



Building on the Foundations: The Need for a Specialist Neuromuscular Service across Wales

A report presented to the
Welsh Assembly Government

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Foreword

In this report produced by the Muscular Dystrophy Campaign and the Genetic Interest Group we set out a clear case for the commissioning of specialist neuromuscular services for people with neuromuscular conditions living in Wales.

There is no comprehensive neuromuscular service for adults or children in Wales and for some parts of the country the picture is very bleak indeed, since services for patients with neuromuscular conditions are extremely limited or non-existent. The result of this situation is that patients in Wales are not receiving specialist care and outcomes are adversely affected

The case made in this document is a very powerful one, and we urge Assembly Members, policy makers and professionals to take note of its findings and to do all that they can to improve patient care for people – both children and adults – living with neuromuscular conditions.

Too often the Welsh Assembly Government has ignored previous reports calling for improvements in neuromuscular services. A commitment to develop a Wales-wide neuromuscular service supported by specialist commissioning is urgently needed.



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Introduction

“We aspire to a world-class health service that is available to everyone irrespective of whom they are or where they live in Wales, and at the time when they need it. Our health services must inspire confidence in the people of Wales that they will receive the best care available.”

Welsh Assembly Government, June 2007¹

The current care for people with neuromuscular conditions in Wales fails to meet the ambition set out by the Welsh Assembly Government last year.

It is simply unacceptable that many patients and their families are not receiving adequate levels of care and cannot enjoy the best possible quality of life.

These problems and shortcomings in NHS Wales’s services in many parts of the country are avoidable – we know the improvements that need to be made and the steps required are described in this paper.

It has been shown that a lack of specialist neuromuscular diagnosis, treatment and care has a direct effect on the quality of life of patients and, for those with life limiting conditions, like Duchenne muscular dystrophy, their life expectancy.

This report describes current service provision in Wales and highlights that the existing system is failing patients. Comprehensive neuromuscular services should be designated within the Health Commission Wales (Specialist Services) Commissioning Plan. A review of services in Wales should be undertaken and the Welsh Assembly Government and NHS Wales should work with the emerging British Myology Society (the professional body for neuromuscular specialists) in approving a Standard of Diagnosis and Care for neuromuscular conditions.

The muscular dystrophies and related neuromuscular conditions

There are more than 60 different types of muscular dystrophy and related neuromuscular conditions. Dr Jader at the National Public Health Service for Wales reported that incidence of the muscular dystrophies in Wales is 50 in 100,000².

However, this figure does not include other related neuromuscular conditions and it is accepted that over 1000 children and adults for every 1 million of the population are affected by muscle wasting neuromuscular diseases in Wales. Therefore, it is estimated that around 3000 people are affected by a neuromuscular condition in Wales. Many neuromuscular conditions are low-incidence conditions and some are ultra orphan.

Neuromuscular disorders can be genetic or acquired. Inherited neuromuscular disorders can 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 can include myasthenia gravis, autoimmune neuropathies and inflammatory myopathies.

A number of these disorders, 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. Neuromuscular disorders such as these are often present in childhood or young adult life.

The majority of muscular dystrophies have related heart problems. Muscle weakness is often associated with poor ventilatory ability and respiratory failure. These are often overlooked by professionals unfamiliar with these conditions. Pain management is a common feature as these conditions can cause nerve deterioration or neuropathy.

The Problem

- Variable – Specialist centre records and patients' experience shows that neuromuscular services are variable across the country and where patients live often adversely affects the care they receive (evidence from a Freedom of Information Request to Local Health Boards and Health Commission Wales in January 2008);
- Vulnerable – Where services are in place in South Wales they are vulnerable given their dependence on a handful of leading clinicians who may in time move on or retire. Historically funding is vulnerable and has been dependent on charitable subsidy;
- Inaccessible – Some Welsh commissioners and non-specialist clinicians are reluctant to refer to Neuromuscular Centres outside but close to Wales.
- Advances in recent years have provided clear evidence that specialist care provided by a multi-disciplinary team prolongs life, reduces morbidity and helps prevent transmission of the genetic disease in patients with neuromuscular diseases. Many patients in Wales do not receive such multi-disciplinary specialist care and outcomes are adversely affected.

