SAT Biology Review: Genetics & Evolution

I. Genetics

A **gene** is a sequence of DNA that encodes a single product, most typically a protein. **Mutations** are changes in the DNA sequence of the gene. Sometimes the mutation causes a slight variation in the protein, making it work slightly better or slightly worse than before. Other times, the mutation can be profound, destroying the protein. Examples of mutations are point mutations, which can be silent, missense, or nonsense, and insertions and deletions which may lead to frameshift mutations.

Genetics is the study of how organisms pass their genes to their offspring. There are many different inheritance patterns, the most common of which are described below.

Mendelian Genetics

The Austrian monk **Gregor Mendel** first described the inheritance patterns of an organism with his study of pea plant *Pisum sativum*. Mendelian genetics studies **traits** that are inherited in a paired, binary fashion. For example, pea plants tend to grow to a set height, and can be either tall or short (there are no intermediate sizes). Tall and short are the two **alleles** for the plant height gene. In the pea, alleles are named using the same one letter abbreviation, with the dominant gene written as a capital letter, the recessive in lowercase. For height, tall is written as T and short as t.

Eukaryotic organisms usually have two copies of each gene (**diploidy**), one inherited from the father and the other from the mother. The combination of alleles in an individual is called its **genotype**. If an individual has two identical alleles, they are **homozygous** (TT or tt). If they have one of each allele, then they are **heterozygous** (Tt). The phenotype of an organism is how it appears (its **ph**ysical appearance= **ph**enotype).

In Mendelian genetics, one allele is always **dominant** to the other, which is called **recessive**. That means that the dominant trait will always be expressed, even if both are present in heterozygotes. In our example, tall is dominant to short, so a heterozygote (Tt) would be phenotypically tall.

Parents give only one of their two alleles to their child. This is termed the **law of segregation**: organisms segregate (divide) their two alleles randomly (with independent probability). Since Mendelian inheritance is nothing more than probability, it is possible to predict all possible offspring of a mating, and the probability of having each type of offspring. This is traditionally done with a **Punnet square**.

50% Chance of having a Tall plant
50% Chance of having a Short plant

If you have many offspring from this mating (as with plants or insects), you would expect about half to be tall, and about half short.

It is possible to study the simultaneous inheritance of two or more genes. **Unlinked genes** (genes that are not located close to each other on a chromosomal arm) obey the **law of independent assortment**. In other words, they behave without regard to the other: they are independent. An expanded Punnet square is used for this analysis. The hardest part of this experiment is determining all possible combinations of the gametes. **Linked genes** are close together on the chromosome, and are inherited together.

Many traits, particularly in humans, do not follow the standard Mendelian rule of one dominant and one recessive allele. These alternate inheritance patterns are termed Non-Mendelian, and the most common are described below.

Polygenic Inheritance

Many human traits are controlled by more than one gene. Examples are human height, human skin color, human muscle mass, and timing for developmental stages such as the onset of puberty, end of puberty, and senescence (old age). If there is a range of many different phenotypes for a trait (think of all the variations in height), then the trait is polygenetic.

Incomplete Dominance

Also known less correctly as blending, this pattern occurs when heterozygotes have a different, intermediate phenotype than the homozygous recessive and homozygous dominant organism. An example is the Japanese four-o'clock flower, which can have three different flower colors: red (RR), pink (Rr), and white (rr). If there are three possible phenotypes for a trait, then it is controlled through incomplete dominance.

Codominance

In certain situations, the alleles for a trait are equally strong, and when both are present in heterozygotes, both are expressed equally. The best-known example of codominance is human blood types. Three different alleles control blood type: I^A, I^B, and i. I^A encodes for the A marker of the surface of red blood cells, and I^B the B marker. i is a damaged gene that cannot create a marker, and thus is useless. When A and B are both present in a heterozygote ($I^A I^B$), both A and B markers are made and placed on the cell surface; this person is blood type AB. Type A blood can be encoded by either $I^A I^A$ or $I^A i$, and Type B by $I^B I^B$ or $I^B i$. Type O people have two copies of the damaged allele, ii. If there are more than three distinct phenotypes (but not a broad range of hundreds as seen in polygenic) than the inheritance pattern is controlled by incomplete dominance. You will usually only see questions on incomplete dominance asked about blood type.

Sex Linked traits.

There are two sex chromosomes in humans, the X and the Y. XX is female, XY is male. The mother gives one of her X chromosomes to her child; the father randomly gives the child either his X or his Y (thus the father determines the gender of offspring in humans). If a trait is carried on either of these chromosomes, it is inherited along with the child's gender and the trait is said to be sex linked. This is particularly important with recessive diseases that are carried on the X chromosome. Hemophilia (X^h) and color blindness (X^c) are two famous examples. Because they have two X chromosomes, females are afforded a certain protection from these diseases: heterozygotes ($X^h X$) are healthy but are called **carriers** because they can pass the gene to their children. The male Y chromosome cannot compensate for a diseased X: $X^h Y$ males have the disease.

