Chromosome Analysis – Amniotic Fluid

**CPT Code(s):** 88235, 88269, 88280, 88285

**Service Code (IU Health):** 53101234, 53100616, 53100715, 53100764

**Ordering Recommendation:** Prenatal (fetal) analysis of amniocytes is a useful diagnostic method for identifying chromosomal abnormalities associated with a large number of congenital disorders and birth defects. This method is used for detection of genetic chromosomal abnormalities in patients with advanced maternal age (AMA), family history of genetic abnormality, abnormal prenatal screening, or abnormal fetal ultrasound. Companion fluorescence in situ hybridization (FISH) testing for prenatal aneuploidy screening (13, 18, 21, X, Y) may also be performed. No additional specimen is required.

**Synonyms:** Karyotype, G-bands, prenatal diagnosis, prenatal chromosomes.

**Methodology:** Tissue culture, microscopic analysis of G-banded chromosomes. If ordered, fluorescence in-situ hybridization (FISH) analysis of interphase cells.

**Performed:** Monday through Saturday

**Reported:** 7-10 days

**Specimen Requirements**

**Patient Preparation:** 14 weeks gestation or greater (or alternatively 1 mL/week of gestation for amniocentesis between 12 and 14 weeks) in a sterile syringe.

**Collect:** Discard first 2-3 mL to avoid maternal cell contamination. Place remaining fluid in 3-4 aliquots, labeled 1st, 2nd, etc in sterile tubes. Sterile Corning centrifuge tubes can be provided upon request.

**Specimen Volume:** 10-25 mL amniotic fluid.

**Storage/Transport:** Refrigerate. Do not centrifuge.

**Unacceptable Conditions:** Frozen. Centrifuged.

**Remarks:** For bloody specimens, use centrifuge tubes containing sodium heparin (can be provided).
Department of Medical and Molecular Genetics
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**Stability:** Ambient: 24 hours; Refrigerated: 48 hours; Frozen: Unacceptable

**Interpretive Data**

**Characteristics:**
Negative: A 46,XX or 46,XY karyotype indicating no apparent chromosomal abnormality is considered negative.
Positive: Identification of any numerical or structural chromosomal abnormality. A report detailing interpretation of results will be provided.

**Incidence:** Variable - Trisomy 21 - 1/700, Trisomy 18 - 1/7,500, Trisomy 13 - 1/15,000, Sex chromosomal aneuploidies ~1/1000 births

**Limitations:** This analysis does not eliminate the possibility of low frequency mosaicism or small structural abnormalities. Living cells are required for chromosome analysis. As such, sample quality can affect the turnaround time. A normal karyotype, i.e. 46,XX or 46,XY with no apparent chromosome abnormality, does not eliminate the possibility that the birth defect may be caused by submicroscopic cytogenetic lesions, molecular mutations, and/or environmental factors such as exposure to teratogens.