

Cytogenetics

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HOURS: M-F: 7 am – 11 pm
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The Allina Medical Laboratories Cytogenetics laboratory is located on the Abbott Northwestern Hospital campus. Our professional staff offers the latest technology available for physicians and patients at a competitive price. We offer a comprehensive testing service including both chromosome and Fluorescence in Situ Hybridization (FISH) analysis for congenital and oncology specimens. We are committed to research and development to ensure the latest technology is available to our customers.

The AML Cytogenetics laboratory provides accurate high quality results with excellent turn-around-times and exceptional customer service including genetic counseling upon request.

For service after hours, leave a voicemail message at 612-863-4541. We will return your call the next working day.

Specimen Collection and Transport Instructions

- Specimen requirements for Cytogenetics testing at AML can be found on our website at: http://www.allina.com/ahs/allinalabs.nsf/page/cytogenetics_home2 or from the AML home page found at www.allina.com/medicallaboratories, click on the *Cytogenetics Laboratory* link located under *Featured Services*.
- Completely fill out a Cytogenetics test requisition including:
 - Patient demographics
 - Billing/Insurance information
 - Genetic indication for testing
 - Test/s requested
 - Answer all questions within each testing section.
- Place the sample in the sealed pocket of a Cytogenetics specimen transport bag, and the original copy of the completed requisition in the outside pouch
- Transport the specimen at room temperature to ANW with your regularly scheduled courier to AML.
- If you do not have a regularly scheduled courier pickup, call Cytogenetics at 612-863-4541 to inform the lab and to initiate a courier pickup. The cytogenetic lab provides transportation of the sample to our lab via an internal or commercial courier.

Congenital Studies

The following section contains congenital testing information regarding chromosome and FISH studies, and send out testing.

CONGENITAL CHROMOSOME STUDIES:

LAB TEST CODE /NAME	SPECIMEN TYPE	REQUIREMENTS	APPROXIMATE TAT
C30 + C31 <i>or</i> C32/ Amnio Chromosome Study	Amniotic Fluid	20-30 cc in sterile tissue culture tube at room temp.	7-14 days
C37/ CVS Chromosome Study	Chorionic Villi	10-30 mg in Hanks Balanced Salt Solution or sterile saline at room temperature.	6-13 days
C34 & C35/ POC/ SKIN / AUTOPSY Chromosome Study	Products of Conception/ Skin Biopsy/ Autopsy	3-5mg abortus tissue (skin and chorionic villi preferred) or punch biopsy in sterile saline at room temperature.	6-21 days
C40/ Blood Chromosome Study	Peripheral Blood or Cord Blood	3-7 cc sodium heparin whole blood sterile and room temperature.	Neonatal STAT: 2 day oral prelim, 7 day final Routine: 7-21 days
C43/ High Resolution Blood Chromosome Study	Peripheral Blood	3-7 cc sodium heparin whole blood sterile and room temperature.	7-21 days
C42/ PUBS Chromosome Study	Percutaneous Umbilical Blood Sampling	3-7 cc sodium heparin whole blood sterile and room temperature.	STAT: 2 day oral prelim, 7 day final

NOTE: For each study described below, additional staining techniques and metaphases studied may be added and charged as deemed necessary by a Cytogenetics Director (see Director Reflex Testing section).

Amniotic Fluid Chromosome Analysis: For all amniotic fluid samples, an aliquot of supernatant is saved for 6 weeks from receipt of specimen. An In situ culture method is performed whenever possible. A minimum of fifteen G-banded metaphases are studied with two karyograms prepared per cell line.

Chorionic Villi Sampling Chromosome Analysis: An In situ culture method is performed whenever possible. A minimum of twenty G-banded metaphases are studied with two karyograms prepared per cell line.

Products of Conception/Autopsy Chromosome Analysis: Several tissues should be sent to ensure the best culture success. Chorionic villi and internal tissue is preferred over skin samples. An In situ culture method is performed whenever possible. A minimum of twenty G-banded metaphases are studied with two karyograms prepared per cell line.

