

**TEST CHANGE**

Ashkenazi Jewish Diseases, 16 Genes

0051415, AJP

**Specimen Requirements:**

**Patient Preparation:**

**Collect:** Whole blood: Lavender (EDTA), pink (K 2 EDTA), or yellow (ACD solution A or B). Fetal specimens: 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, order ARUP test Cytogenetics Grow and Send (test code 0040182) in addition to this test and ARUP will culture upon receipt (culturing fees will apply). If you have any questions, contact ARUP's Genetics Processing at 800-522-2787 ext. 3301. ~~AND WITH maternal whole cell contamination specimen: Whole blood: lavender (K2 or K3 EDTA)~~ Lavender (EDTA), pink (K2 EDTA K-2-EDTA), or yellow (ACD solution A or B).

**Specimen Preparation:** Whole blood: Transport 3 mL whole blood. (Min: 1 mL) ~~Fetal Specimens:~~ Cultured amniocytes OR 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. ~~AND maternal whole blood: transport 2~~ ~~Maternal cell contamination specimen: Transport 3 mL whole blood- (mMin: 1 mL).~~

**Transport Temperature:** Whole blood ~~or maternal cell contamination specimen:~~ Refrigerated. ~~Fetal specimens:~~ Cultured amniocytes OR cultured CVS: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of shipment due to lability of cells ~~ANS maternal whole blood: room temperature. Also acceptable: refrigerated.~~

**Unacceptable Conditions:** Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in glass collection tubes.

**Remarks:**

**Stability:** Whole blood ~~or maternal cell contamination specimen:~~ Ambient: 72 hours; Refrigerated: 1 week; Frozen: ~~unacceptable 1 month~~ Fetal specimens : Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable ~~AND Maternal whole blood: Ambient: 72 hours; Refrigerated: 1 week. Frozen: Unacceptable.~~

**Methodology:** Polymerase Chain Reaction (PCR)/Fluorescence Monitoring

Performed: Varies

Reported: 5-10 days

Note: Cystic fibrosis (CF) carrier testing is NOT included as part of this panel. Please order Cystic Fibrosis (CFTR) Expanded Variant Panel (ARUP test code 2013661) to assess CF carrier status. Any submitted fetal specimens will have Maternal Cell Contamination, Fetal Sample, added on by ARUP. Additional charges will apply.

CPT Codes: 81401, 81209, 81200, 81260, 81242, 81251, 81250, 81479, 81205, 81290, 81400, 81330, 81255

New York DOH Approval Status: This test is New York DOH approved.

Interpretive Data:

Refer to report

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

Reference Interval: