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## ***Test Update: Constitutional Chromosome Analysis***

### **Overview**

**June 8, 2010**

Associated Pathologists / PathGroup Labs are pleased to announce the addition of a new cytogenetic test, chromosome analysis of peripheral blood. Chromosome analysis is the microscopic examination of G-banded chromosomes in dividing cells. Such analysis can detect changes in chromosomal number and structure as well as be used for the diagnosis of constitutional (congenital) chromosome aberrations. The incidence of chromosome abnormalities is relatively common in the population. These chromosome abnormalities may be a genetic cause for infertility, miscarriage, stillbirth, ambiguous genitalia, developmental delay and/or mental disability, and congenital anomalies.

**Methodology:** Microscopic examination of G-banded chromosomes in dividing cells.

**Turnaround Time:** 5-9 days.

**CPT Codes:** 88230, 88262, 88280, 88291

### ***Specimen Requirements, Shipping and Handling:***

A blood sample is collected in a sodium-heparin tube. **Children to Adults:** 3-5 ml of blood. **Newborn:** 1-3 ml of blood. Transport at room temperature, preferably within 24 hours of collection. Specimens may be stored at room temperature or at 4°C until transported to the laboratory.

**Reference Ranges:** The normal reference range is Normal karyotype: 46,XX (female) or 46,XY (male).

### ***Reference:***

Barch M., Knutsen T., Spubeck J. Editors. The AGT Cytogenetics Laboratory Manual, 3<sup>rd</sup> Ed. Lippincott-Raven Publishers. PA, 1997.

Rooney, DE. Human Cytogenetics: Constitutional Analysis: A practical approach. Oxford Press, 2001. Chapter 2.

**For further questions, please contact Client Services at 615-562-9300 or 1-888-474-5227.**