GENETICS IN THE COURTROOM

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I INTRODUCTION

The recent publication of the Australian Law Reform Commission's ('ALRC') very important report *Essentially Yours: The Protection of Human Genetic Information in Australia*[1] and the Issues Paper for the inquiry into gene patenting and human health *Intellectual Property Rights over Genetic Materials and Genetic and Related Technologies*[2] prompts this note about developments in the Federal Court of Australia which led, in September this year, to a notable conference on 'Genetics in the Courtroom' in Sydney.

The conference was organised by the Federal Court under the auspices of the Einstein Institute for Science, Health and the Courts ('EINSHAC'), a voluntary educational and research organisation with close connections to the judicial branch of government in the United States. The aim of EINSHAC is to make science accessible to judges and courts.[3]

The Federal Court's involvement with EINSHAC began in 2001 when two members of the Court along with Justice Kirby of the High Court[4] and a group of distinguished Australian scientists and ethicists took part in the 'Courts First International Working Conversation on Enviro/Genetics Disputes and Issues' held in Hawaii. This was followed by a 'Second International Working Conversation on Biotechnology Issues' in Ottawa in 2002 at which there was a similar Australian representation. At both of these working conversations, judges and leaders in the fields of science and medicine worked through a range of topical issues concerning genetics and biotechnology and their intersection with ethics and the law.

The subsequent Australian 'Genetics in the Courtroom' conference was attended by nearly half the judges of the Federal Court and by judges from the Supreme Courts of New South Wales, Victoria and Queensland and the Family

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1 Australian Law Reform Commission and Australian Health Ethics Committee, *Essentially Yours: The Protection of Human Genetic Information in Australia*, Report No 96 (2003) ('ALRC Final Report').

2 ALRC, *Intellectual Property Rights over Genetic Materials and Genetic and Related Technologies*, Issues Paper No 27 (2003).

3 For more information about EINSHAC see <http://www.einshac.org/> at 20 November 2003.

4 Justice Michael Kirby headed the Australian delegation and delivered one of the keynote addresses.

Court. New Zealand was represented by the Chief Justice, Dame Sian Elias, and the United States by EINSHAC faculty member Judge Joan Zeldon of the Superior Court of the District of Columbia. The scientific faculty included Dr Kerry Breen, Chairman of the Australian Health Ethics Committee, Dr James Evans, Director of Cancer Genetic Services at the University of North Carolina, Professor John Mattick, Director of the Institute of Molecular Bioscience at the University of Queensland, Professor Alan Pettigrew, CEO of the National Health and Medical Research Council, Professor Colin Thomson, Health Ethics Consultant to the National Health and Medical Research Council, Professor Ronald Trent, head of the Department of Molecular and Clinical Genetics at the Royal Prince Alfred Hospital, and some 12 other eminent researchers in the fields of biotechnology, genetics and ethics.[5]

The theme of the conference was the identification of the tools that the courts will need to adjudicate disputes arising from ever-increasing knowledge in the areas of biotechnology and genetics. Judges listened to presentations about the nature of genes; genetic screening; molecular medicine and gene therapy; forensics; the Human Genome Project;[6] heritable conditions, including the heritability of personality traits;[7] predictive medicine; the ALRC Final Report; the use of DNA evidence in court; and developments in the field of embryonic and adult stem cell research.

The judges also took part in adjudication clinics in which scenarios were presented in a way that provoked exploration by small groups of judges, scientists and ethicists of the difficult legal, ethical and scientific issues that the courts may be confronted with in cases involving genetics and biotechnology. The process was dynamic and, as an educational method, effective and very stimulating. The adjudication clinics were followed by a plenary session, at which each group delivered a report of its deliberations. The reports were then debated. The adjudication clinic process is a method of judicial education in science developed very effectively by EINSHAC and is a central element of that organisation's successful work with judges in the United States.

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5 These scientists were from a diverse range of backgrounds and institutions. They included experts in the field of genetics, molecular biology, molecular genetics, molecular medicine, physiology, gene therapy, medical informatics, biotechnology, immunology, microbiology, forensics, applied ethics and genetic counselling. The institutions with which the scientists were associated included: the Department of Immunology and the Department of Molecular and Clinical Genetics, Royal Prince Alfred Hospital; Gene Therapy, Westmead Children's Hospital; Centre for Genetics Education, Royal North Shore Hospital; Kolling Institute of Medical Research; the Victorian Bioinformatics Consortium; Department of Microbiology, Monash University; Mater Research Institute; Institute of Clinical Pathology and Medical Research; Australian Federal Police; the Garvan Institute of Medical Research; and the International Vaccine Institute.

