Scottish Muscle Managed Clinical Network
Service Description Document
and
Patient Expectation of Care
October 2015

Acknowledgments

The Scottish Muscle Network would like to thank all of our partners and stakeholders who contributed to this document. In particular we would like to thank:
Laura Craig (Network Co-ordinator)
Karen Robertson (Senior Programme Manager)
Carsten Mandt (Network Manager)
The Scottish Muscle Network Steering Group
The National Services Division, Scotland.
Charities including: The Muscular Dystrophy Campaign, Action Duchenne, Myaesthenia Gravis, Myotonic Dystrophy Group

We would also like to thank all of our patients and their families and carers for their continued help and support to improve services to patients with neuromuscular conditions across Scotland.

Marina Di Marco
Clinical Lead of the Scottish Muscle Network

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Section One:
Expectations of Care of patients with a neuromuscular condition

I. Introduction

There are more than 6,000 people in Scotland affected by inherited and acquired neuromuscular disorders. Examples include muscular dystrophies, myositis, mitochondrial diseases, motor neuron diseases, peripheral neuropathies and disorders of the neuromuscular junction such as myasthenia gravis. Current patient numbers for the more common of these conditions are available. Although much progress has been made in the diagnosis and management of these diseases, neuromuscular conditions are in large part progressive, debilitating and can frequently be life shortening. Patients commonly require complex and long term management.

Services for those affected by neuromuscular conditions are delivered across Scotland through a collaborative networked approach. Care is routinely delivered as local as possible but travel may be necessary for diagnosis and to access specialist expertise or specialist facilities.

The key aims of the service are to provide:

- Timely access to diagnosis
- Safe, effective and patient-centred management
- Timely access to the relevant specialist expertise when required
- Ongoing support for patients and their families and carers

This document describes the care that all persons who are suspected of having, or have been diagnosed with, a neuromuscular condition can expect to receive in NHS Scotland. This should form the basis of a discussion and shared decision-making between the patient and their carers and the healthcare professionals responsible for their care. However, it is essential that all care is patient-centred, appropriate for and sensitive to the needs of each individual patient. As such there may be good reasons for the actual care provided to an individual patient to differ from what is described in this document.

II. Diagnosis

1. A diagnosis may be established within a Primary Care or non-specialist clinic. If making an accurate diagnosis requires a referral to a specialist with experience in neuromuscular disease, a referral will be made to the appropriate specialist at the earliest possible time to minimise any delay in diagnosis.

2. If further investigations are required to establish diagnosis, a referral will be made to the relevant service at the earliest possible time to minimize any delay in diagnosis. These investigations include: muscle biopsies, neurophysiological studies, genetic studies and muscle imaging.

3. In some of the rarer conditions additional investigations may be required that are only available in other parts of the UK. Where additional investigations are required a referral will be made as soon as possible in accordance with agreed referral pathways.

4. At the time of diagnosis patients and families will be given generic information on services available as well as disease specific information. Some of this information will be summarised in the form of a clinic letter, whilst other information may be in the form of an alert card, information leaflet (SMN , Muscular Dystrophy UK or other charity web site)

5. All persons with a confirmed diagnosis of a neuromuscular condition will prospectively be added to the electronic patient register which is currently being developed. This electronic patient register is held in the Scottish Muscle Network Clinical Audit System (please note that all **patients have the right to opt out** of being added to this register).

III. Management

1. Initial Management

- Depending on the individual diagnosis, a referral to genetic, cardiac, respiratory or other specialist services will be made as required to ensure appropriate holistic management of the individual patient's healthcare needs.

2. Ongoing Management

- Every person with a confirmed diagnosis will have access to a named healthcare professional providing direct support ensuring timely access to relevant information, links into related services (e.g. social care, employability, financial inclusion, housing etc), continuity of care and support at times of crisis or change (e.g. transition to adult life, loss of mobility, loss of employment, death). It is anticipated that this support will be available in the form of a Neuromuscular Care Advisor but equivalent other arrangements maybe in place in some areas.

- Every person diagnosed with a neuromuscular condition will be offered long term follow up to ensure safe and effective ongoing management of their condition and any associated symptoms. Management will be based on existing agreed guidelines and protocols for the particular condition. Further information on agreed protocols and published guidelines is available on the Scottish Muscle Network website: http://www.smn.scot.nhs.uk/

- Wherever possible ongoing management will be delivered close to home. However, if specialist input is needed this may only be available in the four main centres Glasgow, Edinburgh, Dundee and Aberdeen.

An overview of the Neuromuscular Population in Scotland

Describing exact demographic information on patient populations is not possible. Many mildly affected patients do not require to be looked after within a specialist team and can remain well looked after but largely unknown to the specialist team. For the same reason, many patients remain undiagnosed with their neuromuscular disorder as they are so mildly affected that in some circumstances they are unaware they even have a neuromuscular condition. There is also a cohort of patients who are known to have a neuromuscular disorder but their precise diagnosis cannot be determined as the specific genetic mutation has not been identified.

In order to extrapolate the incidence of disease specific populations, it is accepted that statistics outlined by Norwood et al (2009) http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4038491/ can be utilised within the Scottish population however, below is an estimation of three of the most common neuromuscular conditions

Disorder	**Number in Scotland**
Duchenne Muscular Dystrophy	approximately 140
Myasthenia gravis	approximately 1000
Myotonic Dystrophy	approximately 600

Figure One: An overview of the Neuromuscular Population in Scotland (SMN, 2015)

IV. Clinical Genetic Counselling

The majority of diseases are inherited and therefore genetic counselling is important for families. All persons are referred following confirmation of a genetic disease for further advice as appropriate.

Section Two: Scottish Muscle Network Activity

The Scottish Muscle Network supports a number of sub group specialities which include:
The Adult Neuromuscular sub group, the Paediatric Neuromuscular Sub Group, the Myotonic Dystrophy Sub Group, the Education and Editorial Sub Group and the Physiotherapy Sub Group. These sub groups undertake specific work related to the area of expertise as well as collaborating on larger pieces of work of benefit to the neuromuscular population in Scotland led by the Steering Group. The following are examples of sub group activity:

- **Duchenne MD**: The paediatric sub group have developed Scottish standards of care that encompass the UK standards of care (Bushby et al), The North Star protocols and the TREAT NMD guidelines. The Scottish DMD Physiotherapy profile developed by the physiotherapy sub group dovetails with the Scottish standards of care and is updated and audited every 3 years. This profile is being downloaded and used in a variety of different countries such as Australia and Switzerland and has been translated into German.

