

**Constitutional Cytogenetics – Specimen Requirements**

For questions please call:

**North Shore University Hospital Cytogenetics Laboratory** @ **516-562-3863** for prenatal/products of conception/skin biopsy specimens

**Long Island Jewish Medical Center Cytogenetics Laboratory @ 718-470-7065** for

peripheral blood specimens

**Transport Requirements**

Send to the laboratory ASAP at room temperature 20-25˚C.

If there is a delay in transportation, store the sample in a refrigerator at 4˚C- **DO NOT FREEZE**

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| **Test** | | **Specimen type** | **Collection/recommended volumes** | **Average turn around time** |
| **Chromosome Analysis (Karyotype)** | | Chorionic Villi | 5-10 mg in sterile tube with sterile saline *or* transport media | 7-14 days |
| Amniotic Fluid | 20 ml in two sterile conical tubes | 7-14 days |
| Products of conception /  skin biopsy | 5-10 mg in sterile tube with sterile saline ortransport media | 14-28 days |
| Peripheral Blood | Adult: 5-10 ml  Newborn: 2-5 ml  **dark green/sodium heparin tube** | 10-14 days |
| **Fluorescence *in situ* hybridization**  **(FISH)** | | Peripheral Blood | 5-10 ml in **dark green/sodium heparin tube** in conjunction with chromosome analysis | Microdeletion single probe:  7-10 days  Rapid Newborn Aneuploidy:  1-2 days |
| Chorionic Villi | 3-5 mg **in addition** to required amount for chromosomes | Aneuploidy FISH: 1-2 days |
| Amniotic Fluid | 5 ml **in addition** to required amount for chromosomes |
| **Chromosomal**  **Microarray (performed at GeneDx)** | | Peripheral Blood\* | 2-5 ml in **EDTA lavender top** tubes | 2-3 weeks |
| Chorionic\*\* Villi/POC | 10 mg **in addition** to required amount for chromosomes | ~2 weeks on direct specimen  \*Add ~1-2 weeks if direct was not possible and cultures are prepared |
| Amniotic Fluid\*\* | 10-15 ml **in addition** to required amount for chromosomes |
| **Molecular Tests**  **(CF, Fragile X, etc)** | | Peripheral Blood\* | 5-10 ml in **EDTA lavender top** tubes | ~14 days |
| Chorionic Villi/POC\*\* | 5-10 mg **in addition** to required amount for chromosomes | ~14 days on direct specimen  \*Add ~1-2 weeks if direct was not possible and cultures are prepared |
| Amniotic Fluid\*\* | 10-15 ml **in addition** to required amount for chromosomes |
| **\*For peripheral blood:** submit both maternal and paternal bloods (2-5 ml/ **EDTA lavender top** tubes) if needed for result interpretation  **\*\*For prenatals**: submit maternal blood (2-5 ml/ **EDTA lavender top** tube) for maternal cell contamination studies and paternal bloods (2-5 ml/ **EDTA lavender top** tubes) if needed for result interpretation | | | | |

All test results are relayed to the referring physician within the above specified turnaround times.  In some instances, additional time is needed to allow for confirmatory, reflex testing or extensive work-up for a complete diagnosis.  Please note that the above TAT is an internal metric determined by the laboratory.  We adhere to CAP recommendations on TAT for all specimens.