

ZAPOROZHYE STATE MEDICAL UNIVERSITY
DEPARTMENT OF MEDICAL BIOLOGY

Lecture

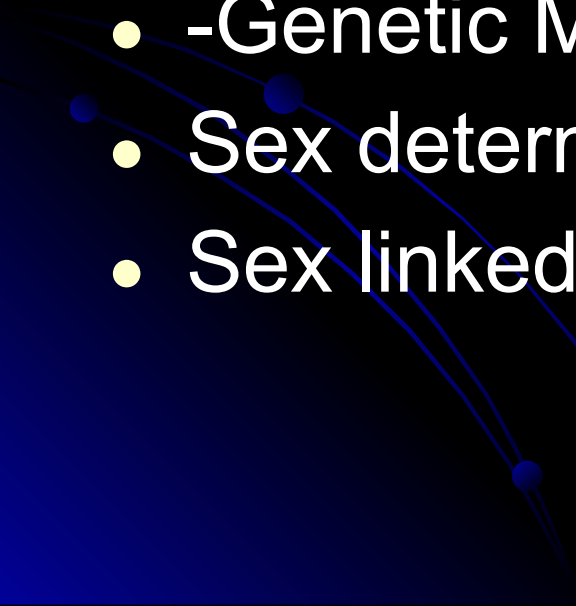
LINKAGE AND GENE MAPS. SEX DETERMINATION

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QUESTIONS

- -Morgan's experiments with fruit flies.
 - -What is Linkage. Types of Linkage.
 - -The main statements of Chromosomal Theory of Linkage.
 - -Genetic Maps.
 - Sex determination and its types
 - Sex linked inheritance
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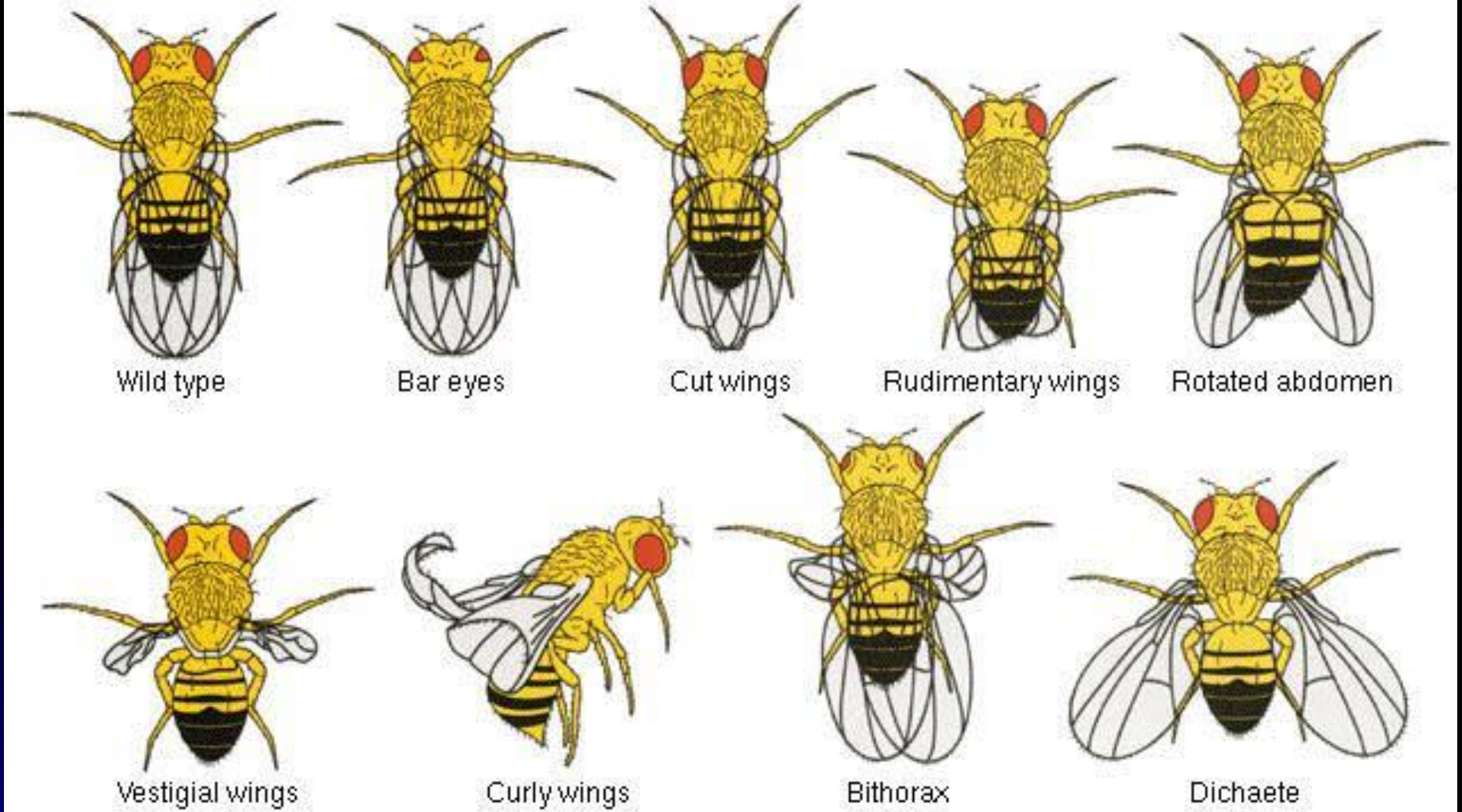
Morgan's Experimental Evidence: *Scientific Inquiry*

