Information on Whole genome array Comparative Genomic Hybridization (aCGH) Testing (Postnatal)

What are the objectives and benefits of aCGH testing?
- aCGH is a special test which finds differences in the amount of genetic material among individuals. It looks for areas of the chromosomes that have gain or loss of genetic material.
- The usual examination of chromosomes under the microscope is called karyotyping.
- aCGH test is much more sensitive than karyotyping. aCGH has the potential to find a chromosome gain or loss even if testing of the chromosomes by karyotyping shows normal results.
- These smaller imbalances, possible gains or losses of genetic material, are called ‘submicroscopic imbalances’ because they cannot be seen through the microscope. They may cause birth defects, delays in development, and genetic syndromes.

Who should consider having this test?
- Whole genome aCGH is the recommended test for individuals with developmental or congenital problems. The clinical geneticist will assess if the test is necessary for your child.

What type of samples is required for this test?
- 3 mL blood from the patient.

How is the test done?
- The DNA of the patient is matched with a control DNA sample on the array to detect gain or loss of genetic material in the patient.

When will the results be available?
- The test will be reported in 21 days.

How do I get to know the results?
- The test result will be reported to your doctor, who will explain the result to you.
- You and your family members may be referred to clinical geneticist for further counselling if there are abnormal or unclear findings.

What are the possible test results?
There are three possible test results:
(The interpretation of results is based on information available at the time of reporting.)
1. Normal – no clinically significant gain or loss of genetic material is detected in the chromosome.
2. Abnormal – gain or loss of genetic material is detected in the chromosome. The clinical features associated with these results depend on the specific genetic material that is gained or lost.
3. Unclear – gain or loss of genetic material is detected in the chromosome but the effect is uncertain. Performing aCGH test on the blood samples taken from both biological parents may help in interpretation.
What are the limitations of this test?

- aCGH test does not look at the structural arrangement of chromosomes.
  *these are supplemented by conventional chromosome study by karyotyping
- aCGH test does not look for individual gene changes.
- aCGH cannot detect low level mosaicism (presence of cells with different chromosomal makeup in an individual).
- A normal aCGH test result cannot exclude all abnormalities.

What are the other important considerations in choosing this test?

- It is possible that a diagnosis unrelated to the reason of testing may be found including predisposition to mental problems, autism, cancer, late-onset diseases or other medical conditions.
- There is a small chance of finding a genetic condition affecting the health of yourself or other family members.
- The test result may still be uncertain even after parental blood is tested.
- The above may impose psychological distress.
- You need to indicate what information you would like to know from the test.

I acknowledge that the above information has been discussed with me by the medical staff and I fully understand the information. I have been given the opportunities to ask questions pertinent to the test and satisfactory answers have been provided by medical staff.

Signature _____________________

Date ________________________