Stanford CS 228, Winter 2011-2012 Programming Assignment 2: Appendix

1 Overview of Genetics

In humans, the DNA is arranged in twenty-three pairs of chromosomes. A person inherits one chromosome in each pair from his/her mother and one from his/her father (there are rare exceptions to this, but we will assume that this is true for the purposes of this assignment). Twenty-two of the pairs of the chromosomes consist of autosomes. The genes on these autosomes are the same across males and females. The final pair consists of the sex chromosomes. Females have two X sex chromosomes, and males have an X sex chromosome and a Y sex chromosome. Thus, every mother passes a copy of autosomes 1-22 and an X chromosome to each of her children. Every father passes on a copy of autosomes 1-22 to all of his children, a copy of the X chromosome to his daughters, and a copy of the Y chromosome to his sons. As a result, each person has two copies of every autosomal gene – one from his/her mother and one from his/her father. Every female also has a copy of each gene on the X chromosome from each parent; every male has a copy of each gene on the X chromosome from his mother and a copy of each gene on the Y chromosome from his father. **Note that, for the purposes of this assignment, we have greatly simplified many aspects of genetics. Therefore, many of the descriptions here and in the assignment are only rough approximations of how genetic inheritance actually works.**

2 Dominant and Recessive Alleles

There can be multiple versions of each gene. Versions of a gene are called alleles. In simple Mendelian genetics, a trait is controlled by one gene with two alleles. For example, there is (as far as we know today) a single gene for face freckles. Say this gene has two alleles, F and f, where F makes someone have face freckles and f makes someone not have face freckles. A person's genotype is the combination of alleles that person has, and a person's phenotype is the physical expression of those alleles. For example, if a person's genotype is FF, meaning that he/she inherited a copy of a gene that is the F allele from each parent, then the person's phenotype is face freckles. If a person's genotype is ff, then the person's phenotype is no face freckles. A person has $\binom{n}{2} + n$ genotypes, where n is the number of alleles, because there are $\binom{n}{2}$ unique combinations of pairs of alleles with no repeats and n combinations of pairs of alleles in which both alleles are the same. A person with genotype FF or ff is called homozygous for the gene for face freckles, meaning that he/she has two copies of the same allele.

What happens if a person inherits an F allele from one parent and an f allele from another, making the person's genotype Ff? In this simple example, we will assume that alleles can be either dominant or recessive. A dominant allele is always expressed, and a recessive allele is expressed only when the dominant allele is not present. We will assume that F is dominant, and f is recessive. Thus, if a person's genotype is Ff, the F allele will be expressed, so the person will have face freckles. A person with genotype Ff is called heterozygous for the gene for face freckles, meaning that he/she has one copy of each allele.

3 Non-Mendelian Inheritance

In addition, inheritance of some traits is not Mendelian. Consider the trait of polydactyly, which is the trait of having an additional finger on one or both hands or an additional toe on one or both feet. (Note that the follwing description is an over-simplification of how the inheritance of polydactyly actually works.) Say there are two alleles for this gene, P and p, where P is dominant. Say that allele P does not cause someone to have an extra finger but is just a risk factor, while allele p might not prevent someone from having an extra finger but just make the person less likely to have an extra finger. As a result, people who are PP or Pp might have an extra finger with probability 0.8, and people who are pp might have an extra finger with probability 0.1.

4 Allele Frequncies

For most genes, some alleles occur more frequently than others. This can happen for many reasons. One reason is that one phenotype might have some kind of evolutionary advantage.

5 Sex-Linked Traits

Some genes are found on the X chromosome. Traits controlled by genes on the X chromosome are called "X-linked." A famous example of an X-linked trait is color-blindness. The allele for color-blindess, call it b, is recessive, and its corresponding allele for non-color-blindness, call it B, is dominant. A female will therefore not be color-blind unless she has two b alleles. A male, however, will be color-blind with only one b allele because he has only one X-chromosome, so he cannot have a corresponding B allele. There are also a few traits that are linked to the Y chromosome, which are called "Y-linked;" women cannot have these traits, and men will have the phenotype for these traits that corresponds to the one allele that he inherited from his father.

6 Multi-Gene Traits

Many traits are controlled by multiple regions of the genome. For example, there is so much variation in hair color because hair color is controlled by multiple genes. For the purposes of this assignment, we will consider only traits controlled by genes that are unlinked, meaning that they are found on different chromosomes or that they are found far enough from each other on the same chromosome that they can be considered independent when computing the probability that their alleles will be passed on to the next generation. There are many other complex forms of inheritance, such as mitochondrial inheritance, co-dominance, and traits controlled by linked genes, but we will not consider these during this exercise.