TEST: Chromosome analysis, peripheral blood lymphocytes

DESCRIPTION: 2 or 3 day PHA-stimulated culture of peripheral blood lymphocytes to produce metaphase cells for Giemsa-banded chromosome analysis. Chromosome examination includes detailed examination of 5 cells and 20 cells total counted. A karyogram is prepared on at least 3 cells.

INDICATIONS:
- congenital anomalies
- developmental delay / mental retardation
- short stature
- ambiguous genitalia
- recurrent miscarriages
- family history of a chromosome rearrangement

SPECIMEN REQUIREMENTS: 5-8 cc (2 cc for infants and small children) in a sodium heparin tube. Use a small sodium heparin tube if the sample volume is $\leq$2cc. The ratio of sample volume to heparin is important. Too much heparin for a small amount of blood often results in a low mitotic index and suboptimal chromosome morphology. Invert collection tube several times to mix blood with anticoagulant. Culture of clotted blood often does not result in dividing cells necessary for cytogenetic analysis. Other anticoagulants such as lithium heparin or EDTA may be detrimental to the preparation of metaphase cells. Send to CompGene at room temperature with a Requisition for Chromosome Analysis form as soon as possible. May be refrigerated overnight. **Specimen must not be frozen.** Label tube with patient's name and medical record number.

REFERENCE VALUE: 46,XX or 46,XY

TURN AROUND TIME: 3 -7 days (2 or 3 days for newborns)

CPT CODES: 88230 - tissue culture for peripheral blood lymphocytes
88262 - chromosome analysis, congenital disorders
88291 - cytogenetic interpretation and report

88271 - fluorescence in situ hybridization probes
88275 - fluorescence in situ hybridization 100-300 cells