



Building on the Foundations in Northern Ireland:

Improving Specialist Care, Support and Independence

February 2009

Muscular
Dystrophy
Campaign



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Foreword

In this report, the Muscular Dystrophy Campaign, together with leading clinicians and health professionals from Northern Ireland, sets out a compelling case for an urgent review of specialised health services for families affected by muscular dystrophy and related neuromuscular conditions.

We are dismayed by the lack of succession planning of professional posts, which are integral to the multi-disciplinary management of these conditions. We are particularly shocked that preparations were not made for the retirement of the paediatric neurologist specialising in muscular dystrophy.

As a result there has been no paediatric consultant specialising in muscular dystrophy in post since July 2008 so some children and their families are forced to seek specialist advice from centres in England. It is clear that the situation undermines the care and could lead to the shortening of lives of these vulnerable children.

Multi-disciplinary specialist care has been developed as the best model for delivering effective clinical care for such complex multi-system conditions. Our findings show that greater co-ordination and specialist care provision improve quality and, where possible, length of life of patients, and their carers, as well as improving cost-effectiveness.

We are calling on the Government of Northern Ireland, the Regional Medical Services Consortium and the four Local Health Boards to work with us to address four central issues:

- Urgently appoint a paediatrician specialising in muscular dystrophy in Northern Ireland.
- Improve current commissioning arrangements for children and adults affected by muscular dystrophy and related conditions to ensure appropriate succession planning for these specialised services.
- Establish a working group, in partnership with health professionals and patient organisations, to review current service provision for patients with muscular dystrophy and related conditions living in Northern Ireland.
- Ensure that all patients have access to a key worker at all stages of their life.

We believe that Northern Ireland can lead the way in providing comprehensive specialist services for people with muscular dystrophy and related conditions.

Executive Summary

What are these conditions?

The report reflects evidence obtained from a patient survey, Freedom of Information requests to Northern Ireland Health Boards and information from the latest research papers on the impact of specialist services on those affected by muscular dystrophy and related neuromuscular conditions.

There are more than 60 different types of muscular dystrophy and related neuromuscular conditions affecting an estimated 1,000 children and adults in Northern Ireland. These conditions can be genetic or acquired.

A number of these conditions, such as Duchenne muscular dystrophy, are aggressive and cause progressive muscle wasting and weakness, orthopaedic deformity, cardiac and respiratory compromise and result in premature death. Other conditions cause life-long disability. Some conditions present in childhood or young adult life while others can be late onset conditions in adulthood. There are currently no known cures for these conditions.

Historical context

Since the 1980s a multi-disciplinary team based at Belfast City Hospital has successfully led the management and care of children and adults affected by muscular dystrophy and related conditions.

However, in recent years several staff have moved to new posts and some have retired with inadequate succession planning in place to replace these posts (paediatric neurologist, occupational therapist, dietician and paediatric dentist.) There is also no dedicated funding for the specialist physiotherapy clinic and this would be vulnerable to changes in Trust priorities.

Currently the muscle clinic for both children and adults based at Belfast City Hospital serves approximately 500-600 adults and 280 children annually. (A number of adults are not reviewed annually.)

Key findings

- There is currently no paediatric consultant, specialising in muscular dystrophy, to provide specialist assessment and care to children.
- There are no speech and language therapists specialising in muscular dystrophy in Northern Ireland.
- There is no dedicated funding available for occupational therapy input as part of the multi-disciplinary clinic, despite identified need.
- There is limited access to rare genetic testing of blood and muscle to confirm suspected hereditary muscle diagnoses.
- Adult rehabilitation services are particularly limited and many adults do not have access to ongoing physiotherapy, or any physiotherapy at all.
- Services are vulnerable given their dependence on charitable funding.
- There is no designated funding for home ventilation services, and the quality of the service depends on where patients live

A survey of patients living with muscular dystrophy revealed vast disparities in service provision for patients living in Northern Ireland:

- two out of five patients feel that they do not receive enough physiotherapy;
- two out of five said their experience of the diagnosis process was either poor or very poor;
- Two of five patients have no access to a specialist neuromuscular consultant.

Action needed

We are calling on the Government of Northern Ireland, the Regional Medical Services Consortium and the four Local Health Boards to:

- urgently appoint a paediatrician specialising in muscular dystrophy in Northern Ireland;
- improve current commissioning arrangements for affected children and adults to ensure appropriate succession planning for these specialised services;
- establish a working group, in partnership with health professionals and patient organisations, to review current service provision for patients with muscular dystrophy and related neuromuscular conditions living in Northern Ireland;
- commission the role of a care advisor at a regional level to ensure that all patients have access to this specialist support at all stages of their life;
- ensure that all patients have access to a specialist multi-disciplinary team who provide timely specialist diagnosis, assessment, treatment and ongoing care;
- conduct an urgent and comprehensive assessment for the development of a home-ventilation service, resourced across Northern Ireland;
- ensure that specialist cardiac support for patients with inherited cardiac disorders is available across Northern Ireland.

