ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

NCBI ClinVar

ClinGen Clinical Genome Resource

CYTOGENOMIC MICROARRAY TESTING AND THE CLINVAR DATABASE

Your doctor has ordered a cytogenomic microarray test as part of a medical evaluation for you or your child. This test is used to look for a genetic cause of problems in the physical, intellectual, and behavioral development of children and adults. It has significantly improved our ability to find the underlying cause of many developmental and medical problems, allowing families to learn about their specific results and make informed plans for medical and/or educational interventions.

The cytogenomic microarray test is a way for the laboratory to look at the entire genetic makeup of a person in order to find missing (loss) or extra (gain) pieces of chromosomes. Many of the losses and gains found by microarray testing are common, and their associations with medical problems are well understood. However, some findings have not been seen before. In these cases, it is not possible to determine if a particular rare result will cause medical problems.

More information is needed to understand rare results, and you can help!

A sample from you or your child will be sent to the ARUP Genomic Microarray Laboratory for microarray testing. ARUP Laboratories is a member of the Clinical Genome Resource (ClinGen), an organization of more than 100 laboratories working together to gather the information needed to understand the meaning of rare cytogenomic microarray results. ARUP contributes to ClinGen by sending results from all microarray tests performed, along with the reason for testing (such as autism or heart defect), to NCBI, the National Center for Biotechnology Information, which advances science and health by providing access to biomedical and genomic information. The central ClinGen database, called ClinVar, is supported by NCBI.

Privacy is of the utmost importance. All patient identity information is removed (de-identified) before results are submitted to ClinVar to ensure confidentiality of patient information. Researchers must request permission from the Data Access Committee, appointed by the National Institutes of Health, to use the de-identified information in the database.

On occasion, the physician who ordered your microarray test may contact you about specific research projects sponsored by ClinGen. You may be interested in participating and/or receiving information about some of these research studies. If so, your physician can give you the researcher's contact information. At no time will any researcher have access to contact information for you or your ordering physician.

The ClinVar database is possible only because of the results received for microarray tests contributed by individual patients. With your help and as the ClinVar database grows over time, laboratories will be able to use this information to determine the meaning of rare results. Patients with rare or uncertain results may then learn the true meaning of their results and optimize their medical care.

Opting out of the ClinVar Database

To learn more about ClinGen, log onto www.clinicalgenome.org. If you do not want your results to be submitted to the ClinVar database, you can choose not to participate (opt out). If you opt out, your microarray test and test results will not be affected. To opt out, contact ARUP Laboratories Genetic Processing at (800) 242-2787, ext. 3301, to request that your test results not be sent to ClinVar.