South West Neuromuscular Operational Delivery Network

NHS

Issue 2 | March 2015

Network Update

There have been a few changes to the Network team since the August newsletter.

New Staff:

We said sad farewells to Vicky Farrell, Dr Sadie Unsworth-Thomas, Jane Kinney and Jess Romero. Their contribution to the Network was much appreciated.

However, we are pleased to welcome Janet McCay, Specialist Physiotherapist for Cornwall, and two Neuromuscular Advisors for Devon and Cornwall; Claire Stayt and Tamara Eaton (see page 3 for details). We will also be welcoming a new Specialist Clinical Psychologist to Bristol in April.

Network Governance:

Dr Andria Merrison, Consultant Neurologist at North Bristol NHS Trust, has taken over as Chair and Clinical Director of the SW Neuromuscular ODN.

An Executive Board has been established and membership expanded to reflect the specialties involved in neuromuscular pathways of care.

NHS England South has been formalising the governance arrangements for the six Operational Delivery Networks (ODNs) in the South West. The ODNs will report three times a year to an Oversight Board which will consist of representatives from Commissioning and Provider organisations.

Making a Difference:

Dr Sadie Unsworth-Thomas, Specialist Clinical Psychologist, led a number of pilot workshops in Bristol. With the support of the Specialist Physiotherapists and Neuromuscular Advisors, pilot workshops were held on:

- Transition from Paediatric to Adult services
- Living Well with Muscle Disease Course
- Duchenne Muscular Dystrophy Parents Day.

Feedback was extremely positive and we plan to develop these educational and support events for patients and their families across the South West in the coming year.

What's next?

The Network has produced a challenging Work Programme for 2015-16. Key projects include:

- A Bristol Pilot for a neuromuscular transition process using the Ready, Steady, Go documents (see page 3);
- Clinical Pathways and Standards of Care for specific neuromuscular conditions;
- Condition specific physiotherapy assessments;
- Commitment to deliver a **SWIM Conference** twice a year (see page 6 for Spring programme) and patient focused events to encourage self-management and increased knowledge about their neuromuscular condition.

Inside this Bulletin

This Bulletin pulls together the themes from the SWIM Winter Conference held last November to give you a taste of what was discussed if you were unable to attend.

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New Research: Men with Duchenne Muscular Dystrophy talk about being men and interactions with social care

Professor David Abbott, Reader in Social Policy,
Norah Fry Research Centre, University of Bristol

The social care sector needs to focus on gender-specific social and sexual needs when looking after men with Duchenne Muscular Dystrophy (DMD), a recent study has found.

Researchers at the School for Policy Studies, University of Bristol (working with Dr Jon Hastie and the Duchenne Family Support Group) looked at the views of 20 men, aged from 21 to 33, who have DMD. The study, funded by the National Institute for Health Research (NIHR) School for Social Care Research (SSCR), revealed that men sometimes found that social care took no account of their male gender and they wanted more support with social activities and sexual relationships. One participant said: "I think the actual impact of social care can be incredibly emasculating, if it is set up in such a way that it takes away your independence and your autonomy. It's not even gender neutral, because gender's not even in there."

Men with DMD in the study described a strong sense of male identity typified by phrases like, "I think of myself as a normal bloke". They also talked about constructions and perceptions of being a man which arguably challenged stereotypes in a positive way. For example: believing that maturity and self-awareness might matter more than having a 'traditional' male body; muscles; emotional strength and maturity developed by facing some of the trials and challenges associated with living with DMD; and understanding that being a man with DMD necessitates care and support and that acceptance of this demonstrated strength rather than weakness.

Participants offered accounts of their similarities and differences from stereotypes of 'ordinary' (usually non-disabled) men. Areas of similarity included: being and wanting to be sexually active; cultivating self-esteem through academic achievement, work and rewarding activity; hobbies and interests such as sports, drinking, computer games, technology, art and design; being a father (as one participant was); living independently and/or owning a home. Areas of difference included: difficulties in attracting sexual/intimate partners; barriers to obtaining paid work; concerns about moving out of the family home relating to care, support and loneliness; restrictions on a social life linked to shortage or inflexibility of support arrangements; physical appearance and muscularity.

