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: 88291 x1, 88230 x1, 88262 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 (PUBS) 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

Submit only 1 of the following specimen types

Type: Cord Blood

Specimen Requirements:
EDTA (Purple Top) or ACD (Yellow Top)
Collect 1 to 3 ml of fetal blood

Specimen Collection and Shipping:
Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

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 PUBS chromosome analysis and is recommended as a first tier diagnostic test.
- When mosaicism is suspected but not detected on a PUBS 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.