

9. HUMAN GENETIC RESEARCH

Genetic research enhances our understanding of how genes and environmental factors interact to influence the health of individuals and communities. Genetic research also has the potential to generate knowledge that could improve the health of individuals and communities.

Genetic research can reveal information about an individual's susceptibility to disease and hence about the individual's future health. Such information may be of interest and benefit to research participants, especially where preventive strategies exist.

In addition to ethical considerations, which apply to all research involving humans, there are ethical issues unique to genetic research. They arise from the nature of genes and genetic information which, although personal, are also shared with other family members and with unrelated individuals in the population.

Participation of families rather than individuals is required for many genetic research studies. Research results and genetic material and information collected for research may be of significance to the health of blood relatives, including some who have not participated in the research. These family members may have an interest in their relative's genetic material or in information that the research generates, because testing that material or acquiring that information may create new options for life decisions, including those with the potential to improve health. However, some family members may prefer not to be given information that might provide knowledge of future health or health risks. 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.

Participants may be at risk of harm arising from the use of genetic information, including stigmatisation or unfair discrimination, and adoption of exclusionary policies. Researchers should recognise that special care must be taken to protect the privacy and confidentiality of genetic information. Research ethics committees should require researchers to consider whether a proposed genetic study might lead to a potential harm to participants, and what steps can be implemented to obviate such harm. The results of genetic tests, especially those that provide information about future health, could be used, potentially, by third parties such as insurance companies and employers to assist with decisions concerning research participants and their families. By participating in genetic research, people should not be put at risk of being deprived of benefits available to other members of the community.

9.1 Social significance and consequences of human genetic research

Researchers should consider the social and cultural significance of their research, especially in the areas of complex socially significant characteristics and the genetic characteristics of collectivities. When such characteristics are the subject of research, Research ethics committees should satisfy themselves that there are no contestable or dubious ethical values subsumed within the research protocol.

When assessing proposals of this type, ethics committees should consider the balance between the contribution to knowledge and the potential for harm to individuals or collectivities. One of the potential harms that may arise from genetic research is the adoption of exclusionary policies. Researchers should be asked by ethics committees whether their studies may lead to such exclusionary policies and what steps they can implement to obviate such policies.

9.2 Privacy and confidentiality

Researchers must ensure the confidentiality and privacy of stored genetic information or research results relating to identified or potentially identifiable participants.

Researchers must keep information provided by participants about family members confidential. Such confidential information must not be revealed either to family members or to persons who are not family members.

The research protocol must specify whether genetic information or genetic material, and any information derived from studying the genetic material, will be stored in identified, potentially identifiable (coded) or de-identified (not identifiable, anonymous) form. (See [Section 7](#)). Researchers should be aware that the rarity of some genetic disorders might allow certain families to be identified by other researchers, and in some cases by members of the community, even where information has been communicated to others in de-identified form.

Researchers should consider carefully the consequences of storing information and material in de-identified form for the proposed research, for future research and for communication of research results to participants.

Identifying genetic information must not be released to others, including family members, without the written consent of the individual to whom the information relates, or a person or institution which may legally provide consent for that person.

A researcher must not transfer genetic material and related information to another research group unless:

- The researcher and the other research group are collaborating on research that has been approved by an ethics committee;
- The genetic material and information is provided in a form that ensures that participants cannot be identified. However, an ethics committee may approve transfer of genetic material and information, which is identified, or potentially identifiable, in certain circumstances. Where this occurs, the other research group must undertake to hold the material and related information in such a manner that there is no reduction in the protection of the privacy of the participants or of the confidentiality of the information.

9.3 Consent

The investigator or other appropriate person or organisation as specified in guidelines of this statement, must obtain a consent for human genetic research unless an ethics committee waives the requirement for consent.

