



THE WINSTON CHURCHILL MEMORIAL TRUST OF AUSTRALIA

REPORT BY KATHRYN MUNRO - 2014 CHURCHILL FELLOW

THE BOB AND JUNE PRICKETT CHURCHILL FELLOWSHIP to evaluate services to improve the management and quality of life for Australian children and adolescents with neuromuscular disorders and their families.

THE UK AND USA

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Signed: Kathryn Munro

Dated: 1st October 2015

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1 INTRODUCTION

The subspecialty of paediatric neuromuscular disease has seen rapid growth within Australia in the last 5-10 years. This has been a result of improvements in the clinical care and management worldwide, increased life expectancy for some conditions, new diagnostic techniques and gene discoveries and the inclusion in clinical trials for novel drug treatments. Additionally, the establishment of the Australasian Neuromuscular Network and NHMRC funding of the Centre of Research Excellence in Neuromuscular Disorders has provided the opportunity for clinicians and researchers across Australia to work collaboratively to lead advancements in clinical care and research.

Neuromuscular disorders are a group of around 60 conditions that affect the muscles and/or nervous system causing significant disability and morbidity and are often progressive in nature and life limiting. It is estimated that around 1:1000 people are affected by a neuromuscular condition with muscular dystrophy affecting 1:3500¹. These conditions usually present in childhood, often as a result of mistakes in the genes. The multi system nature and associated respiratory and/or cardiac complications and natural disease trajectory causes significant burden to the individual and their family and has the potential to decrease quality of life and be costly to health services if diagnosis and treatment is delayed and care is disjointed across the health system.

The most common types of neuromuscular disorders diagnosed in childhood are Duchenne Muscular Dystrophy (DMD) and Spinal Muscular Atrophy (SMA)¹. Other disorders seen in specialist neuromuscular clinics include, Congenital Myopathies, Myotonic Dystrophy, Myasthenia Gravis, Charcot-Marie-Tooth (CMT) and Limb Girdle Muscular Dystrophy (LGMD). Individually, most neuromuscular conditions are considered rare, however collectively they are estimated to affect over 4000 people within Queensland and over 30000 people throughout Australia¹. If the effect on family members is considered and the fact that most of these disorders are hereditary then neuromuscular disease impacts a significant number of the Australian population.

Care and management of neuromuscular conditions, provided within a multidisciplinary specialist neuromuscular service is considered best practice. Multidisciplinary teams should consist of Neurology, Respiratory/Sleep and cardiac specialists with support from specialist neuromuscular Nurses and Allied Health Professionals such as Physiotherapists, Dieticians, Occupational Therapists, Genetic Counsellors and Social Workers/Psychologists. Collaborative care with other specialist teams including Paediatricians, Palliative Care, Orthopaedics and General surgeons is also recommended to ensure timely, predictive and coordinated care and management of treatable complications of neuromuscular disease.

Currently newborn screening programs do not include neuromuscular disorders. Therefore early recognition, diagnosis and timely referral to a specialist multidisciplinary

neuromuscular service is imperative to provide families with the opportunity to make informed family planning choices, optimise care and management outcomes and enrol in neuromuscular registries to facilitate inclusion in clinical trials for potential treatments.

As a Clinical Nurse Consultant working in Paediatric Neurology I have been fortunate to be part of the multidisciplinary neuromuscular team in Children's Health Queensland. The opening of the Lady Cilento Children's Hospital in November 2014 facilitated the expansion of the service to be available for all children throughout Queensland. The Churchill Fellowship gave me the opportunity to visit Centres of Excellence who are recognised internationally for their clinical care and management and research into neuromuscular disease.

2 ACKNOWLEDGEMENTS

Firstly I would like to thank the Winston Churchill Memorial Trust of Australia for providing the opportunity to travel. My Churchill Fellowship was not only professionally rewarding but also such an enjoyable experience.

Secondly I would like to personally acknowledge and express my sincere gratitude to Mr Bob Prickett and his late wife Mrs June Prickett for their continued generosity to the Winston Churchill Memorial Trust of Australia and sponsorship of my fellowship.

Thank you also to my project and professional referees Dr Anita Cairns and Dr Geoff Wallace for their support and vision for the potential of this project to improve neuromuscular services and to my Nursing Director Juliana Buys for her initial encouragement to apply and continued support of my role in the neuromuscular service. I also sincerely thank my colleague and 2012 Churchill Fellow, Anita Inwood for her guidance and mentorship throughout my fellowship journey.

I also would like to acknowledge the clinical and research teams who facilitated and hosted my visit at their centres. For the time they took out of their busy schedules to spend time meeting with me, answering questions and for their dedication to teaching and sharing about their roles and services. I would also like to thank the patients and families at each centre who allowed me to attend their clinic appointments and spend time answering my questions to gain insight about their experience at each service and within the greater health system.

Lastly, I would like to acknowledge and thank my family for their understanding and steadfast support and encouragement and for allowing me the time to devote to this fellowship that will hopefully be of benefit to all families affected by neuromuscular disease.

3 EXECUTIVE SUMMARY

3.1 FELLOWSHIP DETAILS

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3.2 PROJECT DESCRIPTION

The scope of this project was to gain clinical experience and knowledge in the management of neuromuscular disorders. The sites I chose are recognised internationally as leaders in the field. Neuromuscular disorders are a group of rare disorders that affect some part of the neuromuscular system and are generally progressive in nature and shorten life span. My aim was to gain knowledge that will help improve the quality of life for patients and their families and ensure best practice in the delivery and coordination of care.

3.3 HIGHLIGHTS

- Meeting with world renowned Neuromuscular specialists and multidisciplinary teams
- Attending the Muscle Interest Group Meeting, London
- Meeting and establishing professional networks with international Neuromuscular advanced practice nurses
- Observing the Nurse Practitioner role within the Neuromuscular Multidisciplinary team
- Visiting world renowned paediatric hospitals

3.4 RECOMMENDATIONS

- Expand Neurology Fellow trainee positions to include clinical experience in neuromuscular services, inclusive of NCS and EMG training.
- Explore the scope and potential for a neuromuscular Nurse Practitioner role in the tertiary paediatric setting.
- Establish neuromuscular outreach clinics in key regional areas to improve access for children and adolescents with advanced neuromuscular disease and develop clinical networks across Queensland.
- Improve the care and management of adolescents and young people with neuromuscular disease by establishing a ‘Lifespan’ model of care for neuromuscular services, working in partnership with Adult health services.
- Establish multidisciplinary neuromuscular clinics in all paediatric Neurology services in each State of Australia which will optimise clinical care and health outcomes and ensure clinical trial readiness and accessibility.

- Establish nurse led clinics within a ‘lifespan’ model of care to provide coordination and continuity of care and support to adolescents and young people transitioning to adult clinics.
- Development of a Transition Program, Care Pathway and Educational resources for Children’s Health Queensland to prepare young people for Transition to adult health care facilities.
- Development of surgical clinical pathways for neuromuscular/complex care patients, e.g. spinal fusion and insertion of a gastrostomy tube.
- Development of patient/family education/resource materials to support the CHQ Neuromuscular service and provide consistency in care across the State.
- Continue with the trial of streaming neuromuscular clinics based on condition/stage of disease to improve patient experience and use of human resources.
- Development of a ‘Family Care Files’ for all ‘New’ patients coming into the service.
- Investigate the benefit to families and clinicians with the concept of a Personal Complex Care Binder for children with neuromuscular or chronic and complex conditions.
- Build networking relationships with international neuromuscular clinics and Advanced Practice Nurses to support the professional development of neuromuscular Specialist Nurses within Australia.
- Continue to engage with Non-Government and Neuromuscular support organisations to advocate for improved tertiary and community services for this vulnerable patient group.

3.5 DISSEMINATION

- Publication of this report on the Winston Churchill Memorial Trust website.
- This report will be shared with the Children’s Health Queensland Hospital Board and Executive teams.
- This report will be shared with Adult Neurology, Respiratory/Sleep and Cardiac services within Queensland Health.
- This report will be shared with Non-Government organisations and patient support groups such as Muscular Dystrophy Queensland, Montrose Access and the Duchenne Foundation.
- A summary of this report will be presented to the LCCH Neuroscience department and the Medical Division and Nursing forums.
- Recommendations from this report will be presented at the Queensland Neuroscience Meeting, 14th November 2015.
- This report will be shared with Australasian Neuromuscular Network (ANN) board.
- An abstract will be submitted to the ANN annual conference 2016.