Evidence

Children in Northern Ireland are being denied access to specialist assessment and interventions

The effective role of multi-disciplinary specialist care has been developed as the best model for delivering effective clinical care for such complex multi-system conditions.

The provision of expert physiotherapy, orthotics, early cardiac monitoring and intervention and corticosteroids has been shown to improve muscle function and maintain independent mobility¹. The judicious use of spinal surgery and expert respiratory services (including non invasive positive pressure ventilation²) helps to improve quality of life, delay the onset of respiratory failure and prolong the life of these patients³.

At present there is no paediatric consultant specialising in muscular dystrophy. As a result the care of affected children in Northern Ireland is being seriously compromised.

The role of the paediatrician within the neuromuscular multi-disciplinary team includes medical interventions, as well as co-ordinating other specialist medical aspects of care.

The mean age at death in untreated Duchenne muscular dystrophy is 19 years, with roughly 90% of these deaths attributable to respiratory and 10% to cardiac causes. It is a clear testimony to the improvements in clinical care that in many centres the mean age at death in Duchenne muscular dystrophy is now in the late twenties or beyond⁴.

It is clear that the lack of specialist paediatric involvement in the care of children with muscular dystrophy could lead to the shortening of lives of these vulnerable children.

Child A has Duchenne muscular dystrophy and is still walking. This would be an ideal time for him to receive an assessment for the commencement of steroids, which would maintain his mobility (in line with TREAT-NMD standards of care⁵). However, he is being denied vital treatment, as there is no paediatric consultant to oversee and monitor steroid use. This could result in a serious deterioration of his condition.

Child B has Duchenne muscular dystrophy he commenced steroid use as prescribed by the former paediatrician who specialised in muscular dystrophy. However, there is no specialist to monitor his weight gain and care and provide an assessment of the continued use of steroids for this patient.

The loss of the paediatric element of the neuromuscular clinic has also resulted in the loss of specialist orthopaedics as part of the multi-disciplinary set up. Patients are now reviewed separately by orthopaedics.

Plans are being developed to set up a satellite clinic in Belfast from a London centre. It is not clear how often the clinic would be run. The proposal is not a long-term solution to the lack of succession planning in Northern Ireland. A full-time, permanent paediatrician specialising in muscular dystrophy and related conditions is urgently required.

Access to specialist speech and language therapists and dieticians remains poor

A multi-disciplinary team approach, including a speech therapist, dietician, neurologist and pneumologist is needed to evaluate and manage 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.⁶

Historically, a dietician was routinely present at the multi-disciplinary neuromuscular clinic in Belfast. However, this input is no longer present.

There are no speech and language therapists specifically trained in the management of patients with muscular dystrophy and other related conditions despite well-documented evidence of the importance of this input within a multi-disciplinary setting.

There is an urgent need to commission occupational therapy input as part of the multi-disciplinary team in Belfast

Current occupational therapy provision as part of the multi-disciplinary team in Belfast has been identified as inadequate. Attendance at the neuromuscular clinic is only available via a bleep service for referrals.

At present the occupational therapy (OT) team has close liaison with their community OT colleagues and are pro-active in arranging follow-up assessments within their OT department. However, funding for OT support at the neuromuscular clinic in Belfast is required.

The provision of OT input and intervention in the clinic would enhance and ultimately provide a higher quality of care to this client group.

Access to specialist diagnostic services is difficult due to inconsistent funding arrangements

Our patient survey exposed cases of misdiagnosis and patients funding their own diagnostic services.

A woman had been diagnosed with limb girdle muscular dystrophy. Subsequently she and her husband were informed that there was not a risk to having children. She went ahead and had a daughter who also has a muscle disease. Because her initial diagnosis was wrong, the family were not given time to prepare for their child being disabled.

Our research has also revealed inconsistencies in funding arrangements for diagnostic testing. The tests listed on the UK Genetic Testing Network can be sent to any laboratory in the UK. However not all tests are approved by the network, for example some of the rarer genes and some myasthenias, including Dok7 and various types of HMSN, which are very costly. Given that diagnostic services in Northern Ireland operate within a fixed budget they are subsequently limited in the tests they can provide.

Psychology support for this patient group is not well developed, despite clear evidence of need

Psychology support has been identified as an important aspect of multi-disciplinary care⁷. There is pressing need to develop clinical and educational psychology input and support for this patient group.

According to our nationwide survey: ***State of the Nation, two fifths of patients are dissatisfied with the level of emotional support available to their families and to themselves.***

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.

Children with Duchenne muscular dystrophy are at risk of developing pervasive developmental disorders (autism) and since the introduction of steroids to treat Duchenne behavioural problems are emerging as a significant effect.⁸ 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. Other times when input from a psychologist will be helpful include the loss of ambulation, preparation for spinal surgery, the introduction of BiPAP (non-invasive ventilation), which also coincides with transition into adulthood and may include psychosexual issues⁹.

Children and adults with 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 strategies¹⁰.

Adult rehabilitation services are poorly developed in Northern Ireland

There are no adult rehabilitation physicians in Northern Ireland with a special interest in muscular dystrophy and related neuromuscular conditions. The provision of adult rehabilitation services remains an area of significant concern and requires the urgent attention of commissioners.

Adult rehabilitation services in Northern Ireland have prioritised periods of intensive therapy to maximise recovery in areas such as adult brain injury, stroke and spinal injury. Thus there is a very limited rehabilitation service available to adults with muscular dystrophy and related conditions.

Many patients, particularly adults, do not receive continuous specialist physiotherapy or any physiotherapy at all

Current provision:

- Experienced provision is available from a multi-disciplinary team in schools for children; however, our survey showed that families do not feel they are getting enough physiotherapy.
- There is only one specialist physiotherapist employed as part of the multi-disciplinary clinic in Belfast; the post is currently vacant but it is hoped that the position will be filled shortly;
- Many adults receive no physiotherapy at all or, if they do, it is provided in short bursts of therapy with no ongoing input or review.

For adults, referral to physiotherapy in general is mixed and often very limited. Many patients only have access to 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. Indeed, some patients are referred by their GP, others from their consultant neurologist and some have to make a self-referral.¹¹

The following examples illustrate the difficulties adults experience in receiving ongoing and timely physiotherapy:

A man who has mitochondrial myopathy commented on the lack of physiotherapy as an adult: “When I was a child I received ongoing physio but when I became an adult it stopped. I now only receive short spells of physiotherapy and this is not always by a physio who understands my condition.”

A man who has Becker muscular dystrophy commented on the lack of physiotherapy services: “Physiotherapy makes me feel better, like I might be fighting my condition. However, I only see the specialist physiotherapist once every 18 months. When I try to get referred locally I am only entitled to six to eight sessions. This is not sufficient for someone like me with a progressive muscle-wasting condition.”

A woman with muscular dystrophy commented: “Lately I have noticed changes to my joints. I told my GP but my concerns were brushed aside. I would benefit greatly from seeing a physiotherapist but this service has not been offered.”

Services are vulnerable given their dependence on charitable funding

The Muscular Dystrophy Campaign, with funding provided through the Big Lottery Fund, provides direct financial support for the only Care Advisor in Northern Ireland. The Care Advisor has a case-load of 450 child and adult patients. **The funding for this post from the Big Lottery Fund will end in March 2009 and there is no NHS planning in place to ensure future stability and continuity of this post. While the Muscular Dystrophy Campaign itself will fund the post from April 2009, it is clear that NHS funding is required.**

The Care Advisor (or key-worker) plays an essential role in providing a holistic package of care to children and adults with muscular dystrophy and related 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.

Respiratory services

Respiratory muscle weakness is a common cause of morbidity and mortality in the muscular dystrophies.¹² The increased availability and convenience of modern non-invasive ventilation (NIV) has led to a dramatic change in the management of chronic neurogenic ventilatory failure.¹³

Despite this, there is currently no dedicated funding for domiciliary ventilation services for patients with muscular dystrophy in Northern Ireland. Domiciliary support is variable depending on where you live.

An increasing number of young people with muscular dystrophy and related conditions are surviving into adulthood. Respiratory care is becoming critical to the enhanced life expectancy and quality of life for these young people.

The benefits, both in life expectancy and quality of life, of non-invasive ventilation and other technological treatments to support respiratory care has been shown¹⁴, resulting in an incremental growth in demand.

Cardiac services

There is currently only one cardiologist in Northern Ireland providing a specialist cardiac service to patients with cardiomyopathies, inherited cardiac diseases and young sudden death. The service is currently overwhelmed with new referrals.

We are pleased that the Regional Medical Services Consortium has agreed to fund the inherited cardiac diseases service in Northern Ireland. In conjunction with the Genetic Services at Belfast City Hospital, the service will have funding to support specialist nurses, a full-time cardiologist with an interest in inherited cardiac diseases as well as funding for genetic tests. This funding will be rolled out over the next 3 years. Closer collaboration is also planned with the neuromuscular clinic at Belfast City Hospital.

Cardiac involvement is an invariable feature of a number of neuromuscular conditions, including Duchenne muscular dystrophy, Becker muscular dystrophy and myotonic dystrophy type 1. The importance of cardiac monitoring and treatment is clear given that approximately 10-20% of boys with DMD will die from Cardiac involvement.¹⁵ Expert treatment guidelines about the management of cardiac involvement¹⁶ have been developed and published.

Conclusion

We have illustrated the gaps in specialist service provision and the actions necessary to improve the outcomes in care and support for adults and children with muscular dystrophy and related neuromuscular conditions.

Further work is being carried out on the provision of wheelchairs and equipment for patients with these conditions. Our **State of the Nation** survey highlighted some serious deficiencies in the area. If you would like to send us your experiences and comments about wheelchair services and equipment please call 020 7803 4800 or email your details to campaign@muscular-dystrophy.org

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