## Genetic Disorders:

Mistakes in Meiosis – extra, missing or mixed up chromosomes

→autosomal aneuploidy – having an unusual number of chromosomes 1-22; usually fatal

*Trisomy* – have three of a particular autosome instead of just two results from nondisjunction (homologous chromosomes fail to separate)

Examples: 1. trisomy 8
2. trisomy 13 (Patau syndrome)
3. trisomy 18 (Edward syndrome)
4. **trisomy 21 (Down syndrome)**

→ sex chromosome aneuploidy – missing or extra sex chromosome(s)

**Examples: 1. Turner Syndrome - XO**
**→ females who lack ovaries & sex characteristics; short & sterile**
**2. Klinefelter Syndrome - XXY**
**→ taller, longer limbs, sterile, some exhibit mental retardation**
**3. XYY male – 1 in every 1000; normal, fertile, taller than average**

How to Study: take sample cells from fetus & photograph **metaphase** chromosomes – line them up according to length & location of centromere = **karyotype**

A. Dominant Autosomal Heredity

**Huntington's disease** – a rare genetic disorder caused by a dominant gene
→ progressive degeneration of the nervous system causes uncontrolled jerky movements & mental deterioration
→ no effective treatment exists
→ passed on because symptoms don't occur until 30-50, after they have children
→ if you are at risk, you can find out if you are a carrier with a biochemical test

B. Recessive Autosomal Heredity – most genetic disorders caused this way; rare except in some ethnic groups

1. **cystic fibrosis** – most common lethal genetic disorder among Americans
→ 1 in 20 carry the recessive allele; 1 in 2000 has it
→ thick mucous in lungs makes breathing difficult; frequent lung infections
→ thick mucous also slows the secretion of some digestive enzymes
→ physical therapy, special diets, & new drug therapies have raised average life span

2. **sickle-cell anemia** – a blood disorder
→ most common in black Americans (Africa) or white Americans (Mediterranean Sea)
→ red blood cells are shaped like a sickle or half moon – can clog blood vessels
→ rbc's have a shorter life span – people suffer from anemia
*hemoglobin protein in these cells differs from normal by one amino acid
→ treatments include transfusions & drug therapy
→ hetero's can show signs if oxygen availability is low

3. **Tay-Sachs disease** affects the nervous system
→ missing an enzyme that normally breaks down a lipid produced & stored in the central nervous system & it accumulates in the cells
→ results in blindness, progressive loss of movement & mental deterioration
→ symptoms occur within the first year of life & result in death by age 5; no treatment
→ allele common in PA Dutch

4. **phenylketonuria** is a treatable genetic disorder (Norway, Sweden)
→ absence of an enzyme that converts one amino acid, phenylalanine, to tyrosine
→ phenylalanine accumulates & can cause severe damage to the central nervous system
→ newborns appear normal since mother's normal enzyme level prevents accumulation, but when newborn begins drinking milk (high in phenylalanine) accumulation begins
→ PKU tests are now preformed on all newborns a few days after birth to prevent retardation