6 In this presentation we learnt that 99.9 per cent of each individual's DNA is identical to that of all other humans and that human DNA is 98 per cent identical to that of the chimpanzee: see John S Mattick, 'The Human Genome and the Future of Medicine' (2003) 179 *Medical Journal of Australia* 212, 212.

7 Dr Jim Evans, in his presentation 'Predictive Medicine and Behavioural Genetics' at the 'Genetics in the Courtroom' conference, spoke about 'twin' studies which suggest that personality traits are to some extent heritable. Such traits include general intelligence (50 per cent); extraversion (50 per cent); adolescent vocational interests (40 per cent); 'novelty seeking' (40 per cent); juvenile aggression (25–60 per cent); adult criminal behaviour (50–67 per cent); and religiosity (40 per cent). This of course is not to exclude other influences, including environment.

II ESSENTIALLY YOURS OR ESSENTIALLY OURS?

The recent ALRC Final Report titled *Essentially Yours: The Protection of Human Genetic Information in Australia* resulted from a two year inquiry into the laws, institutions and policies required to guide Australia through the genetic revolution. While each individual's genetic makeup is clearly personal, genetic information also has deep relevance for the members of the family of the individual from whom the genetic information originates. This is because genetic information is in large part shared between family members. Genetic information could also be described, therefore, as 'essentially ours'. In the context of genetic testing, this can lead to conflict between an individual's right to privacy, the right to information about one's own genetic status, the right *not* to know, and the confidentiality of doctor-patient relationships. Some of the scenarios presented at the conference raised the complex issue of ownership of genetic information in the context of genetic testing.[8]

One scenario presented the case of a woman who had a family history of breast cancer. She wanted to have a genetic test for the BRCA2 gene, which has been shown to indicate a predisposition to breast cancer, so that she could decide whether or not to have a prophylactic bilateral mastectomy. Genetic diagnosis of a predisposition to breast cancer would not have been reliable if based on a sample of her genetic information alone. It was necessary to test a close genetic relative as well as the patient as a form of crosscheck.[9] The woman's mother and grandmother had both died, and the closest relevant relative was her uncle. Initially, he refused to have the genetic test but, upon his niece's urging, agreed on the condition that neither he, his wife nor his children would be informed of the result. The results of the genetic testing showed that the woman did not carry the BRCA2 gene but that her uncle did. This meant that his children were at a 50 per cent risk of having inherited the cancer predisposing gene. What responsibility did this knowledge place on the niece and on the doctor who undertook the genetic testing, particularly if one of the uncle's children were to contract breast cancer later in life?

A second case scenario involved a situation where a man of 58 had died suffering from Alzheimer's disease and from cancer of the colon. His doctor suspected that he carried genetic predispositions to both diseases, but did not test him because she did not believe this would have influenced his care. At autopsy, samples of blood and tissue were stored. The man's son requested that the father's stored autopsy samples be genetically tested, because if he had indeed had a genetic predisposition to these diseases, it would be relevant to assessing whether or not his children may also have that genetic predisposition. The man's

8 Note that these scenarios have been slightly modified from the actual scenarios used at the 'Genetics in the Courtroom' conference to simplify the factual matrix, but raise the same issues as the scenarios discussed at the conference.

9 See Dean Bell and Belinda Bennett, 'Genetic Secrets and the Family' (2001) 9 *Medical Law Review* 130, 130–1.

daughter sought a court order preventing genetic testing of their father's biological material, claiming that any information so obtained would lead to severe anxiety for all his children, particularly in the context of Alzheimer's disease for which there is no known treatment. She also feared that this knowledge might lead to medical and life insurance discrimination and was opposed to it on religious grounds. The son responded by arguing that cancer of the colon could be treated prophylactically and that there was a new research study into the early treatment of Alzheimer's using adult stem cells, in which he could participate if he knew that he had the relevant gene.

In such a situation, it was clear that even if the results of the genetic test were communicated only to the son, the daughter was likely to be able to infer the results from her brother's conduct.

In a variant of this second scenario, the family was not aware at the time of their father's death that predisposition to these diseases may be genetic. Some 15 years later, the son developed cancer of the colon. He underwent a total colectomy and a six month course of chemotherapy. Having found out that cancer of the colon was largely hereditary, he sued his father's doctor, claiming that she was in breach of her duty to warn him of the genetic risk to his health, and had therefore deprived him of the chance of monitoring, early detection and treatment.[10]

The scenarios raised such challenging questions as:

- Does an individual have the right *not* to know about his or her genetic status? Does an individual have the right to keep his or her genetic status confidential in situations where that information may be medically relevant for others? Does it make a difference if the individual for whom the information may be relevant knows of the existence of the information and wants to know it, or is unaware of it, and it is unclear whether or not they would want to know?
- Do the answers change depending upon whether or not a treatment is available for the disease for which a genetic predisposition may exist? Or do the answers depend upon whether the genetic information is specific (that is, the person has or will develop the disease), or predictive (that is, the person has an unspecified risk of developing the disease)?[11] Is the severity of the disease relevant? Is the accuracy of the genetic test relevant? How should these considerations be weighted?
- How should a doctor balance the common law duty of medical confidentiality and obligations under privacy legislation with any ethical or other legal duties the doctor may owe to family members? Does this differ depending upon whether the doctor has a doctor-patient relationship with both the person who has undergone the genetic test and the family member, or only with the person undergoing the genetic test?

10 These facts were drawn from *Safer v Pack*, 291 NJ Super 619 (NJ, 1996) in which the Superior Court of New Jersey held that doctors have a duty to warn a family member of their patient who is at risk of avoidable harm arising from a genetically transmissible condition.

11 Bell and Bennett, above n 9, 130.

Does this differ depending upon whether the doctor knows that the genetic predisposition exists, or merely suspects it?

- Should a doctor have a duty to warn, as opposed to an ability to warn? If the doctor does have a duty to warn, would it be enough for the doctor to emphasise to the person tested, or to the person opposing testing, the potential relevance of the genetic knowledge to the health of other family members? Or is the doctor under a duty to warn directly those family members?[12] If there is a duty to warn, does it extend to warning about risks to unborn relatives?
- What rights should prevail if there is a conflict between a right to know one's genetic status (for purposes such as decisions about prophylactic surgery, reproductive choices or participation in a research study) and a right *not* to know one's genetic status (because it would cause anxiety, or because there is no current cure for the disease, or because it offends religious beliefs)? How is a court to balance these interests?

One of the conclusions of the adjudication clinics was that some of these difficult questions could be avoided by ensuring that patients were given genetic counselling before or after having genetic tests, and in situations where a genetic predisposition is suspected. Genetic counselling would include making sure that each patient understood that the existence of a genetic predisposition could be relevant to the health of other members of the family and might encourage discussion of the results of the tests, or the origins of the disease, within the family. Genetic counselling is now quite widely available and an expert in this area was one of the members of the science faculty at the conference.

It is of course not surprising that many of the issues that arose for exploration at the conference have been the subject of recommendations by the ALRC in its recent report. For example, the ALRC has recommended that:

> The Commonwealth should amend the *Privacy Act 1988* (Cth) to permit a health professional to disclose genetic information about his or her patient to a genetic relative of that patient where the disclosure is necessary to lessen or prevent a serious threat to an individual's life, health or safety, even where the threat is not imminent.[13]

Another recommendation of the ALRC is that:

12 In *BT v Oei* (Unreported, Supreme Court of NSW, Bell J, 5 November 1999) and *Reisner v Regents of the University of California*, 31 Cal App 4th 1195 (Cal, 1995) it was held that a doctor owes a duty of care to his or her patient's partner in circumstances where the patient has HIV. This duty was satisfied, however, by advising the patient of the dangers of his or her partner contracting HIV and/or suggesting to the patient that the partner should have an HIV test. See also *Pate v Threlkel*, 661 So 2d 278 (Fla, 1995), a case dealing with hereditary cancer. The ALRC concluded that this was an appropriate approach and that doctors should not have a duty to directly warn family members of genetic risk, see ALRC Final Report, above n 1, ch 21, 555–6.

13 ALRC Final Report, above n 1, Recommendation 21.1. See also Recommendation 21.2, which recommends the development of guidelines for health professionals dealing with the disclosure of genetic information to the genetic relatives of their patients.