- The first solid evidence associating a specific gene with a specific chromosome came from Thomas Hunt Morgan, an embryologist
- Morgan's experiments with fruit flies provided convincing evidence that chromosomes are the location of Mendel's heritable factors

Thomas Hunt Morgan in the 'Fly Lab'




Some Mutations



- Morgan found that body color and wing size are usually inherited together in specific combinations (parental phenotypes)
- He noted that these genes do not assort independently, and reasoned that they were on the same chromosome
- Each chromosome has hundreds or thousands of genes (except the Y chromosome)
- Genes located on the same chromosome that tend to be inherited together are called **linked genes**

The tendency of different genes present on a chromosome to be inherited together from one generation to the next generation is called linkage. These genes do not assort independently.



Types of Linkage:

1. **Complete linkage:** the characters are inherited together in their original combinations for two or more generations in a continuous and regular fashion

Parents: P Grey, Long x Black, Vestigial

♀ AA BB

♂ aabb

Gametes: AB

ab

F1 generation

All Grey, Long

AaBb

Test cross: P ♀ aabb x ♂ AaBb
G ab AB, ab
F2 AaBb, aabb
Grey, Long Black, Vestigial

In this type of linkage, genes are closely associated and tend to transmit together.



- However, nonparental phenotypes were also produced
- Understanding this result involves exploring **genetic recombination**, the production of offspring with combinations of traits differing from either parent

Genetic Recombination and Linkage

- Recombinant offspring are those that show new combinations of the parental traits
- Morgan discovered that genes can be linked but due to the appearance of recombinant phenotypes, the linkage appeared incomplete
- Crossing over of homologous chromosomes was the mechanism

2. Incomplete linkage involves separation of linked genes as result of crossing over. They produce some percentages of non-parental combinations.

In this type of linkage, genes are widely located in chromosomes and have chance of separation by crossing over.

Test cross. P Grey, Long (F1) X Black, Vestigial
♀ AaBb x ♂ aabb

G non-crossovers: AB, ab
recombinants: Ab, aB

F2 AaBb – grey, long (41,5%)

aabb – black, vestigial (41,5%)

