Clinical Molecular and Genomic Pathology

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Informed Consent for Chromosomal Microarray Testing

Patient Name: _____ DOB: _____

Parent/Guardian Name: _____

I understand that:

My doctor or my child's doctor has ordered chromosomal microarray analysis (CMA) as part of my or my child's medical evaluation. The *purpose* of this test is to determine whether my (or my child's or my fetus's) sample has changes in the DNA copy number that may explain the clinical presentation. This test will reveal major chromosome abnormalities and sub-microscopic chromosomal imbalances and is considered to be greater than 99% accurate for these disorders.

CMA evaluates the amount of chromosomal information present and CMA test may be ordered to identify any regions of genomic imbalances in patients with dysmorphic features, unexplained mental retardation/developmental delay, autism spectrum disorder, and/or multiple congenital anomalies. Losses or gains of chromosomal information (copy number changes or variants) can result in above mentioned clinical conditions. CMA can detect some deletions or duplications that cause single gene or contiguous gene phenotypes. In addition, CMA can identify regions of homozygosity (ROH) that do not involve a copy number change, but may be associated with having two copies of genes from one parent and none from the other parent in a specific chromosome region. Or, it could mean that both parents share ancestry.

It is possible that the CMA test will detect a genetic abnormality for which there is currently very little medical information available to predict the type of clinical problems that may develop in an individual. While the test is very accurate, not every genomic abnormality (genetic defect) can be detected by a test. For some conditions, genomic gains or losses at a particular locus may represent only a certain percentage of the genetic changes associated with that given disorder. For instance, in some disorders, 99% of the cases may be detected by the test, while for others the detection rate may be 70% or less.

CMA cannot detect balanced chromosomal rearrangements and copy number variants below the stated resolution of the test. In addition, our test is not validated to detect mosaicism.

The results of CMA test may be reported including:

An Abnormal/Positive result indicates the presence of one or more regions of chromosomal imbalance associated with genetic disease, or the presence of one or more regions of homozygosity (ROH), which may represent shared ancestry or the possibility of a type of disorder known as an imprinting disorder.

A Normal result indicates the absence of clinical significant abnormality using this test, even if clinical diagnosis is still correct. This normal result may be due to currently available scientific knowledge. As new information is discovered, our understanding of how genetic changes cause disease improves.

A Variant of unknown significance result indicates that a copy number variant has been identified that is not known to be benign, but has also not been associated any specific disorders. Additional testing of family members may be recommended to help determine the clinical significance of the result.

This test may reveal some unexpected results. This test may show information about me, my child or other family members is not directly related to the clinical condition for which this test was ordered. This information may provide information about the risk for a different genetic disease with symptoms that may or may not be currently evident.

An error in the test interpretation may occur if the biological relationships of the family members are not correct. For example, loss or gain detected in an affected individual may be interpreted as clinical significant, if the abnormality is not detected in the parents. This interpretation is completely depends up on the information from biological parents. If the stated father of an individual is not true biological father, the test interpretation may be incorrect.

The parental testing may reveal that the parents share close common ancestors (consanguinity).

I understand that UKHC CMGP laboratory keeps test results confidential. My or my child's test results will be sent only to the healthcare provider who ordered the test, or his/her agent. My or my child's health care provider is responsible for interpreting the test results and explaining them to me.

No testing will be performed on my or my child's sample apart from that which is ordered by your or your child's physician. Additional testing requires the patient's/guardian's additional, express consent.

Signature: _____ Date: _____

Printed Name: _____Relationship to Patient: _____

Witnessed by: _____

Health Care provider's Statement: I have explained the genetic testing specified to this individual and addressed the limitations outlined above. I have obtained consent from the patient or parent or legal guardian for this testing.

Provider's Signature: _____ Date: _____

Printed Name: _____Phone: _____

Note to Ordering Clinician: UK HealthCare Clinical Molecular and Genomic Pathology (CMGP) laboratory encourages the discussion of the limitations and utility of a genetic test with the patient prior to specimen collection. This form is provided to address pertinent issues regarding prenatal Chromosomal Microarray (CMA) testing. If a signed consent is not submitted with the order, UKHC CMGP laboratory assumes that the ordering clinician has discussed the benefits and limitations of array testing with the patient and obtained the patient's informed consent.