Key Asks

We are calling on the Welsh Assembly Government and NHS Wales to:

- Recognise that current service provision for neuromuscular conditions is failing many patients and acknowledge that all patients with neuromuscular conditions require access to specialist diagnosis, treatment and on-going care;
- Recognise neuromuscular services as specialised services and designate these within the Health Commission Wales (Specialised Services) Commissioning Plan so that these services are strategically planned across Wales;
- Undertake an urgent review of existing neuromuscular services. This would assess the current levels of care across Wales to address any weaknesses in local provision;
- Press Health Commission Wales and NHS Wales to ensure that specialised neuromuscular services are accessible for patients with neuromuscular conditions to overcome current fragmentation, weakness and vulnerability;
- Address the problems arising from the mix of rural, urban and valleys areas that exist across Wales where services are particularly inaccessible and inadequate;
- Work with the Department of Health and the NHS in England to establish how services should be delivered in England and Wales through specialised neuromuscular centres giving support and clinical leadership to a network of local clinics;
- Consider the establishment of a National Neuromuscular Network for Wales to provide clinical leadership, coordination and development of services.
- Ensure that boys diagnosed with Duchenne muscular dystrophy (DMD) by the newborn screening programme receive the necessary follow-on treatment and care.

Issues to be addressed

- Specialised Neuromuscular Services are not recognised on the Health Commission Wales (Specialised Services) Commissioning Plan;
- Most patients in Wales with neuromuscular diseases do not receive specialist multi-disciplinary care;
- Inequality of access to specialist care across Wales leads to a 'postcode lottery' for patients;
- A 'hub and spoke' model to deliver specialist care across all of Wales, utilising specialist centres across the border in England as well as expertise in Wales;
- Welsh commissioners do not consistently fund diagnostic tests for Welsh patients at recognised specialist centres in England;
- There is currently no NHS Wales approved Standard of Diagnosis and Care for neuromuscular conditions;
- Local Health Board commissioning arrangements do not result in sufficient service provision for patients with neuromuscular diseases;
- Local Health Board commissioning arrangements do not result in adequately resourced clinics to care for patients effectively;
- Local Health Board commissioning arrangements for neuromuscular conditions do not result in effective collaboration of services in certain regions;
- Historically commissioning arrangements have meant that some core posts were only provided by fund raising efforts of charities (such as the Muscular Dystrophy Campaign) and patients themselves.

1. Specialised neuromuscular services are not designated on the Health Commission Wales (Specialised Services) Commissioning Plan

Although the Health Commission Wales (Specialist Services) Commissioning Plan prioritises some specialised neuromuscular services, for instance, enzyme replacement therapy, and identifies related services like neuro-rehabilitation and long-term invasive ventilation, the Plan does not recognise or prioritise the specialist nature of care for patients with all types of neuromuscular conditions.

For example, despite the fact that the management of myotonic dystrophy in Wales is internationally recognised, clinicians are unable to improve or expand this service as Welsh health commissioners fail to recognise the specialist nature of this service.

It is clinically recognised that neuromuscular diseases require a multi-disciplinary specialist approach to care. It is clear that services currently fall short of what is needed across Wales with a consequent impact on patient outcomes.

The neuromuscular conditions are low prevalence conditions and specialist services are not consistently commissioned by Local Health Boards or NHS Trusts. Therefore, current commissioning arrangements fail to support services that meet the needs of many children and adults with neuromuscular conditions.

Further, the designation of comprehensive neuromuscular services within the Specialised Services Commissioning Plan would require Local Health Boards and NHS Trusts to establish a broad base-line position making initial comparisons on activity and spend, and provide an important basis for service reviews and strategic planning in addition to the benefits for patients.

