

**Shipping Address:**

UNMC Shipping & Receiving Dock  
 668 South 41<sup>st</sup> Street, Campus Zip 5440  
 Omaha, NE 68105  
 Lab Cell Phone: (402) 203-2110  
 CLIA # 28D0454363  
 CAP # 19749-09



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## Genetics Laboratory SPECIMEN REQUIREMENTS

<b>PRENATAL</b>				
Specimen Type	Amount	Tube Type	Special Instructions	TAT
Amniotic fluid (includes AFAFP & AChE)	15-20 ml ( $\approx$ 10 in 2 tubes)	Supplied by HGL	Room temperature. Deliver ASAP	6-8 days
Rapid Prenatal Aneuploidy Screen (FISH)	5 ml (additional)	Supplied by HGL	Amniotic fluid chromosomes must be ordered with Aneuploidy Screen	1-2 days
Quad Fetal Risk Assessment	2 ml serum or >2 ml whole blood	AFP tube or red top tube	Send serum at room temperature with a completed Quad Screen form (do not freeze).	3 days
Products of Conception	0.5-1.0 cm <sup>2</sup> (sterile)	Tissue culture media	4°C – prevent desiccation. Amniotic fluid – if IUFD or therapeutic abortion. Otherwise chorionic villi &/or fetal tissue. Deliver ASAP.	10-15 days
Paraffin POC Tissue Aneuploidy Screen (FISH)	3-4 paraffin unstained slides		Copy of Pathology Report	7-10 days
<b>POSTNATAL</b>				
Blood (peripheral)	3 ml	Sodium heparin	Room temperature	14 days
Newborn	1-3 ml	Sodium heparin	Room temperature	1-3 days
Deceased	3 ml	Sodium heparin	Collect under sterile conditions. Send specimen at room temperature.	15 days
Skin Biopsy	1-3 mm or more	Tissue culture media	Collect under sterile conditions. Send specimen at room temperature. [Cryopreservation & DNA storage upon request]	15 days
<b>FRAGILE X WITH CHROMOSOME STUDIES</b>				
Blood (peripheral)	3-5 ml	EDTA (purple top)	Room temperature.	7-14 days
Chromosome studies	3 ml	Sodium heparin	Room temperature.	
<b>CGH MICROARRAY ASSAY</b>				
Blood (peripheral)	1-3 ml	Sodium heparin or EDTA (purple top)	Room temperature. Deliver ASAP	10-14 days
Other Tissue	Please inquire – call (402) 559-5070			14-21 days
<b>MECP2 Sequencing for Rett Syndrome</b>				
Blood (peripheral)	1-3 ml	EDTA (purple top)	Room temperature. Deliver ASAP.	21-28 days
<b>MECP2 FISH Duplication</b>				
Blood (peripheral)	3-5 ml	Sodium heparin	Room temperature. Deliver ASAP.	7-10 days
<b>PTEN for Cowden or BRR syndrome – dx Autism Spectrum Disorder with Macrocephaly</b>				
Blood (peripheral)	1-3 ml	EDTA (purple top)	Room temperature. Deliver ASAP.	21-28 days
<b>PTPN11 for Noonan Syndrome</b>				
Blood (peripheral)	1-3 ml	EDTA (purple top)	Room temperature. Deliver ASAP.	21-28 days
<b>METHYLATION for Prader-Willi &amp; Angelman</b>				
Blood (peripheral)	3 ml	EDTA (purple top)	Room temperature. Deliver ASAP.	5-10 days

CANCER				
Bone marrow	2 ml	Sodium heparin	Bone marrow from 1 <sup>st</sup> or 2 <sup>nd</sup> aspirate using a sodium-heparinized syringe. Transport in a 3 ml sodium-heparin tube. Deliver ASAP.	1-3 days
Solid Tumor Tissue	>5 mm <sup>3</sup>	Tissue culture media	Representative tumor tissue collected under sterile conditions & transported in sterile tissue culture media. Deliver ASAP.	10-20 days
Blood – Cancer	5 ml	Sodium heparin	>10% blasts in peripheral blood. Room temperature. Deliver ASAP.	2-5 days
Lymph Node	≥5 mm <sup>3</sup>	Tissue culture media	Room temperature. Deliver ASAP.	10-20 days
FISH STUDIES PARAFFIN TISSUE (HER2/neu / Lymphoma / Solid Tumor)				
Paraffin Embedded Tissue	4-5 unstained slides mounted on + charged slides	H&E slide with the invasive tumor component outlined with a marker. Two stained 3-5 micron sections of formalin-fixed, paraffin embedded tissue on Plus slides, sectioned proximal to the H&E and a copy of the Pathology Report.		5-7 days
FISH for BLADDER CANCER MONITORING - UROVISION				
Urine	30 ml	Sterile container	Room temperature or 4°C – DO NOT FREEZE. Include a copy of the Pathology Report.	5-7 days
MOLECULAR PATHOLOGY (RT-PCR for Rhabdomyosarcoma, Ewing Sarcoma, Synovial Sarcoma & JAK2) - contact Regional Pathology Services at (800) 334-0459.				

