

Chapter 14- Mendel and the Gene Idea

(Key Concepts are Underlined)

Gregor Mendel's Discoveries

Mendel brought an experimental and quantitative approach to genetics: *science as a process*

Character- heritable feature

Trait- variant for a character

- pea experiments: a) peas were available in many varieties, b) sure of parentage of new seeds, c) characters studied were "either-or," and d) began with **true-breeding** varieties (offspring of same variety)

Hybridization- crossing of two varieties

Monohybrid cross- tracks the inheritance of a single character

P generation- true-breeding parental generation

F₁ generation- first filial "P x P hybrid" generation

F₂ generation- second filial "F₁ x F₁ hybrid" generation

By the law of segregation, the two alleles for a character are packaged into separate gametes

- blending model of inheritance is incorrect

- Mendel used the term "heritable factor," which we know now as a gene

Alleles- versions of a gene

Mendel's Hypothesis:

1. Alternative versions of genes account for the variations in inherited characters.
2. For each character, an organism inherits two alleles, one from each parent.
3. If the two alleles differ, then one, the **dominant allele**, is fully expressed in the organism's

appearance; the other, the **recessive allele**, has no noticeable effect on the organism's appearance.

4. The two alleles for each character segregate during gamete production.

Law of Segregation- separation of alleles in separate gametes

Punnett Square

Homozygous- identical pair of alleles for a character

Heterozygous- two different alleles for a gene

Phenotype- organism's appearance

Genotype- genetic makeup

Testcross- breeding of a recessive homozygote with an organism of unknown genotype

By the law of independent assortment, each pair of alleles segregates into gametes independently

Dihybrid cross- mating of parental varieties differing in two characters

Law of Independent Assortment- independent segregation of each pair of alleles during gamete formation

Mendelian inheritance reflects rules of probability

- *independent events* are unaffected by what happens prior

- the probability of two or more independent events occurring in a specific combination is equal to the product of their individual probabilities (a.k.a. the Rule of Multiplication)

- the probability of an event which can occur in two or more ways is equal to the sum of the separate probabilities of those ways (a.k.a. the Rule of Addition)

Mendel discovered the particulate behavior of genes: a review

- the larger the sample size, the closer the results will conform to our predictions

Extending Mendelian Genetics

The relationship between genotype and phenotype is rarely simple

Incomplete Dominance- F₁ hybrids have an appearance somewhere in between the phenotypes of the parental varieties (e.g. red, pink, and white snapdragons)

Complete Dominance- heterozygote and dominant homozygote are indistinguishable

Codominance- both alleles are separately manifest in the phenotype (e.g. M, N, and MN human blood groups)

Organismal Level vs. Biochemical Level vs. Molecular Level issue- e.g. Tay-Sachs disease- lipids accumulate due to a malfunctioning enzyme and eventual death

(*Organismal-Recessive, Biochemical-Incomplete, Molecular-Codominant*)

- dominant alleles do not subdue recessives!

- dominant alleles are not necessarily more common (e.g. polydactyly is dominant and only 1 out of 400 have it)

Multiple Alleles- multiple alleles possible for a single gene (e.g. ABO blood groups in humans)

Pleiotropy- the ability of a gene to affect an organism in many ways (e.g. sickle-cell can have multiple symptoms)

Epistasis- a gene at one locus alters the phenotypic expression of a gene at a second locus (e.g. black **B**₋, brown **bb**, and white mice **cc**)

Polygenic Inheritance- an additive effect of two or more genes on a single phenotypic character (e.g. human skin color)

- *Nature vs. Nurture*: environmental influence gives rise to a range of phenotypic possibilities

Multifactorial characters- phenotype influenced by both genetics and environment

Mendelian Inheritance in Humans

Pedigree analysis reveals Mendelian patterns in human inheritance

Pedigree- family history for a particular trait; used to trace traits back, determine genotypes, and predict genotypes of future offspring

Many human disorders follow Mendelian patterns of inheritance

Recessively Inherited Disorders:

- only show up in recessive homozygotes
- heterozygote individuals are **carriers** (one normal allele is sufficient)

Cystic fibrosis- defective (or missing) chloride channel membrane proteins in certain cells creates an excess of chloride in extracellular fluid, which causes the mucous coating these cells to become thicker and stickier than normal; 1/2500 European white births

Tay-Sachs disease- see “Extending Mendelian Genetics”; 1/3600 Ashkenazic Jew births

Sickle-cell disease- red blood cells become sickle shaped when oxygen level is low, which is caused by the substitution of a single amino acid in the hemoglobin protein; 1/400 African-American births

(tropical Africa vestige); codominant at the molecular level

Dominantly Inherited Disorders:

Achondroplasia- form of dwarfism

Huntington's Disease- degenerative disease of the nervous system, which is phenotypically expressed when the individual is 35 to 45 years of age; offspring has a 50% of have the disease; tip of chromosome 4

Multifactorial Disorders:

- genetic predisposition and environmentally caused (e.g. heart disease, cancer, alcoholism, certain mental illnesses)

Technology is providing new tools for genetic testing and counseling

- genetic counseling (i.e. deciding to have children if potential parents are possible carriers of a recessive disorder)

Amniocentesis- physician inserts a needle into the uterus and extracts 10 millimeters of amniotic fluid between the fourteenth and sixteenth week of pregnancy to karyotype sloughed fetal cells

Chorionic Villus Sampling (CVS)- physician samples a small amount of fetal tissue from the placenta for karyotyping; can be performed earlier than amniocenteses

Ultrasound

Fetoscopy- needle thin camera

- newborn screening can be effective in the proper treatment of genetic disorders (e.g. PKU- deficient in breaking down phenylalanine, which causes mental retardation)