Genetics
3.4- Inheritance

Essential idea:
• The inheritance of genes follows patterns.

Nature of science:
• Making quantitative measurements with replicates to ensure reliability.
  – Mendel's genetic crosses with pea plants generated numerical data. (3.2)

Theory of knowledge:
• Mendel's theories were not accepted by the scientific community for a long time.
  • What factors would encourage the acceptance of new ideas by the scientific community?

Mendel's Laws
• Mendel discovered the principles of inheritance with experiments in which large numbers of pea plants were crossed.
  – Law of Segregation
  – Law of Independent Assortment

Principles of Inheritance
• Gametes are haploid so contain only one allele of each gene.
  • The two alleles of each gene separate into different haploid daughter nuclei during meiosis.
  • Fusion of gametes results in diploid zygotes with two alleles of each gene that may be the same allele or different alleles.
Principles of Inheritance

- **Dominant alleles** mask the effects of recessive alleles but **co-dominant alleles** have joint effects.

Principles of Inheritance

- Many **genetic diseases** in humans are due to recessive alleles of autosomal genes, although some genetic diseases are due to dominant or co-dominant alleles.

Principles of Inheritance

- Some genetic diseases are **sex-linked**. The pattern of inheritance is different with sex-linked genes due to their location on sex chromosomes.

Principles of Inheritance

- Many genetic diseases have been identified in humans but most are very rare.

Mutagens

- Radiation and **mutagenic** chemicals increase the mutation rate and can cause genetic diseases and cancer.
  - Mutations are changes in an organism's genetic code.
  - Gene mutations are changes in DNA nucleotides.
  - Chromosomal mutations are changes in chromosome structure.

Mutagens

- Application: Consequences of radiation after nuclear bombing of Hiroshima and accident at Chernobyl.
  - Radioactive isotopes released into the environment exposing humans and other organisms to potentially dangerous levels of radiation.
Mutagens

- Hiroshima
  - Cancer, especially leukemia and lymphoma
  - Small brain size
  - Mental retardation
  - Lower IQ
  - Delayed development
  - Blindness
  - Spinal Bifida
  - Cleft palate

- Chernobyl Nuclear Power Station
  - Pine forest downwind of the reactor turned brown and died.
  - Horses and cattle near the plant died from radiation damage to their thyroid glands.
  - Bioaccumulation of radioactive cesium in fish (Scandinavia and Germany) and lamb (Wales) - contaminated meat was banned from sale for years afterward.
  - Drinking water (and milk) contaminated with radioactive iodine - at least 6,000 thyroid cancer attributed to radioactive iodine.

Gregor Mendel

- An Austrian monk.
- Formulated two fundamental laws of heredity in the early 1860s.
- Previous theories based on the Blending Concept of Inheritance (Tall + Short = Medium)
- The blending theory did not account for variation, does not explain species diversity
- Mendel’s work was unrecognized until 1900; Darwin was never able to use it to support his theory of evolution.

Mendel’s Work

- Used garden peas
  - Easy to cultivate
  - Short generation time
  - Can be cross-pollinated and self-pollinated
  - Mendel cross-pollinated plants.
    - P1 generation is the parental generation.
    - F1 generation is the first-generation offspring.
    - F2 generation is the second-generation offspring.
  - Mendel’s results were contrary to those predicted by a blending theory of inheritance.

- He found that the F1 plants resembled only one of the parents.
- Mendel’s Conclusions
  - F1 hybrids contained two factors for each trait
  - One dominant and one recessive factors separated when gametes were formed
  - A gamete carries one copy of each factor
  - Random fusion of all possible gametes occurred upon fertilization.
**Mendel’s Law of Segregation**
- Organism contains two factors (alleles) for each trait.
- Factors segregate during formation of gametes.
- Each gamete contains one factor for each trait.

**Mendel’s Law of Independent Assortment**
- Each trait is independent of another.
- Members of one pair of factors assort independently of members of another pair.
- All combinations of factors occur in gametes.
- Meiosis explains these results of independent assortment.

**Monohybrid Crosses**
A. Homozygous x Homozygous
B. Homozygous x Heterozygous
C. Heterozygous x Heterozygous
D. Testcross

**Dominance Has Degrees**
- Incomplete dominance
  - Offspring intermediate between two parental phenotypes.
  - Blending
- Codominance
  - Both alleles of a gene are expressed.
  - F₁ hybrid is different from both parents and not a blending of traits.

