

IDENTITY TESTING: MCC/FETAL SEXING

Individuals can be distinguished from one another by DNA fingerprinting, which compares variable DNA markers in different regions of the genome. Each person has his or her own pattern of DNA markers - a DNA 'fingerprint'. PCR amplification followed by capillary electrophoresis allows comparison of these markers between different individuals.

DNA fingerprinting can also be used to evaluate the purity of tissue samples obtained for other kinds of tests. Maternal cell contamination (MCC) of fetal cell samples obtained for prenatal analysis can be assessed in this way, and DNA fingerprinting can also be used to distinguish between the X and Y chromosomes for fetal sex determination.

TEST METHODS

Identity testing is performed by studying 15 DNA microsatellite markers and a sex-differentiating marker (D5S818, vWA, D13S317, TH01, D7S820, TPOX, CSF1PO, D8S1179, D21S11, D3S1358, D16S539, D2S1338, D19S433, D18S51, FGA and AMELX/Y). PCR amplification of these targeted regions followed by capillary electrophoresis allows comparison of these markers in the samples provided.

The test sensitivity level is 5% for detection of mixed samples.

FETAL SEXING

Fetal sexing tests for the presence of the sex chromosomes in a fetal sample using the sex differentiating markers amelogenin and/or ZFX/ZFY. Males fetuses are identified by the presence of an X and Y chromosome. Female fetuses are identified by the presence of an X chromosome and the absence of a Y chromosome.

The quantity of the X and Y chromosomes cannot accurately be determined by this method, but can be determined by cytogenetic testing.

MATERNAL CELL CONTAMINATION (MCC)

To ensure that a large proportion of maternal cells are not present in the fetal cell sample, maternal cell contamination studies compare the mother's DNA fingerprint to that of a prenatal or perinatal tissue sample.

Fetal cells contain one copy of each marker from each parent. The purity of the fetal sample is confirmed when there is a single maternal contribution at each marker. Maternal and fetal cell mixtures can be detected at levels as low as 5%.

Disease-specific analysis of a contaminated fetal sample may be adversely affected or inconclusive. This may still occur at ratios lower than that detectable by this test.

For More Information

Understanding Gene Testing:
<http://www.accessexcellence.org/AE/AEPC/NIH/index.html>

Waye JS (1993) Forensic identity testing using highly polymorphic DNA markers: current status and emerging technologies. *Transfusion Medicine Review* 7: 193-205.

To locate a genetics center near you, please visit the Canadian Association of Genetic Counsellors website at www.cagc-accg.ca or the National Society of Genetic Counsellors website at www.nsgc.org



1. DNA fingerprinting does not map the entire genome of the samples. The test will detect the presence of sex chromosomes, however it will not detect mutations in genes on the sex chromosomes.

2. Maternal cell contamination of fetal samples is detectable by molecular analysis at levels as low as 5%. The analysis of samples contaminated at ratios lower than this may be inconclusive.

3. This test's performance characteristics were validated by the Genome Diagnostics Laboratory at the Hospital for Sick Children. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes.