

## Chromosome Analysis

**Also known as:** Karyotyping by G-banding

**Clinical Utility:**

Chromosome abnormalities are a common cause of genetic disease, the leading known cause of intellectual disability, and occur in approximately 1/150 live births. There are two major classes of chromosome abnormalities: numerical abnormalities which include aneuploidy (loss or gain of whole chromosomes) and polyploidy (gain of 1 or more entire haploid sets of chromosomes); and structural abnormalities (translocations, inversions, deletions, duplications). Chromosomes are stained by G-banding and arranged in a karyotype for analysis of the number and structure of the individual chromosomes.

Chromosomes can be analyzed on prenatal samples such as amniotic fluid, chorionic villus samples (CVS), or percutaneous umbilical blood samples (PUBS) to determine the fetal karyotype. Chromosome analysis can also be performed on products of conception specimens. Furthermore, postnatal analysis of peripheral blood samples to determine the karyotype in individuals with multiple congenital abnormalities and/or mental retardation, as well as a variety of other clinical indications, can also be performed. Chromosome analysis is an excellent complement to whole genome or targeted array comparative genomic hybridization (aCGH) since it can detect balanced chromosome abnormalities not detectable by aCGH, and can sometimes clarify the mechanisms of chromosome rearrangements detected by the array.

**Test Sensitivity:**

Typically, structural abnormalities <5Mb in size cannot be visualized by G-band chromosome analysis. Mosaicism for additional low frequency cell line(s) present may be missed. If mosaicism for a specific chromosomal abnormality is suspected, a study with analysis of additional cells can be ordered.