

Muscular Dystrophy Campaign



Building on the Foundations: The Need for a Neuromuscular Service serving patients in the NHS Yorkshire and Humber Region July 2009



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

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Executive summary:

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

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

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

Action needed:

- **A short life working group should be established to carry out an in-depth review of current service provision and its vulnerability in Yorkshire and Humberside. This review would involve families, clinicians, PCTs and the Specialised Commissioning Group (SCG), and would bring forward proposals in autumn 2009 to secure and develop the comprehensive, multi-disciplinary service for children and adults, including transition services for young people.**
- **A neuromuscular network should be established on the model of a managed clinical network. This will ensure co-ordination between the existing clinics and ensure that expertise is shared with all clinicians and Allied Health Professionals (AHPs) in each PCT in the region. The network should be supported by a Network Coordinator. This model has been agreed in the South West and has been successful in Scotland with the Scottish Muscle Network. It also reflects the model set out in the National Definition for specialist neuromuscular services.**
- **Five full-time Regional Care Advisors with expertise in muscular dystrophy and related neuromuscular conditions should be established and embedded in the NHS to serve the estimated 5,000 people in the area living with these conditions.**

- Ongoing physiotherapy should be provided to all adults and children with a neuromuscular condition in each PCT, supported and developed in each PCT area by enhanced specialist physiotherapy support from the specialist clinics.
- Psychological support should be provided as part of a multi-disciplinary approach to care for individuals and family members living with a neuromuscular condition in each PCT area across Yorkshire and Humberside.

Our key findings include:

- **Of the 100 boys with Duchenne muscular dystrophy born between January 1980 and December 1999 in the region, there have been 34 deaths. The mean age of death is 17.2 years – this compares with 30 years in the North East. Five deaths were boys under 15 years with cardiomyopathy. Four of these had had no drug treatment.**
- Specialist neuromuscular services are vulnerable due to their reliance on charitable funding. The region's only Care Advisor¹, based in Leeds, is partly funded by the Muscular Dystrophy Campaign and we are calling for this vital post to become fully embedded in the NHS from April 2010.
- Two thirds of patients and their families have no access to a key worker or care co-ordinator. Five full-time Regional Care Advisors are needed to serve the 5,000 children and adults in the area with a neuromuscular condition, many of whom are simply not known to providers of health services.
- Patients have very limited access in particular to ongoing physiotherapy.² Specialist physiotherapists are required to support outreach clinics and provide training and professional development for community physiotherapists. Two out of five patients in Yorkshire and Humberside feel that they do not receive enough physiotherapy.
- There is no dedicated psychology service for neuromuscular patients, despite its importance as part of multi-disciplinary care for this patient group with rare and very rare progressive conditions, often genetic in origin and with no known cures and only limited treatments available.
- Greater support at transition from paediatric to adult services is needed given the evidence of services being removed or greatly reduced when people leave paediatric services even though needs may well increase given the progressive nature of many conditions.
- Adults in the region are particularly poorly served:
 - i) There is only one adult consultant neurologist to serve the entire region, which is having a serious impact on patients and their families.
 - ii) There is no specialist adult physiotherapy or occupational therapy on offer to patients.

- **There is an urgent need for greater clinical time. Currently there is a part-time paediatric neurologist providing services for children, with temporary support from a locum consultant on one day/week. Thus the gap between clinic appointments is often too long, which is a particular worry for children on high dose long-term steroids and those with impending potential respiratory failure. Attempts have been made to promote shared care but without the appropriate support of a managed clinical network the results have been mixed.**
- **There is no succession planning in the region - if existing staff left, the service would fall apart.**
- **There is the particular challenge of providing care for the Asian community in the region (see Appendix 5). The incidence of neuromuscular conditions is twice as high in Bradford, where the Pakisitani population counts for 15% of the total, as in surrounding areas, with a very different spectrum of disease – a trend also seen in other neurological conditions.**
- **Local speech and language therapy and dietetics services are extremely patchy.**

1. Background:

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

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

2. Demographics:

There are some 5,000 people in the Yorkshire and Humberside region affected by a form of muscular dystrophy or a related neuromuscular condition.

The Yorkshire and Humberside region has a population of approximately 5 million.

