



Consent for SNP Chromosomal Microarray

Patient Name: _____ Patient DOB: _____

Ordering Clinician Name: _____

- ❖ Chromosomal microarray (CMA) is a test used to evaluate cells for gains or losses of chromosome material. CMA reveals similar kinds of information as a standard chromosome analysis but at a much higher level of detail.
- ❖ The methods used to collect cells have associated risks and should be reviewed separately. If the sample is insufficient or damaged in shipment, additional sample or growing (culturing) of the sample may be required to complete the test.
- ❖ A potential benefit of CMA is the identification of a chromosomal abnormality. Identifying an abnormality may be useful in directing additional care, or evaluating/monitoring an individual or an ongoing pregnancy.
- ❖ CMA is not able to detect every genetic condition. CMA will not detect balanced chromosome rearrangements, point mutations within a gene, abnormalities smaller than the resolution of the array, and/or low levels of mosaicism.
- ❖ Accurate interpretation of CMA results is dependent upon the accuracy of the information submitted with the sample, including clinical history, previous test/radiographic/ultrasound findings, and family history.
- ❖ Possible CMA results include:
Benign (normal): No known abnormality detected
Pathogenic: The gain or loss is a genetic factor in the medical problem
Uncertain significance: The gain or loss may or may not be the cause of the medical problem
- ❖ SNP (single nucleotide polymorphism) CMA can reveal information that is NOT diagnostic but can provide additional information. For example, excessive ROHs (regions of homozygosity) are reported, *and if clinically indicated*, may help guide further genetic testing (e.g. single autosomal recessive gene, UPD or methylation testing).
- ❖ Unexpected results may be detected that reveal information that is not directly related to the clinical reason for ordering the test.
- ❖ SNP CMA results may reveal that parents are closely related.
- ❖ For an uncertain or pathogenic result, additional testing of parents or other family members may be necessary to determine whether the gain or loss is a new event (*de novo*) or inherited. Follow-up testing may allow for further interpretation of the clinical significance of the finding for the patient, pregnancy, and/or family.
- ❖ Genetic testing of parents has the potential to reveal non-paternity/misattributed parentage. Misinformation regarding the true biological relationships can result in reporting errors.
- ❖ In the case of prenatal CMA testing, a normal result does not guarantee the birth of a healthy child. Birth defects and/or developmental disabilities occur in 3-5% of live births; however, the cause is not always detectable by CMA.
- ❖ Due to the complexity of genetic testing and potential implications, test results will be reported only to the ordering provider(s).
- ❖ Use of this form is highly recommended.

The risks, benefits, and limitations of microarray testing have been explained to me. I have elected to proceed with chromosomal microarray analysis (CMA).

Patient (or Patient Guardian) Signature

Date

I have discussed the risks, benefits, and limitations with the patient/family and all questions have been answered.

Clinician Signature

Date

Laboratories are encouraged to participate in the Clinical Genome Resource (ClinGen) efforts to submit clinical information and test results to a HIPAA-compliant, de-identified public database as part of the NIH's effort to improve diagnostic testing and understanding of the relationships between genetic changes and clinical symptoms. Confidentiality is maintained. **Patients may request to OPT-OUT of this scientific effort by 1) checking this box OR 2) calling the laboratory at (303) 724-5701 and asking to speak with a genetic counselor.**