BACD NEWS

The official newsletter of the British Academy of Childhood Disability

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FOCUS ON...

Recent Advances in Neuromuscular Disorders

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BACD

British Academy of Childhood Disability

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BACD is registered in England and Wales under charity number 1177868

The aims of BACD are:

- Mutual support for all those working in district and tertiary level services for children with neurodevelopmental disability
- Promote communication between Child Development Teams
- Organise regular national multidisciplinary meetings on child development and disability
- Good practice in child development and disability
- Encourage debate and promote research into the many outstanding questions in childhood disability
- Work closely with voluntary organisations and others to advocate for children with disabilities and their families
- Encourage and support research in childhood disability

BACD has close links with the following organisations:

- Association of Paediatric Chartered Physiotherapists
- British Association for Community Child Health
- British Paediatric Neurology Association
- Contact
- Council for Disabled Children
- National Network of Parent Carer Forums
- Royal College of Nursing
- Royal College of Occupational Therapists
- Royal College of Paediatrics & Child Health
- Royal College of Psychiatrists
- Royal College of Speech & Language Therapists

BACD is a specialty group of the Royal College of Paediatrics and Child Health.

BACD is a member of the Health Policy Influencing Group (HPIG), convened by the National Children's Bureau and the Council for Disabled Children.

BACD is also a member of the Disabled Children's Partnership and the #SENDInTheSpecialists coalitions.

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EDITORIAL

In this edition of BACD News, we turn our focus to neuromuscular disorders — a field that exemplifies how advances in science, care, and advocacy intertwine to reshape childhood disability. The progress of recent years has been extraordinary: yet progress also brings complexity. As treatments evolve, so too must our understanding of what it means to live well with a neuromuscular condition.

We begin with Spinal Muscular Atrophy (SMA), a condition that has undergone one of the most dramatic transformations in modern medicine. New therapies have changed the outlook completely; children who once faced a terminal diagnosis are now growing into adolescence and adulthood. This change has been made possible through collaboration. National networks such as SMA REACH UK, its adult counterpart, and the SMA Care UK initiative are helping to build the data systems and care standards that turn scientific success into high-quality practice. But while treatment is extending life, the next challenge is ensuring lifelong, equitable support. Effective transition from paediatric to adult services, sustainable local capacity, and fair access for all families must remain priorities.

The changing role of speech and language therapy for children with SMA is one of the most immediate consequences of therapeutic progress. The SLT contribution in this issue describes working with children who are living longer and presenting with complex bulbar, swallowing and communication needs. Their work helps children not only stay safe but communicate, learn, and express themselves. Yet access to specialist SLT remains inconsistent.

The overview on facioscapulohumeral muscular dystrophy (FSHD) highlights the value of patient partnership in research. Advances in imaging and genetics are opening new opportunities for clinical trials, but what truly stands out is the collaborative model — patient registries, engagement events, and co-designed studies that ensures research stays grounded in real experience. Investment in registries and community involvement is not an optional extra.

The article on Congenital Myasthenic Syndromes (CMS) reminds us that genetic medicine brings both precision and complexity. With many causative genes now identified, diagnosis and treatment require careful interpretation. Some subtypes respond well to specific drugs, while others do not. New targeted therapies offer real promise — but their benefits depend on equitable access and long-term monitoring. The message is simple: precision medicine must be accompanied by clear, compassionate communication.

Another theme running through this issue is the importance of psychosocial support. As children live longer and the pattern of disease changes, families face new emotional and practical pressures. The article on mental-health care within Duchenne services shows how embedding psychologists and neuropsychiatrists in clinical teams can make a tangible difference. Emotional wellbeing is not secondary to medical management; it is part of it. Early psychological input, clear referral routes, and properly funded posts should be standard across neuromuscular services.

Research into wheelchair and equipment use challenges outdated assumptions that mobility aids represent loss. In reality, well-timed and well-fitted equipment can enhance independence, reduce fatigue, and increase social participation. We need to shift the conversation from decline to opportunity. Timely, personalised support helps children and adults maintain autonomy and confidence which are important outcomes.

The feature on Cure Myotonic Dystrophy UK illustrates how specialist charities act as catalysts for progress. They connect families, accelerate research recruitment, and raise awareness, often filling gaps that statutory services cannot. Strong partnerships between clinical centres and voluntary organisations strengthen the whole ecosystem of care. Charities bring lived experience and advocacy expertise; clinicians bring scientific and clinical insight. Together, they create momentum for change.

Understanding Hemiplegia offers practical guidance about assessment and interdisciplinary management that is widely applicable. The focus on function, participation and evidence-based rehabilitation reminds us that many neuromuscular principles — goal-setting, family-centred planning, and adaptive technologies — generalise across diagnoses. We see value in sharing these cross-cutting approaches across networks. Dylan's Muscle Days highlights stories of community events, peer support and younger people taking leadership roles reminding us that lived experience is a core asset. Such grassroots initiatives provide families with practical coping strategies, and help younger people to develop identity beyond diagnosis.

Taken together, the articles in this issue send a clear message: medical breakthroughs are essential, but they must be matched by strong systems, integrated services, and genuine collaboration.

We see three priorities. First, build national, interoperable data systems that inform practice and policy. Second, embed allied health and psychological support within neuromuscular pathways and fund them sustainably. Third, continue supporting family-led initiatives that turn knowledge into lived progress.

YASMIN DE ALWIS AND KATY WOOD
Newsletter Co-Editors

SPINAL MUSCULAR ATROPHY: AN UPDATE ON THE CURRENT INFRASTRUCTURE AND INITIATIVES

Spinal Muscular Atrophy (SMA) is a rare neuromuscular disease that causes progressive weakness in the limbs and trunk and atrophy of the muscles. The hallmark of the disease is degeneration of anterior horn cells in the spinal cord, giving rise to characteristic muscle atrophy of varying degrees[1]. The most common form of SMA, which accounts for over 95% of cases, is the autosomal recessive form that results from a homozygous deletion in the survival of motor neuron 1 (SMN1) gene – also known as 5q SMA. In the absence of treatment, 5q-SMA is one of the most common genetic causes of childhood mortality; however the phenotype of the disease is highly variable and is classified into five main types (type 0 to 4) based on age of onset and achieved motor milestones[2]. An almost identical copy of the SMN1 gene, SMN2 also contributes to the phenotypic classification, with the clinical severity inversely correlating with the SMN2 gene copy number[3].

SMA is a complex disease due to the phenotypic spectrum and multiple system involvement. As such, a multidisciplinary care approach is needed for effective management. In addition to the specialist neuromuscular MDT, patients require a combination of respiratory, nutritional, and orthopaedic care alongside mental health support to aid in the decline of psychosocial wellbeing that may be experienced by young people and adults contemplating the progression of their disease[4]. In addition, services need to be integrated with community teams and primary care. As new treatment options become available, the size of the adult SMA population will continuously grow, therefore establishing a standardised and structured approach to the management of adults is essential[5].

In recent years, the treatment landscape in SMA has rapidly evolved with 3 disease modifying therapies (DMT's) now available[2]. Onasemnogene abeparvovec (Zolgensma) is the first and only approved gene therapy in SMA but is not available for older children or adults[6]. In 2019, The National Institute for Health and Care Excellence (NICE) communicated the conditional approval of Nusinersen followed by the conditional approval of Risdiplam in 2022 under a Managed Access Agreement (MAA) in England, Wales and Northern Ireland[7, 8]. The commencement of an MAA allows for time-limited access to new therapies that have shown promise but lack sufficient evidence to be made routinely available on the NHS whilst further evidence is compiled[9]. The MAA also allowed for treatment in adult patients. Consensus on the management of SMA, particularly the development of standards of care (SoC), has focused primarily on the paediatric population with the latest version being published prior to the availability of DMTs. As new 'treated' phenotypes evolved and with evidence suggesting a lack of co-ordination in the provision of structured care in adults [10, 11, 12, 13], the need for a strong clinical network and robust real world-data (RWD) capturing patient reported outcome measures (PROMs) to monitor and gain a better understanding of the impact of novel treatments on the natural history of the disease, became more evident. There was a gap in clinical networks and data collection for adult neuromuscular services, and the expectations of treatments were largely unknown due to lack of available data[14, 15].

SMA REACH UK, was already well established and, is a UK-wide clinical and research network, led by Professor Muntoni and Prof Scoto at UCL and Great Ormond Street Hospital in London, aiming to provide an ongoing platform to improve and design measures to capture information that is meaningful to SMA patients and families.
SMA REACH (REsearch And Clinical Hub) was established in 2012 as a reboot of the existing SMArtnet registry funded in 2006 and has since expanded to include 23 neuromuscular centres across England, Wales, Scotland and Northern Ireland, with more than 500 patients registered as per October 2025.

SMA REACH UK has facilitated the collection of real-world data on children with SMA in the UK. This includes patients on all 3 disease modifying therapies: Risdiplam (Evrysdi), Nusinersen (Spinraza) and Onasemnogene Abeparvovec (Zolgensma) as well as those who have not received any of these treatments. All data are collected according to the standard of care recommendations.

The aim of this data collection is to better understand the impact these treatments have on patient's motor, respiratory and bulbar function, and other neurodevelopmental aspects. Information is collected on drug use and safety, improving understanding of the impact of disease

modifying therapies on quality of life.

SMA REACH UK was appointed by NICE and NHS England to collect data on children in the UK receiving risdiplam or nusinersen via the MAA to support the regulatory approval of these treatments in the UK, collecting and collating data on the medical and physiotherapy outcome of patients every 6 months. Over the years, SMA REACH UK has led on an increasing number of initiatives to address unmet need across the SMA community, hosting workshops, education and training initiatives for the multidisciplinary network [16, 17, 18, 19, 20, 21, 22, 23].

SMA REACH UK is also part of the International SMA Consortium (ISMAC) alongside the US Paediatric Neuromuscular Clinical Research Network (PNCRN) and Italian SMA Registry (Telethon). Collaborating on international initiatives, such as pioneering new physiotherapy scales and outcome measures, has strengthened knowledge across these networks where there are variations in treatment available and timing of starting treatment. This has fostered learning from the experience across these other networks.

Adult SMA REACH is the equivalent UK adult network of 18 sites launched in 2020 to address these unmet needs using a national network of clinical sites, supported by patient advocacy groups, academia and industry. Adult SMA REACH is led by Prof Marini-Bettolo at Newcastle University and Newcastle upon Tyne Hospitals NHS Foundation Trust. In addition, a RWD collection study was later implemented to collect standardised outcome measures, disease evolution and burden in clinic for adults living with SMA. Similarly to the paediatric network, Adult SMA REACH was also appointed by NICE and NHS E to support the RWD collection in adults receiving Nusinersen and Risdiplam.

Adult SMA REACH works collaboratively with SMA REACH UK and allows for transition of patients from the paediatric to adult database, ensuring continuity in the collection and analysis of their data between paediatric and adult services.

Both networks work collaboratively with the UK SMA Patient Registry to ensure the patient voice compliments the clinical data collected. The UK SMA Patient Registry was established in 2008, and registration is patient initiated. The registry collects patient reported data and PROMs from individuals living with SMA in the UK & Ireland using the TREAT-NMD SMA Core dataset (https://www.sma-registry.org.uk)[24, 25].

Following publication of the 2018 updated recommendations for care [10,11], it was apparent that significant variations in delivery of, and access to, care existed across the UK. As DMTs improved survival and changed the disease phenotype, it became clear that the standards of care needed to be reviewed and updated. In particular, recommendations for care needed to be applicable across the UK in order to meet the needs of all those living with SMA and should address the needs of adults living with SMA and young people transitioning to adult care.

In 2024, taking inspiration from the DMD Care UK project [26], clinicians from both paediatric and adult SMA networks, in collaboration SMA UK, launched the SMA Care UK project [27] to update the standards of care for both children and adults living with SMA in the UK. SMA Care UK is led by Dr Childs and Prof Marini-Bettolo and is a collaborative initiative involving healthcare professionals, people living with SMA and other stakeholders. Its goal is to update evidence-based standards of care and harmonise best practice in response to the priorities of the SMA community and the changing landscape of the condition. The project seeks to update care recommendations following review of the current literature and collaboration with relevant experts. Gaps in the evidence base will be highlighted and consensus to agree UK best practice sought. Guidance will be shared with clinicians through publication in the medical literature and presentation at relevant conferences on specific topics such as respiratory care, spine and hip management, whilst parallel ‘family-friendly’ guidance is shared by SMA UK.