  DM1: In addition to neuromuscular symptoms such as muscle weakness, fatigue and respiratory problems, myotonic dystrophy can be associated with a range of issues affecting all systems of the body which may be overlooked by the inexperienced clinician. Patients in Scotland are therefore invited for annual review appointments co-ordinated by the Myotonic Dystrophy Subgroup of the SMN. Common data relating both to clinical indicators and management are recorded on a standardised database, which has enabled the subgroup to establish and audit standards of care for patients with myotonic dystrophy. This represents one of the few national services for specific management of a neuromuscular disorder that exist in the world.
- **Myasthenia Gravis**: Dr Maria Farrugia leads an expert MG service based in Glasgow and is the Scottish MG expert within the main steering group. Some 500 patients attend the myasthenia clinics in Glasgow. The myasthenia nurse (Sr Caroline Carmichael) provides telephone advice, assists with the MG clinics and also with nurse-led clinics. In the ocular myasthenia clinic, an orthoptist also assists with patients' evaluation. Management of these patients may sometimes include thymectomy which is carried out by a the surgeon Mr Alan Kirk in Golden Jubilee Hospital and plasma exchange with the plasmpheresis team being based at the Beatson and led by Dr Kenny Douglas. Currently national MG guidelines are being developed.
- **Fascioscapulohumeral muscular dystrophy:** A management protocol has been developed and published on the network website by the adult clinicians. This has informed a care pathway under development in Glasgow and Edinburgh. Patients are regularly assessed with regard to muscle function and change but are also monitored by respiratory physicians. Some patients require close input by other Allied Health professionals e.g. physiotherapists, orthotists, occupational therapists and pain management teams.
- **Mitochondrial disease**: The network has well developed historical links with the Rare Mitochondrial Disease group in Newcastle funded by the UK National Commissioning Group "NCG".

In addition to the work of the sub groups, the Scottish Muscle Network host a number of education events for professionals throughout the year.

The Muscle Interest Group: The Muscle Interest Group (MIG) led by Dr. Maria Farrugia is a bi-annual event encouraging professionals to present challenging undiagnosed and diagnosed cases and is hosted in various locations around the country. This enables professionals to highlight difficult patient scenarios to their peers for advice, discussion and information sharing. Each MIG meeting features an invited guest speaker well known in their field of expertise to present and offer comment on patient cases.

The Annual Conference: The annual conference is held in a rotational venue either in the North, East or West of Scotland and features updates on research from around the world as well as research being undertaken in Scotland. This conference invites doctors, nurses and allied health professionals who either specialise or have a special interest in neuromuscular conditions. Each year there are in excess of 80 attendees and an opportunity to those in remote and rural locations via video link access is provided with an average of four sites video-conferencing each year.

The AHP Conference: The Allied Health Professional Conference led by Marina Di Marco is accessed each year by approximately 80 healthcare professionals. This conference is primarily hosted in the West of Scotland where the majority of attendees reside to facilitate ease of access of attending in person and easily reached by public transport. An opportunity to video conference is extended to all areas.

Charity Conferences: The Scottish Muscle Network work in partnership with a number of charities and will present on areas of interest to patient organisations and third sector groups. These groups include Muscular Dystrophy UK, Action Duchenne, Muscular Dystrophy Branch meetings, West of Scotland Cardiomyopathy Group and MDUK patient focus groups.

Further information on agreed protocols and published guidelines is available on the SMN website http://www.smn.scot.nhs.uk/

Section Three: Patient Pathways

Generic Pathway for a Paediatric patient with a Neuromuscular Condition

- *Referral Pathway*: Children are usually referred into the neuromuscular service via Primary and Secondary care from a variety of different healthcare professionals. Some children are referred for specialist opinion when the diagnosis is uncertain, and others are referred to established clinics for on-going medical care.

- *Not all neuromuscular patients have a diagnosis*: If a diagnosis is not clear, referral to NM geneticist may be required and consideration of additional testing, or referral to NM services outwith Scotland. A number of children and young people may still not have a definitive diagnosis within the current diagnostic testing spectrum. All children with a NM condition should be under the care of a Consultant Neurologist or identified paediatrician with specialist neuromuscular interest. The lead paediatrician should coordinate medical care and ensure that all other appropriate referrals are in place.

- *Anaesthesia*: Some anaesthetics are contraindicated in certain neuromuscular disorders. Advice should be sought for both acute and planned admissions.

- *Audiology:* Certain neuromuscular disorders are associated with hearing disorders such as Mitochondrial disorders, Fascioscapulo-humeral muscular dystrophy as well as neurological conditions such as Friedreich's ataxia. Referral for audiology assessment should be considered when risk is identified or if hearing loss is considered.

- *Cardiology:* Some neuromuscular conditions have an associated cardiomyopathy or cardiac rhythm abnormality. Referral to a paediatric cardiologist for cardiac assessment should be made in conditions with a recognised association or if the NM condition has no definitive aetiology. Referral of family carriers should also be considered with conditions like BMD and DMD.

- *Care and Support:* Referral to a Neuromuscular care adviser or local support service should be offered to all families after diagnosis for emotional support, advocacy and advice.

- *Diabetes Service:* Certain conditions have a predisposition to diabetes such as Friedreich's Ataxia and some Mitochondrial disorders. Screening of blood glucose should be considered when appropriate and referral to the diabetes team for advice if indicated.

- *Dietitian:* Refer if patient has problems with dysphagia, aspiration, is under or overweight, has nutritional deficiency or is on long term steroid therapy.

- *Endocrine:* If bone health or growth are problematic such as when on long term steroid therapy, a referral to a paediatric endocrinologist may be beneficial.

- *Genetics:* Genetic counselling for other family members and future pregnancies will be required.

- *Occupational Therapy*: Refer for home and school access assessment, advice about specialist equipment to support posture and mobility. Referral to social work OT for housing assessment/adaptation.

- *Opthamology:* In cases where eyesight can be affected such as those on long term steroid use, mitochondrial disorders, and certain muscular dystrophies such as Fascio-scapulo-humeral muscular dystrophy and nerve disorders such as Friedreich's ataxia, a referral to Opthamology may be necessary.

- *Orthopaedics:* Referral to orthopaedics should be made in the event of fixed contracture, hip subluxation or scoliosis development.

- *Orthotics:* Refer if child requires support at joints for mobility, postural management or specific pain relief associated with joint instability or over use.

- *Pain Service*: In some cases, pain may be a major feature of the condition and a referral to the specialist pain service may be appropriate.

- *Palliative Care:* The palliation service can be involved if the child has deteriorated significantly and not expected to survive into adulthood. End of life may not be imminent but palliative care services such as CHAS can assist children and families to come to terms with the chronic illness and offer support and respite.

- *Physiotherapy*: Refer to the physiotherapy service for postural management, activities and mobility, passive stretching programme, MSK pain, and advice in relation to pain and fatigue. Specialist respiratory advice may also be required for chest care including secretion management, assisted cough techniques, and chest physiotherapy.