2. Most patients in Wales with neuromuscular conditions do not receive specialist multi-disciplinary care

Specialist multi-disciplinary care has been developed by leading clinicians as the best model for delivering effective care for such complex, multi-system diseases. The provision of expert physiotherapy, orthotics, early cardiac monitoring and intervention and corticosteroids has been shown to improve muscle function and maintain independent mobility³. The judicious use of spinal surgery and expert respiratory services (including non-invasive positive pressure ventilation) helps to improve quality of life, delay the onset of respiratory failure and prolong the life of these patients⁴.

The medical specialists that can deliver different facets of diagnosis and care vary from neurologists (adult and paediatric), to clinical geneticists, to paediatricians, rehabilitation physicians, cardiologists, orthopaedic surgeons, pathologists and palliative care inter alia. In addition, specialist physiotherapists, occupational therapists, speech & language therapists and various specialist nurses relating to the above groups have important roles in supporting and monitoring the patient and their family. The rarity of many of the conditions

and specialised nature of diagnosis, care and intervention inevitably require centres of expertise with a relatively large population base.

The development of services for patients with neuromuscular diseases in Wales has been inconsistent and heavily dependent on the research interests of dedicated individuals who have developed a clinical interest in a neuromuscular disease. There is no current strategy in place for succession planning, leaving the services fragile and vulnerable in view of their heavy dependence on the lead clinicians.

The Muscular Dystrophy Campaign undertook a survey of the 22 Local Health Boards (LHB) in January 2008 in order to build a picture of access to healthcare services for people with neuromuscular conditions. The authors asked the Health Boards to provide information regarding services commissioned locally for people with neuromuscular conditions.

Out of the 20 LHBs that have so far responded, the following picture has emerged:

- **75%** of LHBs who responded do not support a muscle clinic that offers a service to adults with neuromuscular conditions.
- **70%** of LHBs who responded do not support a muscle clinic that offers a service to children with neuromuscular conditions.
- **70%** of LHBs who responded do not support any adult or child muscle clinics within their area. [See Appendix 1]

It can be said from the evidence submitted by the LHBs that a large concentration of the services which are available take place in South Wales. Furthermore, the degree to which they provide a comprehensive service is highly variable and further research should be undertaken to identify the clinical workforce at these clinics.

The newly formed NHS Wales Regional Offices for Mid & West Wales, North Wales and South & East Wales were recently established to assist and support the 22 LHBs for implementing policy across Wales and commissioning services uniformly. However, we have concern regarding the effectiveness that these three groups will have in their work when concerned with improving service delivery for rarer conditions such as neuromuscular conditions.

3. Inequality of access to specialist care across different regions in Wales

The recognised specialist neuromuscular centres in the UK providing comprehensive services are found in Oxford, London and Newcastle⁵. First class specialised services are also found in Liverpool and Oswestry and should be fully accessible through specialised commissioning in Wales. Historically, Wales had a strong service for some patients and families with genetic neuromuscular conditions which has been used across the border in England as a model service. The service attracted patients from outside Wales and became internationally recognised for its research. However, the service was extremely vulnerable as it was largely built on the personal commitment of a handful of clinicians with an interest in neuromuscular conditions and certain specialist posts were dependent on charitable subsidy and not strategically planned by the health authorities themselves.

It is very disappointing that due to the vulnerable nature of neuromuscular services in Wales, existing services are fewer and more fragmented than ever before.

In North Wales there is no dedicated service for patients – adults or children – with neuromuscular conditions. In addition, one of the few lead clinicians in South Wales with a special interest in neuromuscular conditions is retiring in autumn 2008. It is essential that this post is incorporated into service planning and a replacement found. In West Wales, a recognised clinic is run in the clinician's own study time. [See Appendix 2 for list of recognised clinics in Wales]

While some patients who live relatively close to one of the few neuromuscular clinics in Wales or are referred to a specialist centre in England receive excellent care from their clinicians, most patients cannot access these services and receive inferior services or, indeed, may receive no services at all.

Furthermore, as the following case study illustrates, some patients in Wales have to travel great distances and at a huge expense to access specialist multi-disciplinary care.

