Neuromuscular Academy

Education with impact: Spinal muscular atrophy

Learning captured from workplace projects 2024

Neurology Academy®

Introduction

This year is the first that we have supported workplace projects for spinal muscular atrophy (SMA) services, and have been delighted to see such a spread of different topics and so many collaborative ventures.

Despite the highly specialist care required by many children, young people and adults living with SMA, the majority of these projects have focused on ensuring comprehensive and coordinated care across the whole spectrum of life, including physical, social, emotional and educational health whatever the specialist aspect of management might be. The importance of this wider consideration of all aspects of life has been clear in the involvement of a huge range of disciplines, and service transformation from specialist and secondary care through to community and primary services.

'I was very pleased to see so many of the projects looking at increasing our understanding in order to improve the experiences of people with SMA. We have had specialist services looking to understand patient experiences which is so important. I think my highlight might be seeing the education to, and upskilling of, primary and community services, which is very close to my heart.'

Dr Vasantha Gowda, consultant paediatric neurologist

'I've really enjoyed hearing about these projects and have particularly noticed the level of deliberate collaboration in so many of them. Several projects were carried out by two or more colleagues as a joint initiative, which really shares the work and embeds it firmly, whilst others have set out to work across specialities or disciplines - and this really elevates care and individuals' experience of it.'

Dr Min Ong, paediatric neurology consultant

From a unique young people's neuro-respiratory clinic through to improved intravenous access for Zolgensma, education for health visitors to multidisciplinary scoliosis management, these projects demonstrate clear steps to improvement, tangible outcomes, and transferable learning which can help transform SMA services across the UK.

At the end of this report are several takeaways that can be learned from, adapted, or put into place in other services and we hope they can inspire and encourage other healthcare professionals to improve services for people living with SMA and their families.

A new approach to transition

'Transition from paediatric to adult health care services is fraught with challenges in multiple domains including future health care use, educational and vocational trajectories, family and social life, self-fulfilment and quality of life' (Racine 2014). The neuroscience transformation programme (NHSE 2024) highlights the importance of transition of paediatric to adult services and has made it a priority for health and care services going forward.

'At an already stressful time of moving from paediatric to adult services navigating round adult services can feel all too much for many young patients. Often transition happens suddenly with little warning. The paediatrician who knew the child so well is replaced by a GP who struggles in the short appointments to grasp the whole complex picture.'

Aware of the challenges for young people living with SMA and the way that these can be compounded by the difficulties of transitioning into adult services, NMD adult physiotherapist Emma Manchester and neuromuscular transition nurse specialist Lisa Cutsey developed a monthly combined neuromuscular-respiratory clinic for 16–24-year-olds with SMA and other complex neuromuscular diseases. They have found positive attendance and re-engagement rates, opportunities to discuss drugs, smoking and vaping, and noticed an increase in wider opportunities such as informal MDT involvement, discussions around advance care planning and engagement of palliative care. Despite practical challenges to obtain administrative support and have the right equipment in the clinic, they hope to continue this work and have aspirations to increase the regularity, and expand the clinic to include a nurse advisor and psychosocial support.