When consent is sought from participants for collection of genetic material and information, they should be informed:

- That they are free to refuse consent without giving reasons. Researchers should be aware that for some genetic research, an individual's participation may be requested by, and may primarily serve the interests of, other family members and the individual may agree to participate out of a sense of obligation;
- About arrangements to ensure the privacy and confidentiality of their genetic information both with regard to other family members and persons who are not family members. Participants should be informed whether their genetic material and information will be used in an identified, potentially identifiable, or de-identified form and, if their material or information is to be de-identified, that it will not be possible to provide them with personal research results;
- Whether or not the research might reveal information of potential importance to the future health of an identified or potentially identifiable participant or the participant's offspring;
- That the researchers will endeavour to provide information about the outcome of the research. Participants should be advised when it is not intended to provide feedback. Participants should

be asked whether they wish to be notified of research results that relate to them as individuals. A decision not to be notified should be respected;

- That if the research generates information that might be relevant to the health of other family members, the consent of participants will be sought before offering to disclose such information to the family members concerned;
- Whether or not information about family members, in addition to that provided by participants, is required for the research;
- That consent to approach relatives will first be obtained from the participant. In deciding to recruit relatives, researchers must consider the privacy and any known sensitivities of the relatives, accepted habits of communication within the family, and the balance of potential benefits and harms that might result from their participation in the research;
- Whether or not the research has the potential to detect non-paternity or non-maternity;
- That genetic material and information may have uses unrelated to research approved by a research ethics committee. Participants should be advised that their material and information will not be released for other uses without their consent, unless required by law. If consent is given, the duration of storage should be specified. If consent for future research use is refused, the genetic material and information should be disposed of at the end of the research, once the sample storage and record-keeping requirements of good research practice have been met;
- Whether or not their genetic material is to be disposed of on completion of the research or after a further period of storage. Some participants or collectivities may have sensitivities regarding disposal of their genetic material. These sensitivities should be established and recorded at the start of the research and account should be taken of them at the time of disposal;
- That they are free to withdraw from the research at any time. This may involve a request that their genetic material and information be disposed of, provided the samples can be identified. Alternatively, samples and information may be retained provided they are de-identified, depending on the wishes of the participants.

When researchers propose to collect genetic material and information from individuals chosen by virtue of their membership of a particular collectivity, consent should be sought from appropriate collectivity representatives as well as from the individuals concerned, in accordance with Research Involving Collectivities.

9.4 Where the requirement for consent could be waived

As a general principle, where a researcher proposes to conduct research using stored genetic material or genetic information, consent is required from the person from whom the material was derived, or to whom the information relates.

An ethics committee may sometimes waive, with or without conditions, the requirement for consent. In determining whether consent may be waived or waived subject to conditions, a research ethics committee may take into account:

- The nature of any existing consent relating to the collection and storage of genetic material and genetic information;
- The justification presented for seeking waiver of consent, including the extent to which it is impossible, difficult or intrusive to obtain consent;
- The proposed arrangements to protect privacy, including the extent to which it is possible to de-identify the genetic material and genetic information;
- The extent to which the proposed research poses a risk to the privacy and wellbeing of the participant;
- Whether the research proposal is an extension of, or is closely related to, a previously approved research project;
- The possibility of commercial exploitation of derivatives of the sample, and relevant statutory provisions.

Institutions or organisations wishing to conduct research on genetic material and on information collected for non-research purposes, should develop and disseminate a general policy that informs patients that such material and information may be used for future research, following research ethics committee approval, subject to the issues raised in the first and second paragraphs of section 9.4. Patients of such institutions or organisations should be informed that this policy exists, and that their privacy and confidentiality will be protected. They should be given the opportunity to refuse consent to the use of their material and information for such research.

9.5 Genetic counseling

When research may reveal information of potential importance to the future health of an identified or potentially identifiable participant's future health or to the participant's offspring, the research protocol must provide for consent procedures, counselling, support, test quality and test result confidentiality, as would apply if the participant sought such information in a clinical setting. Otherwise such research may be performed only if the genetic material has been de-identified. Counselling and information arising from the research must be provided by health professionals who have appropriate training, skills and experience.

Participants who are asked to consent to the use of their genetic material and information for future research, should be counselled about the possible consequences of doing so. In general, their genetic material and information will be used for future research in de-identified form, and feedback will not be possible. However, the research ethics committee may direct the researchers to use the genetic material and information in potentially identifiable (coded) form. In such instances, the views of participants regarding the feedback of information of potential significance to their own or their relatives' future health should be established, recorded and respected. If feedback is requested, the participant should receive information and counselling about the implications of receiving that information. This could be provided at the time of obtaining consent or in the future, prior to the provision of the feedback.