Patient Case No. 1

Rhys is 7 years old, lives in South Wales and has a life limiting disease – Duchenne muscular dystrophy. He had previously lived within the area of a recognised neuromuscular centre of excellence in England – the Hammersmith Hospital in London. Rhys continues to attend this recognised neuromuscular centre for his bi-annual management clinic as there is no equivalent specialist service in Wales.

Even then however, this is a round trip of 300 miles, requiring an overnight stay and time off work for his parents. Despite this, Rhys's parents consider this worthwhile as at the Centre of Excellence he is assessed and reviewed by a multi-disciplinary team of specialists who have expertise and are familiar with neuromuscular conditions like Duchenne muscular dystrophy.

At the Hammersmith, his care is managed by a specialist neuromuscular consultant, a specialist physiotherapist, specialist nurse and a family care officer who are all trained and experienced in dealing with muscle diseases and the related matters. His health and fitness are reviewed – such as his respiratory health, as are his drugs, physiotherapy regime and also any orthotics and equipment used. In addition, the specialists advise of any other aspects of his management, whether he can be referred to other specialists such as ophthalmologists (in case of cataract development as a side-effect from taking steroids), a specialist clinical psychologist and also a general discussion about social care and education.

4. Specialist nature of neuromuscular services and the benefits of a 'hub and spoke' model

It is the nature of the service that is specialised and not its physical location. All care for neuromuscular patients whether within the specialist centre or the shared facility should be

regarded as specialised. Indeed, care and support provided at home will also come within the remit of this specification.

Failure to invest in specialist neuromuscular services will in time lead to growing inequalities for patients in terms of both service provision and long term clinical outcome. Preliminary evidence from a study in progress at the Newcastle Centre in England (highlighted below) has established the benefits for patients of the specialist multi-disciplinary care model.

Benefits of specialised multi-disciplinary care – Preliminary evidence from a Qualitative Research Study:

The care provided to the families attending the paediatric muscle clinic at Newcastle has recently been assessed by a PhD student who studied the availability of and satisfaction with Home and Community Based Services for Children with Neuromuscular Disorders.

The results of this study offer the first concrete evidence of a positive benefit derived from the care model of a Muscle Centre with multi-disciplinary input in improving the experience of patients with chronic disability. These parents were not experiencing the same level of difficulty described in most previous research about the support needs of disabled children and families. Part of the reason for this was felt to be the support provided by the specialist Muscle Team.

This qualitative study, supervised by Professor John Carpenter, initially of the University of Durham but now in Bristol, aimed to explore children and young people with neuromuscular impairments and their parents' experiences with education, health and social care services. The study used the accounts of children and young people themselves and those of their parents or carers, exploring their perceptions of education, health and social care services.

Specialist staff at a Neuromuscular Centre

As these are complex multi-system diseases, a specialist centre requires a multi-disciplinary team approach to care. This team comprises:

- Adult clinician with specific training in muscle diseases including myasthenia
- Adult neurologist with specialist training in neuropathies
- Adult and paediatric neurologist with specialist interest in congenital myasthenia
- Two Paediatric Consultants with specialist training in neuromuscular disease
- Adult and paediatric respiratory physicians who run non-invasive ventilation services and appropriate support staff
- Adult and paediatric cardiologists with specialist interest in NMD
- Clinical neurophysiologists with a special interest in NMD including single fibre EMG
- Clinical geneticist
- Genetic counsellor
- Specialist Neuromuscular physiotherapist
- Occupational therapist with a special interest in NMD

- Neuromuscular Regional Care Advisor/ patient advocate
- Neuromuscular nurse specialist
- Clinical psychologist with a special interest in NMD
- Muscle and nerve pathologist with a special interest in NMD
- Orthopaedic and Spinal surgeons with a special interest in NMD
- Orthotist
- Dietician with a special interest in NMD
- Neuromuscular speech and language therapist

In addition, the recognised specialist centres in the UK largely have a research capacity and a neuromuscular research fellow, a laboratory scientist and an administrator.

