WEST MIDLANDS SPECIALISED COMMISSIONING GROUP

Neuromuscular Services Review and Development Strategy

"Delivering Better Outcomes for Service Users and Better Value for the NHS"

November 2009

Executive Summary

- The West Midlands Specialised Commissioning Group (SCG) called for a review and development plan in relation to services for people with muscular dystrophy and related neuromuscular conditions.

- The SCG sought recommendations that had to be based on best practice and a firm evidence base in relation to service improvement that would lead to better outcomes for service users, and better value for PCTs.

- This report and review is the West Midlands formal response to the requirement of the All Party Parliamentary Group for Muscular Dystrophy and the requirement set out in Lord Walton's report that called for prompt action to address failings in services across the country.

- The review in the West Midlands has revealed that there is a serious risk to service continuity and an urgent need to invest in some key areas, particularly support to current single-handed consultants, and the sole neuromuscular care advisor for the region.

- There is significant inequality in access to services across the region. A person's ability to access specialist care and benefit from better outcomes can be a matter luck and chance. There are no clear referral pathways, and many patients get lost in the system, without receiving the appropriate care from the appropriate specialist.

- There is a wealth of evidence that life expectancy for people with some forms of muscular dystrophy and related neuromuscular conditions can be extended significantly if all people with these conditions access specialist multidisciplinary care.

- For those people with one of these conditions a key issue is slowing the rate of decline that leads to increased and earlier morbidity and mortality and consequential greater cost to the NHS as a result of unplanned and avoidable emergency admissions to hospital.

- More than 50% of people with a neuromuscular condition in the West Midlands (in excess of 2,500 people) do not access specialist multidisciplinary care.

- The consequence of not accessing specialist multidisciplinary care is limited life-expectancy for service users and significant expenditure by PCTs to fund unplanned emergency admissions for people with a neuromuscular condition who experience a respiratory or other crisis.

- In the last financial year, PCTs in West Midlands spent £6.6m on unplanned emergency admissions for people with muscular dystrophy or a related neuromuscular condition. Clinician evidence suggests that a large proportion of this cost could have been avoided through timely access to specialist care.

- The report recommends an investment of £1.26m in total which would address both the cost of taking away risk to service continuity and increasing capacity to deal with the 50% of patients who currently do not access specialist care.

- Local community and primary care services have an important role to play in supporting people with these conditions. However, the commissioning investment plan set out in the report relates solely to the specialised aspects of the service. The way in which specialist and local services work together is a crucial issue and this report does put forward proposals that seek to strengthen this working e.g. developing capacity in specialist practitioners to work with local practitioners to transfer and develop skills in providing routine aspects of treatment people with a neuromuscular condition.

- This report has reviewed a number of examples of good practice and good service models both in this country and abroad and this report advocates a Hub and Spoke service model with capacity for specialist centres to provide local outreach services in main areas of population.

- A regional managed clinical network is being recommended. This would have responsibility for coordinating work between specialist and local services, oversee the implementation of service development plans and ensure maximum impact and optimal service outcomes from current and future investment.

- Current specialist services at the main centres, together with proposed future specialist outreach services will constitute a West Midlands Regional Neuromuscular Service as a single clinical directorate.

Contents

Chapter 1:
Introduction and Purpose of the Report

Muscular dystrophy and related neuromuscular conditions are a collection of rare or very rare conditions, affecting approximately 1 in 1,000 people in the U.K. This year, the National Specialised Commissioning Group designated these conditions within the third edition of the Specialised Neurosciences Services definition. The responsibility for commissioning services now falls with the Specialised Commissioning Group.

Local community and primary care services have an important role to play in supporting people with these conditions. However, the commissioning investment plan set out in the report relates solely to the specialised aspects of the service. The way in which specialist and local services work together is a crucial issue and this report does put forward proposals that seek to strengthen this working e.g. developing capacity in specialist practitioners to work with local practitioners to transfer and develop skills in providing routine aspects of treatment people with a neuromuscular condition.

The West Midlands Specialised Commissioning Group (SCG) agreed in July 2009 to undertake a review of services for patients with muscular dystrophy and related neuromuscular conditions and, on the basis of current best practice and the available evidence base, to produce proposals to improve services in a way that will deliver better outcomes for service users and better value for PCTs

The decision to undertake the review followed the presentation of evidence by NHS West Midlands, to an inquiry carried out by All Party Parliamentary Group (APPG) for Muscular Dystrophy.

The APPG's Inquiry into Access to Specialist Neuromuscular Care was launched in December 2008 to carry out an in-depth investigation looking at access to specialist, multidisciplinary care for people living with muscular dystrophy and related neuromuscular conditions.

In August 2009 the APPG published the inquiry's final report – the Walton Report – which brought forward a number of recommendations which called for prompt action to address the failings in services.

The recommendations put forward in the Walton Report will be followed up by the APPG, to ensure that that action is taken to implement essential improvements in specialist multidisciplinary care for people living with these conditions across the UK

During the presentation of evidence to the APPG NHS West Midlands gave a commitment to undertake a service review and to come forward with a development plan to be considered by the Specialised Commissioning Group.

In a period where the NHS and other public services will experience severe financial pressures, any service review and development plan must be able to demonstrate both that significantly better outcomes can be delivered for service users AND that there is a best-value economic case for investing in service change and service transformation. There is a clear case, demonstrated in this report with full supporting evidence, for an "invest to save" approach to service improvement in this field.

This report covers the following:

- Background, aims and objectives of the neuromuscular services review
- Neuromuscular conditions and their treatment
- Prevalence
- Mapping of current specialist neuromuscular services in the West Midlands
- Outcome from service mapping
- The cost-benefit and evidence case for investment
- Best practice models
- National and regional policy context
- Service vision, service standards and service model
- Recommendations
- Investment plan

Chapter 2:
Background, Process and Overall Aims and Objectives

Following the decision of the SCG to commission the review, a West Midlands Neuromuscular Services Development Group was established to steer and oversee the work.

The Development Group comprised stakeholders from Local Collaborative Commissioning Boards (LCCBs), PCTs, service users, carers, clinicians, provider Trusts, the WM Specialised Commissioning Team and the Muscular Dystrophy Campaign.

The work was led and project managed by Peter Boileau and commenced in July 2009

The remit and aims of the work were as follows:

- To assess need, prevalence and life expectancy rates in the West Midlands
- To compile an evidence base of best practice and service models
- To map current specialist services in the West Midlands
- To assess current services against known best practice and evidence of what works effectively
- To identify gaps in services and service deficiencies
- To develop a service vision/model of service/service strategy for the West Midlands
- To develop and agree recommended standards of care
- To agree priorities for service improvement that must deliver better quality outcomes for patients and better value for PCTs
- To recommend an implementation plan and on-going mechanism to deliver the strategy

Before proposals and recommendations were compiled a stakeholder event took place in September 2009 when a wide range of stakeholders from the NHS-PCTs, providers, clinicians, local government, service users and carers and the voluntary sector were able to have their say and input into the evolving work.

This review and development work was undertaken and completed at a time when PCTs were looking at how they would be taking forward the National Service Framework for Long Term Neurological Conditions. Muscular dystrophy and related neuromuscular conditions are just one relatively small sub-set of those neurological conditions. It was decided to go ahead with the neuromuscular review for the following reasons:

- The need for the NHS in the West Midlands to honour its commitment to the All Party Parliamentary Group and respond to the Walton Report

- A substantial amount of evidence of best practice was already available for this condition and that the Muscular Dystrophy Campaign had already produced a report of services in the West Midlands: Building on the Foundations: the need for a neuromuscular service across the West Midlands (April 2009)[1]

- This review and work could act as a template and support to PCTs in relation to their work on the wider NSF

A further consideration in proceeding early with this specific condition was the contribution this work could make to commissioning development generally in the West Midlands. The work is a very useful exemplar in relation to how knowledge and management of a care pathway, the compilation of cost-benefit evidence/best practice and the active involvement of clinicians can arm commissioners with powerful information to improve outcomes and deliver better value.

This work is very much in line with West Midlands "Investing for Health" strategy and its emphasis on better care pathway management and **Q**uality-**I**nnovation-**P**roductivity-**P**revention-**P**artnerships (QUIPPP)

This is a draft report and is being circulated widely for consultation before a final report is presented to SCG at the end of December 2009.

Chapter 3: Neuromuscular Conditions and Their Treatment

The conditions

- Neuromuscular conditions are disorders of the muscle, peripheral nerve, neuromuscular junction or motor neurone which lead to weakness.

- There are more than 60 different types of muscular dystrophy and related neuromuscular disorders (NMD) caused by 200 known genetic loci.

- Neuromuscular disorders can be genetic or acquired:
  - Inherited neuromuscular disorders include: the muscular dystrophies, the spinal muscular atrophies, the congenital and syndromal neuropathies, congenital myopathies, metabolic myopathies, inherited myasthenic syndromes, channelopathies, mitochondrial disorders, the myotonias and the inherited neuropathies.
  - Acquired disorders include myasthenia gravis, autoimmune neuropathies and inflammatory myopathies.

- There are few treatment options for most of these diseases.

- A number, such as Duchenne Muscular Dystrophy are aggressive and cause progressive muscle wasting and weakness, orthopaedic deformity, cardiac and respiratory compromise, dependency on others for day-to day care and usually result in premature death. Others cause life-long disability.

- They can present from infancy to old age: some neuromuscular disorders can present in childhood (e.g. Duchenne Muscular Dystrophy) or young adult life. Others can be late onset conditions in adulthood (e.g. Inclusion Body Myositis).

Appendix 1 sets out further details of the main neuromuscular conditions

Treatment

Early onset conditions are usually diagnosed in childhood (following the appearance of symptoms of weakness), however in absolute terms, more neuromuscular conditions will present and persist in adulthood. Referral to specialist centre neuromuscular centre will include assessment by a neuromuscular team and confirmation of diagnosis by muscle biopsy and or genetic studies.

The advice of a clinical geneticist to give appropriate genetic counselling is required for other family members who may carry the genetic predisposition.

The impact of the diagnosis is far reaching for the patient and their family, as many of these conditions are severely disabling and life limiting. The family and carers need support from a number of professionals, for instance: health care, social care and later for education services as well as advice on financial support.

Support for these complex care packages is provided through the help of a Neuromuscular Care Advisor or Care Coordinator who provides valuable practical and emotional support for patients, their families and other carers.

Following diagnosis patients and their families need to be seen for regular review and assessment at a specialist centre. Coordinated and comprehensive multidisciplinary specialist care should include a neuromuscular specialist consultant and physiotherapist, and, dependent on medical need, may also include specialist cardiac, respiratory, and orthopaedic care.

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

As these are rare or very rare conditions, the responsibility for commissioning services falls with the Specialised Commissioning Group. Indeed, the National Specialised Commissioning Group has designated muscular dystrophy and related neuromuscular conditions within the third edition of the Specialised Neurosciences Services definition.

