## *Medical Genetics*

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**Test: Microdeletion Syndromes - Amniotic Fluid/CVS/Peripheral Blood**

*Fluorescence In Situ Hybridization (FISH)*

**Purpose of Test:** To identify chromosome anomalies such a microdeletions/ duplication not identifiable by routine cytogenetic analysis.

**Required Information:** Please provide indications (ICD-9) for testing on our referral form with each specimen. Accurate testing and interpretation may otherwise be compromised.

**Note:** Routine cytogenetic testing is **strongly recommended** to identify the possibility of other clinically associated chromosome abnormalities. In addition, an accurate FISH analysis typically requires metaphase chromosomes thus requiring cell culturing. However, FISH analysis can be performed on interphase cells in special cases.

- **Amniotic Fluid (AF)** - see cytogenetics section for collection information.
- **Chorionic Villus Sample (CVS)** - see cytogenetics section for collection information.
- **Peripheral Blood (PB)** - see cytogenetics section for collection information.
- **Stored Pellets** - Samples on previously studied samples for cytogenetic analysis are kept for 6 months. These samples may be used for FISH analysis. After that time, an additional sample from the patient will be required.

**Reflex Testing:** Routine cytogenetic analysis is strongly suggested

**Probes:**

| Disorder | Deleted Region | Disorder | Deleted Region |
|---|---|---|---|
| Wolf Hirschhorn | 4p | Miller-Dieker | 17p13 |
| Cri du Chat | 5p | Smith-Magenis | 17p11 |
| Williams | 7q22 | DiGeorge/VCF | 22q11 |
| Retinoblastoma | 13q14 | STS | Xp22.3 |
| Angelman | 15q12 | Kallmans | Xp22.3 |
| Prader-Willi | 15q12 | SRY | Xp22.3 |

**Turn Around Time:** 2-4 days

**CPT Code Information:**

88271x2 DNA probe, each
88273 Chromosomal in situ chromosomal hybridization analyze 10-30 cells