4 PROGRAMME

Date	Organisation Visited	Address and Contact Person
2 nd – 8 th June 2015	<p>Evelina Children’s Hospital - Neuromuscular Service</p> <p>Lane Fox Respiratory Unit - St Thomas’s Hospital</p>	<p>Westminster Bridge Road, London</p> <p>Dr Elizabeth Wraige Dr Vasantha Gowda Rachel Spahr - Physiotherapist Debbie Clarke – Clinical Nurse Specialist</p> <p>Dr Nicholas Hart – Respiratory Physician Emily Ballard - Neuromuscular Transitional Care Coordinator</p>
9 th June 2015	Muscle Interest Group Meeting at the Institute of Child Health	<p>Guilford St, London</p> <p>Hosted by Dr Adnan Manzur</p>
10 th -18 th June 2015	Dubowitz Neuromuscular Centre Great Ormond Street Hospital	<p>Great Ormond Street, London</p> <p>Professor Francesco Muntoni Dr Adnan Manzur Dr Ros Quinlivan Dr Stephanie Robb Dr Pinki Munot Dr Anna Sarkozy Oxford Congenital Myasthenia Team Ruth Barratt - Nurse Practitioner Karen Roberts-Edema - Clinical Nurse Specialist Naomi Antcliff – Clinical Nurse Specialist Neuromuscular Research</p>
22 nd -26 th June 2015	MRC Centre for Neuromuscular Disease, Newcastle upon Tyne	<p>International Centre for Life, Central Parkway, Newcastle upon Tyne</p> <p>Dr Volker Straub Dr Hanns Lochmuller Dr Chiara Bettolo Dr Michaela Guglieri Dr Teresinha Evangelista Gillian Kenyon - Clinical Trials Coordinator Gail Eglon – Clinical Nurse Specialist Louise Hastings – Clinical Nurse Specialist</p>

		<p>Ruth Wake – Research Nurse Edwina Perkins – Neuromuscular Care Advisor Geraldine Baily – Physiotherapist Dr Anna Mayhew – Physiotherapist Oksana Pogoryelova - Disease Registry</p>
2 nd July 2015	<p>Queen Square Centre for Neuromuscular Diseases at the National Hospital for Neurology and Neurosurgery</p> <p>Transition Clinic</p>	<p>Queen Square, London</p> <p>Dr Ros Quinlivan Suzanne Booth – Clinical Nurse Specialist</p>
9 th July 2015	<p>Queen Square Centre for Neuromuscular Diseases at the National Hospital for Neurology and Neurosurgery</p> <p>Young Adults Neuromuscular Clinic</p>	<p>Queen Square, London</p> <p>Dr Ros Quinlivan Suzanne Booth– Clinical Nurse Specialist</p>
13 th July 2015	<p>Neuromuscular Complex Care Centre (NMCCC) – National Hospital for Neurology and Neurosurgery</p> <p>Multidisciplinary Team Meeting</p>	<p>Queens Square, London</p> <p>Mayumi Rossi – Deputy Sister, NMCCC, MRC Centre for Neuromuscular Disease</p>
20 th -24 th July 2015	<p>Neuromuscular Centre Boston Children’s Hospital</p> <p>Neurology Clinics Ward 9NW Neuroscience ward</p>	<p>300 Longwood Avenue, Boston, Massachusetts</p> <p>Dr Basil Darras Jennifer McGrave – Registered Nurse Regina Laine – Nurse Practitioner Carole Atkinson - Neuroscience Clinical Nurse Specialist - Ward 9NW</p>
26 th -29 th July 2015	<p>Neuromuscular Program Children’s Hospital of Philadelphia</p>	<p>3401 Civic Centre Boulevard, Philadelphia</p> <p>Dr John Brandsema Michelle Bordeleau - Registered Nurse Alan Glanzman - Physiotherapist Beth Kunzelman – Registered Nurse Sarah Stoney - Social Worker Laura McClellan – MDA Healthcare Service Coordinator Nancy Videon – Neuromuscular Clinical Trials</p>

5 REPORT

5.1 ENGLAND

Specialist neuromuscular services in England are delivered by the National Health Service (NHS), one of the world's largest publicly funded health systems servicing a population of over 53 million people². The leading neuromuscular centres are recognised worldwide for their clinical and research excellence. These centres have been integral in advancing the understanding, diagnosis, and care and management of neuromuscular disorders which has led to an increased life expectancy for many conditions diagnosed in childhood.

However, in 2007 it was reported that access to clinical care and life expectancy varied dramatically in different regions of the United Kingdom, particularly for people living in poorer communities and for those that did not have access to a specialist neuromuscular centre^{3,4}. Additionally the audit of unplanned admissions to hospitals for patients with a neuromuscular condition was reported to cost the NHS in excess of £10 000 000 during 2009-2010⁵. Significant reforms since then has seen the commissioning of specialist services all throughout England, with the aim of improving equality of care, access to services and patient outcomes.

Established in 1959 the Muscular Dystrophy UK is a charity supporting over 70 000 people in the United Kingdom affected by a neuromuscular condition⁶. As an organisation they have been instrumental in advocating for improved services across England and continue to work with patients, individual centres, commissioning groups and the NHS to close the gaps in service delivery, clinical care and treatment^{7,8}. Additionally within the United Kingdom there are a number of disease specific charitable patient support organisations.

Whilst in England on my fellowship the media was filled with patients and advocacy groups campaigning to the NHS for access and funding for the medication, Ataluren (Translarna™). This medication is a new treatment for the 10-15% of boys who have Duchenne Muscular Dystrophy caused by a nonsense mutation and already has conditional approval and funding in many European countries⁹. Patients and clinicians are eagerly awaiting the NHS decision and National Institute of Health and Care Excellence (NICE) evaluation due in early 2016¹⁰. This medication is currently in clinical trials at two Australian sites and received orphan drug status in August 2015 which is the first step in gaining regulatory approval in Australia¹¹.

On my fellowship I visited five of the neuromuscular services in England, three of which are recognised as Centres of Excellence. As expected, the centres I observed in London service a much larger population compared to Queensland and as such have larger patient numbers. The population serviced by the Newcastle Muscle centre is closest in size to Queensland, however offers a lifespan service so again the total patient numbers are larger as adults are included in their cohort.

5.1.1 Evelina Children's Hospital – London



Evelina Children's Hospital is a tertiary paediatric hospital and forms part of the Guys and St Thomas's National Health Service (NHS) Foundation Trust and is co-located on site with the St Thomas's hospital in London. It is one of the two specialist paediatric hospitals in London and has a dedicated neuromuscular service within the department of Neurosciences, providing care to children from the South East Thames region of England.

During my time at Evelina I met with Dr Elizabeth Wraige and Dr Vasantha Gowda who are two of the Neurology consultants working in the neuromuscular service, Rachel Spahr, a physiotherapist and Debbie Clarke the Clinical Nurse Specialist and attended the weekly neuromuscular clinic. I also had the opportunity to observe aspects of the general Neurology service, attending the weekly handover and inpatient multidisciplinary team meetings and tour the inpatient Neurology ward which was beneficial given the recent opening of the Lady Cilento Children's Hospital (LCCH).

The service at Evelina hospital is the closest in size to the Children's Health Queensland neuromuscular service, 450:300 respectively. This team consists of 2.5 FTE Neurology Consultants however they are also required to support the general Neurology service and 1 FTE neuromuscular trainee Fellow and a Clinical Nurse Specialist. Clinics are attended by two Physiotherapists who also cover general Neurology inpatient and outpatient workloads.

Neuromuscular clinics are held each Wednesday, all day and fortnightly on Tuesday mornings with Neurology and Physiotherapy providing a joint appointment for patients. Outpatient clinics at Evelina are held in a centralised outpatient department on the ground floor. Other specialist reviews such as Cardiology or Respiratory require a separate appointment. I observed the Wednesday clinic which saw seven patients of varying ages and conditions and was attended by two Neurologists and Physiotherapists. The combined appointment ran smoothly and provided a comprehensive functional assessment during clinic.

The nursing model at the Evelina neuromuscular service differs from all the other services I visited in England and is covered by a 0.6 FTE Clinical Nurse Specialist position, which is shared across the entire Neurology inpatient and outpatient setting and responsible for the discharge planning and complex care management for all General Neurology patients. There was however plans for the appointment of another full time General Neurology Clinical Nurse Specialist position to manage the nursing workload and facilitate greater support for and inclusion in the neuromuscular service. The nursing model however has a very strong outreach focus for neuromuscular patients, with the Clinical Nurse Specialist attending outreach clinics, offering care for families at the time of a new diagnosis, inclusive of home

visits to provide psychosocial support, telephone support with the commencement of steroid therapy for boys with Duchenne Muscular Dystrophy (DMD) and provision of education to schools and local community providers.

Outreach neuromuscular clinics are strongly embedded in the Evelina service. Recognising the physical challenges and barriers for neuromuscular patients and families with travelling into central London, regular outreach clinics are held in local South East Thames communities around 1 hours' drive from London. These are attended by two Neurologists or a Neurologist and Fellow, supported by the Clinical Nurse Specialist and the Physiotherapists who complete the functional assessments and liaises with local teams. These clinics are also attended by the Transitional Care Coordinator from the Lane Fox Respiratory Unit who does respiratory screening assessments.

Whilst neuromuscular Consultants in England guide treatment such as the commencement of steroid therapy, the NHS mandates that the patient's General Practitioner is responsible for prescribing and managing all medications and as the primary care provider, is seen as having an important role in the comprehensive care and management for children with complex conditions. This is very different to the model I have seen in Queensland, where Consultants in the tertiary setting often provide this service. Additionally the choice and availability of Prednisolone or Deflazacort corticosteroid treatment for boys with DMD was significantly different. In Australia, Deflazacort is a second line therapy and is much more expensive than Prednisolone. Within Queensland, Deflazacort is only available through hospital pharmacies and the cost is largely covered by the Hospital and Health Service. In Australia access to Deflazacort is on a case by case basis and requires Independent Patient Authority and Special Access Scheme approvals. Within England, Deflazacort was readily available, and able to be prescribed by GP's with families making the choice of corticosteroid and covering any extra cost.

For adolescents in the Evelina service, transition to adult clinics occurs at around 16-18 years. Options of clinics to transition to include the Lane Fox Respiratory Unit or Neuromuscular Centres at King's College Hospital or the National Hospital for Neurology and Neurosurgery. The co-location of the paediatric service at Evelina and the Lane Fox Respiratory Unit at St Thomas's facilitates a close partnership⁵. The Transitional Care Coordinator is able to work across both services supporting a 'life span' model to support transition between these two tertiary neuromuscular services. My fellowship included visiting both services to gain an understanding of the transition pathway and how paediatric and adult clinics can work collaboratively to provide a life span model of care.

