
GENETICS
ASSOCIATES
INCORPORATED

GENETICS ASSOCIATES, Inc.

1916 Patterson Street, Suite 400

Nashville, TN 37203

Business Hours

Monday through Friday

7:00 a.m. to 6:00 p.m. (CST)

Saturday

8:00 a.m. to 2:00 p.m. (CST)

*After hours, calls are answered by a licensed technologist

Office Phone: 615-327-4532

Office Fax: 615-327-0464

Toll Free: 800-331-4363

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History of Genetics Associates

Genetics Associates was established in 1990, with the goal of providing high quality, efficient, economical, and state of the art cytogenetic diagnostic services. Our owners and directors bring numerous years of experience to GAI and work side by side with their team of cytogenetic technologists and support staff. GAI believes in building relationships by paying close attention to the needs of our clients. Our strengths include providing results in a timely fashion, a highly-skilled, experienced staff and a reputation for quality service.

OUR MISSION

To be the cytogenetic laboratory and information resource of choice for physicians who demand accurate, timely, and state of the art cytogenetic diagnostic services for their patients.

To grow and expand our services while maintaining the excellence in quality expected by our physicians, patients, and staff.

To provide our staff with environment that promotes growth, education, fairness, security, and a sense of well-being through leadership, discipline, and belief in our fellow employees.

LICENSURE AND CERTIFICATION

College of American Pathologists License # 3932401

Clinical Laboratory Improvements Amendments License # 44D0688266

State of TN Board of Health License # 0000002299

State of CA Dept. of Public Health License # COS 00800301

State of FL Agency for Health Care Administration License # 800011959

Maryland Dept. of Health & Mental Hygiene Permit # 1353
Office of Health Care Quality -Medical Laboratory

Pennsylvania Clinical Laboratory Permit Permit # 29942A

Rhode Island & Prov. Plantations Dept. of Health License # LCO 00272

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SPECIMEN COLLECTION

SPECIMEN COLLECTION AND HANDLING GUIDE

Proper specimen collection and handling are essential parts of obtaining an accurate and timely laboratory test result. The purpose of this guide is to describe the proper procedure for specimen collection, labeling, storage, and shipping.

Specimen Identification and Labeling Requirements

Labeling Requirements:

Label all primary specimen containers with at least two patient-specific identifiers.

- Acceptable identifiers include but are not limited to: patient name, date of birth, hospital number, social security number, requisition number, accession number, or unique random number. A hospital room number is not an appropriate patient identifier.
- Label all specimens in the presence of the patient
- Positive identification is the responsibility of the person collecting the sample

Specimen Identification:

Submit a completed order or requisition form with all specimens. Completed orders or requisition forms should contain the following information:

- Full Name of Patient

- Secondary Unique Identifier
- Date of Birth
- Sex of Patient
- Referring Physician
- Facility Name and Address
- Date and Time of Specimen Collection
- Specimen Type
- Referring Diagnosis
- ICD 10 Code
- Appropriate Related Patient History
- Test Requested

Patient Preparation

Many tests require proper patient preparation before collection to ensure a quality specimen for testing. Refer to collection facility's procedures for patient preparation requirements.

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SPECIMEN COLLECTION AND HANDLING GUIDE

Specimen Collection and Storage Requirements for Chromosome Analysis, FISH, ICP, and Microarray

Amniotic Fluid:

- Volume: 10-20 ml; discard the first ml of fluid or use for other testing
- Container: Sterile 15 ml centrifuge tube or container
- Storage Conditions: Room temperature, 20 to 22°C (68 - 72°F) or refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)

Bone Core:

- Volume: N/A
- Container: Sodium Heparin vacutainer or a sterile tube containing sterile transport media

- Invert tube 4-8 times to prevent formation of clots
- Storage Conditions: Room temperature, 20 to 22°C (68 - 72°F) or refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)

Bone Marrow:

- **Adults**
  - Volume: 2 – 5 ml of bone marrow; from the first or at least the second tap
  - Container: Sodium Heparin vacutainer
    - **DO NOT USE LITHIUM HEPARIN**
    - Invert tube 4 – 8 time to prevent clots
  - Storage Conditions: Room temperature, 20 to 22°C (68 - 72°F)
    - **DO NOT FREEZE SPECIMEN**
- **Children**
  - Volume: 1 – 5 ml of bone marrow; from the first or at least the second tap
  - Container: Sodium Heparin vacutainer
    - **DO NOT USE LITHIUM HEPARIN**
    - Invert tube 4 – 8 time to prevent clots
  - Storage Conditions: Room temperature, 20 to 22°C (68 - 72°F)
    - **DO NOT FREEZE SPECIMEN**

Buccal Swab:

- Volume: Entire Swab (2—if performing Microarray testing)
- Container: Original collection tube, dry.
- Storage Conditions: Room temperature, 20 to 22°C (68 - 72°F) or refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)

GENETICS ASSOCIATES INCORPORATED **DO NOT FREEZE SPECIMEN**

Chorionic Villi:

• Volume: 10-20 mg of chorionic villi
• Container: 15 ml sterile centrifuge tube containing sterile transport media
  - Sterile transport media provided by GAI upon request, sterile media such as RPMI, or sterile saline solution may be used
• Storage Conditions: Room temperature, 20 -22°C (38 - 72°F) or refrigerated temperature, 2 - 8°C (35.6 – 46.4°F) o Do Not Freeze
• For locations in the Nashville area and with advanced notice, a cytogenetic technologist may be provided to verify that an adequate sample has been obtained.

