Chromosome Analysis: Targeted Family Member Study

**Test Code:** FS  
**Turnaround time:** 7 days  
**CPT Codes:** 88230 x1, 88261 x1, 88291 x1

### Condition Description

This test is designed to look for a specific chromosomal abnormality that has been previously described in a family member. This is a limited cytogenetic study of a small number of cells and does not constitute a full cytogenetic analysis. Therefore it does not exclude the (unlikely) possibility that other types of chromosomal abnormalities may be present.

### Indications

This test is indicated for individuals who may be at risk of carrying a chromosome abnormality already identified in one or more family members. A targeted chromosome analysis is performed to look for the specific abnormality. Records **must** be available for comparison from the original study. For those cases in which little or no information is available from the original study, routine chromosome analysis is recommended.

### Methodology

PHA stimulated cultures are used for G-banded analysis of a limited number of cells (typically 5 cells; a full chromosome analysis includes analysis of 20 cells). Results are reported using ISCN nomenclature.

### Detection

A minimum band resolution of 550 is required.

### Specimen Requirements

**Additional Specimen Collection/Handling Instructions Required for this Test**

Previous detection of a chromosomal imbalance visible by a standard karyotype is required. Please include the previous report and karyotype (when available) with the sample submission.

**Type:** Whole Blood

**Specimen Requirements:**

- In sodium heparin (green top) tube:  
  - Infants (2 years): 3-5 ml  
  - Older Children & Adults: 5-10 ml

**Specimen Collection and Shipping:** Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

### Related Tests

- Chromosome Analysis (CA)