GENETIC DISORDER AND TREATMENT – SCIENCE & TECHNOLOGY

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.*

WHAT'S IN THE NEWS?

Spinal Muscular Atrophy (SMA)

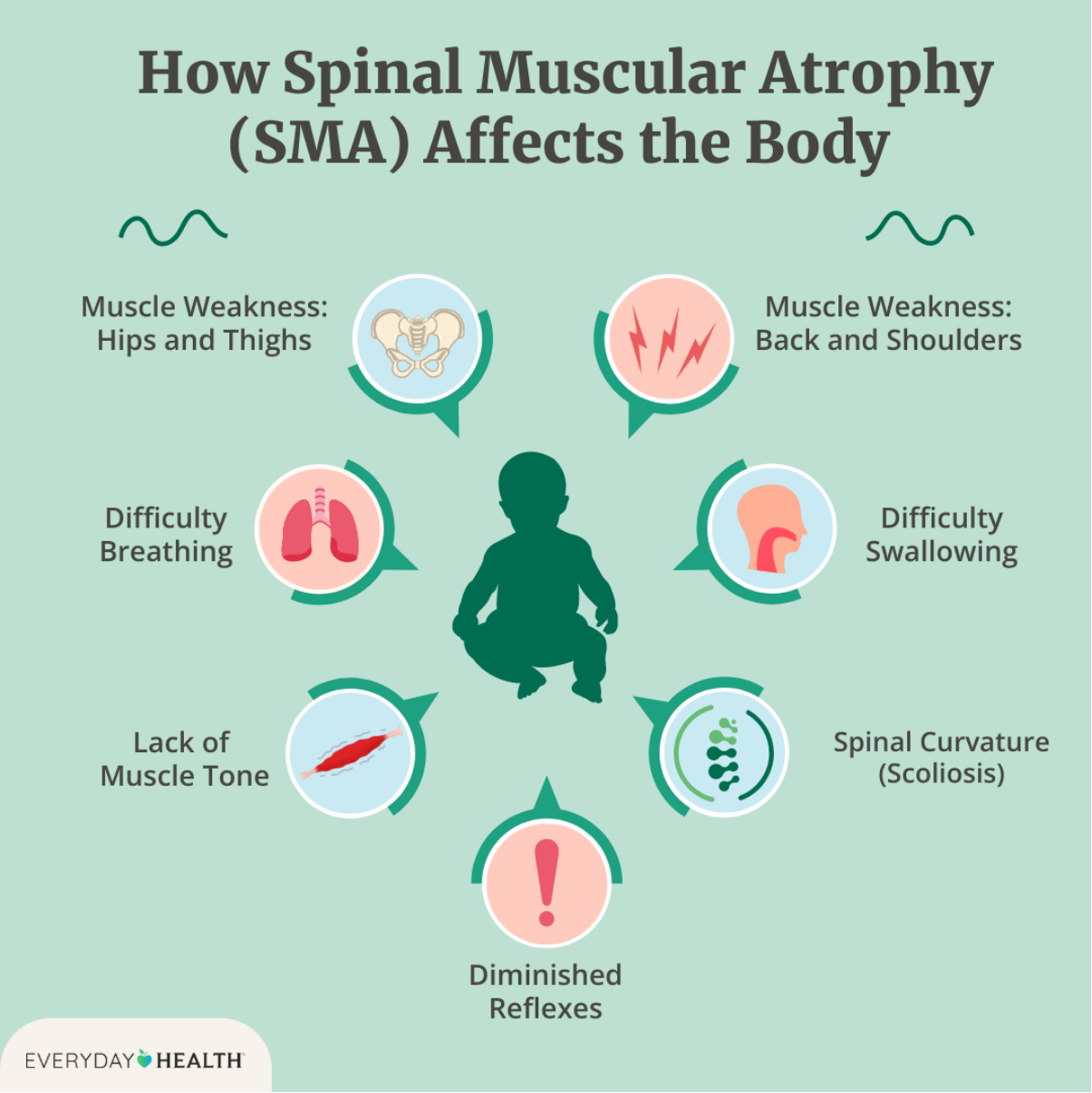
Spinal Muscular Atrophy (SMA) is a **severe genetic neuromuscular disorder** that leads to the **progressive weakening and wasting of muscles** due to the **degeneration of motor neurons** in the spinal cord and brainstem. Motor neurons play a crucial role in controlling voluntary movements such as walking, breathing, swallowing, and other essential muscle functions.

