Chromosome Analysis: Peripheral Blood (Age: Less than 6mo)

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
- 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

Specimen Requirements:

In sodium heparin (green top) tube:

Infants:

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.