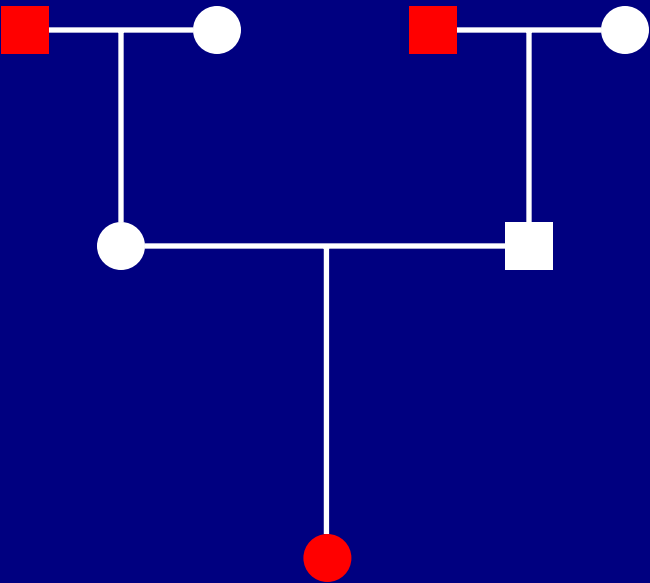


CENTER FOR HUMAN GENETICS LABORATORY

SPECIMEN COLLECTION & HANDLING
PROCEDURE MANUAL



University Hospitals Case Medical Center

To: All Staff

From: P. Michael Smith

Re: Specimen Collection & Handling Procedure Manual

Date: July 14th, 2010

This manual contains the guidelines and instructions for collection and handling of all specimen types for cytogenetics and molecular genetics analysis at the Center for Human Genetics Laboratory.

Reviewed and Approved

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**Center for Human Genetics Laboratory
Specimen Collection and Handling Procedure Manual**

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GUIDELINES FOR SUBMISSION OF AMNIOTIC FLUID SPECIMENS FOR CYTOGENETIC ANALYSIS

Arranging for Analysis:

In order to provide efficient analysis and timely results, this laboratory should be notified of all procedures as soon as possible, in order to make arrangements for an expeditious courier pickup.

Collection:

1. Aspirate amniotic fluid using a sterile Becton-Dickinson 20 cc syringe. It is preferable to use Becton-Dickinson syringes, since some other brands of syringes have been found to be toxic to amniotic fluid cells.
2. It is **very important** to separate the initial 2 - 3 cc's of fluid from the main aspirate, since this "first draw" may contain some maternal cells trapped within the needle during insertion. We recommend that the first 2 - 3 cc's be collected in a separate syringe and clearly labeled "**1st Draw**". This first draw should be sent to this laboratory with the remaining specimen, since it provides an important quality control in cases of suspected maternal cell contamination. If a first draw is not obtained it is essential that the sex of the fetus be identified.
3. After changing syringes, collect the amniotic fluid in one or two additional syringes. The total volume collected is dependent on gestational age:
 - a. 15 weeks gestation or later: 20-25 cc of amniotic fluid.
 - b. Earlier than 15 full weeks from last menstrual period:
 - i. Collect 1 cc for each week of gestation. (i.e., 14 cc for 14 weeks gestation.)
4. The needle **must** be removed before transportation of the specimen is possible. Syringes should be recapped with the original tip cover. Please cap tightly, and be sure the syringe is not leaking. It is not necessary to transfer the fluid into a separate container if the syringes are secure and fluid can be contained during shipment. Capped original syringes are the preferred means of sending the amniotic fluid specimen to the laboratory. If, however, syringes cannot be sent, the specimen may be aseptically transferred to sterile conical centrifuge tubes for subsequent transport to the laboratory. Please keep each draw volume in its own tube (see #5 below). **Do not transfer** the specimen to sterile glass tubes (i.e. Vacutainers) since the amniotic fluid cells in the specimen preferentially adhere to glass.

5. Each syringe or tube **must** be labeled with the patient's complete name and with the number syringe as used in the procedure (i.e., #1 (= first draw), #2 (= second draw), etc.) All syringes from a single gestational sac should be placed in one plastic specimen bag with the accompanying requisition in the side pocket of the bag. **Do not place the requisition in the same side of the bag as the specimen.** Multiple gestations require careful labeling and separation of the specimen containers. Indicate the number and position of the fetus (i.e., twin #A, upper right) as well as the sex of the fetus if known.

6. A genetics lab requisition must accompany each specimen and must include the following information:
 - a. Patient's complete name, address, and date of birth.
 - b. Billing information as follows:
 - i. Complete name, address and telephone of guarantor,
 - For Insurance billing:
Complete name and address of the insurance company, policy number, group number and complete name of the policyholder
 - For Institutional billing:
Complete name, address and telephone number of the institution, and name and telephone number of the contact person for financial inquiries
 - c. For Ohio Medicaid patients include the following information:
 - i. Complete name and address of the guarantor (parent or guardian)
 - ii. Medicaid recipient number of the patient (12 digits)
 - iii. If applicable, complete HMO information
 - d. Ultrasound information including date, gestational age and sex if known
 - e. Reason for referral or clinical indications
 - f. Pregnancy history (gravida, para, number of previous miscarriages, number of living children and any family history of abnormalities seen or diagnosed).
 - g. Referring physician's complete name, address, and telephone number.

7. The following commonly encountered problems should be handled as follows:
 - a. In case of a bloody amniotic fluid tap, heparin **should be added** to the sample. We recommend a concentration of 10 units per cc of fluid (i.e., 0.2 cc of a 1000 u/cc heparin suspension to 20 cc of amniotic fluid). If the sample is grossly bloody, the concentration can be increased to 15 u/cc. Indicate on the requisition that heparin has been added.

 - b. In cases of uterine contraction, only a few cc's of fluid might be obtained. We can attempt chromosome analysis on a small specimen but, a comprehensive study might not be possible. If a repeat amniocentesis is necessary, we will request a repeat sample within 7 days of receipt of the initial specimen.

Transportation:

1. Notify this laboratory (216-983-1134) as soon as it is known that a specimen will be available to be picked up. We will arrange for transportation of the specimen to the lab.
2. Specimens that will be sent through an outside courier should be packaged according to standard medical safety practices, in order to prevent depression of the syringe. In warm weather, a cold (**NOT FROZEN**) gel pack should be placed in the container. Under no conditions should amniotic fluid ever be frozen unless specifically directed by the physician or this laboratory.
3. It is preferable that specimens not remain in the possession of an overnight courier service. If a specimen is collected outside normal business hours, it should be held at room temperature and away from direct light. Mark the specimen "For Genetics Courier Only", and be sure that we are notified at the beginning of the next business day. (Business hours are 9:00 a.m. - 5:00 p.m.).
4. If a specimen is to be shipped overnight via Federal Express, this laboratory can be billed for the Fed Ex shipping fee via Federal Express.

