HOTLINE: Effective September 7, 2021

MEDICARE COVERAGE OF LABORATORY TESTING

Please remember when ordering laboratory tests that are billed to Medicare/Medicaid or other federally funded programs, the following requirements apply:

1. Only tests that are medically necessary for the diagnosis or treatment of the patient should be ordered. Medicare does not pay for screening tests except for certain specifically approved procedures and may not pay for non-FDA approved tests or those tests considered experimental.
2. If there is reason to believe that Medicare will not pay for a test, the patient should be informed. The patient should then sign an Advance Beneficiary Notice (ABN) to indicate that he or she is responsible for the cost of the test if Medicare denies payment.
3. The ordering physician must provide an ICD-10 diagnosis code or narrative description, if required by the fiscal intermediary or carrier.
4. Organ- or disease-related panels should be billed only when all components of the panel are medically necessary.
5. Both ARUP- and client-customized panels should be billed to Medicare only when every component of the customized panel is medically necessary.
6. Medicare National Limitation Amounts for CPT codes are available through the Centers for Medicare & Medicaid Services (CMS) or its intermediaries. Medicaid reimbursement will be equal to or less than the amount of Medicare reimbursement.

The CPT Code(s) for test(s) profiled in this bulletin are for informational purposes only. The codes reflect our interpretation of CPT coding requirements, based upon AMA guidelines published annually. CPT codes are provided only as guidance to assist you in billing. ARUP strongly recommends that clients reconfirm CPT code information with their local intermediary or carrier. CPT coding is the sole responsibility of the billing party.

The regulations described above are only guidelines. Additional procedures may be required by your fiscal intermediary or carrier.

Hotline Page #	Test Number	Summary of Changes by Test Name	Name Change	Methodology	Performed/Reported Schedule	Specimen Requirements	Reference Interval	Interpretive Data	Note	CPT Code	Component Change	Other Interface Change	New Test	Inactive
3	0051265	Achondroplasia (*FGFR3*) 2 Mutations, Fetal			x	x								
4	0091328	Acyclovir, Serum or Plasma		x		x								
4	3003656	Alpha Thalassemia (*HBA1* and *HBA2*) Deletion/Duplication with reflex to Hb Constant Spring, Fetal				x								
4	2008682	Anabolic Steroids, Urine - Screen with Reflex to Confirmation								x				
4	2012232	Angelman Syndrome and Prader-Willi Syndrome by Methylation-Sensitive PCR, Fetal			x	x								
5	0050388	Beta Globin (*HBB*) Sequencing, Fetal				x								

Hotline Page #	Test Number	Summary of Changes by Test Name	Name Change	Methodology	Performed/Reported Schedule	Specimen Requirements	Reference Interval	Interpretive Data	Note	CPT Code	Component Change	Other Interface Change	New Test	Inactive
5	3000480	Comprehensive Systemic Sclerosis Panel					x					x		
6	3002463	Connective Tissue Disease First Line Panel with Reflex					x					x		
7	0051668	Connective Tissue Diseases Profile		x			x					x		
8	3003648	COVID-19 IgG (Spike), Semi-Quantitative by CIA				x								
8	2013562	C-Peptide, 120 Minutes				x								
8	2013564	C-Peptide, 180 Minutes				x								
8	2013558	C-Peptide, 30 Minutes				x								
8	2013560	C-Peptide, 60 Minutes				x								
9	3000529	C-Peptide, Other				x		x						
9	2011231	Duchenne/Becker Muscular Dystrophy (*DMD*) Deletion/Duplication, Fetal				x								
10	3001781	Extended Myositis Panel		x			x					x		
11	0050652	Extractable Nuclear Antigen Antibodies (Smith/RNP, Smith, SSA 52, SSA 60, and SSB)		x			x					x		
11	2001961	Familial Mutation, Targeted Sequencing				x		x	x	x				
12	2001980	Familial Mutation, Targeted Sequencing, Fetal				x		x	x	x				
12	2009034	Fragile X (*FMR1*) with Reflex to Methylation Analysis, Fetal				x								
13	0051270	Galactosemia (*GALT*) 9 Mutations, Fetal		x		x								
13	0091193	Gamma-Hydroxybutyric Acid (GHB), Serum or Plasma - Screen with Reflex to Confirmation/Quantitation				x								
13	0091161	Gamma-Hydroxybutyric Acid (GHB), Urine - Screen with Reflex to Confirmation/Quantitation		x		x								
14	2001755	Hemophilia A (*F8*) 2 Inversions, Fetal				x								
14	0091203	Heroin - Screen with Reflex to Confirmation/Quantitation - Serum or Plasma				x								
14	3002001	Kell K/k (KEL) Antigen Genotyping				x								
15	0091224	LSD, Urine - Screen with Reflex to Confirmation/Quantitation				x								
15	2014704	Maternal T Cell Engraftment in SCID, Maternal Specimen				x								
15	0091551	Phenobarbital, Total/Unbound/Bound, S/P		x						x				
15	3001170	Platelet Antigen 1 Genotyping (HPA-1)			x	x								
16	3000193	Platelet Antigen Genotyping Panel				x								
16	0070256	Proinsulin, Intact/Insulin Ratio				x	x							
16	2014351	Rabies Antibody Screen by RFFIT, Serum	x			x								

Hotline Page #	Test Number	Summary of Changes by Test Name	Name Change	Methodology	Performed/Reported Schedule	Specimen Requirements	Reference Interval	Interpretive Data	Note	CPT Code	Component Change	Other Interface Change	New Test	Inactive
17	3001053	Red Blood Cell Antigen Genotyping				x								
17	3002002	RhC/c (RHCE) Antigen Genotyping				x								
17	0051368	RhD Gene (*RHD*) Copy Number			x	x								
18	3002003	RhE/e (RHCE) Antigen Genotyping				x								
18	3000460	Smith and Smith/RNP (ENA) Antibodies, IgG		x		x	x					x		
19	0050470	Smith/RNP (ENA) Antibody, IgG		x		x	x	x	x			x		
19	2013444	Spinal Muscular Atrophy (SMA) Copy Number Analysis, Fetal				x								
20	0051508	Thanatophoric Dysplasia, Types 1 and 2 (*FGFR3)* 13 Mutations, Fetal				x								
20	0091585	Tin Total Quantitative, Serum or Plasma				x				x				
20	2005476	von Willebrand Disease, Platelet Type (*GP1BA*) 3 Mutations	x	x		x		x						

