DROPLET DIGITAL™ PCR (DDPCR™)

BIO-RAD

What *do you* Gene?

Survival Motor Neuron

Essential for Muscle Movement

WHAT IS IT?

The *SMN1* gene provides instructions for making the survival motor neuron (SMN) protein.

A small amount of SMN protein is also produced from **the *SMN2* gene**.

WHAT IS IT?

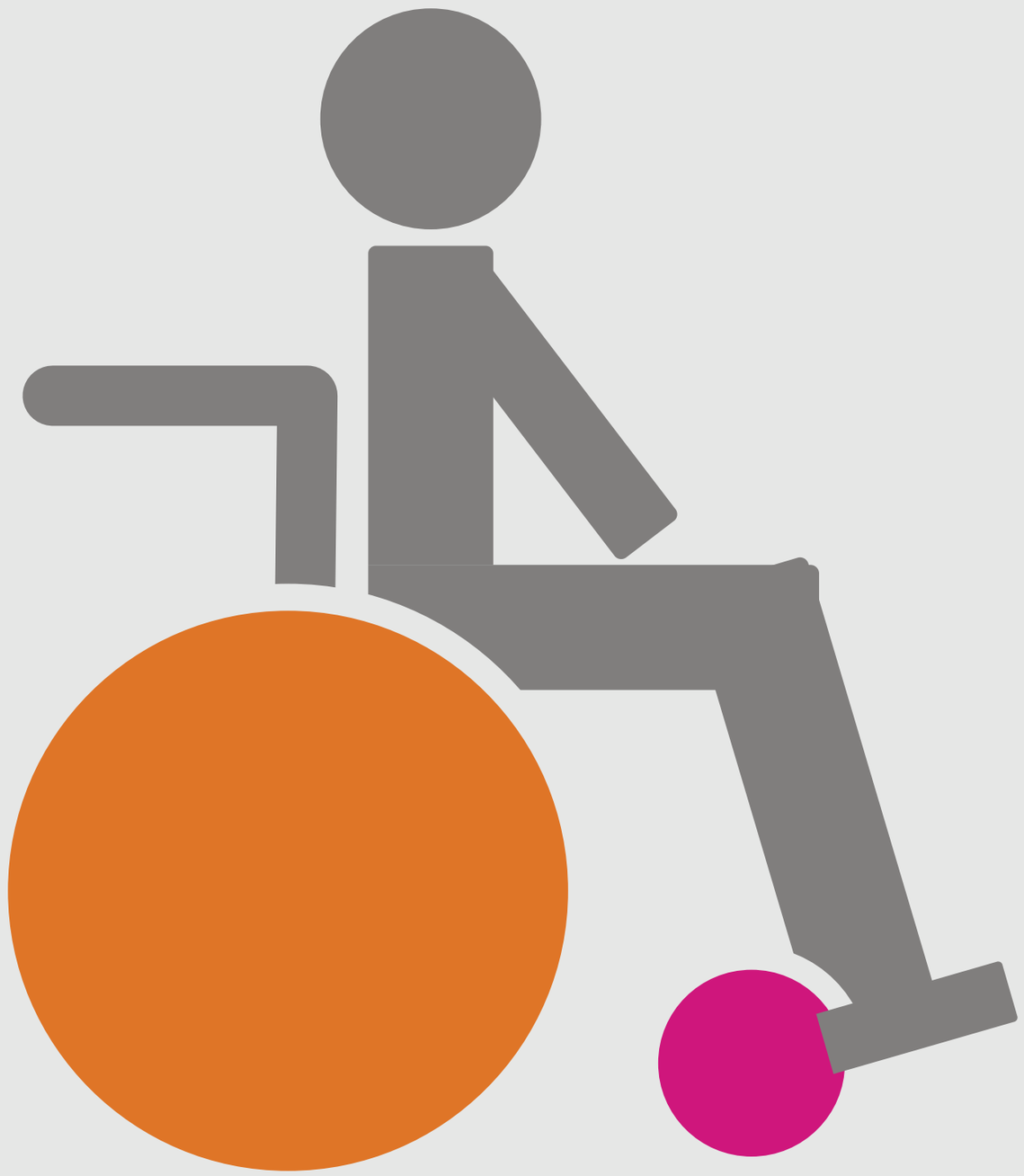
SMN is found throughout the body, with **high levels in the spinal cord and brain**.