Fine Needle Aspirate:

• Volume: Entire aspirate
• Container: 15 ml sterile centrifuge tube containing sterile transport media
    - Sterile transport media provided by GAI upon request, sterile media such as RPMI, or sterile saline solution may be used
    - **DO NOT USE FORMALIN**
• Storage Conditions: Room temperature, 20 -22°C (38 - 72°F) or refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)
    - **DO NOT FREEZE SPECIMEN**

Lymph Node:

• Volume: Entire lymph node
• Container: Sterile container containing sterile transport media
    - Sterile transport media, provided by GAI upon request, sterile media such as RPMI, or sterile saline solution may be used
    - **DO NOT USE FORMALIN**
• Rinse lymph nodes collected in non-sterile conditions with sterile balanced salts solution, Ringer's lactate, or sterile saline.
• Storage Conditions: Room temperature, 20 -22°C (68 - 72°F) or refrigerated temperature, 2 -8°C (35.6 – 46.4°F)
    - **DO NOT FREEZE SPECIMEN**

Masses / Tumors:

• Volume: Entire mass / tumor
• Container: Sterile specimen cup containing sterile transport media
    - Sterile transport media provided by GAI upon request, sterile media such as RPMI, or balanced salt solution may be used
    - **DO NOT USE FORMALIN**
• Storage Conditions: Room temperature, 20 -22°C (68 - 72°F) or refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)
    - **DO NOT FREEZE SPECIMEN**

Peritoneal Fluid:

• Volume: 15 -50 ml peritoneal fluid
• Container: 50 ml sterile centrifuge tube or specimen cup
• Storage Conditions: Room temperature, 20 -22°C (68 -72°F) or refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)
    - **DO NOT FREEZE SPECIMEN**

Peripheral Blood:

• **Children – Adults (8 days and up)**
    - Volume: 2 – 5 ml peripheral blood
    - Container: Sodium Heparin vacutainer; Do Not Use Lithium
        - Invert tube 4 – 8 time to prevent clots

o Storage Conditions: Room temperature, 20 – 22°C (68 - 72°F) or refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)
- **DO NOT FREEZE SPECIMEN**

• **Newborn (0 – 7 days)**
o Volume: 1 – 2 ml peripheral blood
o Container: Sodium Heparin vacutainer
- **DO NOT USE LITHIUM HEPARIN**
- Invert tube 4 – 8 times to prevent clots

o Storage Conditions: Room temperature, 20 – 22°C (68 - 72°F) or refrigerated temperature, 2 – 8°C (35.6 – 46.4°F)
- **DO NOT FREEZE SPECIMEN**

• **PUBS (Percutaneous Umbilical Blood Specimen/Cord Blood)**
o Volume: 1 – 2 ml PUBS or cord blood
o Container: Sodium Heparin vacutainer
- **DO NOT USE LITHIUM HEPARIN**
- Invert tube 4 – 8 times to prevent clots

o Storage Conditions: Room temperature, 20 - 22°C (68 - 72°F) or refreigerated temperature, 2 – 8°C (35.6 – 46.4°F)
- **DO NOT FREEZE SPECIMEN**

Pleural Fluid:

• Volume: 15 -50 ml pleural fluid
• Container: Sterile centrifuge tube or specimen cup
• Storage Conditions: Room temperature, 20 - 22°C (68 - 72°F) or refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)
o **DO NOT FREEZE SPECIMEN**

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Products of Conception:

• Volume: ≥100 mg of appropriate tissue; If volume is inadequate, all tests requested may not be performed.
o Specimen collected should include, by order of preference, one or more of the following: villi, placenta or placental membrane, or recognizable fetal parts.
o **Stillborn:** Placenta containing chorionic villi is the preferred tissue; include other tissue as well
o **Stillborn or infant autopsy:** Preferred tissues in descending order are: lung, kidney, thymus, skin.
• Rinse tissue collected in non-sterile conditions with sterile balanced salt solution, Ringer's lactate, or sterile saline.
• Container: Sterile container containing sterile transport media
o Sterile transport media, provided by GAI upon request, sterile media such as RPMI, or sterile saline solution may be used
o Carefully tighten the lid of container to prevent leakage
o **DO NOT USE FORMALIN**
• Storage Conditions: Room temperature, 20 -22°C (68 - 72°C) or refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)
o **DO NOT FREEZE SPECIMEN**

Slides (Paraffin Embedded Tissue):

- Volume: Submit 2 slides per probe requested
    - o Cut specimen 3 – 4 µ thick.
    - o Use positively charged slides.
- Submit H & E slides marked with the area of interest.
- Storage Conditions: Room temperature, 20 – 22°C (68 -72°F)

Tissue, Solid (Constitutional):

- Volume: 3mm3 tissue biopsy
- Container: Sterile specimen cup containing sterile transport media
    - o Sterile transport media provided by GAI upon request, sterile media such as RPMI, or balanced salt solution may be used
    - o **DO NOT USE FORMALIN**
- Storage Conditions: Room temperature, 20 - 22°C (68 - 72°F) or refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)
    - o **DO NOT FREEZE SPECIMEN**

Urine:

- Volume: ≥33 ml of urine
- Container: Sterile container
    - o Carefully tighten the lid of the container to prevent leakage
- orage Conditions: Refrigerated temperature, 2 - 8°C (35.6 – 46.4°F
    - o **DO NOT FREEZE SPECIMEN**

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Specimen Collection and Storage Requirements for Molecular Testing (DNA Based Testing)

IgVH Hypermutation
JAK2
B Cell & T Cell Clonality
NGS
Thrombophilia
Breast Milk Identity

Bone Marrow:

- Volume: 1.5 – 5ml bone marrow; from the first or at least second tap
- Container: EDTA (purple top) vacutainer
    - o Sodium Heparin tube can be used but is not preferred
    - o Invert tube 4 – 8 times to prevent clots
- Storage Conditions: Refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)
    - o **DO NOT FREEZE SPECIMEN**
- Specimen must be received in the lab within 72 hours of draw

Breast Milk:

- Volume: 5-10 mL
- Container: Sterile DNase/RNase free 15 mL centrifuge tube
- Storage Conditions: Refrigerated temperature, 2-8°C (35.6-46.6°F) or frozen temperature, -25 to -15°C (-13 to 5°F)
    - If specimen is frozen and thawed prior to preparing aliquot for GAI, do not re-freeze specimen, send at refrigerated temperature, 2-8°C (35.6-46.6°F)

Buccal Swab:

- Volume: Entire Swab (1)
- Container: Original collection tube, dry.
- Storage Conditions: Room temperature, 20-22°C (68-72°F) or refrigerated temperature, 2-8°C (35.6-46.6°F)
    - **DO NOT FREEZE SPECIMEN**

Peripheral Blood:

- Volume: 2 – 5 ml peripheral blood
- Container: EDTA (purple top) vacutainer
    - Sodium Heparin tube can be used but is not preferred
    - Invert tube 4 – 8 times to prevent clots
- rage Conditions: Refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)
    - **DO NOT FREEZE SPECIMEN**
- **cimen must be received in the lab within 72 hours of draw**

Specimen Collection and Storage Requirements for Molecular Testing (RNA Based Testing)

BCR/ABL1 p210
BCR/ABL 1 p190

Bone Marrow:

- Volume: 1.5 – 5ml bone marrow; from the first or at least second tap
- Container: EDTA (purple top) vacutainer
    - Sodium Heparin tube can be used but is not preferred
    - Invert tube 4 – 8 times to prevent clots
- Storage Conditions: Refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)
    - **DO NOT FREEZE SPECIMEN**
- **Specimen must be received in the lab within 72 hours of draw**

Peripheral Blood:

- Volume: 2 – 5 ml peripheral blood
- Container: EDTA (purple top) vacutainer
    - Sodium Heparin tube can be used but is not preferred
    - Invert tube 4 – 8 times to prevent clots
- Storage Conditions: Refrigerated temperature, 2 - 8°C (35.6 – 46.4°F)

- **o DO NOT FREEZE SPECIMEN**
- **Specimen must be received in the lab within 72 hours of draw**

Fixed Pellets (Bone Marrow or Peripheral Blood):

- Volume: Cell pellet fixed in Methanol: Acetic Acid (3:1); pellet must be visible
  - o **Pellet must not be older than 1 week**
- Container: Sterile centrifuge tube
- Storage Conditions: Refrigerated temperature, 2 -8°C (35.6 – 46.4°F)
  - **o DO NOT FREEZE SPECIMEN**

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TURN AROUND TIME

Leukemic Bloods	3 Days
Bone Marrows	3 Days
Lymph Nodes	3 Days
Urovysion®	3 Days
Peripheral Bloods	4 Days
Newborn Bloods	4 Days
• 24 – 48 hour prelims	
Paraffin-embedded Slides	4 Days
PCR	4 Days
ICP	5 Days
CVS	7 Days
POC	7 Days
Amniotic Fluid	7 Days

Microarray	7 Days
Mass	10 Days
Tissue	10 Days

REQUISITION INSTRUCTIONS

***Current requisitions may be printed at www.geneticsassociates.com**

ADDITIONAL FORMS: TEST AUTHORIZATION

ADDITIONAL FORMS: PATIENT DATA VERIFICATION

ONCOLOGY TEST	CPT CODES
Chromosome Analysis –Bone Marrow	88237 x 2, 88264, 88280 x 3, 88291
Chromosome Analysis –Bone Marrow Core	88237 x 2, 88264, 88280 x 3, 88291
Chromosome Analysis –Leukemic Blood	88237 x 2, 88264, 88280 x 3, 88291
Chromosome Analysis –Mass/Solid Tumor	88233 x 2, 88264, 88280 x 3, 88291
Fluorescence in Situ Hybridization—Single 88291	88271 (per probe), 88275 (per probe),
Fluorescence in Situ Hybridization—Multi 88291	88271 x 2 (per probe), 88275 (per probe),

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Required	

FISHnet / iFISH (enrichment only)	88112 (FISH codes will also apply—see above)

Microarray (SNP)	81229
PCR	
JAK2	81270
BCR/ABL p210 BKPT	81206
BCR/ABL P190 BKPT	81207

*Please note, codes are report dependent.

CONSTITUTIONAL TEST	CPT CODES
Chromosome Analysis –Amniotic Fluid	88235 X 2, 88267, 88261, 88280, 88291
Chromosome Analysis –CVS	88235 X 2, 88267, 88261, 88280, 88172, 88291
Chromosome Analysis –Peripheral Blood	88230 x 2, 88262, 88280, 88291
Chromosome Analysis –POC	88305, 88233x2, 88261, 88267, 88280
Chromosome Analysis –Tissue	88233 x 2, 88264, 88280 x 3, 88291
FISH (Microdeletion)	88271, 88273, 88291
Aneuvysion® FISH	88271 x 5, 88275, 88291
POC FISH	88271 x 8, 88275 X 2, 88291
ICP FISH	88271 X 24, 88275 x 2, 88291

Microarray (SNP)	81229

*Please note, codes are report dependent.

SHIPPING

SPECIMEN SHIPPING

• **Call Genetics Associates, Inc. at 615-327-4532 for pick up and additional information.**

• Specimen Shipping Kits and FedEx Shipping Bags
  o Specimen shipping kits are provided by GAI.
    o FedEx overnight shipment will be provided for all outlying areas.
    o **Mark "Saturday Delivery" box on the FedEx airbill when samples are shipped on Friday.**
    o **Samples shipped on Saturday by FedEx will not be delivered until the next business day .**

• Enclose completed requisition form with each specimen unless a electronic order has been sent.

• Enclose a refrigerated cold pack in the shipping box for overnight transportation for specimens requesting chromosome analysis, FISH, ICP, or microarray and avoid extreme temperatures to ensure specimen integrity.

• Enclose a frozen cool pack in the shipping box for overnight transportation for specimens requesting PCR testing and avoid temperature extremes to ensure specimen integrity.

• Store samples as stated in the Specimen Collection and Handling Guide for each specific specimen type until pickup by courier

Please note that Federal Express does not deliver on Sunday or holidays.

FEDERAL EXPRESS SUPPLIES

GAI provides shipping supplies for our clients, unless other arrangements are made. Clients may request the following supplies via the website at www.geneticsassociates.com or by calling 615-327-4532 or 1-800-331-GENE (4363).

COURIER SERVICES

GAI provides courier services to the Nashville Metropolitan area. A Better Courier Service

(ABC) couriers are utilized in a 100 mile radius of Nashville, TN.