## Specimen Shipping & Handling Information

1. Notify HGL in advance of specimen arrival and/or to arrange for specimen pickup (pickup provided within the Omaha-Council Bluffs metropolitan area only). Outside of this area, the transport of specimens by overnight express courier service is available and will be paid by HGL. Specimens should arrive within 24 hours of collection.
2. Specimens should be collected under sterile conditions and transported at room temperature unless otherwise indicated. Special arrangements are required if extremes in temperatures or if extended transport times are anticipated. **DO NOT FREEZE.**
3. Label specimen with **two patient identifiers (i.e., patient's full name & date of birth)** as well as date/time of specimen collection. Please use indelible ink or gummed patient labels to label specimens.
4. Send a completed cytogenetics TEST REQUEST information form with the specimen. Include the patient's name, date of birth, diagnosis, physician's name and phone number, billing/insurance information and tissue type. Forms are available on the Home page menu ([www.unmc.edu/geneticslab](http://www.unmc.edu/geneticslab)), or you can call us at (402) 559-5070, and we will fax the appropriate form to you.

## FISH Tests Available

### HEMATOLOGY / ONCOLOGY

<i>CML</i>	BCR/ABL [t(9;22) / LSI 9q34]	<i>Eosinophilia</i>	CHIC2 [4q12]	<i>Neuroblastoma</i>
<i>ALL</i>	TEL/AML [t(12;21)] TEL [12p13] Hyperdiploidy BCR/ABL [t(9;22)] / LSI 9q34 MLL [11q23] p16 [9p21]	<i>Lymphoma Probes</i> ALK [2p23 & variants] IgH / MYC / 8CEP [t(8;14)] IgH / CCND1 [t(11;14)] IgH / BCL2 [t(14;18)] IgH / MALT [t(14;18)]		NMYC [2p24.1] 1p36 deletion
<i>CLL</i>	12CEP D13S319 [13q14 / 13q34] ATM [11q23] IgH translocation p53 [17p13.1]	IgH [14q32 abnormalities] BCL6 [3q27] CMYB [6q23]* CMYC [8q24] CCND1 [11q13] ETV6 [12p13] TCR [14q11]		<i>Rhabdomyosarcoma</i> FKHR [13q14] PAX3 [2q35]
<i>MD/MPD</i>	EGR-1 [5q31] 8CEP 7CEP / D7S486 [7q31] MLL [11q23] D20S108 [20q12] BCR/ABL [t(9;22) / LSI 9q34] JAK2 [9p24.1]*	MALT1 [18q21 / t(11;18)] BCL2 [18q21]		<i>Sarcoma</i> Ewing [t(11;22) & variants] Liposarcoma - LSI CHOP [12q13] Liposarcoma - MDM2 [12q15]* Dermatofibrosarcoma Protuberans [t(17;22)]* Synovial - SYT [18q21 / t(X;18)] FUS [16p11]* (CREB or BBF2H7 [7q33])* Lipoma - HMGIC [12q14.3]* Clear cell sarcoma ATF1-EWS* [12q13-22q12]*
<i>MM</i>	D13S319 [13q14 / 13q34] IgH translocation IgH / CCND1 [t(11;14)] IgH / FGFR [t(4;14)] IgH / MAF [t(14;16)] p53 [17p13.1] 6q21 Hyperdiploidy [CEP 9/11/15q22]	<i>Breast Cancer</i> HER2/neu [17q12] TOP2A [17q11.2-12]		<i>Bladder Cancer Panel</i> CEP3 CEP7 CEP17 p16 [9p21]
<i>AML</i>	7CEP 8CEP ML [11q23] ETO/AML1 [t(8;21)] PML/RARA [t(15;17)] CBFB [16q22]			<i>Neurological Cancer</i> 1p36 / 1q25 19q13 / 19p13 EGFR [7p12] PTEN [10q23]

### CONSTITUTIONAL

#### *Microdeletion / Microduplication*

1p deletion [1p36]	Smith Magenis [17p11.2]
Waardenburg Syndrome I (PAX3) [2q35]*	Miller Dieker [17p13.3]
Waardenburg Syndrome III [2q36-37]*	Charcot-Marie-Tooth / HNPP [17p12]*
Wolf-Hirschhorn [4p16.3]	Saethre-Chotzen (TWIST gene) [7p21.1]*
Cri-du-Chat [5p15.2]	DiGeorge / VCFS Syndrome [22q11.2]
Sotos Syndrome (NSD1) [5q35]	Steroid Sulfatase (STS) [Xp22.3]
Williams Syndrome [7q11.23]	Kallmann Syndrome [Xp22.3]
CHARGE [8q12.2]	Duchenne / Becker Muscular Dystrophy [Xp21.2]*
Langer Giedion [8q24]*	XIST, X inactivation site [Xq13]
Pallister-Killian Syndrome [12p]	MECP-2 duplication [Xq28]
Prader-Willi / Angelman [15q11.2]	SRY [Yp11.3]
Rubinstein-Taybi [16q13.3]*	

\* Home-brew probes

#### **Marker / Derivative Identification**

Centromere enumeration  
Whole chromosome Paint Probes  
M-FISH  
Subtelomere probes for each chromosome

#### **AneuVysion**

**Prenatal Screen**  
(Enumeration for X, Y, 13, 18, & 21)

Revised: 10/1/2009