Chromosomal Microarray: CytoScan SNP Array POC

**Test Code:** CMPOC

**Turnaround time:** 2 weeks - 3 weeks (All abnormal findings are called out immediately. Confirmatory testing for abnormalities will delay reporting.)

**CPT Codes:** 81229 x1

### Condition Description

At least 50% of first trimester miscarriages are caused by a chromosomal imbalance. Chromosomal microarray analysis of uncultured products of conception (POC) samples increases the likelihood of detecting clinically significant genetic imbalances as compared to conventional G-banded analysis. Since the Chromosomal Microarray, CytoScan SNP Array POC uses DNA extracted from uncultured cells, rather than cultured cells, tissue culture failure is avoided. Microarray testing detects smaller imbalances than conventional G-banded analysis, especially since POC samples often have poor chromosome morphology. The CytoScan SNP Array POC detects triploidy, trisomy, monosomy, and chromosome deletions and duplications. This test is a cost effective method to detect chromosomal imbalances, which is important to determine the recurrence risk for future pregnancies.

**Reference:**

### Indications

The CytoScan SNP Array POC is warranted for the evaluation of any spontaneous pregnancy loss and can provide a diagnosis and recurrence risk for future pregnancies.

### Methodology

DNA isolated from the POC sample is hybridized to an array containing oligonucleotide and SNP probes across the genome to detect copy number imbalances and regions of homozygosity.

The CytoScan SNP Array POC consists of 2.6 million markers (including 750,000 SNPs) which allows for the detection of both copy number variation (CNV) and large stretches (>10 Megabases (Mb)) of absence of heterozygosity (AOH), which can result from uniparental disomy (UPD) or common parental descent. The design is based on recommendations from the International Standards for Cytogenomic Arrays (ISCA) Consortium (Baldwin et al. (2008) *Genet Med*; 10(6):415-429).

### Detection

Deletions and duplications of 400 kilobases (kb) or greater are detected and will be reported. Smaller deletions or duplications in regions of known microdeletion/microduplication syndromes or in clinically relevant genes will also be reported.

This test is designed to detect whole and partial chromosome UPD, long stretches of absence of heterozygosity (AOH) greater than 3 Mb, and AOH in clinically relevant regions. In addition, this test can also detect triploidy, a common cause of miscarriages. Possible UPD will be reported when a chromosome has at least one homozygous regions >10 Mb. Homozygosity due to apparent common descent will be reported when >5% of the autosomal genome is present in long stretches of AOH. These regions of AOH will be specified to consider recessive risk alleles.

Chromosomal Microarray, CytoScan SNP Array POC will not detect balanced translocations, balanced inversions, imbalances smaller than the resolution of this array, point mutations, or low level mosaicism (usually less than 25%).

### Specimen Requirements

**Additional Specimen Collection/Handling Instructions Required for this Test**

Tissues fixed in formalin cannot be used.

Submit only 1 of the following specimen types

**Type: Chorionic Villi**

Specimen Requirements:

Collect 15-30 mg of chorionic villi/placental tissue using sterile technique. Place in sterile tube(s) with EGL transport media or other sterile culture media.

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample overnight at room temperature for receipt at EGL within 24 hours of collection.

**Type: Tissue Biopsy**

Specimen Requirements:

Obtain 1-2 cm piece of fetal tissue and place in sterile container with EGL transport media or other sterile culture media. Use sterile dissection (no
prep) for internal tissues.

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample overnight at room temperature for receipt at EGL within 24 hours of collection.

**Special Instructions**

Maternal cell contamination studies are warranted to rule out the possibility of maternal cells in female products of conception. Submit a 5cc maternal whole blood sample in an EDTA tube. Test code: CMCCS. CPT code: 81265 (x1).

Sample Storage and Data Usage: As a participant in the ISCA (International Standard Cytogenomic Array) Consortium, EGL Genetics retains patient samples indefinitely for validation, educational purposes and/or research. The submitted clinical information and test results are also included in a HIPAA-compliant, de-identified public database as part of the National Institute of Health's effort to improve diagnostic testing. For information about the database visit the consortium website at https://www.iscaconsortium.org/. Confidentiality of each sample is maintained.

Patients may request to have their samples discarded upon test completion and to opt-out of participation in the database by:
1) Checking the box provided on the test requisition or consent form
2) Calling the laboratory at 470-378-2200 and asking to speak with a laboratory genetic counselor

**Related Tests**

- Maternal cell contamination studies (CMCCS)
- Targeted testing by FISH is available to family members of an individual with a deletion or duplication detected by microarray.
- Prenatal Chromosomal Microarray (EmArray Cyto) (CMPRE)
- EmArray Cyto (VA)
- EmArray Cyto+SNP (CMSNP)