

<i>Test Name</i>	<i>Specimen Requirements</i>	<i>Turn Around Time</i>	<i>Comments</i>
Chromosome Analysis (<i>Karyotype</i>)			
<i>Referral form must accompany sample</i>			
Amniotic Fluid (716-2545)	2 x 15 ml orange screw-top polypropylene tubes. Discard the first 2 mls; then collect 15-25 mls amniotic fluid.	6-10 days Avg 8 days	Keep at room temperature. Do Not Centrifuge.
Peripheral Blood Routine High resolution Unstimulated (716-2553)	Green-top sodium heparin tubes only Adult: 5-10 mls peripheral blood Child: 2-5 mls peripheral blood PUBS: 1-2 mls percutaneous umbilical blood	48 hr STAT 7-14 days Avg 8 days	Swab with alcohol. Do not swab with Betadine. Keep at room temperature. Do Not Centrifuge.
Bone Marrow (& core) (716-2553)	Aspirate 1-5 mls of a first draw into a syringe coated with 200 units preservative-free sodium heparin.	48 hr STAT 6-10 days Avg 7 days	Add 5 mls media if >2 hr delivery delay to the lab. Keep at room temperature. Do Not Centrifuge.
Chorionic Villi Sampling (716-2545)	Place in laboratory-provided screw-top tubes containing sterile transport media.	6-10 days Avg 8 days	Keep at room temperature.
Products of Conception (716-2545)	Place in laboratory-provided screw-top tubes containing sterile transport media.	10 -14 days Avg 12 days	Keep at room temperature. Refrigerate overnight if necessary.
Solid Tumors (716-2545)	Place in laboratory-provided screw-top tubes containing sterile transport media.	10-18 days Avg 14 days	Keep at room temperature.
Tissues (716-2545)	Place in laboratory-provided screw-top tubes containing sterile transport media.	10-18 days Avg 14 days	Keep at room temperature. Refrigerate overnight if necessary.
AFTER HOURS, IN HOUSE SAMPLES MAY BE LEFT IN THE DROP BOX @ G-09 HANES			

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<i>Molecular Cytogenetics</i> <i>Referral form must accompany sample</i>			
Fluorescence in situ hybridization: FISH (716-2064)	FISH can be performed on specimens used for cytogenetic studies - OR - on slides - OR - specimens for pathology studies.	6hrs to 7 days Avg 48-72 hrs	Call for specific probe details.
Prenatal FISH LSI 13 CEP-X CEP 18 CEP-Y LSI 21	FISH can be performed directly on amniotic fluid specimens used for cytogenetic studies	24 hours or less	Done only in conjunction with chromosome studies. Interphase studies alone not recommended.
STAT Bloods LSI 13 CEP-X CEP 18 CEP-Y LSI 21	FISH can be performed directly on peripheral blood specimens used for cytogenetic studies	6-24 hours	Done only in conjunction with chromosome studies. Interphase studies alone not recommended.
Microdeletion syndromes: Wolf- Hirschorn 4p16 Cri-du-chat 5p15.3 Williams 7q12 Retinoblastoma 13q12 Prader-Willi 15q12 Angelman 15q12 Miller-Dieker 17p13 Smith-Magenis 17p11.2 DiGeorge or 22q11 VeloCardioFacial Syndrome (VCF) Kalman Xp22.3 STS Xp22.3 SRY Yp11.3	Green-top sodium heparin tubes Adult: 5-10 mls peripheral blood Child: 2-5 mls peripheral blood -OR- prepared slides.	After chromosome harvest (72 hours): FISH = 24-72 hrs Total = 5-6 days	Done only in conjunction with chromosome studies. Interphase studies alone not recommended. Metaphase chromosome needed (72hr+). If Prader-Willi is negative, obtain a sample for DNA/methylation studies (see DNA section).
Subtelomeres	Analysis of subtelomeric chromosome rearrangement/deletion/duplication	3-5 days after chromosomes	Done in conjunction with chromosome studies.

