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.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

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

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

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

LOCAL RESOURCES: Insert State newborn screening program web site links

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

Name	
URL	
Comments	

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

Name	
URL	
Comments	

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

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

Clinical Services
http://www.genetests.org/

Find Genetic Services
http://www.acmg.net/gis

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