INTRODUCTION

Over the last 20 years or so there has been a growing realisation among professionals of the need to maintain accurate records of families with genetic disease. This idea is not new, in fact Joseph Adams (1756–1818) wrote in 1814:

That to lessen anxiety, as well as from a regard to the moral principle, family peculiarities, instead of being carefully concealed should be accurately traced and faithfully recorded, with a delicacy suited to the subject, and with a discrimination adopted to the only purpose for which such registers can be useful (Adams, 1814).

The need to maintain records of affected families became especially important with the advent of prenatal diagnosis, which offered the very real possibility of preventing further cases in a family. Recently the introduction of recombinant DNA technology has emphasised even more the importance of genetic registers because they provide the means for storing DNA data on affected families. According to the Oxford English Dictionary, a register is ‘A book or volume in which regular entry is made of particulars or details of any kind which are considered of sufficient importance to be exactly and formally recorded’, but it can also refer to the setting down, entering or recording of facts in a precise manner. A registry on the other hand refers to ‘The act of registering or a place where registers are kept’. The preferred term in the present context is therefore register rather than registry.

There are two types of genetic registers, general and specific. The former is designed to include all serious genetic disorders within a given region. The latter is usually concerned with only a single specific disorder. Each type of register is associated with certain advantages and disadvantages.
A general genetic register in a genetic Centre can reflect its responsibility for inherited diseases in general. It can be argued however that few such Centres have the expertise or laboratory capabilities of dealing adequately with all genetic disorders. But the author’s experience with a general genetic register has shown that 80 per cent of the families included in the system could be accounted for by only 11 disorders. Seven of these were autosomal dominant disorders, often of late onset (Huntington’s chorea, polycystic kidney disease, myotonic dystrophy, neurofibromatosis, polyposis coli, retinitis pigmentosa and Marfan’s syndrome), three X-linked conditions (haemophilia A and B, Duchenne muscular dystrophy and retinitis pigmentosa), and familial translocations in Down’s syndrome. No doubt this particular spectrum of disorders to some extent reflects this Centre’s interests. Nevertheless it seems that even when an effort is made to include all serious genetic disorders in a given population, in effect most attention will still be concentrated on a limited number of conditions. Autosomal recessive disorders are likely to play an important role only when population screening for heterozygous carriers becomes feasible. Until then, effort will usually be limited to preventing second cases in sibships in which an affected child has already occurred.

Several general genetic registers have been established in various Centres throughout the world (Table 12.1). Some of these appear to be maintained largely for administrative purposes for ease of storage and retrieval of clinical, genetic, and laboratory data. The GENFILES system in San Francisco, for example, consists of data files on cytogenetics (CHROMO), genetic diagnosis and counselling (MEGDEN, Mitchell et al., 1980), prenatal diagnosis (AMNIO), and frozen cells (FROZEN). The MEGADATS system has evolved over the years as a very comprehensive system for record-keeping purposes in which is now stored information on individuals who have banked DNA for use in future genetic tests (Yount et al., 1987). This type of system has the great advantage of facilitating the rapid retrieval of patient and family data, particularly from incomplete identifying data. Such dedicated data bases will become increasingly important in future because ‘Not only will they be the means for reducing the clerical work load but they will also become the data bases for research, administrative and service orientated functions’ (Mutton et al., 1988). One such general genetic register (RAPID)