Chromosome Analysis

also known as: Conventional, Standard, Traditional, or High Resolution 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 postnatal diagnostics because it allows for simultaneous characterization of chromosome number and structure.

TEST DETAILS and COMPLEMENTARY TESTING:
- Some chromosome alterations are too small or subtle to detect by Chromosome Analysis. Other testing, such as Postnatal FISH or High Density SNP 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. Depending on the clinical presentation of a patient, Targeted Gene Sequencing or an Indication-Specific Gene Panel test may be most appropriate.
- Chromosome Analysis cannot diagnose Fanconi Anemia (FA), a disorder characterized by physical abnormalities, progressive bone marrow failure, and increased cancer susceptibility. Chromosome breakage studies are performed on patients with a suspected diagnosis of FA, and must be ordered separately.

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:
- Dysmorphic features
- Multiple congenital anomalies with or without intellectual disability
- Intellectual disability
- Failure to thrive
- Short stature, including suspicion of Turner syndrome
- Suspicion of a sex chromosome disorder, including Klinefelter syndrome
- Suspicion of aneuploidy or polyploidy
- Parent or other family member carrying a known chromosomal abnormality
- Family history of a chromosome abnormality
- Recurrent pregnancy losses
- Infertility - see also Male Infertility Panel including YCMD for male infertility

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

SAMPLE TYPE and REQUIREMENTS:
- **blood**: 2-5 ml whole blood in a sodium heparin tube (green top)
  - **newborn minimum requirement**: 1-3 ml
- **tissue / skin**: > 5 mm³ tissue from biopsy or skin punch (transport at room temperature in tissue culture media)
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

REQUIRED FORM: The following form can be downloaded via our website.
- Postnatal Test Request Form

OPTIONAL FORM:
- 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 likely provides an explanation for the clinical indications.
  - Parental testing ([Postnatal Chromosome Analysis](#) on maternal and/or paternal blood) may be recommended in order to clarify whether the result is *de novo* or familial for the purpose of recurrence risk calculation.
- **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, such as the presence of two horizontal bands at a specific location instead of one.
- **Monosomy:** The absence of one whole chromosome. This includes Turner syndrome, a female with a single X chromosome (45,X) instead of 46,XX. Most other monosomies are not compatible with life.
- **Rearrangement:** Genetic material is present on a chromosome but not in its usual location.
- **Translocation:** Translocations result when 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 balanced 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. These include Trisomy 21 (Down syndrome), Trisomy 13 (Patau syndrome), Trisomy 18 (Edwards syndrome), and Klinefelter syndrome, a male with an extra X chromosome (47,XXY) instead of 46,XY, and other less common trisomies.
- In some circumstances, a result will be unavailable.

TURN-AROUND-TIMES: For routine studies, results are typically available in 7-14 days; results are typically available in 2-3 days for newborn blood studies with a preliminary verbal report in 48 hours.
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.

- Insurance Preauthorization Request

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**: 88230, 88262, 88280
- **tissue \ skin**: 88233, 88262, 88280

*When applicable, the following codes may be used in place of or in addition to codes listed above: 88261, 88285, 88289*

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

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