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CELS191 LECTURE MATERIAL

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CELS191 2025

Molecular Biology & Genetics

Lecture 20

X-linked Traits & Recombination

Professor Jon Waters
Te Tari Mātai Kararehe | Department of Zoology

Lecture 20 Objectives

After you have revised this lecture you should be able to:

- Identify the inheritance patterns of genes on sex-linked chromosomes.
- Explain that linked genes show non-independent assortment.
- Explain that crossing over leads to recombination of linked genes.

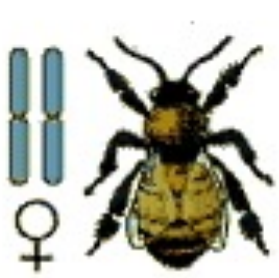
SEX-LINKED TRAITS

- Involving genes on the sex chromosomes (X or Y) (rather than autosomes)
- As a result, their inheritance patterns differ between males (XY) and females (XX)
- Examples: haemophilia; red-green colour blindness

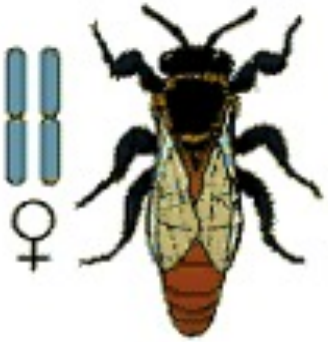
SEX DETERMINATION

Sex chromosomes x,y determine the sex of the individual.

Other chromosomes are called autosomes.



Diploid worker

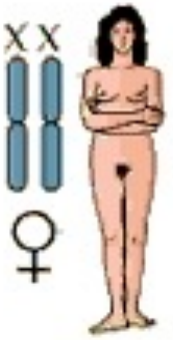


Diploid queen

Haploid male

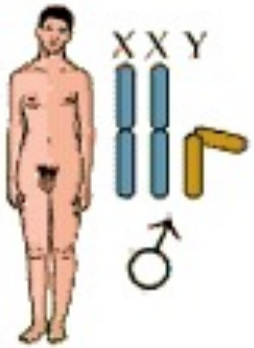
In bees, fertilised eggs develop into diploid females; unfertilised eggs develop into haploid males

Normal



Turner syndrome sterile

Klinefelter syndrome sterile



Normal

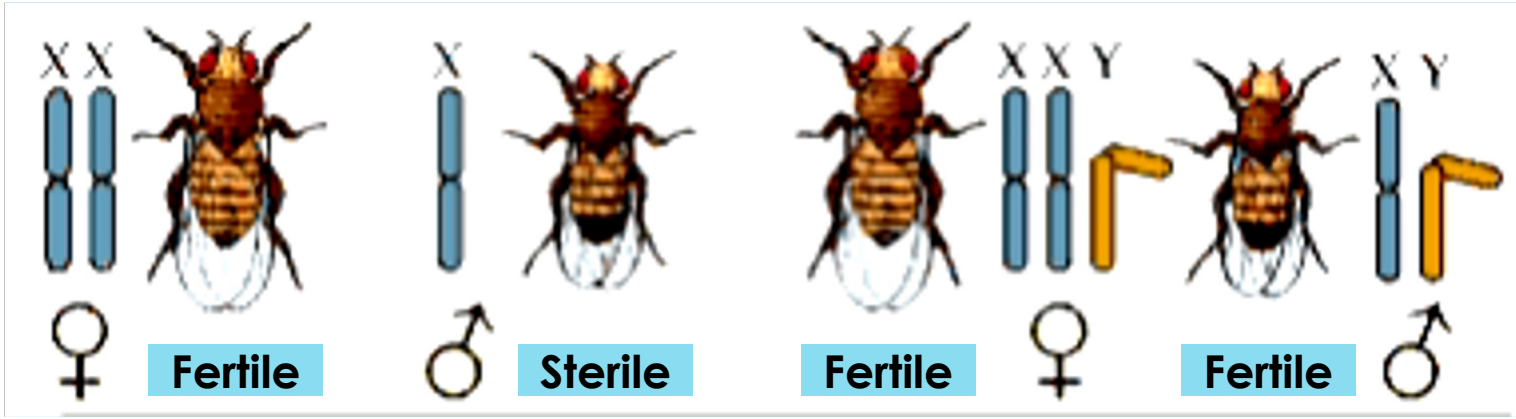
Context Slide

HETEROGAMETIC | **HOMOGAMETIC**

BIRDS

Male birds carry two identical sex chromosomes (ZZ) and females have two different ones (ZW)