However day to day care to help with daily living such as the need for ongoing physiotherapy, specialist mobility equipment in the home or school and help with home adaptations is provided through local health and social care teams for children or adults with disabilities, with advice and support from specialist centre.

Chapter 4: Prevalence

The prevalence of muscular dystrophy and related neuromuscular conditions in the UK is about 1000 children and adults for every million of the population. In the West Midlands therefore we would expect to have about 5000 people with neuromuscular conditions who require access to specialist services.

Prevalence by PCT in the West Midlands is set out in the following table. Resident populations and not GP responsible populations have been used as PCT commissioning and specialised commissioning covers all of the residents in a PCT area irrespective of where a patient is registered for primary care.

Primary Care Trust	Resident Population	Prevalence of neuromuscular conditions (1:1000)
Birmingham East and North	438,000	438
Coventry Teaching	306,726	307
Dudley	300,000	300
Heart of Birmingham	300,000	300
Herefordshire	178,000	178
North Staffordshire	210,000	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	250,000	250
Telford and Wrekin	167,000	167
Walsall	250,000	250
Warwickshire	546,000	546
Wolverhampton City	237,000	237
Worcestershire	553,000	553
TOTAL	**5,549,726**	**5550**

Two main issues arise in relation to this table:

- It demonstrates the frequency of these rare conditions and the need for multi PCT collaborative specialised commissioning to secure the future viability and growth in these services.

Appendix 2 sets out estimates of the prevalence of each of the neuromuscular conditions we are looking at in this report.

- It shows that about 5500 people in the West Midlands need to access specialist multidisciplinary services at a specialist centre if they are to obtain optimum care outcomes. As set out below, we know that currently only 50% of people with these conditions access specialist care and that this impacts not only on their quality of life but on their reduced life expectancy and the cost to the NHS of repeated emergency admissions to hospital for respiratory, cardiac or other crises (see below)

Chapter 5:
Current Specialist Neuromuscular Services in the West Midlands

The tables below sets out the location of main specialist centres in the region, consultant establishment and current workload. Two key issues arise from this information - location and planning of services and workload: prevalence, capacity and demand.

Location and Planning of Services

- Specialist adult and children's neuromuscular services operate in Birmingham (Birmingham Children's, Heartlands and Queen Elizabeth Hospitals) and in Oswestry (Robert Jones and Agnes Hunt Orthopaedic Hospital)

- Specialist neuromuscular respiratory centres that take referrals from the main children's and adult units operate at Birmingham Heartlands and Children's Hospital and at the University Hospital North Staffordshire.

- A number of West Midlands patients travel to the Neuromuscular Centre (NMC) in Cheshire to receive specialist physiotherapy. While the NMC invoices PCTs for this treatment, only 59% of PCTs in the region reimburse the centre.

- A specialist Neuromuscular Rehabilitation Service is provided at the West Midlands Rehabilitation Centre in South Birmingham where the consultant takes adult referrals from the neuromuscular specialists in Birmingham.

- There is no regional pattern to these services and hence these services, although highly specialised have not been developed in line with any regional plan.

- Services in the West Midlands have grown up historically around consultants who have over time developed a special interest and attracted referrals from other consultants.

- Patients have to travel long distances for their care with minimum outreach clinics being provided due to the severe limitation on the capacity of the existing neuromuscular specialists.

- There are large geographic parts of the region that are long distances from a specialist centre AND do not have the benefit of local specialist out-reach clinics (Herefordshire, Worcestershire and North Staffordshire)

- There is no real logic or coherence to referral patterns and referral routes. There are cross referrals across the region between specialists for particular reasons and this reflects the ad hoc and unplanned nature of current services.

Table: Location of specialist neuromuscular centres

Clinician	Speciality	Clinic bases
Nick Davies	Adult neuromuscular	QEH
Ros Quinlivan	Adult and paediatric neuromuscular	RJAH & BCH
Helen Roper	Paediatric neuromuscular	BHH & BCH
John Winer	Adult neuromuscular	QEH
Martina Walsh	Adult neuromuscular rehab	West Mids Rehab Centre

Table: Location of specialist neuromuscular respiratory centres

Clinician	Speciality	Clinic bases
Dev Banerjee	Adult respiratory	BHH
Titus Ninan	Paediatric Respiratory	BHH
Jane Clarke	Paediatric Respiratory	BCH
Martin Samuels	Paediatric respiratory	UNHS
Martin Allen	Adult respiratory	UHNS

Workload- Prevalence and Demand

- Prevalence studies show that there are about 5000 people in the West Midlands with muscular dystrophy or a related neuromuscular condition.

- Workload figures below revel that only 50% of the expected numbers actually access and receive specialist multidisciplinary services. The 2500 people who do not access services experience poor outcomes in terms of quality of life and life expectancy. Clinician evidence has suggested that patients who do not receive specialist care are 20 times more likely to require an unplanned emergency admission to hospital following a respiratory, cardiac or orthopaedic crisis than patients who do receive multidisciplinary care.[2]

- Current capacity as set out below is working to a maximum level dealing with just 50% of the number of people with one of these conditions. There is a serious capacity shortage to pick up the demand that exists.

Table: Workload

Clinician	Speciality	Clinic bases	Total number of patients	Number of new referrals each year	WTE
Nick Davies	Adult neuromuscular	QEH	500	120-140	0.2
Ros Quinlivan	Adult and paediatric neuromuscular	RJAH & BCH	850	200	0.5
Helen Roper	Paediatric neuromuscular	BHH & BCH	415	65	0.4
Martina Walsh	Adult neuromuscular rehab	West Mids Rehab Centre	185	40	0.2
John Winer	Adult neuromuscular	QEH	Approx 500	120-140	0.2
Totals			**2437**	**565**	**1.5**

The number and detail of clinics held at the various centres in the region is set out in the following table:

Clinic	Clinician	Location	Frequency	Adult or paediatric	How many patients at each clinic	Of this, how many are new patients	Is this a condition-specific clinic (e.g. for Myotonic Dystrophy) If so, which condition	How long is the average appointment at the clinic (mins)	Do you have present at clinic: a) Muscular Dystrophy Care Advisor	b) Specialist neuromuscular physio	C) Psychologist	d) Specialist muscle nurse
1	**HR**	BHH	4 per month	Paed	9	0	Follow up, all NM	20 -30	yes	yes	no	no
2	**HR**	BHH	3/4 per month	Paed	3	3	New pts, all NM	45 - 60	yes	yes	no	no
3	**HR**	BHH	15 per year	Paed	7	0	DMD steroid	30	yes	yes	yes	no
4	**HR**	BHH	4 per year	Paed	10	0	SMA	20-30	yes	yes	no	no
5	**HR**	BCH	1 per month	Paed	8-12	2 or 3	All NM	20 -30	yes	No	no	no
6	**HR**	BCH	4 per year	Paed	6-8	0	Myotonia /transition	-	-	-	-	-
7	**HR**	BCH	2 per yr	Paed	6-8	0	metabolic	-	-	-	-	-
8	**RQ**	RJAH	1 per month	DMD steroid	8	1	DMD steroid	60	No	Yes	Yes	Yes
9	**RQ**	RJAH	5 per month	Both	16-22	3	General muscle	New: 60. follow up: 30	No	Yes	Yes	Yes
10	**RQ**	RJAH	3 per year		9 - 12	4	McArdle disease	90	No	Yes	Yes	Yes
11	**RQ**	RJAH	1 per month		4	0	Biopsy results clinic	60-120	No	Yes	Yes	Yes
12	**RQ**	BCH	4 per month	Paeds	6 -7	2		New: 45. follow up: 30	No	no	no	No
13	**JW**	QEH	2 per month	Adult	6	6	All NM	30	no	yes	no	no
14	**JW**	QEH	2 per month	Adult	15	0	All NM	15	no	Yes	no	no
15	**ND**	QEH	2 per month	Adult	6	6	All NM	30	no	yes	no	no
16	**ND**	QEH	2 per month	Adult	15	0	All NM	15	no	yes	no	no
17	**MW**	WMRC	2 per month	Adult – rehab	6-7	2-3	All NM	60 mins new, 30 mins follow up	No	No	No	No

Notes:

1. Total specialist neuromuscular physiotherapists available for whole region = 1.1 Whole Time Equivalent (WTE)

    - This is broken down by centre as follows:
        a) Heartlands: 0.5
        b) RJAH: 0.4
        c) QEH: 0.2
        d) BCH: 0

2. Total specialist psychologists available for whole of region = 0.6 WTE

    - This is broken down by centre as follows:
        a) Heartlands: 0.1
        b) RJAH: 0.5 (charitable funded until end of March 2011)
        c) QEH: 0
        d) BCH: 0

Muscle Pathology Service

Muscle biopsies are processed and reported at 3 centres in the West Midlands:

- RJAH Orthopaedic Hospital, Oswestry (RJAH) 50 biopsies per year
- Birmingham Children's Hospital (BCH) 50 biopsies per year
- Queen Elizabeth Hospital Birmingham (QEH) 200 biopsies per year

Current staff that report muscle biopsies:

- RJAH: 1 clinical scientist (8D) – due to retire
- BCH: 1 consultant paediatric pathologist
- QEH: 2 consultant neuropathologists, 1 due to retire

As stated at the beginning of this report, diagnosis via muscle biopsy is a critically important service and it is essential that early access to diagnosis is made. There is a need for education and higher awareness in non-specialist services particularly in Primary Care and general acute care.

The other main issue relates to the very fragile position regarding dedicated staffing in this service where there are major issues of succession planning. Some of the very few specialist pathology posts about to become vacant due to retirements and no plans in place to replace these posts. Details are set out later in this report together with recommendations about putting this critical service on a sounder regional footing.

Chapter 6:
Outcome from the Service Mapping

General

The Development Group mapped current services and this; together with the survey work completed earlier this year by the Muscular Dystrophy Campaign highlighted the following key service issues:

1) Current specialist services are overstretched and do not have the capacity to deal with current patient numbers, nor any increase in patients:

- The Consultant at RJAH, Oswestry/BCH is single handed and there is no cover when she goes on annual leave or is off sick. This consultant is supported by one paediatric registrar, who is currently working part-time.

- The specialist clinics at the centre are vulnerable as the tariff does not cover all the costs especially for muscle biopsy.

- Clinics at Birmingham Heartlands Hospital are run by a single handed consultant and have not been specifically funded. There is no cover for leave and no registrar support.

- There is only one dedicated neuromuscular specialist rehabilitation clinic in the region.

- The capacity of specialist clinics is further stretched by the longer appointment time each patient requires than in non-specialist clinics. This is because these are very rare and complex conditions, and the clinicians are often the last point of resort for families and patients with undiagnosed conditions.

2) Specialist services are not able to provide a full multidisciplinary team at clinic:

- The specialist neuromuscular physiotherapy service in Oswestry is only funded for 2 days a week, and the unit's specialist nurse is funded for 3 days per week. Furthermore, the psychologist at the unit is funded purely through the Muscular Dystrophy Campaign for just 3 days per week – this funding is due to cease at the end of March 2011. There is no Neuromuscular Regional Care Advisor to support the service.