The importance of specialist professionals should not be understated. Specialist neuromuscular physiotherapy for example, has been shown to prevent and minimise contractures, improve mobility and the quality of life of patients for adults and children. Disappointingly there are no physiotherapists in Wales who have received NHS funded training in neuromuscular conditions.

The following case studies illustrate the importance of specialist neuromuscular physiotherapy and the difficulties patients face in accessing treatment:

Case No. 2

Anne is a community physiotherapist. She has a few children in her care with a muscular dystrophy. In order to provide the best care that she can for her patients, Anne attended a specialist neuromuscular centre at the Hammersmith in England to learn about the physiotherapy sessions there and to receive guidance about best practice. Anne was very impressed by the care and expertise on offer at the Hammersmith and the difference made to quality of life. Anne wishes all patients with neuromuscular conditions had access to this level of support.

Patient Case No. 3

Patrick is a 19 year old young man with Duchenne muscular dystrophy. He is currently in transition from paediatric to adult services. He is in a wheelchair and attends a local college, he uses a computer, and the use of his hands is very important to his mobility and independence.

Patrick is now developing contractures (tightening and stiffening of the muscles) in his fingers. To help counter-act this problem, night splints on his hands are essential.

However, he has been discharged from the paediatric service where there was a community physiotherapist at hand, and no longer has access to on-going physiotherapy. Without specialist physiotherapy intervention to arrest the problem, he will lose the functional use of his hands faster than if he were to receive appropriate and timely specialist physiotherapy.

Clinicians recognise that the most effective way to deliver patient care for people living with a neuromuscular condition follows a 'hub and spoke' model with a specialist centre providing care to patients at the hub while also giving clinical leadership, supervising and sharing care with local hospitals and primary care teams.

The Newcastle Muscle Centre has for more than 15 years been providing 'outreach' clinics to several sites across the Northern Region of England including rural Cumbria, thereby providing specialist services closer to people's homes and allowing the better involvement of local staff.

The All Wales Medical Genetics Service operates on a 'hub and spoke' model and is successful in managing patients in their own community whilst benefiting from the knowledge and expertise of specialists based within the Institute of Medical Genetics, Cardiff.

A review of existing services should be undertaken in order to identify the composition of specialist neuromuscular services across Wales.

The following conditions need to be covered by a specialist multi-disciplinary service but may not necessarily be provided by a single centre:

- All genetic muscle diseases
- All genetic peripheral nerve diseases
- All acquired muscle diseases
- All acquired nerve diseases
- All genetic myasthenia
- All acquired autoimmune myasthenia

5. Welsh commissioners will not consistently fund diagnostic tests for Welsh patients at recognised specialist centres in England. The result is that Welsh patients receive inadequate diagnoses compared with the rest of the UK.

To ensure timely and accurate diagnosis, it is essential that all clinicians in Wales are able to access diagnostic services at recognised specialist centres in the UK.

Unfortunately, our findings reveal that Welsh health commissioners will not consistently finance the sending of lab samples to recognised specialist centres in England for diagnostic services including: specialised diagnostic strains for muscle pathology and Genetic DNA tests for specific rare disorders. Furthermore, while it is standard practice for some samples to be sent to specialist centres outside the UK, Welsh commissioners will not consistently fund this practice. The result is that some Welsh patients receive inadequate diagnosis compared with the rest of the UK.

6. Ventilation prolongs life but is not provided in a systematic way across Wales

Ventilatory support can enable many people with compromised respiratory function to live a longer and better quality life. It is vital that ventilatory services are available for people

with neuromuscular conditions if and when they need it, and that these services are provided by multi-disciplinary teams with experience in managing individuals with neuromuscular disease.

Commissioning of services for non-invasive ventilation across Wales is uncoordinated and is varied. For some areas, the LHBs and the specialist commissioner HCW are unclear on whose responsibility this is. This leads to delays for patients in receiving their treatment and equipment to aid them with their breathing.

