
Lab Dept: Anatomic Pathology

Test Name: SPINAL MUSCULAR ATROPHY DIAGNOSTIC

General Information

Lab Order Codes: SMAE

Synonyms: SMA; Werdnig-Hoffmann disease (SMA type 1); Kugelber-Welander (Type III); Proximal Spinal Muscular Atrophies.

CPT Codes: 81400 – Molecular Pathology procedure, Level 1

Test Includes: SMA type I, SMA type II, SMA type III.

Logistics

Test Indications: Spinal Muscular Atrophy is one of the most common genetic causes of death in childhood. It is inherited as an autosomal recessive gene, with an estimated carrier frequencies of 1 in 40-60 births. SMA is characterized by degeneration and loss of lower motor neurons. The gene responsible for SMA is on the long arm of chromosome 5(1,2). Up to 95% of all patients with SMA types 1 and 2 have a homozygous deletions SMN exon 7. Approximately 85% of those individuals with the less severe form of SMA (type 3) have a deletion in this region. Although the exact functional role of the SMA gene is unknown, detection of the absence of both copies of exon 7 of this gene can be used to confirm the SMA diagnosis in the majority of case.

Lab Testing Section: Anatomic Pathology - Sendouts

Referred to: Ohio State University – Molecular Pathology Laboratory (Thomas Prior's Lab) Test SMAMUT.

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Monday – Thursday by 1500

Turnaround Time: Within 14 days

Special Instructions: Complete [Requisition and Consent forms](#). Submit completed forms with the specimen to the laboratory. ** Draw Monday – Thursday by 15:00 only

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	5 mL blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture, invert specimen gently to mix.
Special Processing:	Lab Staff: Do Not centrifuge. Specimen should be sent in original collection container as whole blood. Specimens may be refrigerated up to 3 days prior to shipping. Ship at ambient temp on MON-THURS via overnight courier to arrive at Ohio State Pathology Lab on a weekday. Label tubes with patient's name, date of birth, date and time of collection. Ship to: Molecular Pathology 680 Ackerman Road, Room 449 Columbus, OH 43202
Patient Preparation:	None
Sample Rejection:	Unlabeled specimens; incorrect tube; mislabeled or unlabeled specimens

Interpretive

Reference Range:	Interpretive report
Critical Values:	N/A
Limitations:	N/A
Methodology:	Polymerase Chain Reaction (PCR)
References:	Ohio State University – The Molecular Pathology Laboratory July 2012 614-366-4557 Fax: 614-366-4589
Updates:	2/11/2013: CPT update