



Targeted Variant

Test Method: DNA extraction (if applicable); amplification of the specified region by polymerase chain reaction (PCR); cycle sequencing of each segment in the forward and reverse directions; analysis of each sequence by capillary electrophoresis; review of sequence data (human reference genome: GRCh37/hg19) by multiple staff members.

Analytical Sensitivity: > 99% accuracy for nucleotides evaluated; our analysis parameters are not designed to detect mosaicism.

Maternal Cell Contamination Study

Test Method: DNA extraction (if applicable); multiplex amplification of fifteen polymorphic markers and one sex determination marker in each sample; capillary electrophoresis of both samples; analysis and review by multiple staff members.

Prenatal Targeted Variant

Test Method: DNA extraction (if applicable); amplification of the specified region by polymerase chain reaction (PCR); cycle sequencing of each segment in the forward and reverse directions; analysis of each sequence by capillary electrophoresis; review of sequence data (human reference genome: GRCh37/hg19) by multiple staff members. MCC: DNA extraction (if applicable); multiplex amplification of fifteen polymorphic markers and one sex determination marker in each sample; capillary electrophoresis of both samples; analysis and review by multiple staff members.

Analytical Sensitivity: > 99% accuracy for nucleotides evaluated; our analysis parameters are not designed to detect mosaicism.