians to be confident that their needs will be met.²³

In the West Midlands clinicians currently coordinate transition between child and adult clinics. However, this service does not receive any designated funding and is vulnerable to service change.

o **Rehabilitation**

There is currently only one neuromuscular specialist rehabilitation clinic in the region. This is an outpatient clinic based at the West Midlands Rehabilitation Centre in Selly Oak. It is the only service for patients affected by muscular dystrophy and related neuromuscular conditions which is currently commissioned by the West Midlands Specialised Commissioning Group.

The clinic aims to help patients maintain independence and adapt to changes which affect their social and domestic life. During an appointment a patient can receive assistance from a number of services including physiotherapy, occupational therapy, speech and language therapy, wheelchair services and orthotics. The service is led by a Consultant in Rehabilitation Medicine and is regarded as extremely successful although current levels of funding do not meet the huge patient demand for this service.

We are calling for an extension of this rehabilitation support service reaching across the West Midlands to ensure that all adults with muscular dystrophy or a related neuromuscular condition can have the best quality of life for the longest possible time.

o **Equipment:**

Children and adults are being forced to wait for essential wheelchairs and community equipment despite the complex and at times life limiting nature of their conditions and the inclusion of specialist wheelchairs in the Department of Health Specialised Services National Definition Set.

In the West Midlands, PCTs do not collaborate to provide specialist wheelchair services. Consequently, 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. 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. This can mean that people are not being properly assessed or being offered appropriate equipment.

A Muscular Dystrophy Campaign Freedom of Information request in February 2009 revealed that the average wait for a powered wheelchair for children in the West Midlands is 33 weeks. This compared to a national average of 19 weeks.

Two of the country's worst performing PCTs for wheelchair waiting times are located in the West Midlands. Both Birmingham East and North PCT and South Birmingham PCT reported that children with muscular dystrophy and related conditions had to wait on average 18 months for a powered wheelchair (73 weeks).

Case studies:

- *One gentleman who has Myofibrillar Myopathy said, "I am still waiting for information from wheelchair services about an appropriate chair which I will inevitably have to buy myself. This is the most inefficient service I have ever come across."*
- *A young girl has Ulrich Congenital Muscular Dystrophy she said "I have to purchase my own chairs as the NHS ones are too big, too heavy and generally inappropriate and impractical."*

Appendix 1

West Midlands PCT populations

Population data taken from PCT websites/annual reports

Primary Care Trust	Resident Population	Prevalence of neuromuscular conditions (1:1000)
Birmingham East and North	437,500	437
Coventry Teaching	326,010	326
Dudley	310,000	310
Heart of Birmingham	300,000	300
Herefordshire	178,000	178
North Staffordshire	210000	210
Sandwell	320,000	320
Shropshire County	295,000	295
Solihull	212,000	212
South Birmingham	383,000	383
South Staffordshire	604,000	604
Stoke on Trent	250000	250
Telford and Wrekin	166,000	166
Walsall Teaching	250,000	250
Warwickshire	540,000	540
Wolverhampton City	237,000	237
Worcestershire	553,000	553
TOTAL	5,571,510	5571

Appendix 2

Summary of Findings from MDC patients survey 'State of the Nation'

68 families from the West Midland completed the survey out of a total of 850 from across the UK

Health and Key Workers

- Two fifths of respondents said that their experience of the diagnosis process was either poor or very poor.
- One third of patients have no access to a specialist neuromuscular consultant and an additional one in ten do not know or do not state whether they have access to a specialist neuromuscular consultant.
- Three out of five of patients are not satisfied with the level of emotional support available to their families and to themselves and an additional one quarter do not state whether they are satisfied with the level of emotional support.
- Nine out of ten patients and their families have no access to a key worker or care co-ordinator and an additional one in ten do not know or do not state whether they have access to a key worker or care co-ordinator.
- Two thirds patients are unsatisfied with the amount and clarity of information available to them.
- Half of patients feel that they do not receive enough physiotherapy.
- Half of patients do not see a physiotherapist.