Results:

1. If desired by the referring physician, our genetics associates will contact the patient directly with normal results. Final results are mailed to the referring physician and upon request, to the referring laboratory.
2. In most cases, abnormal results are known within 7-10 days and are faxed and reported to the referring physician by phone. In all cases of elective termination, a tissue specimen will be requested for confirmation. (Please refer to Guidelines for Submission of Products of Conception).

GUIDELINES FOR SUBMISSION OF CHORIONIC VILLI FOR CYTOGENETIC ANALYSIS

Arranging for Analysis:

The laboratory must be notified in advance of drawing chorionic villi specimens, due to the amount of time involved in dissecting the specimen and initiating the cultures.

Collection:

1. Using a sterile Becton-Dickinson 20cc syringe containing approximately 5ml of sterile physiological saline or transport medium, aspirate 15-26 mg of chorionic villi. If two attempts are necessary to obtain this amount, use separate syringes and submit both of them for analyses. Label each syringe with the patient's full name.
2. After removal of the catheter (transcervical procedure) or needle (transabdominal procedure), firmly replace the tip(s) of the syringe(s), and place the syringe(s) in one plastic specimen bag, with the accompanying requisition in the side pocket of the bag.
3. A Center for Human Genetics requisition must accompany each specimen and must include the following information.
 - a. Patient's full name, address, and date of birth
 - b. Ultrasound dating and any relevant ultrasound information
 - c. Reason for referral
 - d. Gestational age
 - e. Pregnancy history
 - f. Referring physician's name, address, and phone number

Transport:

1. The Center of Human Genetics should be notified (216) 983-1134 as soon as the specimen is ready to be picked up, and we will arrange for pick-up.
2. Specimens to be sent via an outside courier service should be placed in a styrofoam container for the protection of the specimen (i.e. to prevent depression of a syringe plunger). In very warm weather, a cold gel pack should be placed in the container. We prefer not to have specimens in the possession of the courier service overnight, so please call us to arrange pick-up as early as possible.

3. If the specimen is collected outside normal hours, it must be refrigerated until the next business day. Mark the specimen “For Genetics Courier Only”, and be sure that we are notified at the beginning of the next business day. (Hours are 9:00 – 5:00)

Results:

1. Final results are available within 2-3 weeks. If desired by the referring physician, a genetics associates will contact the patient directly with normal results.
2. In most cases, abnormal results are known within 10-14 days and are reported to the referring physician by telephone. In all cases of elective termination, we request a tissue specimen for confirmation (Please refer to Guidelines for Submission of Production of Conception).

GUIDELINES FOR SUBMISSION OF BLOOD SPECIMENS FOR CYTOGENETIC ANALYSIS

Collection:

1. Heparinized blood (sodium or lithium) is currently recommended. We recommend the use of green top vacutainers. However, any blood will be accepted provided that it is not clotted, lysed, or improperly contained (broken or leaking). The section supervisor should be consulted before any specimen is rejected.
 - a. Adults and children over two years of age: 5-8 cc in a large (10 cc) green top tube.
 - b. Children under 2 years of age: 1-2 cc in a small (2 cc) green top tube.
2. Following collection, immediately invert the tube several times to thoroughly mix.
3. Be sure that each tube is labeled with the full name of the patient and the date the specimen is collected. Be sure to designate cord and fetal blood to distinguish it from maternal blood.
4. If both cytogenetic and molecular (Fragile X, uniparental disomy, methylation) studies are ordered, both a heparinized vacutainer and a lavender top EDTA vacutainer are required.
5. A genetics lab requisition must accompany each specimen and must include the following information:
 - a. Patient's full name and address
 - b. Patient's date of birth
 - c. Patient's gender
 - d. Unique identifying number
 - e. Referring physician's name, address, and phone number
 - f. specimen source
 - g. specimen collection date
 - h. test requested
 - i. Clinical indications for the test(s)
 - j. Pregnancy status and history, if applicable

Transport:

1. The Center for Human Genetics Laboratory should be notified (216-983-1134) as soon as the specimen is ready to be picked up and we will arrange for pick-up.
2. If the specimen is collected outside normal business hours, it may be held at room temperature until the next business day. Mark the tube(s) "For Center for Human Genetics Laboratory," and be sure that we are notified at the beginning of the next business day. (Hours are 9:00 a.m. - 5:00 p.m.). The blood section staff is available to pick up late stats at the hospital on a limited basis:

Results:

1. Results of routine studies generally are available in about two weeks.
2. A STAT analysis, identifying numerical abnormalities and gross structural abnormalities, can be accomplished in 3-4 days. Final results are available in about 7-10 days. STAT designation generally is reserved for newborns and cases involving an ongoing pregnancy.
3. Final results are mailed to the referring physician and upon request, to the referring laboratory.

**GUIDELINES FOR SUBMISSION OF
BONE MARROW AND LEUKEMIC BLOOD SAMPLES
FOR CHROMOSOME ANALYSIS**

Collection:

1. Samples can be handled most expeditiously when received before 4 p.m., preferably Monday through Friday.
2. The cytogenetics laboratory personnel must be notified that they will be receiving a sample and provided with a brief clinical description, including chemotherapeutic regimes, transplant status, and suspected diagnosis. The Center for Human Genetics Laboratory's phone number is (216) 983-1134.
3. At times other than regular laboratory hours, hold the sample at **room temperature** (do not refrigerate or freeze) until it can be delivered to the laboratory. If available, the specimen should be aseptically transferred to a conical centrifuge tube containing 5 ml of a tissue culture media such as RPMI 1640 or McCoy's 5A. Hank's Balanced Salt Solution may also be used if no other media is available.

