Objectives

• Describe (in a general way) the basic cytogenetics procedure.
• Be familiar with the layout and organization of a typical karyotype.
• Understand the numbering system for G-banded chromosomes (region, band, sub-band, etc.)
• Describe what FISH is, and compare and contrast it with traditional cytogenetics (karyotyping).
Genetic tests are used in a wide variety of clinical settings.

- Diagnostic testing
- Carrier testing
- Predictive and presymptomatic testing
- Preimplantation testing
- Prenatal testing
- Newborn screening
- Pharmacogenomic testing
- Other stuff (forensics, paternity, genealogy)
Genetic tests can look at:

- Chromosomes
- DNA
- RNA
- Proteins
Q. What is cytogenetics?

A. The study of chromosomes (their structure and inheritance) and how they relate to diseases.
What tissues can you use for cytogenetics?

Pretty much anything that can grow in culture.

- Blood
- Skin
- Bone marrow
- Chorionic villi
- Amniotic fluid
Basic cytogenetics procedure:

1. Draw blood sample.
2. Place the lymphocytes in a tissue culture medium.
3. Wait for them to divide, then arrest in metaphase.
4. Take chromosomes out, fix, put on slide, stain.
Chromosomes in metaphase

Nucleus from adjacent cell still in interphase
Normal karyotype (artistic version)
Normal karyotype (real life version)
Chromosome banding
Every chromosome has a unique G-band pattern
G-banding of chromosome 7: low, medium, and high resolution
Chromosomal location: 7 q 3 1 . 2

How to describe the location of a gene
Fluorescent in-situ hybridization (FISH)
FISH: chromosome 9 = green, chromosome 22 = red
FISH: chromosome 18 = blue, X = green, Y = red