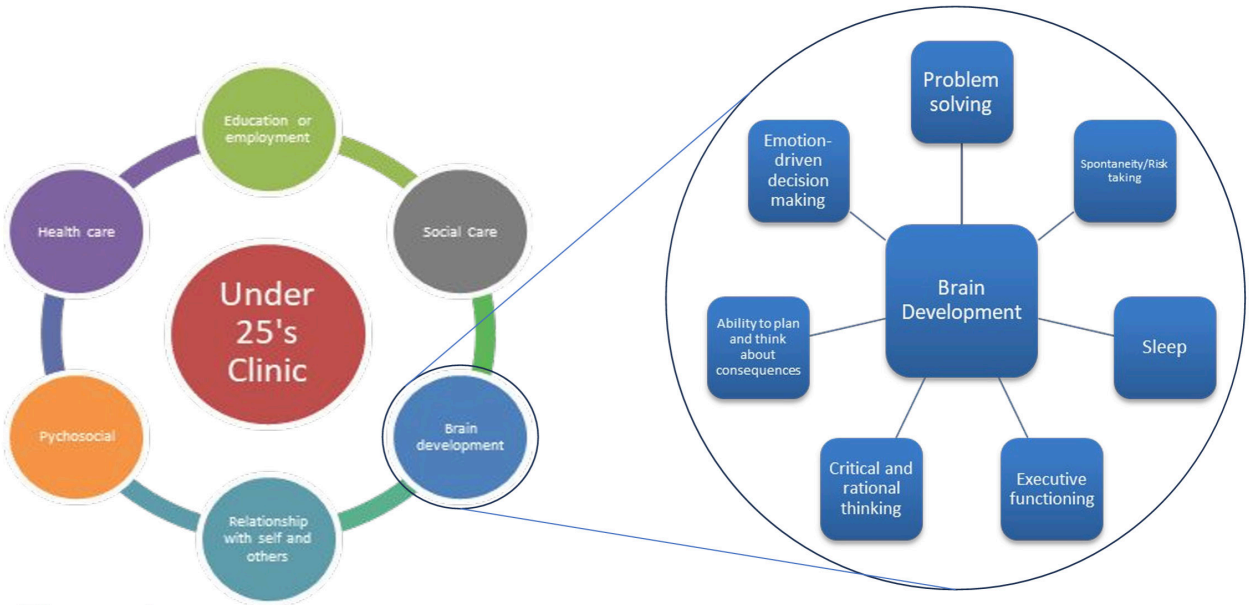

Figure 1: Examples of the complex physical, psychosocial and cognitive challenges to be considered during a clinic for patients aged 16-24.

Collaborative approaches to care

Complex conditions require input from multiple specialisms and communication and coordination across these specialisms is pivotal to ensuring quality care and improves function, longevity and quality of life in the person with SMA (Ropars 2020). Two of the projects carried out this year centred around this principle, one focusing specifically on collaboration, the other using collaborative principles to improve a management of scoliosis.

Clinical research fellow William Quelch, alongside his colleagues across neurology, respiratory and spinal services and supported by faculty member Dr Min Ong, looked at management of scoliosis in SMA patients in Sheffield. Almost all children under 5 to develop SMA will have scoliosis but traditional bracing or surgical interventions have both been found to impair respiratory function in SMA patients. A combined use of multidisciplinary team management across neurology, spinal and respiratory and development of bespoke asymmetric braces with a cut out over the abdomen to allow for full breaths, has been used to improve scoliosis and its impact without restricting respiratory function. William measured progression and compliance in those using the braces and found that the bespoke bracing slowed progression compared to non-bracing with no adverse effects and that there was good compliance in their use.

Neuromuscular specialist nurse and care advisor Louise Burnett and children's neuro-muscular specialist nurse Peter Learmonth worked together to develop a specialist care team to facilitate a coordinated approach to care across physical, social and educational domains for children with SMA and their families.

'The team has evolved from a single care advisor, to include professionals with a range of specialisms including acute, neurology and learning disability nursing, occupational therapy and mental health, and in a very wide range of settings including hospital, community health services, specialist education settings, CCG's and CAMHS.'

They developed a formalised referral and discussion process with the wider team, creating a specialist care team to meet twice monthly as an MDT, and holding joint clinics with respiratory, spinal and palliative teams, as well as transition clinics. Over a three month period, they have seen 126 referrals from a number of routes, including education spaces, health, social care and wellness support, and have found benefits noticed by families and team members alike.

Early access to treatment

With the significant advances in the treatment landscape for SMA in recent years, ensuring the earliest possible recognition and diagnosis of the condition followed by access to the most efficacious drug option available, has become a prime goal for services across the UK. Now that these drugs have been available for a few years, we are able to review the way they are experienced, both in terms of the administration of the drug itself and the impact they are having on individuals' health and quality of life.

