**Cytogenetics Laboratory 275-5859**

### Constitutional Cytogenetic Analysis

**SPECIMEN TYPE**
- Amniotic Fluid
- CVS
- Sterile Container/Media/Rm Temp
- Date of Ultrasound: / / GA on Date of Ultrasound: weeks days
- Peripheral Blood (SMH)
- Lavender/Green Top/Rm Temp (2-5 mL)
- Skin Biopsy
- Sterile Container/Media/Rm Temp
- POC
- Sterile Container/Media/Refrig.

**INDICATION(S) FOR TEST**
- Abnormal Maternal Serum/First Trimester Screen - specify
- Abnormal Ultrasound
- Autism
- Biochemical Testing - specify:
- Congenital Anomalies - specify:
- Developmental Delay - MR
- Dysmorphic Features
- Family History of Chromosome Abnormality - specify
- Failure to Thrive
- History of SAB
- Maternal Age
- Other - specify:

**TEST(S) ** Patient Consent required
- Chromosome G-Banding Analysis
- Mosaicism-Specify
- High Resolution Banding
- Polymorphism Study
- FISH for X, Y, 13, 18 and 21
- Sub-Telomere Analysis
- Other FISH - Specify
- Fibroblast Only
- Chromosome Micro-Array CGH

### Hematology/Oncology Cytogenetic Analysis

**SPECIMEN TYPE**
- Bone Marrow Aspirate - Green Tube Rm. Temp (1 mL)
- Peripheral Blood - Green Tube Rm. Temp (2-5 mL)
- Lymph Node - Sterile Container/Media/Rm Temp
- Solid Tumor - Sterile Container/Media/Rm Temp
- Specify:

**CLINICAL INFORMATIO** (diagnosis under consideration)

### Test(S)
- Chromosome G-Banding Analysis
- FISH-specify
- Chromosome Micro-Array CGH

### Bone Marrow Transplant
- Autologous
- Allogeneic
- Sex Mismatch
- Chromosome G-Banding Analysis
- FISH (X/Y centromere)
- FISH other - specify:

### Patient Consent

I have read the information on the back of this form and discussed it with my health care provider. I have been given the opportunity to ask questions and have them answered about the tests ordered. I authorize collection and analysis of the necessary sample(s).

Patient/Legal Guardian: ______________________ Date: __________ Health Care Provider: ______________________
Important Information about Genetic Testing

1. This test will look for changes in the DNA chromosomes, genes, or gene products which are known to be associated with the specific genetic condition in question.

2. This test may reveal that the individual tested is affected with the condition, carries the genetic pre-disposition for it, or that he/she does not. If a positive result is obtained, a medical and/or genetic counseling follow-up may be advised.

3. Genetic testing is ordinarily highly accurate, however, in some cases results may not be obtained or may be inconclusive. Also, accurate genetic testing depends upon an accurate diagnosis in affected family members. If the diagnosis in a family is not certain, results can be misleading. I have been able to discuss the expected accuracy of the testing in my particular case. Initial: ________

4. Some genetic testing may require comparison of samples from multiple family members with their consent, and in these cases, previous unknown non-paternity can be discovered.

5. Some genetic tests are only done by a few laboratories in the world. This sample may need to be sent out of state to laboratories that are not certified by the New York State Health Department. In these cases, approval for testing will be obtained from New York State.

6. Some types of genetic testing such as fluorescence in situ hybridization (FISH) are considered investigational by the New York State Health Department. Using DNA probes which bind to specific regions of the chromosomes, FISH is helpful in identifying the origin of unidentified "marker" chromosomes, unusual variations in chromosome structure or small chromosomal deletions which cannot be seen by standard chromosome testing. FISH may be used, if indicated on my sample. Initial: ________

7. Chromosome microarray CGH (Array CGH) test is considered to be investigational by the New York State Department of Health. Array CGH is helpful in detecting gains or losses of chromosomal material at the DNA level anywhere in the genome. The method uses cloned DNA probes 'on a chip' to detect deletions or duplications which cannot be seen by standard chromosome and FISH analysis. The purpose of this assay is to detect syndromic microdeletions/duplications and subtelomeric alterations. Copy number alterations of single loci may be false negative due to the limited resolution of the array CGH. Because array CGH is a new technology being used in clinical diagnosis, all abnormal array CGH findings will be confirmed by standard chromosome or FISH analysis. Parental studies and additional assays may be necessary to characterize and interpret the clinical significance of array CGH results. The array CGH is limited to the detection of copy number changes in the genome. It will not detect balanced translocations, inversions, low level mosaicism, point mutations and genomic regions not represented on the array.

Array CGH is used as an adjunct to chromosome analysis and all patients are required to have a chromosome analysis along with array CGH testing.

The patient or their legal counsel is required to sign an informed consent before the array CGH is performed. Due to the complexity of array CGH, the results will be reported directly to the ordering provider. Initial: ________

8. Records of this testing or test results will not be released to anyone other than me, my referring doctors and Strong Memorial Medical Records unless I specify otherwise. Initial: ________

9. No tests other than those authorized shall be performed on the biological sample and that the sample shall be destroyed at the end of the testing process or not more than sixty days after the sample was taken. Any part of the biological sample not used for specific genetic testing may be retained and used for medical research as long as names and other identifying information are not revealed. Initial: ________

10. I indicate my desire to opt out of participation in anonymized research studies using my DNA sample by checking this box: ☐