

COMMISSION DE L'ÉTHIQUE DE LA SCIENCE ET DE LA TECHNOLOGIE

*HU*man *GE*netic Databases

“*A HUGE deal!*”



solidarity
legitimacy and
transparency
autonomy

equity
responsibility
intellectual property
democracy

YOUR
FILE

Québec 

HUman **GE**netic Databases



© Gouvernement du Québec 2007

Legal deposit: First quarter 2007
Bibliothèque nationale du Québec
National Library of Canada

ISBN 978-2-550-49289-4

« **A HUGE deal!** »

FOREWORD¹

This document is specifically designed for CEGEP and university students; it represents a supplement, written in everyday language, to the publication entitled ***Les enjeux éthiques des banques d'information génétique: pour un encadrement démocratique et responsable*** ("The Ethical Issues of Genetic Databases: Towards Democratic and Responsible Regulation"), a CEST position statement released in February 2003.

Since students as a group are especially solicited to participate in clinical tests of all sorts, even DNA testing, the Commission would like to draw their attention to a few of the ethical aspects concerning genetic information and its storage in databases for research purposes.

A special section of the Commission Web site

<http://www.ethique.gouv.qc.ca/hugedatabank/index.html>

contains the PDF file for this document as well as additional information for those wishing to know more about the subject; links to texts and sites related to the subject of genetic information are also available at the site. The French unabridged version of the position statement and the information documents prepared for the Commission are also available on the CEST Web site: <http://www.ethique.gouv.qc.ca/fr/publications.html>. Lastly, the site provides information concerning CEST operations and activities and gives you the option of consulting or downloading these various publications.



¹ Translation from French to English was done in 2004, document edited and put online in 2007.

HUman **GE**netic Databases



PROPERTY

TRANSPARENCY

LEGITIMACY

DISCRIMINATION

DEMOCRACY

an ethical perspective

The history of medicine could be approached in the same way as that of the earth: by dividing it into long periods, geological eras. Concerning medicine, we would no doubt be seen as having entered a new era in these last few years: *the genetic era*. And this classification would be right on the mark! Hasn't the advent of the Human Genome Project – and the sequencing of our 30,000 genes – made a huge splash? And wasn't the 50th anniversary of Watson and Crick's discovery of the structure of DNA celebrated in 2003?

We are the first generation to experience this new era of genetics and also the first to face a whole series of problems, including those arising from genetic screening and cloning. And also from "storing" DNA in so-called **genetic databases**, where human specimens are collected (DNA, cells and tissues), along with personal information of a genetic nature for research purposes.

Since genetic data are very personal indeed, and given that they are more easily processed and accessed in our computer age, the Government of Québec mandated the *Commission de l'éthique* de la science et de la technologie*, known as the CEST, to study the entire problem area of genetic databases and to produce a position statement on the subject. For instance, what exactly are genetic databases? How many are there in Québec? What about the consent of subjects targeted to participate in genetic database research? What is being done to ensure confidentiality as regards the information collected?



* "Ethics": Generally speaking, ethics refers to the process of searching for what constitutes a good and beautiful life. From the Commission's more specific perspective, adopting an ethical approach helps us define and clarify the values upon which to base our decisions as regards the development of science and technology, decisions that will be acceptable in light of the community and individual values of a given society during a specific era.

This brochure looks at what is at stake* by summarizing a few of the major issues addressed in the Commission's position statement: transparency, discrimination, legitimacy, property and democracy. And this will be done in a more direct manner than in the position statement itself, by placing the reader both in the obvious role of gene carrier and in other roles that he or she may play at certain moments in life when genetic information could be of personal importance, i.e. as a parent, patient, research volunteer, insured party or borrower, as well as a worker, researcher-entrepreneur or simple citizen.

It goes without saying that when drafting its position statement, the Commission was guided by a number of different values ranging, most notably, from respect for human dignity to solidarity and to justice as regards genetic information. And all this while putting particular emphasis on respect for democracy, a key element in the development of our society.

Last but not least, it is good to know that in this text the Commission chose to refer to HUMAN GENetic databases as HUGE databanks.