- *Primary and Secondary Care:* Children may benefit from local primary care services in relation to District Nurses, rehabilitation teams, podiatry, wound care, *etc.*

- *Psychology / Psychiatry:* In many chronic paediatric disorders and long term conditions, children may have difficulties adjusting as they get older and their condition deteriorates. Children and young people may live with low mood disorder, anxiety and depression. A referral to Psychology / Psychiatry may be indicated.

- *Renal Service / Continence Care:* In some conditions, bladder and bowel control may be affected.

- *Respiratory:* Refer for respiratory assessment if condition has known respiratory complications. Refer if Epworth sleepiness scale is above 12, daytime somnolence, symptoms of obstructive sleep apnoea, weight loss, poor appetite and repeated chest infections.

- *Speech and Language Therapy:* Refer if patient has signs of dysphagia and / or aspiration. Advice should also be sought if child's communication skills are affected

- *Social Services:* Benefits, carers, housing and respite.

- *Spinal Service:* If spinal scoliosis develops a referral to the spinal service in Edinburgh may be indicated. If the patient is under 18 years of age they can be referred directly to the National Spinal Service, Edinburgh. If 18 years or over, the patient will be referred to the local Orthopaedic Consultant with a special interest in spinal deformity.

- *Transition:* Children will be transitioned to the appropriate adult services between the ages of 16 and 18 years.

- *Wheelchair and Seating Service:* The child may require a wheelchair for energy conservation and fatigue management when out of doors, periods throughout the day or full time. This will include manual and powered chairs with specific and bespoke therapeutic postural management system.

Generic Pathway for an Adult patient with a Neuromuscular Condition

- *Referral Pathway*: Patients are usually referred in to the neuromuscular service via Genetics, Primary and Secondary care from a variety of different healthcare professionals. Young people with a confirmed neuromuscular condition will be transitioned to neurology from children's services.

- *Not all neuromuscular patients have a diagnosis*: If a confirmed diagnosis is not clear, referral to a NM Geneticist may be required and consideration of additional testing, or referral to NM services outwith Scotland appropriate. In some cases, a definitive diagnosis may not be possible within the current diagnostic spectrum. The patient may be referred to a muscle clinic if available.

- *Anaesthesia*: Some anaesthetics are contraindicated in certain neuromuscular disorders. Advice should be sought for both acute and planned admissions.

- *Audiology:* Certain neuromuscular disorders are associated with hearing disorders such as Mitochondrial disorders, Fascioscapulo-humeral muscular dystrophy as well as neurological conditions such as Friedreich's Ataxia. Referral for audiology assessment should be considered if a risk is identified.

- *Cardiology:* Some neuromuscular conditions have an associated cardiomyopathy or cardiac rhythm abnormality. Refer for cardiac screening if condition is undiagnosed or if associated cardiac complications are known for the diagnosis. Referral of family carriers should also be considered with conditions like BMD or DMD.

- *Care and Support:* Referral to care adviser or local support service should be offered if the patient requires emotional support, advocacy and advice especially at time of diagnosis.

- *Diabetes Service:* Certain conditions have a predisposition to diabetes such as Friedreich's Ataxia and some Mitochondrial disorders. Screening of blood glucose should be considered when appropriate and referral to diabetes team for advice if indicated.

- *Dietitian:* Refer if patient has problems with dysphagia, aspiration, is under or overweight, has nutritional deficiency or is on long term steroid therapy.

- *Genetics:* Genetic counselling for other family members and future pregnancies may be required.
- *Occupational Therapy*: Refer to rehabilitation team for home assessment, transfers, work related difficulties and access issues. Refer to Social Work OT for housing assessment / adaptations.

- *Opthamology:* In cases where eyesight can be affected such as those on long term steroid use, mitochondrial disorders, and certain muscular dystrophies such as Fascio-scapulo-humeral muscular dystrophy and nerve disorders such as Friedreich's ataxia, a referral to Opthamology may be necessary.

- *Orthopaedics:* In cases of postural deformity, a referral to orthopaedics may be required such as in Charcot Marie Tooth foot deformity or hip dislocation in SMA.

- *Orthotics:* Refer if patient requires support at joints for mobility, postural management or specific pain relief associated with joint instability or over use.

- *Pain Service*: In some cases, pain may be a major feature of the condition and a referral to the specialist pain service may be appropriate.

- *Palliative Care:* The palliation service can be involved when the patient has deteriorated. End of life may not be imminent but palliation can assist families and patients to come to terms with the chronic illness and offer support and respite.

- *Physiotherapist*: Refer for postural management, mobility, MSK pain and fatigue. Specialist respiratory advice may also be required for chest care including secretion management, assisted cough techniques and chest physiotherapy.

- *Primary and Secondary Care:* Patients may benefit from local primary care services in relation to district nurses, rehabilitation teams, wheelchair services, podiatry and wound and pressure care.

- *Psychology / Psychiatry:* In may long term conditions, coping mechanisms can be affected and patients may live with low mood disorder, anxiety and depression. A referral to Psychology / Psychiatry may be indicated.

- *Renal Service / Continence Care:* In some conditions, bladder and bowel control may be affected.

- *Respiratory:* Refer for respiratory assessment if condition has known respiratory complications. Refer if Epworth sleepiness scale is above 12, daytime somnolence, symptoms of obstructive sleep apnoea, weight loss, poor appetite and repeated chest infections.
- *Speech and Language Therapy:* Refer if patient has signs of dysphagia and / or aspiration. Advice should also be sought if communication skills are affected

- *Social Services:* Benefits, carers, housing and respite.

- *Spinal Service:* In cases of spinal scoliosis as a result of muscle weakness, a referral to the spinal service may be indicated. If the patient is under 18 years of age they can be referred directly to the National Spinal Service, Edinburgh. If 18 years or over, the patient will be referred to the local Orthopaedic Consultant with a special interest in spinal deformity.

- *Wheelchair and Seating Service*: The patient may require a wheelchair for energy conservation and fatigue management when out of doors, periods throughout the day or full time. This will include manual and powered chairs with specific and bespoke therapeutic postural management.

Section Four: Clinical Management Protocols developed by / followed by the Scottish Muscle Network

Facio-Scapulo-Humeral (FSH) Muscular Dystrophy: Standards of care and management in symptomatic adults

Introduction

FSH Muscular Dystrophy is an autosomal dominant disorder with a prevalence of up to 1:20,000. Onset may be at any time in life and some gene carriers may remain asymptomatic into old age. Muscle weakness slowly evolves through life and the management of this and associated features require a co-ordinated approach to long term care. These guidelines and statements are based on a recently published European Neuromuscular Centre (ENMC) evidence based standard and literature review and a consensus within the Scottish Muscle Network. They are applicable to clinically affected adult populations only (over 16 years of age). Some patients may be recognized as gene carriers but show no clinical manifestations (usually identified in the course of genetic counselling). In these patients the nature of any ongoing follow up and management is left to the discretion of the Clinical Geneticist involved to decide on the basis of patient age, wishes and co-morbidity.