Skin Biopsy Chromosome Analysis: It is important to get a deep enough sample to attain viable cells for culture. A 3mm³ sample is preferred. An In situ culture method is performed whenever possible. A minimum of twenty G-banded metaphases are studied with two karyograms prepared per cell line.

Peripheral Blood Standard Chromosome Analysis: Two cultures are set up with each sample. A minimum of twenty G-banded metaphases are studied with two karyograms prepared per cell line.

STAT Neonatal Blood Reporting:

A backup culture is set up with each sample. We provide a STAT service on these samples upon request. This service includes a 48-hour preliminary oral report as well as a 7-day final written report. The preliminary report is based on a six-cell screen and one minimum banded karyogram. The STAT preliminary report should be used only when gross chromosome changes are suspected.

Peripheral Blood High Resolution Chromosome Analysis: A backup culture is set up with each sample. A minimum of twenty G-banded metaphases are analyzed with three karyograms prepared per cell line. A high-resolution study consists of clearing banding of all chromosome pairs ≥ 700 band length. If analysis fails to clear all bands at ≥ 700 bands, the test will be changed to a peripheral blood standard chromosome analysis.

PUBS Analysis: A backup culture is set up with each sample. Twenty G-banded metaphases are studied with a minimum of two karyograms prepared per cell line.

CONGENITAL FLUORESCENCE IN SITU HYBRIDIZATION (FISH) STUDIES:

TEST CODE	TEST NAME	TEST DESCRIPTION
C64	Aneuploidy probe study (X, Y, 13, 18, 21)	Prenatal or congenital blood specimen study to determine aneuploidy of X, Y, 13, 18 or 21
C09	Aneuploidy probe study (13, 21 only)	Prenatal or congenital blood specimen study to determine aneuploidy of 13 or 21.
C10	Aneuploidy probe study (X, Y, 18 only)	Prenatal or congenital blood specimen study to determine aneuploidy of X, Y, and 18.
C88	Digeorge/VCFS study	Microdeletion 22q11.2 probe metaphase and interphase study
C89	Prader-Willi study	Microdeletion 15q11-13 probe metaphase and interphase study
C95	Angelman study	Microdeletion 15q11-13 probe metaphase and interphase study
C96	Cri du Chat study	Microdeletion 5p15.2 probe metaphase and interphase study
C97	Wolf Hirschhorn study	Microdeletion 4p16.3 probe metaphase and interphase study
C98	Williams ELN study	Microdeletion 7q11.23 probe metaphase and interphase study
C99	Smith-Magenis study	Microdeletion 17p11.2 probe metaphase and interphase study
C00	Miller-Dieker study	Microdeletion 17p13.3 probe metaphase and interphase study
C01	SRY deletion study	Microdeletion Yp11.3 probe metaphase and interphase study
C02	Kallman deletion study	Microdeletion Xp22.3 probe metaphase and interphase study
C03	STS deletion study	Microdeletion Xp22.3 probe metaphase and interphase study
C04	Individual sub-telomere study	Single telomere probe metaphase study
C05	All sub-telomere panel	All sub-telomere probes metaphase study
C06	MFISH study	Multi-color paint metaphase study
C07	Individual paint study	Single paint probe metaphase study
C11	Individual centromere study	Single centromere probe metaphase and interphase study
C61	Probe metaphase congenital study	Rare or research congenital metaphase probe studies
C62	Probe interphase congenital study	Rare or research congenital interphase probe studies
C63	Probe extensive interphase study	Rare or research probe study on extended cell populations

Congenital Fluorescent In Situ Hybridization (FISH): FISH is performed using a variety of commercially available probes: individual centromere/ all centromere, individual chromosome paint/ all-chromosome multi-colored paint (MFISH), individual sub-telomere/ all chromosome sub-telomere, along with a multitude of locus specific microdeletion/microduplication probes. FISH is performed on metaphases as well as interphase nuclei. FISH can usually be performed from a chromosome study culture however an additional culture may be deemed necessary by the cytogenetic director (see Director Reflex Testing section) to complete a study.