Sheffield Teaching Hospitals Foundation Trust is one of the main centres for SMA treatment in the UK and two teams from within the hospital collaborated on two different projects to review aspects of two of the treatments available.

Delegate Katie Nevin, a clinical specialist physiotherapist in neuromuscular disorders, worked alongside five other members of her team, supported by faculty member Dr Channa Hewamadduma. They carried out an online survey to understand the impact of risdiplam on their patients. Risdiplam is available to any person of any age with SMA including infants with type 1 or 2 who are not eligible for or able to tolerate nusinersen, and those diagnosed at a later age. The authors note the importance of understanding lived experience of the drug alongside the emerging real world evidence. 67 patients using risdiplam across a wide age range from 0-4 to 65+ responded around their perceived quality of life, health status and side effects. They found that those with type 1 and type 2 SMA perceived a greater health benefit from treatment than those with type 3, but that all perceived increased quality of life with the drug, and that side-effects were experienced in less than half of respondents and were commonly loss of appetite, diarrhoea, headaches, and dermatological problems.

Trainee advanced nurse practitioner Emma Kimpton, clinical nurse specialist Toni Lawford and paediatric neurosciences pharmacist Susan George reviewed their process for securing intravenous (IV) access for Zolgensma, the gene therapy available to infants diagnosed with type 1 SMA (up to 6 months old). They undertook the review after noticing a common trend over the two years that the drug has been administered: it is often difficult to place IV access in these patients. They suggest contributing factors including that the SMN protein is required to promote healthy vasculature, that this is an issue in up to 50% of children and that the difficulties increase in chronically unwell children. A review of those patients who had Zolgensma as an outpatient found 80% had difficulties with IV access often resulting in an anaesthetist being called. They decided to put a process in place to mitigate against difficulties and reduce distress to families and patients. This process included involving the floor anaesthetist in initial discussions, creating a plan jointly with the family, coordinating timings across all involved including pharmacy and aseptic units, and retaining this on electronic records, reviewing as needed.

Comprehensive care

With the advances in treatment that have come about in recent years, the prognosis for people living with SMA is improving significantly whatever type they have been diagnosed with. A number of workplace projects focused on health promotion or maintainance, designed to decrease longer term risk and maximise quality of life. These included measuring maximal mouth opening (MMO), bone health, and reviewing how the whole picture of care can contribute to sustained quality of life.

Specialist neuromuscular physiotherapist Anna Rutherford highlighted the importance of measuring MMO in SMA. She noted that, with patients living longer, their MMO may continue to deteriorate and affect their oral hygiene, nutrition, and ability to clear secretions, as well as impacting risk for future medical procedures where intubation is needed.

'Awareness of this common complication among clinicians is therefore vital, as is the regular assessment and clear documentation of MMO limitation for patients with SMA.'

She standardised its assessment in her service and, after reviewing existing methods for measurement, developed her own easy to use grading scale. She found its use was positive for both professionals and patients and now seeks to embed it in the local care pathway.

Jing Ming Yeo noted an increased risk of developing low bone mineral density, osteoporosis and fragility fractures in people with SMA impacting function, quality of life and long-term health outcomes. Finding a gap in care around this area of health monitoring, he developed a bone health checklist for adults with SMA with advice on review procedure, and recommendations for professionals and the patient around lifestyle and medication changes, services to refer to and steps to take in follow-up.

Specialist physiotherapist Jennielyn Ang reviewed the national picture of what comprehensive care should look like in order to promote a good and sustainable quality of life in all people with SMA (fig 1).

Education & community upskilling

In a complex neuromuscular condition, it can be tempting to focus on specialist care and overlook the role of primary and community services, yet these are essential to optimise both early diagnosis, and ongoing treatment and support for SMA.