Diagnosis

The diagnosis of FSH is established by the identification of a partially deleted D4Z4 repeat array on one chromosome. Patients with FSH have 1 - 10 D4Z4 units . The test for this mutation is readily available but interpretation of these reports is complex and requires further investigation in some 5% of case. It is therefore recommended that

> The diagnosis of FSH can be based on the detection of a short fragment at 4q35 on Southern Blot AND a typical clinical phenotype. In atypical cases the clinical features and molecular result need to be discussed with a clinical geneticist because of the possibility of both false positive and false negative results from the standard Southern Blot test. Advice should always be sought for further genetic input addressing the pedigree. The diagnosis of FSH in patients with intermediate alleles on Southern Blotting requires careful clinical and genetic correlation and should be discussed with a clinical geneticist.

Physical therapy

Axial weakness is a major feature of FSH and pain, especially in the low back a major problem affecting up to 77% of patients. There are no specific therapies for this but education regarding physical fitness and a full physical therapy evaluation is recommended for these patients.

All patients with functional limitations should be considered for a rehabilitation medicine referral.

There is support from one study for the role of aerobic training in FSH and this should be considered in patients able to participate

Aerobic exercise therapy should be considered as part of a neuro-rehabilitation strategy.

There is no evidence to support the role of drug treatment and no specific regimes or drugs are recommended.

No specific drug treatment is recommended.

Cardiac disease
There are isolated reports of both atrial tachy-arrythmias and of right bundle branch block but this has not yet been reproduced in long term or wider studies.

No routine cardiac surveillance in recommended. Any complaints suggestive of a rapid heart beat should be investigated fully for a alternative mechanism. An ECG at diagnosis is recommended.

Respiratory disease
Although it has been stated that respiratory insufficiency requiring overnight ventilatory assistance develops in less than 1%, up to 30% of symptomatic patients have weakness of respiratory muscles.

Respiratory review with ongoing follow up including FVC should be offered to all non-ambulant patients, those with scoliosis or with additional lung disease. In other patients enquiry should be made at review regarding symptoms of respiratory insufficiency.
All non-ambulant patients should have a formal respiratory physician review prior to elective general anaesthesia.

Swallowing
There is no evidence this is a major issue prior to advanced disease and no specific recommendations are made.

Deafness
Sensorineural hearing loss is recognized feature of FSH and may be a major manifestation in paediatric onset cases. In adults however there is debate over frequency (perhaps up to 75%) and severity of deafness. We would agree with the ENMC recommendation that

Adults diagnosed with FSH muscular dystrophy do not require audiological assessment unless they become symptomatic. Symptoms suggestive of evolving deafness should be sought at review appointments.

Visual symptoms
Retinal vasculopathy is common in FSH and in the paediatric group can affect visual function. Progression in adult life is rare but

All adults symptomatic with FSH should have a formal ophthalmological assessment looking for retinal vascular disease. No further follow up is required unless patients develop visual symptoms

Scapular fixation
A Cochrane review concluded that scapular fixation improved shoulder function in muscular dystrophy. However the selection criteria and best technique remain to be defined. Some patients may experience a small reduction in FVC following surgery.

All adults should be referred for consideration of the procedure if upper limb function is impaired by failure of scapular fixation. Referrers should identify a surgeon with a declared interest in the procedure.

Patient information
Many of the recommendations above carry the implication of actions to follow certain events. We would therefore recommend that

All adults with FSH are offered written FSH specific information regarding General Anaesthesia, visual and aural symptomatology with a contact number for further information.

PAEDIATRIC STANDARDS FOR MANAGEMENT OF DUCHENNE MUSCULAR DYSTROPHY IN SCOTLAND

Diagnosis

Diagnosis should be suspected in all pre-school boys with developmental delay. CK should be checked as part of the investigation work up

1. Paediatrician

When diagnosis is suspected all boys should be referred to an identified paediatrician with a special interest in muscle disorders

2. Investigation

When the diagnosis is suspected by grossly elevated CK and clinical history, DNA should be sent to Glasgow for DMD mutation analysis. Point mutation testing will not be done routinely if there is insufficient clinical history. If genetic studies are negative, an open muscle biopsy should be arranged (and sent to Newcastle through NSCAG)

3. Information

All families should be given written information about DMD after diagnosis

4. Genetic counselling

All parents should be offered referral for genetic counselling after diagnosis

Management

1. Paediatrician

All families should have an identified lead Paediatrician with an interest in NM disorders to co-ordinate their on-going care.

2. Physiotherapy

Following diagnosis all families should be referred to a local physiotherapist, who in turn has access to support from a specialist NM physiotherapist.

3. Occupational therapy

Following diagnosis all families should be referred to a local occupational therapist.

4. Steroids

All families with ambulant boys with DMD should have the opportunity of an informed discussion with their Paediatrician about the role of steroid treatment in DMD, and the North Star parent steroid information leaflet should be made available.

5. Cardiovascular management

Following diagnosis all boys should be referred to paediatric cardiology services for cardiomyopathy screening, every 2 years while ambulant, and annually thereafter. Consideration of early treatment should be given when appropriate.

6. Spinal management

Following loss of ambulation, or earlier if clinically indicated, all boys should be referred to the Scottish National Paediatric Spinal Surgical Service, and the parent information leaflet on spinal surgery should be made available.

7. Immunisation

All non-ambulant boys should be offered annual influenza and updated pneumococcal vaccination as recommended by immunisation guidelines.

8. Respiratory Management

Following loss of ambulation, or sooner if clinically indicated, all boys should be referred to a Paediatric Respiratory Specialist for respiratory monitoring and management advice. All families should be offered a timely referral to the appropriate home ventilation services, prior to symptoms of nocturnal hypoventilation developing.

9. Transition

It is the responsibility of the lead paediatrician to ensure that timely and appropriate referrals are made to adult specialist services with clear information to the primary care team.

Section Five: Research and Publications

SCOT-DMD: Research Team: Dr. Shuko Joseph; Dr. Jarod Wong; Dr. Iain Horrocks; Dr. Faisal Ahmed
Funding: Chief Scientists Office, Edinburgh, Muscular Dystrophy UK and Action Duchenne.

As part of the PhD fellowship application, there are 4 designated projects around the area of bone morbidity in DMD involving all of Scotland.