0051265 **Achondroplasia (*FGFR3*) 2 Mutations, Fetal** **AD PCR FE**

Performed: Varies
Reported: 2-7 days

Specimen Required: Collect: **Fetal specimen**: **Cultured amniocytes**: Two T-25 flasks at 80 percent confluency.
OR cultured CVS: Two T-25 flasks at 80 percent confluency.
If the client is unable to culture amniocytes or CVS, this can be arranged by contacting ARUP Client Services at (800) 522-2787.
AND maternal cell contamination specimen: Lavender (EDTA), pink (K_2EDTA), or yellow (ACD Solution A or B).
Specimen Preparation: **Cultured amniocytes AND cultured CVS:** Transport two T-25 flasks at 80 percent confluency filled with culture media. Backup cultures must be retained at the client's institution until testing is complete.
Maternal cell contamination specimen: Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: **Cultured amniocytes and cultured CVS: CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of shipment due to lability of cells.
Maternal cell contamination specimen: Refrigerated.
Remarks: **Maternal specimen is recommended for proper test interpretation. Order Maternal Cell Contamination, Maternal Specimen. Please contact an ARUP genetic counselor at 800-242-2787 x2141 prior to sample submission. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services.**
Unacceptable Conditions: Frozen specimens in glass collection tubes.
Stability (collection to initiation of testing): **Cultured amniocytes and cultured CVS:** Room Temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Maternal cell contamination specimen: Room Temperature: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0091328 Acyclovir, Serum or Plasma — ACYCLOV

Methodology: Quantitative High Performance Liquid Chromatography-Tandem Mass Spectrometry (HPLC-MS/MS)

Specimen Required: Collect: Plain red, Lavender (K_2 EDTA), or Pink (K_2 EDTA).
Specimen Preparation: Separate serum or plasma from cells within 2 hours. Transfer 1 mL serum or plasma to an ARUP Standard Transport Tube. (Min: 0.2 mL)
Test is not performed at ARUP; separate specimens must be submitted when multiple tests are ordered.
Storage/Transport Temperature: Refrigerated. Also acceptable: Room temperature and frozen.
Unacceptable Conditions: Separator tubes.
Stability (collection to initiation of testing): Ambient: 1 month; Refrigerated: 1 month; Frozen: 4 month

3003656 Alpha Thalassemia (*HBA1* and *HBA2*) Deletion/Duplication with reflex to Hb Constant Spring, Fetal — HBA DDCSFE

Specimen Required: Collect: **Cultured amniocytes or Cultured CVS**
AND Maternal Whole Blood Specimen: Lavender (EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
Specimen Preparation: **Cultured Amniocytes or Cultured CVS**: Transfer cultured amniocytes or cultured CVS to two T-25 flasks at 80 percent confluence. (Min: one T-25 flask at 80% confluence). Backup cultures must be retained at the client's institution until testing is complete. **If the client is unable to culture amniocytes or CVS, this can be arranged by contacting ARUP Client Services at (800) 522-2787. Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to test submission.**
Maternal Whole Blood Specimen: Transport 2 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: **Cultured Amniocytes or CVS**: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of collection due to viability of cells.
Maternal Whole Blood Specimen: Room temperature.
Remarks: **Please contact an ARUP genetic counselor at 800-242-2787 x2141 prior to sample submission. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services at (800) 522-2787.**
Stability (collection to initiation of testing): **Cultured Amniocytes or Cultured CVS:** Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Maternal Whole Blood Specimen: Room temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable

2008682 Anabolic Steroids, Urine - Screen with Reflex to Confirmation — STEROIDS

CPT Code(s): 80307; 82570; if positive add 80328 (Alt code: if positive add G0480)

2012232 Angelman Syndrome and Prader-Willi Syndrome by Methylation-Sensitive PCR, Fetal — AS PWS FE

Performed: Varies
Reported: 2-7 days

Specimen Required: Collect: Contact ARUP's genetic counselors at (800) 242-2787 extension 2141 prior to submission for specimen requirements and submission information.

HOTLINE NOTE: Remove information found in the Specimen Preparation, Storage/Transport Temperature, Remarks, Unacceptable Conditions, and Stability fields.

0050388 Beta Globin (*HBB*) Sequencing, Fetal — BGSEQ FE

Specimen Required: Collect: **Fetal Specimen**: Two T-25 flasks at 80% confluent of Cultured Amniocytes or Cultured Chorionic Villus Sampling (CVS).
If the client is unable to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787.
Specimen Preparation: **Cultured Amniocytes or Cultured CVS**: Fill flasks with culture media. Transport two T-25 flasks at 80% confluent of cultured amniocytes or cultured CVS filled with culture media. Backup cultures must be retained at the client's institution until testing is complete.
AND Maternal Specimen: Transport 2 mL whole blood. (Min: 1 mL) Lavender (EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
Storage/Transport Temperature: **Cultured Amniocytes or Cultured CVS: CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of shipment due to liability of cells.
Maternal Specimen: Room temperature
Remarks: **Maternal specimen is recommended for proper test interpretation. Order Maternal Cell Contamination, Maternal Specimen. This can be arranged by contacting ARUP genetic counselors at (800) 242-2787 ext. 2141. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services.**
Stability (collection to initiation of testing): **Fetal Specimen:** Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Maternal Specimen: Ambient: 1 week; Refrigerated: 1 month

Reference Interval: By report

Interpretive Data: By report

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

3000480 Comprehensive Systemic Sclerosis Panel — SCL COMPRE

Reference Interval:

Test Number	Components	Reference Interval	
0050599	Scleroderma (Scl-70) (ENA) Antibody, IgG		
		29 AU/mL or less	Negative
		30-40 AU/mL	Equivocal
		41 AU/mL or greater	Positive
0050470	Smith/RNP (ENA) Antibody, IgG	Effective September 7, 2021	
		19 Units or less	Negative
		20 to 39 Units	Weak Positive
		40 to 80 Units	Moderate Positive
		81 Units or greater	Strong Positive
3000082	Antinuclear Antibody (ANA) with HEp-2 Substrate, IgG by IFA	Less than 1:80	
2012173	Fibrillarin (U3 RNP) Antibody, IgG	Negative	
2003040	PM/Scl-100 Antibody, IgG by Immunoblot	Negative	
2001601	RNA Polymerase III Antibody, IgG		
		19 Units or less	Negative
		20-39 Units	Weak Positive
		40-80 Units	Moderate Positive
		81 Units or greater	Strong Positive

HOTLINE NOTE: There is a unit of measure change associated with this test.
Change the unit of measure for component 0050470, Smith/RNP (ENA) Ab, IgG from AU/mL to Units.