SMA is primarily caused by mutations in the **Survival Motor Neuron 1 (SMN1) gene**, which is responsible for producing **Survival Motor Neuron (SMN) protein**. This protein is essential for the survival and function of motor neurons. When there is a deficiency of SMN protein, motor neurons begin to deteriorate, leading to **severe muscle atrophy and impaired movement**.

Depending on the severity of the condition, individuals with SMA may experience **loss of motor function, difficulty in swallowing and breathing, and in extreme cases, respiratory failure**. SMA is regarded as the **leading genetic cause of death in infants and children**, with **SMA Type 1** being the most severe form. Infants diagnosed with SMA Type 1 typically show symptoms at birth or within the first few months of life, and if left untreated, the condition can lead to death **by the age of 2 to 3 years** due to respiratory complications. The disorder is relatively rare, with an incidence rate of approximately **1 in 10,000 births worldwide**. However, due to advancements in **genetic screening and early treatment options**, the prognosis of SMA patients has improved significantly in recent years.

How Was SMA Treated While in the Womb?

For the first time in medical history, **Spinal Muscular Atrophy (SMA) has been treated in utero (during pregnancy)** as part of an innovative clinical trial. This groundbreaking intervention involved the administration of a **disease-modifying drug called Risdiplam**, which **targets the root cause of SMA** by increasing the production of SMN protein in motor neurons.



Risdiplam is typically prescribed to SMA patients after birth, and research has shown that **the earlier the treatment is initiated, the more effective it is** in preventing irreversible motor neuron loss. In this particular case, the **expectant mother began taking Risdiplam at 32 weeks of pregnancy**, with a **daily dosage for six weeks**. Following birth, the **newborn continued receiving Risdiplam as part of her ongoing treatment regimen**.

The results of this in-utero treatment were remarkably positive:

- The baby showed **higher-than-expected levels of SMN protein**, which is critical for the survival of motor neurons.
- Unlike typical SMA Type 1 patients who display early signs of muscle weakness and atrophy, this child exhibited **normal muscle development** and had **no clinical signs of muscle deterioration**.

- At **30 months of age**, the child continued to show **healthy motor function**, demonstrating that early intervention with Risdiplam could **potentially prevent or significantly delay the onset of SMA symptoms**.

This successful prenatal treatment represents a **major milestone in the management of genetic disorders** and could pave the way for similar early-intervention strategies in the future.

Genetic Disorders

Genetic disorders are medical conditions that result from **mutations in genes or structural abnormalities in chromosomes**. These mutations or alterations can affect how genes function, leading to various inherited or spontaneously occurring diseases.

The **human genome consists of DNA (Deoxyribonucleic Acid)**, which carries **genetic instructions for the synthesis of proteins** responsible for all cellular functions in the body. DNA is inherited from both parents, meaning genetic disorders can be **passed down through generations** or arise due to **spontaneous mutations during embryonic development**. Some genetic disorders manifest **immediately at birth**, while others may only present **symptoms later in life**, depending on the type of genetic mutation involved.

Types of Genetic Disorders

Genetic disorders can be classified into three main categories based on their underlying causes:

1. **Monogenic Disorders (Single-Gene Disorders)**
    - These disorders occur due to mutations in a **single specific gene**.
    - Examples include **Spinal Muscular Atrophy (SMA), Cystic Fibrosis, Sickle Cell Anemia, and Huntington's Disease**.
2. **Multifactorial Disorders (Polygenic Disorders)**
    - These conditions result from **mutations in multiple genes**, often in combination with **environmental factors** such as **diet, exposure to chemicals, smoking, alcohol consumption, or medication use**.
    - Common examples include **Diabetes, Heart Disease, Hypertension, and Certain Types of Cancer**.
3. **Chromosomal Disorders**
    - These disorders arise from **missing, extra, or structurally altered chromosomes**, leading to abnormal gene expression and developmental issues.
    - Examples include **Down Syndrome (Trisomy 21), Turner Syndrome (Monosomy X), and Klinefelter Syndrome (XXY Chromosomal Pattern)**.

Advancements in **genetic research, early screening, and gene therapy** have provided **improved diagnostic and treatment options** for many genetic disorders, offering hope for better management and potential cures in the future.

Source: https://indianexpress.com/article/explained/explained-sci-tech/how-doctors-treated-a-genetic-disorder-in-the-womb-for-the-first-time-9853708/