Chromosome Analysis – Peripheral Blood (RAPID)

**CPT Code(s):** 88230(x2), 88261, 88280, 88289

**Service Code (IU Health):** 53100293, 53100566, 53100715, 53100772

**Ordering Recommendation:** Rapid analysis for detection of numerical and structural abnormalities of autosomes and sex chromosomes for individuals with multiple congenital anomalies. In addition, this test is often requested for infants in distress, with ambiguous genitalia, or suspected aneuploidy. G-banded karyotyping allows for the visualization and analysis of chromosomes for chromosomal rearrangements, including gains and losses, and structural rearrangements. Companion fluorescence *in-situ* hybridization (FISH) testing may also be utilized.

**Synonyms:** Karyotype, G-bands, Constitutional, Congenital

**Methodology:** Tissue culture, rapid, high resolution analysis of G-banded chromosomes. If ordered, fluorescence *in-situ* hybridization (FISH) analysis of metaphase cells.

**Performed:** Monday through Saturday

**Reported:** 48-72 hours (Prelim), 7 days (Final)

**Specimen Requirements**

**Patient Preparation:** Swab area with alcohol and let dry. Do not swab with Betadine.

**Collect:** Whole blood, Green (Sodium Heparin).

**Specimen Volume:** 3 mL (infants).

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

**Unacceptable Conditions:** Frozen specimens. Clotted specimens.

**Remarks:** Post-mortem, obtain by cardiac puncture within 1 hour. Physician notified if results are abnormal or if cultures result in no growth or contamination.
Department of Medical and Molecular Genetics  
Division of Diagnostic Genomics

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

Interpretive Data

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.