> The Commonwealth should amend the *Privacy Act* [*1988* (Cth)] to provide that an individual has the right to access genetic information about first-degree genetic relatives where such access is necessary to lessen or prevent a serious threat to an individual's life, health or safety, even where the threat is not imminent …[14]

The recommendations address barriers to the disclosure of potentially life-saving, or at least welfare-enhancing, genetic information to family members under privacy legislation. They do not, however, modify the common law duty of confidentiality, nor comprehensively define the scope of the right to know. The scope of the duty of confidentiality and exceptions to it in the context of genetic information and the precise boundaries of any new laws enacted pursuant to these recommendations are likely to become increasingly important and will fall to be considered and interpreted by the courts, as matters of this nature come before them.

III UNDERSTANDING THE SCIENCE – A CHALLENGE FOR THE COURTS

Medical genetic testing is of course only one of the many areas in which developments in genetics and biotechnology will be centrally important in cases that come before the courts. These issues already arise in the fields of patents, crime, employment, discrimination, insurance, health, safety and the law of the environment.[15] In all these areas, the rapid advances in genetics and biotechnology will present new challenges to the courts in their understanding of complex and developing science.

The recent 'Genetics in the Courtroom' conference and the methods for judicial education in science developed by EINSHAC show that valuable judicial studies in these complex areas are possible. Although the work is resource intensive and requires the generous cooperation of eminent people in several fields, the dynamic processes that can be put into place when scientists, medical specialists, ethicists and judges meet to explore such issues in a structured way are highly productive.

The approach adopted by EINSHAC involves discursive engagement, as well as very skilled presentations by the experts. It thus allows judges to remove themselves for a while from the role of observing and understanding oral arguments presented or led by lawyers, to a role perhaps more akin to that of a barrister preparing a complex case. Indeed, the EINSHAC process suggests that it may be possible, and necessary, for courts to find ways of enhancing the fact finding role of the judge so as to allow for a deeper immersion in the scientific, social and ethical dimensions of matters that come before them.

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14 Ibid, Recommendation 21.3. Note that it is also recommended that certain procedural safeguards be put in place to ensure that such access does not unreasonably impact upon the privacy of individuals.

15 See *Gene Technology Act 2000* (Cth) and the draft *Environment Protection and Biodiversity Conservation Amendment Regulations 2001* (Cth), which if enacted will regulate bioprospecting in Commonwealth areas.

Courts need, therefore, to be imaginative in developing rules and procedures that will best assist them to understand new issues whilst, at the same time, maintaining the core judicial value of fairness.

The rules of Australian courts already provide for some such useful procedures. The following examples are from the Federal Court Rules:

- Under Order 34, the Court can appoint an expert to inquire into and report upon the questions raised in a matter. The report is provided to the judge and all the parties. The expert can then be cross-examined on the report by the parties. In such cases, the parties can adduce their own expert evidence only with the leave of the Court.[16]
- Order 34A provides for a range of means for taking expert evidence. For example, the Court may direct that all expert witnesses confer prior to the trial and produce a document identifying the matters and issues on which they agree and those on which they disagree. Perhaps most innovatively, the Court can direct experts to provide their evidence in what has become known as 'the hot tub'. This involves the experts being sworn in to give their evidence at the same time, usually after all the lay evidence has been given. One expert will start by giving a brief outline of his or her opinions and answering questions from the other expert(s). The other expert(s) will do the same. Each expert can then give a brief summary. At the conclusion of this process, counsel can cross-examine and re-examine.[17] This method of receiving expert evidence has several advantages. One advantage is that the expert evidence is received after the issues in dispute have been clarified by lay evidence. Another is that the judge has access to opposing expert evidence at the same time, which facilitates comparison between the evidence. Putting experts together in a more discursive context can also help to reduce partisanship in the giving of expert evidence.[18] Finally, experience suggests that this procedure can substantially shorten the time required for the taking of expert evidence.[19]
- Under Order 34B, the Court can appoint an expert assistant with the consent of the parties. The role of the expert assistant is different from that of a court appointed expert. The role is to assist the Court on any issue of scientific fact or opinion identified by the Court or judge – to act, as it were, as a scientific tutor. The expert assistant must provide a report to the judge on the issues identified and a copy of the report to each party.

Perhaps even more useful than the expert assistant provided for in Order 34B of the Federal Court Rules is the expert assessor provided for by s 217 of the *Patents Act 1990* (Cth). This section provides:

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16 See also *Federal Court Rules*, Order 10, Rule 1(2)(cab) under which the Court can direct the parties to consider jointly instructing an expert.