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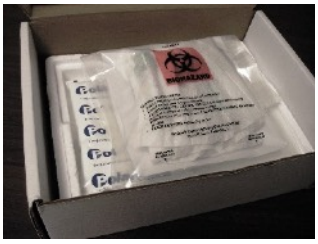
Specimen Packaging Guidelines

1. Use the Genetics Associates, Inc. (GAI) Cytogenetic Specimen box with the Styrofoam insert and lid to send samples.
2. To be compliant with federal regulations, the box must have a UN3373 diamond and should read "Biological Substance Category B". Do not place stickers or labels on the outside of the box or cover up these required logos.
3. **Always use** TWO SMALL OR ONE LARGE REFRIGERATED GEL PACKS**.** NOT frozen or room temperature,
4. Make sure the specimen collection tube/container is fully closed.
5. Place the specimen collection tube in one of the pockets of the included absorbent material (see image below).

6. Insert the tubes and absorbent material in the enclosed 95kpa specimen bag. Seal the bag by removing the tape and pressing down firmly.
   a. If you are sending 5-7ml vacutainers, one GAI specimen box will hold up to 10 tubes.
   b. If you are sending 8-10ml vacutainers, one GAI specimen box will hold up to 6 tubes.
   c. If you are sending 15ml centrifuge tubes, one GAI specimen box will hold up to 6 tubes.
   d. If you are sending 50ml conical tubes, one GAI specimen box will hold up to 3 tubes.
7. Different patient specimens may be included in the same box. Make sure each patient's paperwork is included in the same box as the specimen.
   Note: One patient's specimens are not to be divided between different boxes.

8. Patient test requisitions and corresponding insurance paperwork must be folded and inserted into the pouch at the back of the specimen bag. Fold the paperwork to ensure patient information is not visible.
9. Fold the specimen bag above the tubes and lay flat on the gel pack (see image below).



10. Close the box by inserting the tabs on top lid into side slots.
11. Place in the orange FedEx biological substance shipping pak.
    a. Seal the pak by removing the tape and pressing down firmly.
12. Apply the proper FedEx airbill marked from your facility to GAI.

l 1-800-331-GENE (4363) to inform GAI of the package's shipment and corresponding cking number.

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CONTACT INFORMATION

Executive Team

David Murray, Chief Executive Officer

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Administrative Team

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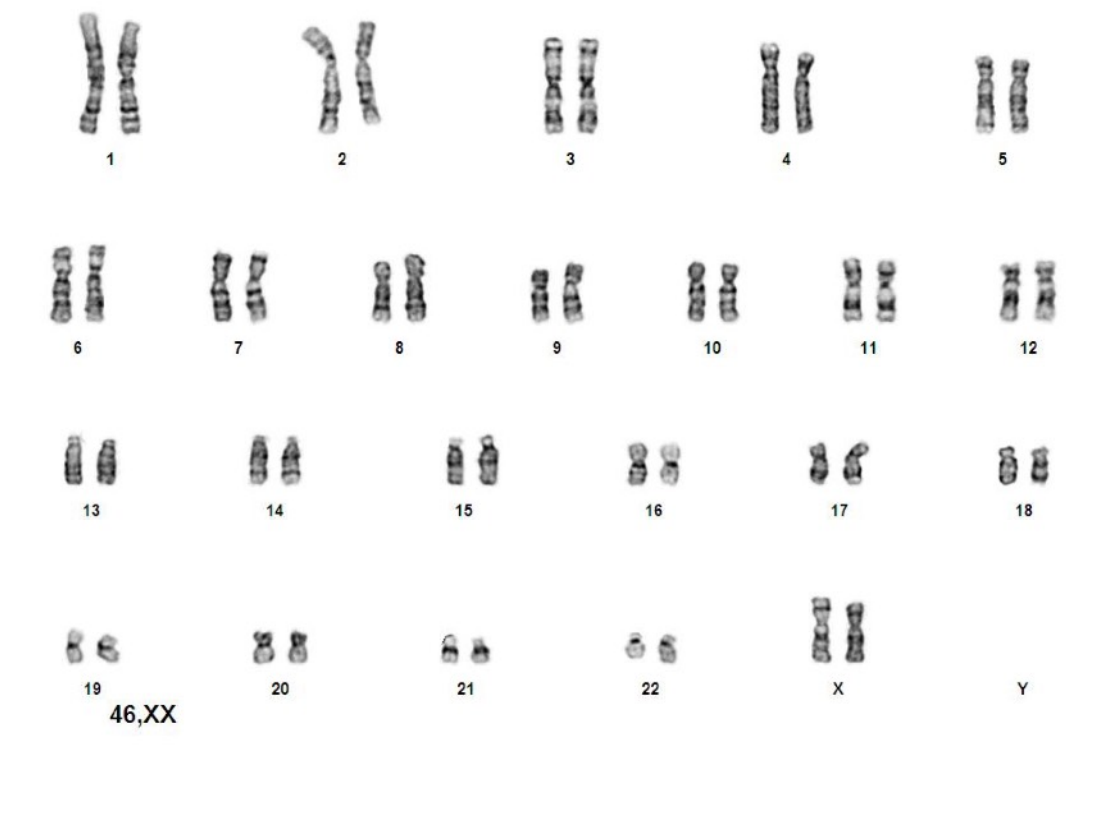
CANCER/LEUKEMIA

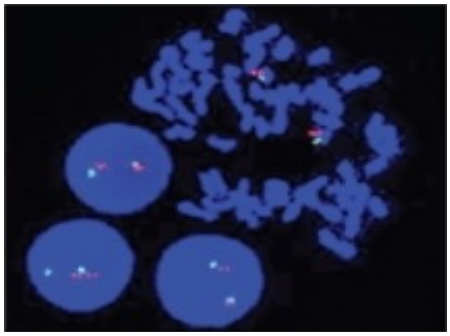
Bone Marrow & Leukemic Blood Chromosome Analysis

w and leukemic blood samples which, contain spontaneously dividing cells cultured short-term without mitogens. Some specimens with B-cell conditions are stimulated with mitogens. Chromosome analysis of the dividing cells is useful in the diagnosis and classification of hematological malignancies.



Fluorescence in Situ Hybridization (FISH)

Fluorescence in situ hybridization (FISH) utilizes molecular biology techniques to detect the presence or absence of, and enumerate specific regions of the genome using fluorescently labeled DNA (probes). Unlike most other techniques used to study chromosomes, FISH does not require actively dividing cells. This adds flexibility and increases the ability to identify and characterize cytogenetic abnormalities.



