

TEST: Chromosome analysis, high resolution banding

DESCRIPTION: Special synchronized 3 day PHA-stimulated culture of peripheral blood lymphocytes to produce metaphase cells for Giemsa-banded chromosome analysis at the 650 or higher band level of resolution. Chromosome analysis includes detailed examination of 5 cells and 20 cells total counted. A karyogram is prepared on at least 3 cells and partial karyotypes are prepared of at least 3 different pairs of homologous chromosomes for each chromosome.

INDICATIONS: High resolution banding chromosome analysis is helpful to identify subtle chromosome rearrangements such as microdeletions and cryptic translocations that may go undetected by routine chromosome analysis. This test is usually ordered by a clinical geneticist who suspects a patient may have a specific chromosomal syndrome when a routine chromosome analysis has not revealed a chromosome abnormality. High resolution banding chromosome analysis may be helpful for certain patients with congenital anomalies, developmental delay / mental retardation and short stature.

SPECIMEN REQUIREMENTS: 5-8 cc blood (2 cc for infants and small children) in sodium heparin tube. Use a small sodium heparin tube if the sample volume is ≤ 2 cc. 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 the collection tube several times to mix blood with anticoagulant. Culture of clotted blood often does not result in spontaneously dividing cells which are necessary for cytogenetic analysis. Other anticoagulants such as lithium heparin and EDTA may be harmful to the viability of the cells. Label tube with patient's name and medical record number. Send to CompGene at room temperature as soon as possible after collection. May be refrigerated overnight. **Specimen must not be frozen.** Indicate suspected microdeletion or other subtle chromosomal disorder on the Requisition for Chromosome Analysis form.

REFERENCE VALUE: 46,XX or 46,XY

TURN AROUND TIME: 2- 4 weeks

CPT CODES: 88230 - tissue culture for peripheral blood lymphocytes
88262 - chromosome analysis, congenital disorder
88289 - chromosome analysis, high resolution
88291 - cytogenetic interpretation and report