What will these results tell me?

A Negative Result

If no unusual findings are detected (a negative result), it does not mean that the health and/or learning problem(s) you or your child are having are not genetic. A negative result would simply indicate that there are no detectable gains or losses of genetic information and no absence of heterozygosity. A patient with a negative result may still need to be followed by a specialist.

A Positive Result

Any gains/losses of genetic information or an absence of heterozygosity (a positive result) will be reported to you. For some individuals, the findings of the microarray will be known to have health and learning effects. These effects will be discussed with you.

(cont.)

For others, the possible effect(s) of the genetic findings may not be completely known. Depending on the finding that is detected, further testing may be indicated. For example, it may be necessary to examine the parents’ DNA to determine if the unusual finding has been inherited.
What are DNA, genes and chromosomes?

DNA is the smallest component of our genetic material. It is found in almost all the cells of our bodies and is made up of a string of letters (A,T,G and C). These letters, when grouped together in a particular way, form our genes. Genes are the instruction manuals for how our bodies grow, develop and are maintained. We receive half of our genes from each parent. Genes are responsible for making proteins that the body needs for proper growth, development and maintenance. Genes make up larger structures in the cells called “chromosomes.” Typically, an individual will have 46 chromosomes in each cell. These chromosomes are found in pairs. We expect each individual to receive 23 chromosomes from each parent. Changes within a gene or a chromosome may result in health and/or learning problems for a particular individual.

Why would my doctor order this test?

Your doctor may order the cytogenomic microarray testing to determine if there is a detectable change in any chromosome(s) that affects the health and/or learning for yourself or your child. To run this test, a small sample of blood is required. DNA will be taken from the blood sample for the test to be completed. Your doctor will call you when the results are completed.

What is a cytogenomic microarray?

The cytogenomic microarray testing uses advanced technology that makes it possible to examine chromosomes in greater detail than is seen under the microscope with traditional chromosome analysis. This testing allows for the detection of small gains or losses of genetic material as well as unusually large areas that are genetically identical (called “absence of heterozygosity”). These changes to the chromosomes can cause health and/or learning problems for an individual.