Spinal Muscular Atrophy (SMA) Carrier Screen

Carrier testing for (Spinal Muscular Atrophy) SMA is offered to asymptomatic individuals with a confirmed or suspected family history of SMA, reproductive partner of known SMA carrier, and/or parents of a child with a deletion of the SMN1 gene.

Testing Method and Background

This test utilizes Multiplex ligation probe (MLPA) assay (MRC-Holland, www.mlpa.com) complements sequence analysis for detection of large genomic rearrangements. The MLPA probemix included in this assay: SALSA MLPA P060-B2 SMA assay for the detection of copy number changes of exons 7 and 8 of SMN1 and SMN2 for patient diagnosis and carrier testing of spinal muscular atrophy (SMA). **Only SMN1 results are reported.**

Spinal muscular atrophy (SMA) is a neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. SMA is inherited in autosomal recessive manner. In most cases, both parents must be carriers of the condition in order to have a child with SMA. The estimated incidence of SMA is 1:6,000 to 1:10,000. Two highly similar genes play a role in SMA: SMN1 and SMN2. Most individuals have two copies of each gene. The SMA region on 5q13.2, containing the telomeric SMN1 and the centromeric SMN2, is a complicated repeat area displaying high instability, leading to frequent deletions and gene conversions. SMA is caused by low levels of SMN protein essential for survival of motor neurons. Majority of SMN protein production comes from SMN1. Increased SMN protein production from SMN2 may partially compensate for SMN protein production missing due to altered SMN1 gene.

Highlights of Spinal Muscular Atrophy (SMA) Carrier Screen

Targeted Region

SMN1: Copy number changes in exons 7 + 8

Ordering Information

Get started (non-HFHS): Print a Genetic Hereditary Disorder requisition form online at www.HenryFord.com/HFCPD

Get started (HFHS): Order through Epic using test "SMA" (DNA2100030)

Specimen requirements:

- Peripheral Blood - 1-3ml in lavender top tube (EDTA) **Specimen sptability: Ambient - 72 hours; Refrigerated - 1 week**
- Extracted DNA - from a CLIA-certified Laboratory

Cause for Rejection: Clotted, hemolyzed, or frozen specimens, improper anticoagulant, tubes not labeled with dual patient identification, non-dedicated tubes.

TAT: 5-7 business days (after Prior Authorization obtained)

Mail test material to:
Henry Ford Center for Precision Diagnostics
Pathology and Laboratory Medicine
Clinic Building, K6, Core Lab, E-655
2799 W. Grand Blvd., Detroit, MI 48202

CPT Codes: 81329, G0452

Contact us: Client Services, Account and Billing Set-up, and connect with a Molecular Pathologist at (313) 916-4DNA (4362)

For more information on Comprehensive Molecular Services, visit our website
www.HenryFord.com/HFCPD

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