The AML Cytogenetics lab utilizes FDA approved protocols when available and uses standardized protocols established in the AML Cytogenetics lab when FDA protocols are not available. Controls are incorporated for all probes used and may include: internal control probes, positive / negative interphase control slides run in parallel, and normal metaphase control slides run in parallel. Normal ranges for interphase analysis have been established when appropriate.

Sample requirements are the same for FISH as those previously described for chromosome studies. We recommend using FISH in conjunction with a chromosome study for best results. To serve you and your patients better, we provide STAT FISH service 7 days a week upon request. A STAT FISH result can be expected within 24 hours for all interphase studies and within 4 days for cultured metaphase blood studies. All other metaphase FISH result TATs are dependent on the chromosome analysis result.

CONGENITAL SENDOUT TESTING:

TEST CODE/ NAME	SPECIMEN TYPE	REQUIREMENTS
C81/ Amnio/CVS send out testing culture - HOLD	Amnio/ CVS	Same as Amnio or CVS studies above. Attach send out lab requirements
C82/ Amnio/CVS send out testing culture -SEND	Amnio/ CVS	Same as Amnio or CVS studies above. Attach send out lab requirements
C83/POC/SKIN/AUTOPSY send out testing culture	POC/ SKIN/ AUTOPSY	Same as POC/ SKIN/ AUTOPSY study above. Attach send out lab requirements
BAA/ Amniotic Alpha-fetoprotein (AFP)	Amniotic Fluid	Amniotic fluid 3 cc supernatant (from chromosome specimen)
ACB/ Amniotic Acetylcholinesterase	Amniotic Fluid	Amniotic fluid 3 cc supernatant (from chromosome specimen)
MSO/ Miscellaneous Send Out	Test dependent	Test dependent
FXM/ Fragile X	Peripheral Blood	15 cc EDTA whole blood sterile and room temp.

Send out cultures for molecular or biochemical testing: The requesting institution is responsible for attaining current molecular or biochemical testing requirements. If the institution requesting send out testing has requested a “hold” on the sample, the requesting institution will be faxed when the specimen has met the culture requirements. If no communication back to the lab occurs within 3 working days from the original fax, the cultures will be cryopreserved (see director reflex testing section).

Alpha-fetoprotein and Acetylcholinesterase Testing: These tests are performed at another reference lab. The cytogenetics lab will prepare the sample and send it to the reference lab. All abnormal results are called directly to the requesting institution from the testing lab.

Peripheral Blood Fragile X Analysis: The Allina cytogenetics lab recommends a Fragile X molecular study be performed in conjunction with a chromosome analysis. The chromosome analysis will be performed by the Allina cytogenetics lab and the molecular Fragile X testing will be sent to another reference lab. The requesting institution is responsible for attaining current testing requirements.

Oncology Studies

The following section contains oncology testing information regarding chromosome and FISH studies.

ONCOLOGY CHROMOSOME STUDIES:

TEST CODE/ NAME	SPECIMEN TYPE	REQUIREMENTS	APPROXIMATE TAT
C51 & C58 Bone Marrow Chromosome Study	Bone Marrow	2-5 cc bone marrow in sodium heparin, sterile and room temp.	1-9 days
C46 & C47 Leukemic blood chromosome study	Leukemic Blood	5-10 cc sodium heparinized whole blood sterile and room temp	1-9 days
C71 & C80 Lymph node chromosome study	Lymph Node	3 mm ³ involved tissue in sterile culture medium or saline.	1-9 days
C71 & C80 Malignant tissue chromosome study	Malignant Tissue	5 mm ³ involved tissue (solid tumor or other) in sterile culture medium or saline.	1-9 days

NOTE: For each study described below, additional staining techniques and metaphases studied may be added and charged as deemed necessary by a cytogenetic director (see director reflex testing section).

