

# CHAPTER 3.5: HUMAN GENETICS

## INTRODUCTION

The genome is an individual's biological inheritance. An individual's biological characteristics are determined by the interaction of his or her genome with the environment. An individual's genome contains all of his or her genes.

Genetics is the study of the structure, location, function, expression, interaction, abnormalities and effects of the genes or genetic material and their products, including but not limited to studies of the structure of the nucleic acids and other molecules that make up the genetic material.

Genes and genetic information are being studied increasingly in clinical, epidemiological and social research, as well as in basic research.

Genetic research may involve study of:

- single or multiple genes, gene-to-gene interaction or gene-environment interaction;
- acquired somatic variation;
- inherited gene sequences, and their variants or their products;
- gene expression, including the influence on those genes of environmental factors, pharmaceuticals and other therapeutic products;
- the genes of individuals, families or populations;
- epigenetics;
- use of informatics and genetic information; and
- clinical phenotypes.

Some research that falls within this broad description of genetic research does not involve information that is relevant to the future health of the individual participant and does not generate sensitivities for the individual, or his or her family or community. The guidelines in

this chapter differentiate between research that necessitates special precautions in that respect, and research that is unlikely to be of concern to individual participants, their families or their communities.

For genetic research using stored data, *see also Chapter 3.2: Databanks*; and for genetic research using human tissue samples, *see Chapter 3.4: Human tissue samples*.

There are ethical issues specific to genetic research because:

- many of an individual's genes are shared with close genetic relatives (commonly called 'blood relatives') and with unrelated people in the population; and
- genetic research can reveal information about predispositions to disease. Although people with such a predisposition may not develop the disease, the information may have implications for their access to employment and education and to benefits or services, including financial services such as banking, insurance and superannuation. The information may also have similar implications for blood relatives.

Research results and genetic material and information collected for genetic research may be significant for blood relatives of research participants. These family members may have an interest in their relatives' genetic material, or in information the research generates, because testing that material or acquiring that information may create new options for life decisions, including those with potential to improve health. However, some family members may prefer not to be given such information, or even not to know of its existence. In addition, other family members who are not blood relatives, such as partners and spouses, may have an interest because of concerns about the health of offspring. Genetic research can also

reveal information about previously unknown paternity or maternity. Genetic research also has uses outside health, such as for tracing migration patterns and in studies of cultural relatedness.

**Research to which this chapter applies must be reviewed and approved by a Human Ethics Research Committee (HREC) rather than by one of the other processes of ethical review described in paragraphs 5.1.7 and 5.1.8 (page 78), except where that research uses collections of non-identifiable data and involves negligible risk, and may therefore be exempted from ethical review.**

**Values, principles and themes that must inform the design, ethical review and conduct of all human research are set out in Sections 1 and 2 of this National Statement. The guidelines and headings below show how those values, principles and themes apply specifically in research that is the subject of this chapter.**

## GUIDELINES

### Research merit and integrity

- 3.5.1 Where research may discover or generate information of potential importance to the future health of participants, or their blood relatives, researchers must prepare and follow an ethically defensible plan to disclose or withhold that information.
- 3.5.2 This plan must take into account the clinical relevance of the research information, the types of genetic test used in the research, and the results of those tests. In addition:
- (a) The plan should:
    - (i) enable participants to decide whether they wish to receive the information and who else may be given the information;
    - (ii) set out a process for finding out whether those other people want to receive information;

- (iii) include procedures to inform participants that the information would remain potentially identifiable;
  - (iv) include measures to protect the degree of confidentiality that participants wish to maintain.
- (b) When participants or their relatives are to be given or notified of genetic information that may be important for their health, the plan should either provide access to genetic and clinical advice and counselling, or clearly recommend to participants that they seek these services. Such advice and counselling should be provided by professionals with appropriate training, qualifications and experience.
- (c) Where participants or relatives prefer not to receive genetic information that is important for their health, they should be advised that they will be approached to confirm this decision when the results of the research are available.
- (d) Where the potential relevance of genetic information to participants' health is not clear until after interim analysis of the research information, participants should again be given:
- (i) the option of being notified of the existence of that information;
  - (ii) the option of receiving the information; and/or
  - (iii) access to, or a recommendation to seek, advice or counselling about the implications of these decisions.

- 3.5.3 Advice about the results of genetic research needs to include a clear explanation of the difference between research and clinical testing, and to clarify any need for clinical testing of research results.

## Justice in the use and disclosure of genetic information

3.5.4 Researchers should consider the potential psychological, social and cultural significance of their research. Where complex socially significant characteristics or the genetic characteristics of communities are being investigated, there is a risk that the research may be misrepresented or misused in ways that lead to prejudice, disrespect or other harm to participants or communities. In designing, conducting and reporting research of this nature, researchers should consider how to counter the possibility of such harm.

## Beneficence

3.5.5 Identifiers of genetic material or related information:

- (a) should not be removed without the consent of participants, if removal would make it difficult to communicate personal results;
- (b) should be removed if participants request it, provided they have been informed that the material or information would remain potentially identifiable.

