CHAPTER 4

THE HUMAN GENOME

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ENCOUNTER WITH THE GENOME

My qualifications to address the ethical problems of the human genome arise out of my work on the International Bioethics Committee of the United Nations Educational, Scientific and Cultural Organisation (UNESCO) in Paris and the Ethics Committee of the Human Genome Organisation (HUGO) in London. Each of these bodies is considering a number of the ethical, social and legal questions which arise out of genomic research and the genetic engineering to which it will give rise. The UNESCO Committee in 1996 adopted a draft of a *Universal Declaration on the Human Genome and Human Rights.* This was, with some modifications, approved by the General Conference of UNESCO in Paris in November 1997. We stand at the brink of remarkable scientific and technological developments concerning our genes. Belatedly governments, and the international community, are beginning to respond. But what path should they take?

DIFFICULTIES IN THE PATH

In the business of the genome, we are talking about nothing less than the future of the human species. It is therefore a topic appropriate to international consideration and, eventually, international law. However, the difficulties of securing a consensus about such a topic are all too obvious. They include the different religious, cultural and legal traditions which must be brought into harmony and the different economic interests of different countries involved in the development of therapies. There are disparities in attitudes to intellectual property protection and in sheer investment and the potential to make profits arising from these scientific developments. The inclination of local law makers is to put such matters in the "too hard" drawer, preferring instead to address more manageable local controversies with greater political attraction.

There is also a feeling of resignation in some quarters arising out of the belief that the tide of science and technology cannot be held back by any law and that any legal attempt to prevent scientists from experimenting is bound to fail and so should not be attempted. Finally, there is a feeling on the part of some that such scientific progress is bound, in the long run, to be for the betterment of humanity and is, in any case, a product of the inherent skills and abilities of the human species and thus an extension of human beings, not something alien to them.

What are we talking about? The Human Genome Project is the largest cooperative scientific activity in history. It is larger by far than the Manhattan Project which resulted in the development of the atomic bomb. Yet its implications are in some ways similar. It is important that the Project should advance with a full understanding of the ethical, social and legal consequences that came in its train. This is recognised by HUGO itself. It gives an impetus to the work of the HUGO Ethics Committee and also to that of the International Bioethics Committee of UNESCO. Yet the reality is that the funds devoted to the ethical, social and legal consequences of genomic research are but a tiny fraction of those devoted to the scientific research itself.

LEGAL & ETHICAL IMPLICATIONS

There are many practical implications which the unlocking of the mysteries of the genome will have for humanity. They include implications for medical therapies, criminal law, privacy and confidentiality, third party interests, intellectual property and human rights.

Medical Therapies: Scientists are now discovering the genes which "trigger" various genetic diseases which, in turn, constitute a large part of the inherited causes of the suffering of humanity. I recently attended an international conference on Huntington's Disease, held in Sydney. The genes which

express that serious affliction have been identified on the genome. Its discovery permits the conduct of extremely accurate tests which can now identify those people who carry and may transmit this genetic disorder. That knowledge would, theoretically, in combination with amniocentesis and abortion, permit the future elimination of carriers of Huntingtons. But is this desirable? Can it be distinguished from the abortion of a foetus with Down Syndrome? Where does this process of medical elimination of "defective" genes begin and end? Is there a less life-destructive means of using the genetic information to delay the onset or diminish the symptoms of Huntington's disease whilst respecting the life of a person born with those genes or others like it?

(2) *Criminal Law:* For the lawyer, the discovery of genetic causes of disorders and of some antisocial conduct may have implications for the future. The criminal law is built upon the hypothesis of free will. For the crime to be established it is normally necessary to prove both the act of the accused and the will (*mens rea*) occasioning that act. But what are the implications for the law of discovering that, in some cases at least, for some people, the act is practically no more than the product of a genetic characteristic? Can we persist with the unquestioned hypothesis of free will in the face of scientific knowledge which casts doubt upon it?

(3) *Privacy and Confidentiality:* The basic rule of the healthcare professions has long been respect for the confidences of the patient. This rule goes back to the Hippocratic Oath. It existed in ancient civilisations. But when a disorder is of a genetic characteristic, is the "patient" the individual or the entire family? Does a family in such circumstances have a right to override the wishes of the patient and to secure data about the patient's genes relevant to genetic features important for them all? Does a patient have a right *not* to know the determinants of his or her future medical conditions?

(4) *Third Party Interests:* This last question leads to the rights of third parties. Should an employer have a right to require an employee to submit to genetic testing to show, with greater perfection, the likely future health status of the employee? Should an insurer be entitled to secure a detailed genetic profile of the insured? Until now, insurance has generally involved the sharing, within the community, of the risks attached to medical conditions that are largely unpredictable. If such conditions can be predicted with perfect or near perfect accuracy, would that not shift the scales unfairly to the advantage of insurers? Yet, where insurers can require those seeking insurance to submit to old-fashioned medical tests, is it sensible, in the future, to close off knowledge of the best medical information that may be available by genetic tests?