Polymerase Chain Reaction (PCR)

The polymerase chain reaction (PCR) is a molecular method that enables the detection and analysis of specific gene sequences in a patient's sample. PCR testing is used to both diagnosis and monitor hematologic malignancies and solid tumors. Peripheral blood, bone marrow, and paraffin embedded tissue are acceptable specimen types for PCR testing.

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Cancer Microarray

Single nucleotide polymorphism (SNP) chromosome microarray (CMA), is a molecular cytogenomic technology that evaluates areas of the human genome for gains or losses of chromosome segments at a higher resolution than traditional karyotyping. Due to the small size, many of these DNA imbalances may not be detected by standard cytogenetics. SNP CGH also will identify marker chromosomes, some cases of mosaicism, and aneuploidy. The performance characteristics of this test, including sensitivity and specificity, have been determined by Genetics Associates. All results are interpreted by board certified and Tennessee licensed cytogenetic directors at our laboratory, who are available to discuss results and offer clinical advice.

Next Generation Sequencing (NGS)

Next-generation sequencing (NGS) is a technology that enables sequencing of multiple regions of the genome in a single assay. Compared to “traditional” or Sanger sequencing method, NGS allows sequencing at higher coverage levels (deep sequencing) that result in greater confidence for calling low-frequency variants. Our targeted NGS panels are designed to detect mutations, substitutions, insertions, and deletions that may have diagnostic, prognostic, and/or therapeutic implications in hematologic malignancies.

Fluorescence in situ Hybridization (FISH) Panels

Please note, FISH probes may be ordered individually or as panel.

Adult B-Cell ALL profile

del(9p) CDKN2A

del(6q) CCND3/SEC63/MYB

t(9;22) BCR/ABL1/ASS1

11q23 KMT2A (MLL) rearrangements

t(1;19) TCF3/PBX1

14q32 IGH rearrangements

Adult T-Cell ALL profile

14q11.2 TRA rearrangements

7q34 TRB rearrangements

10q24 TLX1

5q35 TLX3

11q23 KMT2A (MLL) rearrangements

del(9p) CDKN2A

Pediatric ALL profile

t(12;21) ETV6/RUNX1

11q23 KMT2A (MLL) rearrangements

t(9;22) BCR/ABL1/ASS1

trisomy 4,10,17

Additional probes

t(1;19) TCF3/PBX1

rearrangements

Ph-Like ALL profile

1q25.2 ABL2

5q32 PDGFRB

5q32 CSF1R

9p24.1 JAK2

9q34.1 ABL1

19p13.2 EPOR

Xp22.33/Yp11.3 CRLF2

Acute Myelogenous (AML) profile

t(15;17) PML/RARA

t(9;22) BCR/ABL1/ASS1

t(8;21) RUNX1T1/RUNX1

11q23 KMT2A (MLL) rearrangements

inv(16), t(16;16) CBFB rearrangements

inv(3) MECOM rearrangements

17q RARA rearrangements

Chronic Lymphocytic (CLL) profile

del(11q) ATM

trisomy 12

del(13q) 13q14/13q34

del(17p) TP53

Additional probes

t(11;14) CCND1/IGH

del(6q) CCND3/SEC63/MYB

Chronic Myelogenous (CML) profile

t(9;22) BCR/ABL1/ASS1

Additional probes

trisomy 8

i(17q)

Lymphoma probes

t(8;14) MYC/IGH (Burkitt or Follicular)

8q24 MYC rearrangements

t(11;14) CCND1/IGH (Mantle Cell)

t(11;18) BIRC3/MALT1

18q21 BCL2 rearrangements

18q21 MALT1 rearrangements

t(14;18) IGH/BCL2 (Follicular or Diffuse Large B-Cell)

3q27 BCL6 rearrangements (Diffuse Large B-Cell, Follicular, Marginal Zone B-cell)

T-cell Leukemia/Lymphoma probes

2p23 ALK (Anaplastic) rearrangements

14q11.2 TRA rearrangements

7q34 TRB rearrangements

i(7q) 7cen/7q22/7q31

14q32 TCL1A

10q24 TLX1

5q35 TLX3

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Multiple Myeloma CD138 Enriched (MM) profile (FISHnet™)

1p32.3/1q21 CDKN2C/CKS1B

del(13q) 13q14/13q34

del(17p) TP53

t(11;14) CCND1/IGH

t(4;14) FGFR3/IGH

t(14;16) IGH/MAF

Additional probes

trisomy 5

trisomy 7

t(6;14) CCND3/IGH

t(14;20) IGH/MAFB

Myelodysplastic (MDS) profile

del(5q) EGR1

del(7q) / monosomy 7

trisomy 8

del(20q)

Additional probes

11q23 KMT2A (MLL) rearrangements

t(9;22) BCR/ABL1/ASS1

Myeloproliferative (MPN) profile

del(5q) EGR1

del(7q) / monosomy 7

trisomy 8

del(20q)

t(9;22) BCR/ABL1/ASS1

Additional probes

4q12 FIP1L1/CHIC2/PDGFRA

5q33 PDGFRB rearrangements

8p11 FGFR1 rearrangements

Solid Tumor probes

EWSR1 Ewing Sarcoma

FOXO1 Alveolar Rhabdomyosarcoma

DDIT3 (CHOP) Myxoid Liposarcoma

LOH 1p/19q Glioma

MYCN 2p24.1 Neuroblastoma

SS18 Synovial Sarcoma

UroVysion®

Transplant

XX/XY for sex mismatched transplant

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Individual FISH Probes

(Numerically by Chromosome)