Both SMA REACH networks work closely with the SMA Care UK initiative, being actively involved in ensuring collaborative work and alignment of workstreams in reviewing the current standards of care.

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PAEDIATRIC SPEECH AND LANGUAGE THERAPY FOR CHILDREN WITH SMA: A RAPIDLY CHANGING LANDSCAPE

Speech and Language Therapists (SLTs) have a key, but often underrepresented, role in the neuromuscular team. New treatments for children with neuromuscular conditions such as spinal muscular atrophy (SMA) have led to a shift from palliation to the increasing availability of life-saving medical treatments and the resultant requirement for long term therapy and symptom management. This has led to SLTs redefining their role within the neuromuscular team and reshaping practice with this group of children. Children who would have historically been discharged to palliative care services are now surviving, and in some cases, thriving with therapeutic support. These children have a range of complex presentations, unknown prognosis and require specialist speech and language assessment, monitoring, intervention, and research.

Ollie has SMA1. This is his story... by Amy Williams

Ollie was diagnosed with SMA Type 1 at three months old. He was put on a bi-pap ventilator to help him breathe when sleeping, a chest physio regime with cough assist and suction was introduced to his daily routine, and he was given an NG tube (and later a PEG, then Mini Button) for all his nutrition. Ollie was initially treated with Spinraza but, at nine months old, switched treatment paths to Zolgensma (gene therapy).

Following gene therapy, we saw improvements to Ollie's secretion management. He learnt to swallow and spit these out and was less reliant on suctioning. Ollie had severe bulbar weakness from birth – he struggled to latch whilst breast- and bottle-feeding and was unable to form shapes with his mouth.

At around 20 months old (and approximately one year after gene therapy treatment), we introduced tasters to see if Ollie's swallow had improved. This has been something we've done slowly, as the risk of aspiration is very high for someone with a weak cough. We went at Ollie's pace, starting with purees and yoghurts. Ollie knows his own limits, and whilst he has tried harder foods like apple and strawberries, he often only chews it to get the flavour before spitting the food back out. Yet this is all good practice for him as he has to use his tongue to move the food around and expel it from his mouth.

Recently, nearly two years after gene therapy, Ollie has started to try thin liquids, like milk from a spoon and water from a straw cup.

We weren't sure if Ollie would ever speak, so we started Makaton with him from a young age. Shortly after his first birthday he said "Dada" and this was the only word he said for many months. Yet, since then more words and lots of sounds have followed. Now Ollie uses a combination of speech, sounds, Makaton and gestures depending on what he finds easiest.

What is SMA?

Spinal muscular atrophy (SMA) is a genetic disease which leads to progressive muscle weakness, impaired bulbar function and can result in significantly reduced life expectancy in those with the most severe presentations. It is a spectrum disease which has 5 sub-types: from Type 0, being the most severe and impacting pre-term infants, to Type 4, known as late-onset SMA, which is a less severe adult presentation. People with SMA are monitored by neuromuscular teams around the UK.

SMA is caused by genetic mistakes in both copies of the SMN1 gene which is responsible for producing the SMN protein. This protein is required by motor neurones to survive and function. People with SMA do not make enough SMN protein and although there is a backup gene, SMN2, this only provides a small amount of stable SMN protein. Without enough SMN protein motor neurons do not work and the neurons die resulting in muscle weakness and related comorbidities. Without treatment, babies with the more severe presentations of SMA type 1 are unable to hold their heads up, bring their hands to their mouths or sit independently. They require respiratory support, ventilation support, frequent chest physio and suction to support their respiratory system, and are often unable to swallow.

Recent studies indicate that approximately 1/10,000 babies worldwide are born with SMA and that SMA Type 1 accounts for approximately 60% of cases. SMA UK states that based on current incidence and prevalence rates, in the UK in 2020 approximately 68 babies were born with SMA, with SMA being the leading genetic cause of infant death. Around 1 in 40 people are a carrier of the faulty gene that causes SMA. If both parents who are carriers have a baby, there is a:

- 25% change their child will have SMA,
- 50% chance their child will be a carrier of the faulty gene, but will not have SMA,
- 25% chance their child will not have SMA and will not be a carrier.

In the UK SMA screening is not currently included in the Newborn Screening, meaning treatment is offered following genetic confirmation of the diagnosis, by which time symptoms have generally already developed suggesting that loss of motor neurones will have occurred.

Patients with SMA can present with a range of dysphagia-related and communication needs. Bulbar impairment is universal in SMA 1, and altered feeding and swallowing skills are evident in the SMA 2 population over time, with non-oral feeding being very common in both SMA 1 and SMA 2. In a recent study, patients with SMA 2 self-reported jaw problems (34%), fatigue associated with mastication (44%), choking (56%) and intelligibility problems (27%). The study found that problems with jaw, mastication and swallowing frequently occurred in combination.

In the group of younger, recently treated patients we are observing a need not only for support with eating and drinking but also with a range of speech, language and communication needs and the need for access to AAC.

Treatments

Over the past 6 years, new disease-modifying treatment options have become available. These are now available on the NHS, but are expensive (Zolgensma was labelled most expensive drug in the world in March 2021). Families may take a range of different treatment journeys. Some children will have accessed the first available treatment in the UK, Nusinersen (Spinraza[tm]), but have since received Zolgensma gene therapy. For other children, gene therapy may have been given a few weeks after diagnosis. Currently in the UK, once children have had gene therapy, no further treatments are available, and some families have sought further medical care outside the UK.

Clinicians and families are making decisions on which treatments to opt for based on a new and emerging evidence base. How treatments impact on bulbar function and on the evolution of children's communication remain very uncertain and are active areas of research interest. Current treatment options are:

- **Risdiplam** (Evrysdi [tm])(RG7916, RO7034067), a daily oral medication. Risdiplam modifies the SMN2 gene in such a way that the gene begins to produce increase amounts of SMN protein.
- **Nusinersen** (Spinraza [tm]), a synthetic antisense oligonucleotide (a small piece of man-made genetic material) that targets the SMN2 gene and enables it to produce more functional, full-length SMN protein. It is administered via an intrathecal injection (into the spine). Children receive 4 loading doses and then lifelong maintenance doses every 4 months.
- **Zolgensma** (Onasemnogene Abeparvovec [tm]), a gene replacement therapy, administered at one of the Infusion Centres across the UK. This is a single intravenous infusion. A healthy copy of the human SMN1 gene is put into a viral vector which delivers the SMN1 gene to where it is needed.

Research following one of the treatments (Nusinersen) found that some children (although not the total study cohort) did still feed orally at 24 months, and impaired bulbar function was noted to persist despite the improvement in motor function as measured on the CHOP-INTEND (Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders). This was supported by a more recent study which found that positive changes in CHOP-INTEND scores did not result in an equivalent reduction in the requirement for tube feeding.

Data related to oral feeding following Zolgensma gene therapy is being gathered, and discussion continues about the most appropriate ways to gather wider information related to bulbar function as well as speech, language, and communication.

As a group of paediatric Speech and Language Therapists working with this caseload, we have met to share our experiences, offer peer support, begin to establish consistency of practice, develop pathways, contribute to data collection, ensure there is an SLT presence at national health planning and promotion level, and disseminate information to the wider SLT professional community. The group has members from the initial trial centre, the infusion centres, neuro-muscular centres, and community teams across the UK.

Developing pathways and protocols is challenging as each centre offers different treatments and has different levels of speech and language therapy available within their neuromuscular team. Speech and Language Therapy availability varies widely, and children move between services (local hospital for acute episodes, community services for long term management, neuromuscular centres to specialist services, treatment centres for infusions). With the introduction of new treatments there is a significant increase in the demand for speech and language therapy for this complex and growing caseload. The low incidence of this condition and the very recent developments mean there is an ongoing training need amongst professionals, as well as the need to develop services to adequately meet the needs of these children once they have left the acute hospital or infusion centre. Parental expectations of treatment are high, but early information suggests that each child's response to treatment will be very different. Due to the differences in presentation following treatment children will require individual assessment, including videofluoroscopy where appropriate and ongoing monitoring and support which may fall outside an Episodes of Care model.

The challenge for the speech and language therapy profession is how we provide consistent, equitable support within already stretched services and do not miss out on the opportunity to gain vital data that will allow us to plan treatment and outcomes for these children.

The comments to the right and overleaf have been made by speech and language therapists working with this patient group, and specifically about children with SMA 1.

Babies and children may present with silent aspiration and easy access to VFSS is required. When seeing significant improvements in their child's physical development it is assumed that swallowing abilities will show similar improvements. However, there is still a lot of uncertainty regarding the impact of treatment on bulbar function. This can be emotionally very difficult for the parents and put a strain on therapeutic relationships.

The infusion centre provides the gene therapy and monitoring, but the child's therapeutic care remains with their local community teams throughout. This is explained to families and the importance of close liaison between the child's local SLT and the infusion centre SLT cannot be underestimated.

The experience of working as part of the Specialist Infusion Centre Team has highlighted the many and varied challenges faced by this group of children. Each child requires individual assessment and although there are common themes – such as impaired bulbar function and feeding, each child needs to be considered individually in terms of their speech, language, and communication development and their functional communication skills.

Although there are not high numbers of children, local services do need to be aware that this group of children are likely to require a high level of support from speech and language therapy and that this is likely to have implications for funding.

As a consequence of the complexity and overlapping areas of need, the majority of children will require the long-term involvement of their local community speech and language therapy teams with some children also needing ongoing monitoring regarding dysphagia. Other children are likely to require more specialist services such as AAC. Where a videofluoroscopy is needed this would be best offered locally.

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RECENT ADVANCES IN FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY AND OTHER NEUROMUSCULAR DISORDERS: RESEARCH AND PATIENT ENGAGEMENT AT THE JOHN WALTON MUSCULAR DYSTROPHY RESEARCH CENTRE

Neuromuscular disorders represent a diverse group of conditions that affect the motor neurons, peripheral nerves, neuromuscular junction, or muscles themselves, generally leading to progressive weakness, functional decline, and significant impact on quality of life. Over the past decade, advances in genetics, imaging, and molecular biology have transformed our understanding of these conditions, opening the door to targeted therapeutic strategies. The team at the John Walton Muscular Dystrophy Research Centre (JWMDRC) leads the Highly Specialised Service for Limb-girdle Muscular Dystrophies (LGMDs) and Distal Myopathies, based at the Newcastle-upon-Tyne Hospitals NHS Foundation Trust. The service provides an NHS England-funded national diagnostic and advisory service for patients of all ages with rare neuromuscular disorders and includes in person or remote clinic, MDT case reviews of muscle biopsies, muscle MRI and molecular results, muscle biopsy immunoanalysis, and genetic tests for these conditions. The service also provides advice on patient management and access to clinical research.

In recent years, the team has also expanded their disease area to facioscapulohumeral muscular dystrophy (FSHD), which falls outside the remit of the HSS. FSHD stands out as one of the most common inherited muscular dystrophies, affecting approximately 1 in 8,000–12,000 individuals. It is characterised by weakness of the face, shoulder girdle, and upper arms, and may progress to involve trunk and lower limb muscles. The disease shows marked clinical variability: some individuals remain only mildly affected, while others experience significant disability, including loss of ambulation. It is generally an adult-onset condition, although children might also be affected in the most severe forms.

Historically, the management of FSHD has focused on symptom management and supportive care, with no approved disease-modifying therapies. However, recent advances in understanding disease mechanisms and refining outcome measures are driving a new era of translational research and clinical trials.

At the JWMDRC in Newcastle, we are actively contributing to this momentum through integrated clinical and laboratory research.

1. Progress in Clinical Research and Trial Readiness

A key priority has been the development and validation of robust outcome measures to reliably capture disease progression. Building on large

international collaborations and recent clinical trials, imaging, functional, and patient-reported endpoints have been studied and are now being incorporated into trial protocols worldwide. Advanced quantitative muscle MRI and functional assessments are helping to identify early changes in muscle structure and function, providing sensitive markers to monitor therapeutic effects in upcoming trials, although further research is needed to refine these endpoints.