- The clinics at BCH have no Neuromuscular Regional Care Advisor, psychologist, specialist nurse or physiotherapy support.

- Patients in the region report severe deficits in accessing services provided by Allied Healthcare Professionals - over a third of patients do not see a physiotherapist and half feel that they do not receive adequate physiotherapy support.[3]

3) No workforce planning:

- Existing specialist clinical posts are vulnerable as they are reliant on charitable funding and not embedded within the system: There is only one Neuromuscular Care Advisor/Coordinator in the whole of the region – and this post is reliant on funding by the Muscular Dystrophy Campaign which will cease at the end of the 2009-2010 financial year

- One clinical psychologist post is also reliant on charitable funding from the Muscular Dystrophy Campaign which is due to cease at the end of March 2011.

- There is currently no NHS planning in place to ensure future stability and continuity of these posts.

- 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.

4) Wheelchair services

- There are long waits in the West Midlands for the provision of powered wheelchairs which are essential for mobility and independence.

Specific

a. Specialist Multidisciplinary Care:

There is limited access to specialist multidisciplinary care - with only half of the patients in the region currently having access to a specialist.[4]

Case studies

- *A patient 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.*
- *A patient with Myotonic Dystrophy has not seen a neurologist for ten years, since his previous doctor moved away. In that time his condition has progressed considerably. After a fall in summer 2008 he spent four weeks as an inpatient at Birmingham Heartlands Hospital. During this time he didn't see a neuromuscular specialist or receive any physiotherapy, and by the time he left the hospital, he was unable to stand unassisted. He has still had no contact with a neuromuscular specialist.*

The capacity at the few main centres is overstretched: new appointments are scheduled to always meet the 18 week target, but this is often at the expense of follow-up appointments which can be delayed so they take place at longer intervals than clinicians would advise. Capacity is also stretched by the nature of the conditions which are lifelong, progressive conditions – patients are not discharged, leading to a steady and continual increase in patient numbers.

Case study:
A patient with Facioscapulohumeral muscular dystrophy was advised by her consultant that she should see him every three months due to the progression of her condition. However, she has been limited to two appointments a year. She was due to see her consultant in August 2009, but was informed in June 2009 that this appointment has been cancelled, and rearranged for December 2009. This will have been nearly a year since her last appointment, when expert clinical guidance recommends a gap of no more than three months. She said: "I don't want to end up as an emergency admission because my condition has deteriorated so rapidly without anyone monitoring it."

Local services do not uniformly link effectively with specialist services because there is no managed clinical network in this area.

Case study:
One patient with Limb Girdle Muscular Dystrophy has battled to access a neuromuscular specialist. She was referred from a neurologist to a rehab consultant who has now referred her back to the neurologist she started with. She said "It's like they don't know what to do with you."

b. Diagnosis:

There is no uniform or consistent availability of, and accessibility to, specialist expertise for early diagnosis.

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.

Case studies:
- *There has been one recent 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.*
- *Two patients with Duchenne muscular dystrophy (DMD) were misdiagnosed as having learning difficulties. This had catastrophic implications for the young boys and their families especially as DMD 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.*
- *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. Immunosuppression 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.*

c. Muscle Pathology:

There is only one specialist muscle pathologist in the West Midlands. The clinical scientist at Birmingham Children's Hospital who assisted in reporting and had extensive experience of muscle pathology, including electron microscopy has retired and not been replaced.

At QEH there is a clinical scientist with several years experience of muscle pathology, particularly electron microscopy. Electron microscopy of muscle is highly specialised and performed at the RJAH, QEH and BCH. At QEH and RJAH this is done by the clinical scientist. At BCH electron microscopy is done less often now because of restrictions on the pathologist's time.

BCH and QEH frequently send samples to the clinical scientist at RJAH for second opinion and advice.

The clinical scientist at RJAH, Professor Caroline Sewry, works part time at RJAH and part time at Great Ormond Street Hospital, London, where she supervises a similar muscle pathology service, in particular the NCG funded service for congenital muscular dystrophies and congenital myopathies. She has nearly 40 years experience in the field and has a national and international reputation. Succession planning for her retirement is in place in London, but not at the RJAH.

d. Respiratory Services:

Some respiratory clinics in the region are overwhelmed by demand whilst others report serious concerns about referral practices.

Respiratory care for patients with muscular dystrophy and related neuromuscular conditions is provided at the University Hospital of North Staffordshire. However, this service does not include 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.

Non-invasive ventilation for adult 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 in 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

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 multidisciplinary team apart from a Paediatric Consultant for the muscle clinic at Birmingham Children's hospital.

In September 2007 a new dedicated adult respiratory clinic opened at the Birmingham Heartlands Hospital; previously there was no such service 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 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.

e. Cardiology Services and Spinal Surgery:

Cardiology clinics in the region are overwhelmed by the demand for regular cardiac screening for people with Duchenne muscular dystrophy despite best practice and evidence base that screening should take place before any surgery and that regular screening should be done.[5] There is limited preventative investment in cardiological services for people with neuromuscular conditions

f. Physiotherapy and hydrotherapy:

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.[6]

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

Case study:
One patient said: "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 or a cure could be offered it was a case of go home and do what you can do!"

The result can be that people have respiratory crises and then need more expensive hospital inpatient care as an unplanned emergency admission.

Patients have reported that they are generally denied access to NHS hydrotherapy provision - a service that has great benefits for them. This is largely due to restrictive entry criteria and/or lack of understanding and knowledge of the conditions on the part of local services. This inequality should cease and local services should be required to allow access by people with neuromuscular conditions to hydrotherapy services.

g. Speech and Language therapy and Dietetics:

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 multidisciplinary team at neuromuscular clinics in the region 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.

Case study:
One family had to pay privately for all the speech and language therapy for their young son with Duchenne muscular dystrophy.

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

h. Care Advice and Co-ordination:

There is currently only one Neuromuscular 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 coordinator.[7]

i. Psychology Support:

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.[8] There is a clear and pressing need to develop clinical and educational psychology input and support for this patient group.

j. Young Adults-Transition Services:

In the West Midlands clinicians currently coordinate transition between child and adult clinics, despite the increase in young people with severe neuromuscular conditions living into adulthood. However, this service does not receive any designated funding and is vulnerable to service change.

k. Rehabilitation

This is an out-patient clinic based at the West Midlands Rehabilitation Centre In Birmingham 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, posture mobility 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. There are not enough clinics to cover the whole region 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.

I. Equipment:

In February 2009 Freedom of Information requests revealed that the average wait for a powered wheelchair for children living with muscular dystrophy or a related neuromuscular condition in the West Midlands is 33 weeks. This compared to a national average of 19 weeks.[9]

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).[10]

There is no co-ordination or collaboration between PCTs to provide specialist wheelchairs for patients with these complex conditions.

Case studies:

- *A patient with 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 patient with 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."*
- *A patient affected by FSH muscular dystrophy were told that their mother required an outdoor wheelchair, but Wheelchair Services in Stoke said that they would only give her an indoor chair for six months, and then she would be able to get an outdoor chair. This has severely restricted her independence and quality of life. This six month deadline passed two months ago, and as yet, the family have not received an outdoor chair.*
- *A patient affected by FSH muscular dystrophy requested a powered wheelchair but was told by wheelchair services that she would only receive a manual chair – but her muscle condition has weakened her shoulders and arms so much that she is unable to propel herself.*
- *A patient with Myotonic Dystrophy had to wait over twelve months for his powered wheelchair after leaving hospital after a fall. During this time his independence was severely limited as he had to rely on others to push him in a second-hand manual wheelchair. He did not receive his powered wheelchair until the family had got their local MP involved in the case.*
- *A patient affected by FSH muscular dystrophy waited for two and a half years to get her wheelchair: "I had to jump through hoops and argue all the time to get it. Nothing happens quickly and people die waiting."*
- *One patient with an unknown neuromuscular condition was an inpatient in hospital and required a wheelchair. It took 6 weeks for him to be seen for an assessment, a further 8 weeks for the wheelchair to be made, and then once the wheelchair was in the hospital and ready for him to use, a further 6 weeks wait for an appointment with a wheelchair technician to hand over the wheelchair – all the time while he was occupying a bed in the same hospital: "I could actually see my chair, but wasn't allowed to use it." Once discharged with his new wheelchair, his specialist neuromuscular consultant assessed it as completely inappropriate for his complex needs. He said: "It wasn't even a cheap chair. They spent thousands of pounds on something that is just wrong for my condition." Wheelchair services had not consulted a neuromuscular specialist before providing the chair.*

m. End of Life Care:

There is no strategy for end of life care for people with neuromuscular conditions in the West Midlands.

n. Respite care:

The Region lacks a more modern and innovative approach to respite care. There is the potential to develop a central multi-agency facility that might combine a regional base for a Neuromuscular Managed Clinical Network, Specialist Physiotherapy and a self financing work based facility for people with these conditions that could attract external funding and interest from major voluntary and business organisations.

o: Social Care

There are a number of issues that impact on the care of people with a neuromuscular condition that need to be discussed with social care colleagues.

- Social care operates with different criteria for children and adults in respect of access to care.

- For people with a neuromuscular condition a long-term view of their changing needs is required – in most cases, it is predictable when and how these needs will change. This requires long-term planning and regular reassessment and it would be more helpful if social care processes could incorporate this longer-term view of people's needs. What is also required is more effective longer-term joint planning between health and social care in respect of a care plan for people with one of these conditions. For example, the home adaptations process does not currently reflect the progressive nature of neuromuscular conditions.

Chapter 7:
The Cost-Benefit and Evidence Base for Investing in Specialist Neuromuscular Services

There is an overwhelming evidence base for investing in, and improving the care pathway for these conditions.

The detailed clinical evidence base is set out in Appendix 3.

Similarly, there are many examples of best practice to be drawn on from across the U.K and internationally. Our work has reviewed this to inform our recommendations.

Details of best practice elsewhere is set out in Appendix 4

Effective management of the Care Pathway will deliver better outcomes and better value. The right care in the right place at the right time by the right service professional is good care and good economics.

The Care Pathway can be summarised into the following components:

- Diagnostics, genetic counselling.
- Management of skeletal deformity.
- Nutrition and swallowing.
- Promote independence (orthotics, wheelchairs, prevent falls, liaise with education, access to work, home adaptations etc).
- Investigation and management of osteoporosis.
- Monitoring and treatment of respiratory failure and cardiac complications.

If the aim is to improve outcomes for people with neuromuscular conditions in the West Midlands, then we must base our actions and recommendations on the existing **evidence base,** on proven **best practice** elsewhere and on **national policies** and accepted **standards of care**

There is a growing evidence base that the provision of multidisciplinary specialist services coordinated with and linked to local services can have a significant impact on improving outcomes for people with neuromuscular conditions, particularly in terms of prolonging life expectancy.

For Primary Care Trusts there is also a cost benefit in providing specialised coordinated services within a managed clinical network where local services can be supported in providing local are for people with often complex conditions.