5.1.2 Lane Fox Respiratory Unit (LFU)

Established in 1989 the Lane Fox Unit is a 14 bed purpose built inpatient unit at St Thomas's Hospital, which provides care and management to over 1200 adults with chronic respiratory failure¹². The inpatient unit at Lane Fox is well set up to support the needs of people with

physical disabilities with inbuilt ceiling hoists at each bed. The unit also provides a 24hr medical, nursing and technical back up service and has a direct admission service for patients requiring inpatient care.

The Lane Fox Respiratory unit has a specialist neuromuscular service which provides both inpatient and outpatient care to adults with neuromuscular disorders of which 130 are young adults with Duchenne Muscular Dystrophy aged between 15-35 years. The neuromuscular service at Lane Fox was originally established in response to need, and the increasing trend of unplanned emergency admissions for neuromuscular patients following presentation when acutely unwell and in crisis. This resulted in an increased length of stay and unnecessary costs for the NHS and an audit in the London region during 2009-2010 identified that around 40% of admissions may have been preventable^{5,7}. By 2012, the Lane Fox Respiratory Unit had demonstrated that coordinated respiratory care for people with neuromuscular disease, particularly around the vulnerable time of transition, had reduced the incidence of unplanned admissions, length of stay and need for invasive ventilation, resulting in significant cost savings for the NHS¹³. When first established the Lane Fox Unit provided respiratory care to children with neuromuscular disorders as young as 10, however these are now managed by the paediatric Respiratory team at Evelina.

During my visit to the Lane Fox Unit I met with Emily Ballard the Neuromuscular Transitional Care Coordinator and attended a neuromuscular clinic with Dr Nicholas Hart, the Clinical Director. Neuromuscular clinics are held 2-3 times each month and are attended by Respiratory, Cardiology, Neurology and Physiotherapy with support of the Respiratory nurse, in a 'one stop shop', coordinated multidisciplinary model. Recent changes included streaming patients into the cardiac or non-cardiac clinics, which was reported to have improved the patient's experience. Patients also have access to coordinated care with the orthopaedic, gastroenterology and urology teams as required.

Outreach support is also a large part of this service and whilst there are no regional outreach clinics, patients are supported with home visits to set up invasive and non-invasive ventilation, provide education and improve compliance. The service also provides a Home Care Plan for Chest Crisis Management to all neuromuscular patients to guide them if they become unwell with the Lane Fox Unit providing 24 hour Nurse Advisor support afterhours and on weekends. For neuromuscular patients at an advanced stage of disease they are seen in a home visit by nursing and medical staff and a technician from the home ventilation service once per year. The Lane Fox Unit also supports a satellite centre at East Surrey Hospital in a hub and spoke model to improve patient outcomes in regional hospitals outside London.

As mentioned earlier, transition to adult services at the Lane Fox respiratory unit is supported by a dedicated Transition Coordinator who has a background as a respiratory Physiotherapist. Preparation for adolescents transitioning from the paediatric clinics at Evelina to the Lane Fox Unit starts at around 14-15 years of age with the coordinator

attending the Evelina neuromuscular outreach clinics to meet with families, complete respiratory screening and link with families who have a low engagement in services. Additionally, there are transition clinics with the Evelina respiratory service for neuromuscular patients already using non-invasive ventilation. The role of the Transition Coordinator provides continuity of care across a life span model for neuromuscular patients requiring respiratory care and/or non- invasive ventilation.

5.1.3 Muscle Interest Group Meeting

My visit to London coincided with the biannual Muscle Interest Group meeting, hosted by the team at the Dubowitz Neuromuscular Centre and held at the Institute of Child Health in London. Alternate meetings are held as part of the British Myology Society annual meeting and the other rotates around a number of the neuromuscular centres in England¹⁴. The meeting brought together around 40 neuromuscular specialists, scientists and researchers from all over the England. The meetings are open to paediatric and adult neurologists, paediatricians, clinical geneticists and pathologists¹⁴.

Around 10 clinical cases were presented and the format had both an educational and consultative focus where challenging diagnostic cases were discussed amongst peers. It was very comprehensive and included clinical information, muscle MRI imaging and muscle biopsy images. The day also included updates on current gene testing in the United Kingdom and an overview of the current clinical trials for neuromuscular disorders. The opportunity to attend this meeting was so beneficial to enhancing my knowledge and understanding of neuromuscular disorders from initial assessment through to investigations, diagnosis and potential treatments and demonstrates the strong clinical, diagnostic and care networks across England for people affected by neuromuscular disorders.

5.1.4 Dubowitz Neuromuscular Centre at Great Ormond Street Hospital (GOSH)



The Dubowitz Neuromuscular Centre has been based at Great Ormond Street Hospital, London since 2007 following its move from the Hammersmith Hospital. It is a leading clinical and research centre specialising in paediatric neuromuscular disorders, led by Professor Francesco Muntoni and is recognised as a Centre of Excellence both nationally and internationally. The Dubowitz Neuromuscular Centre is part of Great Ormond Street Hospital NHS Foundation Trust and the UCL Institute of Child Health and a member of the MRC Translational Research Centre. It is also the recognised National Commissioning Group for the specialist diagnosis and assessment of congenital muscular dystrophy and congenital myopathy by the NHS¹⁵.

This service is the largest paediatric centre I visited, with around 2000 patients on their database. On average around 1000 patients are seen in clinics each year. Comparatively, this

service is over three times the size of the service in Queensland. It has a multidisciplinary model supported by a team of five neuromuscular Consultants, two neuromuscular Trainees, two neuromuscular Advanced Practice Nurses, a Family Therapist, Social Work, Physiotherapy, Speech and Language Therapy and a Dietician.

During my time at the Dubowitz Neuromuscular Centre I had the opportunity to work closely with the neuromuscular Nurse Specialists, Ruth Barratt and Karen Roberts-Edema, attend clinics with Professor Francesco Muntoni, Dr Adnan Manzur, Dr Ros Quinlivan and Dr Stephanie Robb, meet with the research teams and lead Research Nurse and attend the various clinical meetings that form part of this comprehensive service. Deservedly recognised as a Centre of Excellence the neuromuscular service is extensive and has number of disease specific and general clinics each week. There are disease specific clinics for Spinal Muscular Atrophy (SMA), Congenital Myopathy, Congenital Myasthenia, Charcot-Marie Tooth (CMT), Becker Muscular Dystrophy and Duchenne Muscular Dystrophy (DMD). The general clinics include 'New' appointments, Results, Complex multidisciplinary patients, Transition and neuromuscular Follow Up appointments. The service also works collaboratively with other departments to offer joint specialist clinics with Spinal, Orthotics and Respiratory. For patients with DMD the clinic appointments are also streamed based on age and disease progression.

The neuromuscular clinics are held in the Royal London Hospital for Integrated Medicine which is next door to the GOSH. The outpatient area is staffed with clinic assistants who are managed by a senior team leader with clinical support provided by the Consultants, Specialist nursing teams and Allied Health Therapists¹⁶. There is a phlebotomy and x-ray service within the same building.

I attended the 'New' appointment, Results, SMA, DMD > 8 years, Congenital Myasthenia and Complex Multidisciplinary clinics during my visit. Each clinic has a multidisciplinary model of care and was attended by the neuromuscular Consultant, neuromuscular Trainee and both Nurse Specialists. Physiotherapy reviews were coordinated on the same day prior to the specialist appointment. This service also has a family therapist that supports the DMD and SMA patients. Dietetic and Speech and Language therapy review are provided on a case by case basis.

Care and management in this service is comprehensive with the members of the multidisciplinary team meeting regularly to discuss case management throughout the week. I experienced the weekly psychosocial meetings with the Clinical Nurse Specialists and Family Therapist, Kate Waters; the weekly multidisciplinary team meetings attended by the entire neuromuscular clinical and research team; and the weekly muscle biopsy meeting prior to the results clinic. Families also have access to an impressive suite of resources to assist with understanding and caring for their child. These have been developed by the Dubowitz team and are readily available on the GOSH website. There is also a family file specifically provided to patients with DMD, which consolidates all the care resources for this

condition. I would like to introduce a similar concept into the CHQ neuromuscular service to consolidate the resources we provide to families.

The neuromuscular team has two Advanced Practice nurses, Ruth Barratt and Karen Roberts-Edema who are integral to the service. Whilst both have the title of Clinical Nurse Specialist, Ruth has a Masters level Nurse Practitioner qualification which allows for an advanced scope of practice. Each attends all the neuromuscular clinics and multidisciplinary meetings and work with the multidisciplinary team to provide care coordination, support for patients and families and case management across the inpatient and outpatient setting. For patients requiring surgical intervention the neuromuscular Nurses work collaboratively with the surgical teams and have developed clinical pathways for neuromuscular patients requiring Orthopaedic, Gastrostomy and Spinal surgery interventions. Aspects of their roles were divided with Karen having responsibility for clinic planning with the administrative team, preparing for the psychosocial meetings and being the neuromuscular nursing lead for the orthopaedic care pathway. Ruth has an extended scope of practice and runs nurse led clinics for the initiation and follow up post commencement of steroid treatment for boys with DMD, recruits and consents for the DMD natural history research project and is the neuromuscular nursing lead for the gastrostomy and spinal surgery clinical pathways. It was certainly a highlight to observe the extended scope of nursing practice and nurse led clinics within the neuromuscular service.

Whilst visiting Great Ormond Street Hospital (GOSH) I was able to attend the multidisciplinary spinal surgery meeting. Held monthly, this meeting brings together Consultants and Nurse Specialists from Orthopaedics/Spinal, Neurosurgery, Anaesthetics, and Cardiology, Respiratory, ICU, Paediatrics and Neuromuscular teams. At GOSH it is mandatory for all complex care patients to have a nominated paediatrician involved in their pre and post-operative care.