2. Clinical Care and the FSHD Registry

The JWMDRC hosts one of the largest specialist neuromuscular clinics in the UK, providing comprehensive multidisciplinary care for children and adults with FSHD. Our clinical activities integrate neuromuscular, physiotherapy, respiratory, cardiac, and genetic services, ensuring coordinated, personalised care for every patient. We have now established dedicated FSHD clinics which provide a comprehensive joint assessment with a neuromuscular consultant and physiotherapist. The clinic provides also access to a nurse specialist and care advisor, and ensure coordination of care is in place when needed. The successful clinic model also ensures access to latest clinical research information and signposting initiatives that capture patient voice, like the UK FSHD Patient Registry or patient events. The UK FSHD Patient Registry, led by the JWMDRC at Newcastle University, is an internationally recognised resource that collects clinical and genetic information from individuals with FSHD across the country. The registry facilitates patient engagement in research and clinical trials, supports epidemiological studies, and provides critical data that help shape the design and delivery of therapeutic studies. By connecting patients, researchers, and industry partners, the registry is an important component of clinical trial readiness in the UK and beyond.

3. Multi-omics and Spatial Technologies

Understanding the molecular underpinnings of FSHD is critical to developing effective therapies. At JWMDRC, we are integrating bulk and single-cell transcriptomics, spatial transcriptomics, and single nuclei RNA sequencing to characterise the cellular landscape of FSHD muscle. These studies are revealing cell-type-specific gene expression patterns and pathways disrupted in the disease, providing insights into novel therapeutic targets, improving patient stratification, and identifying new biomarkers of disease activity and progression.

4. Patient and Family Engagement

Our commitment to meaningful engagement with the FSHD community remains a cornerstone of our work. In 2024, we hosted the first patient and family information day in Newcastle, the FSHD North East & Cumbria Engagement Day. The second event has just taken place in September 2025. These events provided accessible updates on ongoing clinical and translational research, created space for open dialogue about patient needs and priorities, and facilitated stronger collaboration between clinicians, researchers and the FSHD community. Feedback from these sessions continues to shape the way we design studies and deliver care, ensuring that research remains grounded in what matters most to patients and families.

5. Looking Ahead

The coming years are expected to be transformative for FSHD. Several phase II and phase III clinical trials are in progress or planned globally, with the goal of slowing or halting disease progression. At JWMDRC, we are well-positioned to support these efforts, from trial recruitment and biomarker development to data integration that will help predict disease progression and personalise clinical care.

Conclusion

FSHD research is at a pivotal moment. Advances in genetics, imaging, and molecular profiling are converging to accelerate therapy development, offering renewed hope to patients and families. At the John Walton Muscular Dystrophy Research Centre, we remain committed to combining cutting-edge science with deep patient engagement to bring meaningful treatments closer to reality.

For more information about our research or to explore opportunities for collaboration and participation, visit https://newcastle-muscle.org.

PROFESSOR CHIARA MARINI-BETTOLO, Honorary Clinical Senior Lecturer and Consultant Neurologist, Newcastle Biomedical Research Centre/Newcastle-upon-Tyne NHS Foundation Trust

PROFESSOR GIORGIO TASCA, Clinical Professor of Neuromuscular Science, John Walton Muscular Dystrophy Research Centre

CONGENITAL MYASTHENIC SYNDROMES

Congenital myasthenic syndromes (CMS) are rare genetic disorders affecting neuromuscular junction (NMJ) transmission. There are currently 40 genes reported to be associated with CMS[1]. Whilst symptom onset at birth or infancy is most common, later onset in childhood and adult life are well recognised. Early diagnosis is important as there are specific treatments which can improve outcomes in CMS.

With the wider access to genomic testing, several new genes involved in ubiquitously expressed proteins and presenting with multi-system symptoms have been reported to cause CMS [1, 2]. In these cases the abnormal neuromuscular junction transmission is only one component of a more complex phenotype and CMS treatments will not have an impact on the non-NMJ symptoms.[3].

CMS is generally classified into pre-synaptic, synaptic, and post-synaptic subtypes. In most CMS subtypes, the pattern of inheritance is autosomal recessive except for slow channel, SNAP25, PURA and some SYT2 mutations which are dominant [3]. Post-synaptic CMS subtypes accounts for 75% of the cases from large published cohorts with most common variants in CHRNE, DOK and RAPSN. CHAT is the most

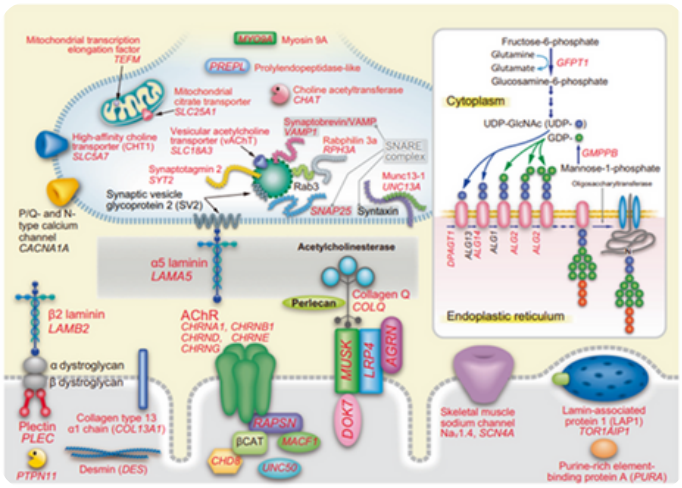

Figure 1- Ohno K et al 2025 (1)

common pre-synaptic CMS and COLQ the commonest synaptic CMS.

Clinical Features

Typical CMS presentation is of fatigable weakness predominantly affecting axial and proximal limb muscles, with ocular (ptosis, ophthalmoplegia, squint), bulbar and or respiratory involvement. Many of the CMS symptoms can overlap with genetic neuromuscular disorders. The clue towards CMS is the fatiguable element of the weakness. This may not be obvious particularly in young children and may become more obvious over time. Episodic respiratory crisis with infections is typical in some CMS subtypes; infants/children may not have significant motor or bulbar issues in between these episodes but usually have a transient motor and/or bulbar deterioration which can take days to weeks to improve.

Diagnosis

Diagnostic tests include electromyography (EMG) and Single fibre EMG (SEMG). EMG typically shows decremental responses of 10% or more on repetitive nerve stimulation (RNS) and SEMG) shows increased jitter and block EMG abnormalities may not be present in all muscles, and it is important to test in the weak muscle and repeat testing especially if initial neurophysiology is negative, particularly in young children. Genetic testing can be performed via CMS gene panel R80. CMS is also included larger panels e.g. R38, R69.

Presynaptic	
Defects in acetylcholine synthesis and recycling	*CHAT, SLC5A7, SLC18A3, PREPL*
Vesicular exocytosis	*SYT2, SNAP25B, VAMP1, MUNC13-1, RPH3A*
Axonal transport	*MYO9*
Synaptic	
Endplate AChR deficiency	*COLQ*
Defects in collagen	*COL13A1*
Defect in laminin	*LAMA5, LAMB2*
Postsynaptic	
Primary AChR deficiency	*CHRNA, CHRNB, CHRND, CHRNE*
AChR kinetics defects (with or without deficiency):	*CHRNA, CHRNB, CHRND, CHRNE*
Fast channel syndrome and	*CHRNA, CHRNB,*
Slow channel syndrome	*CHRND, CHRNE*
Escobar	*CHRNG*
Sodium channel dysfunction	*SCN4A*
AChR-clustering defects	*AGRN*
Agrin	*RAPSN*
RAPSN	*DOK7*
DOK7	*LRP4*
LRP4	*MUSK*
MUSK	*CHRND*
CHRND	
Defects in glycosylation	*ALG2, ALG14, DPAGT1, GFPT1, GMPPB*
Defects in cytoskeleton	*PLEC*
Defects in mitochondrial function	*SLC25A1*
Defects in translational regulation	*PURA*
Defects in nuclear structure	*TOR1AIP1*
Transcription regulation	*CHD8*
Defects in nAChR anchoring to cytoskeleton	*MACF1*

AChR, acetylcholine receptor.

Table 1 - Summary of CMS genes [2]

Treatment

An important aspect to CMS management is that treatment response depends upon the CMS subtype, in particular that drugs that benefit some CMS types can worsen other subtypes. In those awaiting genetic confirmation, empirical treatment must be used with caution. The common drugs used are acetylcholinesterase inhibitors (Pyridostigmine), 3,4 diaminopyridine (3,4-DAP), Salbutamol and Ephedrine. Fluoxetine and Quinidine are used in a specific CMS subtype called slow channel CMS.

Myaware is a UK charity which supports patients with myasthenia, both acquired and genetic.

Recent Developments

Several pre-clinical studies have published promising data on novel therapeutics for CMS. These include adeno associated virus (AAV) gene replacement therapy for DOK7, COLQ and CHAT; and MUSK agonist antibody therapy for DOK7 [4-6]. A Phase 1b of MUSK agonist antibody in DOK CMS in adults demonstrated good safety profile and efficacy data [9], and a further clinical trials are planned which will likely include paediatric population. Other proposed strategies aimed at improving neuromuscular transmission are either inhibiting chloride channels to lower the safety factor or by modulation of AChR activity may be applicable for treating a variety of subtypes of CMS [7, 8].

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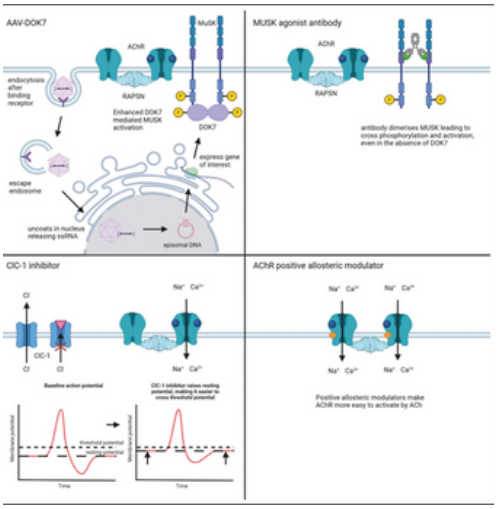

Table 2. Nonneuromuscular junction involvement in congenital myasthenia syndrome genes reported in the literature

Gene	Brain	Facial	Skeletal	Skin	Renal	Other neuromuscular	Gastrointestinal	Other
CHAT	Global developmental delay MRI brain- delayed myelination, thin corpus callosum	–	–	–	–			
SLC5A7	Global developmental delay Learning difficulties Seizures Abnormal EEG MRI Brain – atrophy	Dysmorphic features	–	–	–			
SLC18A3	Global developmental delay MRI brain- atrophy						Necrotising enterocolitis	
SNAP 25	Global developmental delay Ataxia Epilepsy	Dysmorphic features						
SLC25A1	Learning difficulties Epilepsy MRI agenesis of corpus callosum							Optic nerve hypoplasia D-2-hydroxyglutaric and L-2- hydroxyglutaric aciduria
UNC13A	MRI – thin corpus callosum	Dysmorphic features						
LAMB2		Microcoria						Congenital nephrotic syndrome
LAMA5	MRI brain- atrophy Facial tics	Dysmorphic features, myopia					Inflammatory bowel disease	
SYT2						Neuropathy		
MYORA	Global developmental delay							
PREPL	Global developmental delay				Cystinuria			
COL13A1	Mild-to-severe learning difficulties	elongated face, low-set ears, micrognathia and high-arched palate	Pectus carinatum, Moderate to severe Scoliosis with kyphosis, mild spinal rigidity					
RAPSN		Facial dysmorphism	Arthrogryposis					
Glycosylation genes	Learning difficulties					Raised creatine kinase Muscle biopsy – dystrophic changes, tubular aggregates		
PLEC				Epidermolysis bullosa		Muscular dystrophy		
PURA	Global developmental delay Epilepsy							
LRP4			Syndactyly and brachydactyly in upper and lower limbs					Congenital diaphragmatic hernia, Pulmonary hypertension

All features are not reported in all patients.