3002463 **Connective Tissue Disease First Line Panel with Reflex** **CTD PAN**

Reference Interval:

Test Number	Components	Reference Interval
0050215	Double-Stranded DNA (dsDNA) Antibody, IgG by ELISA with Reflex to dsDNA Antibody, IgG by IFA	Effective May 17, 2021
		Test Number / **Components** / **Reference Interval**
		(blank) / dsDNA (Double Stranded DNA) Antibody, IgG / Refer to report
		2002693 / Double-Stranded DNA (dsDNA) Antibody, IgG by IFA (using *Crithidia luciliae)* / Refer to report
2002693	Double-Stranded DNA (dsDNA) Antibody, IgG by IFA (using *Crithidia luciliae)*	Less than 1:10
0050470	Smith/RNP (ENA) Antibody, IgG	Effective September 7, 2021
		19 Units or less: Negative
		20 to 39 Units: Weak Positive
		40 to 80 Units: Moderate Positive
		81 Units or greater: Strong Positive
0050085	Smith (ENA) Antibody, IgG	
		29 AU/mL or less: Negative
		30-40 AU/mL: Equivocal
		41 AU/mL or greater: Positive
2012074	SSA 52 and 60 (Ro) (ENA) Antibodies, IgG	
		Test Number / **Components** / **Reference Interval**
		(blank) / SSA-52 (Ro52) (ENA) Antibody, IgG / 29 AU/mL or Less: Negative 30-40 AU/mL: Equivocal 41 AU/mL or greater: Positive
		(blank) / SSA-60 (Ro60) (ENA) Antibody, IgG / 29 AU/mL or Less: Negative 30-40 AU/mL: Equivocal 41 AU/mL or greater: Positive
0050692	SSB (La) (ENA) Antibody, IgG	
		29 AU/mL or less: Negative
		30-40 AU/mL: Equivocal
		41 AU/mL or greater: Positive
0099592	Jo-1 Antibody, IgG	
		29 AU/mL or less: Negative
		30-40 AU/mL: Equivocal
		41 AU/mL or greater: Positive
0050599	Scleroderma (Scl-70) (ENA) Antibody, IgG	
		29 AU/mL or less: Negative
		30-40 AU/mL: Equivocal
		41 AU/mL or greater: Positive

HOTLINE NOTE: There is a unit of measure change associated with this test.
Change the unit of measure for component 0050470, Smith/RNP (ENA) Ab, IgG from AU/mL to Units.

0051668 Connective Tissue Diseases Profile CONN

Methodology: Semi-Quantitative Enzyme-Linked Immunosorbent Assay
Semi-Quantitative Multiplex Bead Assay

Reference Interval: Effective May 18, 2015

Test Number	Components	Reference Interval
0050085	Smith (ENA) Antibody, IgG	
		29 AU/mL or less: Negative 30-40 AU/mL: Equivocal 41 AU/mL or greater: Positive
0050470	Smith/RNP (ENA) Antibody, IgG	Effective September 7, 2021
		19 Units: Negative 20 to 39 Units: Weak Positive 40 to 80 Units: Moderate Positive 81 Units or greater: Strong Positive
2012074	SSA 52 and 60 (Ro) (ENA) Antibodies, IgG	
		Test Number / Components / Reference Interval SSA-52 (Ro52) (ENA) Antibody, IgG: 29 AU/mL or Less: Negative; 30-40 AU/mL: Equivocal; 41 AU/mL or greater: Positive SSA-60 (Ro60) (ENA) Antibody, IgG: 29 AU/mL or Less: Negative; 30-40 AU/mL: Equivocal; 41 AU/mL or greater: Positive
0050692	SSB (La) (ENA) Antibody, IgG	
		29 AU/mL or less: Negative 30-40 AU/mL: Equivocal 41 AU/mL or greater: Positive
0099592	Jo-1 Antibody, IgG	
		29 AU/mL or less: Negative 30-40 AU/mL: Equivocal 41 AU/mL or greater: Positive
0099249	Ribosomal P Protein Antibody	
		29 AU/mL or less: Negative 30-40 AU/mL: Equivocal 41 AU/mL or greater: Positive
0050714	Centromere Antibody, IgG	
		29 AU/mL or less: Negative 30-40 AU/mL: Equivocal 41 AU/mL or greater: Positive
0050599	Scleroderma (Scl-70) (ENA) Antibody, IgG	
		29 AU/mL or less: Negative 30-40 AU/mL: Equivocal 41 AU/mL or greater: Positive

HOTLINE NOTE: There is a unit of measure change associated with this test.
Change the unit of measure for component 0050470, Smith/RNP (ENA) Ab, IgG from AU/mL to Units.

3003648 **COVID-19 IgG (Spike), Semi-Quantitative by CIA** **COV19G SQ**

Specimen Required: Collect: Serum separator tube (SST). Also acceptable: lithium heparin.
Specimen Preparation: Separate serum or plasma from cells ASAP or within 2 hours of collection. Transfer 1 mL serum or plasma to an ARUP Standard Transport Tube. (Min: 0.5 mL)
Storage/Transport Temperature: Refrigerated.
Unacceptable Conditions: Specimens containing particulate material or otherwise obviously contaminated. Severely hemolyzed, heat-inactivated, severely icteric or lipemic specimens.
Stability (collection to initiation of testing): After separation from cells: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month (avoid repeated freeze/thaw cycles)

2013562 **C-Peptide, 120 Minutes** **C PEP 120**

Specimen Required: Patient Prep: Fasting specimen preferred.
Collect: Serum Separator Tube (SST)
Specimen Preparation: Allow specimen to clot completely at room temperature. Separate from cells ASAP or within 2 hours of collection. Transport 1 mL serum to an ARUP Standard Transport Tube.
Storage/Transport Temperature: Frozen.
Unacceptable Conditions: Grossly hemolyzed specimens.
Stability (collection to initiation of testing): After separation from cells: Ambient: 8 hours; Refrigerated: 7 days; Frozen: 1 month

2013564 **C-Peptide, 180 Minutes** **C PEP 180**

Specimen Required: Patient Prep: Fasting specimen preferred.
Collect: Serum Separator Tube (SST)
Specimen Preparation: Allow specimen to clot completely at room temperature. Separate from cells ASAP or within 2 hours of collection. Transport 1 mL serum to an ARUP Standard Transport Tube.
Storage/Transport Temperature: Frozen.
Unacceptable Conditions: Grossly hemolyzed specimens.
Stability (collection to initiation of testing): After separation from cells: Ambient: 8 hours; Refrigerated: 7 days; Frozen: 1 month

2013558 **C-Peptide, 30 Minutes** **C PEP 30**

Specimen Required: Patient Prep: Fasting specimen preferred.
Collect: Serum Separator Tube (SST)
Specimen Preparation: Allow specimen to clot completely at room temperature. Separate from cells ASAP or within 2 hours of collection. Transport 1 mL serum to an ARUP Standard Transport Tube. (Min: 0.5 mL)
Storage/Transport Temperature: Frozen.
Unacceptable Conditions: Grossly hemolyzed specimens.
Stability (collection to initiation of testing): After separation from cells: Ambient: 8 hours; Refrigerated: 7 days; Frozen: 1 month