At the meeting, each complex care patient assessed as needing surgery is presented by the Spinal team complete with radiology images and any other pre-operative work up assessments. Each team is required to indicate risk/benefit for the patient based on a 'green, amber, red' traffic light system. A 'red' light from any one of the Clinicians immediately means further assessment and consultation is required before surgical clearance. Whilst not limited to complex neuromuscular patients, this meeting demonstrates a strong organisational commitment to patient safety and collaboration between the medical and surgical teams.

For neuromuscular patients requiring spinal surgery there is a Spinal Clinical Pathway coordinated by the Spinal Clinical Nurse Specialist, which guides care from preoperative assessment to discharge. The pre-operative workup is coordinated over one day with consultation from the Paediatrician, Physiotherapist, Occupational Therapist, Anaesthetist and Respiratory Physician. Investigations of the patient include height, weight, blood tests, echocardiogram, electrocardiogram (ECG), lung function, clinical photography and x-rays.

For neuromuscular patients there is the addition of a combined Neuromuscular/Spinal clinic attended by neuromuscular and Spinal Orthopaedic Consultants, Clinical Nurse Specialists and the Spinal Team Psychologist. The aim of this joint clinic is to assess the severity and progression of the spinal deformity and discuss optimal management and risks associated with the underlying neuromuscular condition.

Based on the success of the Spinal Clinical Pathway the Clinical Nurse Specialists at the Dubowitz Centre have adapted this model and now have developed pathways for neuromuscular patients requiring surgery for a gastrostomy feeding device and other orthopaedic procedures. Certainly there is definite scope for the development of similar Clinical Pathways and multidisciplinary pre-surgical models within the neuromuscular service in Children's Health Queensland with the added benefit that this model could be applied to many other services managing complex patient groups.

Great Ormond Street Hospital demonstrates an organisational commitment to supporting and preparing adolescents for transition to adult services. It is anticipated that adolescents will move to adult services between 16-18 years with the Clinical Nurse Specialist or Consultant having the 'Transitional Coordinator' role. The organisation has an integrated clinical pathway for transition to enable clinicians to work with adolescents and families to prepare them for adult services. There is also extensive resources for adolescents and their families which are available on the GOSH website. To support the transition of adolescents with neuromuscular disorders the Dubowitz Centre works in partnership with the Queen Square Centre for Neuromuscular Diseases at the National Hospital for Neurology and Neurosurgery to provide a seamless move to adult clinics in a 'life span' model.

The Dubowitz Centre is recognised internationally for its excellence in neuromuscular research and is involved in international clinical trials, natural history studies, basic research to understand the cause of these disorders and the identification of new therapies. The research program is impressive with over 25 clinical research projects. Around 200 patients at the Dubowitz Centre are enrolled in the research studies and/or clinical trials. During my visit, I was introduced to Naomi Antcliff, the Clinical Nurse Specialist for neuromuscular research and had the opportunity to tour the Somers Clinical Research Facility (CRF) at GOSH. This facility is purpose built to support clinical trials with 8 clinical rooms and specialist preparation rooms and freezers for pathology specimens. The neuromuscular clinical trials team are the largest user of this facility.

The neuromuscular clinical trials are supported by a team of four full time Nurses led by the neuromuscular Research Clinical Nurse Specialist, Naomi Antcliff. Each nurse is allocated to a number of clinical trials, and is responsible for the coordination and provision of nursing care during each trial visit. Patients may be admitted to the CRF, inpatient unit or the ICU depending on the clinical trial protocol. Each clinical trial has a lead Investigator and is allocated a lead Coordinator, lead Nurse, lead Physiotherapist and Data Manager. Research

projects are well coordinated and integrated into clinical care with research teams attending the weekly multidisciplinary team meeting and some of the disease specific clinics.

My last day at Great Ormond Street Hospital coincided with the Congenital Myasthenic Syndromes (CMS) clinic. Held on Thursdays, each month this joint clinic brings together Dr Stephanie Robb from the Dubowitz team and the Oxford clinical and research team. The Oxford service is a nationally commissioned specialised service for the diagnosis and management of children and adults with CMS and autoimmune Myasthenia Gravis. Attending this clinic provided me exposure to a larger number of patients than I would see in Queensland and the teaching provided by the Oxford team was such a beneficial educational opportunity and certainly enhanced my understanding on the diagnosis and management of these conditions.

5.1.5 The MRC Neuromuscular Centre – Newcastle upon Tyne



The Newcastle Muscle Centre based at the International Centre for Life in Newcastle is part of the Northern Genetics Service within the Newcastle upon Tyne Hospitals NHS Foundation Trust. Recognised as a national and international Centre of Excellence in clinical care and research it is one of three centres in England supported by the Muscular Dystrophy Campaign for the diagnosis and management of inherited muscle disease and the Nationally Commissioned Limb Girdle Muscular Dystrophy (LGMD) advisory and diagnostic service for all of England¹⁷. It also forms part of the MRC Centre for Neuromuscular Diseases with the Institute of Neurology.

The centre has a life span model of care servicing a population of around 3 million people in North England and sees around 1800 paediatric and adult patients each year with inherited muscle disease¹⁷. This centre offers an extensive service of paediatric and adult neuromuscular clinics as well as outreach and disease specific clinics at a number of sites in Newcastle upon Tyne and North England. As expected, of a life span service it has a large multidisciplinary team. Professor's Kate Bushby, Volker Straub and Hanns Lochmuller lead the service and have joint appointments between the NHS and Newcastle University¹⁷. The multidisciplinary team consists of another four neuromuscular consultants with a background in Neurology or Genetics, two neuromuscular trainee Fellows, two Neuromuscular Nurse Specialists, four Physiotherapists and a Regional Care Advisor. There is also additional support from another five Neurologists who support specific disease and outreach clinics.

During my visit at the Newcastle Muscle Centre I had the opportunity to meet with the disease registries team, observe the research, clinical and clinical trials meetings, attend the "New" patient and Carlisle outreach clinics and visit the Newcastle Clinical Research Facility (CRF) to observe clinical trial visits and meet the CRF nursing staff. Whilst here I met Dr

Chiara Bettolo and Dr Hanns Lochmuller, Louise Hastings and Gail Eglon the Clinical Nurse Specialists, Edwina Perkins, the Neuromuscular Care Advisor, Gillian Kenyon and Ruth Wake from the Clinical Trials Team, Oksana Pogoryelova from the Registries team, attended clinics with Dr Volker Straub and Dr Teresinha Evangelista and Geraldine Bailey and Dr Anna Mayhew the physiotherapists and observed the Clinical Research Facility and clinical trial visits with Dr Michaela Guglieri.

Clinical meetings are held weekly on Tuesday mornings and open to all the clinic team. Discussion focuses around current and upcoming clinical activity, reviewing clinical cases, triaging urgent referrals and addressing any patient queries or issues. Responsibility for preparing for this meeting is rotated through four of the Clinical Associate doctors.

On the same day I attended the weekly 'New' patient clinic which was held at the Institute for Genetic Medicine. At this clinic 5 patients were seen, varying in age from infancy to adulthood. It is routine for all 'New' patients to be seen in this clinic however for subsequent follow up appointments, the clinic may vary depending on where the patient lives or their diagnosis. This clinic had a multidisciplinary model of care and was attended by two of the neuromuscular Consultant, two Physiotherapists, the Clinical Nurse Specialist and the Research nurse.

Outreach clinics are strongly embedded in the service with over thirty paediatric and adult clinics held at various NHS, hospice or community sites outside of Newcastle upon Tyne each year. The week I visited coincided with the Carlisle outreach clinic, a town 95 kilometres northwest of Newcastle upon Tyne. This clinic is held four times/ year at the Eden Valley Hospice and has a multidisciplinary model of care, attended by two Neuromuscular specialists, a Neuromuscular Fellow, a Clinical Nurse Specialist, two Neuromuscular Physiotherapists, a Neuromuscular Research nurse and the Regional Care Advisor. Thirteen, review patients were seen at the outreach clinic and ranged in age from 17-73 years and had varying neuromuscular conditions such as Myotonic Dystrophy, Facioscapulohumeral Muscular Dystrophy (FSHD), Limb Girdle Muscular Dystrophy (LGMD), Duchenne Muscular Dystrophy (DMD) and Myopathies. All patients were seen by each member of the multidisciplinary team. Respiratory assessments were completed by the Physiotherapists and the Research nurse collected any specimens that were needed. The Eden Valley Hospice provided a relaxed, non-clinical setting and was an accessible and more affordable option for patients who had already travelled 1-2 hours to attend the clinic. This clinic was the only outreach model I had the opportunity to observe during my fellowship but certainly provides an excellent multidisciplinary framework to base future outreach services on.

The Nursing model supporting the muscle service consists of three nurses. There are 1.5 FTE Clinical Nurse Specialists, Gail Eglon and Louise Hastings and a full time Research nurse, Ruth Wake. All are employed by the Newcastle upon Tyne NHS Foundation Trust and as the muscle service is part of the Northern Genetics Service the Clinical Nurse Specialist role has

a strong foundation in genetic counselling. The nursing role is highly valued in this service and it is considered equally important that patients have a nursing consultation at the multidisciplinary clinics. Each nurse has phlebotomy skills which facilitates the collection of pathology specimens at clinics. The role of the Research nurse is diverse and includes supporting outpatient clinics and the various research projects.

As a life span service, all patients move smoothly within the service and ongoing follow up is dependent upon the patient's geographical location and/or disease. With such a large number of outreach clinics the Newcastle Muscle Centre works closely with other hospitals in a hub and spoke model. Whilst adolescents may transfer from paediatric to adult clinicians at their local health services, key members of the Newcastle muscle multidisciplinary team remain constant such as the Clinical Nurse Specialists, Research Nurse, Physiotherapists and the Specialist Neuromuscular Care Advisor. The service also utilises the Muscular Dystrophy UK Guide to Transition to support their adolescent patients transitioning to adulthood.

