



## EmArray Cyto6000 ©2007 Information for Patients & Families

### Frequently Asked Questions:

- **What is the EmArray Cyto6000? How does it work?**

The EmArray Cyto6000 uses the newest advances in a technique called chromosomal microarray analysis. Such array analysis tests for many genetic disorders and can also detect very small missing or extra regions of chromosome material. Array analysis is greatly improved over standard testing for chromosome changes, which may miss a percentage of patients who have changes too small to be identified. Another method, called FISH testing, may also detect small chromosome changes, but FISH is limited in the number of regions that can be tested at one time. Therefore, one EmArray Cyto6000 can give results equal to hundreds of FISH tests combined.

EmArray Cyto6000 begins by collecting a blood sample from an individual. DNA obtained from the blood sample is tested on an array slide that holds selected regions of the chromosomes. The amount of the individual's DNA on the array and the amount of control DNA on the array are compared. Areas where there is less patient DNA represent regions of the chromosome that are missing or deleted. Areas where there is more patient DNA represent regions of the chromosome that are extra or duplicated.

The EmArray Cyto6000 is an oligonucleotide-based array or oligo array. Other names for an oligo array include: oligonucleotide microarray analysis or chromosome microarray. An oligo array allows more regions of the chromosomes to be tested than other array tests on the market, thereby providing a more comprehensive test.

- **What are reasons for using EmArray Cyto6000?**

Extra or missing chromosome material can cause developmental delay, mental retardation, autism spectrum disorders, birth defects or other symptoms of chromosome condition. A person with any of these features may benefit from this test to identify the cause of their delays. If extra or missing material is found, the results may help guide the patient's medical care. In addition, a small number of individuals who already have a known chromosome change may also benefit from this test. In such cases, this can be used to determine the size of a known duplication or deletion, or to test for hidden imbalances in a patient with an 'apparently' balanced translocation. This test is usually performed at the same time as, or following, standard chromosome testing.

- **How can my child/myself/my family members be tested?**

Testing should begin on the person in the family with the symptoms described above. A doctor or geneticist, a doctor who specializes in genetics, can examine an individual and order this test. Results of the test will be given to you by your doctor. Results typically take 1-2 weeks.

- **Are both parents required to give a blood sample?**

It is not necessary to send the parent samples until contacted by the laboratory. Occasionally, blood samples from both parents are requested to help determine the significance of a chromosome change. Without parent samples, it may be difficult to provide an accurate interpretation of the results. Emory Genetics Laboratory will contact the doctor who ordered the test to discuss this request.

Accuracy may also depend on the individual's clinical diagnosis and that reported family relationships are true biological relationships. Genetic testing in family members can sometimes reveal that true biological relationships are not consistent with the reported relationships.

- **What does it mean if they find a chromosome imbalance?**

Finding a chromosome imbalance, or change in the amount of genetic information present, may confirm the reason a person is experiencing symptoms such as developmental delay, mental retardation or other medical problems. If a chromosome change is found, testing for the specific change may be offered to family members.

- **What does it mean if they don't find a chromosome change? (What are the limitations?)**

Not finding a chromosome change does not mean that an individual does not have a genetic condition. Not finding a chromosome change may be because the person truly does not have a chromosome change or because the person carries a change that cannot be found using this test. Examples of genetic changes that cannot be found using this test include very, very small extra or missing chromosome regions, extra or missing single genes, or changes within a single gene. Balanced chromosome changes (when no genetic information is extra or missing) cannot be found using this test.

Though many genetic diseases are caused by extra or missing chromosome material, certain genetic conditions may be caused by other genetic changes. If your doctor suspects a specific condition caused by a single gene, testing other than the array may be considered. Members of Emory Genetics Laboratory are available to discuss these specific cases with your doctor.

- **What is a variant of unknown significance?**

This result means that a chromosome change has been identified, but it is difficult to interpret the meaning of the change. Sometimes there is little medical information about the particular change found. In these cases it may be uncertain whether the change may lead to future medical problems. Often testing the parents of the patient is suggested to see if either parent carries the change. If a parent carries the change, it may be considered a normal variant or change inherited in the family. If the parents do not carry the change, it may be a more likely cause of medical concerns in the person tested.

- **Billing for the EmArray Cyto6000**

The cost of this test is \$1610. With most offices, Emory Genetics Laboratory will bill the hospital or referring laboratory, which may then bill your insurance company. In some cases, Emory Genetics Laboratory cannot bill the referring hospital or laboratory. Therefore, if you choose to have this test performed you should speak with your doctor about insurance coverage and billing options. This test may not be considered standard of care by insurance companies or Medicaid.

The CPT codes are: 83890, 83892(x2), 83894, 83896(x10), 83897, 88384, 88385, 88386, 88230

- **Other considerations:**

Because of the complexity of this testing, some patients may benefit from speaking with a genetic counselor. Genetic counselors at Emory University are available for appointments by calling (404) 778-8500 or (800)366-1502. The National Society of Genetic Counselors provides a searchable address book to find a genetic counselor near you at: [www.nsgc.org](http://www.nsgc.org).