17 This method was first developed by Lockhart J in the Australian Competition Tribunal.

18 Justice P Heerey, 'Expert Evidence in Intellectual Property Cases' (1998) 9 *Australian Intellectual Property Journal* 92, 98–9; Justice P Heerey, 'Expert Evidence: The Australian Experience' (2002) 7(3) *Bar Review* (Ireland) 166, 170.

19 See, eg, *Robert Hicks Pty Ltd (t/as Auto Fashions Australia) v Melway Publishing Pty Ltd* (1998) 42 IPR 627, 645–6 (Merkel J).

A prescribed Court may, if it thinks fit, call in the aid of an assessor to assist it in the hearing and trial or determination of any proceedings under this Act.

In *Genetics Institute Inc v Kirin-Amgen Inc (No 2)*,[20] Heerey J used an assessor to help explain the complicated biotechnology concepts involved in a disputed patent.[21] The assessor sat with Heerey J in court and helped explain the scientific evidence to him as the case progressed. The assessor's role was of course limited to providing advice on the scientific issues.[22]

IV CONCLUSION

The challenges to the courts from the revolutions in genetics and in biotechnology seem particularly acute when we consider the incredible speed at which developments in these areas are taking place. In 50 years, we have moved from the description of the double helix structure of DNA by Watson and Crick, to the mapping of the human genome. The pace continues to accelerate with previously unimagined prospects coming into view.[23] It is not, however, just the unprecedented velocity of change. The enlarging fields in which the advances in genetics and biotechnology are taking place broaden and increase the challenges.

The level at which judges will need to understand the science will of course depend upon whether a case involves technical issues as in a patent case, or whether larger questions are raised. We can expect some cases to assume the science and to turn on the profound issues of ethics and policy that the science has raised.

In whatever form the challenges come, though, it will be essential for judges to be well informed and to develop ways in which, consistent with the fundamental values of the law, the scientific and ethical issues can be fully explored and understood.

The changes that genetics and biotechnology bring are likely to be focused upon our fundamental needs and aspirations, but not necessarily upon those that all would consider good. The genetics and biotechnology of the 21st century are

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20 (1998) 155 ALR 30.

21 Court appointed assessors have also been used in New Zealand, see *Beecham Group Ltd v Bristol Myers Company* [1980] NZLR 185, 192; the United Kingdom, see *Biogen Inc v Medeva plc* [1997] RPC 1; and in the United States, see *Nevamar Corp v Charleswater Prod Inc*, MJG-88-3732, *Association of Mexican-American Educators v California*, 231 F.3d 572 (9th Cir, 2000) and *TechSearch, LLC v Intel Corp*, 286 F.3d 1360 (Fed Cir, 2002). In the United States, court appointed assessors have been described as having a similar role to a judge's clerk (or associate in Australia), but from a scientific perspective.

22 For more about the procedure used in *Genetics Institute Inc v Kirin-Amgen Inc (No 2)* (1998) 155 ALR 30 see Justice P Heerey, 'Expert Evidence: The Australian Experience', above n 18, 166.

23 The draft sequence of the human genome was completed in 2001. By late 2002, the genetic sequence of 125 organisms had been completed, and the genetic sequence of 580 other organisms was underway. These organisms range from the malaria parasite, to tomatoes, to cows: see Mattick, above n 6, 212. The last few years have also seen rapid developments in the fields of embryonic and adult stem cell research and in gene therapy. At the 'Genetics in the Courtroom' conference Dr Ian Alexander, Clinical Head of Gene Therapy at the Westmead Children's Hospital, gave a fascinating presentation about partial success in the treatment of infants with severe combined immunodeficiency (a disease which usually leads to death before the age of one year) using gene therapy.

calculated to interact directly, profoundly, and at astonishing speed with the whole world of economics, culture and belief.

It is in the courts that our society sometimes plays out its most complex, emotive and seemingly unresolvable conflicts. The revolutions in genetics and biotechnology are bound to give rise to these conflicts in the future. We cannot hope to anticipate all the questions that will confront us, but we can, with the assistance of innovative approaches such as those presented by EINSHAC, begin to equip ourselves with the tools necessary to gain the knowledge, understanding and insight needed to help us to answer those questions when they do confront us.