2013560 **C-Peptide, 60 Minutes** **C PEP 60**

Specimen Required: Patient Prep: Fasting specimen preferred.
Collect: Serum Separator Tube (SST)
Specimen Preparation: Allow specimen to clot completely at room temperature. Separate from cells ASAP or within 2 hours of collection. Transport 1 mL serum to an ARUP Standard Transport Tube. (Min: 0.5 mL)
Storage/Transport Temperature: Frozen.
Unacceptable Conditions: Grossly hemolyzed specimens.
Stability (collection to initiation of testing): After separation from cells: Ambient: 8 hours; Refrigerated: 7 days; Frozen: 1 month

3000529 C-Peptide, Other CPEPOTHER

Specimen Required: Patient Prep: Fasting specimen preferred.
Collect: Serum Separator Tube (SST)
Specimen Preparation: Allow specimen to clot completely at room temperature. Separate from cells ASAP or within 2 hours of collection. Transport 1 mL serum in an ARUP Standard Transport Tube. (Min: 0.5 mL)
Storage/Transport Temperature: Frozen.
Unacceptable Conditions: Grossly hemolyzed specimens.
Stability (collection to initiation of testing): After separation from cells: Ambient: 8 hours; Refrigerated: 7 days; Frozen: 1 month

Interpretive Data:
The reference interval for fasting c-peptide is 0.5-3.3 ng/mL. To convert to nmol/L, multiply ng/mL by 0.33.

2011231 Duchenne/Becker Muscular Dystrophy (*DMD*) Deletion/Duplication, Fetal DMD DD FE

Patient History for Fetal Molecular Testing

Additional Technical Information

Test not New York DOH approved at any laboratory. An approved NPL form must accompany specimen.

Time Sensitive

Specimen Required: Collect: **Cultured amniocytes or Cultured CVS**
AND Maternal Whole Blood Specimen: Lavender (EDTA), pink (K_2EDTA), or yellow (ACD Solution A or B).
Specimen Preparation: **Cultured Amniocytes or Cultured CVS:** Transfer cultured amniocytes or cultured CVS to two T-25 flasks at 80 percent confluence. (Min: one T-25 flask at 80% confluence). Backup cultures must be retained at the client's institution until testing is complete. **If the client is unable to culture amniocytes or CVS, this can be arranged by contacting ARUP Client Services at (800) 522-2787. Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to test submission.**
Maternal Whole Blood Specimen: Transport 2 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: **Cultured Amniocytes or Cultured CVS:** CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of collection due to viability of cells.
Maternal Whole Blood Specimen: Room temperature.
Remarks: **Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to sample submission. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services at (800) 522-2787.**
Stability (collection to initiation of testing): **Cultured Amniocytes or Cultured CVS:** Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Maternal Whole Blood Specimen: Room temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable

3001781 Extended Myositis Panel MYOS EXT

Methodology: Semi-Quantitative Enzyme-Linked Immunosorbent Assay/Qualitative Immunoprecipitation/Semi-Quantitative Multiplex Bead Assay/Qualitative Immunoblot

Reference Interval:

Test Number	Components	Reference Interval
2012074	SSA 52 and 60 (Ro) (ENA) Antibodies, IgG	

Test Number	Components	Reference Interval
	SSA-52 (Ro52) (ENA) Antibody, IgG	29 AU/mL or Less: Negative 30-40 AU/mL: Equivocal 41 AU/mL or greater: Positive
	SSA-60 (Ro60) (ENA) Antibody, IgG	29 AU/mL or Less: Negative 30-40 AU/mL: Equivocal 41 AU/mL or greater: Positive

0050470	Smith/RNP (ENA) Antibody, IgG	Effective September 7, 2021
		19 Units or less: Negative
		20 to 39 Units: Weak Positive
		40 to 80 Units: Moderate Positive
		81 Units or greater: Strong Positive
0099592	Jo-1 Antibody, IgG	
		29 AU/mL or less: Negative
		30-40 AU/mL: Equivocal
		41 AU/mL or greater: Positive
2012173	Fibrillarin (U3 RNP) Antibody, IgG	Negative
2003040	PM/Scl-100 Antibody, IgG by Immunoblot	Negative
	Mi-2 (nuclear helicase protein) Antibody	Negative
	PL-7 (threonyl-tRNA synthetase) Antibody	Negative
	PL-12 (alanyl-tRNA synthetase) Antibody	Negative
	P155/140 Antibody	Negative
	EJ (glycyl-tRNA synthetase) Antibody	Negative
	Ku Antibody	Negative
	SRP (Signal Recognition Particle) Ab	Negative
	OJ (isoleucyl-tRNA synthetase) Antibody	Negative
	SAE1 (SUMO activating enzyme) Ab	Negative
	MDA5 (CADM-140) Ab	Negative
	NXP2 (Nuclear matrix protein-2) Ab	Negative
	TIF-1 gamma (155 kDa) Ab	Negative

HOTLINE NOTE: There is a unit of measure change associated with this test.
Change the unit of measure for component 0050470, Smith/RNP (ENA) Ab, IgG from AU/mL to Units.

0050652 Extractable Nuclear Antigen Antibodies (Smith/RNP, Smith, SSA 52, SSA 60, and SSB) ENA ABS4

Methodology: Semi-Quantitative Enzyme-Linked Immunosorbent Assay
Semi-Quantitative Multiplex Bead Assay

Reference Interval: Effective May 18, 2015

Test Number	Components	Reference Interval
0050470	Smith/RNP (ENA) Antibody, IgG	Effective September 7, 2021
		19 Units or less: Negative
		20 to 39 Units: Weak Positive
		40 to 80 Units: Moderate Positive
		81 Units or greater: Strong Positive
0050085	Smith (ENA) Antibody, IgG	
		29 AU/mL or less: Negative
		30-40 AU/mL: Equivocal
		41 AU/mL or greater: Positive
2012074	SSA 52 and 60 (Ro) (ENA) Antibodies, IgG	
		Test Number / Components / Reference Interval
		SSA-52 (Ro52) (ENA) Antibody, IgG: 29 AU/mL or Less: Negative; 30-40 AU/mL: Equivocal; 41 AU/mL or greater: Positive
		SSA-60 (Ro60) (ENA) Antibody, IgG: 29 AU/mL or Less: Negative; 30-40 AU/mL: Equivocal; 41 AU/mL or greater: Positive
0050692	SSB (La) (ENA) Antibody, IgG	
		29 AU/mL or less: Negative
		30-40 AU/mL: Equivocal
		41 AU/mL or greater: Positive

HOTLINE NOTE: There is a unit of measure change associated with this test.
Change the unit of measure for component 0050470, Smith/RNP (ENA) Ab, IgG from AU/mL to Units.