Transport:

1. Fresh sodium heparinized aspirate is required. (Approximately 0.1 ml sodium heparin should be added for every 5 ml bone marrow).
2. The sample should be hand-delivered to the laboratory by courier. The address is: Center for Human Genetic Laboratory, 10524 Euclid Avenue, 6th Floor, Cleveland, Ohio 44106, or call our laboratory at 983-1134 to arrange for the courier service.
3. All samples should be accompanied by a genetics lab requisition which should include:
 - a. Appropriate billing information, including patient's address and insurance
 - b. Patient's name and hospital number
 - c. Clinical history/differential diagnosis
 - d. Chemotherapy information/transplant status
 - e. Date of birth
 - f. Referring physician's name and phone number
 - g. Clear indication of the type of testing requested, i.e., cytogenetic analysis, FISH analysis including probes to be used, or both

4. Two culture methods are routinely set up on patients with hematological disorders. These include either direct or overnight cultures, and 24-hour cultures. Bone marrows are set up without a mitotic stimulant. Peripheral bloods are set up with and without stimulation. The stimulant provides a baseline or constitutional karyotype. Cells of abnormal origin will be detected only in blood that is set up without stimulant. However, if the abnormal cells are not being spilled from the marrow into the blood, these unstimulated cultures simply will not yield cytogenetic results. Therefore, when looking for an abnormal karyotype in the patient, it is preferable to study bone marrow.
5. If special studies are required for donor/recipient heteromorphisms, we must have blood on both donor and recipient prior to transplant. If possible, this should be done 2-3 weeks prior to transplant in case of culture failure.

Results:

1. Completion of a routine analysis takes 7-10 days.
2. A STAT analysis, from which only major structural or numerical abnormalities or identification of the Philadelphia chromosome can be made, can be accomplished in 3-4 days. Stats must be cleared with the laboratory.
3. FISH analysis generally takes 4 working days to complete. Specimens received on Thursday or later will not have FISH results available until the next Tuesday unless special arrangements are made in advance.

GUIDELINES FOR SUBMISSION OF TUMOR SPECIMENS FOR CHROMOSOME ANALYSIS

Collection:

1. Samples can be handled most expeditiously when received before 4 p.m., preferably Monday through Friday.
2. The cytogenetics laboratory personnel must be notified that they will be receiving a sample and provided with a brief clinical description, including suspected diagnosis and site of tumor. The Center for Human Genetic Laboratory's phone number is (216) 983-1134.
3. At times other than regular laboratory hours, hold the sample at **room temperature** (do not refrigerate or freeze) until it can be delivered to the laboratory.

Transport:

1. Specimens should be obtained aseptically and placed in complete media for transport. The Center for Human Genetic Laboratory will provide such media on a prearranged basis.
2. The sample should be hand-delivered to the laboratory by courier. The address is: Center for Human Genetic Laboratory, 10524 Euclid Avenue, 6th Floor, Cleveland, Ohio 44106, or call our laboratory at 983-1134 to arrange for the courier service.
3. All samples should be accompanied by a genetics lab requisition which should include:
 - a. Appropriate billing information, including patient's address and insurance
 - b. Patient's name and hospital number
 - c. Clinical history/differential diagnosis
 - d. Site of tumor
 - e. Date of birth
 - f. Referring physician's name and phone number

4. It is important to provide the sample with adequate nutrients and a good buffering system to maintain viability of the cancerous cells. Generally, cultures harvested within 48 hours of obtaining a specimen will have viable cells present in them only if an appropriate media has been used for transport. The Center for Human Genetic Laboratory provides two forms of transport media, one for tumors and one for tissue specimens. It is important to use the appropriate media for each type of specimen.

Results:

1. Completion of a routine analysis takes anywhere from 2 to 8 weeks depending on the viability of the cells obtained, growth rate and complexity of the karyotype.

GUIDELINES FOR SUBMISSION OF LYMPH NODE SAMPLES FOR CHROMOSOME ANALYSIS

Collection:

1. Samples can be handled most expeditiously when received before 4 p.m., preferably Monday through Friday.
2. The cytogenetics laboratory personnel must be notified that they will be receiving a sample and provided with a brief clinical description, including chemotherapeutic regimes, and suspected diagnosis. The Center for Human Genetics Laboratory's phone number is (216) 983-1134.
3. At times other than regular laboratory hours, hold the sample at **room temperature** (do not refrigerate or freeze) until it can be delivered to the laboratory. The specimen must be aseptically transferred to a sterile container with approximately 5 ml of room temperature tissue culture media such as RPMI 1640 or McCoy's 5A to support the metabolism of the cells present in the sample. Hank's Balanced Salt Solution may also be used if no other media is available. However, media can be provided by our laboratory and stored frozen until needed.

Transport:

1. The sample should be at least a cubic centimeter in size, and placed in growth support media as soon as possible after obtained.
2. The sample should be hand-delivered to the laboratory by courier. The address is: Center for Human Genetic Laboratory, 10524 Euclid Avenue, 6th Floor, Cleveland, Ohio 44106, or call our laboratory at 983-1134 to arrange for the courier service.
3. All samples should be accompanied by a genetics lab requisition which should include:
 - a. Appropriate billing information, including patient's address and insurance
 - b. Patient's name and hospital number
 - c. Clinical history/differential diagnosis
 - d. Chemotherapy information
 - e. Patient's date of birth
 - f. Referring physician's name and phone number
 - g. A clear indication of the type of testing requested, i.e., cytogenetic analysis, FISH analysis including probes to be used, or both

4. It is important to provide the sample with adequate nutrients and a good buffering system to maintain viability of the cancerous cells. Generally, cultures harvested within 48 hours of obtaining a specimen will have viable cancerous cells present in them only if an appropriate media has been used for transport. The Center for Human Genetic Laboratory provides two forms of transport media, one for tumors and one for tissue specimens. It is important to use the appropriate media for each type of specimen.

Results:

1. Completion of a routine analysis takes 7-14 days.
2. A STAT analysis, from which only major structural or numerical abnormalities can be made, can be accomplished in 4-5 days. Stats must be cleared with the laboratory.
3. FISH analysis generally takes 4 working days to complete. Specimens received on Thursday or later will not have FISH results available until the next Tuesday unless special arrangements are made in advance.

GUIDELINES FOR SUBMISSION OF TISSUE BIOPSY SAMPLES FOR CHROMOSOME ANALYSIS

Arranging for Analysis:

1. Samples can be handled most expeditiously when received in the morning, Monday through Friday. Laboratory hours are 8:30 a.m. - 5:00 p.m. Monday-Friday.
2. Notify the Laboratory (216-983-1134) when the sample is available. Provide a brief clinical description and indicate the desired testing.
3. When a specimen is collected outside normal business hours, it can be refrigerated at no less than 40°F (= about 4°C) until the next business day. **DO NOT FREEZE**. Mark the container "For Center for Human Genetics Laboratory", and be sure that we are notified at the beginning of the next business day.