The research suggested men were not being given cues to have discussions about topics such as masturbation when hand function was problematic, getting physical support to facilitate sexual relationships and the use of sex workers (for those for whom this was of interest).

Professor David Abbott, from the Norah Fry Research Centre at the University of Bristol, said: "This group of men is just one example of people living with long-term conditions who need good quality social care. Having support to dress and wash obviously matters, but what about life enhancing activities - sex, relationships, fun, risk – why are these off limits? Imagine if being the man you wanted to be meant you needed flexible and imaginative social care support. Imagine if you didn't have this. The men we spoke to wanted to be treated like men, despite their increasing reliance on physical support. A headline message for the social care sector is to think about gender and the whole range of social and sexual needs men living with long-term conditions may have."

A summary of the research findings is available at: http://www.sscr.nihr.ac.uk/PDF/Findings/RF48.pdf
A short film based on the research findings is available to view online at: https://www.youtube.com/watch?v=eBovuWg-n34

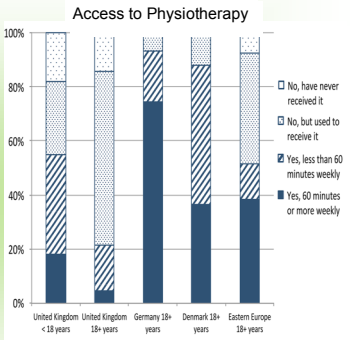
For further information contact: d.abbott@bristol.ac.uk

Related references *(links to abstracts where possible)*:

- Abbott, D. & Carpenter, J. (2014) 'The things that are inside of you are horrible': Children and young men with Duchenne muscular dystrophy talk about the impact of living with a long term condition, *Child Care in Practice*, on-line early DOI: 10.1080/13575279.
- Abbott, D. & Carpenter, J. (2014) 'Wasting precious time': Young men with Duchenne muscular dystrophy negotiate the transition to adulthood. *Disability & Society*, 29 (8) 1192-1205.
- Abbott, D. (2012) Other Voices, Other Rooms: Talking to Young Men with Duchenne muscular dystrophy (DMD) about the transition to adulthood, *Children & Society*, 26 (3) 241-250.
- Abbott, D., Carpenter, J. & Bushby, K. (2012) Transition to Adulthood for Young Men with DMD: Research from the UK, *Neuromuscular Disorders*, 22 (5) 445-456.
- Abbott, D. & Carpenter, J. (2010) *Becoming an Adult: Transition for young men with Duchenne Muscular Dystrophy (DMD)*, Muscular Dystrophy Campaign: London http://www.bristol.ac.uk/media-library/sites/sps/migrated/documents/becominganadult.pdf

Adult Care for DMD in the UK

A recent paper describes how adults in the UK with DMD experience less comprehensive care compared to children in their access to specialised clinics, frequency of cardiac and respiratory assessments, and access to professional physiotherapy.



Read more: "Adult Care for Duchenne MD in the UK"

Preparing Young People and their Families for Adult Services in the South West

Dr Sadie Thomas-Unsworth, Specialist Clinical Psychologist, SW Neuromuscular ODN

SW Strategic Clinical Network

Supporting young people with neuromuscular conditions to make an effective transition between paediatric and adult health care services continues to be a challenge. However, here in the South West we are lucky to have a number of key organisations and professionals who are working together to take this forward. The South West Strategic Clinical Network, Maternity and Children's Network has developed a Transition Working Group comprised of clinicians, AHPs and commissioners who are working together to improve transition across the region. Members of the SW Neuromuscular ODN have played an active part in this group.