Specialised care in the region is commissioned by the Yorkshire and Humberside Specialist Commissioning Group. The region's fourteen PCTs are:

Barnsley PCT
Bradford and Airedale PCT
Calderdale PCT

Doncaster PCT
 East Riding of Yorkshire PCT
 Hull PCT
 Kirklees PCT
 Leeds PCT
 North Lincolnshire PCT
 North East Lincolnshire Care Trust Plus
 North Yorkshire and York PCT
 Rotherham PCT
 Sheffield PCT
 Wakefield District PCT

SCG Budget: In 2007/08 this was £130 million

3. Current level of essential, specialist provision in Yorkshire and Humberside:

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

Specialist multi-disciplinary care can improve quality of life and extend life expectancy. For example, without treatment, the mean age of death in Duchenne muscular dystrophy is 19 years.³ With specialist care and home ventilation, life expectancy is raised to almost 30 years.⁴

Co-ordinated and comprehensive multi-disciplinary specialist care should include a neuromuscular specialist consultant and, dependent on medical need, may also include specialist cardiac, respiratory and orthopaedic care.⁵ Genetic counselling and psychological services should also be offered, together with locality based dietetic, occupational therapy, physiotherapy and speech therapy provision which can both improve the quality of these patients' lives and increase their life spans. Boys with Duchenne muscular dystrophy who are still ambulant should be offered the opportunity to discuss treatment with steroids, such as deflazacort which studies have shown can stabilise muscle strength and delay the loss of ambulation, and may also delay the onset of breathing complications (see appendix 4). For a number of neuromuscular conditions, regular check ups are required irrespective of symptomatology, because deterioration can advance rapidly over the course of months.⁶

o Specialist muscle clinics:

- o Only three out of 14 (20%) PCTs in Yorkshire and Humberside commission adult muscle clinics.
- o Only four out of 14 (less than a third) PCTs in Yorkshire and Humberside commission paediatric muscle clinics.

The Muscular Dystrophy Campaign has noted that there is a significant difference in the standard of clinical care available to paediatric and adult patients. Transition is an area of concern for many families, and this situation requires urgent redress.

Paediatric muscle clinics in the Yorkshire and Humberside region are led by;

- Consultant Paediatric Neurologist, Dr Anne-Marie Childs
- Regional Care Advisor, Sue Manning
- Specialist Physiotherapist, Lindsey Pallant
- Specialist Occupational Therapist, Cathy Szeplaki
- Locum Consultant Paediatric Neurologist, Dr Karen Pysden

Details of paediatric muscle clinics:

Leeds; **Weekly** at the Leeds General Infirmary (LGI)

Hull; **Four times a year**

Halifax; **Three times a year**

Bradford; **Twelve times per year** (dependent on funding for the locum)

Dr Childs, is based at Leeds and manages outreach clinics in Hull, Bradford and Halifax. The service sees patients from West Yorkshire, North Lincolnshire, and North Yorkshire as far as Northallerton and Whitby. Further outreach clinics are necessary, and have been requested in Dewsbury and Airedale.

Dr Childs used to undertake an outreach clinic at York but this ended due to a lack of local support from therapists.

Dr Peter Baxter, consultant paediatric neurologist at Sheffield Children's Hospital covers the wider Sheffield area, Barnsley and Rotherham.

A Joint spinal clinic with teams from Leeds and Manchester is held three times per year at the LGI Joint Respiratory clinic with respiratory clinicians from paediatric and adult services.

A transitional clinic is held twice a year with adult neurologist, Dr Mark Busby.

The provision of cover in Leeds while Dr Childs is attending outreach clinics is a major problem. Dr Childs has currently secured funding for a locum to do this, but the uncertain financial situation at her Trust (Leeds Teaching Hospital NHS Trust has a £50 million deficit) means that this funding isn't guaranteed.

The occupational therapy and physiotherapy department has recently been relocated. The resulting loss of gym space means an inevitable reduction in the provision of therapy. This is an ongoing problem that will have to be assessed.

There is **only one adult clinic held every two weeks** in Leeds – and this clinic has no dedicated therapist or support staff.

There is a recognised need to provide further staff in order to improve the outreach service for all patients with an acute need to develop adult services. Further resources

are needed to ensure multi-disciplinary care with provision of specialist physiotherapists, speech and language therapists, and specialist muscle nurses.

o **Regional Care Advisors**

Regional Care Advisors (RCAs) play an essential role in supporting individuals with muscular dystrophy and related conditions. They successfully co-ordinate their health and social care needs, provide support and information to families and help to ensure a seamless transition from child to adult services. They also save money over the long term, due to freeing up consultants' time and helping to reduce unplanned hospital admissions.

