Chromosome Analysis: Fetal Blood (Percutaneous Umbilical Blood Sampling/PUBS)

Test Code: CP  
Turnaround time: 5 days (Preliminary Report: 2-3 days by request)  
CPT Codes: 88230 x1, 88262 x1, 88291 x1

Condition Description

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

Percutaneous Umbilical Blood Sampling (PUFS) is performed to test a pregnancy for a chromosome abnormality or other genetic condition, depending on the patient’s family history and availability of testing. A karyotype or chromosome analysis is performed on the fetal blood sample.

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: Cord Blood

Specimen Requirements:

In sodium heparin (green top) tube:  
Fetal Blood (PUFS): 1-3 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 (VA) may detect microdeletions/duplications that are not visible on a PUFS chromosome analysis and is recommended as a first tier diagnostic test.
- When mosaicism is suspected but not detected on a PUFS 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.