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Ethical and legal issues and the "new genetics"

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ABSTRACT

- Although uniquely personal, the information from gene analyses impacts on parents, siblings, children and even entire ethnic groups.
- Doctors need to carefully balance the right of a patient to privacy against the wider family and society interests, consistent with ethical standards and their legal obligations.
- Doctors also need to be in a position to advise their patients of potential risks that may result from obtaining predictive genetic information, such as discrimination by third parties.
- While these issues are not new, they occur with new sharpness in the "new genetics", where clinicians have to be familiar not only with clinical significance, but also the ethical and legal implications of genetic analyses and information.

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IN RECENT YEARS there has been an explosion of knowledge in the science of genetics but often less general awareness of the ethical and legal implications of genetic advances. Fueled by sensationalist media reporting, developments are often exaggerated and create unrealistic expectations for the "new genetics".[1] Medicine has a great capacity to test and screen for gene mutations, but currently little ability to cure the clinical consequences of these mutations. Because of the newness of this information, and the deterministic way in which many interpret the data, there is a risk that predictive genetic information will be misunderstood and too much weight will be placed on it.[1] Genetic determinism is particularly unwelcome, because most common diseases involve the interaction of predisposing genes with a facilitative environment — the value of genetic knowledge is usually to allow accurate environmental or pharmacological intervention.

Concerns have been raised about the misuse of genetic information, particularly with computerisation and linkage of health records.[2] These concerns may reduce the willingness of individuals to undergo genetic testing, even when the tests are clearly beneficial. In a recent initiative in Victoria, only a small proportion of people offered free gene screening for haemochromatosis accepted the test. One can only speculate as to the reasons for this low uptake, but fears about confidentiality and potential misuse of the information may have played some part.[3,4] This was in spite of the fact that the clinical consequences of haemochromatosis can be completely averted if appropriate action is taken. Furthermore, a unique agreement had been reached with the Investment Financial Services Association (IFSA), the peak body of the life insurance industry in Australia, that people who tested positive for haemochromatosis and agreed to take preventive measures (regular blood donation) would not be refused life insurance or have their premiums loaded because of their genetic status.[5]

In many cases, ethical concerns about genetics simply underscore existing concerns about marginalisation, stigmatisation and discrimination of disadvantaged groups. Although these concerns may be valid, they are not new or unique to genetics. However, the sheer scope of genetics and the complex nature of genetic information, extending beyond individuals, mean that these concerns are now more pressing. There are also some significant new ethical and legal issues emerging from the application of the human genome project to medicine, particularly with regard to predictive information about common diseases and for traits (such as criminality or ability) that are not diseases at all.

Our aim in this article is to give a broad overview of the main ethical and legal challenges presented by the new genetics and their implications for the medical profession. In many instances, these issues have not been resolved and the full debate remains to be had. Typically, there are no easy answers to the dilemmas raised, but awareness of what the key issues are and sketching of directions will help healthcare professionals understand and participate in these developments.

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Ethical issues

Some basic principles to keep in mind when considering ethics are presented in Box 1. In Australia, ethics usually revolves around informed choices by individuals. However, the pervasive and predictive nature of genetic information means that every clinician has to be familiar not only with its clinical significance, but also the ethical implications.[7] As well as thinking of patients as individuals, doctors must think of families, because gene analyses affect parents, siblings, children, the unborn, and sometimes entire ethnic groups. Doctors must be aware that this responsibility to family may conflict with the individual's right to privacy.

There are some fairly straightforward ethical issues that arise with respect to genetics:

- Is the application of genetics lawful? If it is not, it cannot be offered, as is the case for using DNA to select the sex of an embryo in Victoria, except to avoid transmission of a genetic disorder.[8] Even if it is lawful, do you, as a doctor

1: Ethical principles

There are many religions and belief systems, and it is important that healthcare ethics should be able to inform decisions of the whole community. Four principles that could underpin an ethical approach to healthcare issues are:[6]

1. Respect autonomy: educate, communicate, consult, respect and empower. (Autonomy is both very important and controversial in genetics. Conflict between the rights of the individual, the family and the community arise more often for genetic issues than for most medical procedures.)

2. Beneficence: provide net benefits, but ensure these are realistic.

3. Non-maleficence: do no avoidable harm, to individuals or groups.

4. Promote justice: fair distribution of resources, respect for rights and respect for morally acceptable laws. (One problem of genetics is that it "is not fair". We are not "created equal", because our genetics differs, and with it our health risks. However, the doctor has to try to create a level playing field, in the interests of justice.)

When applied in the context of genetics, beneficence and non-maleficence sit easily, but, for the reasons noted above, autonomy and justice are problematic.

have an ethical objection to this test or procedure, such that you would have to advise the patient to see another doctor?