Bristol Neuromuscular Transition Care Pathway

Within the South West Neuromuscular ODN we are working hard to improve the transition experience for our young people. With this in mind, the Bristol hub will pilot a Neuromuscular Transition Care Pathway over a six month period which will focus on five patients with Duchenne Muscular Dystrophy and trial the "Ready,Steady,Go" transition documents developed by Dr Arvind Nagra, Consultant Paediatric Nephrologist and Clinical Lead for Transitional Care at University Hospital Southampton NHS Foundation Trust. They are a set of care planning documents that are developed in collaboration with the young person and their family to help ensure that they have the knowledge and skills they will require to become active, autonomous participants in their healthcare when they move to adult services.

The Bristol hub will share the results of the Neuromuscular Transition Care Pathway Pilot Trial at the Winter SWIM Conference.

Bristol Transition Workshop

Also, as part of the Neuromuscular Transition Care Pathway, the Bristol hub piloted a "Transition Workshop" in September 2014. Young people between the ages of 15 and 17 were invited to attend the workshop, along with their parents. Members of both the paediatric and adult neuromuscular service delivered the workshop in which families undertook a series of sessions with the goal of ensuring they:

- Felt more prepared for their transition to adult neuromuscular services;
- Were aware of who they could contact to get further information;
- Had realistic expectations for adult services.

The workshop received very positive feedback from the young people and families alike, with 31% "extremely likely" and 63% "likely" to recommend to friends and family needing similar care.

The SW Neuromuscular ODN plan on developing this into a yearly workshop that is integrated into the Neuromuscular Transition Care Pathway.

If you would like any further information about transition and what the network is doing please do get in touch with the SW Neuromuscular ODN office.

Useful resources

http://www.uhs.nhs.uk/OurServices/Childhealth/TransitiontoadultcareReadySteadyGo/Transitiontoadultcare.aspx
http://www.swscn.org.uk/
http://www.togetherforshortlives.org.uk/professionals/projects/project_two

Cornwall - Specialist Physiotherapist

Janet McCay joined the Network on 8 December 2014. She is based at Camborne Redruth Community Hospital and will provide a specialist physiotherapy service for adults in Cornwall as well as providing a training resource for local physiotherapy teams.

Janet's contact details:
e-mail is janet.mccay@pch-cic.nhs.uk
Tel: 01209 318106 / Working days: Tues & Weds.

Ready, Steady, Go Transition Study Days

12 March 2015 & 9 September 2015

http://www.uhs.nhs.uk/OurServices/Childhealth/TransitiontoadultcareReadySteadyGo/ForhealthprofessionalsReadySteadyGostudyday.aspx

New Neuromuscular Advisors for Devon & Cornwall

We are extremely happy to inform you that we now have two Neuromuscular Advisors covering Devon and Cornwall.

Clare Stayt joined the Network on 5 January 2015. She will be providing support and advice for children and adults in Devon. Contact: clarestayt@nhs.net

Tamara Eaton joined the Network on 2 March 2015. She will providing support and advice for children and adults in Cornwall. Contact: tamara.eaton@nhs.net

Both Advisors will be based at Derriford Hospital Plymouth and will be working Monday to Friday. Their office number is 01752 432912.

Genetic Testing for Neuromuscular Disease

Dr Thalia Antoniadi, Principal Clinical Scientist, Bristol Genetics Laboratory, North Bristol NHS Trust

In the last decade, the scope and demand for genetic testing in clinical diagnostics has escalated. Since 2003, when the complete human genome sequence was published, new technological advances have made it possible for scientists to look in to the DNA, in ways that were not possible before, and find out if something is wrong. There are more than 20,000 genes in the human genome, and around a quarter of these have a known disease association.

Neuromuscular diseases affect the muscles and their interaction with the nervous system. They can be acquired or inherited (genetic) conditions. Neuromuscular diseases are rare, but collectively they affect a lot of people.

Some diseases involve the structure of the muscle cells (Duchenne and Becker Muscular Dystrophy), some affect the muscle-controlling nerve cells of the arms, legs, neck, face (Peripheral Motor Neuron Disease), some affect the site where the nerves and muscles meet (Congenital Myasthenic Syndromes), others affect how the muscles control their movements (Myopathies), while others involve errors in producing energy in muscle cells (Mitochondria Disorders).

