

## **Important Information about Array Comparative Genomic Hybridization (array CGH)**

### **What is array CGH?**

Array CGH technology allows for the detection of chromosome imbalances (missing or extra information) that are smaller than can be detected by standard chromosome analysis (karyotype).

Array CGH compares a patient's DNA with control / reference DNA to identify whether there are any imbalances in the patient's genetic information compared to the control DNA.

### **When should array CGH be considered?**

Array CGH may be considered for patients who have intellectual disability (ID) plus or minus congenital anomalies, autism or dysmorphic features that are unexplained after a thorough history and physical examination.

Patients who have features consistent with a known chromosome syndrome (such as Down syndrome) should have routine chromosome analysis as a first line test, NOT array CGH.

### **What are the benefits of array CGH?**

- Provides a detailed study of the chromosomes which may not have been previously possible
- May help understand the cause of the patient's health/developmental problems
- May help guide treatment and care for the patient
- May help gain appropriate services for the patient
- May help understand the family's chance to have another child with the same health/developmental problems

### **What are the limitations and unanticipated outcomes of array CGH?**

- May detect a chromosome imbalance that the laboratory is unable to interpret. These findings may be completely harmless or they may be related to the patient's health/developmental problems but current scientific knowledge is unable to clarify.
- May find an imbalance not related to the patient's current health/developmental problems but that indicates that the patient (and sometimes their family members) is at risk for additional health problems that were not anticipated (e.g., cancer, neurological disease).
- Not all genetic conditions are detectable by array CGH. Some are caused by mutations within a single gene or are multifactorial in nature.
- Cannot detect low levels of mosaicism, polyploidy or balanced chromosome rearrangements such as translocations or inversions which could have significant implications for the patient and their family.
- Not able to detect imbalances in areas of the genome that are not covered by the array CGH platform

### **What does my patient need to know before ordering an array CGH?**

Review the Pre-test Counselling Information (page 2 of the array CGH requisition) with your patient.

- Array CGH detects an imbalance in about 25% of patients (includes pathogenic anomalies and uncertain variants)
- A normal array CGH does not exclude all genetic causes of disease
- Array CGH may detect an imbalance that is not related to the patient's health/developmental problems but that predicts future health problems for the patient and / or their family members. These findings might have insurability implications, for example, a deletion or duplication of a gene (or a part of a gene) that may predispose to developing a disease later in life.
- Array CGH may detect an imbalance that the laboratory is unable to interpret at this time
- Array CGH testing may require testing of parents to help establish the clinical significance of a finding in the patient

### **How do I order array CGH?**

Due to the complexity of potential findings with the need for follow-up genetic counseling and family studies, ordering of array CGH is currently restricted to clinical geneticists.

However, after referring your patient for assessment by a clinical geneticist, if your patient has non-syndromic developmental delay/intellectual disability and/or autism, you may receive a letter requesting that you organize array CGH testing.

Array CGH can only be ordered by completing the array CGH requisition (available at [www.albertahealthservices.ca/3310.asp](http://www.albertahealthservices.ca/3310.asp) in the Cytogenetic Laboratory section). ***The clinical features and information provided is crucial to the interpretation of the results. Please be as detailed as possible in this section.***

Blood can be drawn at a community phlebotomy lab. **Please include the letter from the clinical geneticist requesting that you organize array CGH testing.** Please note that samples received with incomplete requisitions or without the letter from clinical genetics cannot be tested until all information is received.

### **What samples are needed for array CGH?**

A blood sample is required from the patient. Sometimes, samples from the parents or an additional sample from the patient is required and the laboratory will inform you if that is the case.

### **How long will results take?**

The interpretation of array CGH is complex. Results may take at least 3 months.

### **I have array CGH results. Now what?**

If your patient has an abnormal result, it is appropriate to refer them for genetic assessment and counselling. Please include the result with the referral.

If your patient's array CGH is normal, it does NOT mean that your patient does not have a genetic cause for their health problems. Other genetic testing may be appropriate, in consultation with Clinical Genetics.

### **How do I refer to clinical genetics?**

You may make a referral for clinical genetic assessment of your patient by contacting the R.B. Lowry Genetics Clinic at the Alberta Children's Hospital (phone 403-955-7373; fax 403-955-2701) or by contacting one of the private medical genetics specialists listed in the CMA directory.

### **I have questions about array CGH. Who do I talk to?**

Health care providers who have questions about CGH microarray can contact the laboratory genetic counselors at 403-955-3097.