The role of the RCA is crucial to these clinics. Subjects such as work, education, equipment provision and adaptations can be discussed in advance and referrals to local services made when appropriate.

The region has only one RCA, Sue Manning, who is based at the Leeds General Infirmary. The service is highly valued with patients and families describing the RCA service as excellent or good. However, due to the service being over-subscribed, two thirds of patients in the region do not receive the care and support of an RCA. **An extra four WTE posts (each with a caseload of 1,000 patients) are needed to serve the estimated 5,000 people with muscular dystrophy and related conditions in the region, many of whom are simply invisible to providers of health services.**

Commenting on the role of the RCA, one clinician said, "I couldn't run my service without the care advisor".

It is important to note that the current care advisor only sees children – care advisors serving the region's adult population are therefore urgently required.

It is essential that additional RCAs are appointed in order to serve the whole population. We are also calling for the existing RCA to become embedded in the NHS as a matter of urgency.

The provision of a named RCA/Co-ordinator is also stated as an aim by Health Minister Lord Darzi in his final report *High Quality Care for All* which set out how the Government intends to provide this more personalised level of care for people with long-term conditions.⁷ In addition, the need for a RCA was highlighted in the Parker *et al* study of Duchenne patients at the Lane Fox Unit (2005) which noted: "Most patients received full provision of disability allowances, but full access to social services provision was inadequate, and often depended on the input of the muscular dystrophy key worker"⁸.

The results of the Muscular Dystrophy Campaign's Patient Survey carried out in 2008 highlighted the need for an increase in RCAs, with over two thirds of patients reporting that they are not satisfied with the level of emotional support available to themselves and their families. Furthermore, only half of patients are satisfied with the amount and clarity of information available to them.

o **Diagnosis experience:**

Two fifths of patients in the area describe their experience of the diagnosis process as either poor or very poor, with many calling for greater information and support to be given to parents and families after diagnosis.⁹

There is a lack of knowledge among local GPs about these rare conditions, suggesting a need for greater education of the early symptoms of neuromuscular conditions.¹⁰

Clinicians have highlighted that they are unable to get funding for screening – unless there is a family member about to get pregnant – as the cost is £600. This has a knock-on effect on genetic planning and clinical trials.

o **Respiratory clinics:**

Breathing disorders are recognised as the leading cause of mortality in neuromuscular disease.¹¹ Respiratory muscle weakness is relatively common in most neuromuscular conditions and is almost inevitable in the late stages of Duchenne muscular dystrophy.¹² However treatment, including ventilation, has been shown to improve both quality and length of life.¹³

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 multi-disciplinary care reported by the Newcastle group in the most recent study by Eagle *et al* (2007).¹⁴

Regular comprehensive check ups are required with clinicians being instructed to go through a full checklist of signs and symptoms. A study published in 2002 highlighted patients can become too accustomed to their chronic illness and therefore rarely raise complaints about respiratory distress spontaneously.¹⁵

Evidence from a 2003 study highlighted that it is more cost-effective to manage respiratory issues through check ups and home ventilation than through unplanned critical hospital admissions.¹⁶

It is important to note that the paediatric ventilation service based in Leeds is insufficient to meet the current patient demand. Even though she is a neurologist Dr Childs is running the respiratory service – this is unsustainable. A dedicated respiratory service would reduce emergency admissions – a preventative investment.

o **Cardiac clinics:**

As a number of neuromuscular conditions also affect the heart, cardiac monitoring must be part of a multi-disciplinary approach to care. The heart is affected in different ways –

people affected by myotonic dystrophy and Emery-Dreifuss dystrophy are prone to abnormal heart rhythms, while cardiomyopathy is more likely for people affected by Duchenne or Becker muscular dystrophy.

Regular cardiac screenings are crucial even for conditions which appear to cause less severe weakening of the muscles, as “the severity of cardiomyopathy may be out of proportion to that of skeletal muscle involvement.”¹⁷ 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 10.¹⁸ Without screening, cardiomyopathy can progress almost entirely without symptoms until signs of heart failure emerge, when all cardiac reserve has been eroded.¹⁹

Cardiac screening should also be offered to women who are carriers of mutations in the dystrophin gene, who are at increased risk of cardiomyopathy, even if they experience no symptoms.