2001961 Familial Mutation, Targeted Sequencing SEQ FSM

Specimen Required: Collect: **Contact ARUP's genetic counselor at (800) 242-2787 extension 2141 prior to test submission for specimen requirements and submission information.**
Remarks: Documentation of the familial gene variants(s) is required to perform targeted sequencing. Submit a copy of a relative's laboratory test report documenting the gene and specific variants(s) for which testing is requested.
Submit a positive control with the patient specimen for appropriate interpretation, order Sequencing Control (test code 0051650).
Samples tested without a familial positive control may be subject to a disclaimer.
Testing will begin upon receipt of all necessary components, including an original laboratory report detailing the familial variant(s) to be tested and a familial positive control sample.

Interpretive Data: By report

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

CPT Code(s): Varies by gene

HOTLINE NOTE: Remove information found in the Note field.

2001980 Familial Mutation, Targeted Sequencing, Fetal SEQ FSM FE

Specimen Required: Collect: **Contact ARUP's genetic counselors at (800) 242-2787 extension 2141 prior to test submission for specimen requirements and submission information.**
Remarks: Documentation of the familial gene variants(s) is required to perform targeted sequencing. Submit a copy of a relative's laboratory test report documenting the gene and specific variants(s) for which testing is requested.
Submit a positive control with the patient specimen for appropriate interpretation, order Sequencing Control. Fetal samples tested without a familial positive control may be subject to a disclaimer.
A maternal specimen is recommended for proper fetal test interpretation. Order Maternal Cell Contamination, Maternal Specimen.
Testing will begin upon receipt of all necessary components, including: an original laboratory report detailing the familial variant(s) to be tested, a maternal specimen for maternal cell contamination testing, and a familial positive control sample.

Interpretive Data: By report

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

CPT Code(s): Varies by gene; 81265 Fetal Cell Contamination.

HOTLINE NOTE: Remove information found in the Note, Specimen Preparation, and Stability fields.

2009034 Fragile X (*FMR1*) with Reflex to Methylation Analysis, Fetal FX PCR FE

Patient History for Fetal Molecular Testing

Additional Technical Information

Time Sensitive

Specimen Required: Collect: **Cultured Amniocytes or Cultured CVS**
AND Maternal Whole Blood Specimen: Lavender (EDTA), pink (K_2EDTA), or yellow (ACD Solution A or B).
Specimen Preparation: **Cultured Amniocytes or Cultured CVS:** Transfer cultured amniocytes or cultured CVS to two T-25 flasks at 80 percent confluence (Min: one T-25 flask at 80% confluence). Backup cultures must be retained at the client's institution until testing is complete. **If the client is unable to culture amniocytes or CVS, this can be arranged by contacting ARUP Client Services at (800) 522-2787. Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to test submission.**
Maternal Whole Blood Specimen: Transport 2 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: **Cultured Amniocytes or Cultured CVS:** CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of collection due to viability of cells.
Maternal Whole Blood Specimen: Room temperature.
Remarks: **Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to sample submission. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services at (800) 522-2787.**
Stability (collection to initiation of testing): **Cultured Amniocytes or Cultured CVS:** Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Maternal Whole Blood Specimen: Room temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable

0051270 Galactosemia (*GALT*) 9 Mutations, Fetal — GALTDNA FE

Patient History for Fetal Molecular Testing

Additional Technical Information

Time Sensitive

Methodology: Polymerase Chain Reaction/Single Nucleotide Extension

Specimen Required: Collect: Cultured Amniocytes, Cultured CVS, or Amniotic fluid (direct)
AND Maternal Whole Blood Specimen: Lavender (EDTA), pink (K_2EDTA), or yellow (ACD Solution A or B).
Specimen Preparation: **Cultured Amniocytes or Cultured CVS:** Transfer cultured amniocytes or cultured CVS to two T-25 flasks at 80% confluence. (Min: one T-25 flask at 80% confluence). Backup cultures must be retained at the client's institution until testing is complete. **If the client is unable to culture amniocytes or CVS, this can be arranged by contacting ARUP Client Services at (800) 522-2787. Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to test submission.**
Amniotic Fluid (direct): 10 milliliters
Maternal Whole Blood Specimen: 2 mL whole blood (Min: 1 mL).
Storage/Transport Temperature: **Cultured Amniocytes or Cultured CVS: CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of collection due to viability of cells.
Amniotic fluid (direct): Ship room temperature.
Maternal Whole Blood Specimen: Room temperature.
Remarks: Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to sample submission. Patient History Forms are available on aruplab.com or by contacting ARUP Client Services at (800) 522-2787.
Stability (collection to initiation of testing): **Cultured Amniocytes or Cultured CVS:** Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Amniotic Fluid (direct): Room temperature: 48 hours; Refrigerated: 72 hours; Frozen: Unacceptable
Maternal Whole Blood Specimen: Room temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable

0091193 Gamma-Hydroxybutyric Acid (GHB), Serum or Plasma - Screen with Reflex to Confirmation/Quantitation — GHB SP

Specimen Required: Collect: Plain Red, Lavender (EDTA), or Pink (K_2EDTA).
Specimen Preparation: Separate from cells ASAP or within 2 hours of collection. Transfer 5 mL serum or Plasma to ARUP Standard Transport Tubes. (Min: 2.4 mL)
Test is not performed at ARUP; separate specimens must be submitted when multiple tests are ordered.
Storage/Transport Temperature: Refrigerated. Also acceptable: Room temperature or frozen.
Unacceptable Conditions: Separator tubes or citrate buffered tubes.
Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 week; Frozen: 2 months

0091161 Gamma-Hydroxybutyric Acid (GHB), Urine - Screen with Reflex to Confirmation/Quantitation — GHB U

Methodology: Qualitative Gas Chromatography-Mass Spectrometry/Quantitative Gas Chromatography-Mass Spectrometry (HPLC-MS/MS)

Specimen Required: Specimen Preparation: Transfer 5 mL urine to ARUP Standard Transport Tubes. (Min: 2.8 mL)
Test is not performed at ARUP; separate specimens must be submitted when multiple tests are ordered.
Storage/Transport Temperature: Refrigerated. Also acceptable: Room temperature or frozen.
Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 week; Frozen: 3 weeks

