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Caring & Living As Neighbours

# Spinal Muscular Atrophy Club Meeting

April 2024

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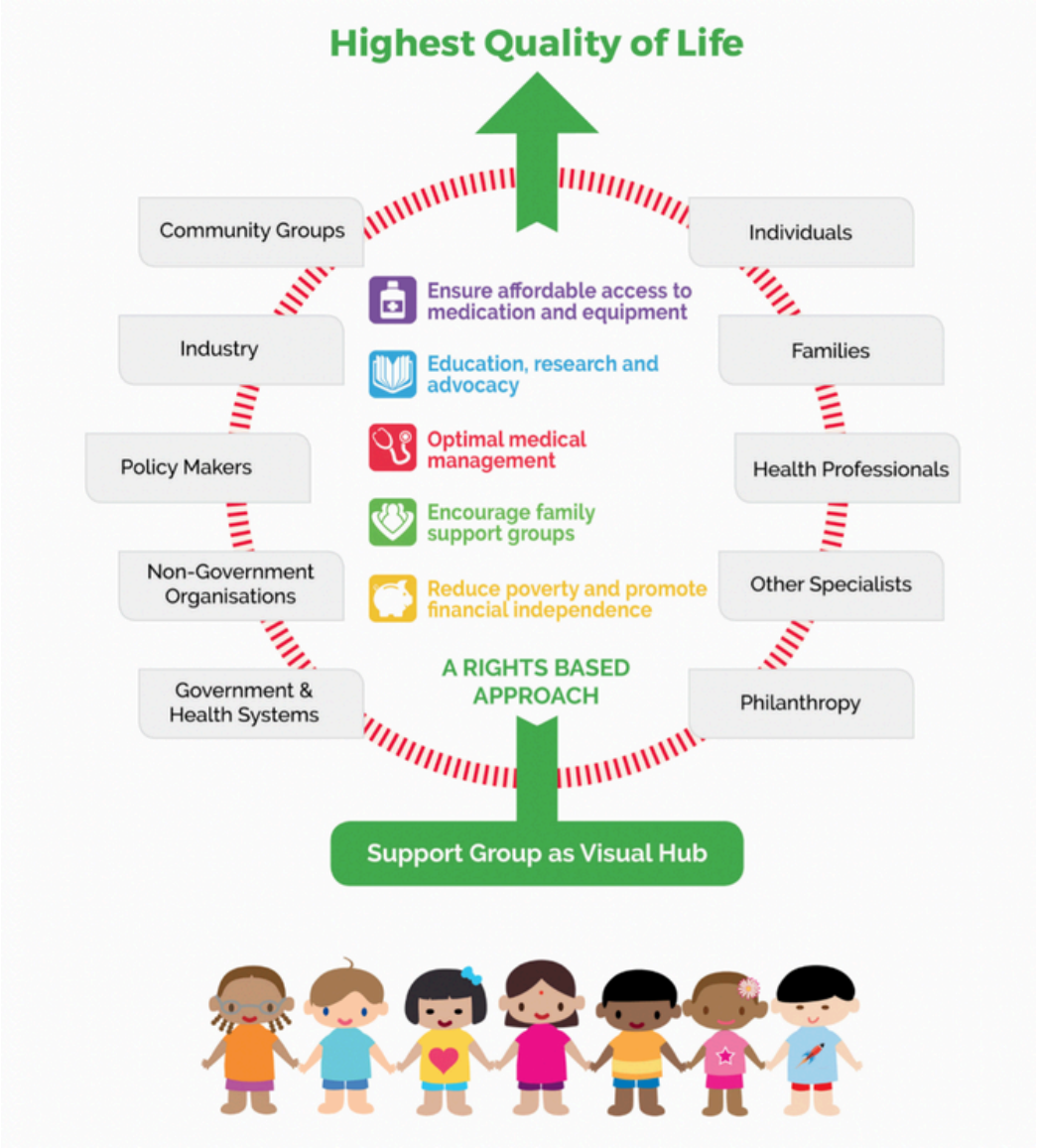
# Every child has the basic human right to health and life

Caring & Living as Neighbours (CLAN) is an Australian based Non-Governmental Organisation formally associated with United Nations Department of Global Communications (UNDGC) and in Special Consultative Status with United Nations Economic and Social Council (ECOSOC).

CLAN's mission is to maximise quality of life for children and their families who are living with chronic health conditions in resource-poor settings of the world. CLAN's five pillars and framework for action are foundational to meeting the needs of communities of children living with chronic health conditions.




# Overview

The Conference of Rare Diseases was held from Thursday 25 March to Saturday 1 April at the Vietnam National Children's Hospital (VNCH) in Hanoi. The conference focused on genetic neuromuscular diseases, specifically, spinal muscular atrophy (SMA), Duchenne muscular dystrophy (DMD), and Pompe disease. Over the three days, presentations were given for the continuing medical education (CME) of health professionals and for training families and individuals living with SMA. The presenters were as follows:

- Clinical Professor Kristi Jones, Senior Staff Specialist and Head of Department of Clinical Genetics at the Sydney Children's Hospital Network - Westmead;
- Dr Michelle Lorentzos, Paediatric Neurologist and Clinical Trials Medical Lead at the Sydney Children's Hospitals Network - Westmead;
- Michelle Williams, Neuromuscular Physiotherapist at the Sydney Children's Hospitals Network in Westmead;
- Lisa Tang, Neuromuscular Clinical Nurse Specialist at the Sydney Children's Hospitals Network in Westmead;
- Dr Vũ Chí Dũng, Director of the Centre for Endocrinology, Metabolism, Genetics and Molecular Therapy at the Vietnam National Children's Hospital;
- Dr Nguyễn Ngọc Khánh, Vice Director of the Centre for Endocrinology, Metabolism, Genetics and Molecular Therapy at the Vietnam National Children's Hospital;
- Dr Nguyễn Lê Trung Hiếu, Head of Neurology Department at Children's Hospital 2; and
- Spinal Muscular Atrophy Patient Group

Key activities during this conference included:

- Continuing Medical Education (CME) for resident paediatricians from Vietnamese provinces;
- Training for families and caregivers with the SMA support group; and
- Training for the VNCH Department of Endocrinology, Metabolism and Genetics.

This event was a collaborative effort between CLAN, VNCH, Taking Paediatrics Abroad (TPA), and the Sydney Children's Hospital Network - Westmead (SCHN-W). We are grateful for this long-standing relationship between parties continuing to contribute to improvements in professional practice and the health of children with genetic and neuromuscular conditions. A special thanks to the SCHN-W team for travelling from Australia to Vietnam, for the team at VNCH for the warm welcome and hospitality, and for the translators that facilitated communications throughout the event. CLAN representative, Anna Rea, attended this conference with the support from the Australian Government's New Colombo Plan scholarship. This report focuses on the activities relating to SMA.

SMA is a neurological condition that causes muscle weakness and atrophy. It occurs in about 1 in 12,000 live births, with 1 in 54 people being a carrier of the gene mutation. There are currently 300 children living with SMA in Vietnam under the age of 15; most are under the age of 5. SMA was previously a fatal condition but thanks to increased newborn screening, early supportive treatment, access to disease modifying therapies, and community support groups, many children are living longer, happy lives.


# Paediatric Resident Training

Resident doctors from Vietnamese provinces were welcomed to the Ho Chi Minh Conference Hall at VNCH to undertake CME on rare genetic neuromuscular diseases. All presentations emphasised the critical role of early detection, where the best outcomes are seen when children receive treatment for SMA within the first 14 days of life.

Dr Jones gave an overview of SMA, the four types of SMA presentation, and described the genetic basis of the condition. This underpinned Dr Lorentzos' presentation on new SMA therapies such as nusinersen and adeno-associated virus (AAV) mediated gene replacement. Dr Jones, Lorentzos, and Williams detailed clinical practices, including floppy infant assessment, SMA diagnosis, multidisciplinary care, and genetic counselling. The multidisciplinary team consists of genetic counselling, physiotherapy, occupational therapy, orthopaedic management, nursing, psychology support, palliative care, pulmonary and acute care, and nutrition with long-term follow up. Dr Hiếu and Dr Khánh presented on SMA from a Vietnamese perspective and reinforced the importance of early detection, treatment, and multidisciplinary teams. The most common cases of SMA in Vietnam are type 2, classified as being non-ambulant but able to sit independently. The presentations were followed by interactive case studies and a question and answer session.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# SMA Club Meeting

On Friday 26 April, VNCH welcomed families and children with SMA to the Hanoi Conference Hall. Approximately 60 people attended the event, which involved presentations from the SMA Patient Support Group, the SCHN-W team, and an open discussion between families and health professionals.