The Newcastle Centre was the only service I visited that had a Specialist Care Advisor. This is a full time position funded by the Newcastle upon Tyne NHS Foundation Trust. Edwina Perkin's role includes supporting families with the wider issues faced following diagnosis such as housing, benefits, education and employment. Patients are seen in the multidisciplinary clinics however the role also has capacity to be mobile seeing patients in the community setting such as the home, workplace, college, care or respite facility and inpatient setting. Other initiatives, to improve the understanding of neuromuscular conditions included information and study days, family weekends and supporting professional networks in the community. Edwina has also been instrumental in working with Muscular Dystrophy UK to develop the self-management pack for people with adult-onset neuromuscular conditions. I was fortunate to be provided with a copy of this excellent resource which I will share with the Neurologists in the adult setting and patient advocacy groups.

The Newcastle Muscle centre is very active in research and leads the 'Treat NMD' patient registries and is involved with a number of clinical trials. The research program has three neuromuscular Consultants who are the research leads as well as a scientific research lead. The clinical trial activity subgroup is led by Dr Guglieri and is supported by three Clinical Trial Coordinators, the Research Nurse and Physiotherapists. During the week I visited I was able to observe the different aspects, through attending the research meeting, clinical trials meeting and visiting the Clinical Research Facility (CRF) to observe clinical trial visits and the model of care.

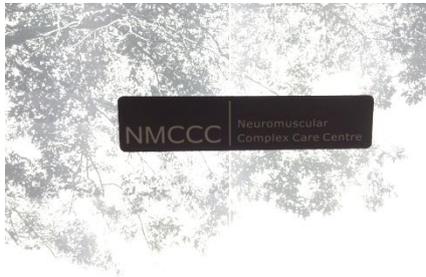
Opened in 2005, the CRF is a purpose built facility located in the Leazes Wing at the Royal Victoria Infirmary and is a partnership between the Newcastle upon Tyne Hospital Trust and the Newcastle University. This dedicated research facility has 10 inpatient beds, consulting rooms, laboratory facilities and a separate paediatric wing. The facility has its own team of

research nurses who have training in research methodology. The CRF research nurses are allocated to specific clinical trials and are responsible for the provision of nursing care, clinical assessments and specimen collections during clinical trial visits.

Whilst the CRF can offer clinical trial coordination, the Newcastle Muscle Centre has a team of three Clinical Trial Coordinators. I was invited to attend the clinical trial meeting which bring together the Trial Coordinators, Neuromuscular Consultants, Physiotherapists and Research Nurse to coordinate and allocate workload for the clinical trial visits. I also observed the fortnightly Research Clinical meeting which brings together the entire clinical and research team to discuss current and potential clinical trials.

5.1.6 Queen Square Centre for Neuromuscular Diseases

Whilst my area of expertise is paediatric nursing, my fellowship included visiting the Queen



Square Centre for Neuromuscular Diseases at the National Hospital for Neurology and Neurosurgery (NHNN) to gain insight into the transition pathway between paediatric services at GOSH and integration into the young adult clinics at Queen Square. The NHNN is part of University College London Hospitals NHS Foundation Trust and is the largest dedicated Neurology

and Neurosurgical Hospital in the UK ¹⁸.

The centre at Queen Square is part of the Medical Research Council (MRC) Centre for Neuromuscular Disease and one of the translational research centres in the United Kingdom. It is a partnership between University Colleges London (UCL) institutes of Neurology and Child Health and the University of Newcastle upon Tyne. The centre offers a number of specialist neuromuscular services for Muscle and Peripheral Nerve Conditions, Myasthenia Gravis and Motor Neurone Disease and sees around 3500 patients each year. Each specialist neuromuscular service is supported by at least one dedicated Clinical Nurse Specialist (CNS)¹⁹.

The transition service has three Neurology consultant leads, Dr Ros Quinlivan for muscle disorders, Dr Shamima Rahman for mitochondrial disorders and Professor Mary Reilly for genetic neuropathies¹⁹. Dr Ros Quinlivan has a joint appointment at the Dubowitz Neuromuscular Centre at GOSH and the Queen Square Centre at the NHNN. The transition service is supported by the Clinical Nurse Specialists at GOSH and the Clinical Nurse Specialist at the Queen Square Centre for Neuromuscular Diseases.

The Transition clinic is a collaboration between GOSH and NHNN and has a lifespan model of care, providing a seamless move from paediatric clinics to the young adult clinics. The centres also have the advantage of being co-located. Held monthly in the GOSH outpatient department, the Transition clinic is for adolescents aged 16-18 years. GOSH has an

integrated transition pathway commencing at age 12 to assist paediatric teams in preparing adolescents and their families for the eventual transfer to adult services.

The Transition clinic is a combined appointment attended by the paediatric neuromuscular team at GOSH and Dr Ros Quinlivan and Suzanne Booth, the Neuromuscular Clinical Nurse Specialist from Queen Square. The consultation is led by the paediatric neuromuscular Consultant who provides a verbal summary and introduces the patient and family to the adult team. The joint appointment provides the opportunity to address any clinical issues prior to discharge from paediatric services, facilitates meeting the adult team, establishes key contacts in the adult service and introduces the adult clinic model. The first clinic at NHNN takes place around 6 months later and is dependent on the young adult making an appointment after receiving a formal invitation to transfer to the adult clinic. The transition clinic was very relaxed and from what I observed all the adolescents and families felt prepared and comfortable with moving to the young adult clinic. The NHNN team expressed that despite this clinic running for a number of years they felt that the model was still evolving and the Suzanne referred me to the Southampton 'Ready, Steady, Go' transition model for additional information around transition as it was a highly regarded transition model within the NHS.

The following week, I attended the Young Adult muscle clinic at Queen Square. This clinic is held monthly and attended by one of the neuromuscular Consultants and the Clinical Nurse Specialist. Patients are generally seen twice a year with coordination of a planned inpatient specialist multidisciplinary review at the Neuromuscular Complex Care Centre (NMCCC). During my visit I was offered the opportunity to see this centre and attend the weekly multidisciplinary team meeting.

Recently opened in September 2014, the NMCCC at the NHNN is the first dedicated neuromuscular inpatient centre in the United Kingdom. In response to the NHS audit identifying fragmented clinical care and avoidable unplanned acute hospital admissions for people with neuromuscular disorders the aim of this service is to improve access to health services and outcomes for people with neuromuscular and inherited metabolic disorders who need highly complex nursing and medical care²⁰.

The centre has six inpatient beds, complete with respiratory and medical monitoring capacity and ceiling hoists and the ability to complete sleep studies. The unit has a 'one stop shop' multidisciplinary model of care and provides a planned, comprehensive, coordinated inpatient assessment.

The nursing team is led by Mayumi Rossi, the Ward Manager, supported by a Neuromuscular Complex Care Clinical Nurse Specialist, a Deputy Ward Sister, 4FTE Senior Staff Nurses, 9FTE Staff Nurses, 1FTE Senior Nurse Assistant and 2FTE Nurse Assistants. The nursing ratio is 1 nurse for 2 patients. The Staff nurses have a background in cardiac or respiratory nursing and have additional training and skills in non-invasive ventilation (NIV),

cough assist, spirometry, respiratory and blood gas monitoring, phlebotomy and intravenous cannulation. Continuity of care with nursing staff is seen as a key factor in providing quality care to patients using this service due to the complex long term needs for those affected by neuromuscular conditions. The unit prides itself on developing strong relationships to foster the patient's compliance and engagement in care.

The multidisciplinary team includes twelve Consultants (Neuromuscular/Neurology/Metabolic), two Cardiologists, a Respiratory Consultant, two Neuro Anaesthetists, a Gastroenterologist, a Respiratory Physiologist, a Physiotherapist, an Occupational Therapist, a Dietician, a Speech and Language Therapist, a Social Worker, a Neuro Psychologist, an admission coordinator, a complex discharge support nurse and five Clinical Nurse Specialists. Multidisciplinary meetings are held weekly on Mondays and attended by all members to plan and coordinate care.

The unit is also able to provide a 24 hour advice line to patients, carers and local community teams. The staff utilise a telephone advice form to guide the call. Local hospitals are also able to contact the unit for support and patient information. Additional initiatives for this relatively new service will be the commencement of seminars in 2016 about the Neuromuscular Complex Care Centre and transition.

On my fellowship journey I did not come across any other unit of this type, and it is most likely that this is not only a first for the NHS but worldwide. I look forward to following the ongoing success of this service as it has real potential as an innovative model of care and could be introduced into adult facilities throughout Australia. It also has a potential application in the paediatric setting as it may increase consumer engagement for those patients who disengage in care due to the physical challenges of accessing multidisciplinary outpatient care at a tertiary setting.

5.2 UNITED STATES OF AMERICA

Unlike the United Kingdom, specialist neuromuscular services in the USA are delivered by individual health care organisations and there does not appear to be a national approach to service delivery by the government. The USA differs from both Australia and the UK in that it does not provide a universal public health care system and is reported to be the most expensive in the world with approximately 18% of GDP spent on health care^{21,22}. Access to health care in the USA is through a complex mix of public and private funding. Significant reforms in the last five years have seen improved equality and access to health care. In the two states I visited whilst on my fellowship, patients had access to health services through Medicaid, a joint federal-state program, private insurance or a combination of both. Despite the complexities of the US health care system, the country is recognised for its innovation in health care. The two hospitals I visited in the US operate as charitable, non-profit organisations and as teaching hospitals have strong academic links.

Founded in 1960, the Muscular Dystrophy Association (MDA) is national charitable organisation dedicated to supporting people affected by neuromuscular disease and funds research to find treatments and a cure²². The organisation has a coordinated management structure with local branches in each state and upholds a network of 200 specialist neuromuscular clinics throughout the USA²². All MDA recognised clinics have a multidisciplinary model of care where patients can see multiple professionals in the one location. The role of the MDA Healthcare Service Coordinator in each state is multifaceted and includes, meeting patients at clinic, providing education to schools, assisting with independent educational plans, coordinating a bank of loan equipment for families in need and organising the annual state neuromuscular camp. Additionally, each state branch provides neuromuscular centres with a discretionary fund which can be used to cover other costs other such as attending clinics, genetic testing and/ or travel to clinics for families without means or private insurance cover.

The Parent Project Muscular Dystrophy (PPMD) established in 1994 is another non-profit organisation in the USA²³. Their focus is advocating for and funding research for males affected by Duchenne Muscular Dystrophy (DMD). They work collaboratively with many organisations and have developed a certification process to recognise centres providing comprehensive care.

5.2.1 Neuromuscular Centre - Boston Children's Hospital



Established in 1869, the Boston Children's Hospital is one of the oldest and largest paediatric hospitals in the United States of America²⁵. It is a comprehensive centre for paediatric healthcare with satellite locations across Massachusetts. The main campus is located in the Longwood Medical district which is home to many world renowned health care institutions and the Harvard Medical School. Recognised for its excellence in health care the Boston Children's Hospital provides care from birth to 21 years of age and occasionally for adults²⁵. As a 'not for profit' organisation, resources are contributed back to the community through the provision of unreimbursed health care, community health improvement programs, education of health care professionals and subsidized health care services²⁰.

The Neurology Department at Boston Children's Hospital is one of the oldest, largest and most experienced in the world and has a team of around fifty Neurologists and thirty neurology Nurses who provide care through many of the subspecialty programs²⁰. The department offers two neuromuscular subspecialty programs, which are directed by Dr Basil Darras. Established in 1977 the Neuromuscular Centre was one of the first clinics providing comprehensive care in the USA. The service provides care to those with Muscular

Dystrophy, Myopathies, Neuropathies and Neuromuscular Junction disorders. For children who have Spinal Muscular Atrophy (SMA) there is a dedicated SMA program.

During my visit to the Boston Children's Hospital I had the opportunity to meet the Neuromuscular Centre team, attend clinics, meet one of the MDA Representatives, work closely with Neurology/Neuromuscular Nurse Practitioner, visit the Clinical Research Facility (CRF) and tour the inpatient neurology ward and spend time with the Clinical Nurse Specialist.

I attended the Neuromuscular clinic which is an all-day clinic held weekly on Mondays. It has a multidisciplinary model of care led by Dr Basil Darras. The clinic is supported by a Nurse Practitioner, two Neurology Fellows and two Neurology Residents, a genetic counsellor and a physiotherapist. An MDA representative also attends the clinic as does the Clinical Trials Coordinator. Around 20 patients were seen at clinic with the Nurse Practitioner, Fellows and Resident leading the consultations, supported by Dr Basil Darras.

During clinic the team is based in a central meeting room which facilitates patient case management discussion. Each patient is seen by the Nurse Practitioner, Fellow or Resident, presented to Dr Darras, who then sees the patient towards the end of their consultation. Patients are also seen by the physiotherapist or genetics counsellor or MDA representative as required. The neuromuscular program at Boston Children's hospital has a fulltime training fellow; however the centre also works collaboratively with Adult Neurology/Neuromuscular trainee programs throughout Boston. Adult trainees attend the clinic on rotation as part of their comprehensive training.

The MDA supports seven neuromuscular services in the state of Massachusetts, with around 2000 people across the lifespan registered as having a neuromuscular disorder. A MDA representative attends the neuromuscular clinics at Boston Children's and is available to answer questions, provide educational materials and coordinate and link with community resources.

Recognising the specific needs for patients with Spinal Muscular Atrophy (SMA), the Neurology department has established a dedicated service. The Spinal Muscular Atrophy program offers a coordinated multidisciplinary clinic each month supported by Neurology, Respiratory, Orthopaedics, Physiotherapy, Dietetics and a Nurse Practitioner. This program is highly regarded and supported by the SMA Foundation but unfortunately my visit did not coincide with this monthly clinic.

Certainly a highlight of my visit to Boston Children's Hospital was the opportunity to shadow one of the Nurse Practitioners, Regina Laine who works with the neuromuscular program. As a Magnet Institution, Nurse Practitioners at Boston Children's Hospital are highly regarded and represent an integral part of the Neurology nursing workforce and the neuromuscular programs. Many of the Neurology nurses have additional accreditation as

Certified Neuroscience Registered Nurses (CNRN). The scope of the Regina's role includes seeing both new and review patients in clinic and also extends to investigative procedures such as lumbar punctures. The neuromuscular service also has a team of senior Registered Nurses who support families and clinicians and coordinate care needs in between clinic visits.

Transition to an adult neuromuscular service is not enforced at Boston Children's Hospital and it seems that there is no organisational mandate to discharge patients from paediatric services. "Once the patient, always the patient" was the philosophy when I enquired about transition pathways for adolescents and young adults with neuromuscular conditions. Like most centres worldwide the concept of transition to adult clinics is a relatively new one and patients continued to be eligible to attend the Neuromuscular Centre and be admitted to Boston Children's Hospital for an inpatient stay well into adulthood if required.

The Neuromuscular Centre and SMA program are very active in research and involved in around 10 clinical studies. The Boston Children's Hospital is also one of the 5 sites in the USA that is funded by the Muscular Dystrophy Association (MDA) to conduct multi-centre clinical trials²⁵. There is a team of three staff and a dedicated clinical trials coordinator to support the neuromuscular research. Boston Children's Hospital is a leader in research and innovation and has a Clinical Research Centre and Clinical and Translational Study Unit that can provide administrative support, research nurses and processing of laboratory and specimen collections²⁵.

On my last day at Boston Children's Hospital, I met with the Clinical Nurse Specialist and spent the morning on 9NW, the Neurology/ Neurosurgical and Neuro-oncology ward. Unlike the UK and Australia, Clinical Nurse Specialists in the USA are required to have a Master's degree.

Whilst not specific to my neuromuscular fellowship, given the recent opening of the Lady Cilento Children's Hospital, it was an opportunity to observe the models of care on the Neurology ward and the coordination and communication between the specific services and ward team. Daily at 10.30am, the Clinical Nurse Specialist, Nurse Unit Manager, Play therapist, Insurance support Nurse and Social Worker would have a multidisciplinary meeting with each Nurse Practitioner following the medical ward rounds. This facilitated timely coordination of care, a comprehensive update and assisted with care and discharge planning. Whilst not relevant for the Australian community, the presence of a dedicated nurse whose role was to liaise with insurance companies to obtain clearance to proceed with treatment plans gave an interesting insight to the USA health system.

5.2.2 Neuromuscular Program - Children's Hospital of Philadelphia - (CHOP)



My visit to Philadelphia was the final leg of my fellowship. As the first Children's hospital in USA opening in 1855, CHOP has a reputation for being an international leader in paediatric health care. The hospital is located within the bustling University City precinct alongside other world renowned institutions such as the Hospital of University of Pennsylvania (HUP) and the University of Pennsylvania campus.

The Neuromuscular Program at CHOP is one of the specialised services within the paediatric Neurology division. The Neurology division is recognised as one of the largest and most comprehensive in the USA and provides paediatric services throughout Philadelphia and the surrounding region²⁶. Families also travel from the state of New Jersey due to the close proximity to Philadelphia.

The Neuromuscular program provides care to over 500 patients each year and is based at the main CHOP campus with clinics held in the Woods Outpatient Building²⁶. It has a multidisciplinary model of care and the team consists of four Paediatric Neurologists, two Registered Nurses, two Physiotherapists, a Dietician, an Occupational Therapist, a Genetic Counsellor and a Social Worker. This program is recognised by the Parent Project Muscular Dystrophy (PPMD), The Spinal Muscular Atrophy (SMA) Foundation and the Muscular Dystrophy Association (MDA). MDA Healthcare service coordinators work closely with clinical teams and attend each clinic. Clinical research and drug trials are also well established in this service, with dedicated Research nurses who are part of the Clinical and Translational Research Centre.

To understand the demand for services in the state of Pennsylvania I spoke with Laura McClellan the MDA Health Service coordinator at clinic who reported that the MDA branch in Pennsylvania has 2400 people across the lifespan with neuromuscular disease registered on their state wide database.

During my visit, I spent time with each member of neuromuscular multidisciplinary team and observed the Seating clinic with Alan Glanzman, the EMG/NCS clinics at HUP with Dr John Brandsema, and the Wednesday Neuromuscular clinic and multidisciplinary meeting. I also had the opportunity to meet with one of the neuromuscular Research Nurses and observe a clinical trial visit for a young boy with Duchenne Muscular Dystrophy (DMD) and visit the Clinical Research Facility and dedicated research pharmacy.

The Neuromuscular program is comprehensive and holds a number of different clinics each week. It incorporates the NCS/EMG service working in partnership with the HUP. Multidisciplinary planning meetings bring together the core neuromuscular team weekly and with other specialists from Cardiology, Respiratory and Orthopaedics on a monthly basis. The neuromuscular program works closely with the child's Paediatrician and/or

Primary Care Provider and has a specific monitoring and review schedule for patients recently commenced on steroid treatment.

Weekly Neuromuscular clinics are held on Tuesday afternoons and a full day on Wednesdays. These are attended by 2-3 Neurologists, the Neuromuscular Fellow and the multidisciplinary team. Complex care clinics are held monthly on Fridays, offering a coordinated multidisciplinary review with Neurology, Cardiology and Respiratory. Patients are seen yearly in this clinic with a 6 monthly review in one of the other multidisciplinary clinics.