Transition

- Three out of five patients rated their transition from childhood to adult services as either poor or very poor.

Wheelchairs and Community Equipment

- Almost half of patients fund their wheelchair out of their own pocket or thanks to a charity.
- Three fifths of patients have experienced delays or difficulties in receiving the appropriate chair.
- One in three rated the service received for assistance in the upkeep of their chair as either poor or very poor.
- Half of patients have experienced delays and difficulties in receiving appropriate home adaptations.
- Half of patients have experienced delays in receiving an appointment with their Occupational Therapist – the key worker most likely to assess their needs.

Appendix 3:

Commissioning specialised services – the Specialised Definition Set:

Since March 2009 neuromuscular services have been recognised as specialist by the National Specialised Commissioning Group, and have been covered in the revised neurosciences definition as follows:

4. Specialist Clinic for Neuromuscular Disorders (children and adults)

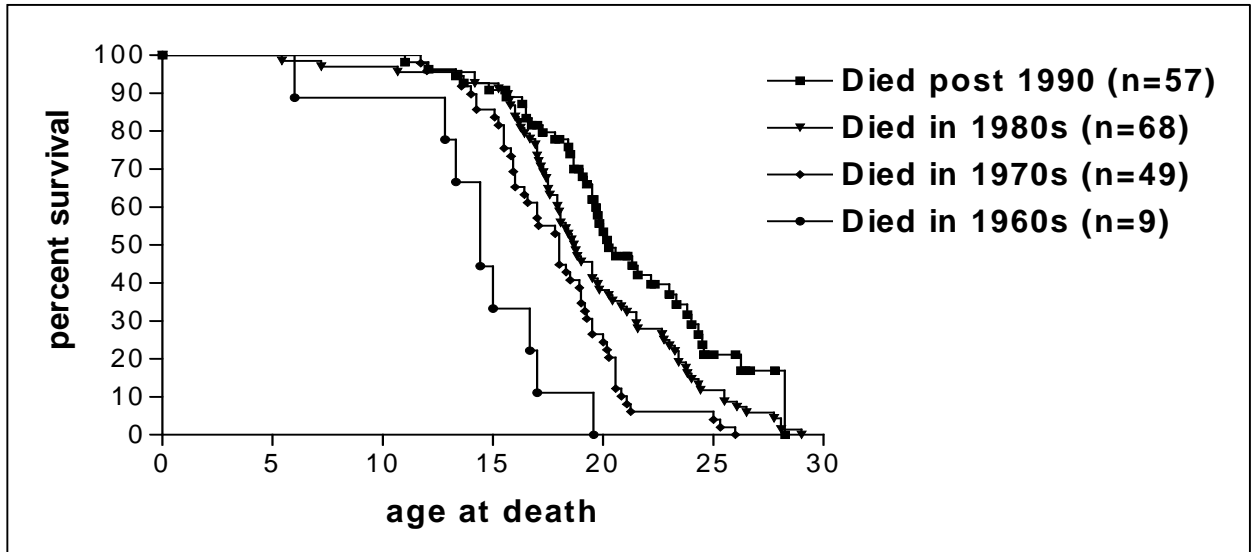
Specialised services for neuromuscular disorders may include:

- **Multiprofessional care including joint involvement of: neuromuscular specialist nurses, professions allied to medicine, dieticians, orthotists, speech and language therapists, psychologists, respiratory care services, orthopaedic or spinal surgical specialist services, cardiac specialist services**
- **Transitional care between paediatric and adult clinicians**
- **Joint neurogenetics services.**

Appendix 4:

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 ventilation²⁴)