WHAT IS IT?

It is particularly important for **the maintenance of specialized nerve cells called motor neurons**, which control muscle movement.

SMN MUTATION AND SPINAL MUSCULAR ATROPHY

Spinal muscular atrophy **is a severe neuromuscular disease** characterized by degeneration of alpha motor neurons in the spinal cord, leading to progressive proximal muscle weakness and paralysis.

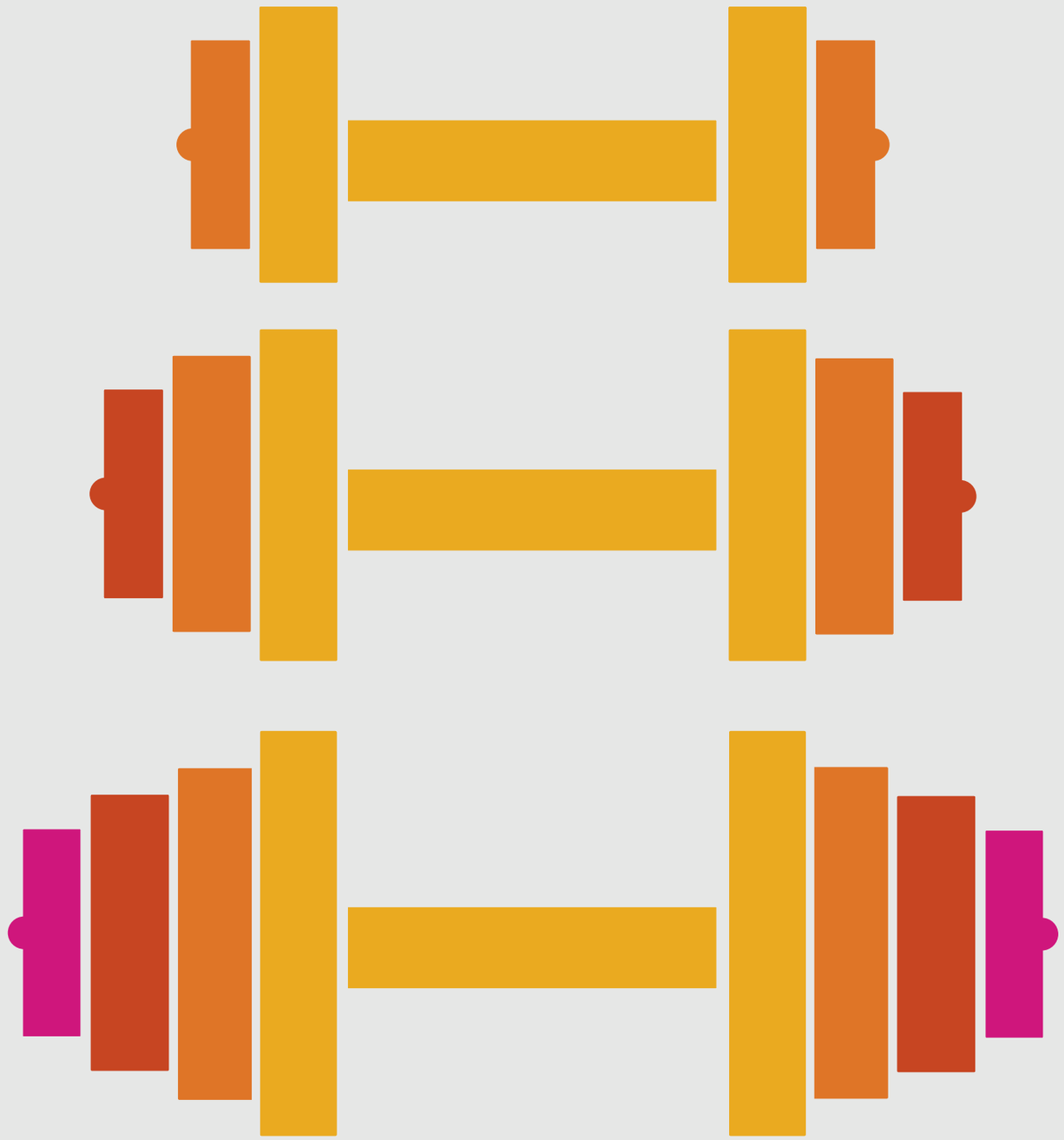


SMN MUTATION AND SPINAL MUSCULAR ATROPHY

It is **caused by a mutation in the *SMN1* gene** that decreases the production of functional SMN protein.

SMN MUTATION AND SPINAL MUSCULAR ATROPHY

More copies of the ***SMN2*** gene results in less severe disease, specifically reduced degeneration of alpha motor neurons and increased muscle strength.



SMN MUTATION AND SPINAL MUSCULAR ATROPHY

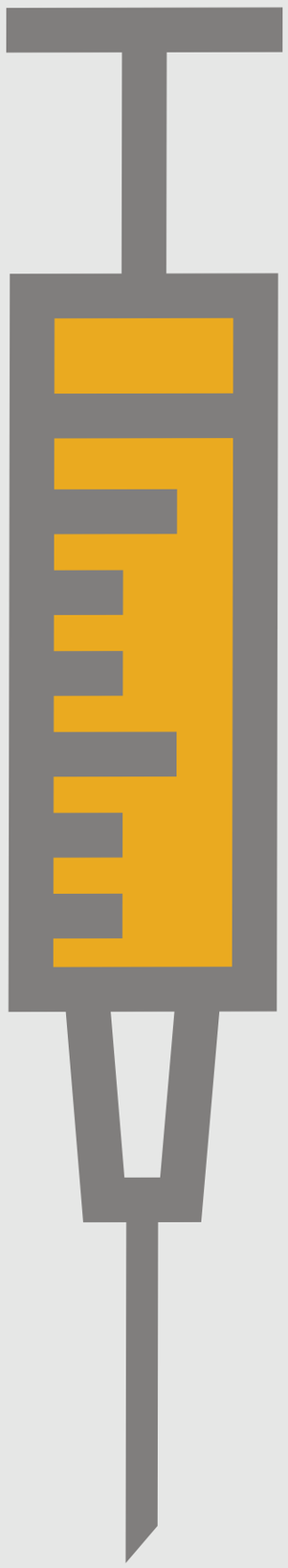
It is the **second most common fatal autosomal recessive disorder,** with an estimated prevalence of 1 in 10,000 live births. It is also the **leading genetic cause of death for children** under two years of age.

DETECTING AND TREATING SPINAL MUSCULAR ATROPHY

The American College of Obstetricians and Gynecologists recommends **screening all pregnant women** for SMN mutations.

DETECTING AND TREATING SPINAL MUSCULAR ATROPHY

There are currently three **FDA-approved therapies for SMA:** Spinraza, an antisense oligonucleotide drug; Zolgensma, a gene therapy; and Evrysdi, an orally-administered mRNA splicing modifier.



DETECTING AND TREATING SPINAL MUSCULAR ATROPHY

The Advisory Committee on Heritable Disorders in Newborns and Children recommends nationwide **newborn screening for spinal muscular atrophy** to help promote life-saving treatments.

SMN1
SMN2

BIO-RAD OFFERS DROPLET DIGITAL PCR (ddPCR) **MN1** AND **SMN2** COPY NUMBER DETERMINATION KITS.

ddPCR is the method of choice for copy number determination.
For research use only.

Visit **bio-rad.com/digital-assays** for more information.

References

Butchbach, M. (2016). Copy number variations in the survival motor neuron genes: Implications for spinal muscular atrophy and other neuro-degenerative diseases. Front Mol Biosci.

American College of Obstetricians and Gynecologists (2022). Carrier screening for spinal muscular atrophy (SMA). https://www.acog.org/-womens-health/faqs/carrier-screening-for-spinal-muscular-atrophy, accessed July 28, 2022.

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