Chromosome Analysis – Bone Marrow (Rapid/Newborn)

CPT Code(s): 88237(x2), 88261, 88280

Service Code (IU Health): 53101259, 53100533, 53100723


Synonyms: Karyotype, G-bands

Methodology: Rapid, tissue culture, microscopic analysis of G-banded chromosomes.

Performed: Monday through Saturday

Reported: 3-4 hours (Prelim)

Specimen Requirements

Patient Preparation: None

Collect: Non-diluted bone marrow aspirate in sodium heparin (green-top) vacutainer, heparinized syringe or flask with sterile media (provided upon request).

Specimen Volume: Preferred: 0.5 mL – 1.0 mL per flask.

Storage/Transport: Room temperature. Do not freeze or expose to extreme temperatures.


Remarks: Physician notified immediately if cultures result in no dividing cells for analysis.

Stability: Ambient: 48 hours; Refrigerated: 48 hours; Frozen: Unacceptable

Interpretive Data
Department of Medical and Molecular Genetics  
Division of Diagnostic Genomics

**Characteristics:** Decisions concerning medical intervention for life-threatening conditions in a newborn depend upon rapid evaluation of the constitutional chromosomal complement. Analysis of STAT bone marrow or peripheral blood specimen will detect gross numerical and structural abnormalities. Identification of subtle chromosome abnormalities is often not possible in these cells. Microscopic or computer analysis of at least five metaphases at 400 bands is completed for the preliminary report. Additional stimulated cultures are used to complete the analysis of at least twenty cells. Additional staining techniques may be utilized. Genetic counseling is recommended for abnormal results.

Negative: A 46,XX or 46,XY karyotype indicating no apparent chromosomal abnormality is considered negative.

Positive: Identification of any numerical or structural chromosomal abnormality. A report detailing interpretation of results will be provided.

**Limitations:** The preliminary report is based on analysis of at least five cells that may have decreased quality. Preliminary results may be updated/altered based on analysis of more cells with higher resolution. This does not eliminate the possibility of low frequency mosaicism or small structural abnormalities. Living cells are required for chromosome analysis. As such, sample quality can affect the turnaround time. A normal karyotype, i.e. 46,XX or 46,XY with no apparent chromosome abnormality, does not eliminate the possibility that the birth defect may be caused by submicroscopic cytogenetic lesions, molecular mutations, and/or environmental factors such as exposure to teratogens.

**References:**