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Précis Paper

Legal and Ethical Issues in Genomic Testing

A discussion of the legal and ethical issues concerned with genomic research and testing, and how clinical practitioners can best respond to these

Discussion Includes

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- Purposes of genomic testing
- National Health Genomics Policy Framework
- Legal and ethical issues concerned with genomic testing:
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Legal and Ethical Issues in Genomic Testing

1. In this edition of BenchTV, Alison Choy Flannigan (Partner – Holman Webb Lawyers, Sydney) and Zara Officer (Special Counsel – Holman Webb Lawyers, Sydney) discuss the legal and ethical issues concerned with genomic research and testing, and advise on how clinical practitioners can best respond to these.

Types of testing involved in genomic technology

2. There are three types of tests that are involved in genomic technology:
   i. DNA Ancestry testing - providing information about a person's ethnic mix;
   ii. Genetic testing - testing for the abnormality of a gene, which could be a pre-indicator of diseases;
   iii. Genomic testing - testing the full DNA of an individual, providing information for a number of medical conditions, research, or personalised medicine.

Purposes of genomic testing

3. Genomic testing can be used for the purposes of personalised medicine and research.

4. Genomic testing for the purposes of personalised medicine helps ensure that medicines that are supplied to specific individuals are targeted to those individuals. This can make health spending more efficient and provide better clinical outcomes for individuals.

5. Genomic testing for research purposes can help identify populations where particular treatment is more effective, or populations where there are specific health issues.

National Health Genomics Policy Framework

6. The Council of Australian Governments has endorsed the National Health Genomics Policy Framework. The Council has identified the following as focus areas of the framework:
   i. a person-centred approach to genomic testing in healthcare;
   ii. building a skilled workforce literate in genomics;
   iii. ensuring sustainable and strategic investment in cost-effective genomics; and
   iv. maintaining quality, safety, and clinical utility of genomics in healthcare.

7. One of the main priorities of the Council's review is to establish a National Genomic Data Governance Framework that aligns with international frameworks. This is considered a good idea, particularly where some DNA testing is undertaken by private companies that are not regulated as much as they should be.

Legal and ethical issues concerned with genomic testing

8. There are a number of legal and ethical issues concerned with genomic testing. These are: consent; duty of care; cost; confidentiality; selection of IVF embryos; privacy; intellectual property in completed genomic research; and emerging issues.

Consent

9. There is not only a legal, but also an ethical, obligation on clinicians to tell patients what their options are, what their information will be used for, and to whom it will be provided.

10. Due to constantly evolving medical technology, the nature of what can be consented to is always changing.

11. Consent raises issues in relation to privacy. For example, as genetic test results are specific to an individual, unlike other health and medical records, they cannot be completely de-identified, such that if a hospital has a database of genetic test results, and someone wants to access those results in the future, the hospital must be aware of who it is consenting to have access to that information, and what it is consenting them to do with the information.

Duty of care

12. Professional obligations will be underpinned by the primary and existing duties of care owed by clinical practitioners. However, where clinical care generates and relies on the collection of data, then the duty of care to patients also extends to data managers and researchers who co-produce information of clinical significance.

13. The analysis of large quantities of genomic data can increase the accuracy of, for example, cancer diagnosis, accelerate diagnostic processes for rare diseases, and enable movement towards personalised medicine. But these developments rely on a blurring of the lines between research and clinical care, which historically have different ethical frameworks and duties of care.

14. The new world of genomics will have an impact on the duty of care as set out in ss 5B and 5O of the *Civil Liability Act 2002* (NSW), and also on the duties to warn as set out in *Rogers v Whitaker* (1992) 175 CLR 479 and *Wallace v Kam* [2013] HCA 19.

15. The duty to warn extends to material risks which may be attendant on a proposed treatment. A material risk is a risk to which a reasonable person in the position of the patient would be likely to attach significance in deciding whether or not to undergo the proposed treatment. With the exception of cases of emergency or necessity, all medical treatment is at the choice of the patient, a choice which would be meaningless if not based on relevant information and advice, so warning a patient of material risks is imperative.

16. Genomic testing will have an effect on the obligation of the duty to warn on clinicians. For example: is there a duty on a clinician to warn the daughters of a woman who has discovered she is predisposed to a particular breast cancer; should the daughters be warned even if the woman does not want her genetic information disclosed; should consent be obtained from the woman before performing the genomic testing; or after receiving the results; and would warning her daughters fall within exceptions to the *Privacy Act 1988* (Cth) and the *Health Records and Information Privacy Act 2002* (NSW), which allow disclosure of health information only in limited circumstances?

17. Another area for consideration concerning duty of care arises in genetic testing prenatally or at birth, and the potential conflicts that can arise between the child and the practitioners looking after the child, on the one hand, and the parents and their needs, on the other.

