DDC clinic
Center for Special Needs Children

MOLECULAR
DIAGNOSTICS
LABORATORY

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INFORMED CONSENT FOR GENETIC TESTING

Genetic testing will look for changes in the DNA, genes, or chromosomes which may be associated with a specific genetic condition. A positive test result is an indication that the individual may be predisposed to or have the specific disease or condition for which we tested. Knowledge of a specific gene change may result in a change in the treatment plan that is currently underway or in medical surveillance for potential future health problems. The changes to care may include increased health screening for a known but healthy carrier of a gene change, a switch in the type of medication used for an affected individual or even a change in the anesthesia options. The possibilities cannot be listed without knowledge of the test result. We will discuss these possible changes once we have a confirmed diagnosis or known gene change.

There are several types of genetic testing. **Targeted Mutation Analysis** is performed when a specific disorder is suspected and one gene change is responsible for the vast majority if not all of the affected individuals. Targeted mutation analysis is also performed when the specific gene change has been identified for a family. **Whole Gene Sequencing** is performed when a specific disorder is suspected, but there is no single gene change associated with the disorder. **Next-Generation Sequencing (NGS)** is performed when there are multiple genes associated with the same or very similar disorders. Sequencing of all of these genes at once is currently cheaper and faster than sequencing each gene individually. Lastly, **Chromosomal Microarray Analysis** is performed when the pattern of medical problems is suggestive of a genetic condition but a single gene disorder is not recognized. The chromosomal microarray is a way for the laboratory to look at the entire genetic make-up of a person in order to find missing (loss) or extra (gain) pieces of the chromosomes. Many of the losses and gains found by microarray are common and have a well understood pattern of medical problems.

As with any test, it is always possible that results may not be obtained and a repeat sample may be requested. With genetic testing, parental samples or samples from other family members may be requested to further understand the findings. When parental testing is performed, it may show changes related to those found in the individual originally tested (i.e. child). In those cases, this information regarding parental test results would become part of the child's test report. Thus it would appear in the child's medical record. Even with or without family testing, it is possible that a test result cannot be explained given the knowledge and literature available at this time.

The genetic test _____ has been explained to me.
I understand the purpose of the testing as well as the limitations and consent to the testing for me/my child.

Patient Name _____ Date of birth _____

Patient or Parent Signature _____ Date _____

(Parent name printed) _____