'Early recognition, diagnosis and access to treatment are essential to ensure the best possible quality of life and health outcomes for an infant with spinal muscular atrophy (SMA)' (SMA paediatric pathway). Often, particularly for type 1 or 2 SMA, the GP, practice nurse or health visitor may be the first person to see signs of SMA, and increasing awareness about these 'red flags' to trigger a referral to paediatric neurology is an important part of preserving as much function in the infant as possible, and getting access to the most effective treatment for them.

Clinical fellow in paediatric neurology Ushma Patel and neuromuscular care advisor Yvonne Julien decided to improve recognition and early referral within primary care by reviewing and addressing education gaps amongst local health visitors. This winning project, as voted by the whole delegate class, saw the colleagues develop an education session for health visitors which they rolled out locally. They created a pre- and post-session questionnaire to measure both the level of understanding amongst those at the session, and the amount of improvement in that understanding as a result of the session.

Ushma and Yvonne found a significant gap in health visitors' awareness of SMA recognition and treatment options but a notable improvement following the education session, particularly in relation to neurological assessments and referral processes. Their work highlights the importance of educational interventions in empowering healthcare professionals to identify and manage SMA cases effectively, stressing the urgency of early intervention for better outcomes.

The current medical advancements in SMA treatment have changed the therapy landscape, requiring both more assessment and increased opportunities for improved function. To provide care as close to home as possible, this is ideally supported locally. Advanced physiotherapist Neil Hinde used a questionnaire to review his regional therapy service in terms of confidence and knowledge around supporting children with SMA. He found that many of the respondents would value more training, and so the specialist neuromuscular therapy team will be liaising with their community therapy teams to provide this upskilling.

Top tips and takeaways

Here are some ideas you could consider replicating based on this project work, summarised in five statements made by the delegates themselves, or featured in recent policy and guidance.

1.Transition is fraught with challenges across multiple domains for young people with SMA. There are lots of ways to improve this, whether with a new or developing an existing initiative, such as

- establishing a specific clinic for 16-24 year olds, spanning the transition period such as the respiratory clinic developed by Emma and Lisa

2. Multidisciplinary management is essential in SMA, and novel collaborations with other disciplines and departments can optimise care further. Collaborations explored by delegates in this MasterClass include:

- a combined approach to scoliosis across neurology, respiratory and spinal departments, like William
- bring different roles together to form a 'specialist care team', like Louise & Peter

3. The treatment landscape has led to great opportunities and new challenges and we are learning more all the time. Using ideas from this years' projects, you could:

- review the outcomes of a treatment such as risdiplam to better understand its benefits across a cohort of patients, like Katie
- develop a collaborative approach across neuromuscular, anaesthesia and the family to optimise Zolgensma administration, like Emma, Toni and Susan

4. With improved prognosis for people living with SMA comes a need to increase preventative care and reduce comorbidities. Ideas from this MasterClass are:

- review assessment for MMO in SMA and standardise its assessment through agreed pathways or protocols, like Anna
- improve health monitoring such as through standardising bone health assessments, like Jing

5. Primary and community services play a vital role in the diagnosis and management of people with SMA. To ensure they have the knowledge and skills needed to support you could:

- develop education for health visitors and measure the improvement it has on understanding, like Ushma and Yvonne
- review community therapists' knowledge and confidence in SMA management and establishing links with specialist teams for upskilling, like Neil

Neurology Academy: education with impact

Neuromuscular Academy is part of Neurology Academy.

Neurology Academy is an innovative educational provider for healthcare professionals including consultants, specialist nurses, pharmacists, therapists and other allied health professionals. We provide practically-applicable events, courses and online education across a wide range of subject areas.

Our education is developed by practicing specialists who combine their experience and expertise into case-based learning designed to create specialists in their field with confidence in effecting change.

We specialise in education, networking and mentorship, encourage the sharing of good practice and promote clinical leadership to enable service transformation and improve outcomes for people living with a neurological condition.

www.neurologyacademy.org

Neurology Academy

1 The Edge Hillsborough Barracks
Langsett Rd
Sheffield
S6 2LR

01143 270 230
info@neurologyacademy.org
@TheNeuroAcademy