

### Module 3D – Chromosomes and Inheritance

- Mendel's experiments with pea plants led him to discover the basic laws that govern how "hereditary factors" are transmitted from one generation to the next.
- However, Mendel did not know where these "hereditary factors" are located, what they are made of, or how they work.

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- During the remainder of this unit, we will examine all 3 of these questions.
- In this module, we will focus on some of the research that led scientists to conclude that genes (Mendel's "hereditary factors") are located on chromosomes. We will also look at how this fact affects the transmission of genetic information.

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### Objective # 20

Explain why some genes do NOT assort independently. Also explain how an experiment by Morgan originally demonstrated this.

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### Objective 20

- In the early 20<sup>th</sup> century, Thomas Hunt Morgan studied patterns of inheritance for many traits in the fruit fly, *Drosophila*.
- At one gene locus controlling eye color, Morgan found a dominant allele for red eyes (R) and a recessive allele for purple eyes (r).

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### Objective 20

- At another gene locus controlling wing length, he found a dominant allele for normal wings (N) and a recessive allele for short wings (n).
- Morgan mated a female heterozygous at both gene loci with a male homozygous for both recessive alleles. Based on Mendel's Laws of Inheritance, what is the expected outcome of this cross?

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### Objective 20

- Genotypes of parents:  
➤ ♀ Rr Nn X ♂ rr nn
- Genotypes of gametes:  
➤ ♀ (RN) (Rn) (rN) (rn) X ♂ (rn)
- Genotypes of offspring:

	RN	Rn	rN	rn
rn	RrNn	Rrnn	rrNn	rrnn

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### Objective 20

- If Mendel's Law of Independent Assortment is correct, we expect equal percentages of all 4 genotypes among the offspring. However, that is not what Morgan observed.

genotype	RrNn	Rrnn	rrNn	rrnn
phenotype	red, normal	red, short	purple, normal	purple, short
expected	25%	25%	25%	25%
observed	47%	5%	5%	43%

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### Objective 20

- Why didn't the observed results match the predicted results?
  - The difference might be due to chance.
  - The Law of Independent Assortment might be wrong.
- Which answer is correct?
  - There is no way to be certain, but a statistical test can tell us the probability of getting a difference as large or larger than the one we observed by chance alone.

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### Objective 20

- A statistical test of Morgan's data gives a  $p < 0.01$ . What should he conclude?
- There is a very small probability the difference was caused by chance alone.
- So, what's wrong with Mendel's Law of Independent Assortment?

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### Objective 20

	RN	Rn	rN	rn
rn	RrNn	Rrnn	rrNn	rrnn
expected	25%	25%	25%	25%
observed	47%	5%	5%	43%

- Examination of Morgan's data shows that the female gametes were mostly RN or rn, while very few had the genotypes Rn or rN. Why?

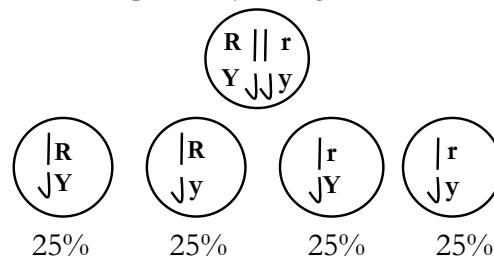
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### Objective 20

- Morgan hypothesized that R and N tend to segregate together because they are both located on the same chromosome, while r and n tend to segregate together because they are both located on the homologous chromosome.
- Genes located on the same chromosome are said to be linked.

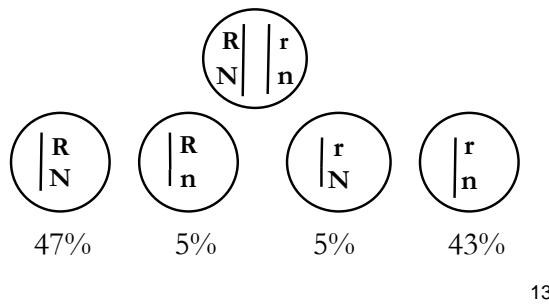
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We now know that unlinked genes (genes located on different chromosomes) assort independently because chromosome pairs assort independently during meiosis:



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On the other hand, linked genes often do not assort independently because they are located on the same chromosome :



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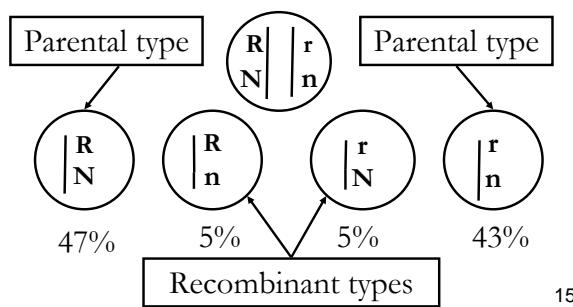
### Objective # 21

Explain how the process of crossing over leads to recombinant types. Explain how the frequency of recombinant types is used by geneticists to construct chromosome maps.