Probe Requisition	Location on Cancer
1p32.3/1q21 CDKN2C/CKS1B	Multiple Myeloma Profile
1q25.2 ABL2	Ph-Like ALL profile
t(1;19) TCF3/PBX1	Adult B-Cell ALL profile

t(1;19) TCF3/PBX1	Pediatric ALL profile
1p/19q LOH (Glioma)	Solid Tumor probes
2p23 ALK (Anaplastic) rearrangements probes	T-cell Leukemia/Lymphoma
2p24.1 MYCN (Neuroblastoma)	Solid Tumor probes
inv(3) MECOM rearrangements profile	Acute Myelogenous (AML)
3q27 BCL6 rearrangements (Diffuse Large B-Cell, Follicular, Marginal Zone B-cell)	Lymphoma probes
trisomy 4,10,17	Pediatric ALL profile
4q12 FIP1L1/CHIC2/PDGFRA	Myeloproliferative (MPN) profile
t(4;14) FGFR3/IGH	Multiple Myeloma Profile
trisomy 5	Multiple Myeloma Profile
del(5q) EGR1	Myelodysplastic (MDS) profile
del(5q) EGR1	Myeloproliferative (MPN) profile
5q32 CSF1R	Ph-Like ALL profile
5q33 PDGFRB rearrangements profile	Myeloproliferative (MPN)
RB rearrangements	Ph-Like ALL profile
	Adult T-Cell ALL profile
del(6q) CCND3/SEC63/MYB	Adult B-Cell ALL profile
del(6q) CCND3/SEC63/MYB	Chronic Lymphocytic (CLL) profile
t(6;14) CCND3/IGH	Multiple Myeloma Profile
trisomy 7	Multiple Myeloma Profile

del(7q) / monosomy 7 profile	Myelodysplastic (MDS)
del(7q) / monosomy 7 profile	Myeloproliferative (MPN)
i(7q) 7cen/7q22/7q31 probes	T-cell Leukemia/Lymphoma
7q34 TRB rearrangements	Adult T-Cell ALL profile
7q34 TRB rearrangements	T-cell Leukemia/Lymphoma probes
trisomy 8	Chronic Myelogenous (CML) profile
trisomy 8	Myelodysplastic (MDS) profile
trisomy 8	Myeloproliferative (MPN) profile
8p11 FGFR1 rearrangements	Myeloproliferative (MPN) profile
8q24 MYC rearrangements	Lymphoma probes
t(8;14) MYC/IGH (Burkitt or Follicular)	Lymphoma probes
t(8;21) RUNX1T1/RUNX1	Acute Myelogenous (AML) profile
9p24.1 JAK2	Ph-Like ALL profile
9p24.1 JAK2	Myeloproliferative (MPN) profile
del(9p) CDKN2A	Adult B-Cell ALL profile
del(9p) CDKN2A	Adult T-Cell ALL profile
9q34.1 ABL1	Ph-Like ALL profile

t(9;22) BCR/ABL1/ASS1	Adult B-Cell ALL profile
t(9;22) BCR/ABL1/ASS1	Pediatric ALL profile
t(9;22) BCR/ABL1/ASS1 profile	Acute Myelogenous (AML)
t(9;22) BCR/ABL1/ASS1 profile	Chronic Myelogenous (CML)

t(9;22) BCR/ABL1/ASS1 profile	Myelodysplastic (MDS)
t(9;22) BCR/ABL1/ASS1 profile	Myeloproliferative (MPN)
10q24 TLX1	Adult T-Cell ALL profile
10q24 TLX1	T-cell Leukemia/Lymphoma probes
del(11q) ATM	Chronic Lymphocytic (CLL) profile
11q23 KMT2A (MLL) rearrangements	Myelodysplastic (MDS) profile
11q23 KMT2A (MLL) rearrangements	Adult B-Cell ALL profile
11q23 KMT2A (MLL) rearrangements	Adult T-Cell ALL profile
11q23 KMT2A (MLL) rearrangements	Pediatric ALL profile
11q23 KMT2A (MLL) rearrangements	Acute Myelogenous (AML) profile
t(11;14) CCND1/IGH	Chronic Lymphocytic (CLL) profile
t(11;14) CCND1/IGH (Mantle Cell)	Lymphoma probes
t(11;14) CCND1/IGH	Multiple Myeloma Profile
t(11;18) BIRC3/MALT1	Lymphoma probes
trisomy 12	Chronic Lymphocytic (CLL) profile
12q13 DDIT3 (CHOP) Myxoid Liposarcoma	Solid Tumor probes
t(12;21) ETV6/RUNX1	Pediatric ALL profile

del(13q) 13q14/13q34 profile	Chronic Lymphocytic (CLL)
del(13q) 13q14/13q34	Multiple Myeloma Profile
13q14.1 FOXO1 Alveolar Rhabdomyosarcoma	Solid Tumor probes

14q11.2 TRA rearrangements	Adult T-Cell ALL profile
14q11.2 TRA rearrangements	T-cell Leukemia/Lymphoma probes
14q32 IGH rearrangements	Adult B-Cell ALL profile
14q32 IGH rearrangements	Pediatric ALL profile
14q32 TCL1A	T-cell Leukemia/Lymphoma probes
t(14;16) IGH/MAF	Multiple Myeloma Profile
t(14;18) IGH/BCL2 (Follicular or Diffuse Large B-Cell)	Lymphoma probes
t(14;20) IGH/MAFB	Multiple Myeloma Profile
t(15;17) PML/RARA	Acute Myelogenous (AML) profile
inv(16), t(16;16) CBFB rearrangements	Acute Myelogenous (AML) profile
del(17p) TP53	Chronic Lymphocytic (CLL) profile
del(17p) TP53	Multiple Myeloma Profile
i(17q)	Chronic Myelogenous (CML) profile
17q RARA rearrangements	Acute Myelogenous (AML) profile
18q11.2 SS18 Synovial Sarcoma	Solid Tumor probes
18q21 BCL2 rearrangements	Lymphoma probes
earrangements	Lymphoma probes
19p13.2 EPOR	Ph-Like ALL profile
del(20q)	Myelodysplastic (MDS) profile
del(20q)	Myeloproliferative (MPN) profile

22q12.2 EWSR1 Ewing Sarcoma	Solid Tumor probes
UroVysion®	Solid Tumor probes
Xp22.33/Yp11.3 CRLF2	Ph-Like ALL profile
Xcen/Yq12 CEPXY (XX/XY for sex mismatched transplant)	Transplant

CONSTITUTIONAL

(Including Prenatal)