A STAT TAT service is provided for acute leukemia specimens.

Bone Marrow Chromosome Analysis: A minimum of twenty G-banded metaphases studied with two karyograms prepared per clonal cell-line.

Note: If patient's indication for study is Chronic Lymphocytic Leukemia (CLL), a leukemic blood is the specimen of choice.

Leukemia Blood Chromosome Analysis: A minimum of twenty G-banded metaphases studied with two karyograms prepared per cell-line.

Note: This test is indicated for patients with Chronic Lymphocytic Leukemia (CLL) or Acute Leukemia.

Lymph Node Chromosome Analysis: A minimum of twenty G-banded metaphases studied with two karyograms prepared per clonal cell-line.

Malignant Tissue Chromosome Analysis: A minimum of twenty G-banded metaphases studied with two karyograms prepared per cell-line. Please indicate tissue type.

ONCOLOGY FLUORESCENCE IN SITU HYBRIDIZATION (FISH) STUDIES:

TEST CODE	TEST NAME	DESCRIPTION
C12	BCR/ABL t(9;22) study	Rearrangement interphase study involving BCR (9q34) and ABL (22q11.2)
C13	PML/RARa t(15;17) study	Rearrangement interphase study involving PML (15q22) and RARa (17q21.1)
C14	TEL/AML1 t(12;21) study	Rearrangement interphase study involving TEL (12p13) and AML1 (21q22)
C16	AML1/ETO t(8;21) study	Rearrangement interphase study involving AML1 (21q22) and ETO (8q22)
C17	MLL study t(11;?) study	Rearrangement interphase study involving MLL (11q23)
C18	CBFb inv(16) or t(16;16) study	Rearrangement interphase study involving CBFb (16q22)
C20	IGH/CCND1 t(11;14) study	Rearrangement interphase study involving IGH (14q32.3) and CCND1 (11q13)
C21	IGH/BCL2 t(14;18) study	Rearrangement interphase study involving IGH (14q32.3) and BCL2 (18q22)
C22	IGH/MYC t(8;14) study	Rearrangement interphase study involving IGH (14q32.3) and MYC (8q24)
C23	BCL6 t(3;?) study	Rearrangement interphase study involving BCL6 (3q27)
C24	ALK t(2;5) study	Rearrangement interphase study involving ALK (2p23)
C25	MALT t(18;?) study	Rearrangement interphase study involving MALT (18q21)
C26	Trisomy 8 study	Detection of trisomy 8 interphase study
C28	Trisomy 12 study	Detection of trisomy 12 interphase study
C29	5q31-/-5 study	Deletion of 5q31 or aneuploidy of chromosome 5 interphase study
C38	7q31-/-7 study	Deletion of 7q31 or aneuploidy of chromosome 7 interphase study
C39	13q14 deletion study	Deletion of 13q14 interphase study
C44	20q12 deletion study	Deletion of 20q12 interphase study
C45	P53 deletion study	Deletion of 17p13.1 interphase study
HFISH	Her-2/neu amplification study	Detection of 17q11.2-q12 amplification interphase in paraffin embedded tissues
UFISH	Bladder cancer aneuploidy detection study	Detection of chromosomes 3, 7, 17 and 9p21 status in urine samples
C59	Oligodendroglioma deletion study	Detection of deletions of 1p36 and 19q13 in neurologic tissues
C08	General morphometric study	Oncology FISH paraffin interphase study
C70	Probe oncology study	Rare or research oncology probe study
C63	Probe extensive interphase study	Rare or research probe study on extended cell populations or paraffin tissue
C56	BM/LB FISH culture	Culture for bone marrow or leukemic blood study
C57	Malignant tissue/lymph node FISH culture	Culture for malignant tissue/lymph node study
TBD	CLL Panel	Probes for detection of +12, 11q22.3-, 13q14.3-, 17p13.1- and t(11;14). Note: Recommended if chromosome results are normal or not available.
TBD	MDS Panel	Probes for the detection of 5q31-, -7, +8, t(11q23;?), 20q12-. Note: recommended only if chromosome results are not available.

