

The Chromosomal Basis of Inheritance

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Slide 1

Mendel's results led him to propose two laws:

- Law of Segregation - When a sexual organism produces gametes, the two alleles for each characteristic separate and end up in different gametes.
- Law of Independent Assortment - If genes occur on separate chromosomes, the alleles for those genes assort independently during gamete formation.

Both of these laws are explained by events that occur during meiosis.

Slide 3

Diploid organisms have two types of chromosomes:

- Autosomes are all of the chromosomes other than sex chromosomes. A diploid cell has a number of pairs of autosomes. Each pair is homologous, with the same number of loci on each homolog in a pair.
- Sex chromosomes are chromosomes that are involved in determining the sex of the offspring created if the organism reproduces. Sex chromosomes also contain a number of genes that have nothing to do with determination of sex. If an individual contains two sex chromosomes, those two sex chromosomes are not the same size, unlike homologous pairs of autosomes. Therefore, there are some genes, called sex-linked genes, for which each somatic cell (in one of the sexes) contains only one allele, because only one of the sex chromosomes has the locus for the corresponding gene.

The gene for eye color in fruit flies is an example of a sex-linked gene, which is a gene whose locus is on a sex chromosome. Since (in male fruit flies) each cell contains only one allele for each sex-linked gene, meiosis will produce some gametes that carry that allele and some that carry no allele for that characteristic. Moreover, since a sex-linked gene is on a chromosome that determines sex, we observe a different phenotypic ratio in males compared to females.

Slide 5

Animals feature a variety of sex-determination mechanisms, depending on species.

- In the X-Y system, a male has one X chromosome and one Y chromosome. A female has two X chromosomes. Since males carry different kinds of sex chromosomes, they make different kinds of gametes with respect to sex determination, and the father determines the sex of offspring.
- In the X-0 system, there is only one kind of sex chromosome (X). Females have two X chromosomes, and males have only one.
- In the Z-W system, the female has both types of chromosomes, and the male has two Z chromosomes. The mother determines the sex of offspring.
- In the haplo-diploid system, there are no sex chromosomes. A male is an individual with a haploid set of autosomes, and a female is an individual with a diploid set of autosomes.

Slide 6

Some genetic disorders in humans are due to a mutant allele for a sex-linked gene. Males (which are hemizygous, because they carry only one allele on their only X chromosome) either have the disorder, or they are normal. A female (having two alleles, because she has two X chromosomes) can be either a fully normal individual, a carrier, or an affected individual, depending on how many of her two alleles are of the mutant form.

Slide 7

In mammals, which have two kinds of sex chromosomes (X and Y), a female (XX) has two alleles for every sex-linked gene, whereas a male (XY) has only one. Therefore, during development in females, one of the two X chromosomes in each cell of the early embryo becomes inactivated, putting the female on an equal footing with males with respect to the number of sex-linked alleles. This causes heterozygous females to develop into phenotypic mosaics, with some parts of the body expressing one of the two traits and the rest of the body expressing the other trait.

Slide 8

If two different genes are located on the same chromosome, they are said to be linked. Since chromosomes (rather than individual genes) independently assort during meiosis, two linked genes are expected to be carried together into one daughter cell or the other. On the other hand, two genes that are not linked (they are on different chromosomes) are able to either go into the same daughter cell or segregate and go into opposite daughter cells. Therefore, the prediction is that if two genes are linked, then a test cross of a dihybrid (an individual that is heterozygous for both characteristics) mated with a doubly homozygous mutant should produce only parental type offspring. If the two genes are not linked, the same cross predicts that offspring can be either of the parental type or of the recombinant type, with equal probability (a 1:1:1:1 ratio). The actual results of such a cross in flies yields a phenotypic ratio that disagrees with both predictions. The explanation is that two linked genes are sometimes able to segregate during meiosis. This happens if crossing over happens in such a way that the chiasma occurs between the two linked genes.

Slide 9

It is possible to calculate the relative degree of linkedness of two linked genes (i.e., how far apart their loci are on a chromosome) by observing the frequency with which recombinant individuals (as opposed to parental individuals) are produced.

Slides 10 & 11

By performing crosses for different pairs of linked genes and comparing the recombination frequencies of the various pairs, it is possible to construct a gene map that shows the positions (loci) of the various genes.

Slide 12

Non-disjunction (the failure of homologs to properly separate during anaphase) can lead to aneuploidy (an incorrect number of chromosomes). Aneuploidy will result whether non-disjunction occurs during meiosis I or meiosis II.

Slide 13

Mutation is any change to the DNA sequence. This can involve long stretches of nucleotides (as opposed to point mutations). Insertion adds a sequence of nucleotides to a chromosome. Deletion removes a sequence from a chromosome. Inversion occurs when a sequence is removed, turned end-to-end, and reinserted. Reciprocal translocation involves two deletions, after which the deleted sequences are inserted elsewhere.