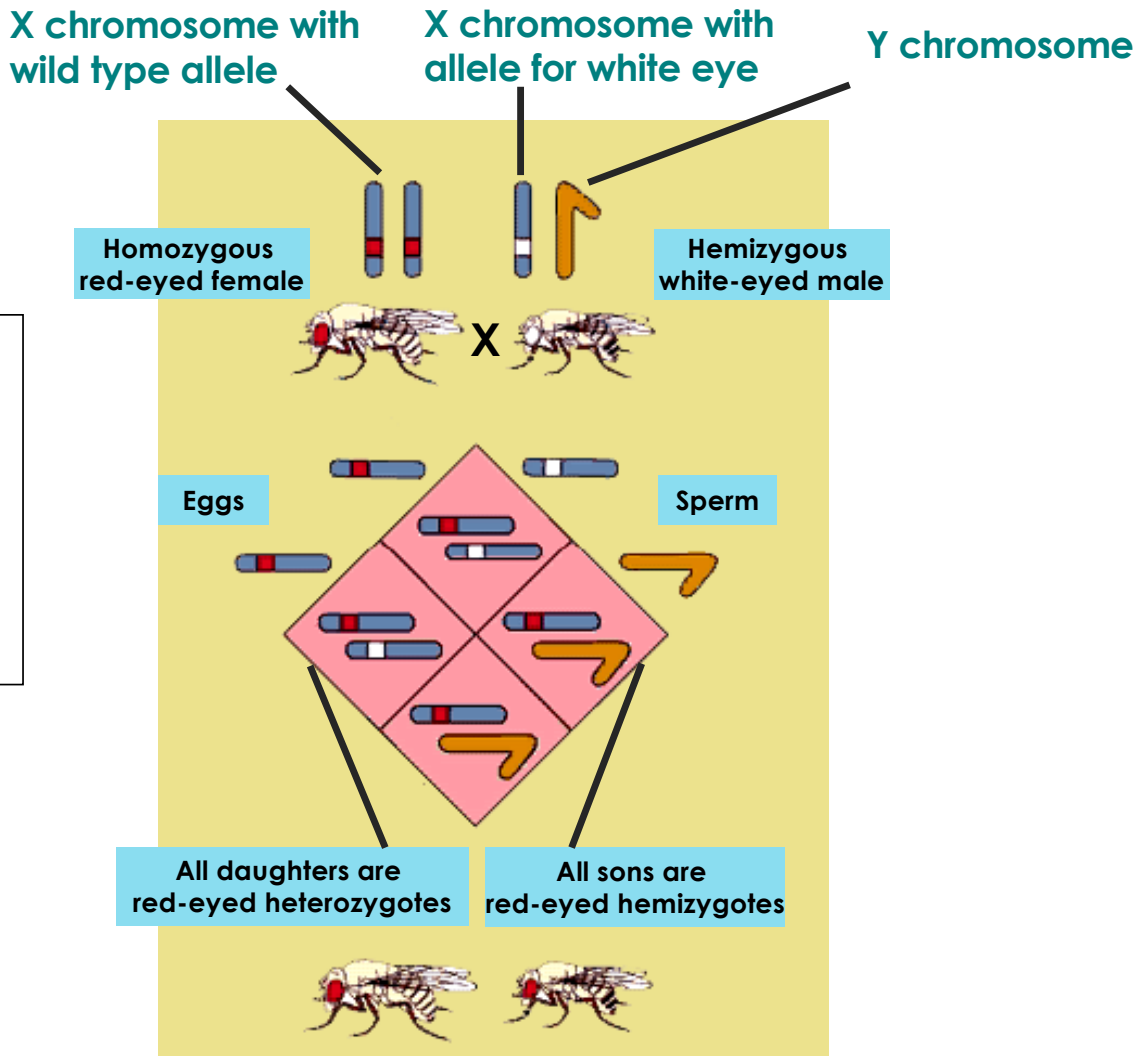
Drosophila



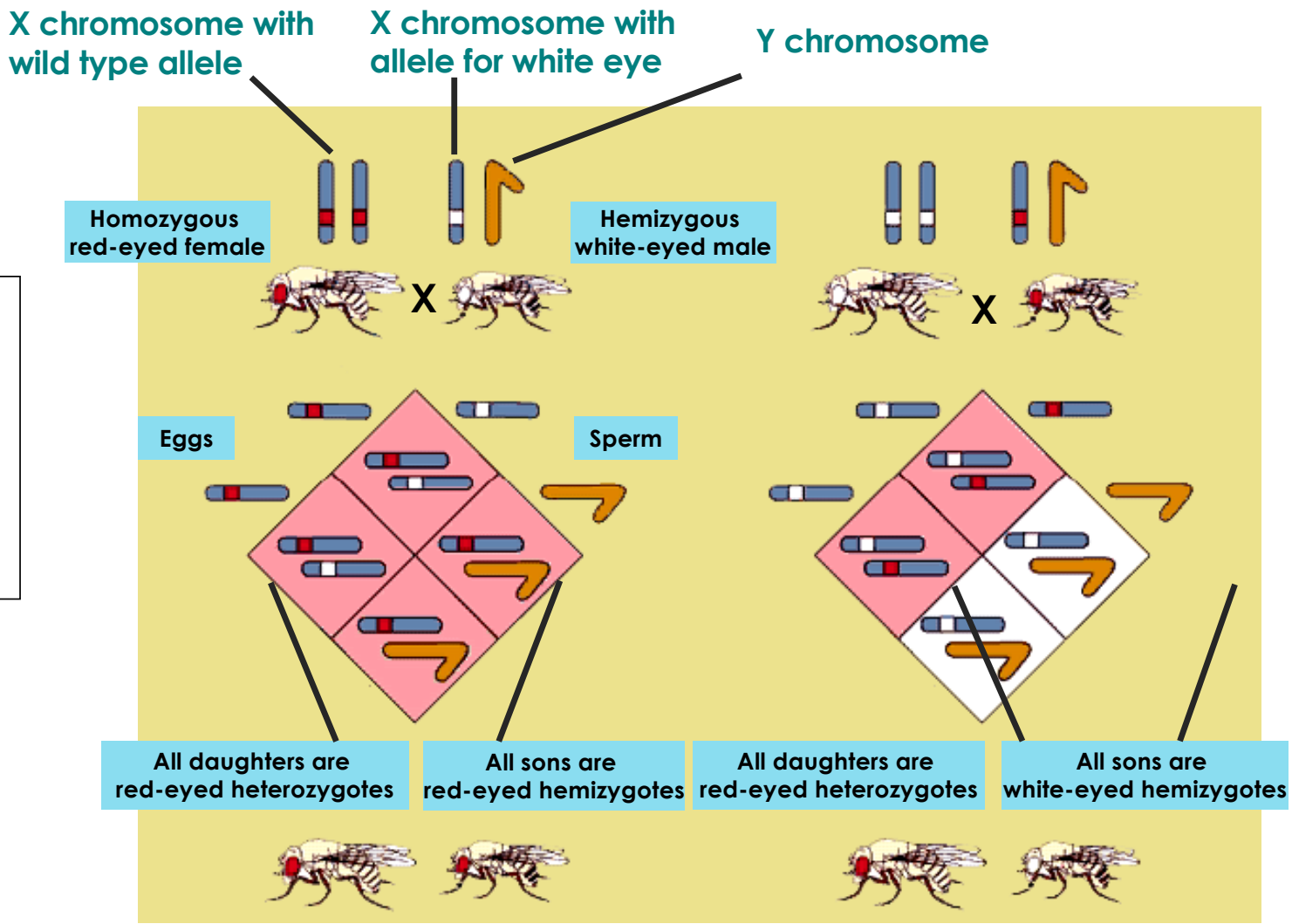
Drosophila **females have two X chromosomes and may also have a Y chromosome; males have an X chromosome and, if they are fertile, a Y chromosome**

Context Slide

Eye Colour is a sex-linked trait in *Drosophila*

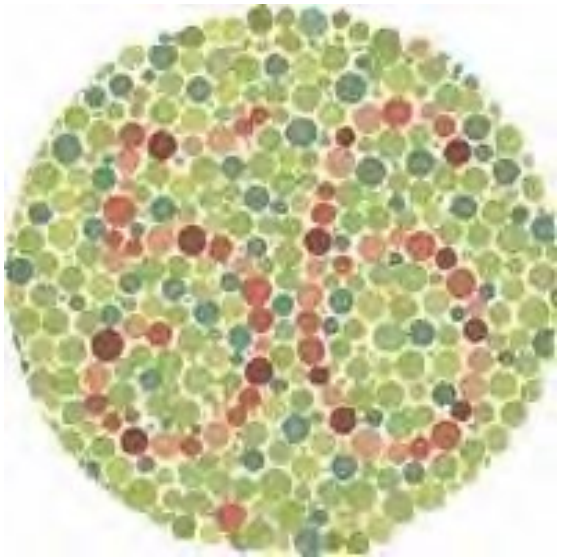


Eye Colour is a sex-linked trait in *Drosophila*

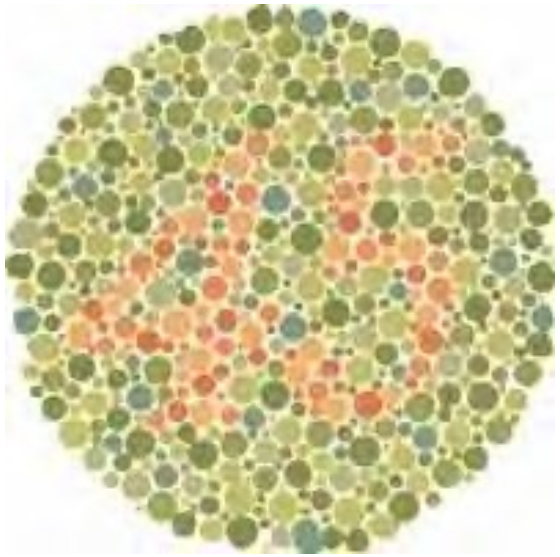


Red-Green colour blindness in humans

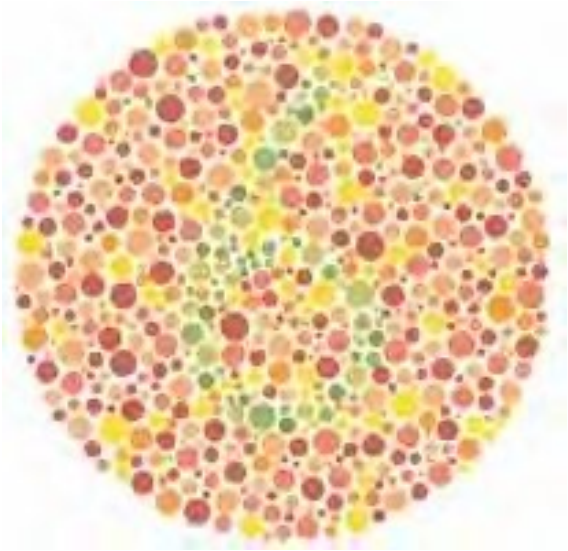
Red-Green colour blindness in humans



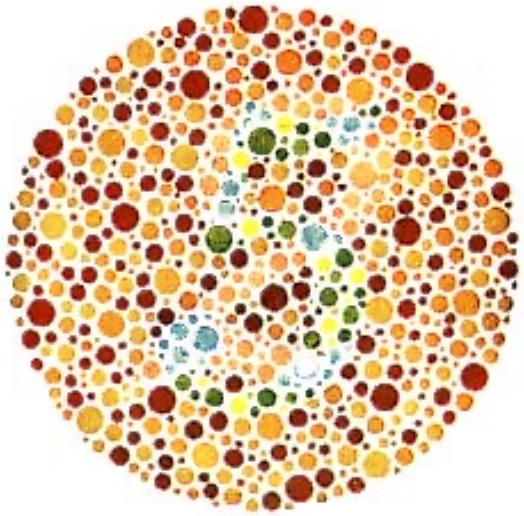
Red-Green colour blindness in humans



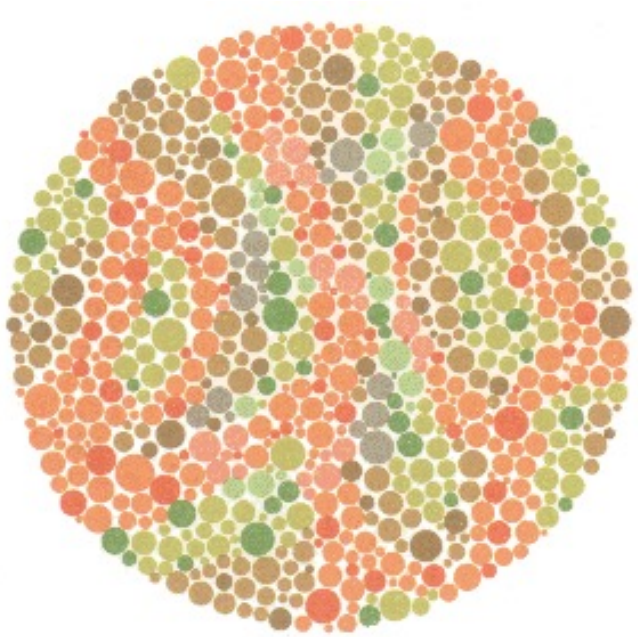
Red-Green colour blindness in humans



Red-Green colour blindness in humans



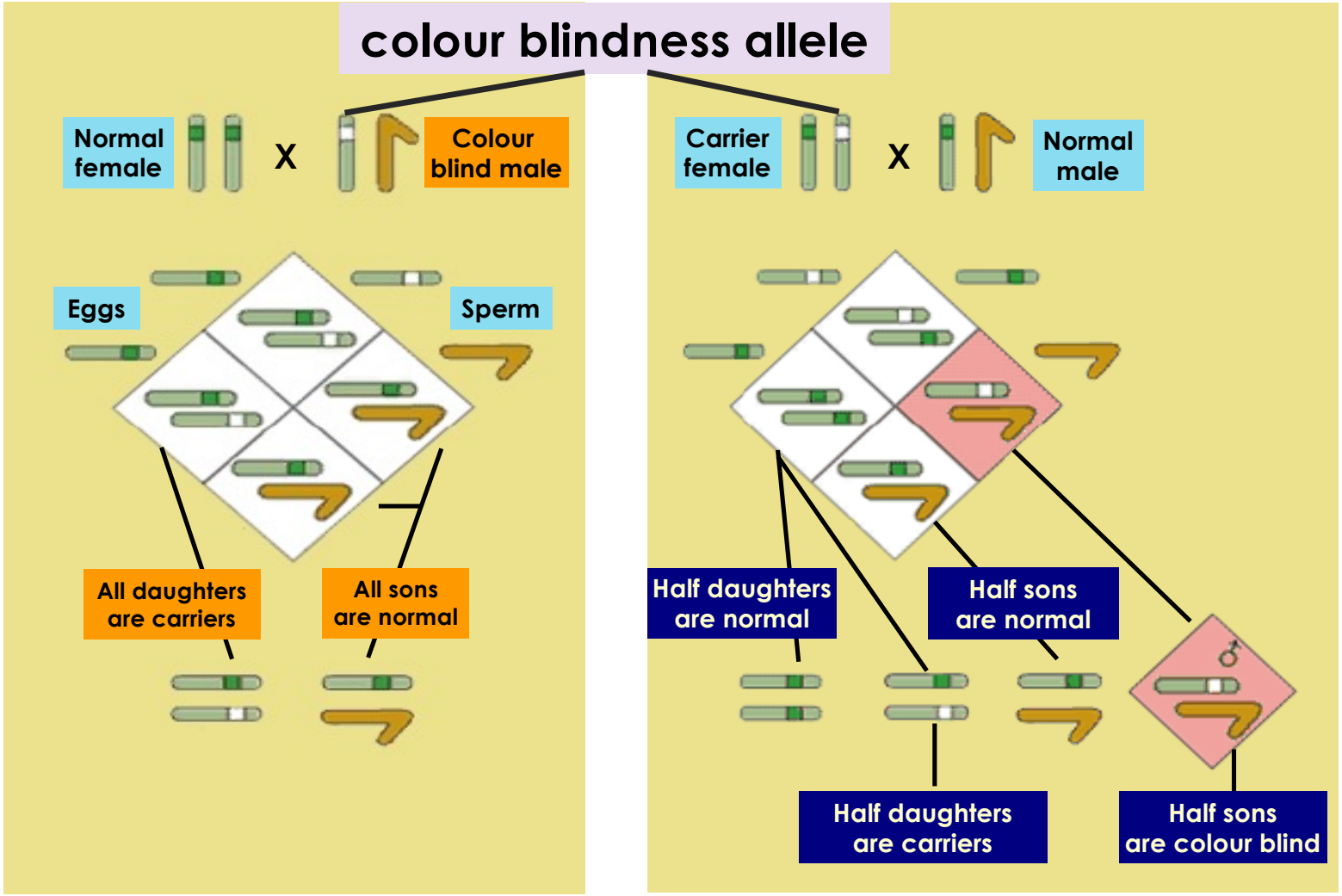
Red-Green colour blindness in humans



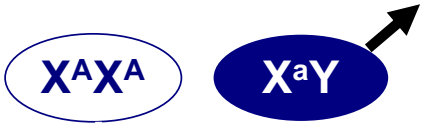
????????

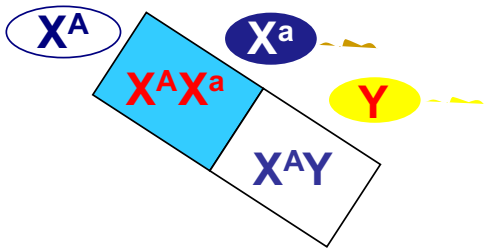
The majority of those with red-green deficiencies should read the number 5

Red-Green colour blindness is a sex-linked trait

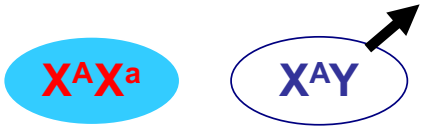


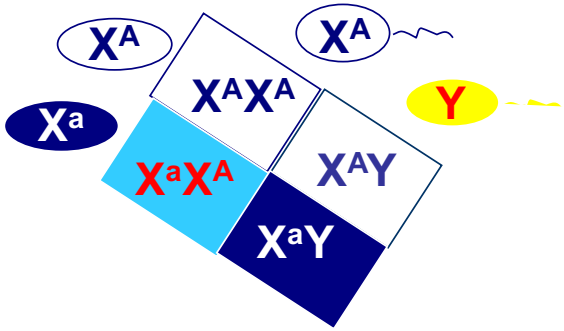
Core Slide



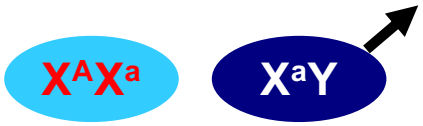


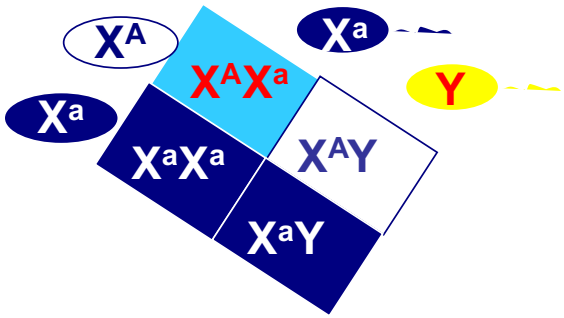
A father with the trait will transmit the mutant allele to all daughters but to no sons. When the mother is a dominant homozygote, the daughters will have the normal phenotype but will be carriers of the mutation.





A carrier who mates with a normal male will pass the mutation to half her sons and half her daughters. The sons with the mutation will have the disorder. The daughters who have the mutation in a single dose will be carriers like their mother.





A carrier mates with a male with the trait, there is a 50% chance that each child will have the trait. Daughters who do not have the trait will be carriers, whereas males without the trait will be completely free of the harmful recessive allele.

Gene Mapping

Q. How many genes are there in humans?

A. Around 20,000

Q. How many chromosomes are there in humans?

A. 23 pairs

That's about 1,000 linked genes per chromosome

Q. How are these linked genes inherited?

A. Linked genes show biases towards parental genotypes

Q. How can we determine the order and location of genes within a chromosome?

A. By using recombination frequencies

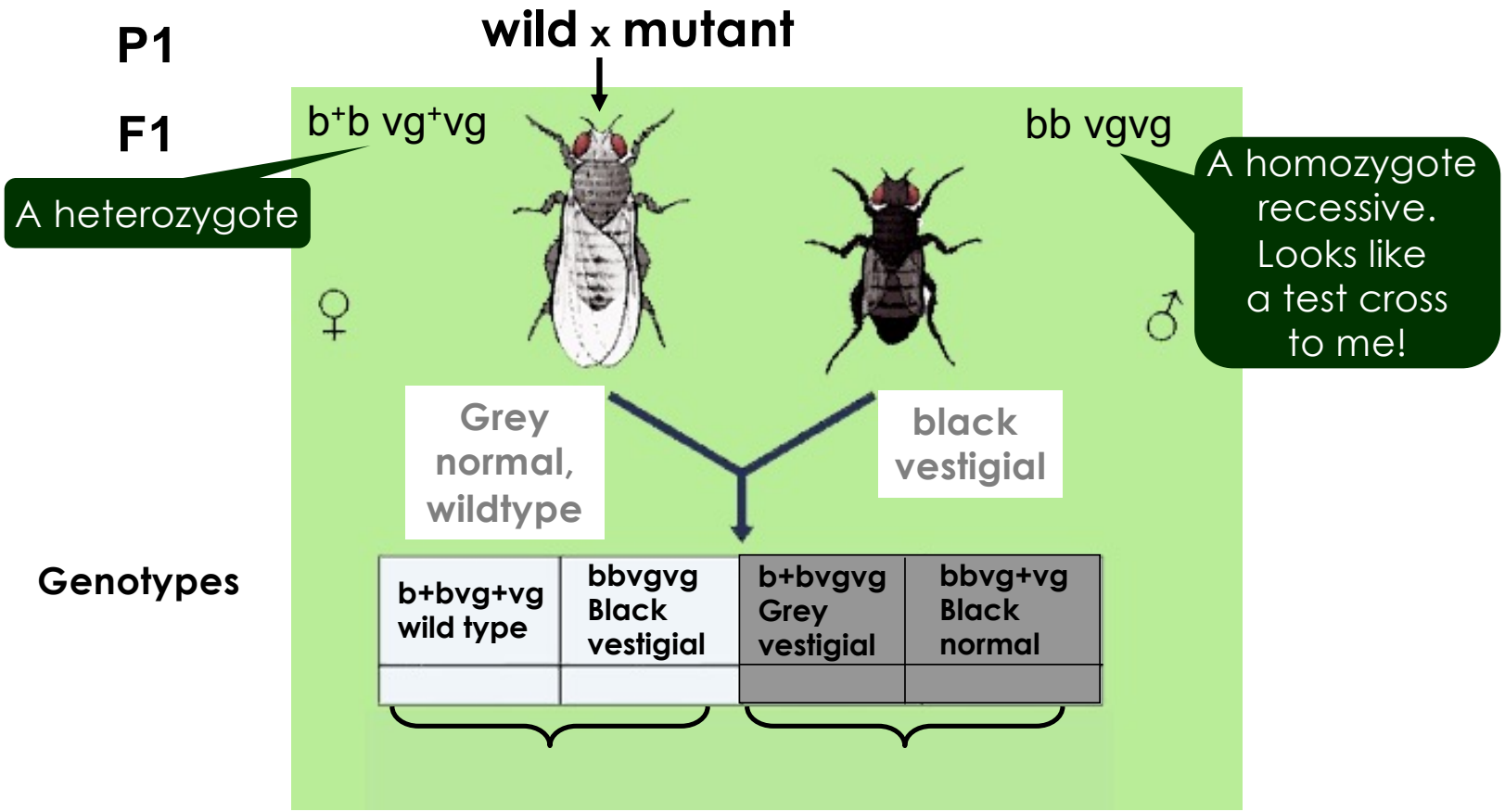
A quick lesson on the convention of how fruit fly (*Drosophila*) genes are symbolized

1. **The first mutant (non-wild type) discovered provides the letter(s). e.g.** 'vg' for vestigial wings

2. **The wild type allele for that gene is given a $^{+}$ after the symbol. e.g.** 'vg^{+}' is wild type wings

3. **vg is recessive to vg^{+}**

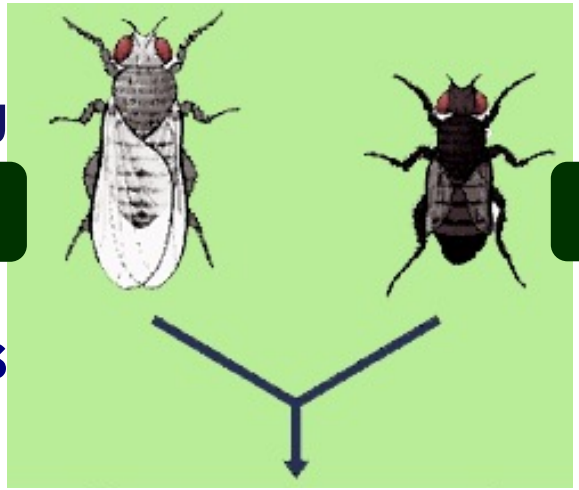
Context Slide



What ratio of phenotypes would we expect?

Context Slide

$b^+b\ vg^+vg$ **WILD TYPE**



bb vgvg BLACK VESTIGIAL

Female gametes

bvg (male gamete)

$b^+\ vg^+$	$b^+b\ vg^+vg$	WILD TYPE
$b^+\ vg$	$b^+b\ vgvg$	GREY VESTIGIAL
$b\ vg^+$	$bb\ vg^+vg$	BLACK NORMAL
$b\ vg$	$bb\ vgvg$	BLACK VESTIGIAL

Expected phenotype ratio 1:1:1:1

P1 wild x mutant

F1 $b^+b\ vg^+vg$ bb vgvg

A heterozygote

A homozygote recessive. Looks like a test cross to me!!

♀ Grey normal, wildtype

♂ black vestigial

Genotypes
2300 offspring

b+bvg+vg wild type	bbvgvg Black vestigial	b+bvgvg Grey vestigial	bbvg+vg Black normal
575	575	575	575

PARENTAL TYPES — RECOMBINANT TYPES

These are the actual results obtained by Morgan

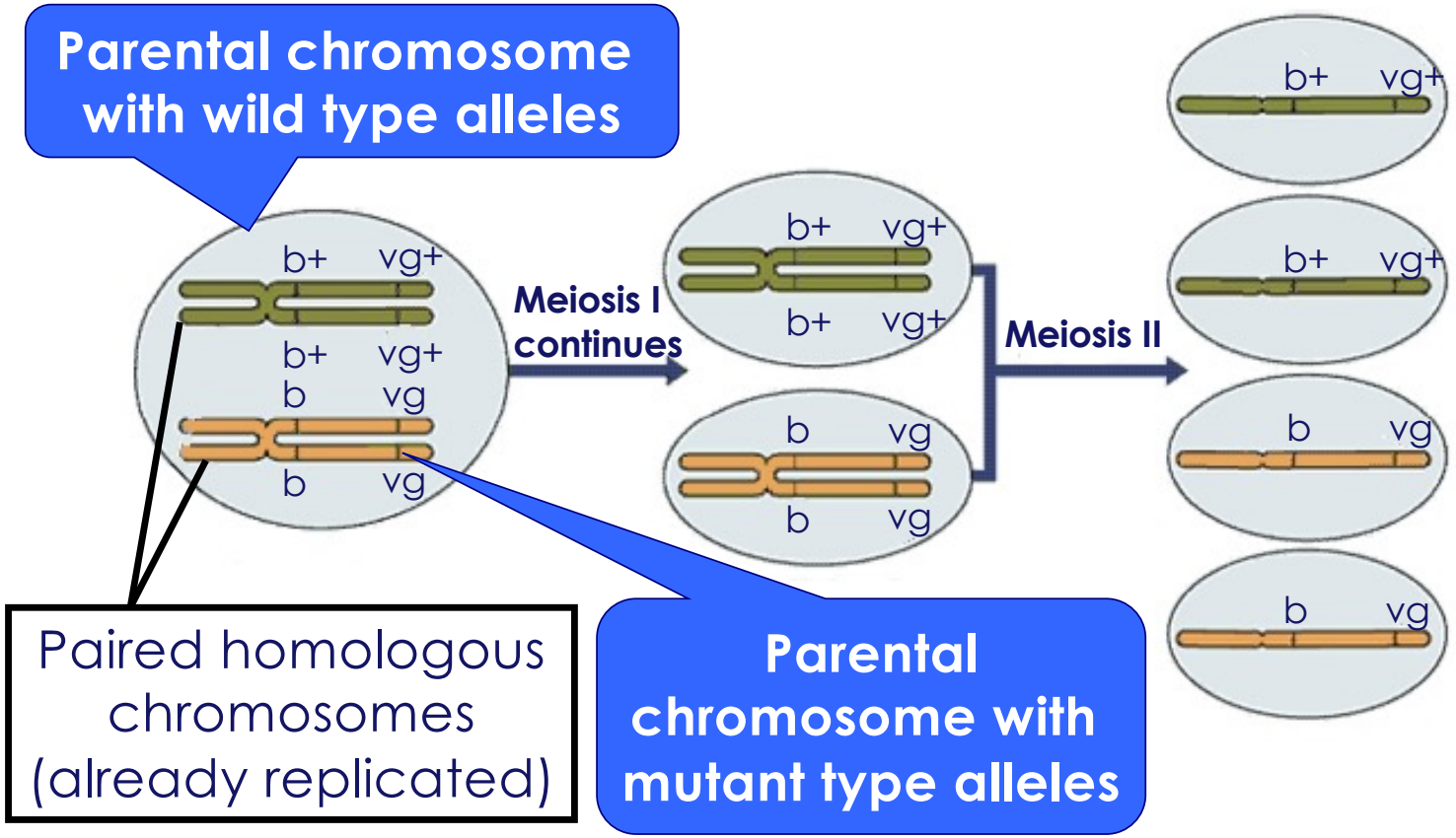
965	944	206	185

These are the results expected from Mendel's 2nd law (independent assortment of alleles)

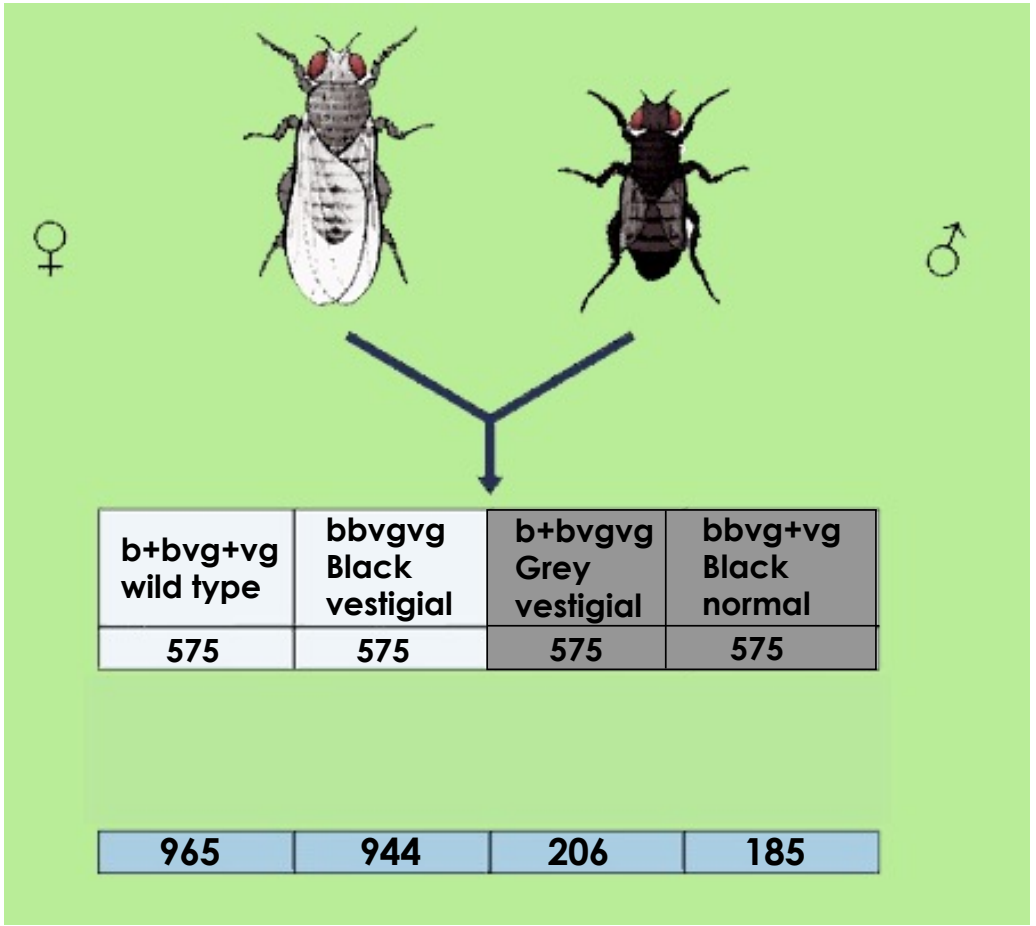
CONCLUSION

- These two genes do not sort independently
- They are linked on the same chromosome

Why don't all gametes have parental genotypes?



Context Slide



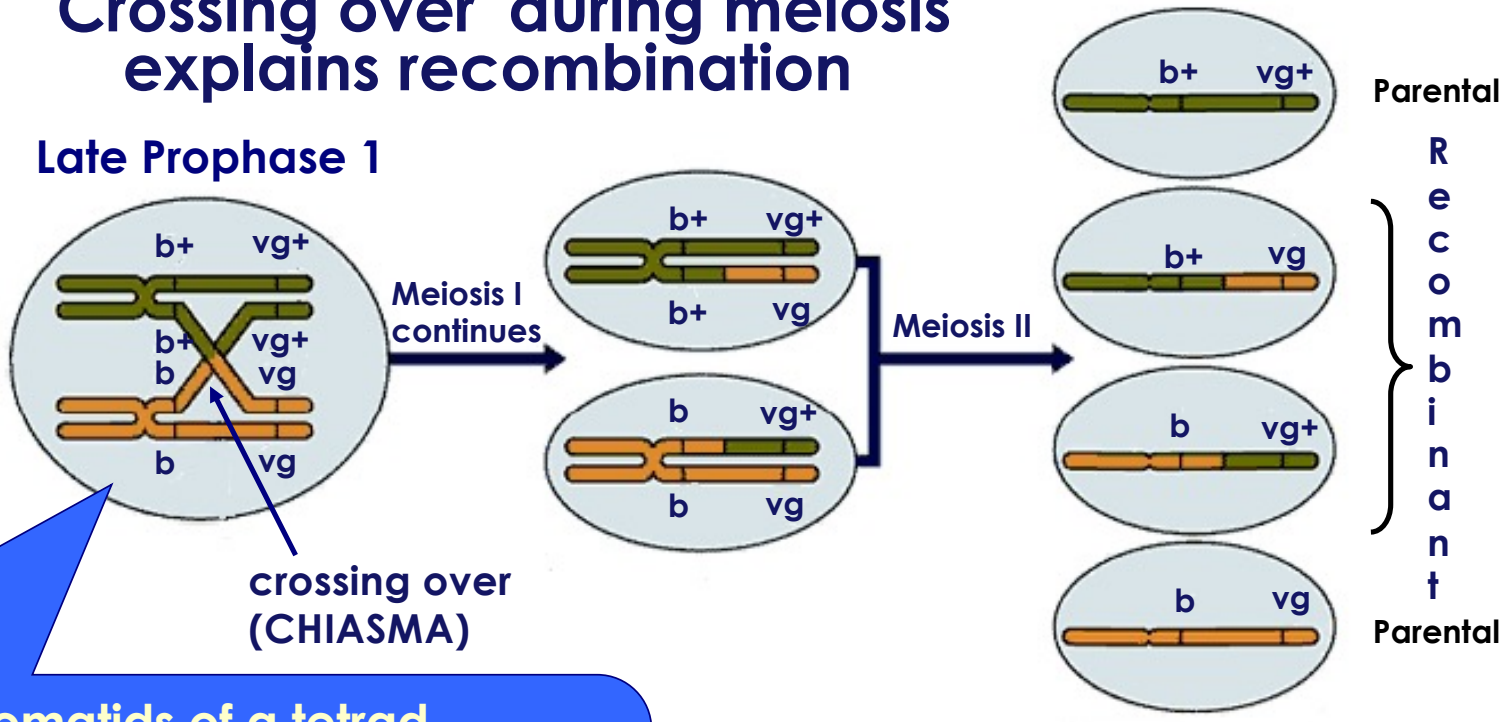
b+bvg+vg wild type	bbvgvg Black vestigial	b+bvgvg Grey vestigial	bbvg+vg Black normal
575	575	575	575

965	944	206	185

We do get some recombinants……why?????