When historians of science study, a hundred years from now, our era of turmoil on the genetic front, let's hope they point out that the first solutions were handed down to future generations at this very point in time. Let's also hope they don't judge us too harshly.

* "Stakes": what can be won or lost in a competition or venture, or during a period of change characterized by an innovative way of doing things.

SOLIDARITY

... YOURSELF AS A GENE CARRIER

The creation of a HUGE population databank

As gene carriers, we all belong to a given population group and as such we could be called upon, in the years ahead, to contribute our genes as part of the development of a *population* genetic database. Those best known and most prominently featured in the media – in 2003, there were seven worldwide – include databases in Estonia, Great Britain, and Newfoundland. Québec would also like to have its own HUGE databank: the CART@GENE project has been designed with this goal in mind.

What is a HUGE population databank?

Population databases are important structures containing biological specimens or personal information (genetic or otherwise). They focus on the entire population or on a statistically representative sample thereof. They aim to identify the genetic characteristics of an entire society.

Such HUGE databanks differ from so-called *sectorial* databases – the other type of large database – which are designed to accumulate information on a specific population segment, for instance, people belonging to a distinct ethnic group or suffering from a particular disease or ailment.



For more information, you can consult the summary regarding population databases available on the Commission Web site at www.ethique.gouv.qc.ca.

Why create banks of this type?

These databases are of great interest for public health researchers since targeting the entire population in this way makes it possible to consider both healthy people and those who are unwell. In so doing, these databases help establish reliable links between certain distinct genetic features and the risk of developing a given disease and also enable us to understand the genetic characteristics of a natural resistance to common ailments such as heart disease, cancer or type-2 diabetes.

Contributing to a *population* database can therefore become a way of collaborating in the struggle to combat diseases with a genetic component.

What is CART@GENE?

CART@GENE is a genetic and demographic database project that aims to study the characteristics of the Québec population. Currently at the preliminary stage of seeking funding, the project proposes to gather some 50,000 blood samples from members of the general public, with these samples to be genetically representative of Québec's 7.4 million residents.

Cart@gene is one in a series of far-reaching human genome projects. Like the other major databases throughout the world, its objectives are as follows: to discover the genetic origins of complex diseases, identify genes that protect us against common diseases, orient research toward new treatments and, based on the regional distribution of various defective genes, help improve the allocation of health-care resources across Québec.

What kinds of question are raised by this sort of project?

The objectives being pursued by the designers of HUGE databanks are highly praiseworthy in themselves from the standpoint of research. However, we have to ask ourselves what other uses could be made of the private, confidential information that these databases contain. We all know very well that no computer system is completely safe from hackers and that genetic information, once divulged, could lead to the marginalizing of certain individuals (carriers of genes who are prone to mental illness, for example) or groups. This information could also become a factor leading to discrimination on the level of employment, insurance, or mortgage loans, adversely affecting those whose genes may reveal their predisposition to a disease that could curtail their expectancy of a normal working life.



LEGITIMACY AND TRANSPARENCY

... YOURSELF AS PATIENT

What's the point of human genetic databases?

Whereas yesterday's scientists could only work with the observable characteristics of people suffering from a given disease so as to seek its causes, their counterparts of today are truly spoiled! They have access to the "intimacy" of human cells, i.e. to the 30,000 or so human genes that make up our 23 pairs of chromosomes. So it's not surprising that accessing genetic databases has become a key priority for researchers since these HUGE databanks very often constitute their main "work tool." If ever, one day, you should happen to suffer from an ailment whose causes are presumed to be genetic in nature – breast cancer, asthma or schizophrenia, to name only a few – you could very well be asked to participate in creating or supplying a so-called sectorial HUGE database.



What's a HUGE databank really like?

HUGE databanks come in all sorts of shapes and sizes. For instance, certain databases contain over 10,000 different DNA samples, while others come in the form of collections of cell lines: since each cell is the carrier of a person's entire genetic baggage, these banks of cells can easily become HUGE. Another example is a brain bank made up of over 1,000 brains from donors, now deceased. Each cell of every one of these brains is also the carrier of all the donor's genetic baggage!

How many HUGE databanks are there in Québec?