18. Genomic testing also gives rise to potential issues of foreseeability. These relate to situations where certain information becomes available after testing is done which might be clinically significant. The issue arising for clinicians here is: what is their duty to report incidental findings not having any clinical significance at present, but that may have in the future - is there a duty to re-contact the patient when new information or technologies come to light that are potentially applicable to the earlier testing?

19. Another issue arising is the question of when will a failure to recommend genetic testing or report findings be the catalyst for a negligence claim?

20. A related issue is what is the duty of the genetic and genomic testers to also offer genetic counselling: should that be an ethical duty, or should that form part of the legal duty of care? Cases have emerged where plaintiffs have sued because they were not offered genetic counselling. This is an emerging area of the law which will continue to evolve.

21. Given the increasing practice of clinicians ordering diagnostic tests as a defensive mechanism, to protect themselves against being sued, this practice may also start being seen more in relation to genetic testing.

Cost

22. The costs involved with genomic testing are a significant ethical issue with this type of technology. Cost is connected to equity of access: should the rich have better services than the poor?

23. Due to limited government budgets, the costs of genomic testing are unlikely to be included in Medicare at present, although, as with all technologies, the cost will reduce over time.

Confidentiality

24. As genetic information cannot be fully de-identified, this gives rise to the ethical issue of maintaining a patient's confidentiality.

25. Confidentiality of information raises the issue of the conflict between individual benefit and the greater good: protecting the privacy of the individual versus the greater good of helping large numbers of people. The increasing prevalence of data banks for DNA information gives rise to the question of whether it is fair to restrict that information for the protection of the individual where that information could be used to potentially save millions of people's lives.

26. However, protecting an individual's privacy and using their information for the common good do not necessarily have to be mutually exclusive - if a governance framework with appropriate protections is in place, researchers should be able to access an individual's data for the purpose of helping people.

Selection of IVF embryos

27. There is a plethora of discussion on the issue of whether genomic testing should be used for couples to choose the sex of their baby, or to choose which embryo should be fertilised and what baby should be born.

Privacy

28. Under the *Privacy Act 1988* (Cth), genetic information falls under the definition of "health information", which is defined in s 6FA as including "genetic information about an individual in a form that is, or could be, predictive of the health of the individual or a genetic relative of the individual".

29. The recent September 2016 Australian Red Cross Blood Service data breach showed that there are databases in place holding the genetic information of a number of individuals. In that case, approximately 550,000 records of potential donors held by the Red Cross were inadvertently placed onto a publicly available website, resulting in a significant data breach.

30. The new mandatory notification of data breaches commencing in February 2018 will result in more disclosure of data breaches concerning information held on large databases.

31. The *Privacy Act* applies only to the Commonwealth government sector and private sector health service providers that collect, use, and disclose health information. Though the *Privacy Act* does not apply to State government, there are various pieces of legislation in Victoria, New South Wales, and the Australian Capital Territory that regulate health information in both the public and private sectors.

32. The *Privacy Act 1988* (Cth) has certain protections and requirements in relation to the collection, use, and disclosure of personal information, under which genetic information falls. Under the Act, genetic information can be used to provide a health service to the individual to which the information relates, which is the primary purpose of collection. Genetic information can also be used for a secondary purpose which is directly related to the primary purpose.

33. Before dealing with an individual's information, it is advised that practitioners obtain that individual's consent where possible. However, s 16B(4) of the *Privacy Act* (and similar provisions in equivalent State and Territory legislation) allows for the disclosure of genetic information to certain individuals without the consent of the individual to which the information relates.

34. Section 16B(4) of the *Privacy Act* allows for the disclosure of genetic information without consent only where:

> (a) the organisation has obtained the information in the course of providing a health service to the first individual; and
> (b) the organisation reasonably believes that the use or disclosure is necessary to lessen or prevent a serious threat to the life, health or safety of another individual who is a genetic relative of the first individual; and
> (c) the use or disclosure is conducted in accordance with guidelines approved under section 95AA; and
> (d) in the case of disclosure - the recipient of the information is a genetic relative of the first individual.

35. The National Health and Medical Research Council has issued, under s 95AA of the *Privacy Act*, guidelines for the use or disclosure of genetic information without consent, which must be followed by medical practitioners under the section. These guidelines apply to health service providers and not to commercial genetic testing companies.

36. The guidelines set out a number of requirements. These include:

i. the "use or disclosure of genetic information without consent may proceed only when the authorising medical practitioner has a reasonable belief that this is necessary to lessen or prevent a serious threat to the life, health or safety of a genetic relative" (this is different from the previous version of this section, which required not only that the threat be serious, but also imminent);

ii. "[s]pecific ethical considerations must be taken into account when making a decision about whether or not to use or disclose genetic information without consent";

iii. further, "[r]easonable steps must be taken to obtain the consent of the patient or his or her authorised representative to use or disclose genetic information".