II. Evolution

Historically, there have been two widely discussed theories of evolution. **Lamarck** proposed that organisms passed on characteristics they acquired during life to their offspring. It was commonly called the theory of use and disuse. If a giraffe constantly stretched his neck, he would pass on this elongated neck to his children. Birds that stopped flying passed on useless wings to their chicks. This theory was proven untrue.

Charles **Darwin** proposed what is now called the theory of **natural selection** or "**survival of the fittest**."
- ❖ Far more organisms are born than can possibly survive
- ❖ All organisms compete with other species and within their own species for resources and mates
- ❖ There is much variation among organisms of the same species. This diversity is heritable.
- ❖ Only those who are best suited to the environment live to pass their better adapted genes to the next generation. Those who are poorly suited to the environment die, taking their bad genes with them.

Mutations, which constantly and normally occur, affect the fitness of different organisms. Mutants that are less suited than their peers die. Those that are more suited to the environment than their peers out compete them, and thus pass on their better genes more often. The modifications that better allow an organism to survive in its niche are called **adaptations**. The **Evolutionary Fitness** of an organism can be measured by how many offspring they leave behind—the more offspring, the greater the evolutionary fitness.

If enough adaptations accrue in an isolated population of organisms, then they may **speciate**, or change into a new species. A **species** is a group of similar organisms that can mate to produce fertile offspring. Usually, in order for speciation to occur, a group of life forms must be **reproductively isolated**. This can occur through

allopatric speciation, where two populations of the same organisms become physically separated (as by a new river, canyon, or mountain range), or by **sympatric speciation**. Sympatric speciation requires that organisms be in the same area, but reproductively isolated (as by changing their mating season, courtship rituals, or through genetic or mechanical incompatibility).

Evolution can be best described as changes in the frequency of alleles in an organism's gene pool. Genetic drift can cause changes in allele frequency without evolution—it does so through random chance. Two common examples are founder's effect and bottlenecks. **Founder's effect** describes the results of a small group of organisms establishing a colony in a new area; by chance, the founders may have a different proportion of alleles than the original population (for example, red hair in the colonists of Ireland). Another way is through a **bottleneck**: a calamity decimates a large group of organism, leaving only a handful behind.

If a group of organisms becomes especially well-suited to their environment, they can quickly spread to new areas. This is called an **adaptive radiation**. Examples of prior adaptive radiations are the spread of the seed plants and the mammals.

III. Hardy-Weinberg Equilibrium

Hardy-Weinberg equilibrium is a mathematical expression that describes the proportion of alleles in a population. It only works in certain situations:

- ❖ Populations are large, and have low mutations rates
- ❖ Mating is random
- ❖ The alleles being studied are NOT under natural selection
- ❖ There is no migration into or out of the area (no new additions or subtractions to the gene pool)

[Note that this equation rarely works with humans: we do not mate randomly. We select mates based on specific criteria, many of which have a genetic basis.]

The Hardy-Weinberg equation is:

(i) $p^2 + 2pq + q^2 = 1$ Which describes AA + 2Aa + aa = 1, all possible genotypes.

(ii) $p + q = 1$ Which describes the distribution of alleles in a gene pool.

where   p = the frequency (written as a decimal) of the dominant allele in the population
q = the frequency of the recessive allele in the population.

If the alleles are in equilibrium, then equation (i) will equal 1. If the alleles are not in equilibrium, which usually means that those alleles are being acted on by natural selection, the equation will not be equal to 1.

Example:

Phenylketonuria (PKU) is a recessive disorder that affects 1 in 10 000 births in the United States. What is the frequency of the carriers (heterozygotes) in the US population?

Since PKU is a recessive disorder, people with PKU must be homozygous (pp). The frequency of the q^2 term, then, is 1/10 000, or q^2 = .0001 and by taking the square root q = .01

You can find the frequency of the healthy, dominant allele with equation (ii).
p + q = 1 → p + .01 = 1 →p = .99

The carriers are heterozygotes, and are described by the 2pq term of equation (i).
2pq = 2•.99•.01 = .0198 or ~2%

Practice problems: (Answers are in the box below)

1. In a population with two alleles for a particular gene, B and b, the frequency of B is 0.7. What is the frequency of heterozygotes if the population is in Hardy-Weinberg equilibrium?

(A) 0.7   (B) 0.49   (C) 0.21   (D) 0.42   (E) 0.09

2. In a population that is in Hardy-Weinberg equilibrium, 16% of the individuals show the recessive trait. What is the frequency of the dominant allele in the population?

(A) 0.84   (B) 0.36   (C) 0.6   (D) 0.4   (E) 0.48

Remember: calculators are not allowed on the SAT II test. Therefore, all math problems must ones you can solve in your head or on paper. This means that H-W math must be simple. For example, if you have to take the square root to find p or q, the number is going to be a perfect root. If you find yourself doing a long, complicated solution, you are making a mistake somewhere.

1.D 2. C