Oncology Fluorescent In Situ Hybridization (FISH): FISH has several applications in oncology studies such as: specific leukemia aneuploidy changes (+12 in CLL, +8 in MDS) specific leukemia deletion detection (20q- in MDS, 16q- in AML M4eo), specific leukemia translocation detection [t(9;22) in CML, t(15;17) in APL], as well as specific solid tissue tumors amplification detection of gene regions (HER-2/neu amplification in breast cancer). We perform FISH using commercially available probes: individual centromere , individual chromosome paint and all-chromosome multicolored paint (MFISH), along with a multitude of locus specific probes specific for structural rearrangements of malignancies (t(9;22) in CML).

We utilize FDA approved protocols when available and use standardized protocols established in the Allina cytogenetics lab when FDA protocols are not available. We incorporate controls for all probe sets used and include: internal control probes as well as positive / negative interphase control slides, and normal metaphase control slides. We have established normal ranges for interphase analysis when appropriate.

Sample requirements are the same as those previously described for chromosome studies. To serve you and your patients better, we provide STAT FISH service 7 days a week. A STAT FISH result can be expected within 24 hours for all PML/RARA studies or other studies upon request. We call STAT FISH results on weekends and evenings.

Director Reflex Testing

The following section contains reflex testing information regarding chromosome and FISH studies and may be performed at the discretion of a Cytogenetics Director to clarify results.

TEST CODE/ NAME	MISCELLANEOUS STUDIES	TEST DESCRIPTION
C84	Thaw frozen culture	Thaw cells for re-culturing.
C85	Cryopreserve culture	Freeze cells for future culturing.
C86	Additional Cells analyzed	One charge for each additional 20 cells analyzed by chromosome analysis beyond a standard study.
C87	Individual Special Stains	AgNOR, C-Band, DA/DAPI, Q-Band or other stains utilized. One charge per stain utilized.
C33 Limited Amnio / CVS Chromosome Analysis	Amniotic Fluid	Ordered by Cytogenetics Director only in place of a full study.
C36 Limited POC/ SKIN/ AUTOPSY Chromosome Analysis	Products of Conception/ Skin Biopsy/ Autopsy	Ordered by Cytogenetics Director only in place of a full study.
C41 Limited Blood Chromosome Analysis	Peripheral Blood	Ordered by Cytogenetic Director only in place of a full study.
C48 Limited bone marrow/ leukemic blood study	Bone marrow or leukemic blood	Ordered by Cytogenetic Director only in place of a full study.
C72 Limited malig tiss chromosome study	Malignant Tissue or Lymph Node	Ordered by Cytogenetic Director only in place of a full study.
C49	Amnio/CVS FISH culture	Culture for amnio/CVS FISH study
C54	POC/ SKIN/ AUTOPSY FISH culture	Culture for POC/SKIN/autopsy FISH study
C55	Blood FISH culture	Culture for blood FISH study

Freeze and thaw cultures: Used for prenatal specimens with send out testing requested. All samples sent for external testing will have a backup sample frozen. For specimens with “hold for send out testing” requested, the lab will alert the requesting institution when cultures are ready to send. If there is no response within 3 working days, the lab will automatically freeze the entire sample for storage. *Note: success in re-culturing frozen cells is not guaranteed.*

Additional cells analyzed: It is sometimes necessary to extend analysis beyond the routine number of cells analyzed for best patient care practices. An example would be to rule out mosaicism at a specific confidence. An additional charge will be added for every additional 20 cells studied.

Individual special stains: It is sometimes necessary to use additional stains to clarify questionable polymorphic chromosome areas or subtle abnormalities. An additional charge will be added for every additional stain performed.

Limited Chromosome Analyses: To be used when less than a full study is performed/needed. This charge will replace a full study ordered.

FISH Cultures: It may be necessary to perform an additional culture to ensure enough sample is available for FISH testing when multiple FISH tests are ordered.