2001755 Hemophilia A (*F8*) 2 Inversions, Fetal — F8 INV FE

Patient History for Fetal Molecular Testing

Additional Technical Information

Time Sensitive

Specimen Required: Collect: **Cultured Amniocytes or Cultured CVS**
AND Maternal Whole Blood Specimen: Lavender (EDTA) or yellow (ACD Solution A or B).
Specimen Preparation: **Cultured Amniocytes or Cultured CVS:** Transfer cultured amniocytes or cultured CVS to two T-25 flasks at 80% confluence: (Min: one T-25 flask at 80% confluence). Backup cultures must be retained at the client's institution until testing is complete. **If the client is unable to culture amniocytes or CVS, this can be arranged by contacting ARUP Client Services at (800) 522-2787. Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to test submission.**
Maternal Whole Blood Specimen: Transport 2 mL whole blood (Min: 1 mL).
Storage/Transport Temperature: **Cultured Amniocytes or Cultured CVS: CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of collection due to viability of cells.
Maternal Whole Blood Specimen: Room temperature.
Remarks: **Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to sample submission. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services at (800) 522-2787.**
Stability (collection to initiation of testing): **Cultured Amniocytes or Cultured CVS:** Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Maternal Whole Blood Specimen: Room temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable

0091203 Heroin - Screen with Reflex to Confirmation/Quantitation - Serum or Plasma — HEROIN SP

Specimen Required: Collect: Plain Red or Gray (Sodium Fluoride/Potassium Oxalate).
Specimen Preparation: Separate from cells ASAP or within 2 hours of collection. Transfer 3 mL serum or plasma to an ARUP Standard Transport Tube. (Min: 1.3 mL)
Test is not performed at ARUP; separate specimens must be submitted when multiple tests are ordered.
Storage/Transport Temperature: Refrigerated. Also acceptable: Frozen.
Unacceptable Conditions: Separator tubes.
Stability (collection to initiation of testing): Ambient: 48 hours; Refrigerated: 2 weeks; Frozen: 1 month

3002001 Kell K/k (KEL) Antigen Genotyping — KEL GENO

Specimen Required: Collect: **Fetal genotyping:**
Cultured amniocytes: Two T-25 flasks at 80 percent confluency.
If the client is unable to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787.
WITH maternal cell contamination specimen (see Note)**:** Lavender (K_2EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
Parental genotyping: Lavender (K_2EDTA), Pink (K_2EDTA).
Specimen Preparation: **Cultured amniocytes:** Transport two T-25 flasks at 80 percent confluency filled with culture media. Backup cultures must be retained at the client's institution until testing is complete.
Maternal cell contamination specimen: Transport 3 mL whole blood (Min: 1 mL)
Whole blood (parental genotyping): Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: **Cultured amniocytes: CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of shipment due to lability of cells.
Whole blood or maternal cell contamination specimen: Refrigerated.
Remarks: Patient History Form is available on the ARUP website or by contacting ARUP Client Services.
Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin tubes.
Stability (collection to initiation of testing): **Fetal specimen:** Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Whole blood or maternal cell contamination specimen: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0091224 LSD, Urine - Screen with Reflex to Confirmation/Quantitation — LSD URN

Specimen Required: Collect: Urine.
Specimen Preparation: Transfer 2 mL urine to an ARUP Standard Transport Tube. (Min: 0.9 mL)
Test is not performed at ARUP; separate specimens must be submitted when multiple tests are ordered.
Storage/Transport Temperature: Refrigerated. Also acceptable: Frozen.
Stability (collection to initiation of testing): Ambient: 24 hours; Refrigerated: 1 month; Frozen: 1 month

2014704 Maternal T Cell Engraftment in SCID, Maternal Specimen — SCID-MAT

Specimen Required: Collect: Lavender (EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A).
New York State Clients: Lavender (EDTA) or Yellow (ACD Solution A). **Collect Monday-Thursday only.**
Specimen Preparation: Transport 2 mL whole blood. (Min: 1 mL)
New York State Clients: Transport 9 mL whole blood. (Min: 4 mL). **Do not send to ARUP Laboratories**. Specimens must be received at performing laboratory within 48 hours of collection. For specimen requirements and direct submission instructions please contact ARUP Referral Testing at (800) 242-2787, ext. 5145.
Storage/Transport Temperature: Refrigerated
Stability (collection to initiation of testing): Room Temperature: 1 week; Refrigerated: 1 month; Frozen: unacceptable
New York State Clients: Room Temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable

0091551 Phenobarbital, Total/Unbound/Bound, S/P — PHENOBAR

Methodology: Quantitative High Performance Liquid Chromatography-Tandem Mass Spectrometry (HPLC-MS/MS)

CPT Code(s): 80184 x2

3001170 Platelet Antigen 1 Genotyping (HPA-1) — HPA-1 GENO

Performed: Varies
Reported: 7-14 days

Specimen Required: Collect: **Fetal specimen: Cultured amniocytes**: Two T-25 flasks at 80 percent confluency.
If the client is unable to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787.
WITH maternal cell contamination specimen (see Note)**:** Lavender (EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
Parental specimen: Lavender (EDTA).
Specimen Preparation: **Cultured amniocytes:** Transport two T-25 flasks at 80 percent confluency filled with culture media. Backup cultures must be retained at the client's institution until testing is complete.
Maternal cell contamination specimen: Transport 3 mL whole blood. (Min: 1 mL)
Whole blood (parental genotyping): Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: **Cultured amniocytes: CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of shipment due to lability of cells.
Whole blood or maternal cell contamination specimen: Refrigerated.
Unacceptable Conditions: Frozen specimens in glass collection tubes.
Stability (collection to initiation of testing): **Fetal specimen:** Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Whole blood or maternal cell contamination specimen: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

3000193 Platelet Antigen Genotyping Panel — HPA GENO

Specimen Required: Collect: **Fetal genotying: Cultured amniocytes**: Two T-25 flasks at 80 percent confluency.
If the client is unable to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787.
WITH maternal cell contamination specimen: Lavender (EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
Parental genotyping: Lavender (EDTA).
Specimen Preparation: **Cultured amniocytes:** Transport two T-25 flasks at 80 percent confluency filled with culture media. Backup cultures must be retained at the client's institution until testing is complete.
Maternal cell contamination specimen: Transport 3 mL whole blood. (Min: 1 mL)
Whole blood (parental genotyping): Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: **Cultured amniocytes: CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of shipment due to lability of cells.
Whole blood or maternal cell contamination specimen: Refrigerated.
Unacceptable Conditions: Frozen specimens in glass collection tubes.
Stability (collection to initiation of testing): **Fetal specimen:** Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Whole blood or maternal cell contamination specimen: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0070256 Proinsulin, Intact/Insulin Ratio — PRO INS