For the moment, it is impossible to complete a portrait of the entire database family or even assess the scope of the phenomenon across Québec. As a matter of fact, most of these HUGE databanks can operate and establish objectives without accountability, monitoring or evaluation constraints. Moreover, a number of researchers possess, in their laboratories, “private collections” of DNA or of tissues, developed over the years, that they do not consider genetic databases and for which no records whatsoever even exist.

How could we get more of a handle on all this?

The Commission believes that all databases, public and private alike, constituted for the purpose of collecting genetic information, should be subject to mandatory registration. It would also like to see a public registry that would list all databases that may contain genetic information. Such a process would facilitate supervision and monitoring activities and help protect the interests of each and every citizen.

In the meantime, what should you do if you're asked to participate in such a HUGE databank?

Until these mechanisms are put in place, if you are asked to provide DNA or biological material for such a database, you should try to sound out those in charge so as to gauge the legitimacy of their project, in other words, to pin down their purpose. You can base your judgment on two points: the project's ends and its necessity. For instance, you could ask: What's the purpose of the proposed database? What sorts of results are expected? Does such a HUGE databank really have to be established for these objectives to be reached? How is my personal information going to be protected?

AUTONOMY

... YOURSELF AS RESEARCH VOLUNTEER

Free and informed consent

"Wanted: persons 18 to 45 years of age, non smokers or light smokers only, for two stays in a clinic, with the possibility of two call-backs."

Offers of this sort, which we regularly hear on the radio and see posted in the subway, on the bus or in the pages of our daily papers, have become all too familiar. They invite us not only to come and test certain new types of medication in return for financial compensation, but also to contribute our DNA to genetic research. Here then is a brief survey of your rights when you, as a volunteer and research subject, agree to give your tissues or DNA to a HUGE databank. But you must also realize that there are possible consequences for the future.

When I agree to participate in genetic database research, just what is the nature of my consent?

When you agree to collaborate with such a database, you allow biological material sampling (tissues or cells) to be carried out for research purposes on your person. Your consent also makes it possible for researchers to obtain information about your health, your family history, and sometimes even your way of life.



In order to be considered valid, your consent must be free, informed and explicit.

Free: You must give your consent without having undergone any pressure, in particular from the research team.

Informed: The nature and goal of the research, as well as the advantages and risks it entails, must have been clearly explained to you from the word go.

Explicit: You will have been asked to sign a mandatory consent form providing an overview of the project and disclosing the use to be made of the information.

*In the particular context of a HUGE databank, one thing above all else has to be taken into account: **the scope of your consent.***

Why? Because the samples to be taken from your body may be kept in the databases for long periods of time and could therefore be used for all kinds of different research projects. But your consent applies to only one such project. To put it bluntly, this means that the samples in question should not be made available for other uses, for instance for purposes of a police inquiry. Nevertheless, the consent form may contain provisions stipulating that you may be contacted at a later date when another research protocol will be presented and explained to you. At this point, you will be free to accept the offer or refuse to participate.

Please realize that in taking this course of action, you in no way limit or hinder the research process; you simply ensure that your autonomy and private life continue to be protected.

The Web site provides an example, among many others, of a typical consent form developed by the Network of Applied Genetic Medicine of the FRSQ (Québec health research fund): <http://www.rmga.qc.ca/en/index.htm>.

EQUITY

... YOURSELF AS AN INSURED PARTY, A BORROWER OR A WORKER

The use of genetic information in fields other than health

Concerning genetics, there are real risks of discrimination and stigmatization once research results are known and linked to persons or communities. For instance, the symptoms of certain diseases, such as *myotonic muscular dystrophy* (*Steinert's disease*), appear only after the age of 40, yet a test can identify carriers of the defective gene at birth, or even earlier.

Who could do the discriminating?

Three groups of social players may engage in what is called a "secondary use of data": **insurers, employers and financial institutions**. In other words, they can use the information concerning your genetic health to their advantage for selection purposes.

Insurers. For the time being, they rely, above all, on your current state of health, your family health history, and a certain number of your lifestyle habits. Nevertheless, they may wish to go further and use the information from your genome (your whole set of genes) to more accurately assess the degree of risk you represent for them.