Clinicians in the region have identified “big holes” in the cardiology services across the region.

o **Physiotherapy:**

It is accepted that all patients with a neuromuscular condition will at some point during the course of their condition require access to ongoing and timely physiotherapy.²⁰ Physiotherapy is the physical treatment and management of a condition which enables people with neuromuscular conditions to reach their maximum physical potential by maintaining mobility, independence and improving quality of life. This should be provided by a specialist physiotherapist, who has skills in both neurological and musculoskeletal physiotherapy, experience in treating muscle conditions and the confidence to treat patients with rare disorders.²¹ Specialist physiotherapy can delay the progression of the condition, reduce pain and minimise emergency hospital admissions.

In April 2008, the Muscular Dystrophy Campaign carried out a Freedom of Information request to all NHS Trusts and Primary Care Trusts across England in order to detail the provision of physiotherapy services. Of the fourteen PCTs in Yorkshire and Humberside, the following picture emerged:

- o Only eight out of 14 PCTs responded. Of these:
 - o seven out of eight (88%) provide ongoing physiotherapy for patients with muscular dystrophy and related conditions where required
 - o four out of eight (50%) have trained physiotherapists available for children.
 - o four out of eight (50%) have trained physiotherapists available for adults.

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

The provision of physiotherapy in short blocks of sessions is problematic for patients and indicates a clinical focus on conditions in which quantifiable improvement can be measured, rather than the maintenance of chronic and progressive conditions. For

example, many hospital Trusts provide physiotherapy in six week blocks with patients having to be referred back for more treatment.

We are calling for ongoing, specialist physiotherapy to be provided to those patients in the region who require it.

o Orthopaedic care

Spinal deformity, such as scoliosis, is common in many neuromuscular conditions, with 90% of people affected by Duchenne muscular dystrophy for example, likely to develop a clinically significant scoliosis.²²

Surgery to correct spinal deformity can improve posture and comfort. It is imperative that the development of scoliosis is monitored by the specialist muscle clinic as success rates are likely to be highest and complication rates lowest if surgery is performed when the spine is still mobile at a Cobb angle of 20-40°. ²³ As it is a major operative procedure, a multi-disciplinary approach, involving the paediatrician/paediatric neurologists and orthopaedic surgeons is essential in the approach to surgery. ²⁴

As an example, the best practice guidelines for patients with spinal muscular atrophy state that evaluation should take place every three to six months, and more frequently in clinically unstable non-sitters. The evaluation should include, depending on clinical need: inspection of the spine, chest x-rays and radiographic evaluations of scoliosis, swallow studies, pulse oximetry and polysomnography. ²⁵

There is no spinal surgery service in Leeds. Families have to travel to Manchester, where two additional spinal surgeons have been recruited to meet the need from Yorkshire.

Specialist orthotists with an interest in neuromuscular services are required. The contracting out of orthotic services to private companies by each PCT means there is a real lack of specialist knowledge. Individual cases can require six letters from consultant teams, and children often have to be sent to London for spinal jackets/ knee-ankle-foot orthoses (KAFOs).

o Rehabilitation and equipment:

Specialist neuromuscular rehabilitation clinics aim to help maintain independence and to adapt to changes which affect social and domestic life and can include a number of services including physiotherapy, occupational therapy, speech and language therapy, wheelchair services and orthotics. Rehabilitation care can improve quality of life and delay progression of the condition. For example, poorly fitting KAFOs can severely compromise mobility and successful care. To avoid this orthotists with specific experience in neuromuscular disorders should be used to measure and supply orthotics. ²⁶

Wheelchairs

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

Currently, as PCTs do not collaborate to provide specialist wheelchair services, children and adults affected by these rare and progressive conditions are competing for equipment with patients who have acute episodes, for example a leg fracture, and are often being forced to wait for long periods for essential equipment. Muscular Dystrophy Campaign Freedom of Information requests have shown that with regards to children's wheelchair:

- 10 out of 14 PCTs responded (71%). Of these:
 - 30% do not provide tilt-in-space wheelchairs
 - Only 10% will fund wheelchairs with a riser function that allows children to elevate their seat to be at eye level with their peers, aiding mental wellbeing.
 - None of the region's 14 PCTs fully fund wheelchairs with a sit-to-stand feature that can maintain a child's muscle strength while only one PCT offers any financial contribution at all.
 - The average wait for a powered chair in the region is 16 weeks. However, the 39 week wait for a powered wheelchair endured by children in Doncaster PCT is more than twice the 19 week UK average.

- **Psychologists:**

Psychology support has been identified as an important aspect of multi-disciplinary care, and as a key part of rehabilitation services.²⁷ There is pressing need to develop clinical and educational psychology input and support for this patient group.