Specimen Required: Patient Prep: Patient must be fasting for 12-15 hours prior to collection.
Collect: Serum Separator Tube (SST).
Specimen Preparation: Separate from cells ASAP or within 2 hours of collection. Transfer 1 mL serum to an ARUP Standard Transport Tube and freeze immediately. (Min: 0.8 mL)
Storage/Transport Temperature: **CRITICAL FROZEN.** Separate specimens must be submitted when multiple tests are ordered.
Unacceptable Conditions: Heparinized plasma. Vitreous or I.V. fluids. Hemolyzed specimens.
Stability (collection to initiation of testing): After separation from cells: Ambient: Unacceptable; Refrigerated: 48 hours; Frozen: 2 months (avoid repeated freeze/thaw cycles)

Reference Interval:

Available Separately	Component	Reference Interval
Yes (0070063)	Insulin, Fasting	Effective September 7, 2021 3-25 µIU/mL
Yes (0070112)	Proinsulin, Intact	0-17 years: Not established Effective May 19th, 2014 18 years and older: Less than or equal to 8.0 pmol/L
No	Proinsulin, Intact/Insulin Ratio Calculation	**Proinsulin, Intact/Insulin Ratio as Percent:** 0-17 years: Not established 18 years and older: 0.8-21.7 percent

2014351 Rabies Antibody Screen by RFFIT, Serum — RABIES AB

Specimen Required: Collect: Plain Red or Serum Separator Tube (SST).
Specimen Preparation: Transfer 2 mL serum to an ARUP Standard Transport Tube. (Min: 0.25 mL)
Test is not performed at ARUP; separate specimens must be submitted when multiple tests are ordered.
Storage/Transport Temperature: Refrigerated. Also acceptable: Room temperature or frozen.
Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 2 weeks; Frozen: 1 month

3001053 Red Blood Cell Antigen Genotyping — RBC GENO

Specimen Required: Collect: **Fetal genotyping: Cultured amniocytes:** Two T-25 flasks at 80 percent confluency.
If the client is unable to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787.
WITH maternal cell contamination specimen: Lavender (K_2EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
OR Genotyping: Lavender (K_2EDTA), Pink (K_2EDTA) OR
Specimen Preparation: **Genotyping:** Transport 3 mL whole blood. (Min: 1 mL)
Cultured amniocytes: Transport two T-25 flasks at 80 percent confluency filled with culture media. Backup cultures must be retained at the client's institution until testing is complete.
Maternal cell contamination specimen: Transport 3 mL whole blood (Min: 1 mL)
Storage/Transport Temperature: **Cultured amniocytes: CRITICAL ROOM TEMPERATURE**. Must be received within 48 hours of shipment due to lability of cells.
Whole blood or maternal cell contamination specimen: Refrigerated.
Remarks: **Maternal specimen is recommended for proper test interpretation if contamination of the fetal specimen from the mother is suspected. Order Maternal Cell Contamination.**
Unacceptable Conditions: Plasma or serum; collection of specimens in sodium heparin tubes. Frozen specimens in glass collection tubes.
Stability (collection to initiation of testing): **Whole blood or maternal cell contamination specimen:** Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month
Fetal specimen: Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable

3002002 RhC/c (RHCE) Antigen Genotyping — RHC GENO

Specimen Required: Collect: **Fetal genotyping: Cultured amniocytes:** Two T-25 flasks at 80 percent confluency.
If the client is unable to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787.
WITH maternal cell contamination specimen (see Note): Lavender (K_2EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
Parental genotyping: Lavender (K_2EDTA), Pink (K_2EDTA)
Specimen Preparation: **Cultured amniocytes:** Transport two T-25 flasks at 80 percent confluency filled with culture media. Backup cultures must be retained at the client's institution until testing is complete.
Maternal cell contamination specimen: Transport 3 mL whole blood (Min: 1 mL)
Whole blood (parental genotyping): Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: **Cultured amniocytes: CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of shipment due to lability of cells.
Whole blood or maternal cell contamination specimen: Refrigerated.
Remarks: Patient History Form is available on the ARUP website or by contacting ARUP Client Services.
Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin tubes.
Stability (collection to initiation of testing): **Fetal specimen:** Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Whole blood or maternal cell contamination specimen: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0051368 RhD Gene (*RHD*) Copy Number — RHD

Performed: Varies
Reported: 7-14 days

Specimen Required: Collect: **Fetal genotyping: Cultured amniocytes**: Two T-25 flasks at 80 percent confluency.
If the client is unable to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787.
WITH maternal cell contamination specimen (see Remarks): Lavender (EDTA), pink (K_2EDTA), or yellow (ACD Solution A or B).
Parental genotyping: Lavender (EDTA), pink (K_2EDTA), or yellow (ACD Solution A or B).
Specimen Preparation: **Cultured amniocytes:** Transport two T-25 flasks at 80 percent confluency filled with culture media. Backup cultures must be retained at the client's institution until testing is complete.
Maternal cell contamination specimen: Transport 3 mL whole blood (Min: 1 mL)
Whole blood (parental genotyping): Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: **Cultured amniocytes: CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of shipment due to lability of cells.
Whole blood or maternal cell contamination specimen: Refrigerated.
Remarks: Maternal specimen is recommended for proper test interpretation if contamination of the fetal specimen from the mother is suspected. Order Maternal Cell Contamination. Patient History Form is available on the ARUP website or by contacting ARUP Client Services.
Unacceptable Conditions: Frozen specimens in glass collection tubes.
Stability (collection to initiation of testing): **Fetal Specimen:** Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Whole blood or maternal cell contamination specimen: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

3002003 RhE/e (RHCE) Antigen Genotyping — RHE GENO

Specimen Required: Collect: **Fetal genotyping**: **Cultured amniocytes**: Two T-25 flasks at 80 percent confluency.
If the client is unable to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787.
WITH maternal cell contamination specimen (see Note): Lavender (K_2EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
Parental genotyping: Lavender (K_2EDTA), Pink (K_2EDTA).
Specimen Preparation: **Cultured amniocytes:** Transport two T-25 flasks at 80 percent confluency filled with culture media. Backup cultures must be retained at the client's institution until testing is complete.
Maternal cell contamination specimen: Transport 3 mL whole blood (Min: 1 mL)
Whole blood (parental genotyping): Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: **Cultured amniocytes: CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of shipment due to lability of cells.
Whole blood or maternal cell contamination specimen: Refrigerated.
Remarks: Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services.
Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin tubes.
Stability (collection to initiation of testing): **Fetal specimen:** Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Whole blood or maternal cell contamination specimen: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

3000460 Smith and Smith/RNP (ENA) Antibodies, IgG — SMITH_RNP

Methodology: Semi-Quantitative Enzyme-Linked Immunosorbent Assay
Semi Quantitative Multiplex Bead Assay