Study 1: Retrospective study of bone monitoring and fractures in all Scottish boys with DMD currently managed in paediatric care.
Study 2: Retrospective audit of fractures of all UK boys with DMD in the Northstar database.
Study 3: Prospective 2 year study of bone morbidity of boys with DMD.
The reason for this prospective study is 2 fold.

1- Fractures especially vertebral fractures occur in DMD. All current studies are retrospective studies reporting only symptomatic vertebral fractures (VF). Nationwide studies from the whole of Canada in other groups of children such as nephrotic syndrome, ALL and inflammatory rheumatic conditions have shown that a proportion of children with VF will be missed if relying on self reported symptoms as some vertebral fractures are asymptomatic. In Glasgow, there is a new DXA machine, the iDXA, only available in a few paediatric centres in the UK which allows for a more specialised assessment of the vertebrae in addition to measurement of bone density and it is the iDXA which will be used in this study. The radiation dose of vertebral assessment using DXA is about 15-20 times less than conventional spinal x ray. Thus, this will be the first prospective large scale study in DMD which can give information on the incidence of vertebral fractures and explore the utility of the DXA for diagnosis of VF in boys with DMD for the first time. Visits for those outside Glasgow will be annually. Clinical care can be delivered in local centres.
2- There is increasing recognition of the limitation of DXA BMD and we have been working on micro MRI to study bone microarchitecture in various conditions like OI, diabetes and growth hormone deficiency. A subset of boys aged 10-16 years will also have a single MRI for bone microarchitecture/muscle and also MRS for bone marrow adiposity. Only a very small handful of centres worldwide are conducting research in bone using MRI in children. The study is fortunate to have access to the 3T research MRI scanner, not used for paediatric bone research any where else in the UK.

Study 4: A survey of patients and families regarding bone health/fractures and to determine if families would be interested to participate in future possible clinical trials on bone protective therapies in DMD. Families would also be asked if they would be willing to input to the design of such possible clinical trials.

The DM1-Neuro Study: Understanding the relationship between genetic factors, and the effects of myotonic dystrophy type 1 on the brain.

Dr Mark Hamilton is Specialty Registrar in Clinical Genetics, who has recently received funding from Muscular Dystrophy-UK and the Chief Scientist Office to undertake a 3 year Clinical Research Fellowship in myotonic dystrophy type 1. His project, titled 'The DM1-Neuro Study' will be supervised by Prof Darren Monckton, Professor of Human Genetics at the University of Glasgow, and Dr Maria Farrugia, Consultant Neurologist and Lead Clinician for the West of Scotland Muscle Service. The study will investigate how the genetic changes that occur in myotonic dystrophy type 1 affect the severity of the brain-related symptoms associated with the condition, which may include excessive sleepiness and impairment of some aspects of thinking. A better understanding of how genetic factors relate to the effects of myotonic dystrophy type 1 on the brain will help to identify new strategies for the treatment of these extremely debilitating symptoms. In addition, improved understanding of the mechanisms which underlie variation in symptoms between individuals will be highly valuable in designing future clinical trials.

What are the researchers aiming to do?

Myotonic dystrophy type 1 is an extremely variable, inherited condition causing muscle weakness. It may also affect other organs such as the heart, eyes and the brain. The effects on the brain can result in slowed thinking and sometimes extreme daytime sleepiness. These symptoms can have profound impacts on the quality of life of people affected and their families.

The condition is caused by an increase in the number of DNA repeats in the *DMPK* gene. A larger number of repeats are associated with more severe symptoms and an earlier age of onset. The number of repeats frequently increases from one generation to the next, causing more severe symptoms and earlier age of onset in succeeding generations. The number of repeats is also seen to increase throughout the life of the person affected. Blood tests that are currently used to diagnose the condition only measure the approximate number of repeats and take no account of the changes during the person's life.

In this study, Dr Hamilton will work with the existing research team led by Prof Monckton at the University of Glasgow to use a more accurate method of measuring the genetic changes that occur throughout the life of a person affected by myotonic dystrophy type 1. The researchers will determine how the genetic changes relate to the severity of the brain-related symptoms using MRI brain scans, sleep studies and tests of memory and thinking. They will then look at how changes in specific structures of the brain relate to impairment of different aspects of thinking and sleep.

How will the outcomes of the research benefit patients?

The results of this study may allow the patients and their families to be offered more specific information about how the condition is likely to affect them in the future based on their genetic results. Understanding how genetic changes and changes in brain structure relate to the symptoms of the condition will also help identify mechanisms that can be targeted by drugs in the future. In addition, this work will be an important step to improve readiness for clinical trials in myotonic dystrophy, by providing new insights into why symptoms vary so considerably between individuals. Data from this study will also help to identify which tests are most relevant to determine whether new therapies are effective in treating the brain symptoms of myotonic dystrophy.

Publications:

These are listed from establishment of the muscle network in 1999

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Poster presentations

2013 IDMS meeting (the Myotonic Dystrophy group)

- Myotonic Dystrophy guidelines and the critical importance of resourcing and support
- Assessing quality of care in myotonic dystrophy:the Scottish experience
- The challenge of increasing attendance and myotonic dystrophy clinics
- Do guidelines guide the generalist? The Scottish experience
- The Scottish myotonic dystrophy database: What can it tell us?
- Patient-reported weakness, myotonia and swallowing difficulties in the UK Myotonic Dystrophy patient registry
- UK Myotonic Dystrophy patient registry: developing standards of care and clinical research

Chapter Six: Mapping of Neuromuscular Services in Scotland

This service map is an example. For the most up to date service map please refer to www.smn.scot.nhs.uk

