Spinal muscular atrophy

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In news- Recently a three-year-old boy from Hyderabad received the single-dose intravenous injection of **Zolgensma, the world's most expensive medical drug**. The drug costs Rs 16 crores and is not available in India.

About the disease Spinal Muscular Atrophy(SMA)-

- SMA is a **life-threatening progressive condition** and Zolgensma is used to cure it.
- It is a genetic **(inherited) neuromuscular disease** that causes muscles to become weak and waste away.
- People with SMA l**ose a specific type of nerve cell in the spinal cord** (called motor neurons) that control muscle movement.
- It's a rare disease that affects one out of 6,000 to 10,000 children.
- A person with SMA inherits two copies of a missing or faulty (mutated) survival motor neuron 1 (SMN1) gene.
- One faulty gene comes from the mother and the other comes from the father.
- An adult can have a single copy of the defective gene that causes SMA and not know it.

Four primary types of SMA:

→ Type 1 (severe): About 60% of people with SMA have type 1, also called **Werdnig-Hoffman disease.** Symptoms appear at birth or within an infant's first six months of life. They don't meet typical milestones like holding up their heads or sitting.

→ Type 2 (intermediate): Symptoms of type 2 SMA (also called **Dubowitz disease**) appear when a child is between six months and 18 months old. This type tends to affect the lower limbs and children with type 2 SMA may be able to sit up but can't

walk.

→ Type 3 (mild): Symptoms of type 3 SMA (also called **Kugelbert-Welander or juvenile-onset SMA**) appear after a child's first 18 months of life. Some people with type 3 don't have signs of disease until early adulthood. Type 3 symptoms include mild muscle weakness, difficulty walking and frequent respiratory infections.

→ Type 4 (adult): The rare adult form of SMA doesn't typically appear until the mid-30s. Muscle weakness symptoms progress slowly, so most people with type 4 remain mobile and live full lives.