Collection:

1. The biopsy must be obtained under sterile conditions. Be careful to remove all betadine with alcohol wipes prior to obtaining the sample.
2. For the biopsy, use a 3 or 4 mm punch biopsy kit for obtaining a skin sample. For stillborns, the following tissue may be used for analysis: skin, kidney, lung, deep muscle.
3. Place the biopsy in a container of sterile HBSS containing gentamicin, which can be provided by the Cytogenetics Laboratory, or in sterile Ringer's Lactate or other physiological solution. **DO NOT USE FORMALIN!**
4. The container should be labeled with the patient's full name, sealed and wrapped in parafilm to prevent leakage or contamination during transport. This container should be placed in a plastic specimen bag with an accompanying requisition in the side pocket of the bag.
5. A Center for Human Genetics requisition must accompany each specimen and must include the following information.
 - a. Full name of the patient, address, date of birth, and hospital number
 - b. Gestational age (if appropriate)
 - c. Any anomalies
 - d. Pregnancy history (if appropriate)
 - e. Sex (if known)
 - f. Referring and/or attending physician's name(s), address, and telephone number
 - g. Source of each sample (e.g. inner aspect of the forearm)

6. Please include copies of the patient's insurance information with the lab requisition and specimen.

Transport:

1. Most tissue specimens should be handled through the Pathology Department/Send-Out Laboratory of the referring institution. You are responsible for following the appropriate procedures within your institution.
2. Telephone (216-983-1134) when a specimen is available. We can arrange for transportation.

Reporting the Results:

1. Routine analysis generally requires 3-5 weeks.
2. Special testing (i.e. biochemical analysis or DNA analysis), building up cell lines or freezing cells must be discussed with Laboratory.
3. A tissue culture is reported as a "failure" if, after at least 3 weeks following culturing of the specimen, there is no evidence of any cell colony growth in any part of any culture vessel, whether petri dish or flask. A culture is also reported as a failure if it is overwhelmed with a contamination that cannot be controlled.

SUBMISSION OF PRODUCTS OF CONCEPTION, FETAL LOSSES OR STILLBIRTHS FOR CYTOGENETIC STUDY

Collection:

1. Select one or more viable-looking samples of tissue.
 - a. For P.O.C., submit about one cubic centimeter of chorionic villi, membrane or sac. Take care to minimize the amount of maternal decidua included with the villi since maternal cells will grow in culture, giving a maternal rather than a fetal karyotype.
 - b. For defined fetus or stillborn, submit a piece of deep muscle, Achilles tendon, lung or kidney about the size of an eraser on a pencil.

****NOTE**** The Center for Human Genetics Laboratory cannot dispose of excess fetal tissue. Large specimens must be returned to you at your expense.

Do not send a specimen **if**:

- a. It has been stored more than five(5) days or has been frozen.
 - b. It is in a container that cannot be kept closed or leaks.
 - c. It is in a non-sterile, non-physiologic medium.
 - d. It is known to be infected with bacteria, fungi, yeast, or mycoplasma.
 - e. **It has been exposed to formalin or formaldehyde.**
1. Transfer each specimen aseptically to a sterile container. Cover the specimen with Hanks Balanced Salt Solution (with Phenol Red), Ringer's Lactate or other physiologic solution.
 2. The container should be labeled with the patient's full name, sealed and wrapped in parafilm to prevent leakage or contamination during transport. This container should be placed in a plastic specimen bag with an accompanying requisition in the side pocket of the bag.
 3. A Center for Human Genetics requisition must accompany each specimen and must include the following information.
 - a. Full name of the patient, address, date of birth, and hospital number
 - b. Gestational age
 - c. Any fetal anomalies
 - d. Pregnancy history (if available)
 - e. Sex of fetus (if known)
 - f. Referring and/or attending physician's name(s), address, and telephone number
 - g. Source of each sample (e.g. inner aspect of the forearm)

4. Please include copies of the patient's insurance information with the lab requisition and specimen.

Transport:

1. Most tissue specimens should be handled through the Pathology Department/Send-Out Laboratory of the referring institution. You are responsible for following the appropriate procedures within your institution.
2. Telephone (216-983-1134) when a specimen is available. We can arrange for transportation.
3. When a specimen is collected outside normal business hours, it can be refrigerated at no less than 40°F (= about 4°C) until the next business day. **DO NOT FREEZE.** Mark the container "For Genetics Center", and be sure that we are notified at the beginning of the next business day. (Hours are 9:00 a.m. - 5:00 p.m. Mon. - Fri.).

GUIDELINES FOR SUBMISSION OF SPECIMENS FOR DNA ANALYSIS

NOTE: In most cases, routine chromosome analysis and FISH studies must be done prior to molecular studies. If these studies have already been completed, please forward a copy of the results to this laboratory. The laboratory must be called prior to any DNA analysis.

Chromosomal Analysis (In conjunction with additional DNA Analysis)

Specimen Requirements: A minimum of 3 – 5cc peripheral blood in a Heparinized (green top) tube (preferably Sodium Heparin) is required. Please send additional blood (6 – 8cc total) for adult and older pediatric patients. Unless directed by a physician, or this laboratory, blood specimens for chromosomal analysis should never be frozen or refrigerated. See specimen requirements list for more details on all offered tests.

Molecular Analysis

Specimen Requirements: A minimum of 3 – 5cc peripheral blood in an EDTA (purple top) tube is required. Please send 6 – 8cc total for adult and older pediatric patients when possible. Smaller children may require less blood depending upon the analysis requested. Please contact the lab for exact requirements before the specimen is drawn. Unless directed by a physician, or this laboratory, blood specimens for DNA analysis should never be frozen or refrigerated. See specimen requirements list for more details on all offered tests.

Fibroblast cultures may be submitted upon prior approval with the laboratory. Please call the laboratory prior to sending 2 T-25 flasks. Unless directed by a physician, or this laboratory, fibroblast cultures in media, for chromosomal or DNA analysis, should never be frozen or refrigerated.

Samples will not be processed without an appropriate laboratory requisition. Printable versions of current laboratory requisitions are available starting on page 23 of this manual.

For a complete listing of tests currently available in our Molecular Diagnostic Laboratory, or for any additional information, please visit our website, www.chglab.com or contact the laboratory at 216-983-1134.