West of Scotland : Neuromuscular Services - PAEDIATRIC

Health Board Area	DGH	Regional Service	Paediatrician / Paediatric Neurologist	Respiratory	Cardiology / Anaesthetics	Physio-therapy	Neuro-muscular Specialist Nurse	Neuromus-cular Care Adviser/ Support Services	Genetics	Neuro-physiology	DM Ser-vice	Neuro-pathology	AHP / Other
Ayrshire and Arran	Crosshouse Hospital, Rainbow House	RHC, Queen Elizabeth University Hospital	Dr Nuno Cordeiro / Dr Iain Horrocks	Dr Phil Davies , RHC, Queen Elizabeth University Hospital	Dr Patrick Noonan Dr Ben Smith (Glasgow) Dr Maria Ilina (Glasgow) Dr. Jon Staines (Ayrshire)	Fiona Gaffney David Cassels Marina Di Marco	Jen Dunne	Wilma Stewart (NCA)	Dr Cheryl Longman Lesley Snadden	Dr Iain Horrocks	Dr Nuno Cordeiro	Dr William Stewart	Clinical Psychologist Alan S. James Rachel Mochrie (Dietitian)
Forth Valley	Forth Valley Royal Hospital	RHC, Queen Elizabeth University Hospital	Dr Ishaq Abu-Arafeh / Dr Iain Horrocks	Dr Phil Da-vies , RHC, Queen Eliza-beth Univer-sity Hospital	Dr Ben Smith	Rachael Livingston Kendra Cleland Marina Di Marco	Jen Dunne	Wilma Stewart (NCA)	Dr Cheryl Longman Lesley Snadden	Dr Iain Horrocks	Dr Ishaq Abu-Arafeh	Dr William Stewart	Rachel Mochrie (Dietitian)
Greater Glasgow and Clyde	RHC, Queen Elizabeth University Hospital	RHC, Queen Elizabeth University Hospital	Dr Iain Horrocks	Dr Phil Davies	Dr. Maria Ilina / Dr Ben Smith	Marina Di Marco Sarah Brown Susanne McKenzie	Jen Dunne	Wilma Stewart (NCA)	Dr Cheryl Longman Lesley Snadden	Dr Iain Horrocks	Dr Iain Horrocks	Dr William Stewart	Rachel Mochrie (Dietitian)
Greater Glasgow and Clyde	Inverclyde Hospital, Panda Centre	RHC, Queen Elizabeth University Hospital	Dr Iain Horrocks Dr Lesley Nairn	Dr Phil Davies	Dr Maria Ilina/ Dr Ben Smith	Marina Di Marco Sarah Brown Susanne McKenzie	Jen Dunne	Wilma Stewart (NCA)	Dr Cheryl Longman Lesley Snadden	Dr Iain Horrocks	Dr Iain Horrocks	Dr William Stewart	Rachel Mochrie (Dietitian)
Lanarkshire	Wishaw General Hospital	RHC, Queen Elizabeth University Hospital	Dr Mary Callaghan / Dr Iain Horrocks	Dr Phil Davies / No identified local service	Dr Maria Ilina / Dr Ben Smith	Sheila Milligan Gillian Sutherland Marina Di Marco	Jen Dunne	Wilma Stewart (NCA)	Dr Cheryl Longman Lesley Snadden	Dr Iain Horrocks	Dr Mary Callaghan / Dr Iain Horrocks	Dr William Stewart	
Dumfries and Galloway	DGH Royal Infirmary/ Galloway Community Hospital	RHC, Queen Elizabeth University Hospital	No identified local service / Dr Iain Horrocks	Dr Phil Davies / No identified local service	Dr Maria Ilina	Marina Di Marco	Jen Dunne	Wilma Stewart (NCA)	Dr Cheryl Longman Lesley Snadden	Dr Iain Horrocks	Dr Iain Horrocks	Dr William Stewart	
Highland and Argyll	Lorn and Islands Hospital, Oban	RHC, Queen Elizabeth University Hospital	Dr Jamie Houston / Dr Iain Horrocks	Dr Phil Davies / No identified local service		Marina Di Marco Liz Taylor	Jen Dunne	Wilma Stewart (NCA)	Dr Cheryl Longman Lesley Snadden	Dr Iain Horrocks	Dr Iain Horrocks	Dr William Stewart	Julie Burslem (PT)

West of Scotland : Neuromuscular Services - ADULT

Health Board Area	DGH	Regional Service (Neuro only)	Neurologist	Respiratory	Cardiology / Anaesthetics	Physio-therapy	Neuro-muscular Specialist Nurse	Neuromuscular Care Adviser/ Support Services	Genetics	Neuro-physiol-ogy	DM Service	Neuro-patholo-gy	AHP / Other
Ayrshire and Arran	Ayrshire Central Hospital, Douglas Grant Rehab Centre	Queen Elizabeth University Hospital	Dr Maria Farrugia	Dr D Sword (Ayr Hospital) Dr Scott Davidson (QEUH)	Dr Iain Findlay	Marina Di Marco / Douglas Grant Rehab Centre		Wilma Stewart (NCA)	Dr C Longman Dr V Murday Lesley Snadden	Dr Arup Mallik	Dr Bob Ballantyne	Dr William Stewart	Douglas Grant Rehab Centre: June Milligan (Dietitian) Alison Clarke (Breathing Support
Forth Valley	Forth Valley Royal Hospital	Queen Elizabeth University Hospital / Western General Hospital	Prof Malcolm Macleod, Dr Christian Neumann, Dr Suvankar Pal /Dr Katy Murray	Dr Scott Davidson (QEUH) / Dr. Anthony Bateman (Edinburgh)	Dr Iain Findlay Dr Allister Hargreaves	Marina Di Marco		Wilma Stewart (NCA)	Dr C Longman Dr V Murday Lesley Snadden	Post vacant	Dr Bob Ballantyne	Dr William Stewart / Dr Antonia Torgersen	Shiona Hogg (AHP Manager Rehab) Donald McLean (AHP Co-ordinator ReACH) Alison Clarke (Breathing Support
Greater Glasgow and Clyde	Queen Elizabeth University Hospital	Queen Elizabeth University Hospital	Dr Maria Farrugia/ Dr Emanuela Molinari	Dr Scott Davidson (QEUH)	Dr Iain Findlay Dr David Murdoch Dr J Byrne	Marina Di Marco	Caroline Carmichael (MG/FSH/ CMT)	Wilma Stewart (NCA)	Dr C Longman Dr V Murday Lesley Snadden	Dr Arup Mallik	Dr Bob Ballantyne	Dr William Stewart	Alison Clarke (Breathing Support)
Greater Glasgow and Clyde	Inverclyde Royal Hospital	Queen Elizabeth University Hospital	Dr Maria Farrugia	Dr Scott Davidson (QEUH)	Dr Iain Findlay	Marina Di Marco	Caroline Carmichael (MG/FSH/ CMT)	Wilma Stewart (NCA)	Dr C Longman Dr V Murday Lesley Snadden	Dr Arup Mallik	Dr Bob Ballantyne	Dr William Stewart	Alison Clarke (Breathing Support)
Lanarkshire	Wishaw General Hospital	Queen Elizabeth University Hospital	Dr Maria Farrugia	Dr K Dagg (Wishaw) Dr Scott Davidson (QEUH)	Dr Iain Findlay	Marina Di Marco		Wilma Stewart (NCA)	Dr C Longman Dr V Murday Lesley Snadden	Dr Arup Mallik	Dr Bob Ballantyne	Dr William Stewart	Alison Clarke (Breathing Support)
Dumfries and Galloway	DG Royal Infirmary/ Galloway Community Hospital	Queen Elizabeth University Hospital	Dr Maria Farrugia	Dr Scott Davidson (QEUH)	Dr Iain Findlay	Marina Di Marco		Wilma Stewart (NCA)	Dr C Longman Dr V Murday Lesley Snadden		Dr Bob Ballantyne	Dr William Stewart	Alison Clarke (Breathing Support)
Highland and Argyll	Lorn and Islands Hospital, Oban	Queen Elizabeth University Hospital	Dr Maria Farrugia	Dr Scott Davidson (QEUH)	Dr Iain Findlay	Marina Di Marco		Wilma Stewart (NCA)	Dr C Longman Dr V Murday Lesley Snadden	Dr Arup Mallik	Dr Bob Ballantyne	Dr William Stewart	Alison Clarke (Breathing Support)