Employers. They could ask to have access to your genetic information before offering you a job, on the pretext that a person like yourself, possessing one genetic characteristic or another, may jeopardize workplace security, or his or her own personal safety.

Financial institutions. They may wish to learn your genetic “profile” in order to evaluate your capacity, as a borrower, to repay any eventual loans that you may be granted. Should they discover that you possess a gene that could curtail your life expectancy, they could consider that your capacity to repay constitutes a risk for the institution.

What’s going to happen?

There are not yet any legislative markers to limit such practices. Moreover, since the predictive value and reliability of genetic tests are not yet absolute, the Commission recommends that the Québec government impose a five-year moratorium on the use, by employers, insurers and financial institutions, of information provided by genetic tests, in this way expressing its wish that citizens and society be given sufficient time to carefully evaluate the situation.

We should mention here articles 2 a) and b) of the *Universal Declaration on the Human Genome and Human Rights* (UNESCO):

- a) “Everyone has a right to respect for their dignity and their rights regardless of their genetic characteristics”; and
- b) “That dignity makes it imperative not to reduce individuals to their genetic characteristics and to respect their uniqueness and diversity.”



RESPONSABILITY

... YOURSELF AS A PARENT

Genetics and reproduction

Never before in human history have parents had at their disposal so many ways to learn about ailments that could affect their unborn child. These means most often take the form of genetic tests designed, among other reasons, to identify genetic imperfections that may cause various diseases.* Researchers wish to create genetic databases in part as a means of developing such tests.

What genetic tests are currently available for future parents?

Genetic tests may be categorized according to when and how they are intended to be used. Among others there are:

diagnostic tests;
presymptomatic tests;
predictive and risk-assessment tests;
carrier-identification tests; and
prenatal tests.



* Over 5,000 genetic diseases have, it seems, already been recorded. Some are monogenetic, in other words due to a single defective gene (such as mucoviscidosis, better known in Québec under the name of cystic fibrosis); others are multifactorial, i.e. caused by an interaction between the environment and a person's genetic heritage (this is the case for cancer, diabetes and some forms of mental illness).

The **prenatal diagnostic test**, for instance, makes it possible to determine whether the fetus is already suffering from a genetic disorder or is the carrier of a gene that increases the risk of developing a disease of genetic origin.

The **presymptomatic test**, for its part, is carried out on healthy individuals to determine whether they are carriers of a genetic imperfection – also known as a mutation – that may trigger the development of a disease of genetic origin.

The **risk-assessment test**, for its part, is used to determine whether individuals are carriers of a mutation that could make them prone to developing a given disease if exposed to certain dangerous substances found in the environment.

How can we find our way in such a labyrinth?

Nowadays, in certain hospital centres, a new type of professional has in fact been hired to help people in this position. Known as **genetic counselors**, they are often trained nurses who then become specialists by completing a master's degree in genetic counseling. They are therefore able to provide information about the positive and negative impacts (benefits and risks) of every one of these tests according to each person's unique, specific genetic heritage. However, since there are as yet few such counselors in the hospital system, more often than not in today's world one must turn to one's own doctor.

Isn't there a risk that one day there will be a selection process designed to create the "perfect baby" ?

Only too often, researchers, journalists and common citizens alike tend to link everything in the human realm to genes, not only the state of one's health, but also various aspects of personality, such as violence, sexual orientation, intelligence and even one's propensity for happiness. This is what some people call the "genetic tidal wave," an ideology based on a simplistic view of genetics, whose proponents talk as if all diseases were of genetic origin only, whereas we know that most have a vast array of causes.

INTELLECTUAL PROPERTY

... YOURSELF AS RESEARCHER-ENTREPRENEUR*

Genetics and the business world

Private enterprise is an essential engine of scientific progress in the field of genetics. Over the last several years, the new research perspectives offered by genetics have propelled the biotechnologies sector to the top of the stock markets. The demand for qualified personnel has increased accordingly, so much so that the next wave of young genetic researcher-entrepreneurs will probably create new HUGE databanks as well. Of course this is a touchy subject from a social standpoint, and all sorts of interests are at play, even contradictory ones. Here is how the situation will look in the short term ...