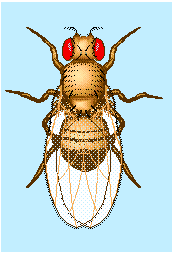
How does recombination occur for linked genes?

'Crossing over' during meiosis explains recombination

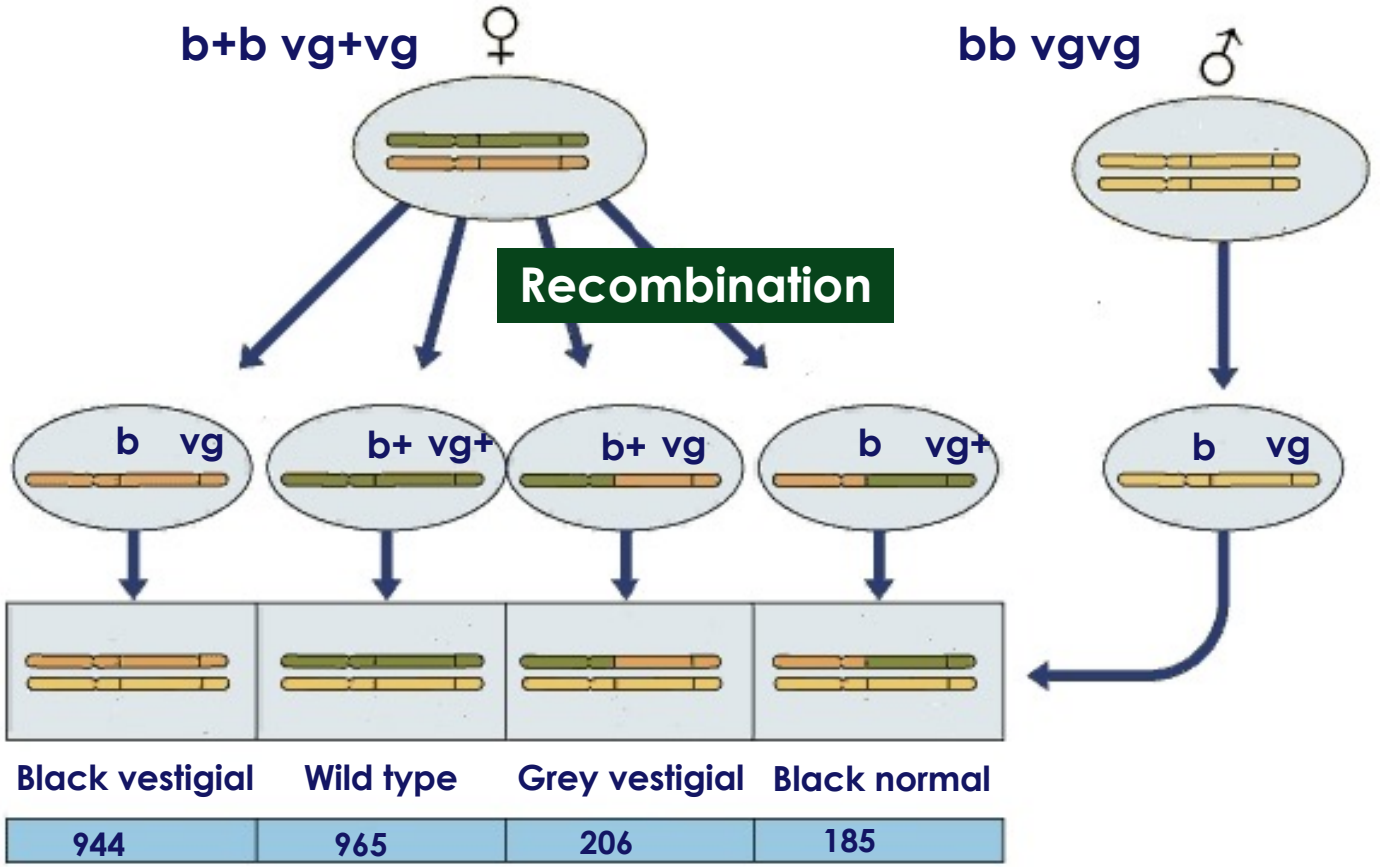


Two chromatids of a tetrad (one from each pair)cross over at random points and swap genetic material. This is a simple cross where only one chiasma has formed.

The proportion of recombinant gametes is termed the recombination frequency



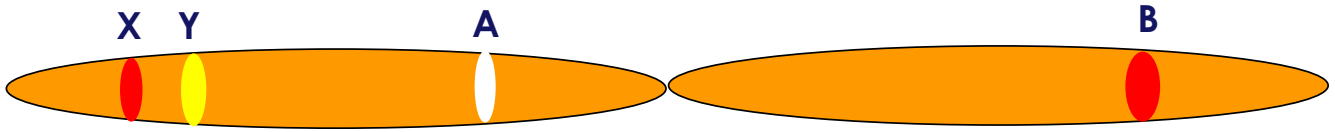
Core Slide



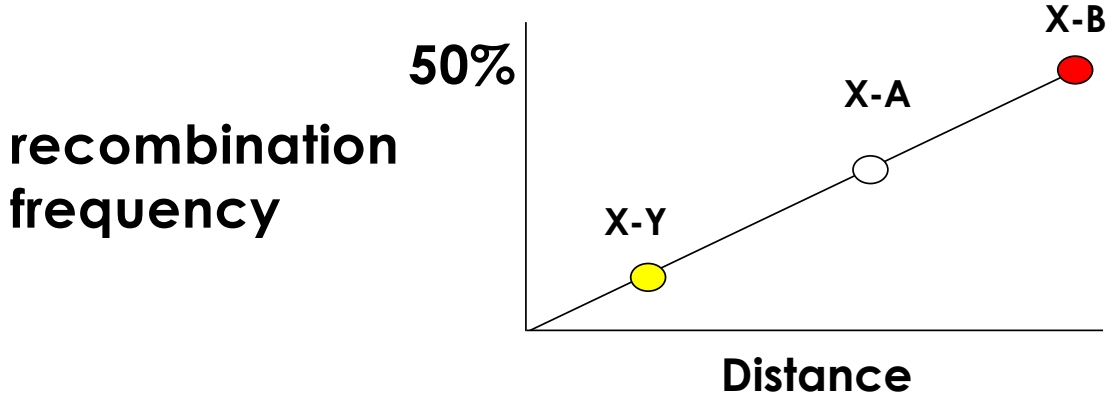
In this case, 17% of the gametes contain a chromosome that has formed a chiasma between the two genes

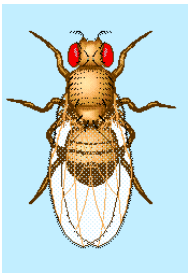
Recombination Frequency = (206+185) / (944+965+206+185) = 0.17 or 17%

The smaller the distance between two genes - the less likely a chiasma will be formed between them

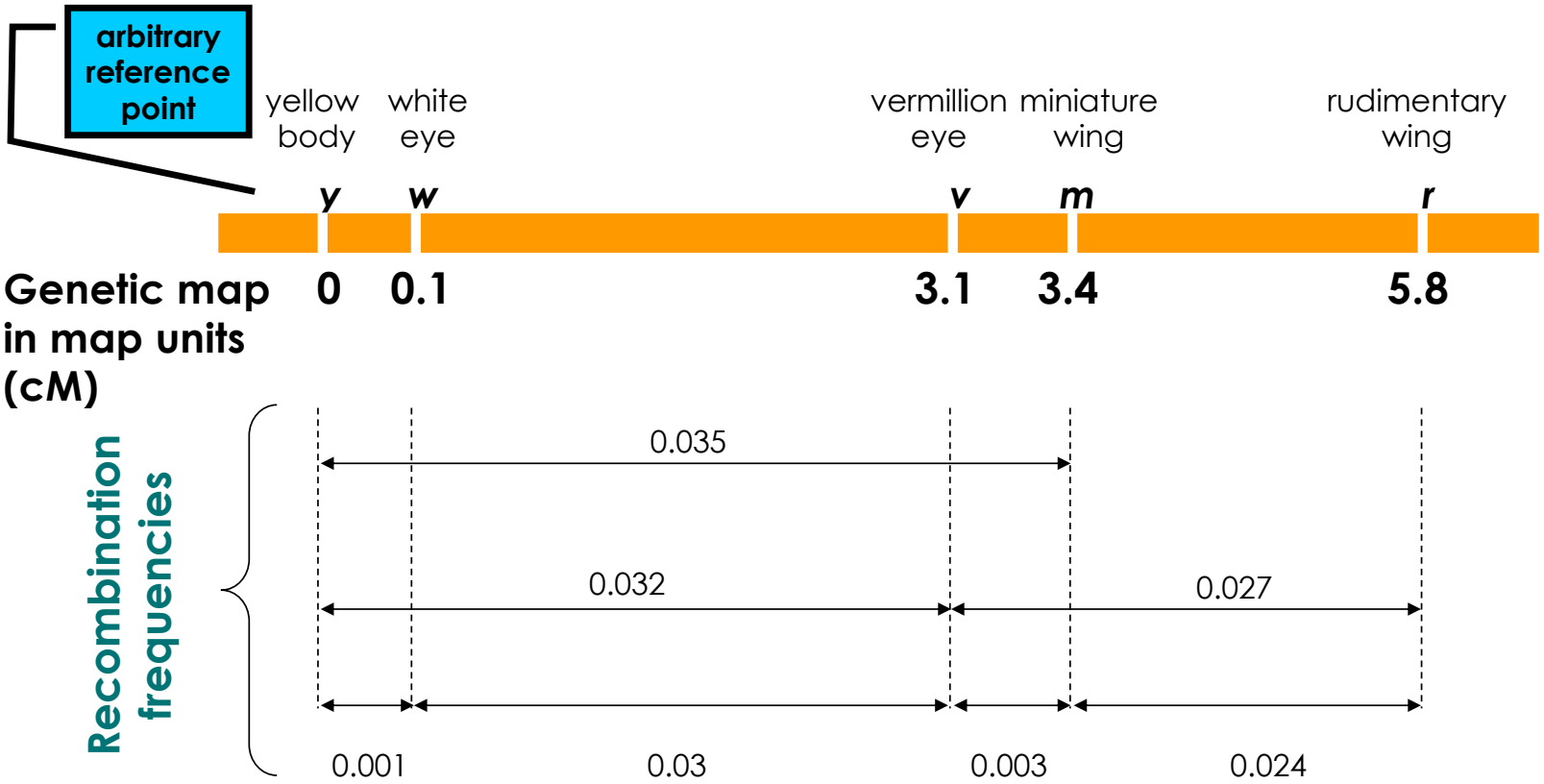


- Distant (unlinked) genes have recombination frequencies around 50% (half recombinant and half parental types).
- Close genes have recombination frequencies of 0-50%.
- There is a near linear relationship between distance and recombination frequency.





Step Towards a Genetic Map



Sturtevant was able to derive a partial map of *Drosophila* by using recombinant frequencies of five traits. He used an arbitrary unit of distance the: **centimorgan (cM) = 100 x recombination frequency**

Example Question

A species of flowering plant has a gene (*purple*) that determines flower colour, and another gene (*tall*) that controls plant height. A scientist performs a test cross between two lines of this species: one heterozygous plant line with purple flowers and tall stems; and the other line pure-bred with white flowers and short stems. The progeny from this cross comprise: 345 purple-flowered tall plants; 340 white-flowered short plants; 255 white-flowered tall plants; and 260 purple-flowered short plants.

1. Give the genotype for each phenotype using standard genetic symbols

PpTt X pptt → PpTt pptt ppTt Pptt

purple tall × white short → purple tall (345), white short (340), white tall (255), purple short (260)

2. Calculate the recombination frequency of these two genes

$(255 + 260)/(345 + 340 + 255 + 260) = 515/1200 = 0.429$ (42.9%)

3. Why is there a bias towards parental phenotypes?

Fewer recombinants means the 2 genes must be linked (on same chromosome)(violates Mendel's 2nd Law (independent assortment))

Lecture 20 Summary

- Sex-linked genes show inheritance patterns that differ between sexes.
- X-linked recessive traits such as red-green colour blindness are more common in males than in females.
- Crossing over during meiosis can shuffle genetic material between the non-sister chromatids of two homologous chromosomes.
- The distance between 2 linked genes can be estimated from the proportion of new phenotypes/genotype combinations produced by a test cross

Objective-Based Questions

- Define the terms carrier, homozygous, recombinant and hemizygous.
- Eye colour is a sex-linked trait in Drosophila i.e., the allele for eye colour is on the X-chromosome.
  a) If you cross a homozygous red eyed female with a hemizygous white eyed male, what are the sex, genotypes and phenotypes of the offspring?
  b) If you cross a homozygous white eyed female with a hemizygous red eyed male, what are the sex, genotypes and phenotypes of the offspring?
- What does it mean when we talk about linked genes? How are linked genes inherited? With linked genes, is the bias towards PARENTAL or RECOMBINANT genotypes?
- Fill in the gaps:
  The GREATER/LESSER the distance between two genes, the more likely a chiasma will be formed between them. Distant genes have HIGHER/LOWER recombination frequencies. There is a near _____ relationship between distance and _____.

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