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<i>Neural Tube / Biochemical Genetics (716-6976)</i>			
Maternal Serum Quad Screen: AFP/hCG/uE3/Inhibin	5-7 mls whole blood in a red top tube -OR- 2 mls serum.	Next working day	4 mls whole blood. 2 mls serum minimum. Okay to spin blood, remove serum and freeze.
Amniotic Fluid AFP	2-3 ml sample from amniotic fluid for cytogenetics.	Next working day	
AChE	Same as AFP.	5-10 working days	
Cancer AFP (x63321)	5-7 mls whole blood in a red top tube -OR- 1 ml serum.	Next working day	2 mls whole blood. 1 ml serum minimum. Okay to spin blood, remove serum and freeze.

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<i>Pediatric Specialty Lab (716-2549)</i>			
L/S ratio / phosphatidylglycerol (PG)	minimum of 8cc amniotic fluid in sterile container transported on ice or with cold packs	3.5 hours	Lab will fax or call results ASAP as indicated
Alpha-1-Antitrypsin	2-5 mls whole blood in red top tube.	72 hours	Spin, remove serum off top, freeze.
Sweat Chlorides for CF testing	>0.0750g of sweat collected in a glass vial, lid on tight.	Same day	Fax report. On positives call Dr. Schechter
Colon Suction Biopsies for Hirsprungs	Tissue, if received from outside need to be flash frozen OCT embedding media and packed on dry ice.	Next day	Tissue flash frozen in liquid nitrogen. Store at -70C Testing for AchE
Human Complement C3	2-5 mls whole blood in red top tube	48 hours	Spin, remove serum off top, freeze

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<i>Molecular Genetics</i> <i>(716-3321)</i>			
Angelman Syndrome Methylation Analysis	2-4 mls whole blood in purple top EDTA tube.	7-10 days	Keep at room temperature.
Chimerism for bone marrow transplantation	2-4 mls whole blood in purple top EDTA tube -OR- 1-3 mls bone marrow aspirate.	Pre: 2-3 weeks Post: 1-2 days	Keep at room temperature.
Cystic Fibrosis Screening and Genotyping	~1 ml whole blood in purple top EDTA tube.	7-10 days	Ethnic background required for proper interpretation.
DNA Isolation Banking/Sendout	2-4 mls whole blood in purple top EDTA tube	5-10 days	
FMR1 Gene Analysis: -Fragile X Syndrome -Fragile X-associated Tremor/Ataxia Syndrome -Premature Ovarian Failure	2-4 mls whole blood in purple top EDTA tube	PCR-based 7-10days Reflex Southern blot 3-4 weeks	Keep at room temperature.
Kennedy Disease/Spinal Bulbar Muscular Atrophy	2-4 mls whole blood in purple top EDTA tube	7-10 days	Keep at room temperature.
Microarray Analysis	2-4 mls whole blood in purple top EDTA tube	2-3 weeks	Keep at room temperature.
Myotonic Dystrophy Analysis	2-4 mls whole blood in purple top EDTA tube	PCR-based 7-10days Reflex Southern blot 3-4 weeks	Keep at room temperature.
Prader-Willi Syndrome Methylation Analysis	2-4 mls whole blood in purple top EDTA tube	7-10 days	Keep at room temperature.
Uniparental disomy * : -Prader-Willi syndrome -Angelman syndrome -Other chromosome specific	2-4 mls whole blood in purple top EDTA tubes. Blood from both parents required.	2 weeks	Keep at room temperature. Blood from both parents required.*

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X Chromosome Inactivation Studies	2-4 mls whole blood in purple top EDTA tube.	7-10 days	Keep at room temperature.
Zygoty Studies – mono- vs di-	1-2 mls whole blood in purple top EDTA tube.	7-10 days	Keep at room temperature.

*Contact the laboratory before submitting samples for uniparental disomy studies