

## Carrier Screening ACT Sheet

# Spinal Muscular Atrophy (SMA)

**Carrier Screening:** Carrier screening for Spinal Muscular Atrophy (SMA) is offered preconceptionally or prenatally to identify individuals at increased risk at having a child with SMA. The disorder is caused by a missing or abnormal gene known as the survival motor neuron gene (*SMN1*). Extra copies of the related gene, *SMN2*, modify the severity of SMA. Carrier screening does not identify all mutations present in individuals.

**Condition Description:** Spinal muscular atrophy (SMA) is an autosomal recessive, neurodegenerative disorder resulting in progressive muscle weakness, atrophy and paralysis. The carrier rate ranges from 1/35 to 1/117 depending upon ethnicity. SMA cases may be divided into three clinical categories on the basis of age of onset and the clinical course of the disease: Type I SMA (Werdnig-Hoffman) characterized by severe, generalized muscle weakness and hypotonia at birth or within the first three months of life. Death from respiratory failure usually occurs before age 2 years. Children with Type II SMA (Intermediate Form) are able to sit unassisted but cannot stand or walk without aid. These children often survive beyond age 4. Type III SMA (Kugelberg-Welander) is milder than the other forms. Onset may be in infancy or childhood, but the affected individual learns to walk unaided.

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### ***YOU SHOULD TAKE THE FOLLOWING ACTIONS:***

- Inform individual of the carrier screening result.
- Refer for genetic counseling.
- Offer partner testing for *SMN* mutations.

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**Reproductive Implications:** When an individual is found to be a carrier of SMA, the reproductive partner should be offered carrier screening for SMA. If both are carriers, the risk of the offspring having the disorder is 1 in 4 (25%); and prenatal testing should be offered. In some ethnic groups, and in some individuals, there can be significant residual risk for an affected offspring, even with a negative carrier screening result. Individuals should be offered pre- and post-screening counseling.

### **Additional Information:**

[Claire Altman Heine Foundation](#)  
[ACMG Practice Guideline](#)

### **Referral (local, state, regional and national):**

[Testing](#)  
[Clinical Services](#)  
[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site (insert state newborn screening program website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

Local Resource Site (insert local and regional newborn screening website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

Claire Altman Heine Foundation

<http://www.clairealtmanheinefoundation.org/>

ACMG Practice Guideline

[http://www.acmg.net/AM/Template.cfm?Section=Practice\\_Guidelines&Template=/CM/ContentDisplay.cfm&ContentID=3979](http://www.acmg.net/AM/Template.cfm?Section=Practice_Guidelines&Template=/CM/ContentDisplay.cfm&ContentID=3979)

Referral (local, state, regional and national):

Testing

[http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical\\_disease\\_id/2256?db=genetests&country=United%20States](http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2256?db=genetests&country=United%20States)

Clinical Services

<http://www.genetests.org/>

Find Genetic Services

<http://www.acmg.net/gis>

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