ARUP GENETICS HOT LINE NEWSLETTER

08.2016

ARUP adds new tests to our genetics test menu quarterly. The test list below is orderable as of August 15, 2016, unless otherwise specified.

Visit the ARUP Genetics website at **www.aruplab.com/genetics** to view our complete test menu and for additional resources, including:

- Letters of medical necessity for genetic testing
- Free webinars on genetic topics
- Access to genetic disease databases that contain variant classification and clinical information in one location.

NEW TESTS

TEST CODE	TEST NAME
2013518	**Fatty Acids Profile, Essential Serum or Plasma**

ARUP is now offering Fatty Acids Profile, Essential Serum or Plasma, by quantitative GC/MS and stable isotope dilution as an in-house assay. This test is used for identifying patients with essential fatty acid deficiency, evaluating nutritional status and diet, and monitoring treatment of patients diagnosed with essential fatty acid deficiency.

2013399	**Hemoglobin S, Sickle Solubility**

Apolipoprotein E (*APOE*) Genotyping, Cardiovascular Risk is performed by PCR and fluorescence monitoring using hybridization probes. Hyperlipoproteinemia III (HPL III) is characterized by increased cholesterol and triglyceride levels, presence of B-VLDL, xanthomas, and premature vascular disease, including coronary heart disease (CHD) and peripheral artery disease. *APOE* Genotyping, Cardiovasuclar Risk can be used to identify a cause for HLP III or premature CHD and to screen individuals with a family history of HLP III or premature CHD.

continued

Visit www.aruplab.com/genetics for continuing updates, important announcements, and further information.

ARUP LABORATORIES | Genetics

A NONPROFIT ENTERPRISE OF THE UNIVERSITY OF UTAH AND ITS DEPARTMENT OF PATHOLOGY

500 Chipeta Way | Salt Lake City, UT 84108-1221
Phone: (800) 522-2787 | Fax: (801) 583-2712
www.aruplab.com | www.arupconsult.com

2013436 Spinal Muscular Atrophy (SMA) Copy Number Analysis

2013444 Spinal Muscular Atrophy (SMA) Copy Number Analysis, Fetal

ARUP is pleased to offer Spinal Muscular Atrophy (SMA) Copy Number Analysis by MLPA. This new test can serve as a screen for prenatal or preconception carrier screening for SMA in the general population, and can confirm a suspected diagnosis of SMA. SMA copy number analysis is also suggested for carrier screening of the reproductive partner of a known SMA carrier, or screening for parents of a child with a deletion of the *SMN1* gene or other family history of SMA. This test includes two SNPs sometimes associated with the occurrence of two copies of *SMN1* on the same chromosome. Ashkenazi Jewish and Asian individuals who have two copies of *SMN1* and are positive for these SNPs are predicted to be silent carriers, although this cannot be definitively determined by this assay.

Spinal Muscular Atrophy (SMA) Copy Number Analysis, Fetal is used as prenatal diagnostic testing for spinal muscular atrophy when both parents carry a deletion of *SMN1* or have a previous child with SMA caused by a deletion of *SMN1*.

2002064 Chimerism, Post-Transplant, Sorted Cells

Chimerism, Post-Transplant, Sorted Cells has a revised sorted cell form called Cell Isolation Request for Chimerism, Post-Transplant, Sorted Cells. Please follow the links to the test directory or to the form for the updates.

Visit www.aruplab.com/genetics
for continuing updates, important announcements, and further information.

ARUP LABORATORIES | Genetics

A NONPROFIT ENTERPRISE OF THE UNIVERSITY OF UTAH AND ITS DEPARTMENT OF PATHOLOGY

500 Chipeta Way
Salt Lake City, UT 84108-1221
Phone: (800) 522-2787
Fax: (801) 583-2712
www.aruplab.com
www.arupconsult.com