Chromosome Analysis – Peripheral Blood (Standard)

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

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

**Ordering Recommendation:** Detection of numerical and structural abnormalities of autosomes and sex chromosomes. G-banded karyotyping allows for the visualization and analysis of chromosomes for chromosomal rearrangements, including genomic gains and losses. Post-natal peripheral blood (leukocyte) chromosomes are indicated for an array of physical and/or mental difficulties. Approximately 7/1,000 live-births each year have a chromosome abnormality. Chromosomal microarray (CMA) is recommended if congenital anomalies are present that are not well defined by a known syndrome. CMA is also recommended for developmental delay and autism spectrum disorders. Companion fluorescence *in-situ* hybridization (FISH) testing may also be utilized.

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

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

**Performed:** Monday through Saturday

**Reported:** 7-10 days

**Specimen Requirements**

**Patient Preparation:** None

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

**Specimen Volume:** 2-4 mL (infants), 7-10 mL (adults).

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

**Unacceptable Conditions:** Frozen or clotted specimens.
Department of Medical and Molecular Genetics
Division of Diagnostic Genomics

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.

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

Interpretive Data

Characteristics:
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: 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.