Spinal Muscular Atrophy

Source: IE

A 2.5-year-old girl is the first person in the world to receive treatment for **Spinal Muscular Atrophy (SMA)** before birth.

- **About SMA:** It is a **genetic disorder** affecting **motor neurons,** leading to progressive **muscle weakening** due to **SMN1 (survivor motor neuron 1) gene mutation** and protein deficiency.
    - **Occurrence:** It affects **one in every 10,000 births,** making it a leading genetic cause of infant and child mortality.
    - **Gene Transfer:** SMA occurs when both **parents pass SMN1 gene mutations**; they are usually **carriers** without symptoms.
    - **Impact**: It primarily **affects muscles**, which **don't receive signals** from nerve cells.
    - **Symptoms:** Weakness in **voluntary muscles** (shoulders, hips, thighs), respiratory and swallowing difficulties, etc.
- Genetic disorders are medical conditions caused by **abnormalities** in an individual's **genes or chromosomes**, either inherited or caused by DNA mutations.

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