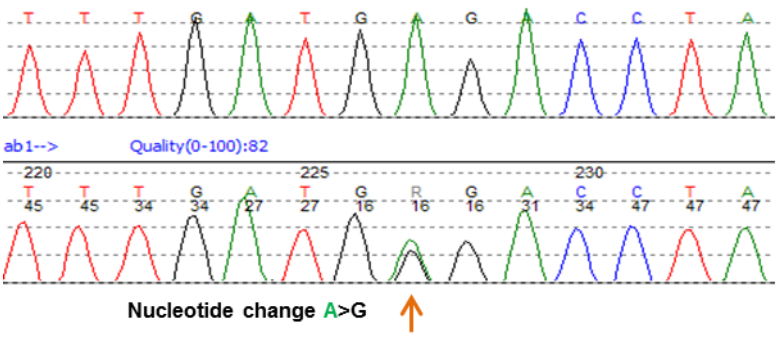
When someone has a genetic neuromuscular disease, somewhere in their genome there is an error (mutation) in their DNA that prevents that gene from functioning properly.

Genetic testing is the process by which we examine the DNA to find the mutation that is causing the symptoms. One disease can be caused by various types of mutations, sometimes in different genes. There are many methods that can be used to look for mutations, and this is because there are many different types of mutations, and there is not a perfect method that can detect them all. For example, DNA sequence analysis is used to look for errors in the order of the bases A,C,G and T that make up DNA. We can also check the number of copies of a whole gene or part of a gene, as deletions or duplications are often disease-causing. A panel of multiple genes, all coding genes (exome), or whole-genome analysis by next generation sequencing is now available for patients where there is no clear link between their symptoms and a particular gene.

Each UK genetics laboratory has expertise in specific disorders; within the neuromuscular service at Bristol we have developed national and international expertise for the rare disorders of Facioscapulohumeral Muscular Dystrophy (FSHD) and Inherited Peripheral Neuropathies (IPN). We also collaborate closely with other laboratories and send and receive DNA samples for testing.

For further information please visit the website of Bristol Genetics Laboratory www.nbt.nhs.uk/genetics, or e-mail: genetics@nbt.nhs.uk

Diagram presenting a small part of DNA sequence



SW Neuromuscular ODN Website

We are pleased to report that the Network's website is now up and running. It still needs a bit of tweaking, but we hope to turn this into a valuable resource for patients and their families and healthcare professionals alike.

Please send us any information about neuromuscular services in your Trust or Community that you think would be of value to people in your area. We would also welcome any suggestions on improving the content. Please tell us what you want!

Thank you to North Bristol NHS Trust for hosting our website and to Jessica Knott, Wed Editor, for the amount of work she put into getting the site live.

www.swneuromuscularodn.nhs.uk

New Support Network for People Newly Diagnosed

Muscular Dystrophy UK (formerly Muscular Dystrophy Campaign) will be introducing a new Community Peer Support Network for patients who have recently been diagnosed with a muscle-wasting condition. This is a UK wide network of peer support volunteers who have a personal experience of living with a muscle-wasting condition.

The Peer Support Network will compliment the work of the already successful Muscle Group Meetings held in the South West. Volunteers will be provided with training and support from members of Muscular Dystrophy UK. Further information is available from Jonathan Kingsley (e-mail: j.kingsley@muscular-dystrophy.org).

Measuring and Improving Quality of Life of those with Muscle Disease

Dr Michael Rose MD FRCP, Consultant and Honorary Senior Lecturer in Neurology,
King's College Hospital and KCL, London

Measuring quality of life (QoL) and understanding the factors that influence it is particularly relevant in a long-term medical condition such as muscle disease (MD). Traditional measures of medicine such as survival, and even impairment measures such as strength, do not capture the real impact of MD on those that live with it. As QoL has a range of definitions, and its measures are used for various purposes, so there is a range of instruments used to measure it.

Common to all of them is that they are attempting to capture a patient's viewpoint and usually take a holistic view rather than just measuring physical impact. Thus they are usually patient reported rating scales that cover social and emotional as well as physical aspects that are felt to contribute to QoL.

