

Medical Genetics

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**Test: Aneuploidy Detection - Peripheral Blood/Bone Marrow/Skin
Biopsy/POC**

Fluorescence In Situ Hybridization (FISH)

Purpose of Test: To rapidly identify one of the primary chromosome aneuploidies involving chromosome 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.

Note: In cases where metaphase cells are necessary for accurate analysis, cells will be cultured.

Peripheral Blood (PB) – see cytogenetic section for collection information.

Bone Marrow (BM) – see cytogenetic section for collection information.

Autopsy Tissue/Skin Biopsy – see cytogenetic section for collection information.

Products of Conception (POC) – see cytogenetic section for collection information.

Stored Pellets

Reflex Testing: **Routine cytogenetic testing is strongly recommended.**

Turn Around Time: 3-5 hours

Probes: 13q14(Rb1), 18cen(D18Z1), 21q22.13-q22.2(D21S259,
D21S341,D21S342), Xcen(DXZ1), Ycen(DYZ3)

CPT Code Information:

88271x5 DNA probe, each

88274x5 Interphase in situ hybridization, analyze 25-99 cells