- Is it safe? If it can cause harm, is the likely harm balanced by the likely benefit?
- Is it helpful in dealing with the problems you perceive as relevant to this patient, this family? Is it helpful in dealing with the problems as perceived by them?
- Is it evidence-based, or still a research procedure?
- Is it cost-effective?

These types of issues arise with every medical procedure, but arise with an unprecedented intensity for genetics.

The availability of genetic testing offers the "capacity to know" about one's genetic destiny with greater certainty than revealed by family history alone, but knowledge will not always be welcome and individuals are generally at liberty to decide whether they want this information. The ethics of testing are different if action can be taken to prevent or treat a disease, as for haemochromatosis, as compared with conditions for which no treatments are presently available, such as Huntington disease. If prevention or early treatment is available, it is unethical not to offer testing. However, some people want and use knowledge even if there is no treatment, for example, for reproductive choice. Particular care needs to be taken with the predictive genetic testing of children: this is widely regarded as inappropriate unless preventive strategies are available.[9,10]

The capacity to use test data for reproductive choice brings a range of ethical dilemmas. Genetic testing of fetuses with a view to termination of pregnancy is met with alarm by some disability advocates, who are concerned about approaches that treat disability as a "problem" that should be prevented using genetic means, rather than dealing with the issue of non-discrimination of people with disabilities.[11] Concerns have been expressed that the range of conditions that may be tested for will extend to good looks and abilities, leading to fears of "designer babies" and the spectre of eugenics.

Society will set the limits within which choices will be made. Although there is some disquiet about genetic interventions, as a society we need to ensure that we have a balanced ethical debate on issues of concern and that we distil the real ethical issues. The challenge ahead is to ensure that the newness of genetics does not unreasonably impede its implementation. There may be a natural resistance to the expansion of genetic science and technology, particularly where it extends beyond the therapeutic model (eg, to enhance appearance or intelligence), but it is important that we not limit the available options unless there is sound justification for doing so. This also underscores the importance of offering appropriate genetic counselling, particularly for the more complex situations in predictive or prenatal testing, so that individuals can make informed choices in both an individual and a social context.

The legal framework

The law needs to set limits within which scientific development and clinical practice can operate. Although the scope and extent of protection that should be provided by the law is a matter for debate, there is consensus that the law should protect individuals from avoidable harm.

The metaphor of the law "limping in the rear" of the march of science[12] is often invoked, and nowhere more so than in the context of the new genetics. There are few laws in Australia regulating the collection and use of personal genetic information, and none that do so explicitly for genetic information. The present legal framework in this area consists primarily of anti-discrimination and privacy legislation. We end up with a complex legislative patchwork, which has some influence on the permissible collection and use of personal human genetic information, but does not effectively "regulate" it. For this reason, there has been much agitation for reform to respond to the new challenges created by the increase in the range of available genetic testing.

The collection and use of human genetic information, and the measures that may be necessary to protect the individual are under intense scrutiny by the Australian Law Reform Commission (ALRC) and the Australian Health Ethics Committee (AHEC).[13] A discussion paper released in August 2002 canvasses a range of issues and makes numerous proposals for regulation.[14] These are being finalised for public discussion and possible enactment.

Individual and family rights

The availability of the new genetics has implications with regard to doctors' legal duty of care to their patients. Doctors have a responsibility to keep up to date with the new genetics so that they can give advice on what tests are available. As with any medical procedure, the law protects the autonomy of competent individuals to decide whether to undergo genetic testing and to accept medical treatment or advice about lifestyle changes arising from such testing. However, there are tensions between the rights of individuals and the rights of the family, for whom this information

2: Genetic discrimination

Genetic discrimination can be defined as different treatment of an individual by a third party such as an insurer or employer on the basis of genetic factors — real, inferred or wrongly imputed. Discrimination can be positive or negative: the concerns relate to *unfavourable* discrimination, involving decisions adverse to the interests of the individuals involved. Unfavourable discrimination can be justifiable and lawful: anti-discrimination legislation, which provides protection for some forms of unfair discrimination, contains exemptions from discrimination by insurers and employers in some circumstances.

A team of researchers, funded by the Australian Research Council, is conducting a major empirical study into the nature and extent of genetic discrimination in Australia and its social and legal implications.[21] The study seeks to gain the experience and perspective of all key stakeholders: "consumers" (those considered to be at risk because of a genetic test result or their family history); third parties such as insurers and employers (the groups against which allegations of genetic discrimination have most frequently been made); and the various organisations within the legal system through which complaints of alleged genetic discrimination may be pursued.

may have relevance to health. Privacy regulation in Australia comprises a combination of common law and legislation.[15] At present, no special status is afforded to genetic information. Strictly, even taking a person's family history involves a potential breach of the privacy of other family members. This difficulty has now been addressed through the Federal Privacy Commissioner making a Public Interest Determination to cover family medical histories.[16]