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

Studies have shown that the incidence of autistic spectrum disorders, attention deficit hyperactivity disorders and obsessive compulsive disorders is higher in males affected by Duchenne muscular dystrophy.²⁸ In addition behavioural changes have been shown to be an adverse side effect of treatment with corticosteroids, which are used to prolong ambulation and preserve muscle strength and respiratory function.²⁹ 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.

- **Transition**

Increasing numbers of young people with complex conditions are reaching transition and living longer because of improvements in therapies and medical care. For young people living with muscle disease, the period between mid and late teens is crucial and the transition from paediatric and adolescent care into adult-oriented healthcare services must be as smooth as possible.³⁰

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

The difficulties are shown by respondents to the Muscular Dystrophy Campaign Patient Survey, with three out of four families describing the transition process as 'poor' or 'very poor'. Only 14% rated the process as good or excellent, with the question not being applicable to the rest of respondents.

Appendix 1:

DemographicsYorkshire & Humber

<u>PCT</u>	<u>Resident population</u>	<u>Prevalence of neuromuscular conditions</u>	<u>Prevalence of muscular dystrophy</u>	<u>2009-10 PCT Revenue Allocations (£ thousands)</u>
Barnsley	224,600	225	113	
Bradford & Airedale	500,000	500 (likely to be higher due to ethnic community)	250(likely to be higher due to ethnic community)	
Calderdale	192,405	192	86	
Doncaster	289,000	280	140	
East Riding of Yorkshire	325,000	325	162	
Hull	249,000	249	125	
Kirklees	402,000	402 (likely to be higher due to ethnic community)	201 (likely to be higher due to ethnic community)	
Leeds	771,597	773	386	
North Lincolnshire	163,000	163	82	
North Yorkshire & York	765,000	765	383	
Rotherham	255,000	255	128	
Sheffield	530,300	530	265	
Wakefield	337,000	337(likely to be higher due to ethnic community)	167(likely to be higher due to ethnic community)	
Yorks & Humber Total	5,003,902	5,004	2,502	

Appendix 2:Background to report:

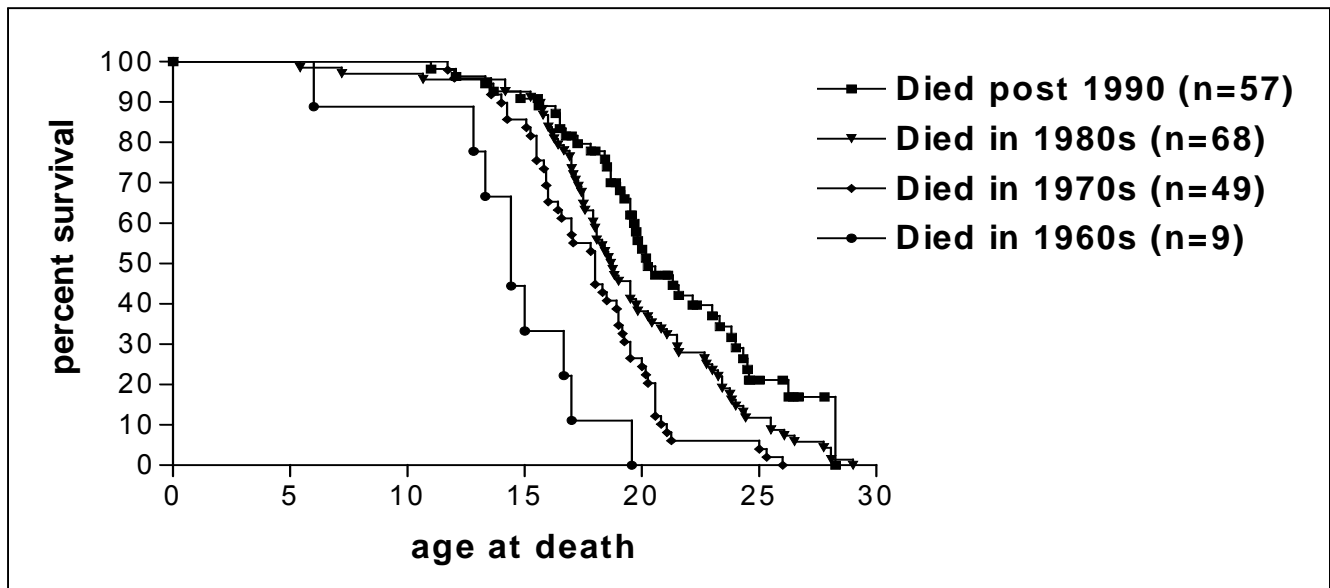
The report contains:

- Evidence from the leading neuromuscular clinicians working in the region.
- Information from the latest research papers on the impact of specialist services on those affected by muscular dystrophy and related neuromuscular conditions.
- Data from the responses to the largest nationwide survey of people affected by muscular dystrophy and related conditions, published in September 2008 by the Muscular Dystrophy Campaign. Eight hundred and fifty people completed the survey from across the UK.
- The responses to Freedom of Information requests to all Primary Care Trusts and Acute Trusts regarding specialist services.

Appendix 3:

Duchenne Muscular Dystrophy Survival data 1960-1990

(Eagle et al *Survival in Duchenne muscular dystrophy: improvements in life expectancy since 1967 and the impact of home nocturnal ventilation*³¹)



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

Results:

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

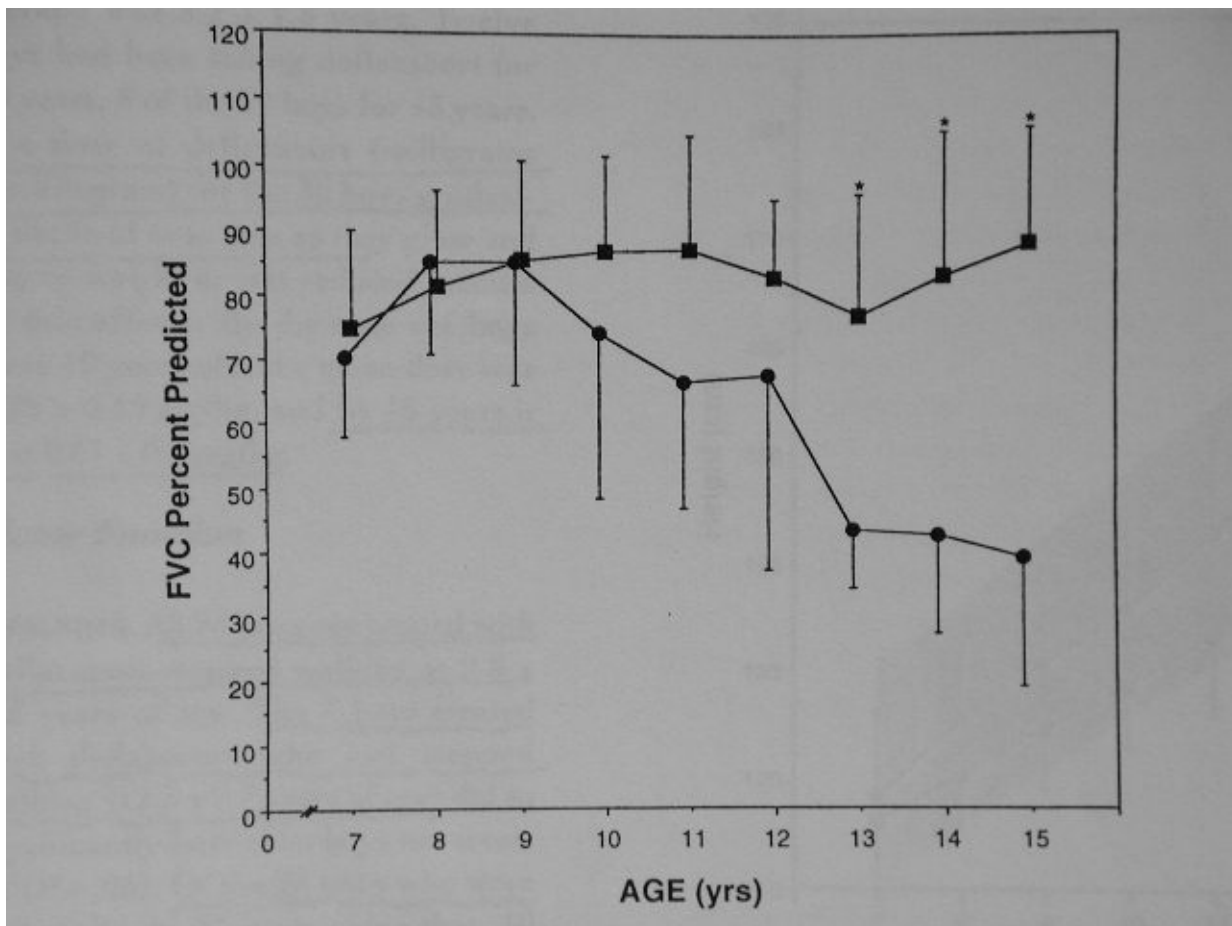
1990s: Mean life expectancy: 19.5 years

Improvement is due to multi-disciplinary care

Appendix 4:

Preserved lung function

(from Biggar WD, Harris VA, Eliasoph L, Alman B. Long-term benefits of deflazacort treatment for boys with Duchenne muscular dystrophy in their second decade. *Neuromuscular Disorders*)³²



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

Results for lung function:

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

Appendix 5:

Current research into the often higher rates of neuromuscular conditions amongst materially deprived ethnic minorities:

- Studies have shown that a higher than expected proportion of families of patients with Duchenne muscular dystrophy seem to be from a deprived background. Duchenne muscular dystrophy is an X-linked disorder affecting approximately one in 3,500 male live births. The incidence remains steady in most populations, maintained by a high rate of new mutations in the dystrophin gene.³³
- Evidence has recently emerged from clinical audits carried out in the Yorkshire and Humber region, which has shown that there is an alarmingly high frequency of cases of inherited muscle disease amongst ethnic minorities in deprived communities.³⁴
- A growing number of ongoing studies have found that marriage patterns, including parental consanguinity and endogamy, can also lead to increased cases of autosomal recessive inheritance, including spinal muscular atrophy and non Duchenne muscular dystrophies.³⁵
- A study by the Wellcome Trust of genetic risk counselling among British families of Pakistani origin estimated that the average national risk of autosomal recessive disorders is about two in every 100 births and that the risk is doubled in first cousin marriages.³⁶
- The national study of Progressive Intellectual and Neurological Deterioration (PIND) provides further epidemiological data on the distribution of neurodegenerative diseases in the UK child population. 1,400 children were reported in the first five and a half years of surveillance. In the five districts with the largest numbers of resident cases the majority not only came from a particular ethnic group but also had high reported rates of consanguinity.³⁷
- Where there has been a longstanding tradition of consanguineous marriage across generations, the resultant level of cumulative homozygosity would predictably be quite high, and is the reason for the high prevalence of recessive disease including neuromuscular conditions, within this population.

Therefore, as many families within Bradford's Pakistani population are materially deprived, young and growing, it is reasonable to anticipate that an already high instance of muscular dystrophy will increase over the next few years to reach a crisis point.

References:

- ¹ Care Advisors may also be known as key workers or neuromuscular care coordinators. They carry out the role envisaged by Lord Darzi in his 2008 High Quality Care for All report.
- ² Muscular Dystrophy Campaign Patient Survey – State of the Nation, September 2008
- ³ 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
- ⁴ Bushby, K. Bourke, J. Bullock, R. Eagle, M. Gibson, M. Quinby, J. 'The Multidisciplinary management of Duchenne muscular dystrophy' Current Paediatrics (2005) 15. 292-300. p. 294.
- ⁵ Bushby, K. Bourke, J. Bullock, R. Eagle, M. Gibson, M. Quinby, J. 'The Multidisciplinary management of Duchenne muscular dystrophy' Current Paediatrics (2005) 15. 292-300. p. 292-293.
- ⁶ A.E. Parker, S.A. Robb, J. Chambers, A.C. Davidson, K. Evans, J. O'Dowd, A.J. Williams and R.S. Howard. Analysis of an adult Duchenne muscular dystrophy population p. 730
- ⁷ Department of Health High Quality Care for All (June 2008):
http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_085825
- ⁸ A.E. Parker, S.A. Robb, J. Chambers, A.C. Davidson, K. Evans, J. O'Dowd, A.J. Williams and R.S. Howard. Analysis of an adult Duchenne muscular dystrophy population p. 734.
- ⁹ Muscular Dystrophy Campaign Patient Survey – State of the Nation, September 2008
- ¹⁰ Heap, R. Mander, M. Bond, J. Bushby, K. (1996) 'Management of Duchenne Muscular Dystrophy in the Community: Views of Physiotherapists, GPs and School Teachers' Physiotherapy Vol. 82. No. 4. p. 258-263. p. 262.
- ¹¹ Benditt J O. Management of pulmonary complications in neuromuscular disease. Physical Medicine and Rehabilitation Clinics of North America 1998; 9(1): 167-185
- ¹² A.E. Parker, S.A. Robb, J. Chambers, A.C. Davidson, K. Evans, J. O'Dowd, A.J. Williams and R.S. Howard. Analysis of an adult Duchenne muscular dystrophy population p. 734.
- ¹³ Bushby, K. Bourke, J. Bullock, R. Eagle, M. Gibson, M. Quinby, J. 'The Multidisciplinary management of Duchenne muscular dystrophy' Current Paediatrics (2005) 15. 292-300. p. 294.
- ¹⁴ 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.,
- ¹⁵ Birnkrant , D.J. 'The Assessment and Management of the Respiratory Complications of Pediatric Neuromuscular Diseases' Clinical Pediatrics (2002) Vol. 41. No. 301 p. 302-303.
- ¹⁶ Howard R.S and Davidson, C. Long Term Ventilation in Neurogenic Respiratory Failure J Neurol Neurosurg Psychiatry 2003;74(Suppl III):iii24–iii30 p. iii30.
- ¹⁷ 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.
- ¹⁸ 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.
- ¹⁹ 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.
- ²¹ Hill, M.E. Phillips, M.F. 'Service Provision for adults with long-term disability: A review of services for adults with chronic neuromuscular conditions in the United Kingdom' Neuromuscular Disorders 16 (2006)107-112. p.110-111.
- ²² Bushby, K. Bourke, J. Bullock, R. Eagle, M. Gibson, M. Quinby, J. 'The Multidisciplinary management of Duchenne muscular dystrophy' Current Paediatrics (2005) 15. 292-300. p. 295.
- ²³ Bushby, K. Bourke, J. Bullock, R. Eagle, M. Gibson, M. Quinby, J. 'The Multidisciplinary management of Duchenne muscular dystrophy' Current Paediatrics (2005) 15. 292-300. p.295.
- ²⁴ Muntoni, F. Bushby, K. Manzur, A. 'Workshop report: Muscular Dystrophy Campaign Funded Workshop on Management of Scoliosis in Duchenne Muscular Dystrophy 24 January 2005, London, UK' Neuromuscular Disorders 16 (2006) 210–219 p. 216.

-
- ²⁵ Ching H. Wang, et al. 'Consensus Statement for Standard of Care in Spinal Muscular Atrophy' Journal of Child Neurology Volume 22 Number 8 August 2007 1027-1049 p. 1033.
- ²⁶ Eagle, M. 'Report on the muscular Dystrophy Campaign workshop: Exercise in Neuromuscular diseases, Newcastle January 2002' Neuromuscular Disorders 12, (2002) 975-983 p. 978.
- ²⁷ Nätterlund, B. and Ahlström, G. 'Activities Of Daily Living And Quality Of Life In Persons With Muscular Dystrophy' Rehab Med 2001; 33: 206–211.
- ²⁸ 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.
- ²⁹ Manzur, AY. Kuntzer, T. Pike, M. Swan, AV. Glucocorticoid corticosteroids for Duchenne muscular dystrophy (Cochrane Review) The Cochrane Library 2009, Issue 1.
- ³⁰ 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.
- ³¹ 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.
- ³² Biggar WD, Harris VA, Eliasoph L, Alman B. Long-term benefits of deflazacort treatment for boys with Duchenne muscular dystrophy in their second decade. Neuromuscul Disord 2006;16:249-5
-

- ³³ Bushby, K., Raybould, S., O'Donnell, S., Steele, J.G., 'Social deprivation in Duchenne muscular dystrophy', BMJ, 323:1035-1036, Nov 2001
- ³⁴ Childs, AM, Rotti, F., Young, S., Corry, P., 'An observational study suggesting significant variation in the presence and spectrum of neuromuscular disease among different ethnic groups', 2009
- ³⁵ Childs, AM et al. 'An observational study suggesting significant variation in the presence and spectrum of neuromuscular disease among different ethnic groups', 2009
- ³⁶ Shaw A, 'Genetic counseling for Pakistani and Bangladeshi origin Muslim families in Britain', The Encyclopedia of the Human Genome, Nature Publishing Group (2003).
- ³⁷ Devereux, G., Stellitano, L., Verity, C.M., Nicoll, A., Will. R.G. and Rogers. P., Variations in neurodegenerative disease across the UK: findings from the national study of Progressive Intellectual and Neurological Deterioration (PIND), Archives of Disease in Childhood, 2004; 89:8-12; See also, <http://bpsu.inopsu.com/studies/PIND/index.html>, accessed July 2009.
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