Chromosome Analysis: Peripheral Blood (Mosaicism)

| Test Code: MM |
| Turnaround time: 7 days - 14 days |
| CPT Codes: 88230 x1, 88262 x1, 88285 x1, 88291 x1 |

Condition Description

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

Analysis of additional cells may be needed to rule out the possibility of chromosomal mosaicism for sex chromosome abnormalities, specific trisomies, and other syndromes, such as Ito syndrome. In EGL’s mosaicism study, 50 cells are examined compared to 20 cells for a routine chromosome analysis.

Indications

This test is indicated when mosaicism, the presence of two or more cell lines with different chromosome patterns, is suspected based upon clinical findings. Infertility may be caused by mosaicism for a sex chromosome abnormality (such as abnormal skin pigmentation). This test is also indicated when clinical stigmata suggest a specific chromosome abnormality, such as trisomy 21, but initial chromosome studies are negative.

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

Specimen Requirements:

- In sodium heparin (green top) tube:
  - Infants (2 years): 3-5 ml
  - Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

Related Tests

- The EmArray Cyto + SNP (CMSNP) or 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. This test may also detect mosaicism not identified by G-banded analysis.
- Chromosome analysis on skin fibroblasts (CSKNC) may be warranted if mosaicism is not detected in blood.
- If there is a known chromosome abnormality in the family, such as a translocation, a targeted, family member chromosome study (FS) may be indicated.