In addition, Dr Carsten Bonnemann, a Paediatric Neurologist and Neuro-genetics specialist leads a consultative complex diagnostic clinic every 1-2 months. Upcoming changes within the service will see the commencement of clustering the Spinal Muscular Atrophy (SMA) patient group into a dedicated clinic with respiratory input, held each month on a Wednesday. This is in recognition of the specific care and management needs for patients with SMA and will assist with coordination of visits for those involved with clinical research.

During clinic the multidisciplinary team are based in a central meeting room which serves as the hub and facilitates communication between each team member. Patients are allocated to their own outpatient room for the duration of their visit with the clinician moving rooms to see each patient. All patients are routinely seen by each member of the multidisciplinary team.

I was impressed by the ease of use, clinical functionality and seamless integration of the electronic medical record at this facility. The electronic medical record allows for direct entry of clinical notes at time of consultation into a combined multidisciplinary template which is then electronically signed by the lead consultant after the final appointment on the day. This then automatically generates a multidisciplinary clinic report ready to be mailed out on the same day negating the dictation, typing and review of the medical letter which is often a barrier to a timely turnaround time in many health services.

The Neurology nurses work in a cluster/portfolio model. Michelle Bordeleau is the primary Registered Nurse for the neuromuscular program supported by a secondary nurse Beth Kunzelman, whose primary Registered Nurse for the Multiple Sclerosis (MS) and Leukodystrophy programs. Michelle then provides secondary support to Beth. This model appears to work well, with both nurses available to support outpatient clinics. The scope of the nursing role within the neuromuscular service includes attending and reviewing patients on clinic day, being a key contact between clinic appointments for families, managing any clinical issues and patient advocacy liaising with insurance companies.

Like many paediatric neuromuscular programs worldwide, transition pathways for young adults in the CHOP service presents challenges. CHOP considers adulthood to be the age of 21 years however; with no specific pathway for neuromuscular patients once they turn 21

some paediatric clinicians retain their patients past this age. Some young adults may be transitioned to adult cardiologists and/or cardiac failure clinics, whilst other clinicians still have the capacity to admit to CHOP for acute admissions and provide follow up in outpatient clinics. Whilst there are some centres in Philadelphia offering adult neuromuscular services they are reported to be quite specialised and focus on particular diseases such as Amyotrophic Lateral Sclerosis (ALS) which is a condition diagnosed in adulthood. Overall CHOP as an organisation clearly supports the concept of preparing children and adolescents for the eventual transition to adult services which is evidenced by the suite of age specific resources online and in brochure format.

The CHOP Neurology division has three social workers supporting their outpatient services. Their caseloads are based on a portfolio/cluster model, with Sarah Stoney the neuromuscular Social Worker also covering other progressive and degenerative conditions such as Leukodystrophy, MS and other Demyelinating disorders. The scope of her role is still evolving and offers psychosocial support for patient and families, advocacy within the school, support with education plans and assisting with accommodation, accessibility, housing and equipment. The inclusion of mental health screening for adolescents and young adults with chronic illness is seen as a key area for further focus.

Sarah has been in the role since early 2015 and has commenced some dynamic initiatives to support young adults with neuromuscular disorders. She coordinates online support group meetings every 2 months and every third meeting the group meet in person to hear from a guest speaker, usually a young adult with a neuromuscular condition. These meetings have been well received by both patients and families. Despite the recognition of potential psychological issues for children and young people with progressive, life limiting neuromuscular conditions it remains challenging to deliver this support in the specialist neuromuscular services within Australia. To find the appropriate personnel with expertise and special interest along with funding such a position could be challenging. Most likely, these services would require initial funding from external sources such as patient support and advocacy organisations in order establish and validate the position.

Another initiative from the greater Social Work department at CHOP is the introduction of customised patient care binders for families with a child who has a chronic and complex condition. These binders contain service specific information and can be adapted for each patient and can include patient resources, clinic letters, genetic and test reports and can evolve over time. These have been well received by families at CHOP and are an example of a practical tool that can assist families who are frequent users of health care services.

The neuromuscular program is active in clinical research and is involved in a number of international neuromuscular research projects and clinical trials. I had the opportunity to meet with Nancy Videon, one of the Research Nurses which was a valuable experience to observe how the research projects integrate with clinical services. There are two Research Nurses involved in neuromuscular trials however their position is separate to the neurology

department as they are managed by the Clinical and Translational Research Centre. This centre also manages all the preliminary work associated with setting up clinical trials such as costings, budget and ethics submissions. Research nurses are then allocated to particular clinical trials. There is a Clinical Research Facility within the main CHOP building which provides a central area for phlebotomy, laboratory staff to process blood specimens and provision for 4-6 day stay patients attending research visits. There is also a dedicated clinical trials pharmacy within CHOP. This model seems similar to one I observed at Newcastle Clinical Research Facility however has the benefit of laboratory technicians working onsite who are responsible for processing the research pathology specimens.

Lastly, another finding that I found particularly interesting whilst at CHOP was the availability of Deflazacort medication in liquid form. In Australia Prednisolone, another type of glucocorticoid is a readily available and inexpensive medication in both tablet and liquid form and is used as the first line treatment for boys with DMD. In Australia Deflazacort is only available in tablet form and is used if side effects from Prednisolone are unmanageable for the patient or family. It is also a more expensive and requires being able to swallow tablets which limits its use in the younger patients. Families in the USA can order directly from the pharmaceutical company in the UK after faxing or emailing their prescription. I aim to work with our hospital pharmacy department to explore the possibility of making this available.

6 CONCLUSIONS

Over nine weeks I travelled to Hospitals and Neuromuscular Centres in London, Newcastle upon Tyne, Boston and Philadelphia. On this journey I met some of the most passionate and dedicated professionals many whom have been instrumental in developing clinical guidelines, advocating for improved access, equity and delivery of clinical care and leading research to find treatments for neuromuscular disorders. It is with pride that I reassure the Australian community that paediatric neuromuscular disease within Australia is managed by highly qualified and extremely talented teams who strive to provide the highest standard of care within a coordinated multidisciplinary framework.

When compared to Queensland all the neuromuscular services I visited service much larger populations and therefore have a greater cohort of patients and number of neuromuscular specialists supporting clinical care and research. Additionally, despite the vastly different health care systems in the UK and USA, health care facilities in both countries have strong partnerships with academic institutions which provide a strong foundation for clinical research. All the centres were well resourced with staff from many disciplines that had such passion for neuromuscular disease.

Throughout my fellowship I did identify several areas which I think would result in improved delivery and coordination of care for Australian children and adolescents with neuromuscular conditions. The key differences I observed were the recognition of neuromuscular disease as a subspecialty of Neurology, England's national approach to the delivery of neuromuscular services, clinical pathways to guide care, organisational commitment to transition through pathways and resources for young adults, patient information resources, psychosocial support in specialist neuromuscular clinics and a coordinated national approach from Muscular Dystrophy groups.

The Australasian Neuromuscular Network has been instrumental in bringing together clinicians, researchers and muscular dystrophy groups and I hope this report can build on that strong foundation and continue to improve care and management for all Australians affected by neuromuscular disease.

7 RECOMMENDATIONS

The subspecialty of neuromuscular disease is rapidly expanding, particularly in the areas of diagnosis, patient numbers, increased life expectancy, multidisciplinary care, and the development of novel therapies. Therefore it is essential to build sustainable neuromuscular services throughout Australia to keep up with this growth. Australia can learn from the UK's national approach to establishing specialist neuromuscular centres and service delivery networks to ensure equity in access and standards of care, regardless of geographical location. State health systems could also benefit from auditing unplanned

admissions for people with neuromuscular conditions to assess the true cost of disjointed health care for this vulnerable patient group and to the Australian community.

It is recommended that all paediatric Neurology services in Australia establish multidisciplinary services to optimise clinical care and health outcomes and ensure clinical trial readiness. The education and development of clinicians with an interest in neuromuscular conditions to support the growth of specialist centres should also be seen as a priority.

It is recommended that Neurology Fellow trainee positions be expanded to include clinical experience in neuromuscular services, inclusive of NCS and EMG training. Ideally a Neuromuscular Fellow Trainee position should be established in each tertiary Neurology service to recruit doctors with a special interest in neuromuscular disease.

As one of four Clinical Nurse Consultants in Australia working in the neuromuscular field, I would like to build on the professional nursing networks I established whilst overseas. The subspecialty of neuromuscular nursing is well recognised in both the UK and USA and as a field that is rapidly growing in Australia, building a sustainable nursing service with an interest and experience in neuromuscular disorders is imperative.

It is recommended that all tertiary Neurology services establish a dedicated Advanced Practice Neuromuscular nursing position and explore the potential for Nurse Practitioners. To observe the Nurse Practitioner model within neuromuscular centres and understand the scope of the role was an inspiration for future benchmarking for neuromuscular services in Australia. Specialist neuromuscular Nurses and Nurse Practitioners were highly regarded in the services I visited and their role was integral to service delivery. Certainly the Dubowitz Neuromuscular Centre and Boston Children's Hospital would be key centres to link with for further development and mentorship of the neuromuscular Nurse Practitioner role.

It is recommended that outreach clinics be established as part of each tertiary neuromuscular service. This will improve the coordination and access to clinical care for all patients particularly those with an advanced stage of disease who disengage in regular follow up due to the physical challenges with travelling to tertiary health care facilities. This will also facilitate the development of clinical care networks across the state with tertiary centres supporting patients and clinicians in regional areas in a hub and spoke model. Where outreach clinics cannot be provided or are unsuitable, organisations should consider planned inpatient admissions for timely, coordinated multidisciplinary review.