3.5.6 Genetic information can sometimes be misused to stigmatise people or to discriminate against them unfairly. Researchers should therefore take special care to protect the privacy and confidentiality of this information. Statutory or contractual duties may require participants to disclose the results of genetic tests or analysis to third parties (for example, insurance companies, employers, financial and educational institutions), particularly where results provide information about health prospects. Genetic research should be designed to minimise any resultant risk that participants will be deprived of benefits available to others in the community. Potential research participants should be advised of any such risks.

3.5.7 Researchers should not transfer genetic material or related information to any researcher not engaged in the research project unless:

- (a) either
  - (i) participants have been informed about and have specifically consented to that transfer and, where the material or information is identified, there is a defensible plan as specified in paragraphs 3.5.1 and 3.5.2 for withholding or disclosing it; or
  - (ii) the provisions for extended or unspecified consent set out in paragraph 2.2.14 (page 21) have been met; or
  - (iii) an HREC has judged that the conditions for waiver of consent have been met (see paragraph 2.3.6, page 24), and has approved the transfer;
- (b) the transferring and receiving researchers are conducting research that has been ethically approved in Australia or through an equally stringent process in another country; and
- (c) the receiving researcher/s undertake/s not to permit attempts to re-identify the material or information or otherwise reduce the protection of the privacy of the participants or of the confidentiality of the information.

## Family involvement

3.5.8 Where people are asked to consent to the collection of their genetic material or information for research, they should be given information required by paragraph 2.2.2 (page 19) and, in addition, be advised:

- (a) that genetic material is in principle re-identifiable, even if identifiers are removed;

- (b) that they are free to decline without giving reasons;
- (c) about arrangements to ensure the privacy and confidentiality of their genetic information with regard to both family members and others, in accordance with the defensible plan for disclosing and withholding information (see paragraph 3.5.2);
- (d) whether information from or about family members, in addition to that provided by participants, is required for the research;
- (e) whether the research may reveal information of potential importance to their future health, or the future health of their blood relatives;
- (f) that, if it is proposed to approach blood relatives, consent to do so will first be sought from the participant;
- (g) that, if the research discloses that a family member may be at risk of a life-threatening or serious illness for which treatment is available or pending, this information may, with the approval of an HREC, be offered by a clinician to the family member, even if the research participant does not consent to this; and
- (h) whether the research has the potential to detect previously unknown paternity or maternity, or non blood-relationship to siblings, and whether, how and to whom this information will be disclosed, according to the approved plan.

3.5.9 In deciding if relatives should be approached, researchers should consider:

- (a) the privacy and any known sensitivities of the relatives;
- (b) accepted habits of communication within the family; and

- (c) whether the harms that might result from the relatives' participation in the research are justified by the potential benefits of their participation.

3.5.10 Where a participant has given consent to approach relatives, the opportunity to make initial contact should be given to the participant or someone else he or she chooses.

### Community involvement

3.5.11 Consent should be sought from appropriate community representatives as well as from the individuals concerned (see paragraph 2.2.13, page 21), where:

- (a) researchers propose to collect genetic material and information from individuals who are chosen because of their membership of a particular community;
- (b) the research involves sensitivities for that community; and
- (c) there is known to be a culturally relevant community structure involved in such matters.

### Other information to be given

3.5.12 Those whose consent is being sought for collection of identified or potentially identifiable genetic material or related information should also be informed:

- (a) if the research has potential to generate information that a participant may be legally required to disclose to a third party, for instance, for the purposes of insurance, employment, finance or education;
- (b) that genetic material and data may have uses unrelated to research. Participants should be advised that their material and data will not be released for such uses without their consent, unless required by law;

- (c) about any proposal, subject to participants' consent, to store their genetic material and data because it might be useful for as yet unspecified future research;
  - (d) that, if such consent is not given, the genetic material and data will be disposed of at the end of the research, once the sample storage and record-keeping requirements of good research practice have been met;
  - (e) that any wishes about the method of disposal will be recorded at the start of the research and taken into account at the time of disposal;
  - (f) that they are free to withdraw from the research at any time. Participants should be informed of any consequences of such withdrawal, including that they may request their genetic material and data to be disposed of, if the samples can be identified. They should also be clearly informed of any practical limitations on the granting of this request; and
  - (g) that, in research studying large numbers of genes simultaneously, participants will not be given the names of all the individual genes to be studied.
- reason, where genetic data are stored, confidentiality might sometimes require restrictions on the release of data for research use (see paragraph 3.2.8, page 31).

## Confidentiality

3.5.13 Researchers must ensure the confidentiality and privacy of stored genetic information or research results relating to identified or re-identifiable participants. Such information or research results should be disclosed to treating clinicians only in accordance with the consent given for the research.

3.5.14 The rarity of some genetic disorders might allow certain families or individuals to be identified by other researchers, and in some cases by members of the community, even if information is given to others in non-identifiable form. For this