Table 2- Summary of CMS genes [2]

CMS subtype	1st line treatment	2nd line treatment	Avoid
Pre-synaptic CMS	Pyridostigmine	3,4 DAP	
COLQ CMS	Salbutamol/Ephedrine		Pyridostigmine, 3,4 DAP
AChR Deficiency CMS	Pyridostigmine	3,4 DAP, Salbutamol	
Fast channel CMS	Pyridostigmine	3,4 DAP, Salbutamol	
Slow channel CMS	Fluoxetine/Quinidine		Pyridostigmine, 3,4 DAP
RAPSN CMS	Pyridostigmine	3,4 DAP, Salbutamol	
DOK7 CMS	Salbutamol/Ephedrine		Pyridostigmine, 3,4 DAP
Glycosylation CMS	Pyridostigmine	3,4 DAP, Salbutamol	

Table 3: Current treatment options in the common CMS subtypes

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ADVANCES IN NEUROMUSCULAR DISORDERS: THE GROWING RECOGNITION OF PSYCHOSOCIAL NEEDS

In the last decade, care for some people living with neuromuscular disorders (NMDs) has changed dramatically. Advances in genetic testing, standards of care, and access to multidisciplinary clinics have improved life expectancy and physical outcomes for many. However, as clinical care has evolved, another need has become more apparent: the need to look beyond physical health and recognise the emotional and psychological impact of living with a progressive condition.

Families affected by neuromuscular disorders often describe their experience as a long journey, one that can be isolating, uncertain, and emotionally demanding. The challenges don't stop with the person diagnosed; they extend to parents, siblings, and carers, who shoulder much of the ongoing care and advocacy. While the medical side of management has advanced, psychosocial support has not kept pace, and this imbalance is now widely recognised by professionals and patient organisations alike.

A recognised gap in care

Clinicians across disciplines have spoken about their growing awareness of the psychological burden faced by families, alongside a lack of confidence in how best to support them. There is variation in access to mental health services, uncertainty about referral routes, and very few professionals with specialist knowledge of neuromuscular conditions. Many families report that when crises occur, they have little choice but to seek help privately (or be added to significant waiting lists for support), often from practitioners without relevant expertise in NMD.

This inconsistent provision has caused inequalities in access, where the type and quality of support a family receives can vary greatly depending on their location or available resources. The outcome is a direct call for more consistent well-integrated and tailored psycho-social care as a standard part of neuromuscular management.

Duchenne Muscular Dystrophy: driving change

Within this landscape, Duchenne Muscular Dystrophy (DMD) has been a leading example of how the community can come together to raise standards of care nationally. DMD Care UK is a collaborative project designed to improve clinical care for everyone living with Duchenne across the country. It brings together clinicians, researchers, and patient organisations under the leadership of Duchenne UK and the John Walton Muscular Dystrophy Research Centre at Newcastle University. The project is embedded within the North Star Network of specialist centres and is funded by Duchenne UK, Duchenne Research Fund, and Joining Jack.

DMD Care UK coordinates 13 expert working groups (with two further subgroups), each focused on a different aspect of care. One of the most important and potentially most challenging aspects is the Psychosocial Working Group (WG). Unlike other WGs, this group could not simply draw on an existing pool of experts, because there were very few professionals in the UK with experience in psychosocial care for Duchenne. This absence speaks volumes about how underdeveloped this area has been until now.

Building capacity and expertise

To address this gap, the Psychosocial WG launched an ambitious project in 2021, funded by the Duchenne Research Fund and Joining Jack. Over three years, this initiative has supported two research clinical psychologists, a neuropsychiatrist, and additional staff at Newcastle University and the National Hospital for Neurology and Neurosurgery, Queen Square, London.

The aim is to build long-term capacity and expertise in psychosocial care - something that has been missing from Duchenne services. The project is working to:

- Strengthen specialist knowledge in the psychological and neuropsychiatric aspects of Duchenne;
- Develop clearer referral pathways and guidance for clinicians;
- Gather evidence on prescribing practices for conditions such as ADHD, and anxiety;
- Identify best-practice approaches to mental health monitoring and proactive intervention.

Together, these steps aim to ensure that emotional well-being is treated as an essential part of comprehensive DMD care, not as an optional extra.

Making a real difference for families

The potential impact is significant. With better access to specialist psychological support, families

can receive help at key transition points, such as diagnosis, loss of ambulation, adolescence, and the move to adult services, rather than waiting for a crisis to occur. Earlier, more consistent support can also reduce untreated anxiety and depression, improve coping strategies, and strengthen family resilience. For clinicians, access to evidence-based guidance and clear referral routes will boost confidence and consistency throughout the UK network. For families, it will reduce uncertainty and provide greater reassurance that emotional well-being is being prioritised as part of holistic care.

Next steps: from guidelines to real-world impact

The development of psychosocial care guidelines within DMD Care UK marks an important milestone. However, as those involved in the project have stressed, guidelines are only the beginning. The next phase must focus on integrating this support into everyday NHS practice, ensuring it is available across all regions and not dependent on a family's postcode or resources. Reducing these inequalities takes time and sustained commitment from funders, commissioners, and policy-makers. But the direction of travel is clear: psychosocial care is a necessity. The DMD Care UK initiative is showing what is possible when clinical, academic, and patient communities work together toward that shared goal.

As advances in treatment continue to increase life expectancy for people with neuromuscular disorders, the focus must now shift to ensuring that those extra years are lived well, with dignity, resilience, and the proper support for both individuals and families.

CHLOE GEAGAN, Clinical Psychologist, John Walton Muscular Dystrophy Research Centre, on behalf of the wider Psychosocial working group:

Catherine Bonney-Murrell, Linda Bouquillon, Rory Conn, Janet Hoskin, Alex Johnson, Emily Reuben, Sheli Rodney, Cathy Turner, Rosaline Quinlivan, Volker Straub

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UPCOMING EVENTS

BACD Events

- 12 Nov: Acquired Brain Injury: Children's Neurorehabilitation (joint regional meeting of BACD & BACCH East Midlands - Loughborough)
- 26 Nov: Sense-ational Kids: Exploring Sensory Worlds in Neurodisability (BACD Online Annual Trainees' Meeting 2025)
- 26 Jan 2026: The Physical Health of Children with Learning Disabilities (one-day online conference organised jointly by BACD and CAIDPN)
- 4 Feb 2026: BACD East of England Regional Meeting 2026 (online)
- 10-11 Mar 2026: BACD Annual Scientific Meeting (Sheffield)

The Neurodisability Community Webinars (register for each individual webinar by clicking the links below)

- 27 Oct: TNC Webinar - Spinal Muscular Atrophy - advances in management (free)

Other Events *(NB: BACD does not endorse external events and these listings are provided for information only)*

- 17 Nov: RCPCH - How to Manage: FASD in Community Paediatric Services (London)
- 5 Dec: DSMIG Winter Meeting 2025 (London)
- 28 Jan 2026: BPNA Annual Conference 2026 (Glasgow/Online)

THE IMPACT OF WHEELCHAIR AND EQUIPMENT USE ON QUALITY OF LIFE IN CHILDREN AND YOUNG PEOPLE WITH NEUROMUSCULAR CONDITIONS: FINDINGS FROM TWO RECENT STUDIES

Background

Loss of ambulation and falls are frequent areas of concern for children and young people with neuromuscular conditions. The transition to requiring a wheelchair or other mobility aids can be distressing and difficult for both the child or young person, parents and professionals involved in their care. There are often worries about how using a wheelchair or mobility aid may impact on quality of life; however two recent studies carried out at the John Walton Muscular Dystrophy Research Centre (JWMDRC) have identified that quality of life, including social participation, can improve with appropriate provision of mobility equipment.

STUDY 1: Assessing the relationship of patient reported quality of life with functional status in a large cohort of adult patients with neuromuscular disorders

Our recent study, based on 580 survey responses from individuals with neuromuscular disorders (NMDs), highlighted the positive impact that wheelchair use can have on health-related quality of life (HR-QoL)1. HR-QoL is known to be compromised in people living with NMDs, but disease severity alone does not reliably predict HR-QoL outcomes [2].

Interestingly, we observed marked variability in HR-QoL between diagnostic groups, which could not be explained by physical disability alone [Figure 1]. For example, individuals with Duchenne Muscular Dystrophy, despite experiencing significant disability from a young age, reported the best HR-QoL amongst the diagnoses included.

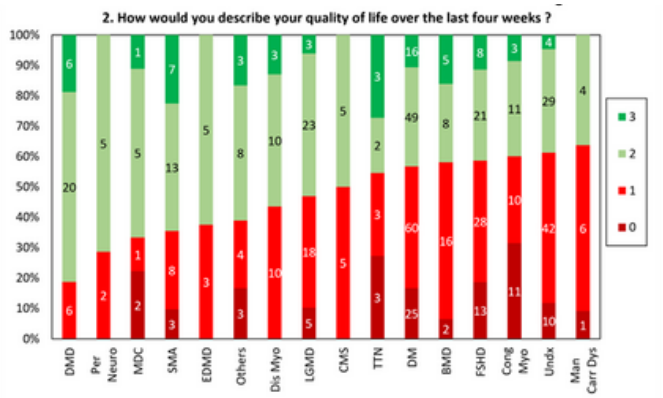


Figure 1. Distribution of responses to the question on general HR-QoL, stratified by disease category and ordered by the highest rate of positive responses. Green represents number of responses with 'good' or 'excellent' HR-QoL; red represents 'Poor' or 'Moderate' HR-QoL.

Beyond physical disability, multiple factors contribute to HR-QoL. Cognitive deficits and psychological wellbeing – such as depressive symptoms, anxiety, and low self-esteem, are key predictors of reduced HR-QoL in NMDs [3], perhaps to a greater extent than physical function [3]. Similarly, social factors, including family and spousal support [4], ability to leave the home [5], and engagement in sports and social activities [6], are all associated with improved HR-QoL. Optimising mental well-being and social participation may therefore enhance HR-QoL, even when underlying muscle weakness and disease progression cannot be altered.

Mobility Status and Wheelchair Use

A key finding of our study was that overall perceived health and HR-QoL were poorest among individuals with "mid-level" mobility – those who walk with aids or rely on a manual wheelchair [Figure 2]. This group typically represents individuals transitioning from ambulant to non-ambulant status, when walking becomes increasingly effortful, painful, and environmentally dependent.

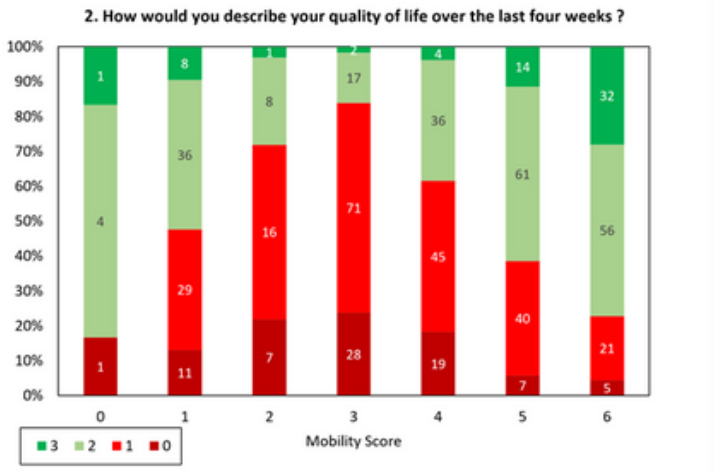


Figure 2: Stacked plots of responses to the question on general HR-QoL, stratified by SOFT mobility scores.* Shades of green represents number of responses with 'good' or 'excellent' health or HR-QoL; pink/ red represents 'Poor' or 'Moderate' health or HR-QoL.

*6 –Runs, 5 –Accelerates, 4 –Walking speed, 3 –Aids, 2 – Manual wheelchair, 1 –Powered wheelchair, 0 – Unable to sit.

Outdoor mobility is often restricted by fatigue, pain, and the risk of falls. Interestingly, HR-QoL improved again among powered wheelchair users, reaching levels comparable to those of more ambulant individuals. Although our study only included adults (aged 18 and above), this positive effect of wheelchair use was even more pronounced among younger participants (under 30 years old).

These findings are consistent with previous research demonstrating that the use of mobility devices enhances participation and activity [7] and is associated with psychological benefits and improved HR-QoL [8]. However, the transition to wheelchair use can be emotionally challenging. For many, it represents disease progression and a shift in self-image or social identity, and it can raise fears of further deconditioning or loss of strength.

