Chromosome Analysis: Amniotic Fluid

Test Code: AD

Turnaround time: 7 days - 14 days. (These cases are processed as STAT. We endeavor to report cases within 7 to 10 days; however, in the case of slow sample growth, this assay may take up to 14 days to complete.)

CPT Codes: 82106 x1, 88235 x1, 88267 x1, 88280 x1, 88291 x1

Condition Description

Prenatal diagnosis for chromosomal abnormalities is the most common indication for prenatal testing. Over 50% of early pregnancy losses result from chromosomal abnormalities in the fetus, including structural and numerical abnormalities. Aneuploidy of the non-sex chromosomes increases in frequency as maternal age increases.

This test will detect abnormalities in chromosome number and large deletions/duplications of chromosome material as well as balanced chromosomal rearrangements. For smaller genetic imbalances, please refer to the Prenatal EmArray Cyto.

The American College of Obstetricians and Gynecologists Committee Opinion on Array CGH in Prenatal Diagnosis, Number 446, November 2009 provides the following practice recommendations:

- Conventional karyotyping remains the principal cytogenetic tool in prenatal diagnosis.
- Targeted array CGH, in concert with genetic counseling, can be offered as an adjunct tool in prenatal cases with abnormal anatomic findings and a normal conventional karyotype, as well as in cases of fetal demise with congenital anomalies and the inability to obtain conventional karyotype.
- Couples choosing targeted array CGH should receive both pre-test and post-test genetic counseling. Follow-up genetic counseling is required for interpretation of array CGH result. Couples should understand that array CGH will not detect all genetic pathologies and that array CGH results may be difficult to interpret.

References:


Indications

Prenatal chromosomal analysis is appropriate for any woman seeking prenatal detection of chromosomal abnormalities. Common indications for prenatal diagnosis include:

- Advanced maternal age
- Abnormal maternal serum screen
- Abnormal ultrasound
- Family history of a genetic imbalance
- Parental concern
- Prior pregnancy with a chromosomal abnormality

Methodology

Amniotic fluid is cultured and used for G-banded analysis. ISCN nomenclature is followed.

Detection

ISCN nomenclature, minimum band resolution of 450.

Specimen Requirements

Type: Amniotic Fluid

Specimen Requirements:

Collect 20-30 ml of amniotic fluid (discard the first 1-2 ml). Place in sterile conical centrifuge tubes.

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

Special Instructions

Please consult with our laboratory genetic counselors prior to submitting prenatal samples. Our genetic counselors are available to help assist in the coordination and ordering of prenatal testing.

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• Prenatal STAT FISH may be warranted for rapid detection of the most common chromosomal aneuploidies.
• The prenatal EmArray Cyto (CMPRE) may detect microdeletions/duplications that are not visible on a peripheral blood chromosome analysis and is suggested in the presence of ultrasound anomalies.
• If there is a known chromosome abnormality in the family, such as a translocation, a targeted, family member chromosome study (FS) may be indicated.