

TEST: Chromosome analysis, skin

DESCRIPTION: In situ culture of skin fibroblasts to produce metaphase cells for Giemsa-banded chromosome analysis. Cytogenetic examination includes detailed analysis 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.

INDICATIONS:

- suspected mosaic chromosome abnormality
- suspected chromosome abnormality despite a normal peripheral blood karyotype

Skin fibroblasts can also be cultured for other tests such as biochemical or molecular analyses as indicated by the clinical situation.

SPECIMEN REQUIREMENTS: 3 or 4 mm diameter full thickness punch skin biopsy collected under sterile conditions and placed in sterile tissue culture transport. A full thickness slice of skin taken from a surgical incision site can also be used. Label tube or container with patient's name and medical record number. May be refrigerated overnight. **Specimen must not be frozen. Specimen must not be fixed in formalin.** Send to CompGene at room temperature as soon as possible after tissue collection. Indicate the reason for chromosome analysis of skin fibroblast culture on the Requisition for Chromosome Analysis form.

REFERENCE VALUE: 46,XX or 46,XY

TURN AROUND TIME: 10-14 days

CPT CODES: 88233 - tissue culture for skin  
88262 - chromosome analysis, skin fibroblasts  
88291 - cytogenetic interpretation and report