

# FACTS ABOUT SPINAL MUSCULAR ATROPHY.

## WHAT IS SPINAL MUSCULAR ATROPHY.

Spinal muscular atrophy is an inherent disorder that destroys the motor neurons and the nerve cells that are present in the brain and spinal cord. This further affects activities like speaking, breathing, walking, swallowing, and general muscle weakness. There hence exist 4 types of SMA which include;

- Werdnig-Hoffmann disease that manifests before 6 months of age.
- SMA type 2 (Intermediate form) manifests before 6 and 18 months.
- Kugelberg-Welander disease which manifests after 18 months.
- SMA type IV develops symptoms after 21 years of age.

## HISTORY OF SPINAL MUSCULAR ATROPHY.

- Spinal Muscular Atrophy was first defined by one Guido Werdnig in the year 1891 in two infant brothers then followed by 7 more cases which were realized by Johan Hofmann between 1893 to 1900.
- Their further analysis further found that the disorder affected the motor neuron cells in babies which degenerated in the anterior horn.
- due to their work, the first two types of the disorder were named after them.
- Eric Kugelberg and Lisa Welander later realized the 3rd and 4th types of the disorder and named them after their names.


## STATISTICAL DATA ON SPINAL MUSCULAR ATROPHY.

Research in the United States of America reveals the following data about spinal muscular atrophy.

- Only 1 out of every 6000-10000 live births are affected with spinal muscular atrophy.
- Around a total of 10000-25000 people are believed to be infected with SMA in the USA.
- the muscles that are mostly affected by SMA are those around the center of the body which include around the hips, shoulder, thighs and the upper back.


## SIGNS AND SYMPTOMS OF SPINAL MUSCULAR ATROPHY.

- Curving of the spinal cord.
- limited or difficulty in mobility.
- weakness of the muscles and decreased muscle tone.
- problems with eating and swallowing things.
- trouble with tongue movement or spontaneous tongue movement.
- Breathing problems in infants.



## **DIAGNOSIS OF SPINAL MUSCULAR ATROPHY.**

SMA is diagnosed through a blood test that looks for mutations of a gene known as the SMN1. The test is hence 95% effective at identifying type I, II and III of SMA. The test is also capable of revealing if an individual is simply a carrier of the defective gene that can be passed down the coming generations.



## **TREATMENT OF SPINAL MUSCULAR ATROPHY.**

- There exists a drug; Nusinersen which is injected into the fluid surrounding the spinal cord to enhance the production of full-length SMN proteins thus treat both children and adults.
- Physical, rehabilitation, and occupational therapy can also be done to the affected to prevent immobility as a result of the disorder.

## **HOW PHARMACOGENOMICS IS APPLIED IN THE TREATMENT OF SPINAL MUSCULAR ATROPHY.**

Pharmacogenomics can be defined as how genes affect people's response to different drugs.

Different people have different genes. Similarly, spinal muscular atrophy is treated by drugs like Nusinersen which works by adjusting the SMN2 gene to make it generate more proteins. Zolgensma drug also works by replacing the SMN1 gene in children under 2 years. Moreover, the Risdiplam drug also stops SMN2 genes from stopping protein production. Hence how best to use these drugs could only be realized when people have perfect knowledge of pharmacogenomics.

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