(5) *Intellectual Property:* One of the key issues of genetic research concerns the desirability of permitting the patenting of human genes or their sequences as the basis for therapeutic applications. Of course, in every country, the patentability of such matter depends upon the terms of the local law on intellectual property protection (patents, copyright etc). That law is itself normally the product of national legislation and is generally influenced by international law. At conferences on the genome, strong views are frequently expressed by participants from developing countries and elsewhere about this topic. They urge that the human genome is the common heritage of humanity. That it belongs to the human species as a whole - some say to God - and not to private corporations engaged in research, however potentially beneficial such research may prove to be. They point to the fact that the great scientists Watson and Crick, who first described DNA, and began mankind's journey a full understanding of to the genome, never attempted to secure commercial advantage for themselves from their discoveries. I will return to this topic.

(6) *Human Rights:* An important element in the UNESCO Declaration is the attempt to reconcile the development of genetic technology and research on the human genome with fundamental human rights and human dignity inhering in every individual. The UNESCO *Declaration* states in Article 6*:*

> "No one shall be subjected to discrimination based on genetic characteristics that is intended to infringe

or has the effect of infringing human rights, fundamental freedoms and human dignity."

The eugenics movement earlier in this century was a doubtless well-intentioned effort to eliminate, in effect, genetic characteristics deemed undesirable to society. For the most part, the movement was targeted at so-called "mental defectives" but it affected (as we now know) large numbers of persons who manifested quite modest mental impairment or none at all. Unsurprisingly perhaps the eugenics movement had strong supporters in the Nazi effort to "cleanse" the German population of so-called undesirables. That effort notoriously attacked people for their genetic identity: specifically their Jewish or gipsy ethnicity. But it also imposed its uniform conception of the human species upon others who presented genetic or other conditions deemed undesirable to the Nazis: homosexuals, the physically disabled and the mentally impaired. The terrible experience of the Holocaust stands as a dire warning to humanity of what can happen when people with a stereotyped view of human existence gain totalitarian political power. We should not consider that this is a problem of ancient history. It endures into our own time. We have recently seen it in one form in the "ethnic cleansing" in Serbia, Bosnia and Rwanda. At the outset of the genomic revolution in medicine, therefore, it is timely to insist that the developments should occur in a context of respect for fundamental human rights and human dignity. I would expect that the Christian Churches would lend their support throughout the world to the effort of the United Nations to insist upon such preconditions.

PATENTING GENES

One advantage of my appointments to the UNESCO and HUGO Committees is that I have the opportunity and obligation to read scientific literature. Not for me is it a single diet of the *Commonwealth Law Reports.* In a recent issue of the journal *Science*[1], the heat of the debate concerning intellectual property law protection of genes and gene sequences is illustrated. The journal records that the National Academy of Sciences in the United States on 14 June 1997 caused its President, Dr Bruce Alberts, to write to the Director of the United States Patent and Trademarks Office about this problem. Particular concern was expressed by the Academy about the willingness of the Office to grant patents on mere fragments of human genes - particularly those known as Expressed Sequence Tags (ESTs). These can be used to identify the presence of full length genes. ESTs are relatively easy to capture. But they reveal little about the biology which they control. Dr Alberts fears that patenting ESTs - a few have been patented so far and thousands of applications are pending - could create a tangled maze of property rights which would actually impede research:

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1. "Academy Joins Debate Over DNA Patents", *Science,* vol 277, 4 July 1997 at p 41.

"It would be sad indeed if patent policies diminished the pace of discoveries or wealth of practical applications."

The National Academy of Sciences appealed to the United States Patent Office to consider granting DNA patents only where "real world" applications are described in the patent application or detailed information about the gene is already known or supplied by the applicant.

The appeal by Dr Alberts parallels one made in March 1997 by the Director of the National Institutes of Health in the United States, Dr Harold Varmus. He wrote to the United States Patent Office after an official of that office had given a speech favouring patents on ESTs as diagnostic or research probes[2]. His concern was that such patent policies might block research and development on more important discoveries such as complete genes and thus stifle beneficial gene-based therapies.