What is meant by “ contradictory interests ? ”

For a while now, the changing context of scientific research, especially the increasing number of public-private partnerships, has been a focus of reflection, and understandably so. A concrete example? The simple **dissemination of research results**, a fundamental practice in public research, is at odds with **industrial secrecy**, a crucial principle for the private sector. Of course each of the players involved appeals to values that are totally legitimate in themselves but may diverge ethically speaking, in other words as to what approach to adopt based on the goals being pursued: to share the information in order to help the research process move ahead or to keep the information confidential with an eye to marketing the results.



* Researcher-entrepreneurs are researchers who not only carry out research but also undertake to market the knowledge that they have developed.

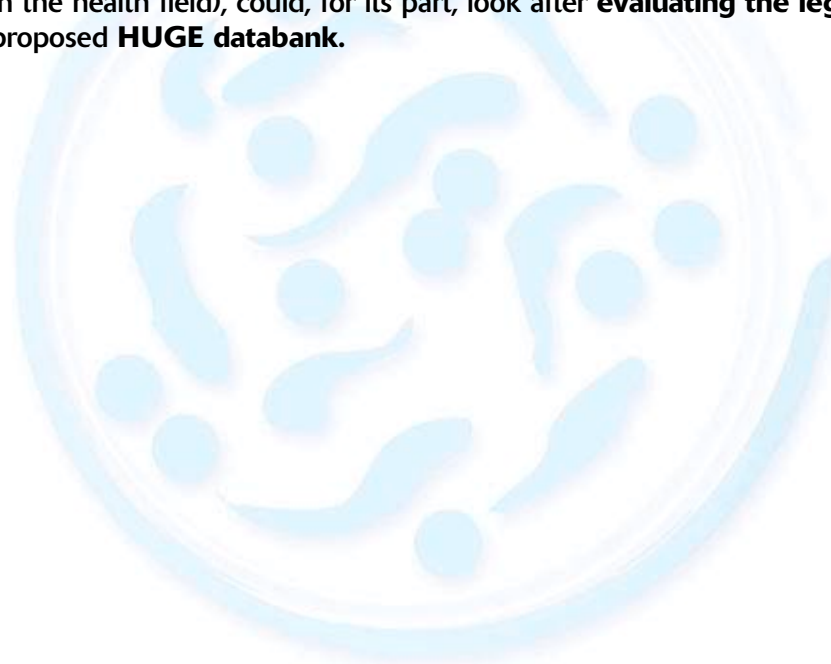
Property: a very thorny problem

A debate is currently raging throughout the world concerning the issue of genetic patenting and the technologies resulting from it (screening tests, for example). Companies argue that in order to survive they need to hold exclusive rights to this type of information and thus that they should be the only beneficiaries of the related marketing possibilities, whereas groups of researchers, for their part, consider that material coming from the human body belongs to the public realm, i.e. to each and every individual.

The process of reflection must continue. The issue of intellectual property, especially as it affects the granting of patents for living things (for example, the development of model animals for research purposes, such as the oncological mouse) is far from being resolved. Canada, like so many other countries, is currently attempting to shed some light on the subject.

How could the government regulate the creation of HUGE databanks

In its position statement, the CEST suggests one path to follow: it recommends that the government change the mandate of the CAI (access to information commission) for the short term, putting it in charge of setting up and managing a **mandatory registry** of genetic databases. This registry would be required to catalogue all existing databases in Québec. Another governmental organization, the AETMIS (agency for the evaluation of technologies and modes of assessment in the health field), could, for its part, look after **evaluating the legitimacy** of any proposed **HUGE databank**.



