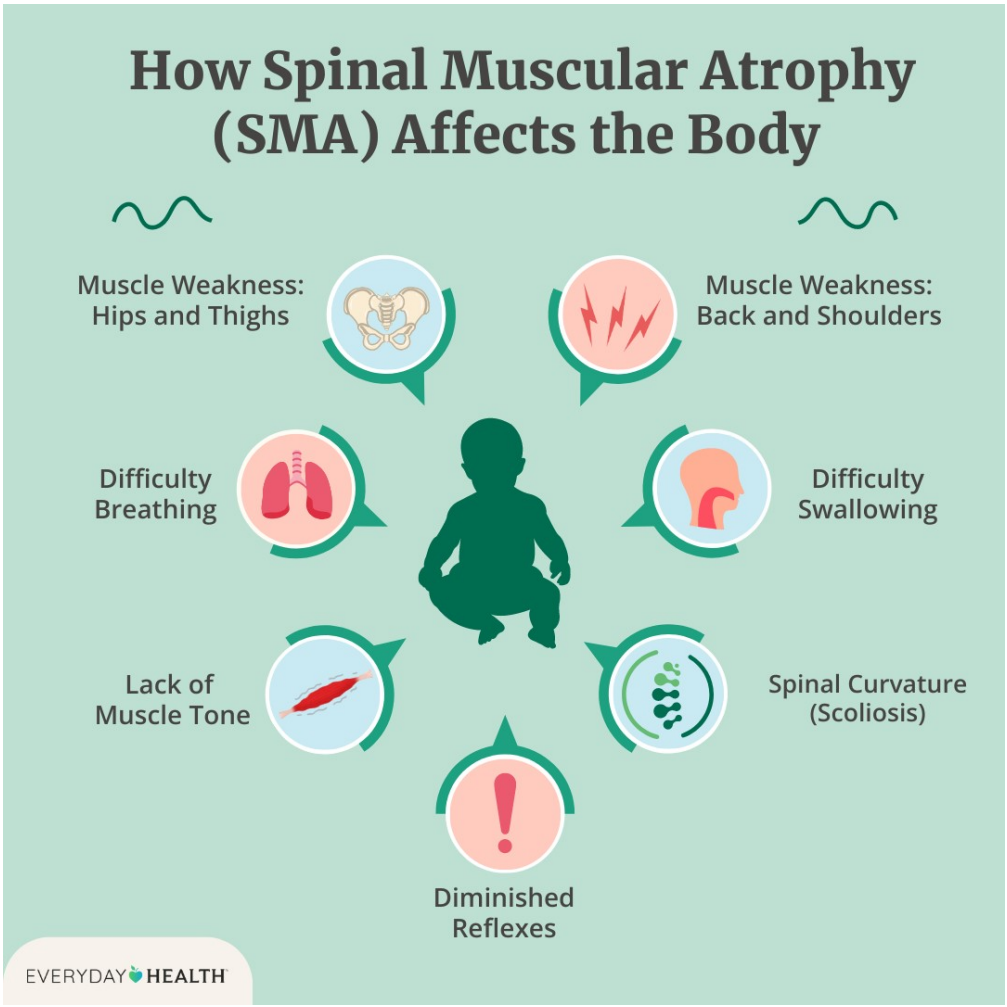
Spinal Muscular Atrophy (SMA)

Why in News?

A two-and-a-half-year-old girl has shown no signs of a genetic disorder, known as spinal muscular atrophy (SMA), becoming the first person in the world to be treated for the disease while in the womb.

- SMA is a debilitating ***genetic condition*** which affects *motor neurons* that control movement, and leads to progressive muscle weakening.
- **Caused by -** Mutations in the survival motor neuron gene (SMN1) which causes a deficiency of a protein crucial for the survival of motor neurons in the spinal cord.
- This prevents muscles from receiving signals from the brain, causing them to waste away.



- **Types of SMA**
  - **Type I** - Usually diagnosed before age 3
  - **Type II** -Begins to affect children between 6–18 months old
  - **Type III** - Also called ***Kugelberg-Welander syndrome*** or juvenile SMA, begins to affect kids as early as 18 months of age or as late as adolescence
  - **Type IV-** The adult form of SMA, symptoms usually begin after age 35.

- In its most severe form, SMA-1, motor skills decline rapidly and patients usually only live two to three years.
- **Prognosis** - About one in every 10,000 births have some form of the condition making it a leading genetic cause of death in infants and children.
- **Recent application in Treatment** - For the treatment, scientists used an oral drug called risdiplam, which is given to patients to slow the progression of SMA.
- *Risdiplam* is typically given to a patient soon after birth.
- The mother, who was 32 weeks pregnant, took Risdiplam daily for 6 weeks.
- The baby started taking the drug from roughly one week old, and will probably continue to take it for the rest of her life.
- The scientists found that the girl had higher levels of the SMN protein in her bloodstream, compared to those usually born with the condition.
- The girl seemed to have lower levels of nerve damage, and even after 30 months had normal muscle development with no sign of atrophy.

Reference

The Indian Express | Spinal Muscular Atrophy (SMA)