Specimen Required: Collect: Serum Separator Tube (SST).
Specimen Preparation: Separate from cells ASAP or within 2 hours of collection. Transfer 1 mL serum to an ARUP Standard Transport Tube. (Min: 0.2 mL)
Storage/Transport Temperature: Refrigerated.
Unacceptable Conditions: Plasma or other body fluids. Contaminated, hemolyzed, or severely lipemic specimens.
Stability (collection to initiation of testing): After separation from cells: Ambient: 48 hours; Refrigerated: 2 weeks; Frozen: 30 days (avoid repeated freeze/thaw cycles)

Reference Interval:

Test Number	Components	Reference Interval	
0050470	Smith/RNP (ENA) Antibody, IgG	Effective September 7, 2021	
		19 Units or less	Negative
		20-39 Units	Weak Positive
		40-80 Units	Moderate Positive
		81 Units or greater	Strong Positive
0050085	Smith (ENA) Antibody, IgG		
		29 AU/mL or less	Negative
		30-40 AU/mL	Equivocal
		41 AU/mL or greater	Positive

HOTLINE NOTE: There is a unit of measure change associated with this test.
Change the unit of measure for component 0050470, Smith/RNP (ENA) Ab, IgG from AU/mL to Units.

0050470 Smith/RNP (ENA) Antibody, IgG RNP

Methodology: Semi-Quantitative Enzyme-Linked Immunosorbent Assay

Specimen Required: Collect: Serum separator tube.
Specimen Preparation: Separate serum from cells ASAP or within 2 hours of collection. Transfer 1 mL serum to an ARUP Standard Transport Tube. (Min: 0.2 mL)
Storage/Transport Temperature: Refrigerated.
Unacceptable Conditions: Plasma or other body fluids. Contaminated, hemolyzed, or severely lipemic specimens.
Stability (collection to initiation of testing): After separation from cells: Ambient: 48 hours; Refrigerated: 2 weeks; Frozen: 30 days (avoid repeated freeze/thaw cycles)

Reference Interval:
Effective September 7, 2021

19 Units or less	Negative
20-39 Units	Weak Positive
40-80 Units	Moderate Positive
81 Units or greater	Strong Positive

Interpretive Data:
Smith/RNP antibodies are frequently seen in patients with mixed connective tissue disease (MCTD) and are also associated with other systemic autoimmune rheumatic diseases (SARDs), such as systemic lupus erythematosus (SLE), systemic sclerosis, and myositis. Antibodies targeting the Smith/RNP antigenic complex also recognize Smith antigens, therefore, the Smith antibody response must be considered when interpreting these results.

Note: An affinity purified RNP/Sm antigen complex is used in this assay.

HOTLINE NOTE: There is a unit of measure change associated with this test.
Change the unit of measure for component 0050470, Smith/RNP (ENA) Ab, IgG from AU/mL to Units.

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

Patient History for Fetal Molecular Testing

Time Sensitive

Additional Technical Information

Specimen Required: Collect: **Cultured amniocytes or Cultured CVS**
AND Maternal Whole Blood Specimen: Lavender (EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
Specimen Preparation: **Cultured Amniocytes or Cultured CVS**: Transfer cultured amniocytes or cultured CVS to two T-25 flasks at 80 percent confluence (Min: one T-25 flask at 80% confluence). Backup cultures must be retained at the client's institution until testing is complete. **If the client is unable to culture amniocytes or CVS, this can be arranged by contacting ARUP Client Services at (800) 522-2787. Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to test submission.**
Maternal Whole Blood Specimen: Transport 2 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: **Cultured Amniocytes or Cultured CVS:** CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of collection due to viability of cells.
Maternal Whole Blood Specimen: Room temperature.
Remarks: **Please contact an ARUP genetic counselor at 800-242-2787 x2141 prior to sample submission. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services at (800) 522-2787.**
Stability (collection to initiation of testing): **Cultured Amniocytes or Cultured CVS:** Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Maternal Whole Blood Specimen: Room Temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable

0051508 Thanatophoric Dysplasia, Types 1 and 2 (*FGFR3*) 13 Mutations, Fetal — TD PAN FE

Specimen Required: Collect: Cultured Amniocytes, Cultured CVS, or Amniotic fluid (direct).
Maternal Whole Blood Specimen: Lavender (EDTA), pink (K_2EDTA), or yellow (ACD Solution A or B).
Specimen Preparation: **Cultured Amniocytes or Cultured CVS:** Transfer cultured amniocytes or cultured CVS to two T-25 flasks at 80% confluence. (Min: one T-25 flask at 80% confluence). Backup cultures must be retained at the client's institution until testing is complete. **If the client is unable to culture amniocytes or CVS, this can be arranged by contacting ARUP Client Services at (800) 522-2787. Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to test submission.**
Amniotic Fluid (direct): 10 milliliters
Maternal Whole Blood Specimen: Transport 2 mL whole blood (Min: 1mL).
Storage/Transport Temperature: **Cultured Amniocytes or Cultured CVS: CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of collection due to viability of cells.
Amniotic fluid (direct): Ship room temperature.
Maternal Whole Blood Specimen: Room temperature.
Remarks: Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to sample submission.. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services at (800) 522-2787.
Stability (collection to initiation of testing): **Cultured Amniocytes or Cultured CVS:** Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Amniotic Fluid (direct): Room temperature: 48 hours; Refrigerated: 72 hours; Frozen: Unacceptable
Maternal Whole Blood Specimen: Room temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable

0091585 Tin Total Quantitative, Serum or Plasma — TIN SP

Specimen Required: Collect: Royal Blue (K_2 EDTA), Royal Blue (Na_2 EDTA), or Royal Blue (No Additive).
Specimen Preparation: Separate from cells ASAP or within 2 hours of collection. Transfer 1 mL serum or plasma to an Acid Washed Transfer Vial (ARUP supply #54350) available online through eSupply using ARUP Connect™ or contact ARUP Client Services at (800) 522-2787. (Min: 0.4 mL)
Test is not performed at ARUP; separate specimens must be submitted when multiple tests are ordered.
Storage/Transport Temperature: Refrigerated. Also acceptable: Room temperature or frozen.
Unacceptable Conditions: Separator tubes.
Stability (collection to initiation of testing): Ambient: 28 days; Refrigerated: 28 days; Frozen: 28 days

CPT Code(s): 83789

2005476 von Willebrand Disease, Platelet Type (*GP1BA*) 3 Mutations — GP1BA SEQ

Methodology: Polymerase Chain Reaction/Sequencing

Specimen Required: Collect: Lavender (EDTA), pink (K_2EDTA) or yellow (ACD Solution A or B).
Specimen Preparation: Transport 2 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: Refrigerated.
Stability (collection to initiation of testing): Ambient: 7 days; Refrigerated: 1 month

Interpretive Data: By report

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.