The availability of multidisciplinary, coordinated care, with an emphasis on early intervention and the provision of the right care in the right place at the right time along the pathway of care, will prevent inappropriate hospitalisation of people when the lack of an early and appropriate response then triggers a far more expensive option. For example the non-availability of specialist physiotherapy will often lead to chest infection requiring a hospital stay. The lateness of diagnosis will result in deterioration and progression of the condition with the result that more resources will be needed to deal with the consequences.

The following table sets out the enormous expenditure incurred by PCTs: £6.6m annually to pay for emergency admissions of people with muscular dystrophy or a related neuromuscular condition who have experienced a respiratory, cardiac, orthopaedic or other crisis:

PCT	Resident Population	Prevalence of neuromuscular conditions (1:1000)	Number of emergency admissions	Total cost of emergency admissions	Number of emergency admissions per NMD patient	Cost of emergency admissions per NMD patient
Telford And Wrekin	166,000	166	41	£113,168	0.25	£681.73
Dudley	310,000	310	83	£276,138	0.27	£890.77
Worcestershire	553,000	553	189	£488,215	0.34	£882.85
South Staffordshire	604,000	604	208	£517,278	0.34	£856.42
Herefordshire	178,000	178	64	£158,274	0.36	£889.18
South Birmingham	383,000	383	143	£413,721	0.37	£1,080.21
Wolverhampton City	237,000	237	89	£231,958	0.38	£978.73
Sandwell	320,000	320	121	£395,952	0.38	£1,237.35
Shropshire County	295,000	295	116	£268,692	0.39	£910.82
North Staffordshire	210000	210	83	£259,592	0.40	£1,236.15
Warwickshire	540,000	540	218	£594,702	0.40	£1,101.30
Walsall	250,000	250	102	£312,836	0.41	£1,251.34
Solihull	212,000	212	89	£271,746	0.42	£1,281.82
Birmingham East And North	437,500	437	184	£680,237	0.42	£1,556.61
Coventry	326,010	326	158	£483,023	0.48	£1,481.67
Stoke On Trent	250000	250	124	£399,724	0.50	£1,598.90
Heart Of Birmingham	300,000	300	181	£445,485	0.60	£1,484.95
Total	**5,571,510**	**5571**	**2246**	**£6,634,090**	**0.39**	**£1,141.22**

Data provided by the Public Health Team at the West Midlands SHA.

The most costly emergency admissions are caused by respiratory crisis, cardiac problems or falls. Although there will always be some emergency admissions for patients with complex neuromuscular conditions, PCTs' expenditure of £6.6m could be substantially reduced. Analysis of one of the specialists neuromuscular consultants patient data base shows that out of 850 patients in receipt of specialist multidisciplinary care only a handful of patients were admitted for an unplanned emergency admission due to a clinical crisis.

Clinical evidence suggests that patients with a neuromuscular condition are 20 times more likely to have an unplanned emergency hospital admission if they DO NOT access specialist multidisciplinary care.

The fact that 2500 people with one of these conditions do not access specialist care would imply that the vast majority of unplanned emergency admissions set out in the above table come from this group AND that PCTs could avoid a substantial amount of the £6.6m expended on these admissions.

All of the clinical evidence set out in Appendix Three shows clearly that access to specialist multidisciplinary care delivers considerably better outcomes measured in terms of increased life expectancy, better quality of life and reduced incidence of medical crises.

An audit of 40 sequential Duchenne muscular dystrophy deaths over 10 years in the South West region showed a median age of death of 18 years. This compares with a mean age of death of almost 30 years in patients with Duchenne muscular dystrophy receiving specialist multidisciplinary care, and, as a consequence of that care, in receipt of home ventilation reported by the Newcastle group in the most recent study by Eagle et al (2007).[11]

Nationally 100 boys die each year with Duchenne muscular dystrophy – two every week - as a result of their condition. Because care is sporadic across the country, some will die in their teens. Some, in receipt of specialist multidisciplinary care, will survive to their late 20s.In Denmark where services are more comprehensive, they will survive to their mid 40s. The evidence is set out in more detail in the Appendix.

In the West Midlands, it is clear that there will be a similar disparity in survival rates given the 50% of patients who do not access specialist care and the pressure on current services to deliver comprehensive and consistent care to all people who can benefit from it. This is a significant issue of equality - a matter of luck about where you live in the West Midlands and how you are, or are not referred into specialist services

In summary, the cost benefit case rests on two issues:

- Significant improvement in patient outcomes in relation to increased life expectancy and better quality of life.
- Significant reduction in the expenditure by PCTs on unplanned emergency admissions to hospital.

Chapter 8:
Main Conclusions and Key Issues from Service Mapping in the West Midlands

- 50% of people do not access specialist multidisciplinary care at a main centre. Evidence shows that their outcomes and life expectancy will be reduced - see below.

- **IF** this 'missing' 50% - 2500 people with neuromuscular conditions - accessed specialist care current service capacity could not cope at all and would be overwhelmed completely.

- Even with current capacity the service is overloaded leading to long waits for follow-up appointments and potential deterioration in the patient's condition as a result.

- There is an unplanned historic development of specialist centres in the West Midlands.
    - There is something of a "hotch-potch" of services with no rationale about the location of and accessibility to services.
    - There is no uniformity of specialisms/specialists available in our centres.

- There is inequity of access to multidisciplinary specialist services - it is largely a matter of luck if a person has access to appropriate care.

- There is no succession planning or cover for overworked specialists who often work in isolation and keep services going through a large mount of goodwill.

- There is a serious risk to continuity of service in the West Midlands if the current fragile base of service is not strengthened.

- Services are not well co-ordinated either on a regional or an individual basis through the non availability of Neuromuscular Care Advisors/Coordinators (only one in the region) and the lack of a managed regional clinical network.

- Specialist and local services are not linked effectively - a network would enable these links to be developed.

- Skills across the system are not being developed - there is insufficient capacity to allow time for specialist staff to transfer aspects of care through training and skill development to local services.

- There is not an appropriate balance between specialist and local services.

- There is evidence that life expectancy could be extended significantly if services were developed on a better base and enhanced.

- There is evidence that PCTs are incurring avoidable expenditure on emergency admissions of people with muscular dystrophy due to the lack of appropriate care which could prevent most of these admissions. Hence there is a cost benefit case to implement the recommendations from this review

- A first priority must be both to protect existing levels of service and guarantee service continuity by having a viable workforce plan in place and ensuring that any first enhancement to existing services seeks to back up existing single-handed clinical staff.

- The future prospects for public finance would indicate that the NHS will have severe restrictions on its expenditure. Service such as neuromuscular will be vulnerable to arbitrary non filling of the very few current single handed clinical posts as any become vacant. The consequences would be non continuity of this vital, specialist service, a rapid decline in care standards leading to increased morbidities and a reduction in survival.

Chapter 9:
National and Regional Policy Context:

This work fits within the following strands of national policy:-

- *The National Service Framework for Long Term Conditions* - Aims to improve the lives of people living with neurological conditions by providing health and social care services. Severe, genetic or acquired life limiting neuromuscular conditions are multi-system disorders that require complex long term surveillance and care to optimise quality of life and life expectancy.[12]

- *The National Service Framework (NSF) for Children, Young People and Maternity Services* - Sets standards aimed at improving the quality of services and care available to children, young people and their families, and to reducing variations in health and social care. Standard 8 states "children and young people who are disabled or who have complex health needs receive coordinated, high quality child and family centered services which are based on assessed needs, promote social inclusion and, where possible, enable them and their families to live ordinary lives". In general, the NSF aims to ensure access to seamless care pathways to the range of health and social care services based on assessed need.[13]

- *Aiming High for Young People* - a ten year strategy for positive activities highlights the importance of considering social and educational opportunities for all young people.[14]

- *Better Care: Better lives* - Improving outcomes and experiences for children, young people and their families living with life-limiting and life-threatening conditions sets out a framework for end of life care support for children.[15]

- *High Quality Care for All: NHS Next Stage Review Final Report* - The final report of Lord Darzi's NHS Next Stage Review. It responds to the 10 SHA strategic visions and sets out a vision for an NHS with quality at its heart. [16]

This work also fits in with a number of national and regional policy drivers as follows:

- *The Walton Report* - Report of the All Party Parliamentary Group for Muscular Dystrophy. The result of a formal Inquiry into access to specialist multidisciplinary care for people living with muscular dystrophy and related neuromuscular conditions. Reviewing best practice, the report published in August 2009 brings forward a number of key recommendations to improve services across the country.[17]

- *Investing for Health Step 2: Delivering our clinical vision for a world class health service* – This report sets out the five to ten year vision of NHS West Midlands, the overall aims of which are as follows:
    - Rebalance a reactive and acute service focus to one which invests in prevention, prediction, early interventions and quality local services.
    - High quality care closer to home, delivered through genuine networks of acute, social and primary care providers.

- To help patients, carers and staff to navigate the system we will develop care pathways that move through every stage from prevention to tertiary care.
- Do the ordinary extraordinarily well. Best practice must be systematically implemented regionally and nationally.[18]

- *Clinical Pathway Report Summary for Long Term Conditions* - The work of the clinical pathway groups set up by NHS West Midlands following the publication of Investing for Health in July 2007. The report envisions "rapid diagnosis with early interventions, provided closer to home, with patients as lead partners in decisions."[19]

Chapter 10:
Service Vision, Service Standards and Service Model

We have looked at service visions, standards of care and service models elsewhere in the country

The South West Specialised Commissioning Team conducted a similar extensive review of neuromuscular conditions in their area. The South West Report refers to a service vision and standards of care that we believe the West Midlands should adopt.

A Service Vision

- The service will provide high quality of care for all individuals within the region for children and adults with attention to transitional care for young adults moving between children and adult services and effective support for families and other carers

- Patients will receive the majority of their care close to home. Specialist services will be available within the region which will support local provision where appropriate through managed clinical networks.

- A multidisciplinary team working at both specialist and community level will provide comprehensive care and support to all individuals and their carers.

- Close links with teams providing community health services and those providing social care and support for children and adults with disabilities in the localities will be essential. Attention to anticipatory care planning will help prepare for future needs.

- End of life care will provide support and care appropriate to the individual and family wishes.