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### Objective 21

- As Morgan demonstrated, linked genes often do NOT assort independently:



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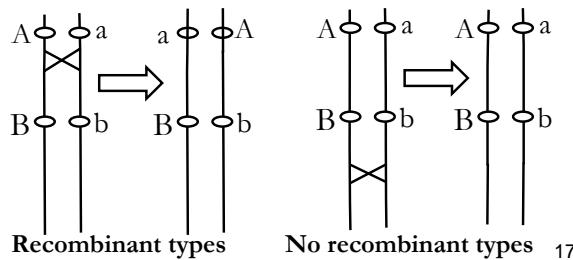
### Objective 21

- However, if R is linked with N and r is linked with n, why do a few gametes have the genotype Rn while others have the genotype rN?
- These recombinant types are produced by crossing over during Prophase I of meiosis.

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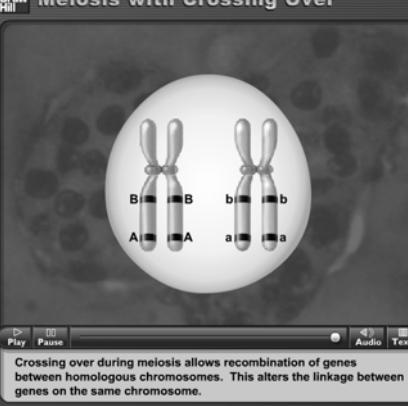
### Objective 21

- Recombinant types are produced only when a crossover occurs between the 2 genes being studied:



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### Meiosis with Crossing Over



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### Objective 21

- Because crossovers occur along the length of a chromosome at random, the farther apart 2 genes are, the larger the chance a crossover will occur between them, and the higher the frequency of recombinant types.
- Therefore, the frequency of recombinant types can be used as a measure of how far apart 2 genes are.

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- This information is used to construct genetic maps. One centimorgan (map unit) is defined as the distance between 2 genes that produces 1% recombinant types.
- Based on this definition, what is the distance between the 2 gene loci Morgan studied:

RrNn	Rrnn	rrNn	rrnn
47%	5%	5%	43%

➤ 10 centimorgans or 10 map units

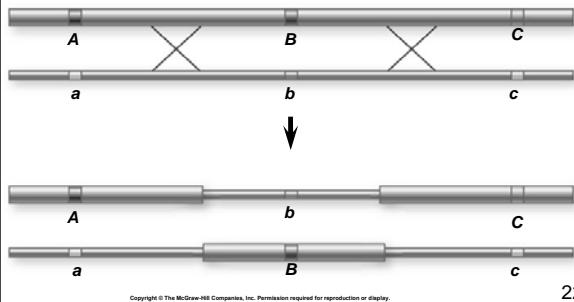
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### Objective 21

- As the distance separating 2 gene loci increases, the probability of multiple crossovers between them also increases.
- While one crossover between 2 genes produces recombinant types, two crossovers between them maintains the parental gene combination.

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Two crossovers between genes A and C maintains the parental gene combination (A with C and a with c):



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### Objective 21

- In general, an odd number of crossovers between 2 genes will produce recombinant types, while 0 or an even number of crossovers produces the parental gene combination.
- At large enough distances, the frequencies of these 2 possibilities are approximately equal.

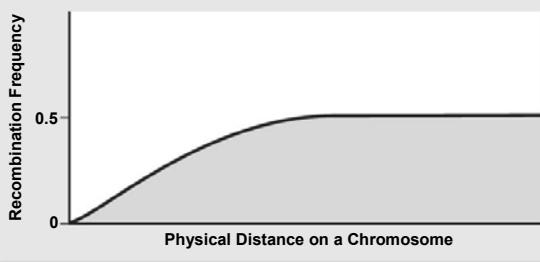
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### Objective 21

- Therefore, if 2 genes are separated by a very large distance, the number of recombinant gametes is approximately equal to the number of parental gametes and the 2 loci will assort independently even though they are located on the same chromosome!

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Genes that are located far apart on the same chromosome will assort independently:



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### Objective 21

- Following the procedures in the class handout, “Predicting the Outcome of Monohybrid and Dihybrid Crosses”, you should now be able to predict the outcome of:
  - monohybrid crosses
  - dihybrid crosses with unlinked genes
  - dihybrid crosses with linked genes and no crossing over.

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### Objective # 22

Describe the process of sex determination in humans and also be able to provide examples of other methods of sex determination.