DEMOCRACY

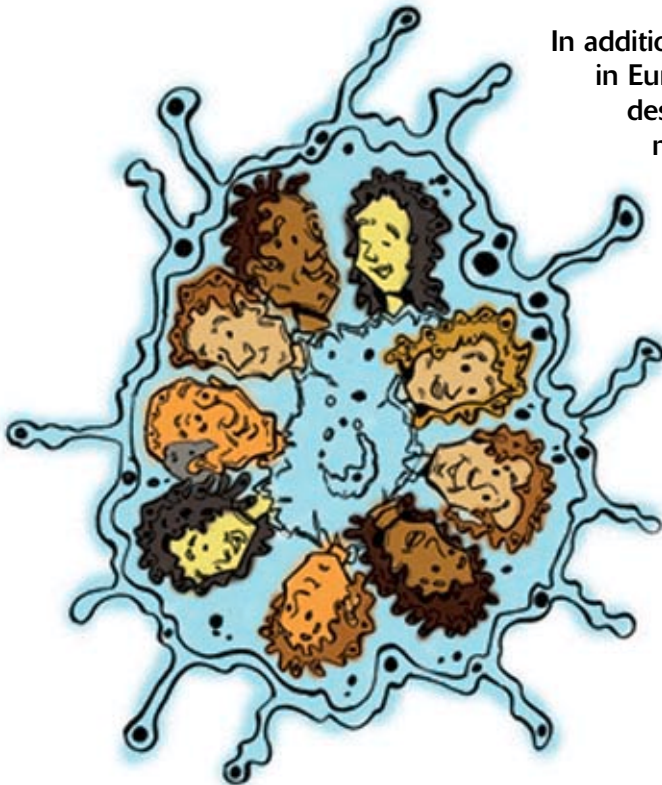
... YOURSELF AS A CITIZEN

Genetic databases: a choice of society

Given the importance of genetic information and its storage in databases for research purposes, the Commission believes that Québec cannot continue further down this road without input from the general public. This is only right and proper, after all, for what is at issue here is the genetic profile of each and every individual – and perhaps therefore your own!

How to promote public involvement in this sort of reflection process?

There are a number of ways for members of the general public to become familiar with ethical issues of concern to them. In Great Britain, for example, where the public consultation process is more advanced with regards to **HUGE databanks**, all sorts of methods have been used. There have been regional workshops at which experts discuss with citizens and confront different points of view. "Summits" and "forums" akin to the *Youth Summit* and the *Forum on Social Development* recently held in Québec have also been organized.



In addition, a tradition has begun to take root in Europe: the holding of annual *ethics days*, designed for the general public but targeting more particularly young people and their teachers.

Would schools have a role to play?

With respect to a field such as human genetics, which may affect all citizens from an individual, family or community standpoint, the Commission considers it fundamental that no student leave school without having acquired at least a minimum amount of knowledge about the subject. Thus the CEST recommends that the minister of Education consider including basic human genetics in the high school curriculum.

What should we expect with regards to public participation in the ethical reflection concerning HUGE databanks?

As André Beauchamp, the 2001-2004 CEST chair, puts it, "above and beyond being an institution, democracy is most of all a state of mind, a practice, an experiment."

This is why political will is required to move from speeches on democracy to making democracy happen through a decision-making process involving the entire population.

André Beauchamp,
Environnement et consensus social,
Éditions l'Essentiel, 1997.

In its position statement, the Commission recommends that the Government of Québec begin public consultations as soon as possible, especially by way of surveys (preceded by the dissemination of information on the subject) and calls for submissions from interest groups and community organizations. Citizens would also be called upon to make their views known in more detail, if they so desire, for instance by regular mail or E-mail.

And in order for the consultation process to be more than another dead-end political operation, the Commission deems it essential that the results of the process lead to the drafting of a **Québec policy statement on genetic information**, which would take into account both the Commission's position statement and input from the general public.

*This edition was produced by the
Commission de l'éthique de la science et de la technologie (CEST)*

Coordination: Diane Duquet, coordinator, CEST

Content edition: Emmanuelle Trottier, ethics counselor, CEST

Communications Coordination: Katerine Hamel, communications consultant, CEST

Writer: Luc Dupont, scientific journalist

Translation: ABACA Traduction Enr.

Graphic design: Vallières Communication Créativité enr.

Secretariat: Annie St-Hilaire, CEST

HUman **GE**netic Databases

“A HUGE deal!”



To learn more don't forget:
www.ethique.gouv.qc.ca

Commission
de l'éthique
de la science
et de la technologie

Québec 