Chapter 3

Gene Identity by Descent

3.1 Kinship and inbreeding coefficients

A *gene*, as opposed to an allele or a locus, is the *DNA segment* that is copied from parents to offspring. Underlying the patterns of phenotypes observed on related individuals are the *genotypes*, but underlying the genotypes are the patterns of gene identity by descent. Phenotypes of relatives are similar because they have similar genotypes and may share a common environment. Genotypes are similar because relatives share genes that are identical by descent (*ibd*) — identical copies of a gene segregating from a common ancestor within the defined pedigree. Although for some microsatellite DNA markers mutation rates are non-negligible (section 1.1), for simplicity we disregard mutation throughout this book. In this case, genes that are *ibd* must be of the same allelic type, while genes that are not *ibd* are of independent allelic types.

Gene identity by descent is defined only within the context of a given pedigree structure. A pedigree specifies the two parents of every non-founder individual. A founder has neither parent specified, and by definition the genes in founders are not *ibd*. It will often be convenient if a pedigree is ordered in such a way that every individual is preceded in the listing by his parents; clearly, this is always possible.

Mendel's First Law (section 1.2) states that:

> a diploid individual receives at any given locus a copy of a randomly chosen one of the two genes in his father and (independently) a copy of a randomly chosen one of the two genes in his mother, and will pass on a copy of a randomly and independently chosen one of these two genes to each of his offspring.

This simple law leads to complex patterns of gene identity on an extended pedigree, due to the huge number of alternative events; 2^m for m meioses, at each locus. The segregating genes determine the patterns of gene identity by descent on the pedigree, and hence the patterns of similarity among relatives.

We start with coefficients of *inbreeding* and *kinship*, since these provide an introduction to the ideas of gene identity by descent, to alternative computational