The SMA Patient Support Group presentation highlighted the progress made with SMA over time, the importance of community support, and the focus on future improvements. The group's goals include SMA advocacy in health leadership and within the community, creating a SMA patient network in Vietnam, and to improve access to SMA medicine and rehabilitation services. The group is active in advocacy , community support, fundraising, and has successfully supported 20 SMA patients to receive regular treatment with Risdiplam from Chinese hospitals. However, the current challenges of the SMA community include:

- getting approved SMA drugs in Vietnam;
- the affordability of medication for families;
- knowledge and skills of parents to care for children with SMA at home; and
- lack of rehabilitation services, including assistive devices.

The SCHN-W addressed one of these challenges by training families with skills and knowledge to care for their children with SMA. Children with SMA are now living longer and stronger with early treatment, gene therapy, and supportive care. Treatment and care options were detailed and exemplified by young patients at SCHN-W seeing positive outcomes and notable improvements. Nurse Tang mapped the landscape of multidisciplinary care, provided useful tips for families navigating clinical time, and recommended reputable resources available in Vietnamese for further information. Dr Lorentzos drew attention to the psychosocial care of the entire family, where community support groups play an important role. For children with SMA, playing with siblings can be the best psychosocial therapy. Presenters acknowledged the power parents and families have in advocacy, the importance of uniting through the support group, and their central role in their child's SMA care.

Families were particularly interested in physiotherapy and assistive devices, which was one of the challenges identified by the SMA support group. Dr Williams, physiotherapist detailed physical assessment, respiratory management, and demonstrated some orthopaedic equipment. She highlighted the importance of weight bearing activities, where possible, for bone health. Some children at the event were using orthopaedic braces and showed their increased mobility. A common concern among families were compromised breathing and Dr Williams demonstrated the technique of percussions to assist with autogenic drainage that is safe for parents to practice at home. Many families were practising the technique after the session.

The presentations were followed by an active discussion session, where parents presented their questions and concerns to the SCHN-W and VNCH teams.



# Paediatric Endocrinology, Metabolism and Genetics Department Training

Approximately 30 medical professionals from the Department of Endocrinology, Metabolism and Genetics Centre for Rare Diseases and Newborn Screening attended the training session. The attendees included paediatricians, physiotherapists, and nurses. The presentations went into more depth about diagnosis, treatment, multidisciplinary care, and communicating with families.

Dr Lorenzos outlined the International Classification of Functioning, Disability and Health Framework, encompassing fitness, functioning, friends, family, fun, and future. This focuses on building strong relationships with families, who are the best teachers for clinical practice specific for patient needs. Health professionals have a role in helping families navigate their experience with diagnosis and care, including psychosocial care. It's beneficial to have a contact person for parents to coordinate multidisciplinary care and communication across all teams. Nurse Tang detailed that improved quality of life and survival rates are seen with multidisciplinary care.

Presenters discussed treatment options, considerations, case studies, and success stories from patients at SCHNW. Dr Williams outlined physical assessments, therapy, respiratory management, and conducted training for autogenic drainage techniques. The VNCH practised percussions and vibrations to assist the dispulsion of respiratory mucous in children with compromised breathing or undeveloped lungs. After the presentations, an open discussion and question time ensued.

# Closing

The conference was a fulfilling experience shared by Vietnamese and Australian health professionals and families of children with neuromuscular conditions. Over the three days VNCH, SCHN-W, resident paediatricians, and families shared their knowledge and experiences to improve care for individuals living with SMA and neuromuscular conditions. Emphasis was drawn to early diagnosis and treatment, multidisciplinary care, and the role of family and community support networks.

Thank you to VNCH for the warmth, hospitality, and invaluable contributions to make this event possible. Special recognition is extended to Dr Dung and Dr Khanh for facilitating this event and exceptional support provided to the SCHN-W team. Thank you to CLAN and TPA for making this international collaboration possible and for the translators that enabled communications. It's an honour to have the opportunity to learn and share experiences with such passionate professionals and families.

Many paediatricians at VNCH have spent some time in Australia during their training and studies, which shows in their strong medical English understanding. This demonstrates the strong relationship between Australia and Vietnam in knowledge exchange, professional development, and patient care.

