Spinal muscular atrophy (SMA) affects approximately 1 in 11,000 live births worldwide. SMA is caused by a mutation in the SMN1 gene, which results in low levels of survival of motor neuron (SMN) protein. SMN protein is found throughout the body and plays a vital role in muscle function.

The development of spinal muscular atrophy

Having low levels of functional SMN protein leads to progressive weakening and wastage of nerve cells, which control muscle movement.

Healthy individuals

The body has two very similar genes that make SMN protein: SMN1 and SMN2.

- SMN1: This gene is able to produce enough protein for the body to function normally.
- SMN2: Only makes a small fraction of the SMN protein we need to function (about 10%).

Individuals with SMA

Mostly non-functional SMN protein

- SMA1: The body has two copies of the SMN1 gene. One copy has a mutation, and the other copy is normal.
- SMA2: The body has only one healthy SMN1 gene and one copy of the SMN1 gene with a mutation.

SMN2 is not normally functional, and most people with SMA are completely unaffected.

- SMA3: The body has only one healthy SMN1 gene and one copy of the SMN1 gene with a mutation.
- SMA4: The body has two copies of the SMN1 gene. One copy has a mutation, and the other copy is normal.

Type 1

Develops in babies less than 6 months old and is life-limiting. Few children survive beyond two years of age.

Type 2

Develops in children who are 7-18 months old, leading to muscle weakness. It may also shorten life expectancy.

Type 3

Develops after 18 months of age and may not be evident until late childhood.

Although SMA is mainly diagnosed in childhood, it can affect people at any age. Even in adulthood, between the usual symptoms in infants, the more severe the disease.

Every person with SMA is different, but...

...there are 3 main types of SMA, based on the age when symptoms first appear.

Type 1

- Requires immediate care
- Typically diagnosed in babies
- Life expectancy is usually short

Type 2

- Requires significant care
- Typically diagnosed in children
- Life expectancy varies

Type 3

- Requires ongoing care
- Typically diagnosed in adults
- Life expectancy may be affected

SIMN1 (full function) needed for normal function

SIMN2 (no function) mostly non-functional

SMN1 (full function) needed for normal function

SMN2 (no function) mostly non-functional

Healthy individuals have two copies of the normal SMN1 gene and one copy with a mutation. They are usually unaware of the mutated gene.

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

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

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:

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

Reference: