

☒NEW TEST ☒TEST CHANGE

NOTIFICATION DATE: 05/08/2025

EFFECTIVE DATE: 05/12/2025

Chromosome Analysis – Constitutional Case

On the effective date Northwell Health Laboratories cytogenetics lab will introduce five separate orderables for this test based on specimen type. This change was implemented to assist with the correct ordering of this test and to reduce ordering errors due to incorrect specimen selection. All test orders must be accompanied by documented informed consent.

Test Requirements	New	New	New	New	New	Current
Test Name	Chromosome Analysis Constitutional Peripheral Blood	Chromosome Analysis Chorionic Villus Sampling (CVS)	Chromosome Analysis Amniotic Fluid	Chromosome Analysis Products of Conception (Reflex Microarray)*	Chromosome Analysis Skin Biopsy	Chromosome Analysis- Constitutional case
Specimen Requirements	Peripheral blood, cord blood	Chorionic Villi	Amniotic Fluid	Placental villi, Umbilical cord, fetal skin	Skin punch core biopsy	Peripheral blood, Chorionic Villi, Amniotic Fluid, POC, Tissue
Submission Container/ Tube:	Green-top (sodium heparin) tube(s) Clotted sample is not acceptable, but an attempt will be made.	Sterile container	Sterile container	Sterile container	Sterile container	Sterile container (prenatal, POC, and tissue specimens); Green-top (sodium heparin) tube(s)
Specimen Volume/ Minimum Volume:	Adult 5-10 mL, Newborn 2-5mL in sodium heparin dark green tube. Invert the tube several times to prevent coagulation	5 - 10 mg Chorionic Villi in sterile saline or transport media	20 mL in two sterile conical tubes	5-10 mg placental villi or 1 cm umbilical cord in sterile tube with sterile saline or transport media (RPMI). *Please contact the cytogenetics lab to order RPMI: NSUHCulturedCellRequest@northwell.edu	Minimum 0.5cm2 (1.0cm2 is preferred) in sterile tube with sterile saline or transport media	Varies by specimen



If you have any questions, please contact the Cytogenetics Department at (516) 562-4359 or 2888

Collection Instructions:	Send to LIJ Cytogenetic 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 NOTE: Pertinent medical information including suspected diagnosis is required	Send to NSUH Cytogenetic 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 NOTE: Pertinent medical information including suspected diagnosis is required	Send to NSUH Cytogenetic 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 NOTE: Pertinent medical information including suspected diagnosis is required	Send to NSUH Cytogenetic 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 NOTE: Pertinent medical information including suspected diagnosis is required	Send to NSUH Cytogenetic 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 NOTE: Pertinent medical information including suspected diagnosis is required Avoid collecting specimens on Fridays.	Send to NSUH cytogenetics 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 NOTE: Pertinent medical information including suspected diagnosis is required
Specimen Stability:	48 hours room temperature If specimen cannot be sent promptly, refrigerate and send ASAP DO NOT FREEZE	48 hours room temperature If specimen cannot be sent promptly, refrigerate and send ASAP DO NOT FREEZE	48 hours room temperature If specimen cannot be sent promptly, refrigerate and send ASAP DO NOT FREEZE	48 hours room temperature If specimen cannot be sent promptly, refrigerate and send ASAP DO NOT FREEZE	48 hours room temperature If specimen cannot be sent promptly, refrigerate and send ASAP DO NOT FREEZE	48 hours room temperature If specimen cannot be sent promptly, refrigerate and send ASAP DO NOT FREEZE
Computer Interface Code:	PDM# 248489	PDM# 248490	PDM# 248491	PDM# 248492	PDM# 248493	PDM# 5160090
Test Order:	CG CONST PB	CG CONST CVS	CG CONST AF	CG CONST POC	CG CONST SB	Chromosome Analysis – Constitutional Case



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