

## Spinal Muscular Atrophy (SMA)

### Use

Qualitative determination of exon 7 of the *SMN1* gene in dried blood spots (DBSs) as an aid in screening newborns for spinal muscular atrophy (SMA).

### Clinical Significance

SMA is a group of inherited (autosomal recessive) neuromuscular conditions that are primarily caused by biallelic mutations of the *SMN1* gene, resulting in deficiency of survival motor neuron (SMN1) protein. Approximately 95% of patients with SMA carry homozygous deletions of exon 7. SMN1 protein plays a major role in the maintenance of motor neurons. The loss of motor neurons leads to progressive muscle weakness and atrophy. The four primary forms of SMA are classified based on clinical severity and age of onset. In general, forms of SMA with an earlier age of onset are more severe and have a greater impact on motor function.

- *SMA type I* is the most common and severe form. Signs and symptoms often begin within the first six months of life. Severe muscle weakness and poor muscle tone leads to significant development delay. Most affected infants are unable to support their heads or sit unassisted. Other signs include breathing problems, difficulty swallowing, poor growth, and joint abnormalities.
- *SMA type II* is generally characterized by muscle weakness that develops between six months and two years of age. Affected individuals can typically maintain a seated position but are unable to walk.
- *SMA type III* is often diagnosed between 18 months and three years of age, with some not developing muscle weakness until adolescence. Affected individuals can stand and walk independently but have increasingly limited mobility with age.
- *SMA type IV* is characterized by mild to moderate symptoms that usually don't develop until adulthood. Mild motor impairment such as gradual muscle weakness, tremor, twitching, and mild breathing problems are common symptoms.

Further information and ACT Sheets can be found at the OSDH Newborn Screening Program [website](#).

### Methodology

Real-time, polymerase chain reaction (qPCR) amplification for qualitative detection of exon 7 in the *SMN1* gene.

### Specimen Type

See [Guidance for Collection of NBS Dried Blood Spots](#)

### Minimum Volume/Size

See [Guidance for Collection of NBS Dried Blood Spots](#)

### Collection Instructions

See [Guidance for Collection of NBS Dried Blood Spots](#)

### Common Causes for Rejection

See [Guidance for Collection of NBS Dried Blood Spots](#)

### Shipping

See [Guidance for Collection of NBS Dried Blood Spots](#)

**Turn-around Time**

Within 5 working days of receipt

**Reference Range**

- Within Normal Limits: Not consistent with SMA

**Reportable Results**

- Within Normal Limits
- Outside Normal Limits
- Unsatisfactory

**Interpretation**

- Within Normal Limits
  - *SMN1* ex7 Detected: Not consistent with SMA
- Outside Normal Limits
  - *SMN1* ex7 Not Detected: Consistent with Spinal muscular atrophy; immediate confirmatory testing required
- Unsatisfactory
  - Poor DNA amplification; submit repeat specimen as soon as possible

**Limitations/Interferences**

- This is a screening test only. A diagnostic procedure should be used to confirm a diagnosis of SMA.
- Do not use heparinized tubes or capillaries to collect blood; heparin is a known inhibitor of PCR.
- Specimens improperly collected, processed or transported may result in erroneous results.

**CPT Code**

81479

**Notes**

This is a laboratory-developed test; performance characteristics have been validated and determined to be suitable for diagnostic purposes by the OSDH PHL. This test has not been cleared or approved by the U.S. Food and Drug Administration.