QoL measure scales may be generic, meaning they are applicable to many different diseases eg SF-36[1], or they can be disease specific. Generic QoL scales have been used in MD and this allows comparison of the impact of MD with that of other diseases.[2, 3] Disease specific measures are felt to be more relevant to those with the specific disease and hopefully more sensitive to change. There is, to date, only one MD specific QoL scale for adults and a couple for children.[4, 5 6] QoL scales for children lacking capacity are usually completed by a proxy such as the parent. QoL in MD is, not surprisingly, related to the severity of the MD, particularly when this is measured using functional assessments.[3] QoL research has also highlighted the impact of symptoms such as pain and fatigue that are often over-looked by clinicians. However, disease severity does not explain all of the influence of MD on QoL.

There are a variety of psycho-social factors that also have an influence on QoL; especially illness perceptions.[7, 8] Illness perceptions are the cognitions formed by an individual in response to a health threat and they include perceptions about the time-course of the disease, its cause, its consequences, the symptoms involved, its control or curability, and how understandable the illness itself is. Such perceptions allow individuals to make sense of the symptoms they experience and may determine the coping strategies adopted.[9]

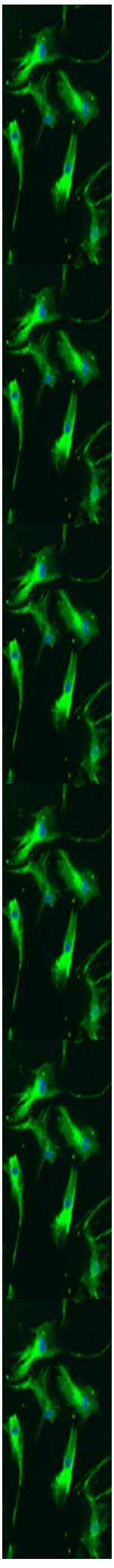
If we want to improve the QoL of those with MD we cannot do so by reducing disease severity, as we do not currently have effective treatments for most MDs. We may, however, improve QoL by better symptom control particularly targeting the oft-neglected issues of pain and fatigue.[10] We might also be able to improve QoL by altering adverse illness perceptions.

We have identified a pattern of illness perceptions particularly likely to worsen QoL of those with MD.[11]

We have used this knowledge to hypothesise that acceptance and commitment therapy (ACT) would be the most appropriate model for altering adverse illness perceptions in those with MDs.[12] A pilot study using a program of ACT specifically tailored to MD suggests that such an approach might indeed improve QoL.[13] We now have NIHR funding for an randomised controlled trial to confirm the effectiveness (or not) of ACT in improving QoL in those with MD and this will be starting in 2015.

References *(links to abstracts where possible)*:

1. Ware JE, Jr., Sherbourne CD. The MOS 36-item short-form health survey (SF-36). I. Conceptual framework and item selection. MedCare 1992;30:473-483.
2. Boyer F, Morrone I, Laffont I, Dizien O, Etienne JC, Novella JL. Health related quality of life in people with hereditary neuromuscular diseases: an investigation of test-retest agreement with comparison between two generic questionnaires, the Nottingham health profile and the short form-36 items. Neuromuscular Disorders16(2):99-106 2006.
3. Sadjadi R, Rose M, Muscle Study Group. What determines quality of life in inclusion body myositis? . J Neurol Neurosurg Psychiatry 2010;81:1164–1166.
4. Vincent KA, Carr AJ, Walburn J, Scott DL, Rose MR. Construction and validation of a quality of life questionnaire for neuromuscular disease (INQoL). Neurology 2007;68:1051-1057.
5. Davis SE, Hynan LS, Limbers CA, et al. The PedsQL in pediatric patients with Duchenne muscular dystrophy: feasibility, reliability, and validity of the Pediatric Quality of Life Inventory Neuromuscular Module and Generic Core Scales. Journal of clinical neuromuscular disease 2010;11:97-109.
6. Ariaudo G, Orcesi S, Gorni K, et al. Importance of self-report in pediatric quality of life assessment: Experience with the SOLE questionnaire in children with neuromuscular disorders. Neuromuscular Disorders 2007;17:868-869.
7. Graham C, Rose M, Grunfeld E, Kyle S, Weinman J. A systematic review of quality of life in adults with muscle disease. Journal of Neurology 2011; 258(9): 1581-.1592.
8. Graham CD, Weinman J, Sadjadi R, et al. A multicentre postal survey investigating the contribution of illness perceptions, coping and optimism to quality of life and mood in adults with muscle disease. Clinical rehabilitation 2014;28:508-519.
9. Moss-Morris R, Weinman J, Petrie KJ, Horne R, Cameron LD, Buick D. The revised Illness Perception Questionnaire (IPQ-R). Psychology and Health 2002;17:1-16.
10. Voet N, Bleijenberg G, Hendriks J, et al. Both aerobic exercise and cognitive-behavioral therapy reduce chronic fatigue in FSHD: an RCT. Neurology 2014;83:1914-1922.
11. Graham CD, Rose MR, Hankins M, Chalder T, Weinman J. Separating emotions from consequences in muscle disease: comparing beneficial and unhelpful illness schemata to inform intervention development. Journal of Psychosomatic Research 2013;74:320-326.
12. Hayes S, Strosahl K, Wilson K. Acceptance and Commitment Therapy: An experiential approach to behavior change. New York: Guilford Press, 1999.
13. Graham CD. Explaining and changing adverse illness perceptions in people with muscle disease [Doctorate]. London UK: University of London, 2012.



South West Neuromuscular Operational Delivery Network

NHS

South West Interest in Muscles (SWIM) Winter Conference

Thursday 23 April 2015 @ Taunton Racecourse, Orchard Portman, Taunton, Somerset, TA3 7BL

PROGRAMME

09.00 ***Registration & Refreshments***

Morning Session: **Neuromuscular Disorders and their impact on child, family and society**
Chair: **Dr Arni Majumdar, Consultant Paediatric Neurologist**

09.25 **Welcome**

09.30 **Case Presentation 1: A Patient with Nemaline Myopathy speaks frankly about her life**
Dr Arni Majumdar, Consultant Paediatric Neurologist, University Hospitals Bristol NHS FT

09.50 **Case Presentation 2: "Why won't young people do what you want them to do?" – A challenging case.**
Dr Roger Jenkins, Consultant Community Paediatrician, Royal Cornwall Hospitals NHS Trust

10.10 **Feedback from "Living Well with Muscle Disease Pilot Workshop" – A patient centred course around adjusting and coping with a neuromuscular condition.**
Nicola Doran, Specialist Neuromuscular Physiotherapist

10.50 *Refreshments*

11.10 **The Socioeconomic Impact of Neuromuscular Disorders - An International Perspective**
Professor Kate Bushby, Professor of Neuromuscular Genetics, Newcastle University

12.00 **Network Update**
SW Neuromuscular ODN Clinical Lead

12.30 *Lunch (provided)*

Afternoon Session: **Neuromuscular Disorders and the Heart**
Chair: **Dr Alison Hayes, Consultant Paediatric Cardiologist**

13.30 **Case Presentation 3: Duchenne MD with Heart Failure**
Dr Robert Spaull, Specialist Registrar in Paediatrics, University Hospitals Bristol NHS FT

14.00 **Case Presentation 4: Sarcoglycanopathy and the heart**
Dr Venkata Paturi, Consultant Paediatrician, Royal Devon & Exeter NHS Foundation Trust

14.45 *Refreshments*

15.15 **The Heart in Neuromuscular Conditions - An Overview**
Dr Matthew Fenton, Consultant Paediatric Cardiologist, Great Ormond Street Hospital

16.00 General discussion/questions/feedback forms

16.15 ***Close***

Directions to venue: http://www.tauntonracecourse.co.uk/directionsfooter

To register to attend, please click on this link:
https://www.eventbrite.co.uk/e/south-west-interest-in-muscles-swim-conference-tickets-15811394306

Registration closes on **Monday 13 April 2015**