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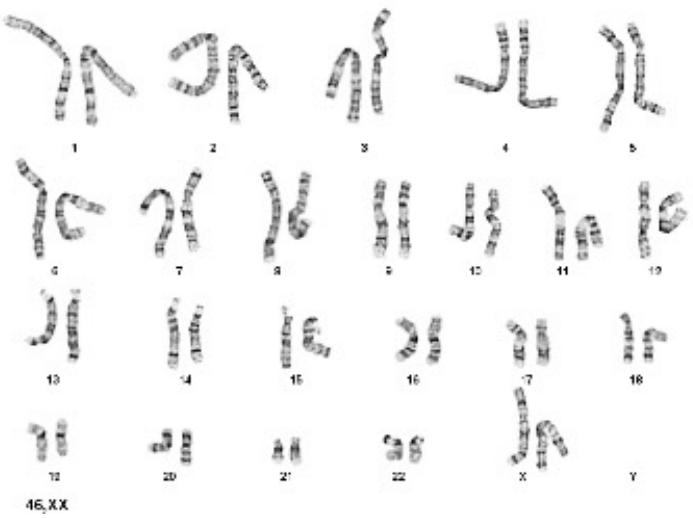
Peripheral Blood Chromosome Analysis

Chromosome analysis (karyotyping) is the microscopic evaluation of metaphase chromosomes for numerical and structural abnormalities associated with disease. A nationally certified and state licensed cytogenetic technologist analyzes the banded chromosomes at a microscope. Digital images of representative cells are captured and the chromosomes are arranged in a standard format known as a karyogram. The karyograms are then reviewed by a board-certified and state licensed clinical cytogeneticist.



Prenatal Chromosome Analysis

The chromosomal constitution of the fetus can be accurately determined from cells derived by prenatal diagnostic techniques. Chromosome studies of the fetus are indicated when there is a significant risk for a chromosome abnormality.

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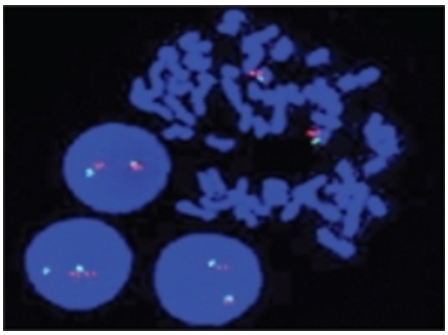
Fluorescence in Situ Hybridization (FISH)

Fluorescence in situ hybridization (FISH) utilizes molecular biology techniques to detect the presence or absence of, and enumerate specific regions of the genome using fluorescently labeled DNA (probes). Unlike most other techniques used to study chromosomes, FISH does not require actively dividing cells. This adds flexibility and increases the ability to identify and characterize cytogenetic abnormalities.



Polymerase Chain Reaction (PCR)

The polymerase chain reaction (PCR) is a molecular method that enables the detection and analysis of specific gene sequences in a patient's sample. PCR testing is used to both diagnosis and monitor hematologic malignancies and solid tumors. Peripheral blood, bone marrow, and paraffin embedded tissue are acceptable specimen types for PCR testing.

Microarray

Single nucleotide polymorphism (SNP) chromosome microarray (CMA), is a molecular cytogenomic technology that evaluates areas of the human genome for gains or losses of chromosome segments at a higher resolution than traditional karyotyping. Due to the small size, many of these DNA imbalances may not be detected by standard cytogenetics. SNP CGH also will identify marker chromosomes, some cases of mosaicism, and aneuploidy. The performance characteristics of this test, including sensitivity and specificity, have been determined by Genetics Associates. All results are interpreted by board certified and Tennessee licensed cytogenetic directors at our laboratory, who are available to discuss results and offer clinical advice.

Noninvasive Prenatal Test (NIPT)

The verifi® from Genetics Associates, Inc. Prenatal Test is a non-invasive test that **screens** for aneuploidy of chromosomes 21, 18, and 13. Additional screening is available for sex chromosome aneuploidies and select microdeletions in singleton pregnancies. In twin pregnancies, screening for aneuploidy in chromosomes 21, 18, and 13 and the option to screen the Y chromosome is available. This test can be performed as early as 10 age (8 weeks of fetal age as determined by date of conception.

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FISH for Microdeletion Syndromes

(Listed Alphabetically)

Angelman Syndrome (15q12)

Cri-du-Chat Syndrome (5p15.3)

DiGeorge Syndrome (22q11.2)

DiGeorge II Syndrome (10p14)

Kallman Syndrome (Xp22.3)

Miller-Dieker Syndrome (17p13.3)

Pallister-Killian Syndrome/Tetrasomy 12p

Phelan-McDermid Syndrome (22q13)

Prader-Willi Syndrome (15q12)

Saethre-Chotzen Syndrome (7p21.1)

Smith-Magenis Syndrome (17p11.2)

Sotos Syndrome (5q35.3)

Steroid Sulfatase Deficiency Syndrome (Xp22.3)

Williams Syndrome (7q11.23)

Wolf-Hirschhorn Syndrome (4p16.3)

1p36 microdeletion Syndrome

ABBREVIATIONS

ABBREVIATIONS and ACRONYMS

FISH chromosome	Fluorescence in situ hybridization. This is a methodology used to detect abnormalities in cells.
FL	Follicular lymphoma
FXS	Fragile X syndrome
GVHD	Graft vs host disease
GVL	Graft vs leukemia or graft vs lymphoma
HCL	Hairy cell leukemia
HCT	Hematocrit; the percentage of red blood cells in the blood. A low hematocrit measurement indicates anemia.
HD	Hodgkin disease
HDC	High-dose chemotherapy
Hem/Onc	Hematologist/Oncologist
HGB	Hemoglobin
HLA bone	Human leukocyte antigen test; a special blood test used to match a blood or

	marrow donor to a recipient for transfusion or transplant.
Ig	Immunoglobulin (IgA, IgD, IgE, IgG, IgM)
ITP	Idiopathic thrombocytopenia purpura
IV	Intravenous
LB	Leukemic blood
LPL	Lymphoplasmacytic lymphoma
MALT	Mucosa associated lymphoid tissue
Mab	Monoclonal antibodies
MCL	Mantle cell lymphoma
MDS	Myelodysplastic syndrome
MM	Multiple myeloma
MS	Multiple sclerosis
	Myeloproliferative disease

