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: 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.

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.

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.