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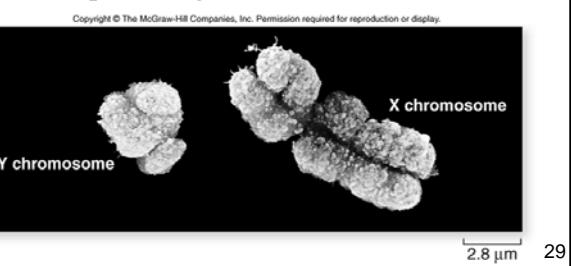
### Objective 22

- In humans, there are 23 pairs of chromosomes; one pair are called sex chromosomes. These 2 chromosomes look identical in females (XX) but different in males (XY).
- All other chromosomes are called autosomes. The 2 autosomes that make up each pair look identical in both males and females.

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### Objective 22

The human Y chromosome is much smaller than the X and contains very few expressed genes.



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### Objective 22

- The “default” setting for human embryonic development is for the production of a female.
- However, a few active genes on the Y chromosome, notably the SRY gene, trigger the development of male genitalia and secondary sex organs.
- Consequently, any individual with at least one Y chromosome is normally a male.

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### Objective 22

- This system of sex determination is shared among mammals, but is not universal.
- In some species of fish and reptiles, for example, environmental factors can determine the sex of individuals.
- There are also other genetic systems for determining sex:

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TABLE 13.1 Sex Determination in Some Organisms

	Female	Male
Humans, <i>Drosophila</i>	XX	XY
Birds	ZW	ZZ
Grasshoppers	XX	XO
Honeybees	Diploid	Haploid

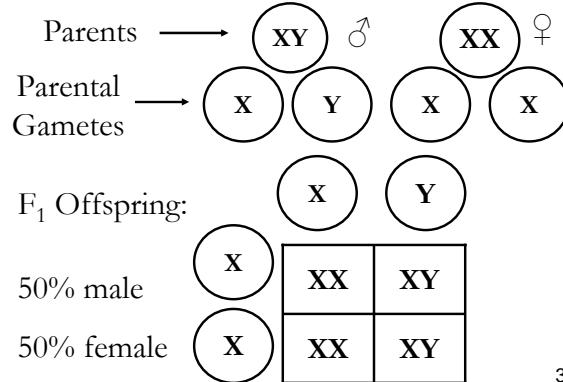
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### Objective 22

- Based on the sex determining mechanism in humans, what is the expected sex ratio among the offspring when we cross a male and female?

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### Objective 22



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### Objective # 23

Explain what sex linkage is, and be able to predict the outcome of crosses involving sex-linked traits.

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### Objective 23

- Other traits, besides sex, are controlled by genes on the sex chromosomes. These are called sex-linked traits.
- Traits controlled by the X are X-linked.
- Traits controlled by the Y are Y-linked.
- Since most sex-linked traits are controlled by the X, you can assume X-linkage, unless it says Y-linked.

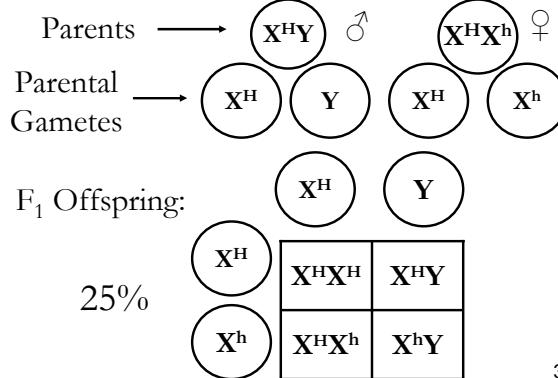
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### Objective 23

- X-linked traits are an exception to Mendel's laws because females have 2 alleles for each X-linked trait, but males have only 1.
- In humans, hemophilia is caused by a recessive allele on the X chromosome. Two normal parents have a son with hemophilia. What is the probability their next child will also have hemophilia?

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### Objective 23



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### Objective # 24

Describe some exceptions to the chromosomal theory of inheritance.

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### Objective 24

- Although most genes in eukaryotes are located on chromosomes within the nucleus, there are some exceptions.
- Primarily, these exceptions are due to the presence of DNA in mitochondria and chloroplasts.
- DNA in these organelles is not partitioned with the nuclear genome by the process of mitosis.

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### Objective 24

- Therefore, traits controlled by genes in the mitochondria and chloroplasts will not show Mendelian inheritance.
- When a zygote is formed, all of the cytoplasm and mitochondria come from the egg cell. Thus, mitochondrial DNA comes solely from the mother. This is called maternal inheritance.

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### Objective 24

- The inheritance pattern for chloroplasts is also usually maternal.
- However, for chloroplasts both paternal (solely from the father) and biparental (from both parents) inheritance have been observed, in different species.

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