Chromosome Analysis: Peripheral Blood (Age: 6 months and above)

**Condition Description**

This test will detect abnormalities in chromosome number and large deletions/duplications of chromosome material as well as balanced chromosomal rearrangements.

For most indications for cytogenetic testing (intellectual disability, developmental delays, autism spectrum disorders, multiple congenital anomalies, etc.) a chromosomal microarray has replaced the G-banded karyotype as the first-tier diagnostic test. For more information about the benefits of the microarray, please click here.

**Indications**

This test is indicated for patients with:

- a known or suspected family history of a chromosome abnormality
- recurrent miscarriage or infertility (ACOG Practice Bulletin)
- suspected trisomy 13, 18, or 21
- congenital abnormalities and/or developmental delay present (chromosomal microarray analysis is recommended as a first-tier test)

**Methodology**

PHA stimulated cultures are used for G-banded analysis. ISCN nomenclature is followed.

**Detection**

ISCN Nomenclature, minimum band resolution of 550.

**Specimen Requirements**

**Type: Whole Blood (Sodium heparin)**

**Specimen Requirements:**

- Sodium Heparin (Green Top)
- Infants and Young Children (2 years of age to 10 years old): 3-5 ml
- Older Children & Adults: 5-10 ml
- Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**

Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze. Not accepted on Saturday. (Late Friday collections may be stored at room temperature over the weekend for Monday receipt.)

**Related Tests**

- The EmArray Cyto (VA) may detect microdeletions/duplications that are not visible on a peripheral blood chromosome analysis and is recommended as a first tier test for patients with congenital abnormalities and/or developmental delay.
- When mosaicism is suspected but not detected on a standard peripheral blood chromosome analysis, a chromosome analysis for mosaicism (MM) in peripheral blood or a chromosome analysis on skin fibroblasts (CSKNC) may be warranted.
- If there is a known chromosome abnormality in the family, such as a translocation, a targeted, family member chromosome study (FS) may be indicated.