The response of the United States Patent Office to pressure of this kind is predictable. It simply says that it will apply the law. If the Congress of the United States wishes to restrict or forbid the patenting of life forms, that is for the Congress to say. There are, of course, many in the United States and elsewhere who assert that

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2 "Renewed Fight Over Gene Patent Policy", *Science,* vol 276, 11 April 1997, p 187.

patenting of genomic discoveries - and even more so gene sequences whose effects are not fully known - should be no part of intellectual property law. That this belongs to all humanity. That no individual or corporation should make a private profit from living matter. However, there are difficulties in such assertions. "Man-made" micro-organisms have been patentable in the United States at least since 1980[3]. The potential for medical therapy of developments arising out of exploration of genes is enormous. The economic profits riding on such discoveries run into billions of dollars. The investment in research said to warrant intellectual property protection is likewise extremely expensive. In these circumstances, striking the right balance between respect for the common genomic heritage of humanity, protection of people in developing and other countries so that they gain some of the benefits, and assurance of a fair economic return to scientific investors is not an easy dilemma to solve[4].

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3 *Diamond v Chakrabarthy*, 447 US 303; and 65 L Ed 2d 144; 206 USPQ 193 (SC 1980). Cf *Ex parte Latimer* 46 AG 1638, 1640 (1889); *Funk Brothers, Seed Co v Calo Inoculant Co* 333 US 127 (1948).

4 J C Venter, "The Patentability of Genetic Discoveries" in BBV Foundation (Spain) *The Human Genome Project: Legal Aspects*, vol 2, p 123; and C Byk, "Patenting Human Genes", BBV Foundation (Spain) The Human Genome Project: Legal Aspects, vol 2, p 127.

THE GENOME AND EVOLUTION

In another article in *Science*[5], the authors appeal for the development of a new view of evolution arising from the contemporary study of genes. It was in the late 1970s that scientists at Harvard University began to focus on genes in order both to understand evolution, including human evolution. But it was not until the mid-1980s that the new tools for studying developmental genes began to generate the data which could explain how, in the comparatively short period of the Earth's existence, such a remarkable myriad of living creatures found on earth - vertebrate and invertebrate - could have developed, presumably from the basic living cells present at the beginning.

Recent research has shown a number of genes to be common across a very wide range of animals. They have similar or related functions across completely disparate species. For example, a gene which stimulates the development of eyes but may cause no more than a photosensitive area in a very primitive animal, may stimulate the development of a compound eye in an insect or the highly developed eye of a mammal, such as a human being. The same or a very closely similar gene can operate in a related fashion across

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5 E Pennisi and W Roush, "Developing a New View of Evolution", *Science,* vol 277, 4 July 1977 p 34.

vast periods of evolutionary history. This discovery has obvious relevance to the patenting of human genes. If the same, or a closely similar, gene in an animal has the same, or closely related, functions across a very wide range of living species, and it is suggested that the patenting of human genes is somehow repugnant or socially undesirable, would a distinction between the human and non-human gene be a way out of this dilemma? Or would the recent discoveries indicate that if human genes are not to be patentable then no genes of living matter (human or animal) may be patented?

The exploration of the genome has also offered a possible answer to a dilemma about evolution which has puzzled biologists for some time. If evolution proceeded by a process of substitution in DNA chains of particular species, then our current knowledge of mutation rates makes it absolutely clear that four billion years (the Earth's estimated existence) is simply not long enough to arrive at the richness and variety of the species now existing. If, however, species can use a modular genetic approach to building new genes and gene functions, this would permit the rapid speeding up of the process of genetic change. A comparison has been drawn between one team of computer programmers, starting from scratch to design a whole series of programmes to carry out a variety of widely different functions, while another team starts with a number of

already developed programme parts with known functions and whose task is merely to put the modules together in new ways[6].

AN ADJUNCT TO MEDICINE OR A NEW WORLD?

The fundamental question which is presented by genomic research is this. Should genetic research be seen as no more than an adjunct of improving the health of the current human species? Should it be limited by law, and otherwise, to removing this or that disease from human beings but keeping them, in every other way, basically as they are? In short, should genomic research and genetic engineering be viewed as nothing more than an assistant to established medical science? To provide tests for genetic maladies? To provide the foundation for treatment of genetic disorders? According to moral conviction and law, to provide a basis for eliminating foetuses demonstrating grave genetic disabilities or potentialities?

These questions are hard enough. But the lessons of science and technology are that to foresee developments of the future we must engage in a leap of the imagination. It seems unlikely to me that genomic research will stop at being a mere adjunct to current

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6 Letter to the author from Dr J R Coulter, Adelaide, 29 August 1997.

medicine. If it becomes possible to alter the human species in particular potentialities, are we really talking about an aid to the human species? Or are we on the brink of considering something which may actually change the human species itself? A kind of scientific speeding up of evolution?

