

## Unit 5: Mendelian Genetics

### Content Outline: Mendelian Genetics (5.2)

- I. **Complete Dominance**
  - A. The dominant allele has information and it is *expressed*, even if there is only one copy. Geneticists call this “100% penetrance”.
- II. **Incomplete Dominance**
  - A. Information from *both alleles is expressed* in the cell. Neither phenotype is *completely* penetrant, therefore the heterozygous phenotype appears “blended”. (Red + White = Pink)
- III. **Codominance**
  - A. *Both alleles are expressed* in the cell. They are both equally present in terms of phenotype, i.e. black and white coats in animals, AB blood type in humans.
- IV. **Multiple Alleles**
  - A. There are *multiple (more than 2)* versions of the *same basic allele*.
  - B. The glycoprotein “hands” of red blood cells is a classic example. These “hands” (antigens) identify the blood types. One dominant allele results in the presence of A antigens on the surface of red blood cells. Another dominant allele results in the presence of B antigens on the surface of red blood cells. A *third* allele results in no antigen and is a recessive allele. This condition is referred to as Type O. Since it is recessive, the gene or stretch of DNA that codes for these proteins has no code for A or B antigens, so *neither is made*.
  - C. **Universal Donor** – Can **give** blood to anyone. Since the O type blood has neither A nor B antigens, it does not cause a reaction in the bodies of people who have A, B, AB or O blood types.
  - D. **Universal Recipient** – Can **receive** blood from anyone. Since the blood cells have both A and B antigens, there is no reaction if those cells are mixed with A, B, AB, or O blood types.
  - E. **A bag of golf tees in two different colors works well here. A is one color and B is another color; but both are still tees. O would be like a hand holding no tee. Show A hands (tee in hand). Show B hands. Show AB hands. Show O hands. Now talk about various situations with donating blood.**
- V. **Pedigree**
  - A. This is a *family history* of trait occurrence in chart form.
  - B. These help tell the *past occurrences* of traits and can be useful in *predicting the future occurrence* of traits.
  - C. The power of pedigrees is their ability to predict possible genotypes and thus phenotypes of future generations.
- VI. **Recessive Disorders**
  - A. Only occur in the *homozygous recessive* state.
  - B. **Carriers** – These are organisms that are *heterozygous* in genotype. (They are 50/50 in terms of passing on an allele for the trait.) These organisms usually *appear normal* for the trait as they possess one dominant allele.
  - C. Human recessive disorders: **(Students just need to be familiar with the disorder and not memorizing numbers.)**
    1. **Cystic Fibrosis** (Also referred to as “CF”).
      - a. This is the *most common lethal genetic disease* among people of European descent.
      - b. This disorder affects 1 in 2,500 births.
      - c. In Caucasians, 1 in 25 people is a carrier for the disorder.
      - d. The disorder creates a faulty Chloride ion (Cl<sup>-</sup>) protein carrier on cell membranes in the lungs. This causes fluid (water) to build up in the lung tissues.
        - i. People drown in their own fluid.
        - ii. They are also prone to get multiple infections in the lungs.
      - e. Treatment? Since it is *genetic* there is no cure. Patients have to get the fluid drained from the lungs periodically for their *entire* life. There are medicines to help *reduce the number of times* this has to occur. **(Students should understand the difference between disease and disorders.)**
    2. **Sickle-cell Disease**
      - a. This disorder is the *most common genetic disorder among people of African descent*.
      - b. It affects 1 in 400 births.
      - c. The 6<sup>th</sup> Amino Acid in the hemoglobin molecule is changed (Glutamine → Valine) in the **primary sequence** needed to make red blood cells. (The easy way to remember this is: 666 is the number of the beast. 6 is the amino acid that changed to create this horrible disease. It went from good [glutamine] to very bad [valine].) **(Quickly review protein structure and point mutations.)**
      - d. **Sickle-cell trait** (“trait” is used to refer to individuals that are **carriers**).
        - i. These individuals have resistance to Malaria because of the *one* recessive allele they possess but mainly have normal red blood cells for carrying oxygen.

- ii. This is referred to as the **Heterozygote Advantage**. They have an *advantage* over individuals that are homozygous dominant or homozygous recessive. Homozygous dominant are not resistant to Malaria. *Homozygous recessive are also resistant to Malaria; BUT they have the disease to contend with.*
- e. These sickle shaped cells have *reduced oxygen carrying ability*. They also are painful when the points of the cell jab into the walls of the blood vessels.
- f. Treatment? There is no cure as it is genetic. Some medicines help with the pain or low oxygen levels.

VII. Dominant Disorders

- A. Only need *one* allele for these disorders to be present or “expressed”.
- B. If an individual is homozygous dominant, usually the disease is much worse and often fatal.
- C. Human Dominant Disorders: **(Again understand the concept; not memorizing numbers.)**
  - 1. **Achondroplasia** (This is referred to as **Genetic Dwarfism**.)
    - a. This disorder affects 1 in 10,000 births.
    - b. Most people are *homozygous recessive* and there for much taller than these individuals.
  - 2. **Huntingdon’s Disease**
    - a. This disorder affects 1 in 10,000 births.
    - b. It has a late life onset – usually in the 40-50 age range. (Usually *after* children are born.)
    - c. **(Please talk about the need to have counseling (below) if it exists in the family history.)**
    - d. The dominant gene has a locus on tip of Autosome 4.
    - e. Family history is important in diagnosis of this disorder. (Pedigree can help.)
    - f. It is a slow *degenerative disorder affecting the brain* that is almost always fatal.

VIII. Genetic Counseling and Counselors

- A. These are individuals who run pedigrees, using family history data, to look at potential outcomes or carrier recognition and give advice on whether or not to pursue trying to have children based on the “probability” of a genetic disorder occurring within the offspring.
- B. *Remember, chance has no memory*. Each mating can potentially have a different outcome.
- C. Need to perform karyotypes to help with confirm diagnosis sometimes.