**Tay-Sachs Disease (Chromosome 15)**
- Autosomal Recessive
- Usually among Jewish people in the U.S. of central and eastern European descent
- Lipid accumulation in the brain
- Symptoms not initially apparent
- Development slow at 4-8 months, child gradually becomes blind, develops seizures, eventually becomes paralyzed, dies by age of three or four.
- No treatment or cure

**Cystic Fibrosis (Chromosome 7)**
- Autosomal Recessive
- Most common lethal genetic disease in Caucasians in U.S.
- About 1 in 20 Caucasians is a carrier, and about 1 in 2,500 births has this disorder.
- Production of viscous form of mucus in the lungs and pancreatic ducts.
- New treatments have raised average life expectancy to 28 years.
Huntington Disease (Chromosome 4)
- Autosomal Dominant
- Affects one in 20,000.
- Progressive degeneration of brain cells, which in turn causes severe muscle spasm, personality disorders, and death.
- Most appear normal until they are of middle age.

Sickle Cell Anemia (Chromosome 11)
- Autosomal Codominance
- Blood cells are abnormal
- Symptoms are anemia, weakness, heart attack.

Sex-Linked Traits
- Chromosomes can be categorized as two types
  - Autosomes
    - Nonsex chromosomes
    - Same number and kind between sexes.
  - Sex chromosomes
    - Human female is XX
    - Human male is XY.
    - Carry genes for traits unrelated to sex.

Sex-Linked Traits
- Thomas Hunt Morgan
  - Work with fruit flies showed XY system similar to humans.
  - Newly discovered mutant male fruit fly had white eyes.
  - Cross of F1 male with F1 female
    - Yields 3:1 red-to-white ratio
    - All white-eyed flies were males
  - An allele for eye color on the X but not Y chromosome supports the results of the cross.

Color Blindness (X Chromosome)
- Sex-Linked Recessive
- Mutations of genes coding for green or red-sensitive cone cells.
- Inability to perceive green or red.

Hemophilia (X Chromosome)
- X linked recessive
- About 1 in 10,000 males.
- Impaired ability of blood to clot.
- Hemophiliacs bleed externally after an injury and also suffer internal bleeding around joints.
Dihybrid Cross

Feather color = ORANGE or Blue, CREST or no crest

Trihybrid Cross

Moving Away from Punnett Squares

A. Multiplicative Law of Probability
   1. The chance of two or more independent events occurring together is the product of the probability of the events occurring separately.
   2. Chance of inheriting a specific allele from one parent and a specific allele from another is 1/2 x 1/2 or 1/4.
   3. Possible combinations for the alleles $Hh \times Hh$ are the following:
      - $HH = 1/2 \times 1/2 = 1/4$
      - $hH = 1/2 \times 1/2 = 1/4$
      - $Hh = 1/2 \times 1/2 = 1/4$
      - $hh = 1/2 \times 1/2 = 1/4$

B. Additive law of probability
   1. The probability of an event that occurs in two or more independent ways is the sum of individual probabilities of each way an event can occur.
   2. In the above example where Tall is dominant ($HH$, $hH$, and $Hh$), chance for Tall is $1/4 + 1/4 + 1/4 = 3/4$.

C. EX: $SsYyAa \times SsYyAa$; odds of child being $SSyyAa$ (1/32)

A. Show pattern of inheritance within a family.
   1. Males are designated by squares, females by circles.
   2. Shaded individuals are affected.
   3. A carrier has no apparent abnormality but can pass on an allele for a recessively inherited genetic disorder.
   4. Autosomal dominant and autosomal recessive alleles have different patterns of inheritance.

D. Characteristics of autosomal dominant disorders
   1. Affected children must have one affected parent.
   2. Heterozygotes are affected.
   3. Two unaffected parents can produce only unaffected child.

E. Characteristics of autosomal recessive disorders
   1. Affected children can have normal parents.
   2. Two affected parents always produce an affected child.
   3. Close relatives who reproduce together are more likely to have affected children.

A. Trait is controlled by several allelic pairs at different loci.
B. Allele can be at different loci on a chromosome or on different chromosomes.
C. Examples include skin color, eye color, and height.
Sickle Cell Anemia (a SNP)

- Causes of sickle cell anemia
  - A base substitution mutation of DNA.
  - Changes the base sequence of mRNA transcribed from it.
  - Produces a different AA. (Val instead of Glu)

Mutagens

- Agents that cause gene mutations:
  - Chemicals (carcinogens)
  - High energy radiation such as X-rays
  - Short-wave ultraviolet light
  - Some viruses (HPV)