Service Standards

Although specific aspects of a person's treatment are underpinned by specific standards of care (for example as those laid down in Treat-NMD - standards of care for Duchenne Muscular Dystrophy[20]) from a generic perspective the following key standards are commonly adopted in areas where a coordinated plan to improve care and outcomes for people with neuromuscular conditions has been put in place:

- **Standard 1** - guidance for primary care and referral pathways will be in place to ensure early recognition of families that may need expert help, rapid access to diagnostic services and specialist treatment and care. *NSF Quality Requirement 2*

- **Standard 2** - families will have access to a key worker, with regular monitoring of the person's condition and associated needs (e.g. health and social), including a multidisciplinary review at a specialist centre at least annually *NSF Quality Requirement 1*

- **Standard 3** - a patient's care will be lifelong care and should be continuous and seamless irrespective of age and whether or not transition is a part of the patient's care pathway. Lifelong care includes, where appropriate, enabling patients and their families to work with the multidisciplinary team to become expert at managing their chronic conditions because specialist care through a multidisciplinary team is the most effective way to extend life expectancy and improve quality of life. *NSF Quality Requirement 1*

- **Standard 4** - patients will have timely and appropriate access to support services such specialist physiotherapy and wheelchair services. *NSF Quality Requirements 4, 5, 7*

- **Standard 5** - support to the individual and their family in making plans and expressing their preferences for end of life care and help and support for the individual and their family; a proactive approach to supporting families following bereavement. *NSF Quality Requirement 9*

Elements of Care

The Muscular Dystrophy Campaign report Building on the Foundations: The Need for a Specialist Neuromuscular Service across England (December 2007)[21] sets out the following key elements of care:

- The care of all patients with a neuromuscular condition should be led from a regional Specialist Neuromuscular Centre with specialist multidisciplinary teams providing regular local clinics.
- Specialist multidisciplinary care may be lead by neurologists, clinical geneticists, paediatricians, paediatric neurologists or rehabilitation physicians. This specialist supervision supports and oversees local provision.
- Agreed standards of diagnosis and care should be developed and agreed and disseminated across all regions including to patients and patient groups to ensure equity of care.
- Provision of expert physiotherapy, early cardiac monitoring and intervention and corticosteroids to improve muscle function and maintain independent mobility.
- Judicious use of spinal surgery and expert respiratory services (including non invasive positive pressure ventilation to improve quality of life and delay the onset of respiratory failure to prolong life.

Service Model

We have looked at the appropriateness of a number of service models and looked for one that would better build on and reflect the particular way services have developed historically in the West Midlands.

In overall terms we are proposing a "hub and spoke" model for specialist neuromuscular services. We envisage three levels of service:-

Level 1:

Specialist Care provided at a main centre, fully equipped to carry out complex procedures such as muscle biopsies and early access to other specialist clinical services such as respiratory, cardiac and orthopaedic. Collectively the 4 main centres in Birmingham and Oswestry will form the service "Hub"

Level 2:

Multidisciplinary specialist outreach clinics to be developed in main population centres e.g. Coventry, Herefordshire, Worcestershire and Stoke-on-Trent. These outreach services, provided by specialists visiting from the Hub centres, will constitute the “Spokes”

Level 3:

Local community and primary care services e.g. physiotherapy and community nursing where skills can be developed with the assistance of specialist staff from the Hub centres in order to provide on going basic maintenance for people with neuromuscular conditions

In our model levels 1 and 2 will constitute a “single” **West Midlands Specialist Neuromuscular Service**. This service will operate from the collective Hub as a single clinical directorate.

Over time this will require joint contracts for staff working at the different main centres and flexibility of working between the centres that comprise the Hub and Spoke. This will ensure better value, better quality and better patient outcomes.

A regional managed clinical network will be established to manage and develop the Hub and Spoke model, ensure that local and specialist service network together effectively and provide confidence in commissioners that any new investment will be managed in ways that will deliver best value.

The proposals in this report are not advocating any service reconfiguration. Over time, as service capacity is increased to meet demand and as more of the specialist service outreaches into centres of population around the region, PCTs, Providers and Clinicians who make up the managed clinical network may take a view about the benefits, appropriateness and timing of any further consolidation and coordination of level 1 hub services perhaps bringing them together into a single centre in Birmingham.

The model would be supported by the following practice:-

- Clinical Network - a managed clinical network to support multidisciplinary and cross organisational working to provide effective and efficient treatment, care and support to patients and their families.

- Shared care with services delivered as close to home as possible as well as access to a specialist centres in the West Midlands, supported by multidisciplinary team working - each person will be supported by an individualised package of care across primary, secondary and tertiary services.

- Individualised care supported by ongoing care coordination from the point of diagnosis.

- Close working between paediatric and adult services to ensure smooth transition between services. Neuromuscular Care Advisors/Coordinators will play a key role in both initiating the best time for transition in consultation with the multidisciplinary team, but also continuing to support the patient, thus ensuring continuity of care.

Chapter 11: Recommendations

It is recommended:

- **That** PCTs sign up to the recommendations and investment plan set out in this report.
- **That** the Specialised Commissioning Group assumes responsibility for the commissioning of specialised neuromuscular services in line with the national decision to designate these services as specialised services. These services include the main neuromuscular centres and the multidisciplinary teams that operate within them.
- **That** the Specialised Commissioning Group enhance infrastructure within the Specialised Commissioning Team to develop and manage the regional contract for these services and associated work, including develop a service specification including agreement on outcome measures, clinical standards and performance monitoring.
- **That** all people with muscular dystrophy and related neuromuscular conditions have equal access to specialist multidisciplinary services at a main specialist centre.
- **That** a Hub and Spoke model of service delivery be adopted with specialised services supporting local provision.
- **That** a managed regional clinical network be established to coordinate specialist and local services, to be the central office for the Hub where a regional database, quality audit and research activity can be managed and to develop and project manage an implementation in respect of the recommendations in this report.
- **That** a network manager be appointed together with administrative support and data input facilities.
- **That** the HR arrangements are put in place immediately to secure the appointment of the Network Manager from the 1st April 2010. In the meantime it is proposed that SCG put in place interim arrangements for a Network Manager, possibly via a secondment, to ensure essential planning takes place.
- **That** current specialist neuromuscular services be brought together into a single clinical directorate and function as one single West Midlands Regional Neuromuscular Service. That arrangements are put in place for all consultant posts to be joint and shared consultant contracts between main centres with all contracts held by one of the main centres. Details to be worked out and agreed by the Regional Network.
- **That** one of the current neuromuscular main centres hosts the network and provides, via the network, the management coordination of the regional service and clinical directorate. This centre will also hold future joint consultant contracts. This should be either Heart of England NHS Foundation Trust or University Hospitals Birmingham NHS Foundation Trust.
- **That** in the short term the priority is given to the stabilisation of current services by increasing capacity in some key areas revealed by the service review and detailed elsewhere in this report. This will involve an increase in specialist consultant staff, and the multidisciplinary team.

- **That** existing capacity in main centres (the Hub-Tier 1) be enhanced to meet the latent unmet demand and allow the development of outreach services (spokes-Tier 2) and outreach clinics commencing in Coventry, Worcester, Stoke and Hereford that will provide more local access to sub areas of the region.

- **That** 4 Neuromuscular Care Advisors/Coordinators should be appointed and be based sub-regionally to support local patients and coordinate the care pathway.

- **That** earlier access to diagnosis be a priority. Education and training of primary care and community health staff backed up with clear referral protocols would support this aim.

- **That** specialist muscle pathology services be strengthened with an appropriate manpower plan to assure succession planning and service continuity. That priority is accorded to the replacement of the clinical scientist post in Oswestry with the appointment of a specialist Consultant Muscle Pathologist who can network with the other two muscle biopsy centres in Birmingham and lead on the development of a coordinated regional muscle biopsy service where cover, support and shared work will greatly strengthen this critical service.

- **That** capacity in the Hub centres is developed to enable specialist staff f to train and pass on enhanced skills to staff in local services (Tier 3) e.g. physiotherapy, speech therapy, community nursing etc.

- **That** opportunities for research be pursued to allow West Midlands patients to access clinical trials and to allow consultants and other staff to develop their expertise and interests.

- **That** a workforce plan is developed to aid succession planning and to guarantee in such a small but specialist service, continuity of service.

- **That** an end of life strategy is developed with the full involvement of all stakeholders.

- **That** an in-depth review of the provision of specialist wheelchairs is undertaken with a view to SCG agreeing a formal process for PCT collaboration in the commissioning of specialist wheelchairs to eliminate long waiting times.

- **That** a task group is established with social care colleagues to examine and come forward with proposals in relation to ways in which collaborative long term planning of care plans for individuals with neuromuscular and other long term deteriorating and disabling conditions can be achieved.

- **That** hydrotherapy facilities, where provided or commissioned by the NHS, be made available to people with neuromuscular conditions on an equal basis to people with other disabling conditions.

- **That** interested parties are engaged in a feasibility study in relation to the possible development of a specialist neuromuscular physiotherapy centre along the same lines as the national centre in Cheshire, established as a social enterprise bringing together a partnership of the NHS, voluntary and business sectors. As well as providing specialist physiotherapy services the centre could generate income through the provision of commercial activities by people with a neuromuscular condition e.g. graphic design. In this way such a centre could offer more innovative day and respite care support.

Chapter 12:
Investment and Implementation Plan

The first priority is to formally establish a regional managed clinical network with the appointment of a network manager, administrative support and data input support. As well as managing the coordination and development of the service, the network will have responsibility to develop and project manage a more detailed implementation plan.

In parallel with this the priority will be to commence the appointment process for the additional key clinical staff to ensure that service continuity ceases to be a risk and that service capacity can be enhanced to meet unmet demand.

The next order of priority will be to further enhance clinical and practitioner capacity in order to be able to both meet the currently access specialist multidisciplinary care) and to enable outreach spoke specialist spoke services to be established in Coventry, Worcester and Hereford initially.

On the basis of comparison with the neuromuscular service in the North East, widely recognised as having a good solid base of service that meets demand, and on the basis that an acceptable workload is 500 patients per WTE Consultant then the long term plan in the West Midlands is to have 10 Consultants. Implementation of the recommendations in this report will bring the Consultant establishment up to 5.5 WTE.

In view of the recommendation that a single clinical directorate for specialist neuromuscular services be created, it would perhaps be appropriate for one of the current main centres to hold what will be the joint contracts in respect of the 4 additional Neuromuscular Consultants and the regional network management staff.

Key new posts to achieve the short/medium term objectives and which need to be actioned now are:

- 2 Consultant Post –children
- 2 Consultant Posts-Adult
- 1 Consultant Muscle Pathologist
- 4.5 Care Coordinators (including pick up of charitable funding for 0.5wte of existing care coordinator
- 3 Specialist Physiotherapists
- 2 Psychologists
- 1 Network Manager
- 1 Network Admin Support
- 1 Network data input clerk

Investment Plan

Investment (provisional figures requiring validation)	**£'s Recurring**
1 Network Manager (band 7 - mid range) 1 Administrator (band 5 - mid range) 1 Data Input Clerk(band 3 mid range)	£43,012 £29,181 £20,872
Network non-pay support costs for clinical. IT equipment and software for data base. Travel expenses /sundry. Patient & Carer involvement x 3 (1for paediatric, Adult and Transition) Training & Development.	£30,000 £10,000 £20,000 £15,000 £20,000
2 WTE Paediatric Consultant specialising in Neuromuscular conditions (inc. secretarial support), or equivalent sessions to attract part-time posts.	£260,000
2 WTE Adult Consultants specialising in Neuromuscular conditions (inc. secretarial support), or equivalent sessional time to attract part-time posts.	£260,000
1 WTE Consultant Pathologist (including secretarial support)	£130,000
4.5 WTE Care Coordinators (Band 7 mid range –incl .5 wte pick up of charitable funding for current post	£193,554
3 WTE Specialist Physiotherapists (Band 7 mid range)	£129,036
2 WTE (in sessional time) Psychologists(mid grade band 8a)-incl pick up of Oswestry 0.5 in April 2011	£102,132
TOTAL INVESTMENT REQUIRED (full year cost)	**£1,262,796**

In view of the national decision to designate the specialised aspect of neuromuscular services as a regional specialised service, the £1.26m investment required has been divided between the 17 PCTs pro rata to their start year revenue allocation. This is set out in the table below.

