FISH Analysis with Culture

Test Code: PA

Turnaround time: 7 days - 10 days (3-4 week TAT for custom probes)

CPT Codes: 88230 x1, 88271 x2, 88273 x1, 88291 x1

Condition Description

Genes

HMGC

Indications

Fluorescence In Situ Hybridization (FISH) may be used to confirm and/or detect microdeletions and duplications identified by microarray or G-banded chromosome analysis. This test is appropriate for affected individuals with a cytogenetic finding that needs confirmatory testing. This test is also appropriate for parents and other relatives of an individual with a cytogenetic abnormality confirmed by FISH.

Methodology

FISH analysis is performed on cultured peripheral blood lymphocytes using a probe corresponding to a specific microdeletion/duplication region identified by microarray or G-banded chromosome analysis.

EGL performs FISH analysis using both standard and custom probes. EGL's standard probes are listed below. EGL can also create custom probes for any region of the genome to be used for microarray confirmation or family studies.

- Wolf-Hirschhorn (4p-)
- Cri-du-Chat (5p-)
- Williams (7q11.2 del)
- Prader-Willi/Angelman
- Smith-Magenis (17p11.2 del)
- Miller-Dieker, lissencephaly (17p13)
- 17q21 Microdeletion Syndrome
- 22q11.2 deletion, DiGeorge/VCF

Detection

Validation for specificity and sensitivity performed on each probe. Control probes are present in all probe sets.

Specimen Requirements

Type: Whole Blood

Specimen Requirements:

5 ml blood in a sodium heparin (green top) tube.

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

Special Instructions

Please submit a copy of the cytogenetic report with the sample.

Related Tests

- Chromosomal Microarray: [EmArray Cyto (VA), EmArray Cyto + SNP (CMSNP), EmArray Cyto POC (CMPOC), and EmArray Cyto Prenatal (CMPRE)]
- Chromosome Analysis: [blood (CA, CB), skin (CSKNC), CVS (CV), amniotic fluid (AD), POC (CO), PUBS (CP)]