Problems can also arise with the disclosure of an individual's genetic information to other family members. At present, standard rules regarding the disclosure of health information apply, limiting disclosure to circumstances where there is a threat of serious and imminent harm to others or a serious public health risk.[17] However, the familial nature of genetic information demands some modification of the usual principles of privacy and non-disclosure, in both directions. The information should be able to be shared with family members whose health may benefit from access to this information by alerting them to the risk of genetic disease and enabling them to institute preventive or therapeutic strategies,[18] but be protected more carefully from outsiders. One aspect of the ALRC/AHEC proposals that is likely to be of practical relevance to doctors is the proposal to expand the circumstances in which genetic information may be released to other family members.[14]

Third-party access to genetic information

There are also vexing questions about whether third parties should be entitled to access personal genetic information. When applying for insurance, individuals are required to disclose family history and the results of any genetic tests,[19] and insurers are entitled to take this information into account for the purposes of underwriting for life insurance and related products. Insurers are exempt from disability discrimination,[20] but must be able to justify the way in which they use the genetic information with regard to actuarial, statistical or other data. There are concerns about the adequacy of available data for underwriting purposes and the potential for unfair genetic discrimination (Box 2).[22] In several jurisdictions, including the United Kingdom, moratoriums have been introduced on the use of genetic test information by insurers, or such use has been prohibited by legislation. Current proposals for reform put forward by the ALRC/AHEC retain the insurers' entitlement to use genetic test information for risk assessment, but seek to regulate more stringently what genetic tests can be used by devolving this responsibility to the proposed Human Genetics Commission of Australia.[14]

In the sphere of employment, the challenge is to ensure that legitimate uses of genetic test information are permitted, such as offering screening for susceptibility to workplace hazards that cannot otherwise be avoided, but to protect employees and job seekers from unfair discrimination motivated by employers' expediency and profit. The proposals advanced by the ALRC/AHEC seek to strike a proper balance to allow uses of genetic testing which are consistent with occupational health and safety interests, but prohibit other uses.[14]

Genetic samples

Fundamental questions are also being raised about the status of genetic samples collected for pathology examination, such as blood or other sources of DNA, including pathological tissue blocks and human tissue on microscope slides. At present, these are generally regarded as the property of the hospital, over which the donor may have no legally enforceable rights. Although such samples have no clear legal status as property, opinions are divided over whether it is appropriate to create legally enforceable rights, especially if the sample proves to have a commercial value.[23,24] The line between research and clinical care is ethically blurred when a sample is studied by a specialist or a pathologist, and becomes even more confusing as we move towards an increasingly commercialised environment in which the potential for profit from genetic knowledge is real and the clamour for patents resonates.[25]

Reproductive technologies

When it comes to regulation of artificial reproductive technology, the situation is even more confused. In Victoria, South Australia and Western Australia, there is legislation regulating this area.[26-28] These Acts, to varying degrees, restrict the circumstances in which genetic testing can be undertaken. The other States and Territories have no legislation, and this lack of uniformity invites "doctor shopping".[29]

There is great concern that genetics will be used for "designer babies", but no one knows whether this will be possible, economically viable, or wanted by anyone. Couples are now being allowed to choose pregnancies that will provide an infant with a particular genetic make-up in the context of a sibling with a very serious illness.[30] The baby can then be a donor of cord blood stem cells to the seriously

ill sibling. Although this is occasionally described as "designer babies", it is clearly far from what concerns the public. It is important for doctors to be aware of and emphasise the difference between the use of clinical interventions to save the lives of children with serious diseases, as compared with the use of procedures for trivial purposes such as choosing hair or eye colour. The former is generally thought to be ethical, the latter unethical. There is concern that over-the-counter DNA tests will soon be available, but knowledge has for generations been regarded as a positive, not a negative, commodity. Doctors may be the gatekeepers of the new genetic knowledge, but they will not be its owners.

Maintaining flexibility in regulation

The draft sequence of the human genome is now on the Web, and advances in our understanding of the relationships between genes and environment and disease occur almost daily. Challenging issues lie on the horizon in terms of defining the role of doctors with respect to their patients and the extent of their duty of care, as we learn more of the relationship between genes, the environment and complex diseases and behaviours. Because of community concerns, there will be pressure for laws to regulate the application of genetics in medicine. However, knowledge of genetics and the methods of applying it to people are changing rapidly, so there is an overwhelming need for flexibility in the development of solutions. This may encourage the use of regulations and other "soft" laws, such as guidelines and codes of practice, in preference to statute law, as the former are easier to adapt to new situations.

Conclusion

The new genetics has enormous potential to confer clinical benefits. The challenge is to harness these benefits and to minimise the risk of harm. Fortunately, most doctors, patients and families want to make sensible choices. The ethical interpretations we offer, and the legal framework that is used to interpret these ethical principles, must ensure that application of the new genetics is not unreasonably restricted as it develops. The better informed doctors are of the ethical and legal issues arising from the new genetics, the better equipped they will be to give appropriate information to patients and the community.

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