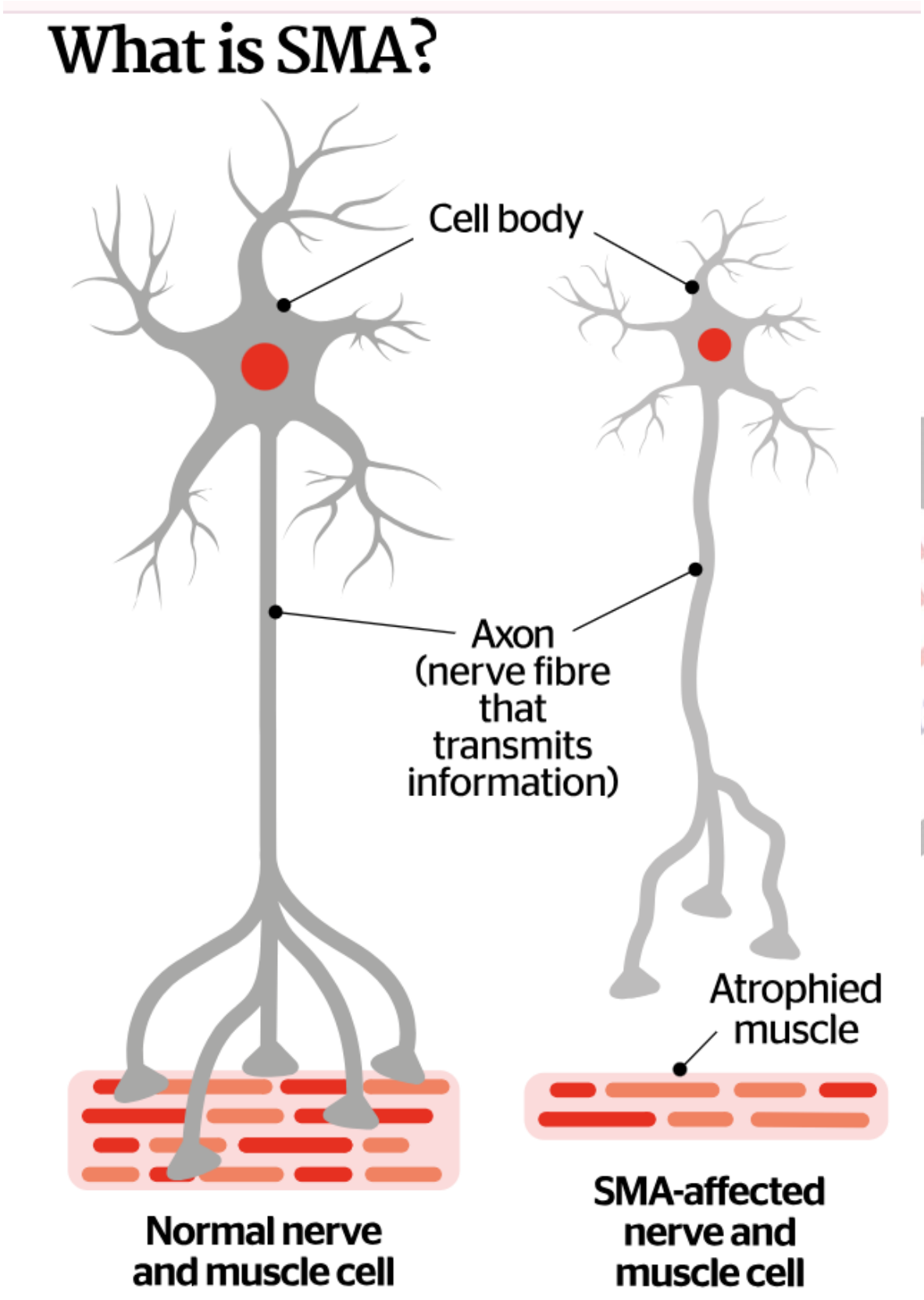
Spinal Muscular Atrophy

Source: TH

In a **first-of-its-kind** medical intervention in India, a **newborn with the SMN1 gene mutation for Spinal Muscular Atrophy (SMA)** is receiving **presymptomatic treatment** using **Risdiplam**, a rare disease-modifying drug given to **prevent motor neuron degeneration.**

Spinal Muscular Atrophy

- **About:** It is a **genetic disorder** caused by an **SMN1 gene mutation** and **protein deficiency**, leading to the **progressive weakening of muscles** due to damage to motor neurons.
    - Genetic disorders are caused by **abnormalities in genes or chromosomes**, either **inherited** or due to **DNA mutations.**
- **Occurrence:** It affects **one in every 10,000 births** and is a **major genetic cause of infant and child mortality.**
- **Gene Transfer:** SMA occurs when **both parents pass on SMN1 gene mutations**, though they are typically **carriers without showing symptoms.**
- **Impact: It mainly affects muscles that fail to receive signals from nerve cells.**
- **Symptoms: It causes weakness in voluntary muscles like the shoulders, hips, and thighs, along with breathing and swallowing difficulties, while involuntary muscles (heart, blood vessels, digestive tract) remain unaffected.**



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