



## **PEDIATRIC POPULATION CHROMOSOMAL SNP MICROARRAY TEST**

Page 1 of 2

### **INFORMATION AND CONSENT FORM**

#### **WHY IS SINGLE NUCLEOTIDE POLYMORPHISM (SNP) CHROMOSOMAL MICROARRAY (CMA) PERFORMED?**

Each segment of DNA is usually found in two copies, one inherited from each parent, in all the cells of the human body. Many genetic diseases and syndromes are caused by variations in the number of copies of these DNA segments. Occasionally, the number of copies for a DNA segment is normal (two) but both copies are identical in nature (stretch of homozygosity). In rare instances, this may cause a disease. SNP CMA is used to determine whether the number of copies is lower (DNA loss) or higher (DNA gain) than normal or identical. To perform SNP CMA, a biological sample (blood) must be collected, from which DNA will be extracted and analyzed.

#### **WHAT TYPE OF RESULTS MAY BE OBTAINED WITH SNP CMA?**

- A. The result of this test may be normal. This means that no clinically significant DNA losses, DNA gains or stretches of homozygosity have been identified, according to the laboratory reporting criteria and current guidelines. A normal test does not exclude the possibility that your child's clinical findings are due to a genetic cause, not detectable by this test (see test limitations).
- B. The result of this test may be abnormal. This means that the test identified a loss and/or gain of DNA that most likely explains the clinical condition of your child.
- C. The result of this test may be of uncertain clinical significance, meaning that a DNA loss and/or DNA gain and/or long stretch of homozygosity has been identified, but that additional tests may be required to assess whether this variation is related to your child's condition. This may also mean that the current state of knowledge does not allow for a conclusion to be drawn (variants of unknown significance or VUS).

#### **SECONDARY/INCIDENTAL FINDINGS**

- A. CMA could identify DNA gains or losses that are not related to the clinical condition for which the test is requested, but that may predispose or be associated with a risk of developing a disease. DNA losses and DNA gains associated with conditions presenting in childhood/adolescence will be reported only if preventative treatment is available or surveillance is recommended. Incidental findings associated with adult onset conditions will not be reported.
- B. SNP CMA may reveal genomic homozygosity suggestive of close genetic relationship (identity by descent/consanguinity) between the biological parents of the patient.

#### **ADDITIONAL TESTING**

A follow-up testing by a complementary technique on your child and or her/his biological parents may be required. In this case, additional blood samples will be requested. Misinformation regarding the accurate biological relationships of the parents, such as non-paternity, can result in significant misinterpretation of findings or the impossibility to interpret results.

**CMA TEST LIMITATIONS**

- A. CMA cannot detect all genetic changes.
- B. CMA cannot detect balanced rearrangements (structural anomalies without loss or gain of genetic material).
- C. CMA cannot detect DNA losses and gains smaller than the technical resolution of the microchip used.
- D. CMA may not detect DNA gains or losses present in only a minority of cells (low level mosaicism).

**DNA SAMPLE RETENTION AFTER TESTING (choose one of two options)**

Your child's DNA sample will be kept for a minimum period of six months in the event that further analysis is required. After that period, do you accept that your child's DNA sample is anonymized and included in a local DNA sample bank for clinical test development or validation?  YES  NO

**I understand that I am free to give my consent and that a refusal on my part will not deprive me of health or social services required by the state of health of my child.**

**CONSENT**

I, the undersigned, \_\_\_\_\_, consent to the analysis of the genetic material of my child, \_\_\_\_\_, by chromosomal microarray (CMA) with SNP.

I acknowledge that I have read and understood the information presented in this consent form and that I have obtained, where applicable, all explanations necessary for its understanding. This consent has been given in a free and informed manner, without undue coercion or pressure, and I acknowledge that I have received all the information necessary to be able to consent to the acts to be performed.

\_\_\_\_\_  
Patient's parent /legal guardian signature

\_\_\_\_\_  
Date (YYYY-MM-DD)

***I have explained the proposed SNP CMA analysis to the person who has consented to the test and I have provided answers to her/his questions.***

\_\_\_\_\_  
Name (in print letters)  
Physician/Resident/  
Genetic counsellor

\_\_\_\_\_  
Signature  
Physician/Resident/  
Genetic counsellor

\_\_\_\_\_  
Date (YYYY-MM-DD)

\_\_\_\_\_  
Physician's/ Resident's  
licence N#