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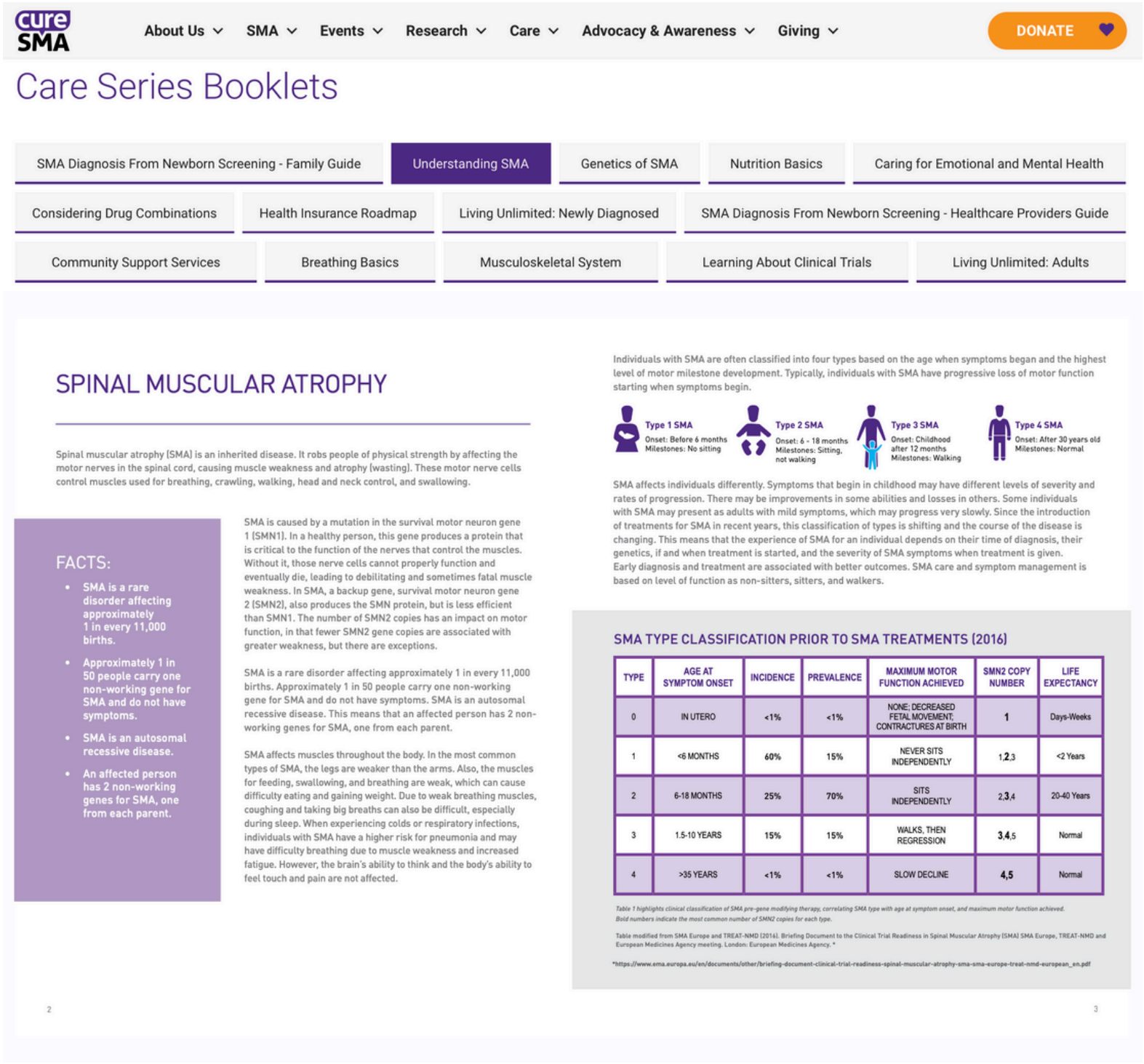
*Special thanks to Cure SMA for providing trusted resources for families and healthcare providers about caring for individuals living with SMA. These resources are available on their website in a number of languages including English and Vietnamese. Please visit the Cure SMA website for further information.*

**https://www.curesma.org**




## SPINAL MUSCULAR ATROPHY

Spinal muscular atrophy (SMA) is an inherited disease. It robs people of physical strength by affecting the motor nerves in the spinal cord, causing muscle weakness and atrophy (wasting). These motor nerve cells control muscles used for breathing, crawling, walking, head and neck control, and swallowing.

