

Microdeletion Syndromes (fluorescence *in situ* hybridization)

DESCRIPTION:

- This test applies to Wolf-Hirschhorn Syndrome, Cri du Chat, Williams Syndrome, Retinoblastoma, Prader-Willi/Angelman Syndrome, Miller-Dieker Syndrome, Smith Magenis Syndrome, DiGeorge/Velocardial Syndrome, X-linked Ichthyosis, Kallmann's Syndrome, and Sex determining Y.
- Confirms/identifies deletions below the resolution of cytogenetics.

METHOD OF ANALYSIS:

- Metaphase fluorescence *in situ* hybridization
- Detects deletions of specific chromosome regions. Cannot rule out the presence of other chromosome abnormalities.
- For Angelman and Prader-Willi Syndrome, more comprehensive testing is available by DNA methylation studies. Pertinent clinical diagnosis should be included with specimen. For leukemia/lymphoma studies please refer to Oncology Fluorescence *in situ* hybridization.

SAMPLE REQUIREMENTS:

- Fixed-cell pellet from a cytogenetic analysis. Or collect 1-10 ml. of blood into a green top (sodium heparin) tube. Invert the tube several times to prevent coagulation. A charge (CPT 88237) for lymphocyte culture will be applied if chromosome studies have not been ordered. It is recommended that FISH studies be done in conjunction with routine cytogenetic studies.
- Maintain blood specimen at room temperature. Do not freeze or refrigerate.

TEST CPT CODES:

- CPT 88271 Probe
- CPT 88273 Met analysis 10-30 cells
- CPT 88291 Cyto and molecular interpretation and report

Discounts from list price are available for institutional billing under contractual arrangement with the laboratory. Contact Ellen Livers at 800-447-6614 ext 7523.