There must be coordinated agreement between commissioning bodies on whose responsibility it is for ventilation services. Organisations must work together to ensure that patient well-being does not suffer and lives are not at risk through unclear commissioning procedures.

Consideration should also be given to establishing a coordinated ventilation service for individuals with neuromuscular disease, to ensure that individuals are assessed and treated efficiently and appropriately.

7. Service standards and protocols to ensure adequate care is given to patients with for the neuromuscular conditions

To ensure that the much needed improvements are made in service provision, work should be initiated without delay to develop an NHS Wales approved Standard of Diagnosis and Care for neuromuscular conditions, to be endorsed by the emergent British Myology Society. This would provide Health Commission Wales and LHBs with a benchmark against which neuromuscular services can be regularly assessed and is essential in maintaining and driving up standards across the country.

Between 2003 and 2006 the Muscular Dystrophy Campaign provided a three-year grant to Cardiff University (previously UWCM) to establish and coordinate a Wales Neuromuscular Network. The aim of the network was to improve neuromuscular services, knowledge and awareness about primary muscle disease amongst professionals and in doing so improve access to specialist care and advice.

The Network successfully organised meetings and seminars across Wales and they also began to develop much needed care pathways and standards of care. The service was well supported by leading clinicians, professionals, patient groups, patients and their families. Despite the advances made the Network did not receive any support from NHS Wales and its financial vulnerability led to its closure.

The major TREAT-NMD initiative which is co-ordinated from the Newcastle Centre (www.treat-nmd.eu) brings together some of the world's leading neuromuscular specialists in a pan-European 'network of excellence' and engages leading specialists worldwide to create international consensus on care and management for patients suffering from neuromuscular diseases. Consensus has already been agreed by an international group of experts for the standard of care of patients with spinal muscular atrophy, and this will follow for other diseases as the network develops.

Further, it is planned to launch the British Myology Society (BMS) in early 2008. The BMS will ensure that standards of care and best practice guidelines are developed and introduced in the UK for all neuromuscular diseases and will also support and facilitate relevant aspects of the work of TREAT-NMD. The BMS will also take on the role of the professional body in the UK that provides the clinical lead to commissioners regarding specialist service provision.

8. Local Health Board commissioning arrangements do not result in sufficient service provision for patients with neuromuscular diseases.

It can be said from the evidence in the MDC Survey that where neuromuscular services exist in Wales they are largely concentrated in South Wales. However, this does not equate to a comprehensive dedicated muscle clinic. Indeed, a patient might be able to see a clinician with specialist knowledge of neuromuscular conditions but will not have access to other specialist care and therapies.

Without the adoption of a Wales-wide strategy for the planning and development of specialist neuromuscular provision, services have evolved on an ad hoc basis dependent on interested and committed clinicians leading to the inherent fragility of this approach (as emphasised in this report).

9. Welsh commissioners are unable to fund treatment at centres in England which border with Wales

Some Welsh health commissioners are unable to refer patients with neuromuscular conditions to the specialist neuromuscular centres in England which border with Wales.

Specialist clinicians have identified one LHB bordering England which is unable to commission services from Oswestry due to funding constraints. Clinicians at this specialist centre are very concerned about how patients in the LHB area will receive adequate and essential specialist treatment in their area. Furthermore, the lack of contracts set up with the level of specification sought after is of serious concern.

10. Local Health Board commissioning arrangements do not result in adequately resourced clinics to care for patients effectively

There is no requirement within the commissioning structure to ensure that the staffing levels within paediatric neurology departments include a specialist in neuromuscular diseases and most departments rely on the individual interests of applicants for posts rather than recruiting directly a specialist with an interest in a neuromuscular disease.

In addition, the MDC survey of LHBs shows that where there are specialist clinics they are not consistently able to provide a comprehensive multi-disciplinary approach to care. The survey findings support those reported by Hill and Phillips (2006) who identified that 32% of clinics did not routinely monitor respiratory function and 10% did not routinely perform Electrocardiograms (ECGs). They further recorded that follow-up was variable; physiotherapists only offered long-term review of patients in seven centres, only five of which had a specialist muscle physiotherapist⁶.

