

Chapter 2 Outline and Notes

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MENDEL OBSERVED PHENOTYPES AND FORMED HYPOTHESES

INHERITANCE FOLLOWS THE SAME LAWS IN MOST ORGANISMS.

THE BASIC LAWS OF GENETICS were first established by Gregor Mendel in 1865.

Mendel used peas which could either be crossed or self-fertilized.

Earlier observers looked at many traits at once. Mendel focused on one at a time.

Earlier observers used parents of unknown hereditary background. Mendel bred PURE LINES first until they bred true.

Earlier observers only observed a single generation at a time. Mendel extended his observations over several generations.

Earlier observers failed to quantify their results. Mendel counted offspring and established ratios.

TERMINOLOGY:

PHENOTYPE = appearance ("pheno-"=visible, as in "phenomenon"); GENOTYPE = genetic make-up, not always visible, but detectable by performing crosses

ALLELES = variants of a gene.

HOMOZYGOUS = having two alleles that are alike; HETEROZYGOUS = having two unlike alleles

DOMINANT = showing a phenotypic effect in heterozygous form; RECESSIVE = showing a phenotypic effect only when homozygous

MENDEL'S FINDINGS:

Crosses between PURE LINES produce offspring of one (dominant) phenotype only.

Crossing of first generation plants produces 3:1 ratio of dominants to recessives in the second generation (F₂).

Explanation of 3:1 ratio in terms of PARTICULATE INHERITANCE.

"LAW OF SEGREGATION" = dominant and recessive alleles of heterozygote separate from one another during meiosis.

"LAW OF INDEPENDENT ASSORTMENT" for 2 genes at a time: genes at different locations are chosen (sampled) independently of one another during gamete formation.

THE CHROMOSOMAL BASIS OF INHERITANCE EXPLAINS MENDEL'S HYPOTHESES:

Genes are located on chromosomes within the nucleus of each cell. Behavior of genes follows behavior of chromosomes. (This includes an exception to independent assortment in the case of "linked" genes on the same chromosome.)

MITOSIS:

Normal cell division is called MITOSIS; chromosome number doubles and is then halved, so the resulting chromosome number remains unchanged in the two daughter cells.

MEIOSIS:

Most animal and plant cells are DIPLOID (their chromosomes occur in pairs); the major exceptions are the egg and sperm cells, called GAMETES; gametes are always HAPLOID (their chromosomes occur as singletons).

The cell division that produces haploid cells (such as gametes) is called MEIOSIS. During meiosis, the chromosomes double once and divide twice, resulting in four haploid cells that each have half of the original chromosome number, including one chromosome from each pair.

The separation of chromosome pairs during meiosis is responsible for segregation.

GENE LINKAGE:

The independent separation of different pairs of chromosomes is responsible for independent assortment.

Genes on the same chromosome segregate together (LINKAGE), unless a chromosomal cross-over brings about their recombination.

CONFIRMATION OF THE CHROMOSOMAL THEORY:

Experiments have shown that the inheritance of genes parallels the inheritance of visible chromosomes.

When chromosomes have visible markers at opposite ends, recombination of genes (as observed in crosses) is always accompanied by the rearrangement (recombination) of the visible chromosome markers.

GENES CARRIED ON SEX CHROMOSOMES DETERMINE SEX AND SEX-LINKED TRAITS.

SEX DETERMINATION:

Humans and most other species have an XX / XY form of sex determination: XX usually produces female (with two copies genes on the X-chromosome); XY usually produces male (only one copy of most sex-linked genes).

Occasional anomalies resulted in discovery of the sry gene carried on the Y chromosome; this gene determines maleness, presumably by regulating production of the hormone TESTOSTERONE.

Two other forms of sex determination are WW (male) versus WZ (female) in birds and haplodiploidy (males are haploid and females are diploid) in the insect order Hymenoptera.

SEX-LINKED TRAITS are those carried on the X chromosome.

Because males only have one X chromosome, they only have one copy of any sex-linked gene, and thus only a single allele. The product of this allele is always displayed phenotypically.

Females have two copies of each sex-linked genes. If a sex-linked allele is recessive, females will not exhibit the phenotype unless they have two copies of that allele.

Females heterozygous for a recessive sex-linked trait are called carriers. They do not exhibit the trait phenotypically, but they can pass it on to their descendants.

Recessive sex-linked alleles for uncommon conditions show up much more often in males and only rarely in females. Males with such traits may have affected grandfathers or great-grandfathers, and the connecting individuals in the intervening generations are carrier females.

Red-green colorblindness and hemophilia are examples of sex-linked traits in humans.

CHROMOSOMAL VARIATION:

The pattern of chromosomes visible under a light microscope is called a KARYOTYPE.

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Most people have 23 pairs of chromosomes. These include the sex chromosomes and 22 pairs of other chromosomes, called autosomes.

Trisomy is an uncommon condition in which an extra chromosome is present, making a triple instead of the usual pair.

The most frequent form of trisomy is trisomy of chromosome 21, resulting in one form of Down's syndrome.

Other examples of trisomy include Patau's syndrome (trisomy #13).

Trisomy of the sex chromosomes can result in:

XXY (sterile males with Klinefelter's syndrome),

XYY (males with an extra Y), or

XXX (sterile females with an extra X).

Chromosome numbers may also be lower than 46. Females with only one X chromosome (XO) have Turner's syndrome and are sterile.

Both Turner's and Klinefelter's syndromes result from a type of abnormal cell division called nondisjunction.

Chromosomal translocations occur when a piece of one chromosome is attached to another.

SOCIAL AND ETHICAL ISSUES:

Rare individuals can be XX but not female, or XY but not male.

Rare individuals show ambiguous indications of sex.

Some people say it is unfair to forcibly assign an individual to one sex or another because many variations naturally exist.

THE MOLECULAR BASIS OF INHERITANCE FURTHER EXPLAINS MENDEL'S HYPOTHESES.

DNA AND GENETIC TRANSFORMATION:

Griffith's experiment established the phenomenon of BACTERIAL TRANSFORMATION: dead bacteria of a virulent strain called IIIS were able to transform the nonvirulent strain IIR into IIIS.

Avery, MacLeod & McCarty established that bacterial transformation required DNA.

Hershey & Chase demonstrated that BACTERIOPHAGE viruses reproduced by using genetic material made of DNA. The viruses injected this DNA, but not their protein, into host bacteria during viral reproduction.

THE CHEMICAL COMPOSITION OF DNA:

DNA is composed of phosphate groups, deoxyribose sugar, and nitrogenous bases of four types (abbreviated A, G, C, and T).

Chargaff discovered that the amount of adenine and thymine were equal (A=T) in DNA from a given species, as were the amounts of guanine and cytosine (G=C).

THE THREE-DIMENSIONAL STRUCTURE OF DNA:

Rosalind Franklin used X-RAY DIFFRACTION to study the 3-D structure of DNA. Watson & Crick described double-helix model of DNA structure: Building blocks: phosphate groups, deoxyribose sugar, and nitrogen-containing bases (A = adenine, G = guanine, C = cytosine, and T = thymine). One phosphate + one deoxyribose sugar one base = a NUCLEOTIDE. Nucleotides are connected by alternating chain of phosphates & sugars. There are two strands of nucleotides, arranged in opposite directions. Base-pairing of (A with T) and (C with G) holds the two strands together. The two strands are twisted to form a DOUBLE HELIX.

A gene is a sequence of bases in DNA. The location of the gene on the DNA is called its LOCUS.

DNA REPLICATION: DNA is made from DNA, using one strand as a TEMPLATE (pattern) to synthesize the missing strand one base at a time.

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