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)