Chapter 1

Genes, Pedigrees and Genetic Models

1.1 DNA, alleles, loci, genotypes, and phenotypes

The *DNA* in the nuclei of cells of an individual consists of about 3×10^9 base pairs (bp). This *DNA* is packaged into *chromosomes* each of which has a linear DNA sequence in a twisted double-helical structure. There are 46 chromosomes in the nucleus of each normal human cell, 22 pairs of *autosomes* and a pair of *sex chromosomes*. Of the two chromosomes of a pair, one derives from the DNA of the mother of the individual and the other derives from the DNA of the father. In this book, we will restrict attention to the autosomes, which contain the majority of the DNA coding for the proteins and affecting the characteristics of individuals. Similar approaches would apply to the sex chromosomes, but the details differ. There is additional DNA in the mitochondria, which are located in the cytoplasm of the cell; mitochondrial DNA is maternally inherited.

Any small segment of the DNA of the chromosome is known as a *locus*. Typically, a locus used to refer to the segment of DNA coding for some functional protein, but it is now used to refer to any position characterized by a specific DNA sequence, or by specific forms of variation in the sequence. These loci exhibiting observable variation in the DNA are *DNA marker loci*, and a *locus* simply indicates a particular position on a particular one of the pairs of chromosomes. The DNA at a locus may come in a variety of forms, or *alleles*. Any individual has two chromosomes of a given pair, and thus has two (possibly identical) alleles at each locus. The unordered pairs of alleles that an individual has is the individual's *genotype* at this locus. If the locus is one relating to a functional gene, the resulting potentially observable characteristic of the individual is the *phenotype*. A locus exhibiting non-negligible variation in a population is known as a *polymorphism*, or the locus is said to be *polymorphic*. Classically, the frequency of the the most frequent genotype should