

# STATEMENT OF PRINCIPLES ON THE ETHICAL CONDUCT OF HUMAN GENETIC RESEARCH INVOLVING POPULATIONS



## WHY ANOTHER STATEMENT FROM THE RMGA?

Recent advances in genomics and bioinformatics allow for genetic analyses on whole populations. In the fields of human genetic research and genetic medicine, such advances raise ethical issues. Indeed, consultation, governance and benefit-sharing, although not entirely new, are not central to the usual genetic studies of families and cohorts.

Because the “subject” of research is now a whole population or community, the principles that should guide such research range from the respect of the individual to the recognition of his/her participation in the pursuit of knowledge for humanity.

The Quebec Network of Applied Genetic Medicine (RMGA) of the FRSQ, as the project manager of the proposed genetic map of the Quebec population (CARTaGENE), presents to the international community a *Statement of Principles*. This framework seeks to direct and harmonize such large genetic studies where the population is a partner in the research.

– CLAUDE LABERGE, DIRECTOR RMGA

This *Statement of Principles on the Ethical Conduct of Human Genetic Research Involving Populations* is based on a framework of ten fundamental principles giving rise to specific recommendations and procedures for their implementation.

Population genetic research includes research projects that investigate the dynamics and the structures of genetic variation in either populations or subsets thereof, generally defined by their geography, ethnicity, language, religion, etc.

It is the intention of the Quebec Network of Applied Genetic Medicine (RMGA) that this *Statement* is to be interpreted consistent with its previous *Statement of Principles: Human Genome Research* (2000) on DNA sampling and banking in individuals and families.

In the following *Statement*, the expression “genetic data” means genetic material as well as genetic information.

Furthermore, RMGA recognizes the fundamental principles of bioethics: respect for persons, beneficence, non-maleficence and justice as well as those relevant to genetic research from the following international organizations: United Nations Educational, Scientific and Culture Organization (UNESCO), Human Genome Organization (HUGO) and the World Health Organization (WHO).

All human rights and ethical principles flow from respect for the inherent dignity of the person. Population research is impossible without the trust of participants. RMGA members consider participants as research partners.

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# FRAMEWORK OF PRINCIPLES

**INDIVIDUALITY:** Recognition of the uniqueness of the person mandating respect for the autonomy of the individual within a given group.

**DIVERSITY:** Recognition and respect for difference through the fostering of a multidisciplinary approach.

**COMPLEXITY:** Interpretation of genetic information that recognizes its multi-variable nature and expression.

**RECIPROCITY:** Mutual exchange through consultation and communication.

**SOLIDARITY:** Protection from discrimination and from stigmatization.

**SECURITY:** Confidentiality of genetic data and strict control in the use or exchange thereof.

**ACCOUNTABILITY:** Adherence both to the recognized body of legal and ethical norms applying to research as well as to transparency and representation of the population in decision-making.

**EQUITY:** Participation, access, and the sharing of benefits and risks, taking into account the characteristics of each population.

**CITIZENRY:** Contribution to the public good and to the health of the population.

**UNIVERSALITY:** Knowledge dissemination and international collaboration.

These ten fundamental principles lay the foundations for the following recommendations and procedures.

## RECOMMENDATIONS

### 1. CONSULTATION

Respecting the principles of **RECIPROCITY** and **ACCOUNTABILITY** requires that research on a given population be based upon open dialogue between the population and the research team. A guiding mechanism for population genetic research is prior and ongoing public consultation.

#### IMPLEMENTATION

- ▶ Consultation and information should be offered to the population before recruitment begins.

- ▶ The role of the population in the process should be clarified at the outset.
- ▶ Continuous, meaningful dialogue should be maintained throughout the project.
- ▶ The values and cultural worldviews of the population should be taken into account at every stage of the research.
- ▶ The possible risks and benefits of research for the public good as well as for the population studied should be discussed.

### 2. RECRUITMENT

Genetic data about a population is a social and community resource that must be protected. Taking into account the principle of **EQUITY**, recruitment should ensure a process for participation that spreads the risks and benefits.

#### IMPLEMENTATION

- ▶ Researchers should justify the chosen methods of sampling and banking.
- ▶ Recruitment should be organized so as to respect privacy.
- ▶ Over-recruitment of any given population should be avoided.

### 3. CONSENT

Each individual carries a unique copy of the human genome. Yet, genes are shared by all humans as a species, as the common heritage of humanity. Research on any population should neither lose sight of the **INDIVIDUALITY** of the person nor of the **UNIVERSALITY** of the human genome.

#### IMPLEMENTATION

#### AUTONOMY OF THE PERSON (INDIVIDUAL CONSENT)

- ▶ Even in a population study, free, informed, and written consent of each individual is required, with legislative exceptions (for the surveillance of disease prevalence in a population, for example). It should be made clear to participants in a population research project that they are considered as a representative of their population.
- ▶ Consent is a continuing process and must be reconfirmed for instance in the case of significant changes to the research protocol, to the conditions of banking, in the research partnerships, and in the management of the bank.

- ▶ In all population studies, the individual retains the right of withdrawal except in the case of anonymization or where the law foresees otherwise.

#### GROUP INTERESTS (POPULATION SUPPORT)

- ▶ The right of expression (of both those who participate and those who do not) should be recognized through:
  - Support of representative and legitimate group(s) or absence of significant opposition;
  - Withdrawal by the population of its support for the research through mechanisms determined at the outset of the research.

## 4. CONFIDENTIALITY

Mutual confidence between the researcher and the population is essential to **RECIPROCITY**. To respect this confidence, the researcher should ensure the **SECURITY** and confidentiality of population data.

#### IMPLEMENTATION

- ▶ Regardless of the size of the data bank, measures must be instituted to ensure the safe conservation of the information.
- ▶ Management and exchange of data should be protected by appropriate mechanisms of confidentiality, such as coding or anonymizing the samples, confidentiality agreements binding the users, identification of the person accountable for the use of data, etc.
- ▶ Confidentiality, dissemination, and access mechanisms should be communicated to the population.
- ▶ An independent authority should oversee the overall protection of personal information.

## 5. GOVERNANCE

The creation of a genetic data bank and its use in population genetic research should respect current legal and ethical norms. **ACCOUNTABILITY** to the public and to a research ethics board is mandatory.

#### IMPLEMENTATION

- ▶ A banking policy should be created and made public (*see Statement, 2000*).
- ▶ Evaluation by an independent research ethics board should precede recruitment. This ethics board should collectively be knowledgeable in genetics and population research. Its composition should be multidisciplinary and include representatives from the population studied. A research ethics board should evaluate and monitor the constitution of the bank and also approve any research projects as well as requests for access.

- ▶ A committee should also be set up to oversee the creation and management of the bank. This committee will provide for the long-term adherence to the policy. The composition of this committee could include for example: a representative of the population studied, a participant from the group under study, a databank manager, a representative of the social sciences and humanities, and experts in human genetics and computer sciences.
- ▶ In the case of international partnerships, the highest ethical standards in force among the countries involved should apply, taking into account their values.

## 6. COMMUNICATION OF RESEARCH RESULTS

**RECIPROCITY** implies that researchers will regularly share data out of respect for the participation of the population. Researchers should exercise caution in the communication and interpretation of their results due to the **COMPLEXITY** of genetic information and its personal, familial, and social impact. They should adopt a multidisciplinary approach to reflect the values of the population and avoid misleading the public in the interest of **SOLIDARITY**.

#### IMPLEMENTATION

#### COMMUNICATION OF GENERAL RESULTS

- ▶ Results should be made public.
- ▶ By communicating results in a timely and diligent fashion, researchers contribute to a better understanding of the determinants of health.
- ▶ It is reasonable to expect that researchers should communicate with the population and the relevant governmental authorities regarding results that are pertinent to the improvement of health and/or the prevention of disease.
- ▶ Where appropriate, researchers, in collaboration with the population concerned, should facilitate the development and the implementation of a follow-up plan.

#### INTERPRETATION OF THE RESULTS

- ▶ The population studied should be made aware of possible socio-economic discrimination or group stigmatization as a result of perceptions of genetic risks. The population should also be informed of the means taken to minimize the risks.
- ▶ To avoid misleading or unrealistic expectations, the researchers should make the limits of the results and their applicability known.

## 7. COMMERCIALIZATION

Researchers and those responsible for the bank do not own the genetic material collected but may aspire to the acquisition of intellectual property rights over inventions derived from genetic data. The research team should be transparent in its operations. For the sake of **EQUITY**, population research should promote the attribution of benefits to the population.

### IMPLEMENTATION

#### BENEFIT SHARING

- ▶ The eventual sharing of any benefits with the population should be discussed at the outset. This sharing could take different forms such as: an access to medical care, to future treatments or drugs developed; a contribution of a portion of the benefits to a humanitarian organization; support for local needs, or support for technological infrastructures or health services to the population, etc.
- ▶ Benefit sharing cannot be limited to the individuals who participated. The research must, in consideration of the principle of equity, provide advantages for the whole population.
- ▶ Benefit sharing should not constitute undue inducement to participate.

#### FREEDOM OF RESEARCH

- ▶ Freedom of research should be promoted by respecting the principle of public access to the bank.

#### CONFLICT OF INTERESTS

- ▶ All perceived or real conflicts of interest should be disclosed.
- ▶ Mechanisms should be foreseen to take into account the interests of the population in any commercialization. For example, an independent body could be created for its management.

## 8. CONTRIBUTION TO THE WELFARE OF THE POPULATION

Population genetic research should endeavour to promote health and prevent disease, especially for the population studied. The principle of **CITIZENRY** requires the recognition of participation in health research. Given the **COMPLEXITY** of population genetic research, a multidisciplinary approach is essential.

### IMPLEMENTATION

- ▶ The research team should possess proven expertise in the field of population research.
- ▶ Partnerships with local research teams should be established and copies of samples must remain in the province/state/country of origin.
- ▶ Research should serve the interests of the population and be used for peaceful means.
- ▶ Research should be for the acquisition of scientific knowledge for the eventual improvement of health.

## 9. CONTRIBUTION TO THE WELFARE OF HUMANITY

The **UNIVERSALITY** of the human genome mandates the sharing of knowledge at an international level.

### IMPLEMENTATION

- ▶ Collaboration with researchers from other countries should be encouraged and results should be broadly disseminated.
- ▶ Scientists should also be encouraged to pursue research on all types of populations including those with rare or endemic diseases.