East of Scotland : Neuromuscular Services - PAEDIATRIC

Health Board Area	DGH	Regional Service	Paediatrician / Paediatric Neurologist	Respiratory	Cardiology / Anaesthetics	Physio therapy	Neuro-muscular Specialist Nurse	Neuromus-cular Care Adviser /Support Services	Genetics	Neuro-physiology	DM Ser-vice	Neuro-pathology	AHP / Other
Borders	Borders General Hospital	Royal Hospital for Sick Children, Edinburgh	Dr K McWilliam/ Dr. Alex Baxter /Dr John Stephen	Dr Don Urquhart Dr Kenny McLeod Dr Steve Cunningham Dr Catherine McDougall Dr Krishnan Ani (Fife)	Dr Dzung Dr M Walayat		Yvonne Robb (Neuro-muscular Genetics)	Gill Paton (NCA)	Dr C Longman	Dr. Kenneth McWilliam	Yvonne Robb	Dr Antonia Torgersen (Edinburgh)	Linda McCarthy (Ventilation Nurse Specialist)
Fife	Victoria Hospital / Queen Margaret Hospital	Royal Hospital for Sick Children, Edinburgh	Dr K McWilliam Dr Jamie Cruden / Dr Robert Humphreys		Dr Dzung Dr M Walayat		Yvonne Robb (Neuro-muscular Genetics)	Gill Paton (NCA)	Dr C Longman	Dr. Kenneth McWilliam Dr Eleonora Saturno	Yvonne Robb	Dr Antonia Torgersen (Edinburgh)	Linda McCarthy (Ventilation Nurse Specialist)
Lothian	Royal Hospital for Sick Children, Edinburgh	Royal Hospital for Sick Children, Edinburgh	Dr K McWilliam Dr Alex Baxter		Dr Dzung Dr M Walayat		Yvonne Robb (Neuro-muscular Genetics)	Gill Paton (NCA)	Dr C Longman	Dr. Kenneth McWilliam	Yvonne Robb	Dr Antonia Torgersen (Edinburgh)	Linda McCarthy (Ventilation Nurse Specialist) Hannah Waugh (PT) Lesley Harper (PT)

East of Scotland : Neuromuscular Services - ADULT

Health Board Area	DGH	Regional Service	Neurologist	Respiratory	Cardiology / Anaesthetics	Physio-therapy	Neuro-muscular Specialist Nurse	Neuromus-cular Care Adviser / Support Services	Genetics	Neuro-physiology	DM Ser-vice	Neuro-pathology	AHP / Other
Borders	Borders General Hospital	Western General Hospital	Dr David Simpson	Dr Anthony Bateman Dr S McLellan Dr Kalliroi Kefala	Dr Neary Dr Anthony Bateman (Anaesthetics)	Post vacant – Under discussion	Yvonne Robb (Neuro-muscular Genetics)	Gill Paton (NCA)	Dr C Longman / Yvonne Robb	Post vacant	Yvonne Robb	Dr Antonia Torgersen (Edinburgh)	B. Forrest /Colin McPherson / Claire Hogg / Marian Dingwall / Alina Macleod Dr. Stewart Donald (Astley Ainslie) Dr Lance Sloan (Rehab Team, Fife)
Fife	Victoria Hospital, Kirkcaldy	Western General Hospital	Dr Martin Zeidler		Dr C Mark Francis Dr Anthony Bateman (Anaesthetics)		Yvonne Robb (Neuro-muscular Genetics)	Gill Paton (NCA)	Dr C Longman / Yvonne Robb	Dr Eleonora Saturno	Yvonne Robb	Dr Antonia Torgersen (Edinburgh)	
Lothian	Western General Hospital / St John's Hospital, Livingston	Western General Hospital	Dr Richard Davenport		Dr Martin Denvir Dr A Bateman (Anaesthetics)		Yvonne Robb (Neuro-muscular Genetics)	Gill Paton (NCA)	Dr C Longman / Yvonne Robb	Post vacant	Yvonne Robb	Dr Antonia Torgersen (Edinburgh)	
Lothian	Edinburgh Royal Infirmary	Edinburgh Royal Infirmary		Dr T Mackay	Dr Martin Denvir Dr Anthony Bateman (Anaesthetics)		Yvonne Robb (Neuro-muscular Genetics)	Gill Paton (NCA)	Dr C Longman / Yvonne Robb	Post vacant	Yvonne Robb	Dr Antonia Torgersen (Edinburgh)	

