FACT SHEET

SPINAL MUSCULAR ATROPHY TYPE 1

MUSCULAR DYSTROPHY NSW BUILDING STRENGTH REACHING POTENTIAL

Spinal Muscular Atrophy (SMA1) is a type of childhood motor neurone disease.

INCIDENCE: 1 : 10,000 live births
AVERAGE AGE OF DIAGNOSIS: 3 - 9 months of age (birth - 12 months)
OCCURRENCE: affects males and females
LIFE EXPECTANCY: usually 1 - 2 years of age

Spinal Muscular Atrophy (SMA1) is a genetic condition caused by a faulty gene (SMN1) carried by each parent that is then passed on to the child. SMA1 is caused by a lack of a protein called survival motor neuron (SMN), in the motor neurons within the spinal cord that control voluntary muscle movement. Without the SMN protein to support growth of the motor neurons coming from within the spinal cord they do not survive and this results in decreased muscle function as the muscle cells die (atrophy). SMA has a continuum from the most severe (Type 1) to the mildest form (Type 4,) and the individual types are defined by the maximum motor skills gained.

SMA1 is a severe progressive condition that primarily affects the muscles for movement and breathing in babies. Infants born with SMA1 will not reach the normal motor skill milestones like sitting, crawling or walking, and will have a shortened life expectancy. The brain is not affected in SMA1 and these babies are engaging, bright eyed and intuitive. Babies with SMA1 are usually floppy at birth compared to their peers. In the early stages, SMA1 can be very subtle and delays in motor skills may not to be evident until a few months of age. It can be difficult to support intellectual growth in the presence of physical decline. There is no current cure.

Basic motor functions become affected such as swallowing, feeding, breathing and independent muscle movement. The muscles that control breathing and coughing are significantly weak and the major risk to babies with SMA1 is complications from coughs, colds, chest infections and pneumonia which can quickly become life threatening. These babies require services and equipment to ensure they can interact with their surroundings, maintain good nutritional status, optimise their breathing and support their positioning in the context of muscle weakness.

Recent advancements have seen some gene modifying drugs being trialled in infants with SMA1 and there is hope that the progression of motor neuron atrophy can be paused and infants may gain motor skills. Currently the availability of these gene modifying treatments is limited. Treatment options require a huge amount of time and regular hospital visits and assessments. This should be considered when providing resources to support attendance for treatment. Some SMA1 babies who are receiving gene modifying treatment are living longer.

Planning ahead for services and equipment is essential, particularly given the progressive nature of SMA1 in infants

Despite being a rare disease, SMA1 is the largest genetic cause of infant death. About 1 in 50 people in the general population carry the faulty SMN1 gene.

Genetic testing and counselling should be offered to parents and family members for future pregnancy planning. Parents and families experience enormous grief in the dealing with the diagnosis and decline in function of their child with SMA1, and the knowledge that their child will have a shortened life expectancy.

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The need for "end-of-life" care planning should be discussed with the parents early on or when deemed appropriate. Timely referral to palliative care services for symptom management and comfort should be considered. Sometimes care planning allows the parents to focus on what can be done rather than the end of their child's shortened life. It is up to the parents how they want to proceed with the care options for their child and planning will reflect the needs of each individual situation. Referral to palliative care hospices may be required. Enough support needs to be provided to those with SMA1 so that their carer can have a break.

It is important that service provision is centered around quality of life outcomes and meets the needs of the individual

Families and individuals affected by chronic conditions need to be empowered in order to have control over their life choices. Through seeking guidance and support regarding treatment and care options, parents will be empowered to make the right decisions for their child. Despite the physical challenges a child with SMA1 faces, there are opportunities available to support infants with SMA1 to have a happy and interactive life.

CONSIDERATIONS FOR PLANNING SHOULD INCLUDE BUT ARE NOT LIMITED TO THE FOLLOWING:

- A consistent General Practitioner (GP)
- Specialists for monitoring and medical care planning eg. neurologist, respiratory physicians
- Support coordination for complex care needs
- Physiotherapy programs eg. hydrotherapy, cough assist, chest physio
- Occupational therapy to support daily activities eg. modified car seat
- Speech therapy for swallowing and speech problems
- Comfort/sleeping eg. special pillows to support head and torso
- Continence management planning eg. nappies
- Nutritional support eg. dietician, feeding pumps & consumables, high calorie formulas
- Assistive technology eg. iPad
- Assistive ventilation devices eg. CPAP or BiPAP machines
- Adapted equipment eg. bathing equipment such as a bath bean chair
- Surgery and hospital admission planning eg. permanent feeding tubes (PEGs)
- Professional psychological support for family eg. counsellor or clinical psychologist
- Future planning eg. palliative care
- Financial entitlements for parent as carer eg. Centrelink, Medicare

USEFUL RESOURCES

MDNSW www.mdnsw.org.au
MDNSW NDIS Toolkit www.mdnsw.org.au/ndis
Spinal Muscular Atrophy Australia (SMAA) www.smaaustralia.org.au

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