### FACTS:

- SMA is a rare disorder affecting approximately 1 in every 11,000 births.
- Approximately 1 in 50 people carry one non-working gene for SMA and do not have symptoms.
- SMA is an autosomal recessive disease.
- An affected person has 2 non-working genes for SMA, one from each parent.

SMA is caused by a mutation in the survival motor neuron gene 1 (SMN1). In a healthy person, this gene produces a protein that is critical to the function of the nerves that control the muscles. Without it, those nerve cells cannot properly function and eventually die, leading to debilitating and sometimes fatal muscle weakness. In SMA, a backup gene, survival motor neuron gene 2 (SMN2), also produces the SMN protein, but is less efficient than SMN1. The number of SMN2 copies has an impact on motor function, in that fewer SMN2 gene copies are associated with greater weakness, but there are exceptions.

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SMA affects muscles throughout the body. In the most common types of SMA, the legs are weaker than the arms. Also, the muscles for feeding, swallowing, and breathing are weak, which can cause difficulty eating and gaining weight. Due to weak breathing muscles, coughing and taking big breaths can also be difficult, especially during sleep. When experiencing colds or respiratory infections, individuals with SMA have a higher risk for pneumonia and may have difficulty breathing due to muscle weakness and increased fatigue. However, the brain's ability to think and the body's ability to feel touch and pain are not affected.



Individuals with SMA are often classified into four types based on the age when symptoms began and the highest level of motor milestone development. Typically, individuals with SMA have progressive loss of motor function starting when symptoms begin.

**Type 1 SMA**
Onset: Before 6 months
Milestones: No sitting

**Type 2 SMA**
Onset: 6 - 18 months
Milestones: Sitting, not walking

**Type 3 SMA**
Onset: Childhood after 12 months
Milestones: Walking

**Type 4 SMA**
Onset: After 30 years old
Milestones: Normal

SMA affects individuals differently. Symptoms that begin in childhood may have different levels of severity and rates of progression. There may be improvements in some abilities and losses in others. Some individuals with SMA may present as adults with mild symptoms, which may progress very slowly. Since the introduction of treatments for SMA in recent years, this classification of types is shifting and the course of the disease is changing. This means that the experience of SMA for an individual depends on their time of diagnosis, their genetics, if and when treatment is started, and the severity of SMA symptoms when treatment is given. Early diagnosis and treatment are associated with better outcomes. SMA care and symptom management is based on level of function as non-sitters, sitters, and walkers.

### SMA TYPE CLASSIFICATION PRIOR TO SMA TREATMENTS (2016)

| TYPE | AGE AT SYMPTOM ONSET | INCIDENCE | PREVALENCE | MAXIMUM MOTOR FUNCTION ACHIEVED | SMN2 COPY NUMBER | LIFE EXPECTANCY |
|---|---|---|---|---|---|---|
| 0 | IN UTERO | <1% | <1% | NONE; DECREASED FETAL MOVEMENT; CONTRACTURES AT BIRTH | **1** | Days-Weeks |
| 1 | <6 MONTHS | 60% | 15% | NEVER SITS INDEPENDENTLY | 1,**2**,3 | <2 Years |
| 2 | 6-18 MONTHS | 25% | 70% | SITS INDEPENDENTLY | 2,**3**,4 | 20-40 Years |
| 3 | 1.5-10 YEARS | 15% | 15% | WALKS, THEN REGRESSION | **3**,4,5 | Normal |
| 4 | >35 YEARS | <1% | <1% | SLOW DECLINE | **4,5** | Normal |

*Table 1 highlights clinical classification of SMA pre-gene modifying therapy, correlating SMA type with age at symptom onset, and maximum motor function achieved. Bold numbers indicate the most common number of SMN2 copies for each type.*

Table modified from SMA Europe and TREAT-NMD (2016). Briefing Document to the Clinical Trial Readiness in Spinal Muscular Atrophy (SMA) SMA Europe, TREAT-NMD and European Medicines Agency meeting. London: European Medicines Agency. *

*https://www.ema.europa.eu/en/documents/other/briefing-document-clinical-trial-readiness-spinal-muscular-atrophy-sma-sma-europe-treat-nmd-european_en.pdf