GUIDELINES FOR SUBMISSION OF SPECIMENS TO THE FISH LABORATORY

1. A Center for Human Genetics Laboratory Requisition, specifying a clear indication of test request, should accompany all samples. If specimen is a follow-up, previous karyotype and/or FISH results should be included.
2. FISH testing needs to be requested by the referring physician or laboratory director. No other requests (by a technologist) will be accepted unless approved by a laboratory director.
3. The specimen of choice for metaphase analysis is a slide or coverslip with fixed cells. See the appropriate blood/amnio/tissue/cancer protocol for the slide or coverslip preparation. Slides requiring metaphase analysis (structural abnormalities, microdeletions) should contain at least 15 metaphases per 22mm X 22mm area. Slides of a mosaic karyotype (i.e. markers) should have enough metaphases to yield 5 metaphases with the abnormality. (Example: a marker present in 20% of metaphases examined by cytogenetic analysis should have at least 25 metaphases per 22mm X 22mm area.) Slides with less than the required number of metaphases will be designated as “sub-optimal” and evaluated on a case-by-case basis.
4. The specimen of choice for AneuVysion™ is fresh amniotic fluid. Before planting the amniotic fluid sample, the recommended volume (see below – adopted from AneuVysion™ package insert) should be set aside for AneuVysion™ in a centrifuge tube labeled with specimen name and number and stored at room temperature.

<u>Gestation Weeks</u>	<u>Minimum Volume (ml)</u>
11 – 15	2
16	3
17	3
18	4
19	4
20 – 27	5+

The sample may sit for up to 3 days (over weekend) at room temperature. Bloody samples and samples with a blood-tinged pellet should be noted in the comment section on the FISH Lab Requisition, and caution taken when results are reported since these samples could contain maternal cells.

5. The peripheral blood specimen of choice for rapid aneuploid detection is a fresh blood smear (see blood protocol for smear preparation.) The cancer specimen of choice for interphase analysis is a fixed slide. The slide should contain approximately 300 interphase cells per 22mm X 22mm area. Slides with less than 300 cells will be designated as “sub-optimal” and evaluated on a case-by-case basis.
6. Attempts will be made to complete an analysis whenever possible (i.e. using double probe volume to increase chances of completion).

UroVysion™ Specimens

1. The UroVysion™ assay is a genetic test designed for recurrence of transitional cell carcinoma of the bladder by detected aneuploidy for chromosomes 3, 7, 17 and the loss of the 9p21 locus. UroVysion, along with cystoscopy, is a noninvasive test for monitoring recurrence in patients diagnosed with bladder cancer.
2. UroVysion requires voided urine, in an amount greater than 33 ml. Mix voided urine 2:1 with Carbowax preservative and ship on ice packs immediately or refrigerate and ship on ice packs within 24 hours. Samples must be received by the Center for Human Genetics Laboratory within 72 hours of collection.
3. For non-UH institutions, UroVysion collection kits can be obtained by contacting the Center for Human Genetics at (216) 983-1134. For UH entities, UroVysion collection kits can be obtained by ordering through ORACLE iProcurement. The ORACLE internal catalog number is: 328137
4. All UroVysion specimens must be accompanied by a Center for Human Genetics Laboratory UroVysion specimen requisition. This specific requisition can be faxed or emailed upon request by calling the laboratory at (216) 983-1134, or by printing and/or downloading from our website, www.chglab.com

Medical Record Number: _____

SPECIMEN INFORMATION

Type: Peripheral Blood Bone Marrow Lymph node Solid Tumor (specify) _____ Other (specify) _____

★ **Date of specimen collection:** _____ ★ **Where drawn (institution):** _____

Post-treatment Y / N Date of last treatment _____ Medication/treatment used _____

PATIENT INFORMATION

Name (Last, First) _____ Phone (H) (____) _____ DOB ____/____/____

Address _____ (W) (____) _____ SS# ____-____-____

City/State/Zip _____ Sex: Male Female

★ **Transplant patient Y/N** Donor Recipient ★ **Sex of transplant match:** Male Female

REFERRING PHYSICIAN

Name _____ Results also sent to _____

Phone: _____ Fax: _____

BILLING INFORMATION

Bill: Insurance Referring Institution Patient Other Party

Ins.Co./Instit. _____ Name _____

Please attach appropriate billing information if available

INDICATIONS FOR TESTING (ICD9 Codes are in parentheses)

- | | |
|---|---|
| <input type="checkbox"/> Acute lymphocytic leukemia (ALL) (204.00) | <input type="checkbox"/> Lymphoproliferative disorder (238.79) |
| <input type="checkbox"/> Acute myelocytic leukemia (AML)(205.00) | <input type="checkbox"/> Monoclonal Gammopathy (273.1) |
| <input type="checkbox"/> Acute promyelocytic leukemia (APL) (205.00) | <input type="checkbox"/> Plasma Cell Dyscrasia (273.9) |
| <input type="checkbox"/> Anemia (suspected leukemia) (285.9, 208.80) | <input type="checkbox"/> Multiple Myeloma (203.00) |
| <input type="checkbox"/> Burkitt's Lymphoma (200.20) | <input type="checkbox"/> Myelodysplastic Syndrome (288.75) |
| <input type="checkbox"/> Chronic myelogenous leukemia (CML) (205.10) | <input type="checkbox"/> Myelofibrosis (suspected leukemia) (289.83, 208.80) |
| <input type="checkbox"/> Chronic lymphocytic leukemia (CLL) (204.10) | <input type="checkbox"/> Myeloma (203.0) |
| <input type="checkbox"/> Hodgkin's Lymphoma (201.9) | <input type="checkbox"/> Myeloproliferative Syndrome (238.79) |
| <input type="checkbox"/> Non-Hodgkin's Lymphoma (202.80) | <input type="checkbox"/> Neutropenia (suspected leukemia) (288.0, 208.80) |
| <input type="checkbox"/> Lymphoma (202.80) | <input type="checkbox"/> Pancytopenia (suspected leukemia) (284.0, 208.80) |
| <input type="checkbox"/> Leukemia (known or suspected) (208.80) | <input type="checkbox"/> Polycythemia vera (suspected leukemia) (238.4, 208.80) |
| <input type="checkbox"/> Leukopenia (suspected leukemia) (288.0, 208.80) | <input type="checkbox"/> Thrombocytopenia (suspected leukemia) (287.5, 208.80) |
| <input type="checkbox"/> Leukocytosis (suspected leukemia) (288.8, 208.80) | <input type="checkbox"/> Thrombocytosis (suspected leukemia) (289.9, 208.80) |
| <input type="checkbox"/> Lymphocytosis (suspected leukemia) (288.8, 208.80) | <input type="checkbox"/> Other _____ |