MUD	Matched unrelated donor of bone marrow
MZL	Marginal zone lymphoma
NHL	Non-Hodgkin lymphoma
OR	Overall remission
OS	Overall survival
PD	Parkinson’s Disease
PB	Peripheral blood
PLL	Prolymphocytic Leukemia
PMF	Primary Myelofibrosis
PR	Partial remission
PV	Polycythemia Vera
RARS	Refractory anemia with ringed sideroblasts
RBC	Red Blood count
Rx	Prescribed medication
SB	Spina bifida
SBMT	Syngeneic bone marrow transplantation (identical twin transplant)
SLE	Systemic lupus erythematosus
SLVL	Splenic lymphoma with villous lymphocytes

SMA	Spinal muscular atrophy
SMCD-eos	Systemic mast cell disease with eosinophillia
SMS	Smith Magenis syndrome
SPS	Stiff Person syndrome
SRBCT	Small round blue cell tumor
T-ALL	T-cell acute lymphoblastic leukemia
t-AML	Therapy-related acute myeloid leukemia
TTP	Thrombotic thrombocytopenia purpura
	Turnaround time

VCFS	Velocardiofacial syndrome
VHL	Von Hippel-Lindau disease
WBC	White Blood cell
WD	Well differentiated
WM	Waldenstrom’s macroglobulinemia
WS	Williams syndrome
XRT	External radiation therapy
XP	Xeroderma pigmentosa

CYTOGENETIC ABBREVIATIONS

AI	First meiotic anaphase
AII	Second meiotic anaphase
Ace	Acentric fragment
add	Additional material of unknown origin
amp	Denotes an amplified signal
(~)	Approximate sign denotes intervals and boundaries of a chromosome segment or number of chromosomes, fragments, or markers; denotes a range of number of copies of a chromosome region when the exact number cannot be determined
arr	Microarray
b	Break
(< >)	Brackets, angle; surround the ploidy level
([ ])	Brackets, square; surround number of cells or genome build
c	Constitutional anomaly
cen	Centromere
cgh	Comparative genomic hybridization
chi	Chimera
chr	Chromosome
cht	Chromatid
(:)	Colon, single; break, in detailed system
(::)	Colon, double; break and reunion, in detailed system

(,)	Comma; separates chromosome numbers, sex chromosomes, chromosome abnormalities
con	Connected signals
cp	Composite karyotype
cth	Chromothripsis
({})	Curly braces; indicates differences in the altered segment compared to the reference sequence in duplications, in versions, conversions, insertions, etc.

cx	Complex rearrangements
(.)	Decimal point; denotes sub-bands
del	Deletion
der	Derivative chromosome
dia	Diakinesis
dic	Dicentric
dim	Diminished
dip	Diplotene
dis	Distal
dit	Dictyotene
dmin	Double minute
dn	de novo; designates a chromosome abnormality that has not been inherited
dup	Duplication
e	Exchange
end	Endoreduplication
enh	Enhanced
(=)	Equal sign; number of chiasmata
fem	Female
fib	Extended chromatin/DNA fiber
fis	Fission, at the centromere
fra	Fragile site

g	Gap
GRCh	Genome Reference Consortium human; human genome build or assembly
h	Heterochromatin, constitutive

hmz	Homozygous, homozygosity; used when one or two copies of a genome are detected, but previous, known heterozygosity has been reduced to homozygosity through a variety of mechanisms, e.g. loss of heterozygosity (LOH)
hsr	Homogeneously staining region
htz	Heterozygous; heterozygosity
i	Isochromosome
idem	Denotes the stemline karyotype in a subclone
ider	Isoderivative chromosome
inc	Incomplete karyotype
inh	Inherited
ins	Insertion
inv	Inversion
ish	In Situ hybridization
lep	Leptotene
MI	First meiotic metaphase
MII	Second meiotic metaphase
mal	Male
mar	Marker chromosome
mat	Maternal origin
med	Medial
min	Minute acentric fragment
(-)	Minus sign/loss
mos	Mosaic
(x)	Multiplication sign; multiple copies of rearranged chromosomes
neg	No presence of the rearrangement for which testing was conducted

neo	Neocentromere
nuc	Nuclear or interphase

oom	Oogonial metaphase
or	Alternative interpretation
p	Short arm of chromosome
PI	First meiotic prophase
pac	Pachytene
()	Parentheses; surround structurally altered chromosomes and breakpoints
pat	Paternal origin
pcc	Premature chromosome condensation
pcd	Premature centromere division
pcp	Partial chromosome paint
(.)	Period; separates various techniques
Ph	Philadelphia chromosome
(+)	Plus sign; additional normal or abnormal chromosome; increase in length; locus present on a specific chromosome
(++)	Plus sign, double; two hybridization signals or hybridization regions on a specific chromosome
pos	Detection of a rearrangement for which testing was conducted
prx	Proximal
ps	Satellited short arm of chromosome
psu	Pseudo-
pter	Terminal end of the short arm
pvz	Pulverization
q	Long arm of chromosome
qdp	Quadruplication
qr	Quadriradial
qs	Satellited long arm of chromosome

qter	Terminal end of the long arm
(?)	Question mark; questionable identification of a chromosome or chromosome structure
r	Ring chromosome
rec	Recombinant chromosome
rev	Reverse, including comparative genomic
rob	Robertsonian translocation
I-IV structures	Roman numerals; indicate univalent, bivalent, trivalent, and quadrivalent
rsa	Region-specific assay
s	Satellite
sce	Sister chromatid exchange
sdl	Sideline
(;)	Semicolon; separates altered chromosomes and breakpoints in structural rearrangements involving more than one chromosome; separates probes on different derivative chromosomes
sep	Separate signals
seq	Sequencing
sl	Stemline
(/)	Slant line, single; separates clones or contiguous probes
(//)	Slant line, double: separates chimeric clones
spm	Spermatogonial metaphase
stk	Satellite stalk
subtel	Subtelomeric region
t	Translocation
tas	Telomeric association
ter	Terminal (end of chromosome) or telomere
tr	Triradial
trc	Tricentric chromosome

underline (single)	Used to distinguish homologous chromosomes
(_)	Underscore; used to indicate range of nucleotide positions
upd	Uniparental disomy
var	Variant or variable region
wcp	Whole chromosome paint
xma	Chiasma(ta)
zyg	Zygotene