Spinal muscular atrophy (SMA) affects approximately 1 in 10,000 live births worldwide. SMA is caused by a mutation in the SMN1 gene, which results in reduced levels of survival of motor neuron (SMN) protein. SMN protein is found throughout the body and plays a vital role in muscle function.

The body has two very similar genes that make SMN protein: SMN1 and SMN2. These genes are located near each other on the same chromosome (number 5). SMN1 is a normal gene and is the fully functional copy of the gene. SMN2 is a modified, or ‘splice mutant’, version of SMN1, which only produces a small fraction of the normal SMN protein (about 10%).

Healthy individuals

The body has two copies of the SMN1 gene which produces all the SMN protein we need to function

Individuals with SMA

The body has two copies of the SMN1 gene which produces insufficient amounts of functional SMN protein

Healthy individuals are at a reduced risk that the child will have SMA, but there is a 25% chance that the child will also be a carrier and a 25% chance the child will be completely unaffected.

A carrier is someone who does not have SMA, they have one ‘normal’ copy of the SMN1 gene and one copy of the SMN1 gene with a mutation.

Carriers are usually unaware of the mutated gene.

When two people who are carriers of the affected gene have a child, there is:

- A 25% chance the child will have SMA
- A 50% chance that the child will also be a carrier
- A 25% chance the child will be completely unaffected

Although SMA is mainly diagnosed in childhood it can affect people at any age, from childhood to adulthood. However, the earlier symptoms appear, the more severe the disease.

You can find more information on SMA at Cure SMA: www.curesma.org/carriers-of-sma/

Every person with SMA is different, but there are 3 main types of SMA, based on the age when symptoms first appear:

**Type 1**

- Develops in babies more than 6 months old and is the leading cause of death before two years of age.
- Children with SMA Type 1 cannot sit or walk, or breathe on their own. Extra medical treatment may also be necessary.
- The most severe form of SMA.

**Type 2**

- Develops in children who are 7-18 months old, leading to muscle weakness. It may also shorten life expectancy.
- Children with SMA Type 2 can sit and walk, but may need to use a walker or wheelchair. They can breathe on their own, but may need extra medical treatment.
- faster progressions.

**Type 3**

- Develops after 18 months of age and may not be evident until late childhood.
- Most children with SMA Type 3 can sit and walk, but may need to use a walker or wheelchair. They may breathe on their own, but may need extra medical treatment.
- Often the slowest progression of SMA.

Ongoing research is exploring how other cells and organs, including the heart, vascular system, and bone may also be affected by reduced levels of functional SMN protein.

For more information on SMA, go to SMA Europe: www.sma-europe.eu/essentials/