

Maternal Cell Contamination Test

Background

The contamination of fetal samples with maternal cells is cause for concern in prenatal testing. The frequency of maternal cell contamination varies considerably due to sampling protocol, operator and culturing of cells from the fetal sample. The presence of maternal cells does not always lead to a prenatal diagnostic error. The level of maternal cell contamination that could adversely affect a prenatal test result varies among different genetic tests, as it is highly dependent upon the technology employed by the testing laboratory. Maternal cell contamination testing detects the presence and estimates the percentage of contamination. This test result, in conjunction with the prenatal test result, the sensitivity of the prenatal test to maternal cell contamination, and the known genotype of the mother, provides the basis for accurate prenatal diagnosis. At least five polymorphic loci located on different human chromosomes are amplified by PCR from the fetal and maternal samples. After capillary electrophoresis, the alleles in the maternal and fetal samples are compared and the percent contamination determined.

Indications for Testing

-Prenatal diagnosis (amniocentesis or chorionic villus sampling)

Genetic Counseling

Genetic counseling can be useful to patients and families considering genetic testing. The laboratory can provide referrals to genetics clinics in the patient's locale or a listing can be found at www.genetests.org

Ordering

Call the Genetics Lab with information on the fetal and maternal samples and expected dates of receipt.

1. Obtain a blood sample from the mother of the fetus. Provide chorionic villus tissue for direct testing, or send the tissue or amniotic fluid to a cytogenetics laboratory for cell culturing.
2. Fill out a Clinical Lab Request - Genetics for each patient
(available at <http://depts.washington.edu/labweb/Divisions/MolDiag/MolDiagGen/index.htm>).
-Request: "Other _____" and write in **MCC**; also select the test for prenatal diagnosis.
Provide items needed for test interpretation:
-Clinical and genotypic information for mother and father of the fetus
3. Call Laboratory Medicine Community Services at (206)598-6066 to arrange the best method of shipment.

Sample Requirements and Specimen Handling

TWO samples are required:

1. Fetal sample: cultured amniocytes in T25 or T75 flask; Chorionic villus tissue in sterile tube or culture media; or purified DNA (3 ug). Amniotic fluid is not acceptable.
2. Maternal Sample: Whole blood - EDTA (purple top) - adults - 5 mL or purified DNA (3 ug)
Blood sample should be received within 72 hours of collection; heparin (green top) tubes are not acceptable.

Optional: Paternal sample: Often helpful if provided; sample requirements as for maternal sample.

Test Frequency and Reporting

Prenatal testing and maternal cell contamination testing are both expedited. A written interpretative report is issued.

References

Steinberg S, Katsanis S, Moser A, Cutting G. Biochemical analysis of cultured chorionic villi for the prenatal diagnosis of peroxisomal disorders: biochemical thresholds and molecular sensitivity for maternal cell contamination detection. *J Med Genet* 42:39-44, 2005

Frederickson RM, Wand HS, Surh LC. Some caveats in PCR-based prenatal diagnosis on direct amniotic fluid versus cultured amniocytes. *Prenat Diagn* 19:113-117, 1999.

Stojilkovic-Mikic T, Mann K, Docherty Z, Mackie Ogilvie C. Maternal cell contamination of prenatal samples assessed by QF-PCR genotyping. *Prenat Diagn* 25:79-83, 2005.