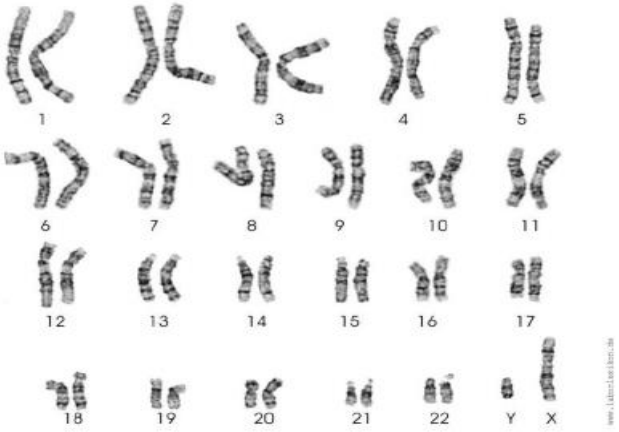
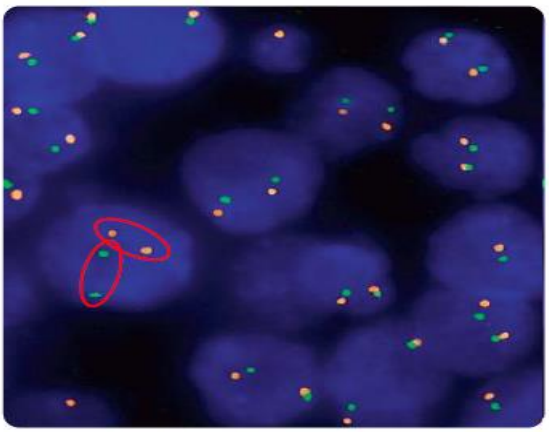
The FML Clinical Cytogenetics Laboratory offers comprehensive cytogenetic testing, including standard chromosome analysis (karyotyping), fluorescence in situ hybridization (FISH), and array CGH. Cytogenetic studies show the significant contribution of chromosome aberrations to infertility, pregnancy loss at all stages of gestation, developmental disabilities, and congenital malformations in the newborn. The study of human chromosomes also contributed to understanding the relation of genetics to the origin of cancers, the physical mapping of genes, and the prevention of genetic diseases by prenatal testing.

Our cytogenetics laboratory offers constitutional studies from amniotic fluid, chorionic villus samples, tissue biopsies, products of conception, and peripheral blood. A standard peripheral blood constitutional analysis examines 20 Giemsa trypsin-banded (G-banding) metaphase cells. All metaphases are microscopically analyzed at the 550-band level of resolution, where possible, and at least two karyotypes are prepared for each case. When chromosome analysis is requested to rule out certain conditions, such as Turner syndrome or suspected mosaicism, an additional 10-30 cell count and/or special stains will be performed. FISH testing is available as an adjunct to chromosome analysis for a wide range of microdeletion syndromes, such as DiGeorge, Prader-Willi, Angelman, and others. FISH is used to clarify chromosomal rearrangements and identify the origin of marker chromosomes.



Human genome: **diploid, 22, XX(Y)**



Normal interphase nucleus:
2 signals /chromosome pair