Primary Care Trust	**Cost per PCT**
Birmingham East and North	£100,862
Coventry	£79,243
Dudley	£69,114
Heart of Birmingham	£78,320
Herefordshire	£38,420
North Staffordshire	£47,319
Sandwell	£78,326
Shropshire County	£61,730
Solihull	£43,992
South Birmingham	£87,874
South Staffordshire	£123,622
Stoke on Trent	£67,536
Telford and Wrekin	£35,533
Walsall	£63,614
Warwickshire	£110,694
Wolverhampton City	£61,128
Worcestershire	£115,468

In terms of timing and phasing of the programme the following considerations should be noted:

- The absolute first priority is to remove the risk to continuity of current services is funding to increase paediatric and adult consultant sessions, the appointment of a Consultant Pathologist with a special interest in muscle disease and the picking up of charitable funding for a 0.5 WTE neuromuscular care advisor and increasing the numbers of neuromuscular care advisors to ensure that patients and families receive vital support across all seventeen PCT areas.

- The establishment and staffing of the regional clinical network is essential infrastructure to shape and manage the service developments set out in the report.

- For financial planning purposes approximately 50% of the funding agreed would be required in 2010/2011 with the balance to full year cost in 2011/2012.

Appendix 1: Neuromuscular Conditions

The term 'neuromuscular conditions' is used to describe a group of mostly genetic disorders, which are generally progressive. They cause loss of muscle strength and sometimes this deterioration can happen quickly.

There are over 60 types of neuromuscular condition, including muscular dystrophy. The age of onset varies between, and within, conditions. Some conditions, such as Duchenne muscular dystrophy, always begin in childhood while others start to affect individuals in adulthood.

BECKER MUSCULAR DYSTROPHY

Becker muscular dystrophy is an X chromosome condition. It is a milder variant of Duchenne and those boys/men with the condition experience similar problems to those with Duchenne. The condition varies in severity. It can be almost as severe as Duchenne or mild enough to only be diagnosed later in adult life. Some of those with Becker muscular dystrophy will lose the ability to walk in early adult life, but others remain ambulant into middle age and beyond.

CHARCOT-MARIE-TOOTH DISEASE, ALSO KNOWN AS HEREDITARY MOTOR AND SENSORY NEUROPATHY

Charcot-Marie-Tooth disease has a variable inheritance pattern. It may first be noticed in childhood, affecting the small muscles of the hands (and fine motor movements) and feet (high arches, foot drop and 'club foot' may be symptoms).

Some people with the condition experience mild weakness and may not even be aware they have the condition, while others are severely affected and have great difficulty in walking.

CONGENITAL MUSCULAR DYSTROPHIES

These are a collection of different muscular dystrophies characterised by weakness at birth or soon after. Occasionally a congenital muscular dystrophy may be diagnosed a little later.

The severity of the condition depends on the type of congenital muscular dystrophy diagnosed. Early life problems include floppiness (hypotonia), poor head control, contractures, respiratory problems, swallowing and feeding difficulties. Some children may also have learning difficulties. While many children are never able to walk, others do achieve delayed walking but lose this ability as they grow older.

CONGENITAL MYOPATHIES

This group of conditions usually causes muscle weakness in children, although in some cases there are no symptoms until adulthood. Respiratory problems are a common feature and scoliosis, cardiac problems and contractures can also be factors, depending on the type of myopathy.

DUCHENNE MUSCULAR DYSTROPHY (DMD)

Duchenne muscular dystrophy is a serious condition and the most common of the childhood onset muscular dystrophies. It is caused by a fault on the X chromosome so the condition only affects boys, although their mothers may be carriers. About 100 boys with Duchenne are born each year in the UK and, at any one time, approximately 1500 boys are living with the condition. The risk for the general population of having an affected child is 1:3500 male births.

Duchenne is often, although not always, characterised by late walking (after 18 months). Further early signs include calf hypertrophy (enlarged calves) and muscle weakness in the lower limbs causing loss of balance. An affected child also finds it difficult to get up from the floor or use stairs. As the condition progresses, a distinctive walk emerges; to compensate for the increasing weakness in the hip and pelvic muscles the boy will walk on his toes with his abdomen pushed forward. A child may also have contractures (stiffness) of the heels and ankles, which may require surgery.

Some boys with Duchenne have learning difficulties, particularly with language and communication skills. These difficulties are rarely severe and do not worsen over time.

Boys with Duchenne lose their ability to walk, usually after the age of nine, and then become full-time wheelchair users. From this point onwards, they may experience scoliosis (curvature of the spine), cardiac problems, weakness of the shoulders, arms and hands, chest infections and, at a later stage, respiratory problems. Life expectancy is reduced.

In the past, most boys died in their late teens. Today, with assisted non-invasive ventilation, they can survive into their late twenties and beyond, which poses new long-term management issues.

FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY (FSH)

FSH is an inherited condition that can affect men and women. The first signs are usually weakness in the face and shoulder muscles which make it difficult for the individual to raise his or her arms. A ‘winging’ of the shoulder blades is also apparent.

Weak facial muscles can affect speech, communication and feeding. The muscle which raises the foot is often affected early on in the condition - causing people to trip – along with the lower (distal) leg. This weakness can then spread to the larger hip girdle muscles. The rate at which the condition progresses is variable, although it is usually true that the earlier the symptoms, the more severe the eventual muscle problems. A minority of people will experience complete loss of walking, but others may also need a wheelchair for long distances and to prevent fatigue. Some people may have hearing loss.

INCLUSION BODY MYOSITIS

This is not an inherited condition. Inclusion body myositis is the most common form of muscle weakness acquired in later adult life. It causes substantial disability and is characterised by weakening muscles in the hands and thigh, which can cause falls. Swallowing may also be affected. Although other forms of myositis (muscle inflammation) respond to treatment, it is generally ineffective with this form of the condition.

LIMB GIRDLE MUSCULAR DYSTROPHIES (LGMD)

These are a group of progressive muscle conditions affecting both males and females. The limb girdle group of muscular dystrophies are so called because they usually cause weakness in the shoulder and pelvic girdle. Weakness in the legs generally occurs before weakness in the arms.

The muscles of the face are usually unaffected. The condition progresses at a variable rate. Some children will be severely affected while others will not be diagnosed until adulthood. Some forms of limb girdle muscular dystrophies affect the heart and breathing.

METABOLIC MYOPATHIES

Contracting a muscle requires energy. The body metabolises the food eaten into a form of energy the muscle can use. Many inherited disorders affect the metabolic pathways. Some, such as McArdle's disease or mitochondrial myopathies, cause exercise intolerance (the person has no symptoms at rest but develops muscle pain and weakness if he or she attempts activity). Others, such as Pompe's Disease or debrancher enzyme deficiency, cause progressive weakness regardless of whether exercise is undertaken or not. Some metabolic myopathies also affect the heart and respiratory muscles.

MYOTONIC DYSTROPHY AND CONGENITAL MYOTONIC DYSTROPHY

Myotonic dystrophy and congenital myotonic dystrophy are dominantly inherited conditions that tend to increase in severity from one generation to the next. The majority of those affected will begin showing symptoms in early adulthood but it is not uncommon for children of affected mothers to have the more serious congenital form of the condition. Both men and women are equally likely to be affected.

A characteristic feature of these conditions is the myotonia or muscle stiffness (a delayed relaxation of the muscle following contraction) which is often worse in cold weather and more of a nuisance than a disability. More problematic, however, is muscle weakness in the hands, ankles, face and neck. Affected individuals may also experience smooth muscle problems causing trouble with the gut (pain, similar to that experienced in Irritable Bowel Syndrome, has been known to occur).

A significant feature of the conditions, particularly congenital myotonic dystrophy, is learning difficulties, which can be severe. People with myotonic dystrophy may also exhibit sleepiness; tiredness, lethargy and cataracts at an unusually early age are not uncommon. The heart may also be affected and cardiac monitoring is recommended for both adults and children. It is unlikely, however, that an individual would have all the symptoms and problems associated with such a variable and complex condition.

SPINAL MUSCULAR ATROPHY (SMA)

SMA is a recessively inherited condition that causes muscle weakness. It affects both boys and girls equally. The severity of the condition depends on the type of SMA and age of onset.

SMA type I is the most severe form of the condition. Children with type I are very weak and lack motor development. They cannot sit unaided and have difficulty breathing, sucking and swallowing. Most do not survive beyond their first birthday.

SMA with Respiratory Distress Type 1 (SMARD1) causes muscle weakness but the predominating symptom is severe respiratory distress due to paralysis of the diaphragm. Babies between one month and six months old experience respiratory failure and progressive muscle weakness, mainly in the distal lower limbs. Sensory and autonomic nervous systems may also be involved. SMARD1 is distinguishable from SMA Type I by the paralysis of the diaphragm and distal muscle weakness. Infants with SMA Type I become floppy due to weakness of the proximal limb muscles, assuming a 'frog leg' position, before they suffer respiratory failure due to paralysis of intercostals muscles.

SMA type II is slightly less severe than SMA type I. Children with type II can sit unaided and even stand with support, but cannot walk. They do not usually have difficulties with feeding and swallowing but are at increased risk of complications from respiratory infections. Some children will not live into adulthood. SMA type III is milder than SMA type II. Children with type III can stand and walk. They may outgrow their muscle strength and many do eventually need to use a wheelchair. SMA type III affects children after 18 months of age.

SMA type IV is the least common form of the condition and usually begins in late adolescence or adulthood. It has a similar clinical course to Becker muscular dystrophy and was often misdiagnosed as such in the past.

