HOTLINE: Effective August 17, 2020

3001053 Red Blood Cell Antigen Genotyping RBC GENO

Performed: Sun-Sat
Reported: 3-10 days

Specimen Required: Collect: **Genotyping:** Lavender (K_2EDTA), Pink (K_2EDTA) OR
Fetal Genotyping: Amniotic fluid OR two T-25 flasks at 80 percent confluency of cultured amniocytes. 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 (K_2EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
Specimen Preparation: **Genotyping:** Transport 3 mL whole blood. (Min: 1 mL)
Amniotic Fluid: Transport 10 mL unspun fluid. (Min: 5 mL)
Cultured Amniocytes: Fill flasks with culture media. Transport two T-25 flasks at 80 percent confluency of cultured amniocytes 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: **Whole Blood or Maternal Cell Contamination Specimen:** Refrigerated.
Amniotic fluid: Room temperature.
Cultured Amniocytes: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of shipment due to liability of cells.
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 specimen in sodium heparin 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

Interpretive Data:

Background Information for Red Blood Cell Antigen Genotyping:
Characteristics: Erythrocyte alloimmunization may result in hemolytic transfusion reactions or hemolytic disease of the fetus and newborn (HDFN). Clinical presentation is variable and dependent on the specific antibody and recipient factors.
Incidence: Erythrocyte alloimmunization occurs in up to 58 percent of sickle cell patients, up to 35 percent in other transfusion-dependent patients, and in approximately 0.8 percent of all pregnant women.
Inheritance: Typically co-dominant for red blood cell (RBC) antigens, autosomal recessive for hemoglobin S (HbS).
Cause: Antigen-antibody mediated red-cell hemolysis between donor/recipient or transferred maternal antibodies.
Variants Tested: See the "Additional Technical Information" document.
Clinical Sensitivity: >99 percent for c (RH4), C (RH2), e (RH5), E (RH3), k (KEL2), K (KEL1), Jka (JK1), Jkb (JK2), Fya (FY1), Fyb (FY2), M (MNS1), N (MNS2), S (MNS3), s (MNS4). Unknown for Kpa (KEL3), Kpb (KEL4), Jsa (KEL6), Jsb (KEL7), Lua (LU1), Lub (LU2), Dia (DI1), Dib (DI2), Coa (CO1), Cob (CO2), Doa (DO1), Dob (DO2), Joa (DO5), Hy (DO4), LWa (LW5), LWb (LW7), Sc1 (SC1), Sc2 (SC2), U (MNS5), V (RH10), VS (RH20), Hemoglobin S (HbS).
Methodology: Immucor PreciseType™ HEA Molecular BeadChip which is FDA-approved for clinical testing. Predicted phenotypes are reported for each antigen and HbS based on the variants tested.
Analytical Sensitivity and Specificity: >99 percent for c (RH4), C (RH2), e (RH5), E (RH3), k (KEL2), K (KEL1), Jka (JK1), Jkb (JK2), Fya (FY1), Fyb (FY2), M (MNS1), N (MNS2), S (MNS3), s (MNS4). Unknown for Kpa (KEL3), Kpb (KEL4), Jsa (KEL6), Jsb (KEL7), Lua (LU1), Lub (LU2), Dia (DI1), Dib (DI2), Coa (CO1), Cob (CO2), Doa (DO1), Dob (DO2), Joa (DO5), Hy (DO4), LWa (LW5), LWb (LW7), Sc1 (SC1), Sc2 (SC2), U (MNS5), V (RH10), VS (RH20), Hemoglobin S (HbS).
Limitations: Only the targeted variants will be interrogated. Rare nucleotide changes leading to altered or partial antigen expression and null phenotypes may not be detected by this assay. This assay does not assess for RhD nor is it designed to diagnose sickle cell disease. Patients who have had hematopoietic stem cell transplants may have inconclusive results on this test. Abnormal signal intensities may result in indeterminate genotyping results for all tested antigens/HbS.

For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

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