HOTLINE: Effective August 15, 2022

New Test **3004778** **Fetal Aneuploidy Screening with 22q11.2 Microdeletion** **FAS 22**

Methodology: Targeted Sequencing with SNPs
Performed: Sun-Sat: Varies
Reported: 12-14 days

Specimen Required: Collect: Maternal whole blood in Cell-Free DNA BCT tube. A kit must be ordered prior to specimen collection (ARUP Supply #50223) available online through eSupply using ARUP Connect™ or by contacting ARUP Client Services at (800) 522-2787.
Specimen Preparation: Transport 20 mL maternal blood in Cell-Free DNA BCT tube. (Min: 16 mL)
Storage/Transport Temperature: Room temperature.
Remarks: Patient history form is required prior to testing.
Stability (collection to initiation of testing): Ambient: 5 days; Refrigerated: Unacceptable; Frozen: Unacceptable

Reference Interval: By report

Interpretive Data:
Refer to report.

Note: Testing utilizes a single-nucleotide polymorphism (SNP)/informatics-based approach to detect fetal copy number for the five chromosomes responsible for most live-birth aneuploidies (chromosomes 13, 18, 21, X, Y, and triploidy) and certain specific microdeletion syndromes (see current list of microdeletion syndromes listed under "Ordering Recommendations"). This is a screening test to help identify fetuses at risk for Down syndrome, trisomy 18, trisomy 13, and Turner syndrome, as well as fetuses affected with the specified microdeletion syndromes listed. Test should not be considered diagnostic. All positive results should be confirmed by amniocentesis or CVS.

CPT Code(s): 81420; 81422

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.