SMART MOVES

EARLY ACTION. EARLY TREATMENT. SAVES LIVES.

Spinal Muscular Atrophy

Early Diagnosis and Access to Treatment Saves Lives!

Diagnostic delays in SMA are common. However, early provision of treatment is critical to modifying the rapid and irreversible loss of motor neurons and exponentially increasing chances of survival and functional gains to patients.

In December 2016, Nusinersen (Spinraza ™) was approved to treat all SMA types; other potential treatments are on the horizon. Early treatment has a substantial beneficial impact on clinical outcomes.

WHAT IS SMA?

- Autosomal recessive neuromuscular disease caused by deletion/mutation in SMN 1 gene
- Hypotonia and weakness in the trunk and limbs are the predominant symptoms
- Symptoms present with acute onset and rapid deterioration followed by a prolonged plateau
- One of the most common monogenic disorders in humans

CLINICAL PRESENTATION BY TYPE

SMA is categorized in five types based on age of onset and highest motor milestone achieved. The most common 3 types present as follows:

TYPE I (SEVERE/MOST COMMON FORM)

Symptom onset < 6 Months

- Hypotonia
- Typical supine position is bent elbows and frog legs
- Difficulty lifting extremities against gravity
- Rapid paradoxical breathing or belly breathing
- Bell-shaped chest
- Poor head control/head lag
- Bulbar weakness/dysphagia
- Never sits

TYPE II

Symptom onset occurs from 6 to 18 months

- Hypotonia
- Difficulty lifting extremities against gravity
- Possible bell-shaped chest
- Possible paradoxical breathing or belly breathing
- Tongue fasciculations
- Absent deep tendon reflexes
- Delayed sitting and never walks

TYPE III

Symptom onset occurs from 1.5 to 10 years

- Hypotonia
- Muscle tremor
- Fatigue with exertion and frequent falls
- Gower's sign, Trendelenburg gait, hyperlordosis, and overpronation observed
- Delayed walking

Genetic testing is required to confirm SMA.

RESOURCES

SMA Diagnostic Toolkit: //cureSMA.org/SMArtmoves-toolkit

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