



The development of Spinal Muscular Atrophy

Spinal muscular atrophy (SMA) affects approximately...



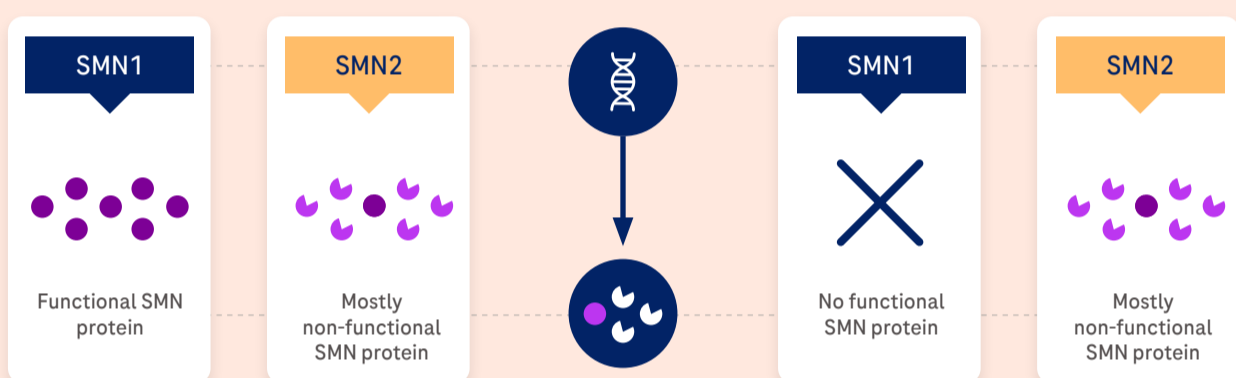
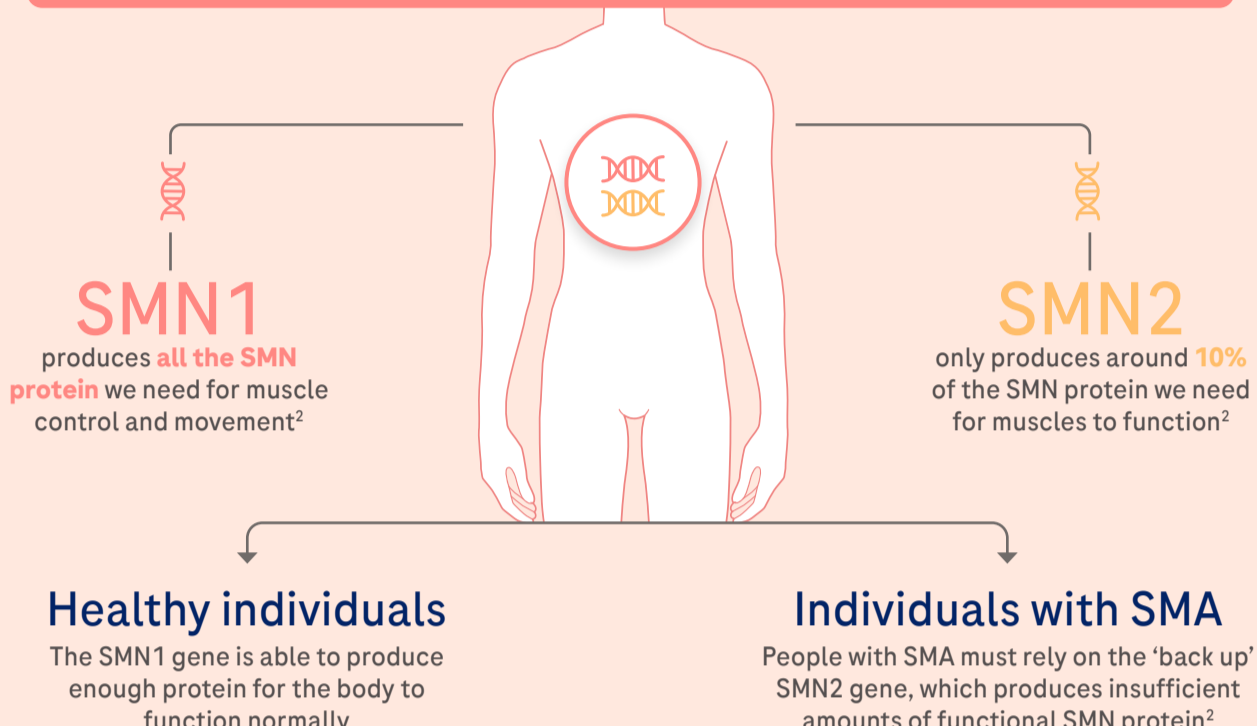
1 in 10,000
live births worldwide¹



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



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 carrier¹



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

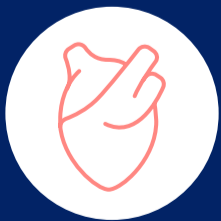
The most common symptoms of SMA, such as progressive muscle weakness, reduced stamina and fatigue, result in:⁴



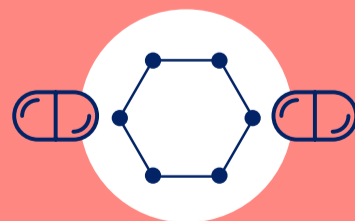
Trouble breathing, coughing, and swallowing^{4,5}



Losing the ability to walk, sit or stand unaided⁵



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
7. Muramatsu H, et al. Scientific Reports. 2021; 11(1)