11. Local Health Board commissioning arrangements for neuromuscular conditions do not result in effective collaboration of services in certain regions

The MDC survey of LHBs revealed a worrying lack of communication internally between LHBs over their collaboration and commissioning arrangements.

While some LHBs replied with clear concise information, it was apparent from certain LHB responses that there was a lack of knowledge of what was provided in their own area and even of the definition of the clinic which we were enquiring about. This was reflected in referrals to both NHS Trusts and HCW.

The LHBs who responded in detail by providing information about the services in their own area and also supplying new information about surrounding areas whose responses had already been received reflected a fundamental breakdown of communication between LHBs. Patients are therefore severely inconvenienced by the fact that they are currently having to travel much further than should have to in order to receive treatment.

12. Boys diagnosed with Duchenne muscular dystrophy (DMD) by the Welsh newborn screening programme must receive specialist follow-on treatment and care

The newborn screening programme for DMD in Wales is the only one of its kind in the UK. Established since 1990 the programme tests for DMD as part of the newborn blood spot screening programme as an additional parental opt-in test. The aim of the programme is to ensure that, where parents request this, boys with DMD are diagnosed early enough so that their condition may be managed effectively and thoroughly. It can also help families avoid a prolonged diagnostic process and can help them plan for the future. They are also in a position to benefit from reproductive choice in any future pregnancies.

Currently, a number of boys in Wales are not receiving access to adequate follow-up treatment and specialist care. Clinicians agree that boys with DMD and their families need specialist healthcare in addition to accurate and timely information, support and follow-up provided by a multi-disciplinary team of specialist professionals.

Action required

- An urgent review of existing specialist neuromuscular services should be undertaken. This will assess current gaps in service provision and lead to action to address any weaknesses in local provision.
- Many neuromuscular conditions are low-incidence conditions and some are ultra orphan. Neuromuscular conditions require a multi-disciplinary specialist approach to care and comprehensive neuromuscular services should be designated within the Health Commission Wales (Specialised Services) Commissioning Plan. This would provide an important basis for commissioning, service reviews and strategic planning and enable commissioners to establish a broad base-line position and make initial comparisons on activity and spend.
- In collaboration with existing centres and clinics across the UK, patient care should be delivered by a 'hub and spoke' arrangement with the specialist centre at the hub providing care and also sharing care provision with local clinics based in hospitals and primary care teams. While details of the service model may vary between patients, the key is that specialist supervision supports and oversees local provision.
- An NHS Wales approved Standard of Diagnosis and Care for the neuromuscular conditions should be developed and agreed. The British Myology Society, to be launched in 2008, will endorse such standards for diagnosis and care and it will also support the work of TREAT-NMD, an international initiative backed by NHS Wales and the Department of Health in England, so that agreed standards of care and best practice guidelines are introduced in the UK for all neuromuscular diseases. These steps will provide standards against which the service can be assessed by commissioners and lead to consistent, high quality care with improved patient outcomes.
- The agreed Standards of Diagnosis and Care should be disseminated throughout Wales, including to patients and patient groups. This would ensure equity of access both to specialist multi-disciplinary services and to non-specialist services for general medical problems, and provide assurance to patients of the level of care they can expect.
- The needs of patients also require a renewed focus on workforce planning with, in particular, specialist posts to be designated and established within the specialist centres and clinics. Further, the issue of succession planning should be addressed urgently especially for the lead clinician posts which are vital to the specialist neuromuscular service.

Appendix 1 – MDC Survey of Local Health Boards in Wales

Introduction

The purpose of the survey was to identify which areas in Wales commission specialist muscle clinics for children and adults with neuromuscular conditions.