North of Scotland : Neuromuscular Services - PAEDIATRIC

Health Board Area	DGH	Regional Service	Paediatrician / Paediatric Neurologist	Respiratory	Cardiology / Anaesthetics	Physio-therapy	Neuro-muscular Specialist Nurse	Neuromus-cular Care Adviser /Support	Genetics	Neuro-physiology	DM Service	Neuro-pathology	AHP / Other
Grampian (Aberdeen & Moray)	Royal Aber-deen Children's Hospital	Royal Aberdeen Children's Hospital	Dr Martin Kirkpatrick Dr A Jollands Dr Elma Stephen	Dr Richard Brooker Dr Mustafa Osman	Dr Detlev Rogahn Dr Richard Brooker	Jane Tewnion		Support services are being met locally through paediatric teams, Dis-trict Nursing services, AHP's and GP services. A more informed pathway is currently a work in progress.	Dr Alison Ross Dr Helen Gregory Dr Cheryl Longman	Post vacant – awaiting appointment	Dr Martin Kirkpatrick Dr Alice Jollands	Dr Antonia Torgersen (Edinburgh)	
Grampian (Aberdeen & Moray)	Dr. Gray's Hospital, Elgin	Royal Aberdeen Children's Hospital	Dr Martin Kirkpatrick Dr Alice Jollands Dr Elma Stephen	Dr Richard Brooker Dr Mustafa Osman	Dr Detlev Rogahn Dr Richard Brooker	Post vacant			Dr Alison Ross Dr Helen Gregory Dr C Longman	Post vacant – awaiting appointment	Dr Martin Kirkpatrick Dr Alice Jollands	Dr Antonia Torgersen (Edinburgh)	
Highland	Raigmore Hospital Inverness	Royal Aberdeen Children's Hospital	Dr Martin Kirkpatrick Dr Linda MacLellan	Dr Richard Brooker RHSCE Resp Team Yorkhill Resp Team	Dr. Walayat	Julie Burslem			Dr Alison Ross Dr Helen Gregory Dr C Longman	Post vacant – awaiting appointment	Dr Martin Kirkpatrick Dr Linda McLellan	Dr Antonia Torgersen (Edinburgh)	
Orkney	Balfour Hospital (RGH)	Royal Aberdeen Children's Hospital	Dr Alice Jollands		Dr Detlev Rogahn Dr Richard Brooker				Dr Alison Ross Dr Zosia Miedzybrodzka	Post vacant – awaiting appointment		Dr Antonia Torgersen (Edinburgh)	
Shetland	Gilbert Bain Hospital, Lerwick	Royal Aberdeen Children's Hospital	Dr Martin Kirkpatrick Dr Susan Bowie		Dr Detlev Rogahn Dr Richard Brooker				Dr John Dean Dr Cheryl Longman	Post vacant – awaiting appointment		Dr Antonia Torgersen (Edinburgh)	
Tayside (Perth, Angus, Dundee and NE Fife)	Ninewells, Hospital, Armitstead Child Develop-ment Centre, Perth Royal Infirmary	Ninewells, Hospital	Dr Karen Naismith Dr Martin Kirkpatrick Dr Ann Scott (Neuro-disability)	Dr Jonathan McCormick / Edinburgh Home Vent service	Dr K MacLeod Dr Walayat (Cardiology) Dr Grant Rodney (Anaesthetics)	Nicola Whatley (Dundee)	Lynn Willox		Dr Catherine McWilliam Dr Cheryl Longman		Dr Karen Naismith Dr Martin Kirkpatrick Dr Ann Scott	Dr Antonia Torgersen (Edinburgh)	Lynne Rule (OT) Louise Taylor (Dietitian)
Western Isles	Western Isles Hospital	RHC, Queen Elizabeth University Hospital	Dr Iain Horrocks	Dr Phil Davies		Marina Di Marco	Jen Dunne	Wilma Stewart (NCA)	Dr Alison Ross	Dr Patrick Fox (Raigmore	Dr Iain Horrocks (Glasgow)	Dr W Stewart (Glasgow)	

North of Scotland : Neuromuscular Services - ADULT

Health Board Area	DGH	Regional Service	Neurologist / Rehab Consultant	Respiratory	Cardiology / Anaesthetics	Physio therapy	Neuro-muscular Specialist Nurse	Neuromus-cular Care Adviser/ Support	Genetics	Neuro-physiology	DM Service	Neuro-pathology	AHP / Other
Grampian (Aberdeen & Moray)	Aberdeen Royal Infirmary	Aberdeen Royal Infirmary	Dr Gillian Hall Dr A Cozens	Dr Patrick Fitch / Dr Joy Miller	Dr Paul Broadhurst	No specialist service (Referral to local adult PT)		*Susan Stewart (Genetic Specialist Nurse	Dr A Ross Dr Helen Gregory	Post vacant – awaiting appointment	Dr Gillian Hall Dr Helen Gregory	Dr Antonia Torgersen (Edinburgh)	Jacqueline Burnett / Jenny Robinson (PT's) Claire Fitzsimmons (OT), Rhoda Downey (SLT) Susan Stewart (Genetic Specialist Nurse)
Grampian (Aberdeen & Moray)	Dr. Gray's Hospital, Elgin	Aberdeen Royal Infirmary	Dr Gillian Hall (referral only) Dr A Cozens	Dr. Patrick Fitch / Dr Joy Miller	Dr Paul Broadhurst	No specialist service (Referral to local adult PT)		*Susan Stewart (Genetic Specialist Nurse	Dr A Ross Dr Helen Gregory	Post vacant – awaiting appointment	Dr H Gregory	Dr Antonia Torgersen (Edinburgh)	
Highland	Raigmore Hospital Inverness	Raigmore Hospital Inverness	Dr K Taylor Dr Javier Carod-Artal Dr Barbara Chandler (Rehab)	Dr Lorna Murray	Dr Stephen Cross	No specialist service (Referral to local adult PT)		*Susan Stewart (Genetic Specialist Nurse	Dr A Ross Dr Helen Gregory	Dr Patrick Fox (Raigmore)	Dr K Taylor Dr H Gregory	Dr Antonia Torgersen (Edinburgh)	Susan Stewart (Genetic Specialist Nurse)
Orkney	Balfour Hospital (RGH)	Aberdeen Royal Infirmary				No specialist service (Referral to local adult PT)		*Susan Stewart (Genetic Specialist Nurse	Dr Zosia Miedzybrodzka Dr Alison Ross	Post vacant – awaiting appointment		Dr Antonia Torgersen (Edinburgh)	Susan Stewart (Genetic Specialist Nurse)
Shetland	Gilbert Bain Hospital, Lerwick	Aberdeen Royal Infirmary / Perth Royal infirmary	Dr Jim Unsworth (Consultant Physician)			No specialist service (Referral to local adult PT)		*Susan Stewart (Genetic Specialist Nurse	Dr John Dean	Post vacant – awaiting appointment	Dr Jim Unsworth (Consultant Physician) Dr. Helen Gregory	Dr Antonia Torgersen (Edinburgh)	Susan Stewart (Genetic Specialist Nurse)
Tayside (Perth, Angus, Dundee and NE Fife)	Ninewells Hospital	Ninewells, Hospital	Dr Gillian Stewart and neurology team Dr O' Riordan Dr Gentleman Dr Rabia Din	Dr Robin Smith; Home Ventilation Dr A Bateman (Edinburgh)	Dr A. M. Choy	No specialist service (Referral to local adult PT)		*Kirsten Patterson (Genetic Specialist Nurse)	Dr Catherine McWilliam Dr Cheryl Longman		Dr Catherine McWilliam	Dr Antonia Torgersen (Edinburgh)	Betty Forrest (Home ventilation) Perth CPDT Carol Greig (PT) Kirsten Patterson (Genetic Specialist Nurse)
Western Isles	Western Isles Hospital	Queen Elizabeth University Hospital	Dr Maria Farrugia Dr Richard Petty	Dr Scott Davidson (QEUH)		Marina Di Marco		Wilma Stewart (NCA)	Dr Alison Ross	Dr Patrick Fox (Raigmore)		Dr W Stewart (Glasgow)	Alison Clarke (Respiratory Support Nurse)

*Genetic Specialist Nurses will accept referrals for complex cases.