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

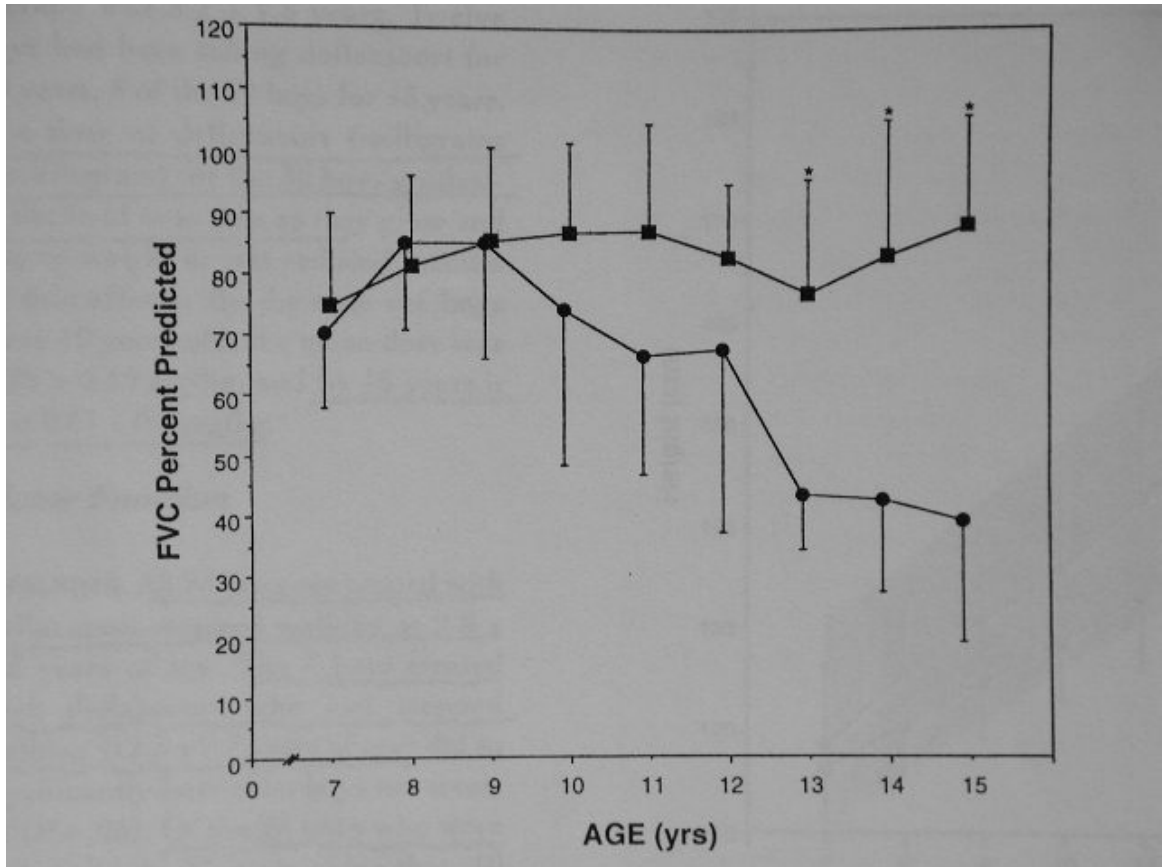
1990s: Mean life expectancy: 19.5 years

Improvement is due to multi-disciplinary care

Appendix 5:

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*)²⁵



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:

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

References

- ¹ In 2008 the Muscular Dystrophy Campaign undertook the largest nationwide survey undertaken of people affected by muscular dystrophy and related neuromuscular conditions. 850 people completed the survey from across the UK – including 68 families from the West Midlands region.
- ² Pane, M., Vasta, I., Messina, S., Sorleti, D., Aloysius, A., Sciarra, F., Mangiola, F., Kinali, M., Ricci, E., Mercuri E. Feeding problems and weight gain in Duchenne muscular dystrophy *European Journal Of Paediatric Neurology* 10 (2006) p. 231 – 236. Philpot J, Bagnall A, King C, Dubowitz V, Muntoni F. Feeding problems in merosin-deficient congenital muscular dystrophy. *Archives of Disease in Childhood*, Volume 80, Issue 6, 1999; 542-547. 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.
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