

## Chromosomal Basis of Inheritance

## I. Chromosomes Contain Genes

**A. Chromosomal Theory of Inheritance**

- Independently proposed in 1902 by Theodor Boveri and Walter S. Sutton.
- Genes are located on chromosomes.

**B. Chromosomes can be categorized as two types**

- Autosomes
  - Nonsex chromosomes
  - Same number and kind between sexes.
- Sex chromosomes
  - Human female is XX
  - Human male is XY.
  - Carry genes for traits unrelated to sex.

## I. Chromosomes Contain Genes

**C. Thomas Hunt Morgan confirmed genes were on chromosomes.**

- Work with fruit flies showed XY system similar to humans.
- Newly discovered mutant male fruit fly had white eyes.
- Cross of white-eyed male with hetero red-eyed female
  - Didn't produce expected results.
  - Yields 3:1 red-to-white ratio
  - All white-eyed flies were males
- An allele for eye color on the X but not Y chromosome supports the results of the cross.

	w	w			
W	Ww	Ww	H	Hh	Hh
w	Ww	Ww	h	hh	hh

## II. Genes Are Linked

**A. Fruit flies have four pairs of chromosomes**

**B. Sutton realized each chromosome must hold many genes.**

**C. All alleles on a chromosome make up a linkage group.**

**D. Linked alleles do not obey Mendel's laws because they go into the gametes together.**

**E. Except when crossing over occurs**

- In a dihybrid cross a heterozygote forms only two types of gametes and produces offspring with only two phenotypes.
- Crossing-over causes recombinant gametes and at fertilization, recombinant phenotypes.

## II. Genes Are Linked

**F. Mapping the Chromosomes**

- Linked genes can show the distance between genes on the chromosomes.
- Percentage of recombinant phenotypes measures distance between genes to map the chromosomes.
- If 1% of crossing-over equals one map unit, then 6% recombinants reveal 6 map units between genes.
- If crosses are performed for three alleles on a chromosome, only one map order explains map units.

gene pair	Recombination frequency c
A - D	18.5 %
A - B	13.2 %
B - D	6.4 %

$\frac{A}{\quad} \xrightarrow{13.2\%} \frac{B}{\quad} \xrightarrow{6.4\%} \frac{D}{\quad}$

Note  $c(A,B) + c(B,D) = 19.6\% > c(A,D)$

## III. Chromosomes Undergo Mutations

**A. Mutations increase variation among offspring.**

**B. Nondisjunction**

- Changes Chromosome Number
- Failure of chromosomes to separate
- More common during meiosis I than meiosis II
- Can occur in mitosis.

### III. Chromosomes Undergo Mutations

5. Types

- Monosomy: missing one chromosome
- Trisomy: three of one type of chromosome.
- Polyploidy
  - More than two complete sets of chromosomes
  - Create triploids [3n], tetraploids [4n], etc.
  - Does not increase variation in animals
  - A major evolutionary mechanism in plants.

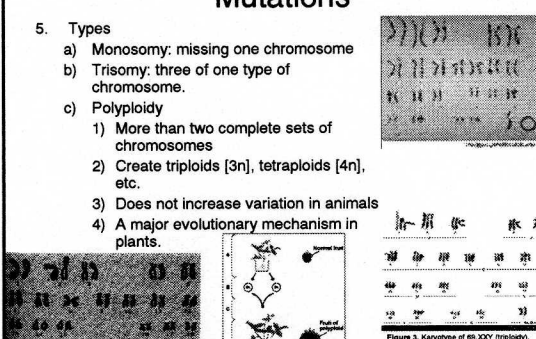


Figure 3. Karyotype of 69,XXX (triploidy), common finding in spontaneous abortion.

### III. Chromosomes Undergo Mutations

C. Changing Chromosomal Structure

- Causes: radiation, chemicals, viruses
- Inversions: segment separates and is reinserted in reverse.
- Translocations: segment separates and inserted into another, nonhomologous chromosome.

