Expanded Carrier Screening

QUICK FACTS

ARUP LABORATORIES

WHAT IS EXPANDED CARRIER SCREENING?

Expanded carrier screening is a blood analysis that detects carrier status of 100+ recessive genetic diseases. It includes diseases endorsed by the American Congress of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG), such as cystic fibrosis, spinal muscular atrophy and Tay-Sachs disease. It also includes more genetic diseases that have traditionally been precluded from population carrier screening such as Smith Lemli Opitz because of cost-prohibitive screening methodologies.

WHAT ARE THE TESTING OPTIONS FOR EXPANDED CARRIER SCREENING?

ARUP has partnered with Counsyl, the leader in expanded carrier screening, to offer customers the choice of two expanded carrier screening panels (see disease list for all conditions and number of mutations). Fragile X can be added to both panels.

1. UNIVERSAL PANEL – 101 diseases

2. PRENATAL PANEL – 83 diseases – CVS/Amnio available for all diseases on this panel

WHAT IS THE ANALYTICAL VALIDITY OF THE TEST?

The analytical performance of the test has been rigorously confirmed in a publication by Counsyl and by three external regulatory agencies: CLIA, CAP, and NYS CLEP. The latter accreditation is particularly noteworthy, as NYS holds genetic testing laboratories to the most rigorous validation standards. For reference, the CLIA license number is 05D1102604, the CAP accreditation number is 7519776 (AU-ID: 1586635), and the NYS CLEP PFI is 8535.

WHAT IS THE CLINICAL VALIDITY OF THE TEST?

The Counsyl platform detects over 400 clinically significant mutations with at least the same level of accuracy and precision as current testing methodologies. Each mutation is backed by one or more publications supporting an established clinical phenotype, to the extent that genotype-phenotype correlations are possible. This is consistent with ACMG's Category 1 variant classification system. Likewise, the screening methodology for spinal muscular atrophy is consistent with the 2011 ACMG guidelines, and produces results equivalent to standard gene dosage quantification for the SMN1-specific region of exon 7. For diseases on the panel that are traditionally ethnic-specific, founder mutations are tested with detection rates comparable to those reported by other CLIA-approved laboratories.

DOES INSURANCE COVER THE TEST?

Most insurance plans that cover the cost of cystic fibrosis testing will also cover the cost of expanded carrier screening. Reimbursement may vary based on particular fee schedules. A list of billable CPT codes is available on the ARUP technical bulletin.

180 Kimball Way | South San Francisco, CA 94080
www.counsyl.com | support@counsyl.com | (888) COUNSYL

Counsyl