## *Medical Genetics*

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# Test: Prenatal Aneuploidy Detection - Amniotic Fluid/CVS/PUBS

*Fluorescence In Situ Hybridization (FISH)*

**Purpose of Test:** To rapidly identify one of the primary chromosome aneuploidies involving chromosomes 13, 18, 21, X, and/or Y. This analysis allows the identification of the following chromosomal syndromes: Down syndrome - trisomy 21; Edwards syndrome - trisomy 18; Klinefelter syndrome - 47,XXY; Patau syndrome - trisomy 13; Turner syndrome - 45,X; other sex chromosome aneuploidies including 47,XXX and 47,XYY; and to resolve cases of ambiguous genitalia.

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

**SUBMIT ONLY 1 OF THE FOLLOWING SPECIMENS:**

**Amniotic Fluid** - see cytogenetic section for collection information.

**Chorionic Villus** Sample - see cytogenetic section for collection information.

**Fetal Blood or PUBS** - see cytogenetic section for collection information.

**Reflex Testing:** Routine Chromosome analysis
Confirmation will be performed on metaphase chromosomes thus requiring culturing of cells.

**Turn Around Time for FISH:** 6-18 hours
**Probes:** 13q14(Rb1), 18cen(D18Z1), 21q22.13-q22.2(D21S259,D21S341,D21S342), Xcen(DXZ1), Ycen(DYZ3)

**CPT Code Information:**

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|---|---|
| 88271x5 | DNA probe, each |
| 88274x5 | Interphase in situ hybridization, analyze 25-99 cells |