If you alter a large number of features of the human species - eliminating Huntington's Disease, expelling the potential to Alzheimer's, excluding Parkinson's, removing Down Syndrome - where does this path lead? Certainly it leads to the reduction of much human pain and misery which presently affect patients and their loved ones. But taken to extreme, might it not also lead to a change of what it is to be a human being? Add to the exclusion of serious genetic disorders the elimination of baldness, the removal of a potentiality to obesity, the exclusion of undue height or undue shortness and you are well on the way to redesigning the human species. The experiments of Dr I Wilmut and his colleagues[7] demonstrate that sheep embryonic eggs can reproduce the nuclei of differentiated cells, enabling the cells to develop into any type. This showed that it may now be possible to envisage cloning of adult humans in a completely asexual fashion. If it can be done with

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7 "Clone mammals ... clone man?", *Nature,* vol 380, 13 March 1997 p 119.

sheep, given time, it can undoubtedly be done with humans. And what or who will stop it?

FORBIDDEN TERRITORY OR THE NEXT STEP FOR HUMANITY?

With catchy phrases, writers in the scientific literature talk of our era as one where human beings will pass from Genesis to genetics[8]. Obviously, the developments of scientific knowledge have large implications for religious faiths which accept as doctrine the teachings of a Holy Book. As scientists and technologists report their discoveries, it becomes necessary for religious teachers and theologians to explain and justify the revealed scientific truths, reconciling them with the previous understanding of Scripture and the teachings of the religious faith which were expressed in an earlier time when the scientific truth was completely unknown.

In the summer of 1993, a team of researchers at the United States National Cancer Institute announced that they had evidence linking male homosexuality to a gene in the region of the X chromosome[9]. It looks increasingly likely that sexual orientation is, in part at least, a genetic phenomenon and thus beyond the "wicked"

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8 Ted Peters, "From Genesis to Genetics", *New Scientist,* 15 March 1997, p 42.

9 *Ibid,* p 42.

choosing of a "wilful" individual. If this is the case, then prima facie to discriminate upon that basis would be as morally impermissible, and even repugnant, as to discriminate upon any other genetic basis such as gender, race, skin colour or a pre-programmed disease or characteristic over which the individual has no control.

It might be said that, exceptionally, sexuality is a genetic condition that the individual should just try to struggle against and to deny. It might even be said that this is one genetic condition that should be eliminated in whatever way possible. The Chief Rabbi of the Commonwealth of Nations, controversially, suggested that this should be done to get rid of homosexuals, thereby provoking cries of outrage from Holocaust survivors and other Jewish intellectuals. But if sexual orientation is, indeed, part of the genome of our species, a serious moral question is plainly presented. By what right can we say that it is not part of Nature's - or God's - great purpose? That purpose, as the Church has taught, is not always clear to us, mere mortals. We see through a glass darkly. But will it be the Human Genome Project as it develops that helps us to see[10]:

> "Face to face: now I know in part, but then shall I know even as also I am known".

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10 1 Corinthians 13 xii.

An even more fundamental question than this is presented. For all those (including in the Church and the United Nations and its agencies) who urge that we should keep genetic alteration as an adjunct of human existence *as it now is*, others dispute. For the disputants, genetic discoveries arise out of the intelligence of human beings. That intelligence is given by Nature - or God - to discover reality as it exists. The genome and DNA existed for millennia before we discovered them, in our generation, through the intelligence of Watson and Crick.

If the genome is discovered, and is there, that discovery is, arguably, the outgrowth of a human development which was ordained for us in this era. That development will itself not stand still. It will take us further down a path that might indeed be called "evolutionary", which is itself the product of our human intelligence. It may be a path that involves leaps of evolutionary history - a type of fast forward of the kind that seems somehow to have occurred without human intervention in the past. It may even be a path that involves a reconsideration of what it is to be a human being and what, if any, are those characteristics of the human species that are to be regarded by scientists as absolutely forbidden territory. In any case, no law can stop science and technology completely. There will always be a small corner of the world that will give sanctuary to the free spirit of the enquiring scientist and the technologist at work in the laboratory. Especially will this be so if profits dangle tantalisingly at the end of the endeavour.

If the Christian Churches or other religious teachers take a different view, they must explain that view and argue for it. It seems unlikely that dogmatic assertion or even scriptural texts will win the argument today. Reason and a return to fundamental wisdom may help in the persuasion as may an appeal to universal notions about the things that all human beings share in common. But if we do not join this debate it will surely go by default.

INFORMED DECISIONS

This is why I consider that the work of the UNESCO International Bioethics Committee and the Ethics Committee of HUGO as amongst the most important that I have been involved in. For a lawyer, like a theologian, it is somewhat intimidating to stand staring at the brink of a new era of genetics. The scientist and the technologist rush ahead. The lawyer, the ethicist and the theologian amble slowly along, their heads full of puzzlement at the problems which seem so insoluble. Yet to do nothing is to make a decision. It is to permit science and technology to take our species where they will. We know enough now to realise that there are quandaries here for human beings to answer. The ultimate question is whether we will have the will and the means and the wisdom to afford the answers.