

TEST: Chromosome analysis, amniotic fluid

DESCRIPTION: In situ culture of amniotic fluid cells to produce metaphase cells for Giemsa-banded chromosome analysis. Chromosome examination includes detailed examination of 5 cells and 20 cells total counted from approximately 15 colonies in 2 or more different cultures. A karyogram is prepared on at least 3 cells. Often fluorescence in situ hybridization (FISH) analysis of interphase amniotic fluid cells is also performed to rapidly exclude the common autosomal trisomies and sex chromosome numerical abnormalities. Amniotic fluid alpha-fetoprotein (AFP) determination can also be performed. Acetylcholinesterase determination is performed if the AFP is found to be elevated.

INDICATIONS:

- advanced maternal age
- fetal anomalies on prenatal ultrasound
- polyhydramnios
- intrauterine growth retardation
- abnormal maternal serum triple screen results
- previous child with a chromosome abnormality
- familial chromosome rearrangement
- biochemical or molecular diagnosis of a genetic disorder

Cultured amniotic fluid cells can also be used for other tests such as biochemical or molecular analyses as indicated by the clinical situation.

SPECIMEN REQUIREMENTS:  $\geq 15$  cc amniotic fluid collected in a sterile Becton-Dickinson syringe (other syringes may have lubricants which may be harmful to the viability of the amniocytes) and transferred to two sterile polypropylene screw-cap tubes. Polystyrene tubes should not be used because cells adhere to polystyrene causing the number of viable amniotic fluid cells available for culture to be significantly reduced. Label tubes with patient's name and medical record number. Send to CompGene at room temperature with a Requisition for Chromosome Analysis form as soon as possible after collection. May be refrigerated. **Specimen must not be frozen.**

REFERENCE VALUE: 46,XX or 46,XY

TURN AROUND TIME: 6-10 days (usually 7-8 days)  
1 -2 days for fluorescence in situ hybridization (FISH) analysis

CPT CODES: 88235 - tissue culture for amniotic fluid  
88269 - chromosome analysis, amniotic fluid  
88291 - cytogenetic interpretation and report  
  
88271 - fluorescence in situ hybridization probes  
88275 - fluorescence in situ hybridization 100-300 cells  
  
82106 - amniotic fluid alpha-fetoprotein  
82013 - acetylcholinesterase