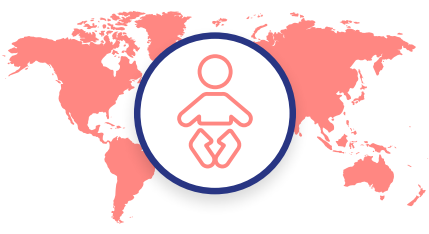
The development of **Spinal Muscular Atrophy**

Spinal muscular atrophy (SMA) affects approximately...

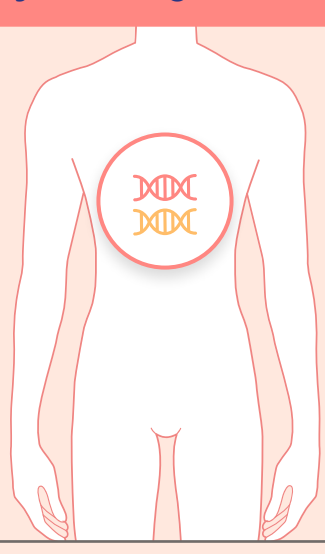


1 in 10,000
live births worldwide[1]

Typically diagnosed in childhood, it is the **leading genetic cause of death in infants**[2]

SMA is caused by a mutation of the survival motor neuron 1 (SMN1) gene, **which leads to a deficiency of SMN protein.**

The body has **two very similar genes** that make SMN protein:[2]

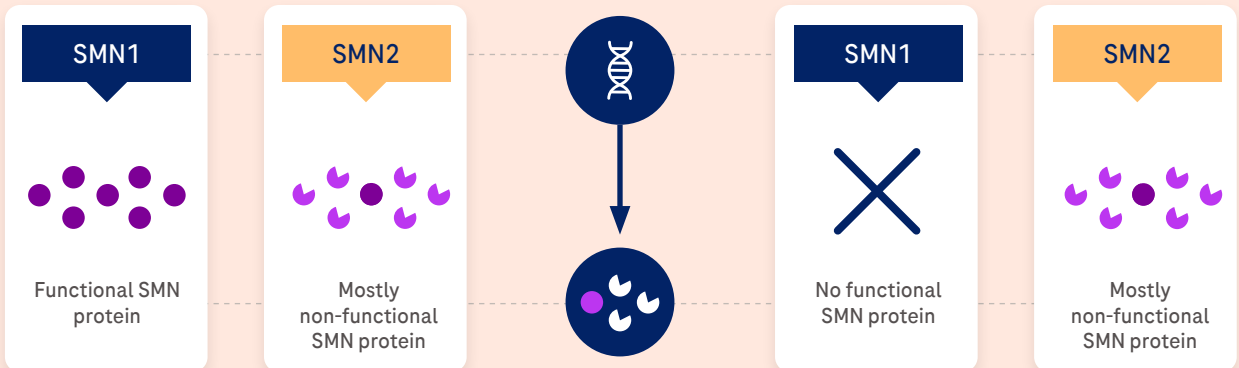


SMN1
produces **all the SMN protein** we need for muscle control and movement[2]

SMN2
only produces around **10%** of the SMN protein we need for muscles to function[2]

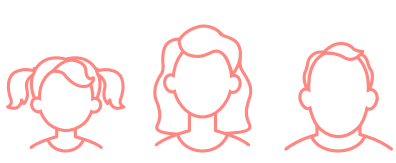
Healthy individuals
The SMN1 gene is able to produce enough protein for the body to function normally

Individuals with SMA
People with SMA must rely on the 'back up' SMN2 gene, which produces insufficient amounts of functional SMN protein[2]



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

~1 in 45 people is a carrier[1]



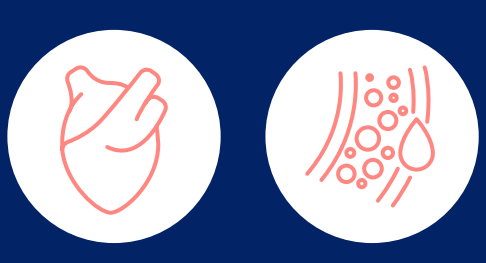
Although SMA is mainly diagnosed in childhood, it can affect people at any age. However, the earlier symptoms appear, the more severe the disease.[2]

An SMA diagnosis must be confirmed through genetic testing. SMA is usually diagnosed after noticing symptoms of SMA, through newborn screening, or via prenatal testing.[3]

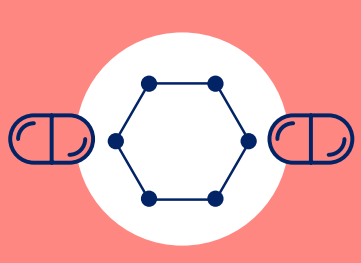
The most common symptoms of SMA, such as progressive muscle weakness, reduced stamina and fatigue, result in:[4]

Trouble breathing, coughing, and swallowing[4,5]

Losing the ability to walk, sit or stand unaided[5]



Ongoing research is exploring how other cells and organs, including the vascular system and heart may also be affected by reduced levels of functional SMN protein.[6]



New approaches to treatment, including combination therapies, are being explored to help muscles grow in size and strength.[7]

References
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6. Simone et al. Cell Mol Life Sci. 2016; 73(5): 1003–1020
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