BAYLOR MIRACA GENETICS LABORATORIES
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PRENATAL CMA CONSENT FORM

Consent Form for Prenatal Studies Using Chromosomal Microarray Analysis

Background

You are considering the genetic test called Chromosomal Microarray Analysis (abbreviated CMA) for your current pregnancy. The purpose of this document is to provide information about the test so that you can decide whether it is right for you. This information is meant to be used in addition to your discussion with a genetic counselor or genetic physician and an information brochure on the CMA test that you may have received. Only after you have been offered professional genetic counseling and when you understand all the information and your questions have been answered should you make your decision about whether or not you wish to have the CMA test. If you decide to have the CMA test, you will be asked to sign the last page of this document, indicating that you understand the information provided and wish to have testing. You will be given a copy of this document for your records.

CMA is a method of looking at the structure and number of the chromosomes in our body. All cells in our body, except for red blood cells, contain 46 chromosomes (23 pairs). Chromosomes are the structures in cells that carry our genetic information. The development of our bodies and brains is very dependent on having the correct number and proper structure of chromosomes. When a chromosome, or even a very small piece of a chromosome, is extra or missing, problems in development and/or birth defects may result. An example of a problem due to an extra copy of an entire chromosome 21 is Down syndrome.

Cells or blood from our body can be used to look at our chromosomes. The most common way to look at chromosomes is in a test called a karyotype analysis. In a karyotype, the number of chromosomes can be counted and the structure of chromosomes can be determined, but in a limited way. Another method, FISH testing, looks at very small areas on the chromosomes that cannot be seen on a karyotype test.

A simplified way of thinking about the new CMA method is that all the different chromosome tests, such as a karyotype and hundreds of FISH tests, have been combined in a single test. In this method, many pieces or spots of DNA (the material that carries the genetic code that makes up our body), from specific parts of all of the chromosomes are placed on a glass slide, called a microarray or DNA chip. The DNA from the person or fetus to be tested is matched up against the normal DNA spots on the chip. This test is able to determine whether the tested DNA from the person or fetus has any missing or extra copies (copy number) of the parts of the chromosomes that are represented on the glass slide.

In addition, the CMA test can detect an abnormal inheritance pattern of the chromosomes called uniparental disomy (UPD). UPD occurs when both of the chromosomes in a pair (or part of a chromosome) are inherited from one parent and no copies are inherited from the other parent. Most of the time, UPD does not cause any abnormalities. However, in some instances, UPD may result in a genetic disorder.

The CMA test can also detect regions of genetic similarity, called absence of heterozygosity (AOH). Genetic similarity is an area of the chromosome that does not show the normal differences we expect to see between the material passed down (inherited) from the mother and the father. If multiple regions of genetic similarity (AOH) are found in a person's CMA, this suggests that the person's parents are biologically related to one another (called consanguinity). The amount of AOH observed can also provide information about how closely related the parents might be however, the data does not determine the exact identity and relationship between the parents. If a region of genetic similarity is found by CMA, it could provide a clue to a specific gene of interest and more genetic testing may be recommended.

Purpose

The CMA analysis is performed as a diagnostic test in the Baylor Miraca Genetics Laboratories on a fee for service basis. The purpose of this consent form is to explain the risks and benefits of having CMA testing for your current fetus.

Procedures

You are eligible to have CMA testing if you are pregnant and you and your doctor agree that the pregnancy will be monitored by amniocentesis or chorionic villus sampling. You and your doctor should discuss and agree on a plan for prenatal diagnosis in your pregnancy before considering whether you should have CMA testing. If you still have questions about whether amniocentesis or chorionic villus sampling is right for you and your pregnancy, we ask that you speak to your doctor. If you decide to learn more about CMA testing, you will undergo the following procedures:

1. Pre-test counseling and informed consent.

In most cases, your doctor has referred you to speak to a genetic counselor about prenatal diagnosis and CMA testing. Professional genetic counseling is recommended prior to consenting to CMA testing. You will have a standard genetic counseling session and your insurance will be billed for the professional fee of the genetic counselor. The genetic counselor will ask you questions about your health information and the health information of your family, including your parents, siblings, aunts, uncles, cousins, grandparents, your children and previous pregnancies. The same information will be collected about your partner and your partner's family. The genetic counselor will also ask you about your current pregnancy and discuss with you the risks and benefits of the amniocentesis or chorionic villus sampling procedure, if you still have questions about these. After reviewing the information that you provide, the genetic counselor will discuss with you any other genetic risks and tests, in addition to the amniocentesis or chorionic villus testing and chromosome analysis, that may be appropriate for your pregnancy.

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If you would like to learn about CMA testing, you will be given this document to read. The genetic counselor will explain CMA testing to you and answer your questions. You can make your decision about whether you want the CMA test at any time up until you have the amniocentesis or chorionic villus sampling.