Appendix 2: Prevalence

The below table sets out the prevalence of neuromuscular conditions in the U.K[22]:

Group of conditions	Patient Number in UK	Percentage of total (%)
Muscular dystrophies	8,000-10,000	12.8
Myotonic disorders	7,500	11.0
Congenital myopathies	1,000	1.5
Distal myopathies	300	0.4
Mitochondrial myopathies	3,500	5.1
Metabolic myopathies	700	1.0
Periodic paralysis	900	1.3
Myositis	5,000-6,000	7.9
Spinal muscular atrophies (SMA)	1,200	1.7
Hereditary Neuropathies	23,000	33.0
Inflammatory and autoimmune neuropathies	6,400	9.2
Disorders of the neuromuscular junction	10,500	15.0
Myositis ossificans progressiva (MOP)	60	0.1
Total number (prevalence)	**68,060-71,000**	**100**

Appendix 3:
Evidence Base for Multidisciplinary Specialist Services

There is evidence that when people living with severe disabling and/or life limiting neuromuscular conditions have access to the appropriate interventions and support as their condition progresses not only have a better quality of life but also have their life expectancy lengthened

An audit of 40 sequential Duchenne muscular dystrophy 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 Duchenne muscular dystrophy receiving home ventilation and specialist multidisciplinary care reported by the Newcastle group in the most recent study by Eagle *et al* (2007).[23]

Multidisciplinary specialist input must include:

Medical	MDT Team
Neuromuscular	Physiotherapy
Respiratory	Nurse specialist
Cardiac	Care advisor
Pathology	Psychology
Orthopaedic/spinal	Dietetics
Genetics	Occupational therapy
Metabolic	Speech therapy
	Social work/education advisor

It is evident from a number of studies that better outcomes in terms of increased life expectancy have been achieved as a direct result of specialist multidisciplinary care delivered within a managed clinical network of specialist centres and coordinate local services.

Best practice shows that essential, specialist services should be delivered by a range of professionals from primary, secondary and tertiary providers working within a managed clinical network.

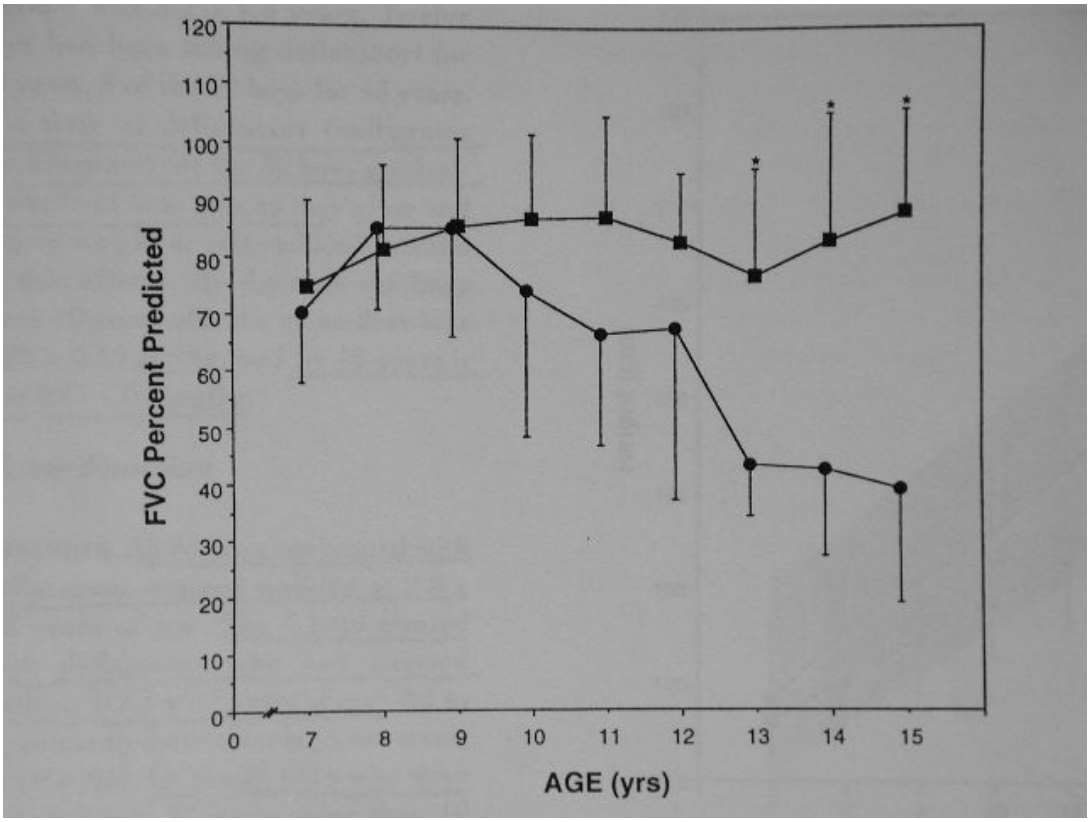
Respiratory Services

Respiratory monitoring and support is an essential part of multidisciplinary specialist neuromuscular care - roughly 90% of deaths among untreated Duchenne muscular dystrophy are attributable to respiratory causes.[24] Studies have shown that non-invasive ventilation and other technological respiratory treatments can extend life expectancy and improve quality of life.

The graph below shows the impact of steroid treatment on preserving lung function in Duchenne muscular dystrophy

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]

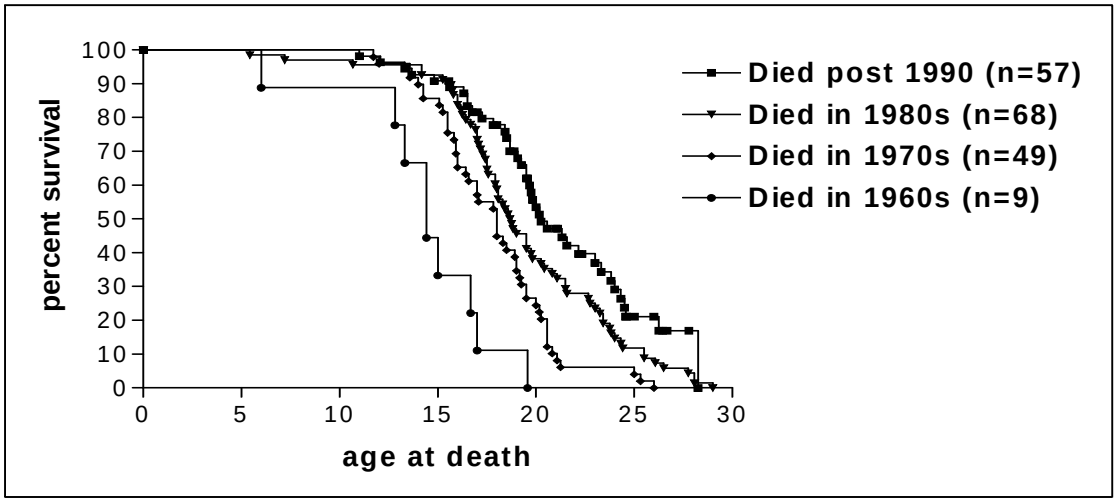


The article compares the clinical course of 74 boys 10-18 years of age with Duchenne muscular dystrophy (DMD) treated (40) and not treated (34) with deflazacort.

Results:
Deflazacort group: 88% (± 18%)
No treatment Group 39% (± 20%)

The importance of correct diagnosis leading to the correct early intervention is demonstrated by the graph above. Further evidence shows that when ventilation plays a part in multidisciplinary care life expectancy is increased further. [26]

The following graph shows life expectancy extended to 25.3 years in 2000 compared to 19years in 1990:



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

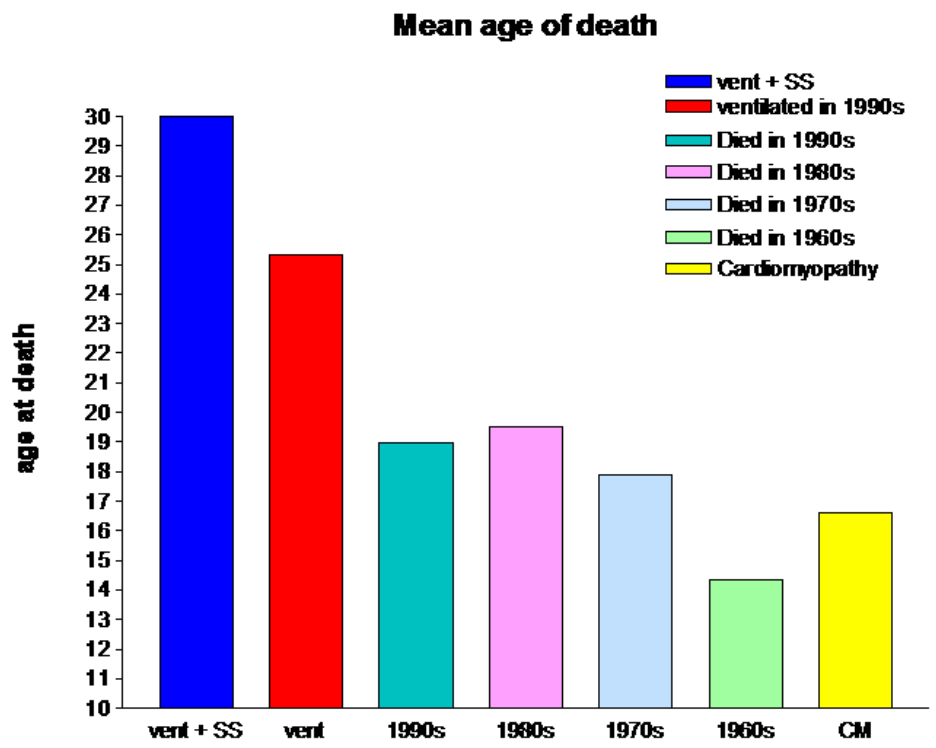
Indeed life expectancy is increasing now to the late 30s for some patients with Duchenne muscular dystrophy and cases are reported of patients aged 40 and above.

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.

Spinal Surgery

Spinal Surgery is an important service for people with neuromuscular conditions and needs to be made available as a preventive form of investment. The evidence is that cardiology screening should take place before any surgery and that regular screening should be done

Studies such as that carried out by Eagle et al 2007[27] on the combined additive effect of spinal surgery and ventilation show a significantly improved life expectancy when these services were available along the care pathway:



In a peer-reviewed study from 2007, the Newcastle Centre's Michelle Eagle *et al* highlighted that patients with Duchenne muscular dystrophy treated at the Newcastle Centre and receiving home ventilation had a mean age of death of almost 30 years.[28] The survival rate has continued to improve.

Cardiology Services

As a number of neuromuscular conditions affect the heart, cardiac monitoring should be recognised as an essential part of a multidisciplinary 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.[29]

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".[30]

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.[31] Without screening, cardiomyopathy can progress almost entirely without symptoms until signs of heart failure emerge, when all cardiac reserve has been eroded.[32]

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.[33]

Evidence shows the beneficial effect of proactive cardiac management. In terms of effective cardiac protection, a randomised placebo control trial (Duboc et al 2005, 2007) in relation to the prescribing of Perindopril showed that administration of this medication to people with DMD delayed the onset of left ventricular failure.[34]

Silversides et al showed that 5 years treatment with Deflazacort preserved heart and lung function:[35]

	Control group	Treatment group
FVC (lung function)	41%	83%
Mean Fractional Shortening (normal>28%)	21%	33%

Physiotherapy

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."[36]

The non availability of specialist physiotherapy can and will result in people with these conditions having problems such as respiratory crises that could lead to an emergency hospital admission with associated costs (see below) that could have been avoided

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 dieticians and speech and language therapists has impacted on the morbidity of children with neuromuscular conditions, and can have substantial impact on quality of life.[37]

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.

Psychology Support

Psychology support has been identified as an important aspect of multidisciplinary care.[38] Children and adults with neuromuscular conditions would benefit from the input of a clinical psychologist to help families develop management strategies.[39] Specific issues include support at the time of diagnosis, chronic illness, loss of ambulation, transition to adulthood, times of crisis and bereavement.[40]

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.[41] 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.[42] 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.

Rehabilitation

Neuromuscular specialist rehabilitation clinics that aim to help adult 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.

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.[43]

The care programmes developed and implemented for young people should include the arrangements that will be in place to ensure a safe transition from paediatric to adult services, at an appropriate time. This time will be determined by the individual young person's development. It is anticipated that transition may take place between the years of 16 to 18 as reflected by the requirements of the Children's Act (1989), the need to safeguard children (the Children's Act 2004) and the standards set out in the National Service Framework for Children, Young People and Maternity Services (2004).

End of Life Care

Care for someone in the terminal stages of a progressive chronic illness focuses on sustainment of quality of life for the patient and their family. An interdisciplinary approach is required, including primary and specialist physicians, hospice/palliative care specialists, social services, and psychological care, family members, and others appropriate to the patient's cultural, religious and spiritual background.

The goals of end of life care for patients with a neuromuscular condition include:

- Treating conditions (pain, dyspnea) that cause distress (palliative care).
- Attending to the psychosocial and spiritual needs of the patient and their families.
- Respecting the patient and family's choices concerning testing and treatment.

Coordination of Care

As early as 2005 the Department of Health recognised that: “Some people with more complex needs requiring skilled multidisciplinary 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).

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. They also help to coordinate community-based services such as occupational therapy and social care support and signpost service users to benefits advice or employment support, for example.

Equipment

Children and adults should not be forced to wait for essential specialist wheelchairs and community equipment in view of the complex and at times life limiting nature of their conditions. The Department of Health has included specialist wheelchairs in the Specialised Services National Definition Set.[44]

PCTs should collaborate to provide specialist wheelchair services. Children and adults affected by these rare and progressive conditions should not be 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.

Managed Clinical Networks

Most best practice models are based on a “managed clinical network” model. Managed clinical networks are linked groups of health professionals and organisations from primary, secondary, and tertiary care, working in a coordinated manner that is not constrained by existing organisational or professional boundaries to ensure equitable provision of high quality, clinically effective care.

These networks are seen as a vehicle for providing an integrated approach along the continuum of care from community to acute care settings, within which shifts in the balance of care would be possible. The emphasis within a managed clinical network shifts from buildings and organisations towards services and patients.

A managed clinical network would aim to improve patient care in terms of quality, access and appropriateness.

Appendix 4:
Best Practice: Models of service elsewhere in the United Kingdom and internationally:

- Historically a number of regions have seen the development of specialist centres of excellence based on university research departments or existing specialist centres - Newcastle, London, Oxford and Oswestry.

- In Scotland a managed clinical network has been developed that provides support from lead centres to the clinical teams across the rest of the country. These arrangements form the Scottish Muscle Network. The geographical challenges and population demographics are similar in Scotland to those experienced in the West Midlands. Thus it will be appropriate to consider the lessons learned in Scotland when developing the service model for the West Midlands.

- Over the last few years diagnosis and ongoing management of neuromuscular conditions have improved leading to the increasing life-span of individuals, so that patients who had previously only survived into their mid teens are now living into their 30s and, in some cases, beyond.

- The introduction of standardised care that includes the use of steroids, respiratory support, orthopaedic interventions and multidisciplinary care as well as technological development, have all contributed to this improvement in survival rates. However, increasingly complex care packages and comprehensive local support for patients and their families requires specialist support.

- The Treat NMD consortium – a European Union Network of Excellence for the development of translational research in rare neuromuscular diseases has created international consensus on care and management for patients with Duchenne muscular dystrophy and Spinal Muscular Atrophy. Treat NMD is an international initiative bringing together some of the world's leading neuromuscular specialists in a pan-European 'network of excellence' aimed at improving treatment and finding cures for patients with neuromuscular disorders.[45]

- Further afield, in Quebec, the Conseil d'Evaluation des Technologies de la Sante du Quebec considers that the conditions are in place for recommending that a service be instituted for the genotypic diagnosis of patients with Duchenne or Becker Muscular Dystrophy. The service would include screening for women carrying a mutation on the dystrophin gene and, if necessary, the prenatal diagnosis of the disease.[46]

- International Guidelines recommend cardiac care in patients with Duchenne Muscular Dystrophy (DMD) or Becker Muscular Dystrophy (BMD), which includes details of the cardiac assessment and clinical management by a cardiac specialist with an interest in the management of cardiac dysfunction and/or neuromuscular disorders.[47]

- The Neuromuscular Centre in Winsford: The Neuromuscular Centre (NMC) based in Cheshire provides ongoing specialist physiotherapy, in addition to

training and employment within a small social enterprise. There are currently 360 registered patients at the NMC who, following a detailed assessment, are provided with an individual treatment programme which targets their main problems and deals with the areas that cause them most concern. There is also an advice and information service.

A number of physiotherapy treatments are offered at the NMC including:

Passive stretches	Hydrotherapy
Assisted standing	Mobilisation techniques
Active exercises	Acupuncture for pain relief
Core stability exercises	Intermittent compression
Walking re-education	Electrotherapy

The benefits of the specialist physiotherapy are clear: 100% of service users said NMC physiotherapy treatment helped them stay out of hospital and 80% of those who are able to walk said that the physiotherapy had helped them to stay on their feet and walking.

In addition to physiotherapy, the NMC provides training and employment through a small, user-led social enterprise which it hosts, NMC Design+Print. It has recently been awarded the Social Enterprise Coalition's Best Social Enterprise, Enterprising Solutions 2008 award.[48]

- The North Star project aims to optimise the care of young patients with Duchenne muscular dystrophy (DMD) by achieving and practicing consensus on best clinical management, with agreed assessment and treatment protocols, no matter which clinical centre they attend. Clinical Leader for the Project is Dr Adnan Manzur, Consultant Paediatrician with a Special Interest in Neurology, based at the Dubowitz Neuromuscular Centre, Great Ormond Street Hospital, London. The project is coordinated by Elaine Scott, research physiotherapist.

  Part of the North Star work has been to develop a standardised physiotherapy assessment protocol for ambulant children with DMD via a lengthy process of consensus and review. Tests include assessment of functional abilities, muscle strength and pulmonary function. Both medical and physiotherapy assessment proformas have been produced for these children.[49]

[1] West Midlands: Building on the Foundations: the need for a neuromuscular service across the West Midlands (April 2009)
[2] Spelman, C Westminster Hall debate – Muscular Dystrophy 3 Nov 2009 : Column 234WH
[3] Muscular Dystrophy Campaign State of the Nation (2008)
[4] Muscular Dystrophy Campaign State of the Nation (2008)
[5] 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.
[6] Muscular Dystrophy Campaign State of the Nation (2008)
[7] Muscular Dystrophy Campaign State of the Nation (2008)
[8] Muscular Dystrophy Campaign State of the Nation (2008)
[9] Muscular Dystrophy Campaign Equipment Shortfall: How disabled children are being failed (2009)
[10] Muscular Dystrophy Campaign Equipment Shortfall: How disabled children are being failed (2009)
[11] 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.,
[12] DH The National Service Framework for Long Term Conditions (2005)
[13] DH The National Service Framework (NSF) for Children, Young People and Maternity Services (2004)
[14] HM Treasury and DCSF Aiming High For Young People (2007)
[15] DH Better Care, Better Lives (2008)
[16] DH High Quality Care for All: NHS Next Stage Review Final Report u(2008)
[17] All Party Parliamentary Group for Muscular Dystrophy The Walton Report (2009)
[18] NHS West Midlands (2008)
[19] NHS West Midlands (2008)
[20] Treat NMD Standards of Care for Duchenne Muscular Dystrophy (2008)
[21] Muscular Dystrophy Campaign Building on the Foundations: The Need for a Specialist Neuromuscular Service across England (2007)
[22] Muscular Dystrophy Campaign (2009)
[23] 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.,
[24] Benditt J O. Management of pulmonary complications in neuromuscular disease. Physical Medicine and Rehabilitation Clinics of North America 1998; 9(1): 167-185
[25] 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 2006;16:249-5
[26] 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.
[27] M. Eagle, J. Bourke, R. Bullock, M. Gibson, J. Mehta, D. Giddings, V. Straub, K. Bushby (2007) Managing Duchenne muscular dystrophy – The additive effect of spinal surgery and home nocturnal ventilation in improving survival Neuromuscular Disorders, Volume 17, Issue 6, Pages 470-475.
[28] 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.,
[29] Muscular Dystrophy Campaign Factsheets (2009)
[30] 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.
[31] 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.
[32] 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.
[33] 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.
[34] Denis Duboc, Christophe Meune, Bertrand Pierre, Karim Wahbi, Bruno Eymard, Annick Toutain, Carole Berard, Guy Vaksmann, and Henri-Marc Be´cane Perindopril preserves left ventricular function in X-linked Duchenne muscular dystrophy European Heart Journal Supplements (2007) 9 (Supplement E), E20–E24
[35] Silversides et al Effects of deflazacort on left ventricular function in patients with Duchenne muscular dystrophy The American Journal of Cardiology, Volume 91, Issue 6, Pages 769-772
[36] Muscular Dystrophy Campaign Focus on Physio (2008)
[37] 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.
[38] Bothwell JE et al, Duchenne Muscular Dystrophy—Parental Perceptions, Clinical Pediatrics, Vol. 41, No. 2, 105-109 (2002)
[39] Yoshimura I, A case of Congenital myotonic dystrophy with infantile autism, 1989 Jul;21(4):379-84
[40] Reid, D T & Renwick R M, Relating familial stress to the psychosocial adjustment of adolescents with Duchenne muscular dystrophy, International Journal of Rehabilitation Research: Volume 24(2)June 2001pp 83-93
[41] 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.
[42] Manzur, AY. Kuntzer, T. Pike, M. Swan, AV. Glucocorticoid corticosteroids for Duchenne muscular dystrophy (Cochrane Review) The Cochrane Library 2009, Issue 1
[43] 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.
[44] DH SPECIALISED SERVICES NATIONAL DEFINITIONS SET (2nd Edition) The Assessment and Provision of Equipment for People with Complex Physical Disabilities (all ages) - Definition no. 5 (2001/2002)
[45] Treat NMD http://www.treat-nmd.eu/
[46] Conseil d'Evaluation des Technologies de la Sante du Quebec. Duchenne and Becker muscular dystrophies: family screening and molecular diagnosis - nonsystematic review. Conseil d'Evaluation des Technologies de la Sante du Quebec (CETS), 1997; CETS 97-1 RE: 56
[47] 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.
[48] The Neuromuscular Centre www.nmcentre.com
[49] Dubowitz Neurtomuscular Centre: SMArtNet and The North Star Project http://www.ich.ucl.ac.uk/gosh/clinicalservices/neuromuscular_services/smartnet_and_the_north-star_project