Our findings highlight the importance of timely and appropriate wheelchair provision for individuals living with NMDs - to preserve independence and mitigate the negative impact on HR-QoL.

Addressing Broader Quality of Life Concerns

In our study, participants identified difficulties with future planning and increased dependence on others as the most problematic aspects of their HR-QoL. The introduction of a wheelchair, by improving independence and mobility, may help alleviate some of these concerns, particularly when outdoor mobility becomes unsafe or too physically demanding. In addition, physical symptoms such as poor sleep quality, fatigue, and pain were highlighted as the most problematic areas. These issues should form a key part of clinical discussions, including the potential role of wheelchair use in reducing fatigue and pain associated with overexertion.

STUDY 2: Is there a relationship between the ability to sit to stand and social participation in people with dysferlinopathy and what is the impact of this?: A mixed methods approach

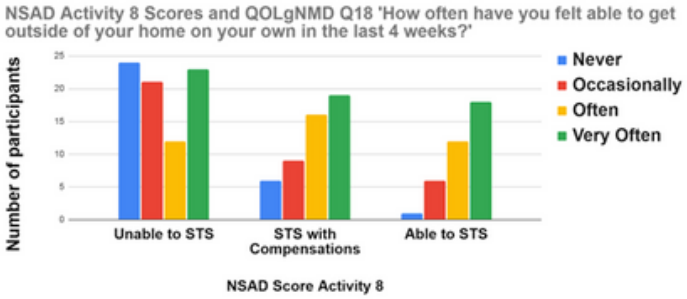
This study explored whether there is a relationship between losing the ability to stand up from a standard chair and social participation in a subtype of limb girdle muscular dystrophy (LGMD) called dysferlinopathy (LGMDR2), the impact of this and how it may be managed.

LGMDs are characterised by a slow progression of muscle weakness, mainly affecting the pelvis and/or shoulder girdle [9]. The condition presents in ambulant older teenagers/early adults, with patients reporting problems from an average of 19 years and being diagnosed around 25 years[1] [10]. Typical muscles affected include some around the hips, calf muscles, the hamstrings and quadriceps [11]. The hip abductors and flexors, however, are less affected [11 12], meaning that people with LGMDR2 often lose the ability to sit-to-stand (STS) before losing ambulation [10, 13-14]. Compensation strategies are adopted to enable STS in the absence of sufficient knee and hip muscle power [15], including the typical Gower's manoeuvre, yet all strategies are effortful, noticeable and increase the risk of falls.

If ambulation is maintained, even just around the house, people with LGMDR2 do not meet wheelchair service criteria. This means that this population often struggle to get on and off standard chairs and toilets when they are out in the community, rather than being able to remain seated in an appropriate wheelchair or benefit from a riser function, which can be provided on some powered wheelchair models. Clinically, the population report that these difficulties lead to reductions in social activities even though they are still able to mobilise.

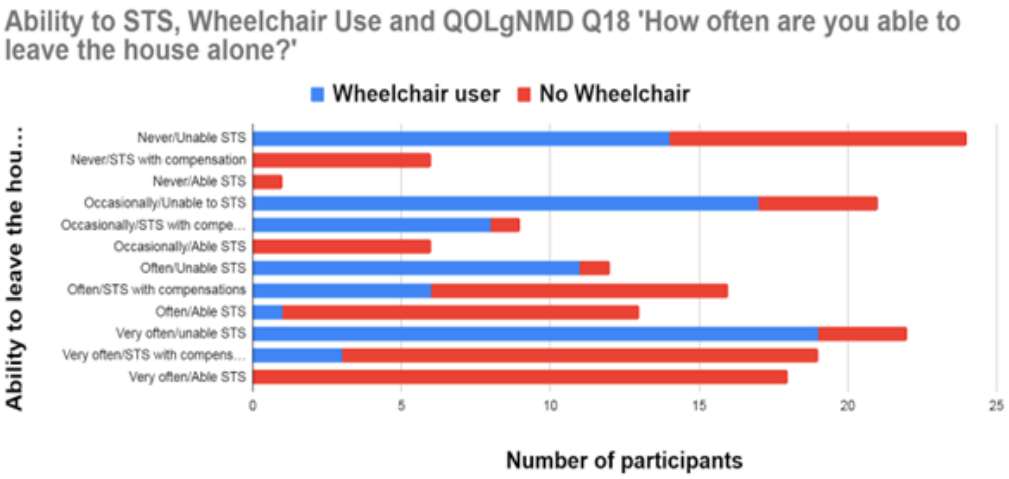
The study used baseline data from 205 ambulant and non-ambulant participants aged 10 or over with a genetically confirmed diagnosis of dysferlinopathy that had been recruited to the clinical outcome study in dysferlinopathy (COS II); a longitudinal natural history study that took place internationally between 2019-2024. The Chi square test for association (χ^2) was used to test for a relationship between clinician-reported ability to STS and participant-reported ability to complete activities requiring STS (for example getting on and off a toilet), with participant-reported responses to the social participation domains of a quality-of-life measure. In addition, a small focus group explored the impact of losing the ability to STS and any management strategies.

We found that participants who were unable to STS report difficulties getting on and off the toilet and that these difficulties restricted activities outside of the home. Participants who were unable to STS also reported difficulties getting in and out of the car and more difficulties leaving the house alone, which led to restrictions in social activities. Interestingly, some participants who were unable to STS still felt able to leave the house frequently [Figure 3].



On further analysis, we found that out of the participants who were unable to STS but able to leave the house very often, the majority had a wheelchair [Figure 4].

his suggests that the use of equipment for those unable to STS is a huge enabler; however, those who are unable to STS but remain ambulant are less likely to have access to this equipment.



Data from the focus group concurred with these findings as all participants described how equipment and support made a big difference to their social lives. Five themes were identified in relation to the impact of losing the ability to STS, with equipment and support being the most prevalent followed by coping/ compensation strategies. Those with no access to equipment described how difficulties with STS had a social and emotional toll on daily activities.

We summarised that the ability to STS does impact on social participation, but the use of equipment and support can enable continued social participation and better quality of life. Clinically this means that early referrals to wheelchair and equipment services, regardless of ambulatory status, are therefore recommended. In addition, further exploration of equipment prescription and support is required to evidence what equipment is most helpful and when to inform standards of care and wheelchair/equipment providers.

Conclusions

Evidence from both studies shows that equipment use can enhance, not hinder, quality of life and social participation. In some cases, early equipment provision may offer the greatest benefits. Clinicians should engage individuals with NMDs in open, proactive discussions about the use of equipment to help them navigate the progressive journey that they face.

Thank you to all patients who participated in the studies. Funding for the COS study was by the Jain Foundation.

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CURE MYOTONIC DYSTROPHY UK CHARITY (CUREDM)

What is Myotonic Dystrophy?

Myotonic Dystrophy Type 1 (DM1) is a life-limiting, multi-systemic neuromuscular disease, which affects whole families. It is caused by a CTG triplet repeat on the DMPK gene, and inherited dominantly, giving each pregnancy a 50% genetic chance of being affected. In practice we support families where multiple children are severely affected, so this statistic should not be interpreted that if you have a child with DM the next one won't. Severity of the parent is not a factor (mildly affected parents can have severely affected babies). Interestingly, DM1 worsens with each generation due to 'anticipation', meaning that very often, families are not aware of the condition until a severely affected baby is born with Congenital DM (CDM).

Everybody will have several DMPK CTG repeats; however, once this number hits 37 it is deemed 'unstable'. People with 37-49 repeats are diagnosed as 'pre-mutation' and whilst they themselves are un-symptomatic, due to anticipation, following generations will inherit more severe forms of the condition. This is how we continue to get 'new families' with the disease. Greater than 50 CTG repeats is classed as a positive genetic result for DM1. DM1 is not sex-linked; however the congenital form holds a maternal bias, with approximately 13% of CDM cases being paternally inherited.

DM1 can be split into categories depending on when symptoms appear:

- Congenital DM: symptomatic at birth or within the first month of life (with repeats often numbering over 1,000)
- Early childhood onset: symptoms before 10 yrs
- Late childhood onset: symptoms appearing between 10-18 yrs
- Adult onset: symptoms beginning 18-40 yrs
- Late adult onset: symptoms beginning after the age of 40 yrs

Previously reported as affecting 1:8,000 people worldwide, it is now believed that DM1 affects as many as 1:2,100. This condition is extremely under-diagnosed and, despite being the most common form of adult-onset muscular dystrophy (arguably the most common form of MD overall), it is often not fully understood within the clinical community; therefore many with DM continue to lack the necessary care and treatments.

Who are CureDM?

'Cure Myotonic Dystrophy' is a charity for people in the UK living with Myotonic Dystrophy (Types 1 or 2), their families and caregivers. The aims of the charity are to raise awareness, facilitate research and support families. Run by trustees living with varying stages of the condition themselves or within their own families, it provides a fountain of knowledge to patients, medical professionals, and researchers/ industry working in the field of DM research.

With a specialist interest in congenital and early childhood onset, CureDM advocates for families and patients in areas often overlooked, raising the voice of the UK DM community which is often unheard. Type 2 DM has a different genetic cause, but is very similar in symptoms, and patients living with Type 2 are welcomed by the charity and included in events and support. Families living with all types and stages of DM can become lonely and isolated due to multiple people being affected. CureDM is succeeding in bringing the community together for peer support, as well as improving the medical and social care which is needed to attain the best possible quality of life.

How it Began

We (Mum Emma-Jayne and stepdad Peter Ashley) had been actively searching for an answer for our son's deteriorating health for almost 15 years, when we received the diagnosis of Congenital Myotonic Dystrophy. Hundreds of appointments over the years led to over 20 separate 'diagnoses', yet the clinical teams had failed to connect symptoms and reach the accurate diagnosis, which is simple to spot when you know what to look for. Dregan inherited CDM from his biological father, which at the time was thought to be only a 1% chance (now shown to be around 13%), who was later diagnosed with late childhood onset DM and passed away from a DM-related sudden cardiac event aged 39. The condition was unknown to the family until Dregan

was diagnosed, but once identified, answered many years of questions around his increasing difficulties. Receiving the diagnosis was, therefore, a relief at first, until we realised that we had been fighting through what was his 'best years' and now we were told he was unlikely to survive into adulthood. We are very proactive people, and thought 'we can't just sit back and let other families go through this'. So much precious time had been wasted in those uneventful hospital appointments, and that seed of thought grew into the CureDM we know now. The knowledge we have built in this time has helped us to break down barriers, and whilst Dregan has had some horrible life-threatening periods of illness over the years, he is now 26 years old and has a great quality of life.

What Do We Do?

The charity's three aims are Raising Awareness, Facilitating Research and Supporting Patients and Families. It would be impossible to go through everything we do in a few short words, so you can see more on our website and our main Social Media Page.

CureDM were delighted to be co-founders of the International Myotonic Dystrophy Awareness Day five years ago. This awareness day is held on 15th September every year, Dregan's birthday! We will always be enormously proud of this day, which has seen over 60 myotonic dystrophy-focused organisations come together, including worldwide Charities, clinics, research labs, and pharmaceutical companies. This will be Dregan's legacy, and we love celebrating it on his birthday every year, after all, he is the reason we do this. We encourage you to add your voice, and join the Global Alliance. CureDM marks the day by asking buildings to light up green every year. This year we also produced an easy to understand video for schoolchildren, which was presented at primary schools who joined in to support the day, and collaborated on an NHS Myotonic Dystrophy Animation. This encourages DM patients and families to raise awareness their way within THEIR communities. Social media is a hive of activity, and it is tremendous to see the worldwide participation.

Raising awareness (both in the community and clinical practice) is vital not only to support patients with their day-to-day living, but also to share information around recent advances in research, and the many upcoming clinical trials which are being developed for Myotonic Dystrophy. Preparing and helping the community to learn about and access clinical trials will pave the way to having new treatments approved and available, hopefully in the very near future.

