INTRODUCTION
You and your doctor have decided to do chromosomal genetic testing. Your doctors usually offer these tests only if they suspect a problem after looking at results of blood tests, ultrasound results or your family history. Pregnant women can do these tests to see if the fetus has changes in its chromosomes that could cause serious problems in a child, such as intellectual disability or birth defects.

Prenatal Chromosome Microarray (CMA) is a type of genetic testing. It is used to look for certain kinds of genetic changes in the fetus. We recommend that you meet with a professional genetic counselor before you have this test. A genetic counselor helps you understand the possible risks and benefits of having prenatal chromosome microarray testing.

Prenatal CMA testing uses cells from the water surrounding the fetus in the womb (obtained by a procedure called amniocentesis), or from the placenta (obtained by a procedure called chorionic villus sampling). Before you decide to have these procedures you should discuss their risks with the doctor who will perform them or with a genetic counselor.

Your signature below indicates that you understand the risks and benefits of CMA testing, that you were able to ask questions and received satisfactory answers.

OVERVIEW OF PRENATAL CHROMOSOMAL MICROARRAY (CMA)
• Our bodies are made up of billions of cells. Inside these cells are chromosomes, which are thread-like strands of DNA that contain genes.

• A child’s cells contain chromosomes inherited from each parent. However, sometimes there are abnormalities in a child’s chromosomes that develop just before or after conception that are not present in the parents.

• A standard chromosome test, also called a karyotype, uses a microscope to see whether there are extra or missing whole chromosomes or large pieces of chromosomes.

• Chromosomal microarray (CMA) can look for much smaller gains or losses of chromosome material. When used to look at chromosomes from fetal cells, CMA testing can find changes that a standard chromosome test cannot detect that could cause serious diseases or birth defects in a child.

• When the CMA test shows a gain or loss in chromosome material there are 3 possibilities:
  1) The gain or loss may be known to cause problems such as birth defects or disability
  2) The gain or loss may be known not to cause problems
  3) It may not be known whether the gain or loss can cause problems

• When a gain or loss of chromosomal material is inherited from a healthy parent it is much less likely to cause a problem for a fetus or a child. Testing the parents or other family members will tell whether the gain or loss has been inherited or is a new change in the fetus. We often ask for parental blood samples with the prenatal sample, to allow parental testing to be done quickly if needed.

• If there is a gain or loss, the laboratory may do more tests to understand an unusual result. In this case, the laboratory may need additional fetal or parental samples.

• Some CMA gains or losses can cause diseases that vary from mild to severe. It may not be possible to predict what the outcome will be in a particular individual.

• A CMA may show changes that will not affect health during infancy or childhood, but may cause illness in adult life. These findings in the fetus may also show that one of the parents has the same problem but has not yet had signs of the disease.

• CMA testing cannot detect all chromosome changes that can cause genetic disease. Changes that occur in some, but not all cells in the sample may not be detected. Very small changes within a single gene will not be detected. It cannot detect changes that occur in parts of chromosomes not included on the microarray.

• A normal CMA result does not guarantee a healthy child. Not all birth defects, genetic disorders or health problems result from extra or missing or interrupted segments of DNA.
MORE INFORMATION ABOUT GENETIC TESTING

- It is important that you provide your health care team with **accurate medical information about the pregnancy, the fetus and your family medical history**. Correct interpretation of test results depends on this information.
- Genetic testing of parents may reveal information about yourself, your fetus or your family. **For example, the test may reveal a different father, the use of an egg or sperm donor, or that the parents are close relatives.** It is important to disclose this information before the test, to fully understand the results.
- CMA is a complex test using advanced technology. There are some times when results cannot be interpreted.
- Your OB-GYN provider and the physician who ordered the test or your genetic counselor will receive the results of the tests. The results are confidential to the extent allowed by law. Participation in genetic testing is completely voluntary.
- Health care insurance sometimes pays for this testing, but not always. You give permission to Brigham and Women’s Hospital to bill your insurance and provide information necessary for reimbursement. You understand that you are responsible for amounts not paid by insurance, for services that are not covered and not authorized or for other reasons. You authorize a copy of this consent form to be used in place of the original.

**Use of specimens:** The BWH Center for Advanced Molecular Diagnostics may keep patient samples for validation, educational purposes, and/or research. For cytogenetic and molecular genetic tests, the clinical information and test results are included in public databases that do not contain any personal information, in respect of privacy regulations. Each sample is kept in strict confidence. This information will help the National Institute of Health’s effort to improve diagnostic testing and our understanding of genetic changes and illness. (For information about the National Institute of Health database visit http://www.iccg.org/)

**Signatures:** My signature below shows that I choose voluntarily to have chromosomal microarray testing for my pregnancy. I understand that the genetic analysis performed by the BWH Center for Advanced Molecular Diagnostics in no way guarantees my health, the health of an unborn child, or the health of other family members.

Patient Name (printed) __________________________________________

Patient Signature ________________________________________________  Date ________  Time ________ AM/PM

Clinician Signature

I understand that interpretation of the results of microarray testing on fetal cells may require similar testing of my own chromosomes. I hereby give permission for the laboratory to use my blood sample for this testing if indicated.

Patient Name (printed) __________________________________________

Patient Signature ________________________________________________  Date ________  Time ________ AM/PM

Father of Fetus’s Signature _________________________________________  Date ________  Time ________ AM/PM

Clinician Signature