Method

In December 2007, the Muscular Dystrophy Campaign contacted by email 22 Local Health Boards in Wales and under the Freedom of Information Act asked the following questions:

- *Does your Local Health Board currently support a muscle clinic that offers a comprehensive service to (a) children and (b) adults with a neuromuscular condition?*
- *If you do support a muscle clinic for children and/or adults, where is the clinic located and who is the lead clinician/Head of service?*
- *If patients are referred out of the local area, please could indicate this and provide details.*

Local Health Boards who responded

Out of the 20 LHBs that have so far responded, the following picture has emerged:

- **75%** of LHBs who responded do not support a muscle clinic that offers a service to adults with neuromuscular conditions.
- **70%** of LHBs who responded do not support a muscle clinic that offers a service to children with neuromuscular conditions.
- **70%** of LHBs who responded do not support any adult or child muscle clinics within their area.

Services identified

The following table and graph set out the percentage of responding LHBs who commission a muscle clinic.

Age group	Percentage of LHBs without clinics
Have child clinic but no adult clinic	5%
Have neither adult or child clinic	70%

It can be said from the evidence submitted by the LHBs that there is limited provision of muscle services in Wales, with a large concentration of the services in the southern regions. Where services are provided, it is often unclear how specialised the service is and patients have to travel long distances within Wales and to London to receive this treatment.

Approximately 50% of the LHBs responded by saying that Health Commission Wales should be contacted for information on the services provided.

The lack of contracts set up with the level of specification sought after is of serious concern. Action is urgently required regarding the paucity of clinics in Wales and the reluctance to commission specialist English services situated close to the Welsh border. The lack of contracts set up by the Health Commission Wales with the level of specification sought after is of serious concern.

Appendix 2 – Clinics for Welsh Patients with Neuromuscular conditions

Paediatric Management Clinics			
Area	Clinic type	Frequency	
South East Wales			
Cardiff	Muscle	Fortnightly	
Aberdare	Muscle	6 monthly	*
Nevill Hall Hospital	Muscle	6 monthly	
South West Wales			
Swansea	Muscle	Monthly	*
Haverfordwest	Muscle	6 monthly	*
Llantrisant	Muscle	3 monthly	*
Mid Wales			
No local services. Patients attend Oswestry Neuromuscular Centre.			
North Wales			
No local services.			
Patients attend Oswestry Neuromuscular Centre or Alder Hey Hospital, Liverpool.			
Oswestry	General	1-2x weekly	**

Adult Management Clinics			
Area	Clinic type	Frequency	
South East Wales			
Newport	Neuromuscular	Monthly	
Cardiff	Peripheral nerve	Fortnightly	
	Muscle	Fortnightly	
	Neuromuscular ventilation	Every other month	
South West Wales			
Swansea	Neuromuscular	Fortnightly	
Haverfordwest	Muscle	6 monthly	
Mid Wales			
No local services.			
Patients are known to attend the Oswestry Neuromuscular Centre or the Walton Centre, Liverpool			
North Wales			
No local services.			
Patients are known to attend the Oswestry Neuromuscular Centre or the Walton Centre, Liverpool.			

* *Physiotherapy intervention exists in some paediatric clinics. However, not all physiotherapists are specially trained nor do they have expertise in neuromuscular conditions. These services are reliant on the interest of the therapist and are not strategically planned.*

The services that are available are dependent on the locality of the patient. A patient from outside the Local Health Board area who is attending the clinic would not be eligible to receive any physiotherapy.

** A specialist physiotherapy services and specialist muscle pathology services exists at the Oswestry Neuromuscular Centre.

Glossary

BMS	British Myology Society, emerging professional body
DMD	Duchenne Muscular Dystrophy
ECG	Electrocardiogram – a test that measures the electrical activity of the heart
FOI	Freedom of Information Act
FTE	Full time equivalent – used in assessing staffing levels
GIG	Genetic Interest Group
LHB	Local Health Board
MDC	Muscular Dystrophy Campaign
NMD	Neuromuscular disease
OT	Occupational therapy and occupational therapist
RCA	Post of Regional Care Adviser
TREAT-NMD	Major European network aiming to improve treatments and find cures for neuromuscular disorders
UWCM	University of Wales College of Medicine
WTE	Whole time equivalent - used in assessing staffing levels

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