2. Sampling Procedures.

If you decide to have CMA testing, you will then have an amniocentesis or chorionic villus sampling procedure by your physician according to his or her usual practice standards. The amount of sample that can be obtained influences how quickly the laboratory will be able to complete the CMA test and report the results. We request that your doctor obtain a small amount of extra material for the CMA test. If you are having an amniocentesis, we request an amniotic fluid sample of approximately 30 cc and we recommend that your amniocentesis procedure not be performed before 16 weeks. We realize that the timing of your amniocentesis is a decision that will be made between you and your doctor; however, it is important to understand that your results may be delayed if a small amount of fluid (less than 4 teaspoons or 15 cc) is obtained. If you are having a chorionic villus sampling procedure, we will request that your doctor obtain an additional 30 mg of material. If your doctor feels that obtaining the extra material may pose a risk to your pregnancy, he or she can decide not to obtain the extra material. This may result in the CMA test taking longer to complete. Potential risks to obtaining the additional amniotic fluid or chorionic villi include cramping, or possibly miscarriage.

After the prenatal material is collected, a portion of the sample will be used by Baylor Miraca Genetics Laboratories for CMA testing. Chromosome analysis by karyotype will be performed as part of clinical care. The CMA test will be performed using materials and protocols developed at the BMGL and validated by the laboratory. This laboratory is certified by standards set by the Clinical Laboratory Improvement Acts (CLIA) and the College of American Pathologists. Both biological parents will also be asked to give a blood sample (approximately 3 teaspoons of blood) that may be used to help interpret the CMA results. It is important that both biological parents submit a blood sample to the lab. If a sample is not submitted from both the biological mother and father, the interpretation of the results may be compromised. If an abnormality is detected on the array that is too small to be detected by routine chromosome analysis, additional confirmation studies, such as FISH analysis, will be performed if technically possible.

3. Test Reporting.

It is anticipated that the CMA result will be available within 7 to 14 days from the date your amniotic fluid or chorionic villus sample reaches the laboratory. As stated above, the time to obtain results is dependent on the amount of sample the laboratory receives. If the laboratory determines that the size of the specimen will result in a reporting time of greater than 14 days, your doctor will be notified.

When the CMA results are complete and ready to be reported, we will send the results to your physician. Professional genetic counseling to discuss the CMA test results may be warranted. There are several categories of results that may be reported: these include 1) No clinically significant abnormality detected; 2) Clinically significant abnormality detected, known to be associated with a genetic condition; 3) Variation of uncertain significance detected in the fetus, but also in a parent. Based on our experience thus far, this has been seen in about 10% of cases. It is generally of low concern, but should be discussed with a genetic counselor; and 4) Variation of uncertain significance detected in the fetus, but not present in either parent. This is relatively rare (seen thus far in about 1% of cases) and requires detailed discussion with a physician or genetic counselor. In addition, regions of genetic similarity (AOH) may be reported if the CMA results indicate the possibility of uniparental disomy (UPD) or consanguinity. If a clinically significant abnormality has been detected, the genetic counselor and a physician who specializes in genetics will discuss the information with you. These individuals will review the results and the medical problems that may go along with the abnormality (For additional discussion, see Potential Risks section below). In the case of abnormal CMA results, a sample may be requested for confirmation of the prenatal diagnosis either at spontaneous or induced termination of the pregnancy or after the baby is born. Because the CMA test is new and very sensitive, there is the chance that you may be told that the CMA test has detected variations of uncertain significance as described under "Test Reporting," above. This means that the CMA test has found a variation (change or difference from the usual result), but that there is little or no medical knowledge about the particular change. In other words, the change may never have been described before and therefore, it cannot be determined, based on current medical information, whether it may cause medical problems. In these cases, the doctors and counselors will explain all of the available information about the change to help you understand the information and make a decision about the pregnancy. In most cases, it can be determined if the change in question is also present in one of the healthy parents. If this is the case, the change is less worrisome. This is the reason for requesting blood samples from both parents. Depending on your decision about the pregnancy, we may ask for a sample to confirm the lab finding and may ask to examine the baby after delivery and to review results of certain special tests ordered by the pediatrician as part of the clinical care of the baby.

The CMA test is a clinical diagnostic test and not a research test. However, we wish to continue to gather information about prenatal cases studied by CMA analysis. The information that will be collected will include the CMA test results and information about your pregnancy and your fetus. This information may be compiled in a cumulative report and published in the medical literature. Your identity will remain anonymous and no personal information that may identify you will be included in the report. We also ask your permission to retain your laboratory sample for internal laboratory quality assurance studies. If your sample is retained in the laboratory, the sample will be identified by number only.

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Potential Risks and Discomforts

1. While the CMA test is very accurate, it is possible that your fetus could have one of the medical conditions included in the CMA test, but the CMA test was unable to detect the condition. This is possible because many genetic syndromes have more than one cause. For some conditions included in the CMA test, 99% of the cases can be detected by the test, while for others it may be 70% or less. Therefore, it is possible that your pregnancy may be affected with one of the conditions tested by CMA, but that the test did not detect the condition.

2. Due to the fact that many different regions of the chromosomes and many different conditions are being analyzed, there is a risk that you will learn genetic information about yourself, your fetus, or your family that is not directly related to the reason for monitoring your pregnancy. This information might relate to diseases with symptoms that may develop in the future in your fetus or possibly yourself or other family members. It is important that the results of the CMA test be told to you by your doctor and/or the genetic counselor and genetic physicians and that if necessary, additional referrals to other medical professionals be made to explain the results and their effect on the health of your fetus, yourself or family members.

3. If the results of this testing are normal, your fetus could still have a birth defect(s) or mental retardation from causes that are not detected by the CMA test.

4. As with any genetic test, results may be unclear or indicate the need for further testing on other family members. In about 10% of cases, we will find a change in the fetus that is different from the most usual finding, but not clearly associated with a known genetic condition. In these cases, we will test the parents' blood sample in order to help determine if this change runs in the family. If we determine that the change runs in your family and is likely to be benign (it is also present in a healthy parent), you will be informed of these results. If study of the parents does not clarify the origin of the variation, additional studies may be recommended in order to give you the most accurate information about what the lab finding may mean for the health of your fetus. It is possible that additional information may come to light during these studies, such as non-paternity (the father of the fetus is not the real father). If additional studies are recommended, the lab will ask your physician or genetic counselor to discuss these studies with you and for you to give your permission before the lab proceeds. If you decide not to give the lab permission for additional testing, the lab may, in the future, perform studies on a research basis. In this case, the results will not be reported to you and will not be reported in any way that could identify the family.

5. Alternatively, it is possible that the test will detect an abnormality that is not present in the parents and for which there is very little or no medical information available to predict the type of problems that may develop. This may result in uncertainty as to the seriousness of the condition that has been found. This result is likely to occur in about 1% of cases in our current experience. The genetic counselor and genetic physicians will discuss with you all of the available information regarding your results to help you understand them and make an informed decision about your pregnancy.

6. The CMA results are confidential: they will only be released to your doctor and other medical professionals or other parties with your written consent. All laboratory data is confidential and will not be released within legal limits. You should understand that in rare cases, persons with genetic medical conditions have experienced problems with insurance companies, employers, and others.

7. You can withdraw your consent for testing at any time. If you sign the consent form, but you no longer wish to have your sample tested by CMA, you can contact your doctor to cancel the test. If testing is complete, but you have not received your results as yet, you can inform your doctor that you no longer wish to receive the results. If you withdraw consent for testing more than 24 hours after the sample is received by the laboratory, you will be charged for the full cost of the test.

8. As with all types of laboratory tests, there may be problems during the laboratory testing that result in delays in completing the test. In very rare instances, the test may not be completed due to technical problems or small sample size. If the laboratory is delayed in completing the test or may not be able to complete the test, you will be informed of this as soon as possible. At that point, you can decide whether or not you wish to continue with the testing. You will not be charged for the test if you withdraw from testing because the delay is due to a technical problem in the laboratory.

9. Information obtained from the CMA test may be used in scientific publications or presentations, but the identity of all persons studied will not be revealed in such publications/presentations. You should understand that your sample may be kept by the laboratory for quality assurance testing. In addition, your sample may be shared with other scientists who are doing research in genetic problems. If your sample is retained in the laboratory or shared with other scientists, your name and any other identifying information will be removed from the sample.

10. There is a small risk of bruising and bleeding at the puncture site when you (the parents) give a blood sample.

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Potential Benefits

The benefits of selecting this testing are that your prenatal sample will be tested by a highly sensitive method for finding chromosomal problems. This test is an expanded version of the currently available cytogenetic tests. Thus, this test may discover an abnormality that would not have been detected by the standard test. If an abnormality is found, this information may be important for you in many ways: it may give you important information about your pregnancy or it may help guide the medical decisions about your baby after delivery. However, you may receive no benefit from selecting this test.

Potential Conflicts and Financial Disclosure

In this section, we provide information regarding potential sources of conflict of interest. The performance of CMA testing provides a source of income for the Department of Molecular and Human Genetics (DMHG) at Baylor College of Medicine. This is similar to the usual conflicts in the practice of medicine as when a surgeon recommends a procedure for which there will be a substantial surgical fee.

The physicians and/or genetic counselors who have explained the CMA test to you today may be members of the Department of Molecular and Human Genetics of Baylor College of Medicine. They, as individuals do not receive financial incentives to refer specimens to the Baylor Miraca Genetics Laboratories. These physicians and other members of the Department of Molecular and Human Genetics may have given lectures about the use of CMA testing to professional groups and referring physicians and have published articles about the use of CMA in many clinical settings.

I have read the above consent form and hereby give permission for my prenatal sample to undergo testing by chromosome microarray analysis.

Patient Name

Patient Signature DATE: ____ / ____ / ____
MM DD YY

LAST UPDATED: 2/2015