Relationship identification using linked autosomal markers

Nuala A Sheehan [1] and Øivind Skare[2] and Thore Egeland[2]

[1] Department of Health Sciences, School of Medicine, University of Leicester, UK
[2] Department of Medical Genetics, Ulleval University Hospital, Oslo, Norway

Likelihood-based approaches to determining the relationships between specified individuals from DNA marker data are of central importance in many situations. Applications are diverse in behaviour, evolution and conservation research and include forensic problems ranging from standard paternity cases to inheritance claims, immigration cases and identification of remains following disasters. For human applications, standard forensic unlinked micro-satellite markers often suffice. These are selected to be highly polymorphic and hence so informative that typically no more that 15 markers are required. However, it is not possible to distinguish between certain relationships, such as half-sibling and uncle-nephew, on the basis of *any* number of independently segregating loci. Large numbers of linked markers are possibly required to distinguish between alternative relationships with equal likelihoods for independent loci. We argue that linked markers should be given greater attention for relationship estimation, especially now that the required laboratory and statistical analyses can be performed using existing technology and freely available software. We also note that SNP markers, although individually less informative than micro-satellites, can also be used because of the large numbers of SNPs that are now readily available.

Although our primary focus is on forensic applications where the precise relationship is often crucial, the exploitation of linked markers for relationship estimation is also relevant for other applications. For example, identification of sets of related individuals from large population biobanks for homozygosity mapping is also important as they are more likely to share larger segments of genome around disease susceptibility alleles with relatively small effects on the common complex diseases of current public health interest.