Advances in DM Research

We are at a pivotal point for research in Myotonic Dystrophy, with many worldwide pharmaceutical companies, specialists, and researchers working together to develop the first systemic treatments. There are currently 6 clinical trials in the UK. Avidity (phase III), Dyne (phase III) and Pepgen (Phase I/II) are intravenous gene therapies, using different delivery vectors and technologies to target the cells in the most affected parts of the body. Arthex (Phase I/II) have a dual-action injected drug in a trial and are aiming to use the same one to also treat DM2 and Congenital DM1. Lupin are trialling their prolonged release Mexiletine for Myotonia in DM1 and DM2 (standard mexiletine is currently used off-license in the UK). There is also AMO Pharma, which completed their Newcastle trial on adults living with Congenital and juvenile-onset DM1 in 2018. They currently have the ongoing open label extension (phase II/III) for CDM in the USA, Canada, Australia and New Zealand, with plans to start an adult (phase III) study soon. AMO use Tideglusib, a GSK-3β inhibitor, to reach the whole body including the brain, taken simply as a daily drink. Other companies are planning clinical trials in the UK for next year.

There is also much work ongoing for Quality-of-Life improvements. Increased knowledge of this complex and heterogeneous condition is leading to improvements in clinical care, with many consensus-based care guidelines available now. More are being produced, including international insights and recommendations for diagnosing, understanding and treating gastrointestinal symptoms, GI being recognised as the largest area of unmet clinical need in DM.

CureDM keep an ear close to the ground and have developed a medication and research snapshot - a horizon scan. This includes all organisations that we

have found to be publicly working on potential treatments for DM, together with details of their work and links to further information. The number of organisations involved is growing exponentially, with over 50 on our snapshot. For the first time, there is real hope in the DM community for not one, but a choice of systemic treatments, together with rapidly improving clinical knowledge, care and support.

Support and Events

And of course, we mustn't forget the reason we do all this, and that is the patients themselves and their families. The patient support we provide can be face to face/1:1 disease-specific support, information for schools to help education, helping signpost to support for DM-specific health and social care, support applying for adaptations, mobility aids and benefits - the list goes on. We have two regular get-togethers each year: our large Families Day event, which is always on the Saturday around 25th July, and our smaller Christmas get-togethers.

The Families Day events have become extremely popular, with 166 attendees joining us from 41 family groups at our most recent event held at Chester Zoo. Over the years these events have gone from a handful of people meeting in a park for a picnic, to huge 2-day events with hundreds of people from all over the UK. Held in a different geographical area every year, we aim to reach as many people as we can because this peer support is so important. The Charity facilitates, organises and pays for the attendees to get together: personal finances should not be a barrier to attending. During the event we provide information on current research, trials and distribute information and care guidelines. This year 41% were new attendees, and the community reach is increasing all the time. You can see more about our charity, what we do and the events we provide

on this video (filmed at our 2023 Families Event at the Calvert Trust).

Recent feedback from the events includes the following words from Becky, who lives with DM herself, and has a young son with CDM:

> "CureDM are a continued beacon of light and hope, so knowledgeable about not only the condition but the research and support out there. We have been lucky enough to go on two family fun days with CureDM, The Polar Express and Chester Zoo weekend. These events allow us to make such wonderful family memories, meet other families and individuals that really understand your circumstances and also speak to other members of the charity. It really does feel like such a supportive and wonderful community of individuals, and it really has brought light to us in some of our darkest days."

Finally, none of the work we do would be possible without the partnerships, sponsorships, fundraising, and support we get from people like you. When presenting, we always say "please tell one other person about Myotonic Dystrophy and the work we do as a charity. That makes it two more people who are aware and who might just be able to make a difference to somebody's life", and we would like to share that sentiment with you. Thank you!

EMMA-JAYNE ASHLEY Founding Trustee, CureDM
curedm@outlook.com

Reference 1: Johnson NE, et al. Neurology. 2021;96:7.

DYLAN'S MUSCLE DAYS AND FINDING STRENGTH TOGETHER

Joshua was originally one of our foster children and diagnosed with Duchenne muscular dystrophy (DMD) aged 21 months. We decided to adopt him and make as many magical memories as possible. Like many parents, I have searched for ways to help people understand what Duchenne is, and more importantly, what life looks like for a child living with it. Each year on his diagnosis day anniversary I try to do something positive to raise awareness. We've had a 'Jingle in June' 2nd Christmas party, a Facebook launch of 'Journey With Joshua', and a fun run challenge. This year I decided to publish 'Dylan's Muscle Days'.

I wanted to write something that children could relate to - a story that gently explains why Dylan might get tired more quickly, why he sometimes needs a wheelchair, or why his muscles work differently. My hope was that the book would help family, classmates, friends, and even teachers see past the condition and just see Dylan for who he is: a bright, funny, and adventurous boy.

Above: 'Duchenne mums' enjoy themselves at one of their weekends away

Karen Jessop with her book Dylan's Muscle Days

But as the years have gone on, I also learned that support isn't just about raising awareness—it's about looking after each other as parents too. Life with Duchenne can feel relentless, and sometimes mums like me don't get the chance to breathe, let alone rest. That's why I started organising weekends away for Duchenne mums. These weekends are so much more than a break. They're a chance to sit with women who just "get it," without having to explain. We've taken a mini cruise to Amsterdam, stayed in a Welsh Castle, and a manor house in Stafford-shire, and even sipped cocktails at the Savoy after a night at the theatre. We laugh, we cry, we share experiences and stories, and most of all, we remind each other that we're not alone on this journey. Both the book and the mums' weekends grew from the same place in my heart - the need to connect, to share, and to create spaces where Duchenne families can feel seen and supported.

Dylan's Muscle Days can be found and ordered on Amazon.

KAREN JESSOP, author and DMD parent advocate

UNDERSTANDING HEMIPLEGIA: SUPPORTING FAMILIES FROM DIAGNOSIS

Background

Cerebral palsy affects approximately 1 in every 400 children in the UK, with an overall prevalence of 2-3.5 per 1,000 live births [NHS England, 2025]. Hemiplegia (also referred to as unilateral cerebral palsy) is the most common type of cerebral palsy and causes weakness or paralysis on one side of the body. Around 80% of hemiplegia cases are a form of cerebral palsy present from birth, while around 20% are acquired later. Hemiplegia frequently impacts movement, coordination, and everyday tasks such as dressing, eating, and playing. Early intervention for infants and children with hemiplegia is critical to ensuring they reach their full potential.

Evidence-based intervention can improve function, particularly for upper limb skills, but it often requires more time than health services can provide. Parents are increasingly asked to deliver therapy at home, yet many report feeling underprepared and uncertain where to start. Without clear guidance, families can experience confusion, stress, and delayed intervention.

However, it is not just a lack of time within health services that creates challenges. Some health professionals report limited knowledge and skills regarding evidence-based interventions and confidence when coaching families to deliver intervention [Massey et al 2025a]. This may be linked to the broad caseloads faced by community therapists, who often work across a diverse range of conditions [Atun Einy et al 2019; Scott et al 2023]. Supporting community-based therapists, particularly those in isolated roles with diverse caseloads, is crucial for maintaining fidelity and effective delivery [Massey et al 2025b].

To address these gaps, the Understanding Hemiplegia guide – your guide to supporting your child with hemiplegia, with a focus on strategies to improve hand and arm use – was developed as a co-designed, evidence-based resource to support families from diagnosis onward. The guide provides practical advice, emotional reassurance, and strategies to enable parents to confidently support their child's rehabilitation at home.

Co-design and Collaborative Development

Funded by the National Institute for Health Research [NIHR 300556], this guide was developed as part of a doctoral study using the Experience-based Co-Design (EBCD) method [Bate & Robert, 2006]. The project involved parents with lived experience and clinicians, facilitated by a clinician researcher. This partnership ensured the resource reflected real-world experiences and needs. The process included:

- Interviews with parents to understand their challenges and information needs.
- Co-design workshops to set shared priorities.
- Iterative co-design workgroups to develop content and format, review drafts and incorporate feedback.
- Celebration event

This inclusive and collaborative method resulted in a guide that is both informative and user-friendly, balancing clinical knowledge with lived experience.

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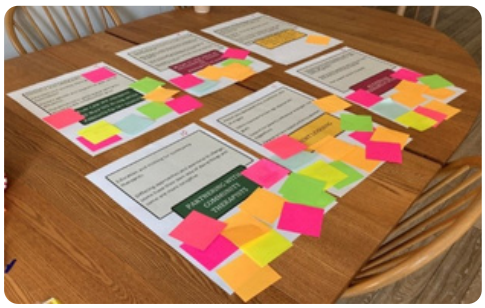


Top Photo: Group of parents and therapists in a co-design workshop; bottom photo: priority-setting activity

The Understanding Hemiplegia Guide

The guide offers families a comprehensive, accessible resource for supporting their child's rehabilitation:

- Information about hemiplegia – Clear explanations of what hemiplegia is and how it can affect a child's movement, coordination, and daily activities.
- Practical guidance for home-based therapy – Step-by-step advice to support upper limb development, aligned with interventions introduced in clinical settings.

- Emotional support and reassurance – Insights into common feelings following diagnosis, helping parents feel empowered and confident.
- Evidence-based interventions – Summaries of the research behind recommended strategies, supporting parents in understanding why and how therapy works.
- Signposting to organisations that provide additional family support.
- An easy-to-read guide to health professionals families may meet during their child's rehabilitation journey, plus a glossary of terms they may encounter.

Parents report that the guide provides crucial clarity at a challenging time:

"After my child's diagnosis, I didn't know where to turn or what to do. This guide would have given me practical steps and reassurance, making such a difference."

"It will help countless families gain knowledge, confidence, and hope, providing tools to start therapy correctly from the beginning."

While this resource was originally intended for parents, clinicians also recognise its value:

"The guide supports parents and professionals alike, helping us understand how to provide clear, timely guidance and strengthen the partnership in care."

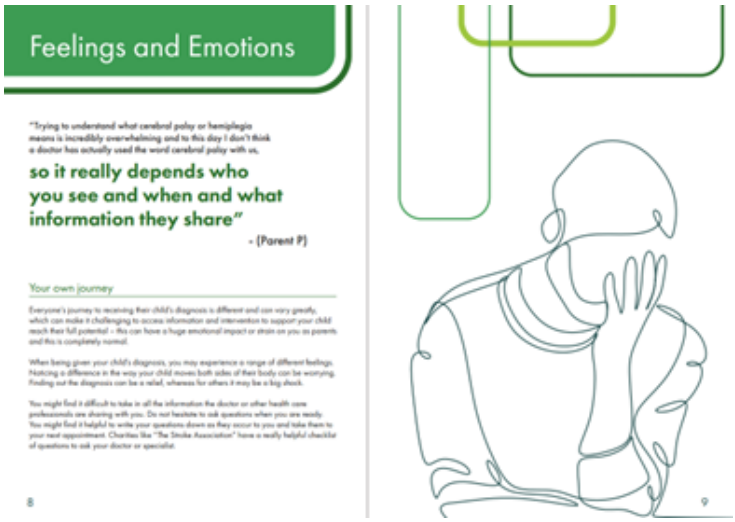
Evaluation and Feedback

Initial feedback from families and clinicians has been overwhelmingly positive:

- Parents reported feeling better informed, reassured, and capable of supporting therapy at home.
- Clinicians noted that the guide helps them communicate clearly with families, reducing confusion and increasing engagement.
- The co-design process itself was praised for creating a safe, inclusive environment where all perspectives were valued.

Recent Launch Event

On Saturday 4 October (the closest weekend to World Cerebral Palsy Day), families of children with hemiplegia joined professionals and representatives of four national charities to celebrate the launch of the Understanding Hemiplegia guide. The day combined learning, support, and community, with children enjoying toys and games while parents heard about the guide's development and the additional support available from partner organisations such as HemiHelp.



The event was co-led by Katrina Galvin, a parent with lived experience who played a pivotal role in co-designing the guide. She shared her family's journey, including her son's diagnosis, and emphasised the importance of clear, practical resources for families at such a critical time.

Left photo Katrina Galvin (parent with lived experience) and Jill Massey with the Guide to Understanding Hemiplegia

Right photo: Katrina Galvin presenting at the launch event

This event marked not only the formal introduction of the guide but also a celebration of collaboration – between families, clinicians, and charities – in creating a resource that truly reflects the needs of those it is designed to support.

Next Steps

Having now launched the guide, future plans for the guide include:

- A national roll-out to community therapy teams and rehabilitation services;
- Integration into policy and practice guidance via engagement with key stakeholders and organisations;
- Evaluation of long-term impact.