Advances in care and management have led to improved health outcomes and an increased life expectancy for children with neuromuscular disorders²⁷. Despite there being no cure and many neuromuscular conditions being progressive in nature, children are now living into adulthood, creating the need for transition pathways to support adolescents moving from paediatric to adult health services^{27,28,29}. To support and improve the care and

management of adolescents and young adults with neuromuscular disease in Queensland the establishment of a 'lifespan' service is recommended. Paediatric clinics and adult care providers need to work in partnership to provide seamless care for adolescents and young people within a 'lifespan' model of care. Based on the models I observed in London and Newcastle upon Tyne this would provide continuity and the continued engagement of young adults in their own care. With Queensland's population size being comparable with the population serviced by the Muscle Centre in Newcastle upon Tyne, this could be achievable. Since a paediatric neuromuscular service is already established, adult services could build on this foundation, establishing transition and nurse led clinics, sharing expertise and resources. The added benefit is that this would reduce the unexpected and preventable acute admissions associated with disjointed adult services and/or loss to follow up and justify any costs associated with establishing the service.

It is recommended that Queensland Health and Children's Health Queensland consider the establishment of a transition program, resources and pathways to support clinicians in preparing adolescents for transfer to adult services. It is well recognised in the literature that how the young person is transitioned to adult services is crucial to long term well-being and health outcomes and "education of patients, families and healthcare professionals on the concept of health care transition is critical"³⁰. Currently paediatric services in Queensland are mandated to transition patients by 18 years of age, however unlike NSW and Victoria there is no state-wide or organisational support or resources available to facilitate this. As the Clinical Nurse Consultant coordinating the neuromuscular service at Children's Health Queensland I would like to work with the executive teams to establish a transition program and or pathway to support adolescents, clinicians and families.

It is recommended that clinical pathways be developed for neuromuscular patients undergoing common surgical interventions such as spinal fusion, insertion of a gastrostomy device and orthopaedic surgery. This will improve coordination and quality of care and communication between the neuromuscular and surgical teams, provide support for surgical nursing staff from the specialist neuromuscular team during the inpatient admission and provide quality and safe care for this complex patient group. I aim to work with the Clinical Nurse Consultants in the surgical teams to develop these pathways. It is also recommended that Children's Health Queensland consider establishing a multidisciplinary framework for all complex patients requiring surgery. The multidisciplinary pre spinal surgery meeting that I attended at GOSH demonstrated an organisational commitment to safe and timely care and in my opinion a benchmark for paediatric hospitals.

The review and development of additional patient and family resources and emergency management guidelines to support the neuromuscular service at Children's Health Queensland and provide consistency in care across the state is recommended. As a centre of excellence, the Dubowitz Neuromuscular Centre has developed a suite of resources to support patients, families and clinicians and should be used as benchmark for tertiary

services. The Treat NMD network has also developed excellent resources which we currently provide to families at time of diagnosis and the Australasian Neuromuscular Network is currently developing best practice guidelines.

Based on my experience visiting other neuromuscular centres, I will continue with establishing specific condition and disease stage clinics, particularly for DMD and SMA. This has already been well received by our multidisciplinary team and has improved the coordination of care and use of resources. The service will continue to monitor feedback from our patients, families and Muscular Dystrophy Queensland. I will also continue developing 'Family Files' to compile resources for all new patients introduced to the service. Both GOSH and CHOP had great examples to model.

Since returning from my fellowship I have commenced fortnightly multidisciplinary neuromuscular meetings, which are in addition to our weekly pre clinic meeting. This has improved the case management for our patient group. Establishing this meeting has been rewarding as we now have input from the non-invasive ventilation and spinal/scoliosis specialist nursing staff and have also engaged the Neurology Social Workers to increase the psychosocial support provided at a tertiary level. The need for psychosocial support for neuromuscular patients has well been recognised and I hope to establish links for our Neurology Social Workers with the Family Therapist, Specialist Care Advisor and Psychologist from the centres in England and with the Social Worker at the Children's Hospital of Philadelphia.

It is recommended that neuromuscular services continue to engage with Non-Government and patient support organisations and work collaboratively to provide patient care and advocate for essential tertiary and community services. The delivery of evidence based care within a multidisciplinary setting is essential to ensure best practice and quality of life outcomes for patients and families.

Lastly, I was impressed with the coordinated and national approach of Muscular Dystrophy UK and the MDA with their role in advocacy and supporting patients and neuromuscular centres. I think it would be worthwhile for charitable patient support organisations in Queensland and Australia to explore the business models and governance structure of these non-profit organisations to ensure a cohesive and collaborative approach to supporting those affected by neuromuscular disease in Australia.

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9 APPENDIX

9.1 GLOSSARY OF NEUROMUSCULAR TERMS

Antibodies	Protein produced by the body's immune system in response to a 'foreign' material in the body.
Aspiration	Food, liquid or saliva entering the trachea (airway), instead of the oesophagus. It can cause choking or in more severe cases respiratory infections.
Autoimmune disorders	Conditions in which the immune system produces antibodies that attack the body's own cells. Example, Autoimmune Myasthenia Gravis
Becker Muscular Dystrophy	A genetic disorder in which the muscle cells break down causing progressive muscular weakness. Becker muscular weakness usually affects only boys/men and is caused by a lack of dystrophin protein. The disorder resembles Duchenne muscular dystrophy but is less severe.
Cardiomyopathy	A heart condition. The most common type in children or young people with neuromuscular conditions is dilated cardiomyopathy. This involves enlargement and thinning of the heart muscle, deterioration of the muscle and loss of reduced movement of the heart wall.
Charcot-Marie-Tooth disease	A genetic condition that affects the peripheral nervous system characterised by progressive weakness and wasting of the muscles in the calves, lower arms, hands and feet.
Clinical Trials	A test in humans of an experimental medicine or therapy.
Congenital	Present at birth or soon afterwards.
Congenital Muscular Dystrophy	A group of genetic conditions which share common muscle pathology but present with different clinical symptoms and are caused by a mutation in different genes.
Duchenne Muscular Dystrophy	A genetic disorder in which muscle cells break down and are eventually lost, causing progressive muscle weakness. The condition usually affects only boys and is caused by a lack of dystrophin protein.
Echocardiogram (Echo)	The use of ultrasound to examine and measure the structure and function of the heart.
Electrocardiogram (ECG)	A test to measure the electrical activity of the heart.
Electromyogram (EMG)	An examination of muscles by insertion of a very fine needle electrode. Interpretation of electrical patterns at rest and activity can assist with the diagnosis of various diseases of the muscle or nerve.
Facioscapulohumeral Muscular Dystrophy	A muscle wasting condition, caused by a genetic defect. The name describes the distribution of weakened muscles: 'facio' – facial; 'scapulo'- shoulder blade; 'humeral' – upper arm.
Gastrostomy	An opening through the abdominal wall into the stomach. A feeding device is inserted through this opening, which allows a person to be fed directly into his or her stomach, bypassing the mouth and throat. Some neuromuscular disorders cause dysphagia and increase risk of aspiration.
Genes	Genes are made of DNA and each carries instructions for the production of a specific protein. Genes usually come in pairs, one inherited from each parent. They are passed on from one generation to the next, and are the basic units of inheritance. Any alterations in genes (mutations) can cause inherited disorders.

Genetic counselling	Information and support provided by a specialist doctor, nurse or counsellor, to people who have genetic conditions in their families, or are concerned about a genetically transmitted condition.
Genetic disorders	Conditions resulting from alterations in the genetic make-up on an individual. They may be caused by defects in single genes or whole chromosomes. Parts of which may be lost, duplicated, misplaced or replaced. Genetic disorders can be caused by defects in one or more genes.
Limb Girdle Muscular Dystrophy	A group of conditions affecting the limb girdle muscles (shoulders and hips) which present with different clinical symptoms and are caused by mutations in different genes.
Mitochondrial myopathies	Genetic disorders of muscle which affect the mitochondria, caused by either a fault in the DNA of the mitochondria or the nucleus. Without a sufficient supply of energy the cells fail to function properly, causing disease.
Muscle Biopsy	Surgical removal of a small amount of muscle tissue for examination in the laboratory.
Mutation	The alteration of a gene. Mutations can be passed on from one generation to generation.
Myasthenia Gravis	A chronic, autoimmune disorder which causes muscle weakness and excessive muscle fatigue, thought to be caused by a faulty immune system.
Myopathy	Muscle weakness
Myotonic Dystrophy	A genetic disorder with symptoms of myotonia, progressive muscle wasting and weakness, usually affecting the face, jaw and neck muscles. Caused by an increased number of repeated units of genetic material (called triplet repeats) within a gene.
Neuromuscular	Anything that pertains to the nerves, muscles or the nerve-muscle junction.
Non-Invasive Ventilation (NIV)	The administration of mechanical breathing support with positive airway pressure delivered through a face mask.
Scoliosis	Curvature of the spine.
Spinal Muscular Atrophy	Genetic conditions resulting in progressive muscle wasting and loss of lower motor neurons in the spinal cord. Spinal Muscular Atrophy is caused by the absence of (Survival Motor Neuron) SMN1 genes and follows an autosomal recessive pattern of inheritance.
Steroids	Drugs that are similar to some of the hormones produced by the body. They are classed as corticosteroids and are similar to natural hormones produced by the adrenal glands. They are often prescribed to boys with Duchenne muscular dystrophy. These steroids may have an effect on stabilising or even improving muscle strength for a period of time but side effects are relatively common. The main steroids that used are Prednisolone or Deflazacort.

Reproduced from:

1. Muscular Dystrophy Campaign. Self- Management pack for people with an adult-onset neuromuscular condition.
2. Great Ormond Street Hospital for Children. Glossary of terms used in the Neuromuscular Disorders Service.
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