Spinal Muscular Atrophy: *SMN1* Common Deletion Testing

<table>
<thead>
<tr>
<th>Test Code:</th>
<th>SM</th>
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<td>Turnaround time:</td>
<td>2 weeks</td>
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<td>CPT Codes:</td>
<td>81400 x1</td>
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**Condition Description**

Spinal muscular atrophy (SMA) is the second most common lethal, autosomal recessive disorder in Caucasians. SMA is characterized by anterior horn cell degeneration which causes a symmetrical muscle weakness and wasting. Three types of SMA are described:

- **Type I (Werdnig-Hoffman disease):** most severe form of SMA with an onset of symptoms before 6 months of age; affected individuals usually die by 2 years of age.
- **Type II (intermediate form):** intermediate in severity between Types I and III, with an onset of symptoms between 6 and 18 months of age; death occurs after 2 years of age.
- **Type III (Wohlfart-Kugelberg-Welander disease):** mildest form of childhood onset SMA, with symptoms beginning between 18 months and 17 years of age; affected individuals survive into adulthood.

All three SMA types are linked to chromosome 5q11.2-q13.3. A telomeric gene, known as the *survival motor neuron (SMN1)* gene, was found to be deleted in about 98% of patients. Point mutations are also known in this gene.

*SMN1* is deleted in about 95% of individuals with SMA. This assay tests for the common *SMN1* deletion only; other pathogenic variants will not be detected. Note that approximately 5-8% of individuals that are carriers of SMA carry two copies of *SMN1* on one chromosome with a deletion on the second chromosome, and will not be detected with this assay. This assay will not report *SMN2* copy number.

Please [click here](#) for the GeneReviews clinical summary on this condition.

Visit [www.ThinkGenetic.com](http://www.ThinkGenetic.com) for patient-friendly information on *spinal muscular atrophy*.

**Genes**

*SMN1*

**Indications**

**Methodology**

*SMN1* gene deletions were quantified by multiplex ligation polymerase chain reaction amplification (MLPA) of exons 7 and 8. Gene dosage ratios of *SMN1* are calculated relative to the average of 16 reference loci and are expressed as gene dosage, and/or copy number, according to the SALSA protocol available from MRC Holland. Two copies of the *SMN1* genes most often indicate normal (not affected) status and one copy of a deletion of this region most likely indicates carrier status.

**Detection**

Deletions of the *SMN1* gene are found in approximately 95% of individuals with SMA. This carrier assay tests for the common *SMN1* deletion only; other pathogenic variants will not be detected. Approximately 5-8% of carrier individuals will have a normal *SMN1* copy number of two, but both copies will be on the same chromosome (in cis) with a deletion on the second chromosome. This assay will not detect these carrier individuals. *SMN2* copy number is not assessed.

**Reference Range**

Qualitative assay.

**Specimen Requirements**

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.
Diagnostic testing only. No prenatal or carrier testing available.

### Related Tests

- Congenital Hypotonia Panel