TEST REQUESTED

Cytogenetics: (Requires heparinized sample or Green Top tube)		
<input type="checkbox"/> Chromosome Analysis		
FISH: (Requires heparinized sample or Green Top tube) ★ <input type="checkbox"/> FISH for previous abnormality ★		
<input type="checkbox"/> t(8;21) (AML, ETO/AML1)	<input type="checkbox"/> Myeloma Panel (including the following)	<input type="checkbox"/> MDS Panel (including the following)
<input type="checkbox"/> t(9;22) (CML and ALL, BCR/ABL)	<input type="checkbox"/> t(4;14) (FGFR3/IGH)	<input type="checkbox"/> -5/5q- <input type="checkbox"/> 13q-
<input type="checkbox"/> t(15;17) (APL, PML/RARα)	<input type="checkbox"/> -13/13q- (D13S319, LAMP1)	<input type="checkbox"/> -7/7q- <input type="checkbox"/> 20q-
<input type="checkbox"/> Inv 16/t(16;16)/del16 (AML-M ₄ , CBFB)	<input type="checkbox"/> 17p- (p53)	<input type="checkbox"/> +8
<input type="checkbox"/> 9p-, +9 (p16 probe)	<input type="checkbox"/> Lymphoma Panel (including the following)	<input type="checkbox"/> CLL Panel (6q-,11q-,13/13q-,17p-,+12)
<input type="checkbox"/> Pediatric Pre-B ALL Panel (COG)	<input type="checkbox"/> t(14;18) (BCL2/IGH)	<input type="checkbox"/> CHIC2 deletion (FIP1L1-PDGFRα)
<input type="checkbox"/> MLL involvement (11q23)	<input type="checkbox"/> t(11;14) (CCND1/IGH)	
<input type="checkbox"/> t(12;21) (Pediatric ALL, TEL/AML1)	<input type="checkbox"/> t(8;14) (c-MYC/IGH)	<input type="checkbox"/> Other _____
Molecular: (Requires Purple Top tube--EDTA)		
<input type="checkbox"/> Factor V Leiden (ICD9=286.3)	<input type="checkbox"/> Factor V HR2 (done if Leiden positive)	<input type="checkbox"/> Quantitative PCR for PML/RARA
<input type="checkbox"/> Prothrombin (ICD9=286.3)	<input type="checkbox"/> Hereditary Hemochromatosis	<input type="checkbox"/> Quantitative PCR for BCR/ABL
<input type="checkbox"/> MTHFR (ICD9=286.3)	<input type="checkbox"/> NPM1 Mutation Analysis	<input type="checkbox"/> JAK2 Mutation Analysis
<input type="checkbox"/> T-Cell Gene Rearrangements	<input type="checkbox"/> DNA Extract and Store	<input type="checkbox"/> FLT3 Analysis
		<input type="checkbox"/> Other _____
Pre-Transplant: - - CHIMERISM STUDY - - Post-Transplant:		
<input type="checkbox"/> Donor (Requires Purple Top tube—EDTA)	<input type="checkbox"/> FISH (X/Y Sex Chromosomes) (Requires Green Top tube--NaHep)	
<input type="checkbox"/> Recipient Blood (Requires Purple Top tube--EDTA)	OR -	
<input type="checkbox"/> Recipient Buccal Swab	<input type="checkbox"/> DNA (Microsatellite Analysis) (Requires Purple Top tube--EDTA)	

CENTER FOR HUMAN GENETICS LABORATORY
CYTOGENETICS AND MOLECULAR GENETICS REQUISITION

SPECIMEN INFORMATION

*Collection date _____ Institution _____ Phlebotomist _____

Peripheral Blood Cord Blood from (Circle) Liveborn / Stillborn/Demise/Ongoing Pregnancy DNA Tissue Other _____

PATIENT INFORMATION

Name (Last, First): _____

DOB: ____/____/____ Medical Record Number: _____

Address: _____ Phone: _____

City, State, Zip: _____ SS #: ____/____/____

Sex: Male Female Ambiguous Unknown **Pregnant:** Yes / No Gestational age: _____

Ethnicity: Caucasian (N. and S. European) Ashkenazi Jewish Hispanic Asian Afr. American Other: _____

REFERRING PHYSICIAN

Name _____ Phone: _____ Fax: _____

Name & Phone of person completing requisition: _____ Informed consent obtained (if appropriate)

BILLING INFORMATION

Bill: * Insurance Referring Institution Check enclosed for \$ _____

* If Insurance will be billed, please attach a copy of current insurance card (front and back), which should include:
Patient Name, Insurance Provider address & phone #, Policy #, Group #, Relationship to Patient

TEST INDICATION

ICD9 CODES (Required): _____

CYTOGENETICS TESTS

(5mL in green top Sodium-Heparin Tube)

CHROMOSOME ANALYSIS, HIGH RESOLUTION
(also known as karyotype or cytogenetics)

With five-cell, lower resolution preliminary result called within 48-72 hours
(extra charge, done for newborns only)

With extra 10 counts for sex-chromosome mosaicism
(used with Q. 45,X or Q. 47,XXY)

FISH with selected probe(s)

Probe: _____

Other _____

MOLECULAR TESTS

(5mL in purple top EDTA Tube)

- Hearing Loss Panel
 - Aminoglycoside Induced Deafness
 - Connexin 26 (sequencing)
 - Connexin 30 (deletion testing)
- Thrombophilia Panel
 - Factor V Leiden Mutation
 - Reflex to Factor V HR2 if Needed
 - Prothrombin Mutation Analysis
 - MTHFR C677T Mutation Analysis

Hereditary Hemochromatosis

Uniparental Disomy, Chrom # _____

Y Deletion for Male Infertility

Zygosity

Cystic Fibrosis Carrier Screen
(41 Mutations)

DNA Extract/Store

Other _____

COMBINED PANELS

(5mL in purple top EDTA & 5 mL in green top)

Fragile X Syndrome Analysis
(Both tests done unless otherwise specified)

- Molecular Analysis ONLY
- Chromosome Analysis ONLY

Prader-Willi Syndrome Analysis
(Both tests done unless otherwise specified)

- Methylation Analysis ONLY
- Chromosome Analysis + FISH ONLY

Angelman Syndrome Analysis
(Both tests done unless otherwise specified)

- Methylation Analysis ONLY
- Chromosome Analysis + FISH ONLY

CENTER FOR HUMAN GENETICS LABORATORY
PRENATAL TESTING REQUISITION

www.chglab.com

SPECIMEN INFORMATION

*Collection date _____ Institution _____ Time of Collection _____
 Amniotic Fluid ____cc's (1st cc's separated Y/N) CVS Cord Blood Products of Conception (specify) _____
 Peripheral Blood Tissue (specify) _____ Other _____

PATIENT INFORMATION

Name (Last, First): _____
DOB: ____/____/____ **Medical Record Number:** _____
 Address: _____ Phone: _____
 City, State, Zip: _____ SS #: ____/____/____
Pregnant: Yes / No **Gestational age:** _____ **Gender by U/S:** Male / Female / Unknown **Twins? Y/N**
Ethnicity: Caucasian (NW European SW European) Ashkenazi Jewish Other Jewish Hispanic Asian Afr. American
 Native American Other: _____