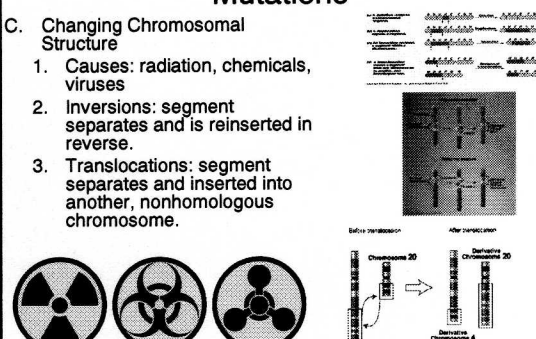
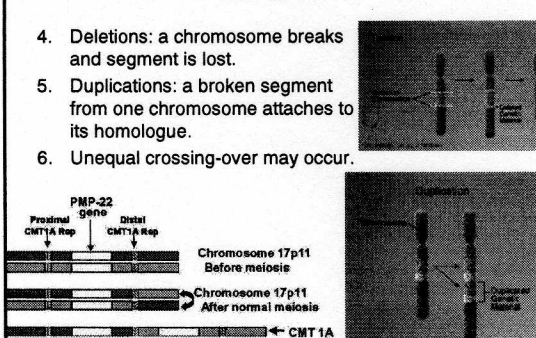


Figure 3. Karyotype of 69,XXX (triploidy), common finding in spontaneous abortion.

### III. Chromosomes Undergo Mutations

- Deletions: a chromosome breaks and segment is lost.
- Duplications: a broken segment from one chromosome attaches to its homologue.
- Unequal crossing-over may occur.



Chromosome 17p11 Before meiosis

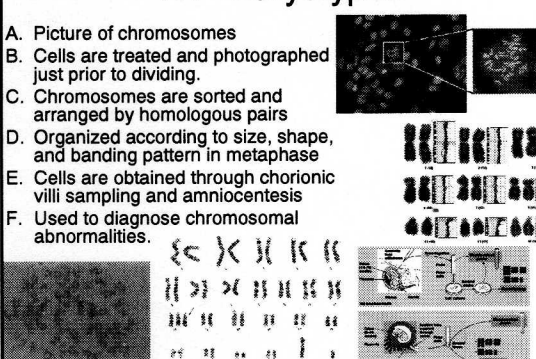
Chromosome 17p11 After normal meiosis

Chromosome 17p11 After unequal crossing-over

Labels: PMP-22 gene, Proximal CMT1A Rep, Distal CMT1A Rep, CMT1A, HNPP Chromosome

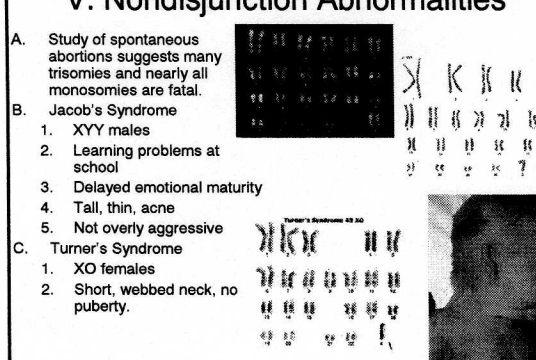
### IV. Karyotypes

- Picture of chromosomes
- Cells are treated and photographed just prior to dividing.
- Chromosomes are sorted and arranged by homologous pairs
- Organized according to size, shape, and banding pattern in metaphase
- Cells are obtained through chorionic villi sampling and amniocentesis
- Used to diagnose chromosomal abnormalities.



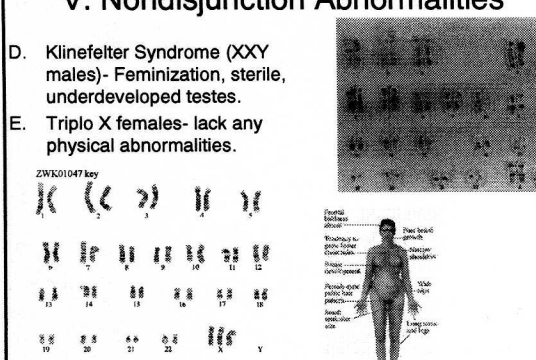
### V. Nondisjunction Abnormalities

- Study of spontaneous abortions suggests many trisomies and nearly all monosomies are fatal.
- Jacob's Syndrome
  - XYY males
  - Learning problems at school
  - Delayed emotional maturity
  - Tall, thin, acne
  - Not overly aggressive
- Turner's Syndrome
  - XO females
  - Short, webbed neck, no puberty.



### V. Nondisjunction Abnormalities

- Klinefelter Syndrome (XXY males)- Feminization, sterile, underdeveloped testes.
- Triplo X females- lack any physical abnormalities.



ZWK01047 key

Distal bands absent

Distal bands present

Washing to give color bands

Stain

Photograph

Mount

Slide

Trisomy 21 and 18

