Do any of your patients have SMA?

Spinal muscular atrophy (SMA) is a rare neuromuscular disease that affects a broad range of individuals. For patients with SMA, progression can be continuous and cumulative. Identifying patients with SMA as early as possible is critical.

REFERENCES
11. Butchbach ME. Copy number variations in the survival motor neuron genes: implications for spinal muscular atrophy and other neurodegenerative diseases.
Neuromuscular disorders are a group of diseases characterized by weakness and loss of muscle tissue.

- Because symptoms can present similarly, there is potential for misdiagnosis
- Neuromuscular disorders include muscular dystrophy, myopathies, amyotrophic lateral sclerosis (ALS), and SMA
- There are genetic tests available to confirm an SMA diagnosis

SMA is characterized by the degeneration of motor neurons in the spinal cord and brainstem due to insufficient levels of survival motor neuron protein.

- This leads to skeletal muscle atrophy and general weakness

SMA may present similarly to other neuromuscular disorders. Some patients with SMA may have been misdiagnosed in the past or remain untreated.

Identifying patients with SMA as quickly as possible is critical. SMA affects approximately 9000 individuals in the United States. ~85% of individuals living with SMA are later-onset. Positive results from genetic testing are typically required to initiate treatment. Participation in the SMA Identified Program does not guarantee access to treatment.

### SMA Identified Program

If you suspect SMA, Biogen, together with Invitae, offers no-charge genetic testing for your patients through SMA Identified.

To order a test online, or to learn more about the program, please visit SMAidentified.com/awareness. Always use the partner code “SMA” when filling out the online form.

### SMA Disease Characteristics

#### Infantile-onset SMA (Symptom onset <6 months of age)
- Typically don’t achieve major milestones such as sitting and crawling
- Severe with rapid progression

#### Later-onset SMA (Symptom onset ≥6 months of age)
- Motor function and strength decrease over time
- Variable rate of progressive decline from patient to patient

#### Disease progression

- High mortality within first 2 years of life
- May require ventilation support to assist with breathing
- A leading genetic cause of infant death
- Likely to require assistive devices at some point
- Muscle strength or function loss occurs for many during adulthood, despite apparent plateaus
- May be able to sit or walk independently, but as the disease progresses, can lose those abilities over time

#### Disease impact

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### Questions to ask your patients about their SMA disease progression.

- Have you or your friends and family noticed any changes in your ability to do certain things from last year or even a few months ago?
- Have you noticed you’ve been walking less lately, or doing certain activities slower than before?
- Think back a year, 3 years, 5 years ago. What was your function level then versus now?
- What are your concerns if you remain untreated?
Progressive muscle weakness, motor function impairment, and disability can affect all individuals with SMA.2

The broad range of SMA symptoms includes the following:

- Weakness that is usually symmetrical and more proximal than distal.6,7
- Absent or diminished tendon reflexes7
- Difficulty or inability to walk7
- Respiratory issues that may require tracheostomy or ventilation7

Motor function declines in all individuals with SMA, though the rate and severity vary.2,6

The clinical course of SMA is highly variable. The age at which patients with later-onset SMA develop significant muscle weakness and lose ambulation is unpredictable.15-17

Some Type 3 patients can lose ambulation as adults, 20-30 years after the initial diagnosis.14

Motor function loss is unpredictable.14,15 There is no way to predict when motor function losses will occur or who will experience them.

Some with later-onset SMA may progress slowly.2,3 Declines may not be apparent over the course of a single year, but can become more apparent with longer-term follow-ups.

Loss can occur without clinical symptoms.15,17 The objective decline of muscle strength can be detected in the absence of clinical signs and symptoms.

Adults with later-onset SMA are at risk for progression.2,3,14,16 Despite apparent plateaus, many adults with later-onset SMA may continue to lose muscle strength or function.
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(Using Watermark
Symptom onset >6 months of age)
Later-onset SMA
(Symptom onset <6 months of age)
Infantile-onset SMA

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According to a survey, the majority of individuals with later-onset SMA say stabilization of their current motor function would be a success.

Some of your patients may have SMA and may have been previously classified with these ICD-9/10 codes.

Unlike many other neuromuscular diseases, there is a validated genetic test to confirm SMA.

ICD-9 Diagnosis Codes
- 335.0 Werdnig-Hoffmann disease
- 335.1 Spinal muscular atrophy
- 335.11 Kugelberg-Welander disease
- 335.19 Other spinal muscular atrophy
- 335.21 Progressive muscular atrophy

ICD-10 Diagnosis Codes
- G12.0 Infantile spinal muscular atrophy, Type 1 [Werdnig-Hoffmann]
- G12.1 Other inherited spinal muscular atrophy
- G12.2 Motor neuron disease
- G12.8 Other spinal muscular atrophies and related syndromes
- G12.9 Spinal muscular atrophy, unspecified

Identifying patients with SMA as quickly as possible is critical. According to a survey, the majority of individuals with later-onset SMA say stabilization of their current motor function would be a success.
Identifying patients with SMA as early as possible is critical.²

- All individuals with SMA can experience progressive muscle weakness, motor function impairment, and disability²
- The rate of motor-function decline varies, but affects all individuals with SMA²,⁶
- Some patients with SMA may have been misdiagnosed in the past or remain untreated⁸

Test now if you suspect a patient has SMA. Learn how at SMAidentified.com

REFERENCES