By prioritising co-design and accessible support, the Understanding Hemiplegia guide represents an important step toward empowering families, strengthening clinician partnerships, and improving outcomes for children with hemiplegia.

Get Involved and Stay Connected

We warmly welcome contact from anyone interested in the Understanding Hemiplegia guide and in helping us share it widely. On the back of the guide, you will find a QR code that links to a short form.

By scanning the QR code (right), families, clinicians, and organisations can let us know:

- How the guide is being used.
- Who it is reaching.
- What difference it is making.

This information will help us gauge the wider impact of the guide, strengthen our evaluation, and ensure it continues to meet the needs of families and professionals.

If you would like to connect with us, share your experience, or help spread the word, we'd love to hear from you. Together, we can build stronger support for families navigating hemiplegia.

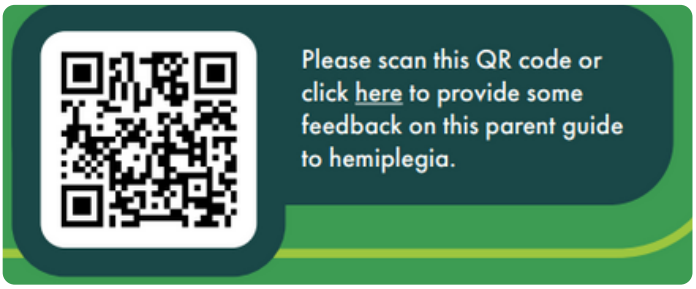
Find out more

The new Understanding Hemiplegia guide is now available for families to download. For further information or future collaborations please contact jill.massey6@nhs.net or jill.massey@kcl.ac.uk.

JILL MASSEY, Consultant Clinician Researcher, Evelina London Children's Hospital / Cerebral Palsy Alliance, Sydney, Australia

Acknowledgements

Sincere thanks to all the parents and therapists who generously shared their experiences and invested their time and energy in the development of this guide. Particular thanks to Katrina Galvin and Natalie Sadler (parents with lived experience) for not only their involvement in the project, but also for their ongoing commitment to sharing this work with families and professionals through presentations, publications, and by sharing their personal stories.



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TRAINEE UPDATE: INTRODUCING YOUR NEW BACD TRAINEE REPRESENTATIVES

Our names are Beth and Charlotte, and we are delighted to introduce ourselves as your new BACD Trainee Representatives. Although we have been floating around since the summer, we officially took over the role in September!

Charlotte is an ST7 Neurodisability GRID trainee based in London, and Beth is an ST5 Community Child Health GRID trainee in Severn. We both feel very privileged to be taking on this role and are honoured to represent trainees across the UK who are working in the field of childhood disability. We are passionate about making sure trainee voices are heard and look forward to working with colleagues to strengthen and support our community.

Stay Connected

If you haven't already, please do join the National Paediatric Neurodisability Trainee Network WhatsApp group – it's a great way to connect with peers.
We now also have a dedicated BACD Trainees Instagram account (@bacdis_org), where we share trainee news, research, educational opportunities, events, and more. Please follow us and encourage colleagues to do the same – it's the easiest way to stay informed and connected.

What's Coming Up?

In the next few months, one of our main priorities is planning the Annual Trainee Meeting. This year's theme, chosen by trainees, is "Sense-ational Kids – Exploring Sensory Worlds in Neurodisability". The meeting will be held online on 26th November 2025. You can sign up via the BACD website.

The final programme will be circulated ahead of the event, but we hope to include: Sensory Processing Disorder, Technology in Sensory Loss, a visually impaired teacher's perspective, and a lived experience talk from a young person with sensory loss.

Looking Ahead

We are also working with the team on the upcoming BACD Annual Scientific Meeting in Sheffield in March 2026. This promises to be a fantastic event with high-quality academic content and brilliant networking opportunities. We really hope to see many of you there.
We would love to hear from as many of you as possible – whether to share ideas, raise concerns, or get involved with any of the projects. Together, we can continue to strengthen the trainee voice within BACD and build a supportive and vibrant community.

Warm regards,

BETH AND CHARLOTTE
Your BACD Trainee Representatives
charlotte.sayer2@nhs.net
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SUPPORTING ORAL HEALTH IN AUTISTIC CHILDREN: INTRODUCING TOOTHPASTE

Following its introduction at the recent BACCH ASM, we're delighted to share toothPASTE, a free online resource to support the oral health of autistic children and their families (www.autismtoothcare.com). Developed in collaboration with autistic children, families, and professionals, toothPASTE provides practical, autism-informed guidance around toothbrushing, eating and drinking, and visiting the dentist.

Colleagues who have already used ToothPASTE describe it as accessible, family-centred, and complementary to existing work:

"I really think this is going to be such a useful resource – this is an area of health where it's so important to try and build good healthy behaviours early on, but often children with autism may not be able to access dental care until the problems are severe or are an emergency. Being able to access practical advice, share experiences, watch videos created by professionals and parents who have really understood the challenges will make such a difference….I've already had 2 phone consultations with parents where the

issue of toothbrushing has come up – so I've given them the website and will put it in my clinic letter too." - **Dr Alexandra Hardisty, Paediatric consultant, Harrogate District Hospital**

Why it matters

Oral health challenges remain a significant public health issue: one in four children in England has tooth decay by age five, rising to almost half in more deprived areas [1]. Similar figures are seen among children with Special Educational Needs and Disabilities (SEND). Tooth decay is the leading cause of hospital admissions for children, costing the NHS over £50 million annually in preventable extractions [2].

For autistic children, barriers such as sensory sensitivities, communication differences, and rigid routines can make oral health care particularly difficult [3]. Research shows autistic children are less likely to attend for routine care and receive preventive treatment, and are twice as likely to need treatment under general anaesthetic [4].

However, the impact goes far beyond the health system. Tooth decay can cause severe pain, difficulty eating and speaking, disrupted sleep, and recurrent infections [1]. These problems often lead to missed days at nursery or school, and widening educational inequalities [5]. Families also experience emotional and financial strain, with parents frequently taking time off work for appointments or procedures [1]. Although tooth decay is almost entirely preventable through regular brushing with fluoride toothpaste, reduced sugar intake, and regular dental visits, these habits can be particularly hard to establish for children with additional needs.

toothPASTE aimed to co-develop an intervention that enables parents of young autistic children to feel confident and empowered to look after their child's oral health.

The Research Behind toothPASTE

From the outset, families were involved to ensure the resources were practical, respectful, and worked for them. A panel of six parents with lived experience of autism guided the research throughout.

Using a complex intervention approach, the team conducted a scoping review and interviews with autistic children (n=10), families (n=14), and professionals (n=29), including community paediatricians, dietitians, school SENCOs, and speech and language therapists. Families described toothbrushing as a daily "battle" with oral health advice being too generic. The research found that many early years professionals, including paediatricians, are asked by families about oral health but lack the confidence and training to engage with these conversations. Key themes reported by families and autistic children included sensory sensitivities, the emotional strain of daily toothbrushing, while helpful strategies involved finding toothbrushes that suited sensory needs and using visual supports to build consistent routines around brushing, eating and drinking, and dental visits.

Findings were refined through four co-design workshops with families and professionals (across health education and third sector). This process directly shaped the design of the toothPASTE website, ensuring it reflected real-life experiences and multidisciplinary input.

Implications for Practice

Community paediatricians and allied health professionals may find that oral health concerns naturally emerge during consultations. However, with the breadth of competing priorities, such discussions can be difficult to explore in detail, particularly when families are already navigating multiple care pathways. The toothPASTE website provides a reliable and accessible starting point for families to engage with at their own pace. Available in eleven languages, it offers downloadable leaflets, videos, and visual resources designed to support both professionals and families.

Clinicians can incorporate toothPASTE into their routine practice in several ways, such as including the website link in information packs provided when a child receives an autism diagnosis or is added to a waiting list for assessment, displaying posters in waiting rooms for families awaiting appointments (available on request), sharing the resource with multidisciplinary team members including speech and language therapists, occupational therapists, school nurses, and other professionals involved in the child's care, or by adding the website link to clinic letters where appropriate.

Future Directions

The next phase of the research will focus on refining toothPASTE through user testing to better understand how parents and carers engage with the website in everyday contexts. Approximately 30 parents and carers, primarily from West Yorkshire, will be invited to take part. This phase aims to explore their experiences of using toothPASTE, identify aspects they find helpful or challenging, and gather insights to inform further improvements to the website's design and content. The research team will also examine whether toothPASTE is beneficial for families of older autistic children (up to the end of primary school) and for those caring for children with other Special Educational Needs and Disabilities (SEND).

The team welcomes contact from those interested in the study and encourages feedback from clinicians on how toothPASTE is used in practice and what further support may help its implementation.

Accessing the resource
Visit www.autismtoothcare.com
A poster featuring a QR code and link is also available on request.

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Competing Interests

None declared.

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CDC HEALTH POLICY UPDATE

Plenty of updates since the last newsletter, and plenty more expected over the coming months, including the NHS Workforce Plan and Autumn Budget, with indications the government may lift the two-child benefit cap.

The Schools White Paper has now been delayed to early next year. While a further delay will clearly frustrate many, there is now an opportunity for the sector to work together to shape and articulate a plan that addresses short-term challenges and offers a coherent vision for the future of the SEND system. Read Bridget Phillipson's letter to the Education Select Committee explaining the delay here, and the CDC response to the news here.

Child Health Workforce Alliance

The Child Health Workforce Alliance, launched in response to the crisis in children's health services, brings together leading twenty-five leading organisations across health, education and social care to ensure that the currently underfunded and overlooked child health workforce receives the attention and strategic focus in policy making needed to help realise the government's ambitions for children.

A key policy briefing published by the Alliance outlines the serious challenges faced by the child health workforce and highlights the importance of a whole-system approach to workforce planning. The policy briefing sets out a series of cross-sector solutions including investment in workforce planning, improved career pathways, and stronger integration across services.

Read the policy briefing here.
Read the NCB article here.

SEND system reform: key insights from our roundtables and principles for change

The government is preparing to set out its proposals to reform the system which supports disabled children and those with special educational needs in the upcoming Schools White Paper.

To help think about what change should look like, the Council for Disabled Children hosted three roundtable workshops in the summer, in partnership with the Association of Directors of Children's Services, National Association of Special Schools, Royal College of Paediatrics and Child Health, Alliance for Youth Justice and the Michael Sieff Foundation.

The SEND system reform roundtables underscored the urgent need for change and the potential of a new approach centred on collaboration, early intervention, co-production, shared accountability, and coordinated funding. Further detail can be found in the links below.

Individual roundtables: Special Schools, Youth Justice, Health
Summary of roundtables discussions
NCB article

Supporting the mental health of children with social care involvement

Despite evidence that one in two young people with a social worker experience mental health difficulties, and higher referral rates, research shows that those with social care involvement are three times more likely to not receive the Child and Adolescent Mental Health Services (CAMHS) they require.

Research from the NIHR-funded COACHES project found that young people with social care involvement were frequently rejected by mental health services because their needs were viewed as too 'complex' or caused by their current situation. It was often cited that 'social' factors were to blame caused by instability at home, but it was unclear what this meant. Nine recommendations have been suggested to improve services, including a review of 'social stability' as the basis for mental health access, better integration of child and adult mental health services, and automatic access to a mental health assessment for any child in contact with social care. Please see the links for more detail.
Read the full NCB article here
Read the research report here

Designated Clinical Officer for SEND Handbook

The new DCO handbook has been developed by the Council for Disabled Children in partnership with DCOs and ICB Heads of SEND and NHS England. It contains the key information all ICB Board Executive leads, and senior leaders need

to know about the critical ole Designated Clinical Officers (DCOs) for SEND play in supporting ICBs to fulfil their statutory requirements for children and young people aged 0-25 with SEND.

CDC and NHSE are grateful for the contributions and sharing of expertise of everyone who has been involved. We recognise that there are different arrangements across the country and hope that the new handbook is an opportunity to consolidate learning from experience and best practice including further detail of the intended purpose and impact of the DCO role.