REFERRING PHYSICIAN

Name _____ Phone: _____ Fax: _____
 Name & Phone of person completing requisition: _____ **Informed consent obtained (if appropriate)**

BILLING INFORMATION

Bill: * Insurance Referring Institution Patient Other Party

* If Insurance will be billed, please attach a copy of current insurance card (front and back), which should include:
 Patient Name, Insurance Provider address & phone #, Policy #, Group #, Relationship to Patient

TEST INDICATION

ICD9 CODES (Required): _____

Cyto/Molecular Tests

-- CYTOGENETICS

- CHROMOSOME ANALYSIS/KARYOTYPE
(Amniotic Fluid AFP done automatically unless otherwise specified) No AFP
- Aneuvysion FISH (prenatal screen for X, Y, 13, 18, 21)
- FISH with Selected Probe (Specify) _____
- Save Cells Temporarily (Reason) _____

-- MOLECULAR (purple top EDTA required)

- Uniparental Disomy, Chrom # _____ (purple top tube)
- Thrombophilia Panel (purple top tube)
 - Factor V Leiden Mutation with reflex HR2
 - Prothrombin Mutation Analysis
 - MTHFR C677T Mutation Analysis
- Other _____

-- Additional tests on Amnio/CVS to be sent out:

- CF CMV Toxoplasmosis Parvovirus
- Herpes I/II RhD genotyping Sickle Cell

Ethnicity-Based Screening

(5mL in purple top EDTA Tube)

- Cystic Fibrosis Carrier Screen (41 Mutations)
- Ashkenazi Jewish Panel (CF NOT included)
 - Bloom syndrome
 - Canavan Disease
 - Familial Dysautonomia
 - Tay-Sachs
 - Fanconi Anemia Type C
 - Gaucher Disease Type 1
 - Mucopolidosis Type IV
 - Niemann-Pick Type A & B

Complete Required Information:

____ Patient/Couple is Pregnant
 ____ Family history of Disorder and/or Mutation
Specify _____
 ____ Abnormal Ultrasound _____
 ____ Absence of Vas Deferens
 ____ Other Infertility

Maternal Serum Screening

(Red or Yellow Gel Separator Tube Required)

- Quad Check (AFP/UE3/hCG/Inhibin A)
 - Triple Check (AFP/UE3/hCG)
 - AFP Only
 - Repeat Test at This Laboratory
- Complete Required Information:**
 Patient Current Weight _____
 Insulin-Dependent Diabetic: ____ Yes ____ No
 Twin Pregnancy: ____ Yes ____ No ____ Unknown
 Previous Child with ONTD: ____ Yes ____ No
 Family History of ONTD: ____ Yes ____ No

Gestational Age Dating:

Last Menstrual Period _____
 Date of Ultrasound _____
 Gestational Age on that date _____
 EDC (By US Dating Only) _____
 By Physical Exam: _____ weeks
 Date of Exam _____

Center for Human Genetics Laboratory

Urovysion FISH Test Requisition

University Hospitals Case Medical Center
10524 Euclid Avenue, 6th Floor
Cleveland, OH 44106 Tel: (216) 983-1134 Fax: (216) 983-1144

SPECIMEN INFORMATION

Urine Other (specify) _____ Date of collection _____

PATIENT IDENTIFICATION

PATIENT NAME _____ SEX Male Female (circle one)

DATE OF BIRTH _____ ADDRESS #1 _____

SOCIAL SECURITY # _____ ADDRESS #2 _____

MEDICAL RECORD NUMBER _____ PHONE _____

REASON FOR REFERRAL

_____ HEMATURIA (ICD9 599.70) _____ OTHER (SPECIFY)

_____ BLADDER CANCER (ICD9 188.9, 239.4) Diagnostic or recurrence? (circle one)

REFERRING PHYSICIAN

Name _____ Phone _____ Fax _____

Address _____

Results also sent to: _____

REFERRING INSTITUTION

Name _____

BILLING INFORMATION

Bill: Referring Institution

Patient directly

Patient Insurance (Please attach insurance information)

Center for Human Genetics Laboratory, Specimen Requirements

Test/Syndrome	Cytogenetics	Molecular	FISH	Sendout	Specimen Type	Media	Infant	Child	Adult	Comments
Ambiguous Genitalia	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	Possible additional cells; possible FISH
Amenorrhea	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	Possible additional cells; possible FISH
Aminoglycoside Induced Hearing Loss		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Maternal Inheritance
Anemia (Chromosomes)	X				Bone Marrow	Heparinized	2-3	2-3	2-3	Insufficient reason for referral
Anemia (Chromosomes)	X				Peripheral Blood	Heparinized	3-5	3-5	5-7	Insufficient reason for referral
Angelman Syndrome (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	Requires phenotype
Angelman Syndrome (FISH)			X		Peripheral Blood	Heparinized	2-3	3-5	5-7	Requires phenotype
Angelman Syndrome (DNA Methylation)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Requires phenotype
Angelman Syndrome (UPD)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Requires phenotype
APC (I307K)		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
Ashkenazi Jewish Carrier Screening (Full Panel)					Peripheral Blood	EDTA	2-3	3-5	5-7	
Autism (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	Possible DNA for Fragile X
Autism (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Requires concurrent chromosome study
Autistic Spectrum Disorder (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	
Autistic Spectrum Disorder (FISH)			X		Peripheral Blood	Heparinized	2-3	3-5	5-7	FISH for dup (15)
Bcr/abl (FISH)			X		Peripheral Blood	Heparinized	2-3	5-7	5-7	May include chromosomes
Bcr/abl (CML) (FISH)			X		Bone Marrow	Heparinized	2-3	5-7	5-7	May include chromosomes
Bcr/abl (PCR) (Major / Minor)				X	Peripheral Blood	Heparinized	2-3	5-7	5-7	Call Lab
Bcr/abl (PCR) (Major / Minor)				X	Peripheral Blood	EDTA / ACD-A	2-3	5-7	5-7	Call Lab
Bcr/abl [t(9;22)] (Chromosomes)	X				Bone Marrow	Heparinized	2-3	3-5	3-5	May include FISH
Bcr/abl [t(9;22)] (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	May include FISH
Beckwith-Wiedemann (UPD 11)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Call Lab prior to sending sample
Bladder Cancer Screening (UroVysion™) (polysomy for chromosomes 3, 7, 17 and deletion of 9p21)			X		Urine	Fixative	>33	>33	>33	Please collect specimen using urine cytology collection kit (call Lab for more information)
Bloom Syndrome					Peripheral Blood	EDTA	2-3	3-5	5-7	
Canavan Disease					Peripheral Blood	EDTA	2-3	3-5	5-7	
Chimerism Post-transplant		X			Peripheral Blood	EDTA	2-3	2-3	2-3	Pretransplant required
Chimerism Post-transplant		X			Bone Marrow	EDTA	2-3	2-3	2-3	Pretransplant required
Chimerism Pre-transplant		X			Peripheral Blood	EDTA	2-3	2-3	2-3	Pretransplant required
Chimerism Pre-transplant		X			Bone Marrow	EDTA	2-3	2-3	2-3	Pretransplant required
Chromosome Analysis	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	
Cleft Lip	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	Possible FISH #22
Connexin 26		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Cri-du-chat	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	