83% - parental combination showing linkage

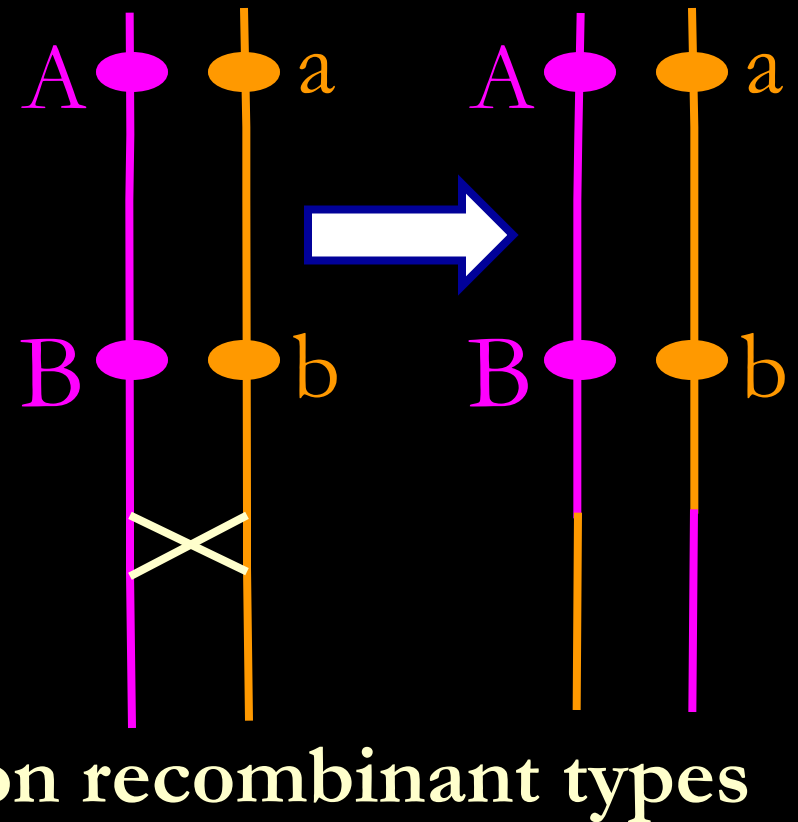
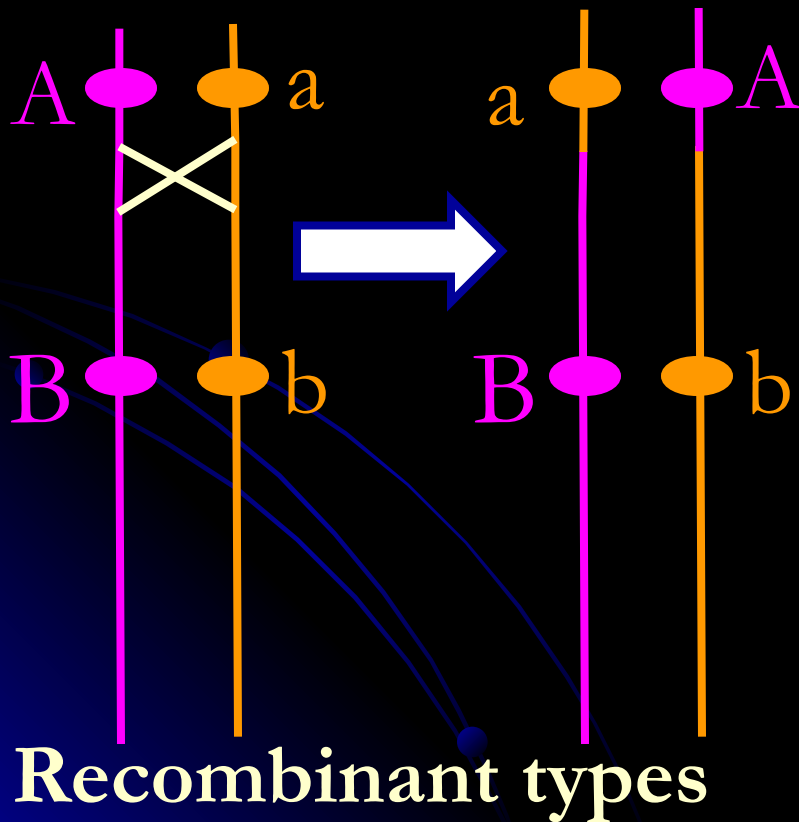
Aabb – grey, vestigial (8,5%)

aaBb – black, long (8,5%)

17% - recombinants due to crossing over

Recombinant Types

- Produced when a crossover occurs between the 2 genes being studied:



Morgan formulated “**The chromosome theory of Linkage**” according to which:

Each chromosome bears many genes. Genes are arranged in a linear fashion in the chromosome.

The strength of linkage depends upon the distance between the linked genes in chromosome. The closely located genes show strong linkage while the widely located genes show weak linkage.

Linked genes remain in their original combination during the course of inheritance.

It becomes possible to determine the distances between the genes in a linkage group, their order and may give diagrammatic representation of chromosomes showing the genes as points separated by distances proportional to the amount of crossing-over. Such a diagrammatic representation of the relative distances between linked genes of a chromosome is called genetic map.

- Sturtevant used recombination frequencies to make linkage maps of fruit fly genes
- Using methods like chromosomal banding, geneticists can develop cytogenetic maps of chromosomes
- **Cytogenetic maps** indicate the positions of genes with respect to chromosomal features

- A **linkage map** is a genetic map of a chromosome based on recombination frequencies
- Distances between genes can be expressed as **map units**; one map unit, or centimorgan, represents a 1% recombination frequency
- Map units indicate relative distance and order, not precise locations of genes

Map Units

- The farther apart genes are on a chromosome the more likely they are to be separated during crossing over
- 2 genes on the same chromosome can be located so far apart that the frequency of recombinant types reaches 50%
- Same as for genes located on different chromosomes.
- These genes will assort independently, even though they are on the same chromosome.

Sex determination

Genetic information on the sex chromosomes is responsible for the primary sex determination. The development of gonads in the body is the main character for sex determination.

If sexually reproducing organisms two types of chromosomes are recognized: autosomes and sex chromosomes.

Sex chromosomes are responsible for the sex determination.

If two sex chromosomes are similar (XX) the individual is described as homogametic. It produces similar gametes.

If two sex chromosomes are different (XY) or it contains only one sex chromosome (XO) the individual is described as heterogametic and it produces two types of gametes.

Sex determination based on:

- ❖ number of sex chromosomes
- ❖ differences in sex chromosomes

1. Number of sex chromosomes.

In this method of sex determination, chromosomal number is different in male and female. Ex.: in bugs, grasshoppers and cockroaches females are with two X-chromosomes and males are with one X-chromosome.

So females are homogametic and males are heterogametic. The sex of the offspring depends on the fertilizing sperm:

$$\begin{array}{rcc} \text{P} & \text{♀ } 2A + XX & \times & \text{♂ } 2A + XO \\ \text{G} & A + X & & A + X \quad A + O \\ \text{F1} & 2A + XX & & 2A + XO \end{array}$$

2. Differences in sex chromosomes

❖ XX – XY method

In this type of sex determination, both males and females have the same number of chromosomes:

Females – XX – chromosomes, males-XY chromosomes

$$\begin{array}{rcccl} \text{P} & \text{♀} & 2A + XX & \times & \text{♂} & 2A + XY \\ & \text{G} & A + X & & & A + X \quad A + Y \\ \text{F1} & & 2A + AA & & & 2A + XY \end{array}$$

Ex.: in human beings and Drosophila females are homogametic and males are heterogametic

❖ ZW – ZZ method

In birds, reptiles, some fishes, butterflies, female are heterogametic with ZW-chromosomes and males are homogametic with ZZ-chromosomes

SEX - LINKED INHERITANCE

Sex chromosomes carry genes for some characters. Such characters are said to be sex – linked and may be possessed by either sex.

The genes which occur exclusively on the X-chromosomes are called X-linked genes. They determine X-linked characters:

- Duchene muscular dystrophy (DMD)
- Hemophilia
- Colour blindness
- Night blindness

The genes which exclusively occur in Y chromosome are called holandric genes. They inherited from father to son. Y-linked characters in man are:

- Hypertrichosis (growth of hair on the rim of pinna)
- Porcupine man (growth of hair on the body)
- Webbing of toes
- Testis determining factor (TDF)

XY – linked characters are controlled by genes located in the homologous regions of X and Y-chromosomes.

EX.: skin diseases - Xeroderma pigmentosum
- Retinitis pigmentosum