Chromosome Analysis
also known as: Cancer Cytogenetics, Karyotyping, G-bands

TEST DESCRIPTION:
Conventional cytogenetics (chromosome analysis) analyze the whole genome in a single assay. Developed over 50 years ago, this assay still plays a vital role in cancer diagnostics because it allows for simultaneous characterization of chromosome number and structure. This assay can provide diagnostic and prognostic information as well as be utilized to monitor response to therapy, disease progression, and treatment-related secondary malignancies.

TEST DETAILS and COMPLEMENTARY TESTING:
- Some chromosome alterations are too small or subtle to detect by Chromosome Analysis. Other testing, such as FISH or Microarray, may be indicated to further investigate chromosomal abnormalities.
- Chromosome Analysis only provides information relating to the number and structure of chromosomes; it does not evaluate for single gene disorders.

ADVANTAGES:
- Detects changes in chromosome number, including aneuploidy and triploidy
- Characterizes both balanced and unbalanced structural rearrangements
- Identifies large-scale deletions and duplications

LIMITATIONS:
- Requires mitotic (dividing) cells from a fresh specimen
- Limited resolution as compared to newer technologies such as microarray

INDICATIONS FOR TESTING:
- Leukemia
- Lymphoma
- Malignant hematologic disorders
- Solid tumor

SPECIMEN COLLECTION & TRANSPORT:
Complimentary test kits are available upon request, but are not required.

SAMPLE TYPES and REQUIREMENTS:
- **blood [cancer]:** 3-5 ml whole blood in a sodium heparin tube (green top)
- **bone marrow:** 1.5-3 ml bone marrow in a sodium heparin tube (green top)
  - Collect marrow from 1st or 2nd aspirate using a sodium-heparinized syringe
- **bone marrow core:** collect, store, and transport in tissue culture media – room temperature
- **lymphatic tissue / node:** ≥5 mm³ collect, store, and transport in tissue culture media – room temperature
- **solid tumor tissue:** ≥5 mm³ representative tumor tissue / collect, store, and transport in tissue culture media – room temperature
- **other - body fluid:** 1-50 ml fluid in a sterile container

SHIPPING:
- Maintain and ship samples at room temperature.
- Coordinate transport for sample to be received in our laboratory within 24-48 hours of collection.
  - **LOCAL:** Call 402-559-5070 (option 1)
OUT OF AREA: Prior to shipment, please fax the completed test request form to 402-559-7248, including the FedEx® airbill tracking number.
- Saturday delivery MUST be checked when sending FedEx® on Friday.
- Please include Internal Billing Reference # 3155070600 on the FedEx® airbill.
- Ship To: Human Genetics Laboratory – Zip 5440
UNMC Shipping & Receiving Dock
601 S. Saddle Creek Road
Omaha, NE 68106

FORMS FOR TESTING: The following forms can be downloaded via our website.
- Required: Oncology Test Request Form
- Optional: Informed Consent for Genetic Testing

POTENTIAL TEST RESULTS:

NORMAL:
- A normal result indicates no clinically-significant chromosome abnormalities were identified.
- 46, XX (female)
- 46, XY (male)

ABNORMAL:
- An abnormal result indicates that a chromosome abnormality was identified that may provide an explanation for the laboratory results.
- Deletion: Missing pieces of chromosomes and/or genetic material. Some may be small and difficult to detect.
- Duplication: Extra genetic material which may be present on any chromosome.
- Monosomy: The loss of one whole chromosome.
- Rearrangement: With this, genetic material is present on a chromosome but not in its usual location.
- Translocation: With translocations, pieces of chromosomes break off and reattach to another chromosome. If it is a one-to-one switch and all of the genetic material is present (but in the wrong place), it is said to be a reciprocal translocation. If it is not, then it is called an unbalanced translocation.
- Trisomy: The presence of an extra chromosome; a third instead of a pair.
- Loss of Y: A phenomenon seen in males as they age which may represent a neoplastic state if ≥75%.

TURNAROUND TIMES: Results are typically available in 2-5 days when performed on bone marrow and bone marrow core, cancer blood, and other body fluid. Results are typically available in 15-25 days when performed on lymphatic tissue/node and 20-26 days when performed on solid tumor tissue.

BILLING: Our laboratory offers patient/self-pay, insurance (including Medicare/Medicaid), and client/institution billing options. Verifying coverage requirements or obtaining preauthorization PRIOR TO OR AT THE TIME OF SPECIMEN COLLECTION is often necessary. We provide preauthorization services upon request by calling 402-559-5070 (option 3); the following form is helpful for obtaining the information required by insurance providers and can be downloaded via our website.
- Request for Pre-Authorization for Genetic Testing (Hematology/Oncology)
In some circumstances, a test may be warranted even though insurance coverage is denied or not guaranteed. For these situations, we request the following form be signed by the patient and submitted with the sample. This helps inform patients of their potential financial responsibility, should the costs of genetic testing not be paid by their insurance provider.
- Advanced Beneficiary Notice of Noncoverage (ABN) – required when billing Medicare

CPT CODES:
Though some may not be billed, we recommend submitting all listed codes (for sample type being sent) for preauthorization.
- blood, bone marrow and core: 88237, 88261, 88262, 88280, 88285
- lymph node / solid tumor tissue: 88239, 88261, 88262, 88280, 88285

PRICING: For current costs contact the laboratory billing staff at 402-559-5070 (option 3).

REFERENCES:
- Clinical Significance of Y Chromosome Loss in Hematologic Disease; Genes, Chromosomes & Cancer. 27:11-16 (2000).
updated 3-2019