Center for Human Genetics Laboratory, Specimen Requirements

Test/Syndrome	Cytogenetics	Molecular	FISH	Sendout	Specimen Type	Media	Infant	Child	Adult	Comments
Cystic Fibrosis Screening		X			Peripheral Blood	EDTA	N/A	N/A	2-3	
Deep Vein Thrombosis		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Developmental Delay (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Developmental Delay (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Concurrent chromosome study highly recommended
DiGeorge Syndrome	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH #22
DNA Extraction & Storage		X			Peripheral Blood	EDTA	2-3	5-7	5-7	Must have indications
Down Syndrome	X				Peripheral Blood	Heparinized	2-3	3-5	3-5	
Edwards Syndrome	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Factor V HR2		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Only for Factor V Leiden Heterozygotes
Factor V Leiden Gene Mutation		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Failure to Thrive	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	
Familial Dysautonomia					Peripheral Blood	EDTA	2-3	3-5	5-7	
Fanconi Anemia Group C					Peripheral Blood	EDTA	2-3	3-5	5-7	
FISH (not including telomeric probes)			X		Peripheral Blood	Heparinized	2-3	2-3	2-3	Must be specific
Fragile X (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	
Fragile X (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Chromosomes recommended
FXTAS (Fragile X tremor/ataxia syndrome)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Gaucher Disease					Peripheral Blood	EDTA	2-3	3-5	5-7	
Gonadal Dysgenesis	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Habitual Aborter	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	
Habitual Aborter	X				Tissue	Sterile Saline				Refrigerated
Habitual Aborter (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Hearing Loss (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	
Hearing Loss (DNA) (Connexin 26 Analysis)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Hemochromatosis		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Hypotonia (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH #15
Hypotonia (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Infertility (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Infertility (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Iron Overload		X			Peripheral Blood	EDTA	2-3	3-5	5-7	For hereditary hemochromatosis
Karyotype	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Klinefelter	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	
LCHAD Deficiency		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	

Center for Human Genetics Laboratory, Specimen Requirements

Test/Syndrome	Cytogenetics	Molecular	FISH	Sendout	Specimen Type	Media	Infant	Child	Adult	Comments
Leukemia	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	
Lymphoblast Line (Cell Line Build Up)	(x)	(x)	(x)	(x)	Peripheral Blood	EDTA	2-3	3-5	5-7	CLBU then used for specific, designated test
MCAD		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
MELAS		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
Mental Retardation (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Mental Retardation (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Concurrent chromosome study highly recommended
MERRF		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
Methylation		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Requires Phenotype
Microdeletion Study	X		X		Peripheral Blood	Heparinized	2-3	3-5	3-5	
Miller-Dieker Syndrome	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH / Possible UPD
Mitochondrial Panel		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
Monosomy Study	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	Possible FISH
MTHFR Gene Mutation (C677T)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Mucopolidosis Type IV					Peripheral Blood	EDTA	2-3	3-5	5-7	
Multiple Congenital Anomalies	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	
Multiple Pregnancy Losses (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Multiple Pregnancy Losses (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
NARP (Leigh) Syndrome		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
Niemann-Pick Disease					Peripheral Blood	EDTA	2-3	3-5	5-7	
Transient Neonatal Diabetes (UPD 6)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Patau Syndrome	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Pellet Buildup	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
PML (rar alpha)	X		X		Peripheral Blood	Heparinized	2-3	3-5	3-5	Possible FISH #15
Prader Willi Syndrome (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Requires Phenotype
Prader Willi Syndrome (FISH)			X		Peripheral Blood	Heparinized	2-3	3-5	5-7	Requires Phenotype
Prader Willi Syndrome (DNA Methylation)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Requires Phenotype
Prader Willi Syndrome (UPD)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Premature Ovarian Failure		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
PRION Mutation Analysis		X			Peripheral Blood	DNA				Contact lab first
Prothrombin Gene Mutation (G20210A)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Pulmonary Embolisim		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Retinoblastoma	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH #13
Rubenstein Taybi	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH Rubenstein region

Center for Human Genetics Laboratory, Specimen Requirements

Test/Syndrome	Cytogenetics	Molecular	FISH	Sendout	Specimen Type	Media	Infant	Child	Adult	Comments
Russel Silver (UPD 7)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Sex Chromosome abnormality	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH Sex chromosomes
Short Stature	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Spongiform Encephalopathy		X			Peripheral Blood	DNA				Contact lab first
SRY (Y microdeletion)			X		Peripheral Blood	Heparinized	2-3	3-5	3-5	
t(9;22) (CML)			X		Peripheral Blood	Heparinized	2-3	5-7	5-7	
Tay-Sachs Disease					Peripheral Blood	EDTA	2-3	3-5	5-7	
Thrombophilia		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Translocation	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	
Translocation Carrier	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	
Translocation Carrier (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	UPD for Chromosomes 6,7,14,15
Trisomy	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	
Turner Syndrome	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	
Unbalanced Translocation Carrier	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	
Uniparental Disomy		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Call Lab prior to sending sample if chr. 11 analysis
Uniparental Disomy (Amnio)		X			Cultured Cells					2 T-25 flasks at 75% confluency
Uniparental Disomy (CVS)					Cultured Cells					2 T-25 flasks at 75% confluency
UPD (14)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Rob. Translocations & Isochromosomes involving 14
Velocardiofacial Syndrome	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH #22, #10
Venous Thrombosis		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Williams Syndrome	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH #7
Wolf-Hirschorn (4p deletion)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH #4p
X Inactivation		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
Y Deletion		X			Peripheral Blood	EDTA	2-3	3-5	5-7	

NOTE A Minimum of 2 ml is required for any Molecular Testing

X* Indicates Temporary Sendout because of low volumes of requested test

For more information, please contact the Lab:

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