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Editorial

The Human Genome Project is an ambitious joint venture between the UK and numerous centres in the USA which commenced in 1990. The aim is to provide comprehensive and detailed information about the DNA making up the 23 pairs of human chromosomes. The sequence of base units of which DNA is composed code for the 70 000–100 000 individual genes which form the human genome. At the end of the initiative each gene will be identified, its position mapped and information about its precise function will be determined. It has already been established that, for many genes, deviation in the sequence of base pairs will result in abnormal function which in some cases is sufficient to cause the clinical manifestations of disease.

The Human Genome Project is the largest co-ordinated biological project which has ever been undertaken. It is anticipated that by the year 2005 the entire human genome will have been sequenced and the functions of every gene will be known. Currently at least 5000 single gene disorders have been identified. Soon it will be possible to specify many more conditions that to a greater or lesser degree are genetically influenced.

The information which has so far been generated by the Human Genome Project is already creating significant changes in health care (Bell, 1998) and has been identified as the major factor now driving organizational change in the National Health Service in the UK (Kirk, 1999). For example, it has become apparent that most of the common serious diseases such as diabetes, cardiovascular conditions and many cancers are to some extent inherited (Kinmouth, 1998). This does not mean that the individual carrying the affected gene will inevitably develop the condition it determines. Instead the person concerned will be placed at increased risk of acquiring symptoms and will do so if exposed to an environmental insult which operates as a trigger. This explains why some smokers do not develop lung cancer (those having an affected gene and meeting the trigger in cigarette smoke will develop lung cancer; those without the affected gene will not be at the same degree of risk despite exposure to the same carcinogens). It also explains why many conditions, such as cancers, tend to develop with increasing age (the longer an individual lives, the greater their chance of encountering the environmental trigger which will interact with their affected genes). To make matters more complicated, most of the common conditions are coded not by a single gene, but by many small genes operating together and with the environment. This is the situation with hypertension. Many factors, both genetic and environmental, contribute to this condition, but the clinically significant outcomes – elevated blood pressure and its sequelae – are the same in each case.

The availability of new technology to locate and identify genes will provide opportunities for genetic risk assessment. Ultimately it will even be possible to develop specific, targeted treatment regimes to prevent many genetic conditions (Kinmouth, 1998). It will also be possible to establish sub-classifications for known diseases that at present are clinically indistinguishable. Thus we will be able to predict which individuals will develop hypertension as they age. Treatment and preventative strategies can then be employed accordingly. In

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some cases of hypertension, perhaps where genetic influence is slight, dietary modification and stress reduction can be recommended as the most effective approach. In other cases, where different genes are contributing, medication will be advisable. As almost everybody has some genes which place them at increased risk of developing some disorder with a genetic component, the scope for prediction will be enormous.

Advances in genetics research have major implications for society and will be associated with ethical and legal scenarios of which all health professionals need to be aware. Many such issues relate to third party access to information (such as employers and insurance companies). Other issues raise concern over revealing possibly unsolicited genetic information to individuals, especially where treatment will not become available for some time or is limited through cost. Special problems will arise in relation to screening children and others with a limited ability to comprehend complex genetic issues, raising questions about informed consent (Patenaude, 1996). This issue is likely to be of particular concern given the large number of disorders which result in learning difficulties and problems related to mental health. Changes will also occur in the way in which health promotion and preventative care are delivered (Patterson *et al*., 1999), demand for services (Graham *et al*., 1998) and thus for public health policy (Kinmouth, 1998).

At present, predictive testing is available for only a few conditions in the UK and there is worrying evidence that for these, clients' needs are not being met, particularly in terms of the availability and quality of information provided (Atkin & Anionwu, 1998; Harris *et al*., 1999). This is a situation which urgently needs to be addressed because in future all members of society will require adequate information to enable them to make informed genetic choices appropriate to their specific circumstances (Rieger & Pentz, 1999). One of the first hurdles to tackle may be that of commercially available genetic testing kits. Some are already obtainable and the range is likely to increase (Kirk, 1997). Being able to determine their genetic status may be of interest to individuals and of benefit to their families, but will introduce novel problems. To whom does the information belong? Suppose it provides one member of the family with information about another which he or she may not want known or not wish to know themselves? The results of a survey conducted in the USA indicate that members of the public who know they are likely to be at increased risk of developing an inherited condition (cancer of the colon) are interested in commercial testing to take preventative action if necessary, for peace of mind and for reasons of curiosity (Graham *et al*., 1998). The same may well be true for other genetic conditions and for people in the UK.

It is becoming apparent that clinical genetics can no longer be regarded as a peripheral speciality concerned with rare disorders, but has implications for all health professionals. Nurses will be particularly affected through their responsibility to provide health promotion at the level of the individual and their contribution to public health (Kirk, 1999). In future they will be required to help people decide whether to undergo genetic testing, to help them understand the results and to come to terms with the implications for themselves and their families. Education is clearly needed to equip nurses for this demanding role and must go far beyond mere descriptions of genetic conditions and mechanisms of inheritance.

The need for nurses to achieve competency in genetics has been recognized in the USA (Rieger & Pentz, 1999) but so far there appear to be few plans to extend genetics education to non-specialist nurses in the UK, according to the results of

a recent survey of pre-registration diploma nursing curricula (Kirk, 1999). Its findings indicated that, while genetics is generally included, the content and mode of delivery vary and compulsory assessment is omitted. This survey examined only a single type of programme, ignoring the needs of qualified nurses undertaking post-registration courses to prepare them for work in particular clinical settings (e.g. cardiac care, cancer services), those pursuing studies at degree level, midwives, and nurses preparing to work in primary care. Information relating to these programmes is vital in order to ensure that the future workforce is receiving adequate preparation to meet health care needs. Nurses employed in primary care are in a key position to provide health promotion, whilst midwives help families to make choices related to reproductive health. These groups would appear to have definite and particular needs for genetics education. The content of degree programmes is also of interest, given the numbers of graduates who seek employment in primary care and other clinical areas, where they retain high levels of patient contact.

Meanwhile, the first complete chromosome (number 22) was completely mapped by scientists in the UK at the beginning of this year. In fact the Human Genome Project is probably the only research project which is expected to deliver its findings before the original target date. It is hoped that before 2005 the nursing profession will have seriously considered the impact of this purely scientific research on the lives of ordinary people and will have made provision for future patient needs.

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