37. Practitioners are advised to encourage their patient to talk to their relative first, if possible, before the practitioner tries to talk to the relative without the patient's consent. In deciding whether to speak to the patient's relative, practitioners must take into consideration the following: what are the chances of the relative to have the disease, what treatments are necessary, etc. If there is a small chance that the patient's relative may have the disease in spite of their genetic predisposition, then there would be less of a case for the practitioner to inform the relative.

38. The Guidelines say that there is currently no legal obligation on practitioners to tell a family member that they have a predisposition to a genetic disease where the practitioner knows the existence of that genetic trait in the family, however, the law may change in this regard.

39. In relation to research and the privacy of genetic information, any research must comply with the privacy legislation, and undergo ethical review through a relevant human research ethics committee (particularly in the case of clinical trials). Researchers are advised that patient information sheets and consent forms should go through the ethics committee for approval.

40. The *Privacy Act* and related State legislation allow for health and medical research to be conducted without the consent of the individual, provided that certain requirements are met. These include: human research ethics committee approval, and ensuring that the information is protected (for example, any final report of that research must not name any individuals).

41. Research clients are advised to consider whether the patient information they wish to use for research can be de-identified, as the privacy legislation does not apply to de-identified information. Even though genetic information is not truly de-identifiable, de-identifying as much of it as possible will enable the research to proceed with significantly less regulation, and will make a difference to how that information can be used and who can access it.

Intellectual property

42. The High Court case of *D'Arcy v Myriad Genetics* [2015] HCA 35, unanimously allowing an appeal from the Full Court of the Federal Court, provides guidance on the issue of the intellectual property arising in genomic research after it has been completed. In that case, the High Court held that an isolated nucleic acid (coding for a BRCA1 protein with specific variations from the norm that are

indicative of susceptibility to breast cancer and ovarian cancer), was not a patentable invention within the meaning of s 18(1)(a) of the *Patents Act 1990* (Cth), as it did not define a "manner of manufacture".

43. Following *D'Arcy v Myriad Genetics*, the Commissioner of Patents has established a revised patent examination practice, recommended to practitioners practising in this area. This examination practice provides that the law is to be applied on a case-by-case basis, taking into account the principles and approach of the High Court in *D'Arcy v Myriad Genetics*, subject to other requirements of patentability, such as utility, novelty, and inventiveness.

44. It is useful to ask the following questions:

i. What is the substance of the claim?
ii. How is the substance of the claim made, or changed by man, or is it artificial?
iii. Does the invention have economic utility?
iv. Does the invention, as claimed, represent a new class of claim?

45. Determining whether the substance of the claim is made or is artificial involves a comparison of the state of affairs before the invention, and the state of affairs as a result of the invention. Relevant factors include:

i. whether the substance of the claim was made, created, or modified by human action;
ii. what are the physical differences between the claim and the natural state;
iii. what was the labour required to produce the product.

Emerging issues

46. Infrastructure is a key emerging issue in genomic testing. Apart from the responsibilities to ensure accountable governance, data security and regulation, care will need to be taken with respect to establishing databases and infrastructure sufficient for handling the vast quantities of genomic data. Quantum computing may be of use in this area, as current computing may be unable to deal with the amount of data available.

47. Another emerging issue is that curators of the data must keep ahead of hackers. This will be an ongoing issue.

48. As results can vary in terms of clinical utility, another emerging issue is the question of what are the duties of clinicians in cases where results cannot be acted upon at present, but are able to be acted upon at a future point in time.

49. A final issue is potential discrimination on the basis of genetic information, including for insurance products.

50. All of these emerging issues tie back to the content and scope of consent from patients who give their genetic information. Special care needs to be taken because it is very difficult to control what that information will be used for.

Summary

51. Genetic and genomic testing has placed us at the doorstep of a new generation of technology in relation to healthcare. As the legal and ethical issues involved in genetic and genomic testing are constantly evolving, this gives rise to more questions than answers at present.

BIOGRAPHY

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Alison is a partner in the Sydney Office of Holman Webb and has over 20 years of corporate, commercial and regulatory experience. She also She is also a company secretary for the National Foundation for Medical Research and Innovation. Alison was previously Company Secretary for Research Australia Limited and partner in two law firms specialising in health, aged care and life sciences.

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Zara is Special Counsel of Holman Webb and focuses on litigation, medical negligence, insurance, and discrimination. She was previously Special Counsel of Henry Davis York and a Senior Associate at Hunt and Hunt. Zara holds a Bachelor of Arts (Honours) and a Bachelor of Laws from Monash University.

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