SEND Quality Assurance Framework

The NHS England South East Mental Health, Learning Disability, Autism and SEND programme, in partnership with colleagues from the North East and Yorkshire region, have developed and launched the SEND Quality Assurance Framework (QAF) which aims to support ICBs in delivering SEND services. The QAF was co-produced through engagement with the Council for Disabled Children (CDC), health professionals, and regional parent carer leads as well as children and young people and has been designed to ensure systems understand their overall position at a place and system level to inform improvement plans. The information gathered through the framework provides the ICB with assurance and can be used to develop areas that are either location specific or that can be considered as a system wide priority.
The framework and guidance can be found here.

BACD and the Castang Foundation are pleased to announce Round 4 of the BACD-Castang Fellowship Programme for 2026 for UK-based candidates from a relevant professional discipline (i.e. within neurodisability-related health care, education, social care, or research).

Pipeline Fellowship for candidates who want to secure a funded PhD/MD award:

- For applicants at different stages of their clinical/practice career, ranging from relatively newly qualified to well-established clinicians and practitioners, or applicants from an academic, teaching, or research background (i.e. non-clinicians / non-practitioners).
- For applicants who want to secure a pre-doctoral funding award or equivalent (e.g. an NIHR Pre-doctoral Clinical and Practitioner Academic Fellowship).
- For candidates who want to secure a doctoral funding award or equivalent (e.g. funded MD).

Advanced Fellowship (for research-experienced candidates who want to secure their next award):

- For applicants who are close to finishing their PhD or MD (i.e. submitting within the next 6 months approximately) and who want to secure their next research funding award.
- For post-doctoral applicants in health or social care settings or universities or other settings who want to secure their next research funding award.
- For research-experienced applicants (with or without a PhD or MD) with strong early-career researcher CVs who want to secure their next research funding award.
- For established researchers, or clinical/practitioner academics, or educators with experience in leading research who want to secure their next funding award and/or re-work funding applications that have not yet been successful.

The Fellowship programme includes:

- Individual mentoring from a member or collaborator of the BACD Strategic Research Group (SRG)
- Interactive learning workshops about key childhood neurodisability research topics, methodologies, and funding streams
- An intensive grant sprint experience, where candidates iteratively present and develop their grant applications with support from expert clinicians, practitioners, methodologists, and BACD-Castang Fellowship peers
- Multidisciplinary networking with experienced childhood neurodisability researchers and collaboration with BACD-Castang Fellowship peers.
- National opportunities to disseminate Fellowship outputs and raise their professional profile within the childhood neurodisability research community.

Fellowships include financial support to cover attendance at in-person sessions in Sheffield on 9th -10th March 2026, and the BACD annual scientific meeting on Wednesday 11th March 2026. This includes all travel, accommodation and subsistence up to the value of £1,000 for each successful candidate.

Full guidance on the Fellowship programme, including aims, output, programme content, eligibility and assessment criteria can be found here.

"The Fellowship has been instrumental in my early career research journey so far. I have made a network of peer connections that has extended into all areas of my work. The experiences in the Fellowship have supported me with my academic studies. Check points in the programme have embedded a time frame for me to complete reflections and specific actions. A fully immersive learning experience for me as a compete novice. Would highly recommend." - *Round 3 Fellow*

How to apply: Applications should be made on the BACD-Castang Fellowship application form which consists of a short CV and a covering letter which should be emailed to kelly.robinson@rcpch.ac.uk by 1700 on Thursday 11 December 2025.

Candidates are required to attend the in-person sessions in Sheffield on 9th-11th March 2026 inclusive. Please do not apply if you are not available to attend these dates in Sheffield in person.

Our goal is to identify and encourage people from diverse professional backgrounds with the potential, intention, and commitment to lead UK research in childhood neurodisability. We aim to be inclusive and to grow the childhood neurodisability research community. Please do get in touch if you're not sure about your eligibility or whether one of these awards is for you. Applicants are welcome to contact Jennifer.McAnuff1@nhs.net or Jeremy.Parr@ncl.ac.uk with any queries.

BACD ANNUAL SCIENTIFIC MEETING 2026

Tuesday 10th – Wednesday 11th March, Sheffield

BACD is pleased to present the programme for next year's inaugural two-day annual scientific meeting, to be held in Sheffield. Please note that the first day will be for in-person delegates only, and the second day will be a hybrid conference.

ABSTRACT SUBMISSION

BACD is calling for abstracts for presentation at the 2026 Annual Scientific Meeting in the following categories:

- Original Research or Quality Improvement
- ·Novel Multi-Disciplinary Team Working - showcasing innovative MDT working practices and the impact of different roles on clinical practice and for patients and their families

Why should you submit an abstract to the BACD Annual Scientific Meeting 2026?

- Opportunity to share your research or innovative working practices at the UK's leading multi and interdisciplinary scientific conference about childhood disability
- Chance to win one of the Mac Keith Press Best Presentation Prizes - best oral presentation will win a cash prize of £350 and the best poster presentation will win £200 of Mac Keith Press vouchers
- Meet and network with leading experts in the field and colleagues from around the country

As in previous years, we encourage abstracts from all members of the MDT. DFundation doctors and undergraduate students will receive complimentary registration for the second day of the event if selected for presentation.

Key Dates

- **3 October 2025** - abstract submission open
- **9 December 2025** - abstract submission closes
- **13 January 2026** - abstracts selected for oral and poster presentation confirmed
- **23 February 2026** - all presenters to have registered to attend the conference on Wednesday 11 March, and poster presenters to submit jpeg/png file of poster

Full details are available here - please ensure you read the Abstract Guidelines before submitting your abstract(s).

TUESDAY 10 MARCH – PERSONAL PRACTICE WORKSHOPS

- The first day will focus on interactive, personal practice workshops.
- Registration is from 10.15am with the first workshop starting at 10.50am, and the day will close at 4.20pm
- Delegates will receive refreshments throughout the day and a cooked lunch
- Delegates can claim 4 CPD for this activity

Confirmed workshops are:

Developing a national guideline for the identification of learning disability in children
Dr Catherine Tuffrey, Consultant Paediatrician, HIOW Healthcare NHS Trust and BACD Chair

Doing better by our children: transforming SEND support and building needs-led care within Local Area Partnerships
Samantha Armitage, Advanced clinical practitioner occupational therapist in neurodisability, Sheffield Children's Hospital; Nick Whittaker and Emma Stevenson, Learn Sheffield

How to set up a district sleep service
Dr Hemant Kulkarni, Consultant in Paediatric Respiratory and Sleep Medicine; Janine Reynolds, Lead Paediatric Sleep Disorders Nurse Specialist; and Ruth Kingshott, Clinical Sleep Physiologist, Sheffield Children's Hospital

Managing conflict: multi-team working across boundaries and perspectives
Dr Mary Salama and Dr Hannah Nicholson, CoLab Partnership

Research-focused workshops
BACD Strategic Research Group

Using Artificial Intelligence in your clinical practice
Dr Venkat Reddy, Consultant Neurodevelopmental Paediatrician, Peterborough & Cambridgeshire NHS Foundation Trust, and RCPCH Officer for Digital Health and Technology

- Each workshop will last 60 minutes and repeated once.
- There is time allocated between workshops to enable delegates to move to their next workshop.
- Delegates will be able to choose to attend two of four workshops in session one and two, and two of three workshops in session three and four. The schedule of workshops will be confirmed shortly.

Gala Dinner

Join us for an evening of celebration and dancing! The Gala Dinner will include the PDDL diploma graduation and awards ceremony, a 3-course sit down meal, followed by a disco. The Gala Dinner will take place at the event venue, the Royal Victoria Crown Plaza Sheffield, arrival from 7pm, dinner from 7.30pm and carriages at 11pm. The Gala Dinner is open to all delegates, as well as colleagues, family and friends who are not attending the ASM but would like to come along.

WEDNESDAY 11 MARCH – CONFERENCE

This year's conference will focus on Mental Health and Wellbeing.

- The second day will take the form of the usual conference format, and delegates can choose to attend in-person or virtually via Zoom
- In-person registration is from 08.30am, with the first session at 09.20am, and the day will close at 4.30pm
- In-person delegates will receive refreshments throughout the day and a cooked lunch
- Delegates can claim 5 CPD for this activity

REGISTRATION – note early bird deadline 15 December!

Registration is now open! Discounted rates are available for BACD members, and there are additional discounts for those attending both days in-person.

	Day One & Two (9 CPD) IN PERSON	Day One Only (4 CPD) IN PERSON	Day Two Only (5 CPD) IN PERSON	Day Two Only (5 CPD) VIRTUAL
Doctor	£195	£110	£125	£90
Trainee / Resident Doctor	£120	£65	£75	£50
AHP / Non-Medical Professional	£120	£65	£75	£50
Undergraduate student / Foundation Dr	£75	£40	£45	£20

To register click here

For all aspects of the Annual Scientific Meeting 2026 visit the BACD website https://www.bacdis.org.uk/pages/asm-2026

PROGRAMME

We are pleased to present the programme, featuring speakers from across the multi-disciplinary team, overleaf.

BACD ASM 2026: PROGRAMME

08.15-09.15	**Trainees' Network Meeting** *A chance to meet and network with fellow trainees from across the country. Open to all paediatric trainees with an interest in childhood disability.*
08.30	Registration and Refreshments
09.20	Welcome and Housekeeping
09.25	**Healthy Parent Carers: peer-led group-based health promotion programme to improve health and wellbeing parent carers of disabled children** *Prof Chris Morris, Professor of Child Health Research, University of Exeter*
09.45	**"I am the only man in the room." The psycho-social experiences of fathers caring for children diagnosed as autistic** *Dr Louise Cooper, Lead Children's Learning Disability Nurse, Northern Care Alliance NHS Foundation Trust*
10.05	**Parent Carer Trauma: Insights for Transforming Practice** *Rachel Wright, parent and Founder of Born at the Right Time*
10.25	**Panel Q&A**
10.40	Refreshments, Exhibition, Posters
11.20	**BACD Strategic Research Group Update**
11.35	**Oral Presentations**
12.10	**Paul Polani Lecture: The impact of language disorders on mental health and wellbeing** *Prof Courtenay Norbury, Professor of Developmental Language & Communication Disorders, University College London*
12.55	Lunch, Exhibition, Posters BACD Annual General Meeting
14.00	**Prevalence of mental health conditions in adults with CP** *Dr Jennifer Ryan, Senior Lecturer, Royal College of Surgeons in Ireland and Director, Cerebral Palsy Lifespan Health and Well-being (CP-Life) Research Centre*
14.20	**Addressing health inequalities to improve mental health and wellbeing** *Dr Guddi Singh, Consultant in Neurodevelopmental & Social Paediatrics, Broadcaster, Founder, Wellbeing and Health Action Movement (WHAM)*
14.40	**Integrating Psychological Care into Acute Paediatrics: Promoting wellbeing beyond the bedside for hospitalised children and young people** *Dr Isabel Paz, John Radcliffe Hospital Oxford*
15.00	**Panel Q&A**
15.15	Refreshments
15.30	**Mental health of children with complex physical disabilities: the importance of meaningful activity and participation** *Annemarie Simms, Occupational Therapist, and Dr Ashley Liew, Consultant Paediatric Neuropsychiatrist, Evelina Children's Hospital*
15.55	**The Lucy Project – drop in service for CYP with mild to moderate psychological difficulties** *Dr Steve Jones, Principal Clinical Psychologist, Sheffield Children's Hospital and Sophie Bennett, Reader in Clinical Psychology, Kings College London*
16.15	**Panel Q&A**
16.25	Mac Keith Press Prizes Awarded
16.30	Close of Annual Scientific Meeting

Endorsed by BACD British Academy of Childhood Disability

Sheffield Children's NHS
NHS Foundation Trust

Paediatric Disability
Distance Learning

Introduction to Childhood Disability and Clinical Assessment

The Epilepsies . ADHD - A Clinical Guide

Autism, Speech, Language and Communication . Working with Families and Service Networks

Acquired Disabilities and Wider Needs . Links Beyond the Disability Team

Intellectual Disabilities . Vision . Motor Disorders . Hearing

Distance learning resources to support practitioners working with disabled children, young people and their families

Applications now being accepted for the Paediatric Neurodisability Diploma 2025

For all course information please scan the QR code, or email the support team at scn-tr.PDDLC@nhs.net