# Nusinersen, Newer Drug for Spinal Muscular Atrophy

**Bijaylaxmi Behera**

Maulana Azad Medical College, India

**Short Communication**

## Abstract

Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disease, with incidence of 1 in 5,000 to 1 in 10,000 live births [1,2]. It is caused by homozygous deletion of exons 7 and 8 in the SMN1 gene [1,2]. It is a rare genetic disease resulting from degeneration of motor neurons in the spinal cord and brainstem [3]. This disease can have onset of signs and symptoms from before birth to early adulthood and manifests with symmetrical, proximal, and progressive muscle weakness and atrophy. Other complications encountered are poor weight gain, restrictive lung disease, scoliosis, joint contractures, and sleep complications, typically without affecting cognition. SMA causes mutation in chromosome 5q11.2-q13.3, affecting the Survival Motor Neuron (SMN) gene, resulting in absence of SMN1 exon 7. SMN1 and SMN2 encode the SMN protein, which is essential for the maintenance of motor neurons. A homozygous deletion of SMN1 exon 7 is confirmatory for the diagnosis of SMA. A typical patient with SMA have zero copies of SMN1 but the number of SMN2 copies are variable [3-6]. Type 0 is associated with prenatal onset and is the severest form of SMA whereas Type 1 is the most common type [4]. SMA types 2 present around 6 to18 months, type 3 after 18 months, and type 4 after 5 years, respectively [7]. The introduction of Nusinersen ,from early infancy has shown improvement in clinical outcomes of SMA Type 1 patients. With open label phase two and phase three trials, this drug has demonstrated efficacy and an improved life expectancy in most of the patients [8].

## Biography

Bijaylaxmi Behera is working at Maulana Azad Medical College, India.

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