Spinal Muscular Atrophy AUSTRALIA INC.

FACT SHEET

Spinal Muscular Atrophy (SMA) is an inherited genetic (it is NOT contagious) muscle wasting disease. Muscles affected are not only external muscles like arms and legs, that we can see, but also all internal muscles; including breathing, coughing and swallowing. As time progresses coughing becomes difficult and breathing becomes laboured.

SMA FACTS

- SMA is the childhood version of Motor Neurone disease.
- 1 in 35 people in Australia unknowingly carry the faulty SMA gene. Being a carrier does not mean you are affected by the disease.
- One in 10,000 live births in Australia are affected by SMA.
- 60-70% of all SMA patients have the most severe form (Type 1).
- SMA is a physical disease only. Children with SMA have reduced movement.
- There is no known cure for Spinal Muscular Atrophy but with recent drug advancements there is some new treatment options for SMA.
- Babies (Type 1) don't often reach milestones like sitting or rolling in early infancy, have hypotonia (weak muscles), progressive weakness and loss of motor function.
- Babies born with SMA appear perfectly normal in every way except they are extremely weak. They are bright, alert, interested in people and what's going on around them. They enjoy music and being played with – just like other babies.
- SMA Children's intelligence is unaffected. Many people with SMA have above average intelligence. Children go to main stream schools, adults work (ie: graphic design, lawyer) and even have children themselves.
- A person is born when BOTH parents are carriers of this gene, neither parent is to blame. There is a 1 in 4 chance of this couple having future babies with SMA.
- New genetic testing technology using saliva samples is now available Australia wide. The test costs $385 and is available through www.vcgs.org.au (this test screens for SMA, Cystic Fibrosis and Fragile X).
- Children with SMA catch germs very easily so washing hands before having contact is very important. Avoid visits by anyone suffering a cough / cold or anything contagious, due to their weakened immune system.
- Infants / Children / Adults diagnosed with Type 2 and Type 3, have a good survival rate, but mobility and dexterity are compromised. Most sufferers are wheelchair bound by early childhood. With some requiring steel rods in their back to allow them to sit upright and prevent scoliosis. All sufferers require assistance with activities of daily living such as grooming and feeding etc.
- Around 630 people die from Motor Neurone disease every year in Australia. (ABS)

This Fact Sheet was prepared by Spinal Muscular Atrophy Australia Inc.

For further information please call:

Spinal Muscular Atrophy Australia Inc.
Unit 7, 16-28 Melverton Drive
Hallam VIC 3803

PO Box 5245, Hallam VIC 3803

Phone 03 9796 5744
Email reception@smaaustralia.org.au
Web www.smaaustralia.org.au

ABN 82 885 991 569
A0047660D

Our SMA goals #beSMAaware

"Timely access to treatment for SMA families"

"SMA to be added to the newborn screening list for earlier intervention"

"Clearly promote reproductive choice through available carrier screening"