"PERSONALIZED" HEALTH CARE:

prudence and guidance

COMMISSION DE L'ÉTHIQUE
EN SCIENCE ET EN TECHNOLOGIE

Québec

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For ease of reading, the masculine gender is used throughout
the text without any discriminatory intent.

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List of initialisms and acronyms

ACFAS:	Association francophone pour le savoir [Francophone association for knowledge]
CAI:	Commission d'accès à l'information du Québec [Québec commission for access to information]
CEST:	Commission de l'éthique en science et en technologie [Commission for ethics in science and technology]
CIHR:	Canada Institutes of Health Research
CIRANO:	Centre interuniversitaire de recherche en analyse des organisations [Interuniversity centre for research on the analysis of organizations]
DNA:	Deoxyribonucleic acid
FMG:	Family medicine group
INESSS:	Institut national d'excellence en santé et en services sociaux [National institute for excellence in health and social services]
MSSS:	Ministère de la Santé et des Services sociaux [Ministry of health and social services]
PHC:	"Personalized" health care
QHR:	Québec Health Record
QNPHC:	Quebec Network for Personalized Health Care
UQAM:	Université du Québec à Montréal

Executive summary

A "personalized" health care approach in medicine would allow the diagnosis, treatment and prevention of diseases with a genetic component to be tailored to each individual. However, contrary to what the term *personalized* suggests, the person is not at the centre of this new approach, but rather his genetic make-up.

Although it gives hope to people who are sick or may have – or pass on – a serious disease with a genetic component, "personalized" health care involves ethical issues that must be taken into consideration. For instance, it may have a significant effect on the quality of the clinical relationship and medical practice. It also raises concerns about the possibilities of genetic discrimination by employers and insurance companies. Furthermore, since "personalized" health care will generate a large volume of data that will circulate among many people, it is important to ensure the confidentiality of this information. Lastly, these technological advances will put considerable pressure on the limited financial resources of Québec's health care system.

The Commission has identified possible solutions and has made a number of recommendations to the people, organizations and decision makers concerned:

- Promote the development of evidence-based "personalized" health care;
- In clinical practice, use only genetic tests with proven scientific validity and clinical utility;
- Inform the population about the limitations, reliability and dangers of genetic tests;
- Ensure health professionals are provided with appropriate genetics training;
- Reaffirm the criterion of clinical utility and show the importance of quality genetic information in clinical decision making;
- Encourage information sharing with family members while respecting the patient's wishes and professional secrecy;
- Determine proven efficacy thresholds and increase post-market surveillance of new drugs;
- Negotiate an agreement with private insurers on the use of genetic tests;
- Promote the development of drugs for therapeutic orphans;
- Ensure the population has access to basic care;
- Adopt a strategy for implementing "personalized" health care that will maximize its health benefits;
- Provide a framework for information sharing that will ensure the confidentiality of data;
- Recognize access to care as a guiding principle of the strategy for implementing "personalized" health care.

Summary

Since 2010, the Government of Québec has been engaged and made significant investments in a strategy to promote "personalized" health care (PHC).

The purpose of this position statement is not to express an opinion on PHC, but to ensure that its integration into Québec's health care system respects our society's values and is adapted to the State's limited resources. The Commission believes that accessible health care, which the population has a right to expect, must be maintained. It insists on the importance of the criteria of scientific validity and clinical utility that new technologies must satisfy before being introduced into the health care system.

It believes that PHC will not revolutionize medical practice, but – thanks to genetic data – will allow more accurate diagnoses and more effective targeting of treatment.

The following definition, proposed by the Quebec Network for Personalized Health Care (QNPHC), served as the basis for the Commission's ethical analysis of PHC:

> « Personalized health care relies on more comprehensive information and a better knowledge of patients, their genetic profile, environment, behaviour, medical history and certain metabolic characteristics to identify the most appropriate methods of treatment and therapeutic and preventive solutions for each group of individuals profiled (QNPHC, 2011). [Translated from the French.] »

A guiding principle Health is of core importance to human well-being. The development of new technologies and new therapies must be encouraged if it translates into better health care for the population at the best possible cost.			
A vision to share Improve health care, in particular through more accurate diagnoses, better targeted drugs and a concomitant reduction of side effects	**A phenomenon to understand** **A characteristic of PHC** The probabilistic nature of genetic information **Challenges to overcome** - Ensure innovations introduced into the health care system are based on evidence - Ensure the scientific validity and clinical utility of PHC applications - Ensure health professionals are able to interpret genetic tests correctly	**An ethical framework to support decision making** **A central principle to respect** Accessible health care for all based on individual needs **Values to promote** - Autonomy - Protection of privacy - Equity and justice - Solidarity towards and protection of vulnerable people - Health, safety and well-being - Freedom - Confidentiality	**Issues that must be addressed as a priority** - Data quality and patient safety - The repercussions on the clinical relationship and medical practice - The social effects (discrimination, stigmatization, orphan phenomenon) - Data management - The management of Québec's health care system resources

The priority issues and recommendations

I- Data quality and patient safety

The Commission considers that the first issue – both scientific and ethical – is the quality of evidence on the safety and efficacy of PHC interventions. The main challenge in producing this type of data for PHC is related to the fact that a much smaller number of patients is targeted by each intervention. Interventions associated with PHC are usually introduced in clinical practice as "innovative", experimental or pilot interventions without really knowing the beneficial or harmful effects… and sometimes without their being part of properly regulated research projects. Thus patients are exposed to interventions whose advantages, disadvantages and risks are not yet known.

The importance of evidence

A growing number of scientists question the quality of data supporting PHC interventions. Firstly, because to be considered conclusive, data must be corroborated by several independent teams and come from well-designed studies, randomized controlled trials and meta-analyses. A systematic review of several independent and scientifically sound studies must be conducted before determining that safety and efficacy data for a health care intervention is definitive.

Secondly, in a context where it is difficult to recruit the very large cohorts needed to produce evidence in accordance with applicable standards, new high-quality study designs must be developed to demonstrate the efficacy of very specific interventions. Reviewing the level of evidence required to conclude that an intervention has clinical utility is therefore also an important issue.

The clinical trial model

By identifying patients more likely to benefit from a particular drug, pharmacogenomics would allow smaller cohorts to be used in clinical trials, thereby shortening their duration. These changes would allow promising drugs to be brought to market more quickly and reduce costs for pharmaceutical companies. However, the smaller the size of the study population, the greater the risk that some side effects will go undetected.

The validity of genetic tests

The predictive power and clinical value of genetic tests have not yet been clearly proven. This is particularly true for tests available online without the involvement of a health care provider. Furthermore, companion tests and screening tests are not foolproof; there will be a certain number of false positives due to the misinterpretation of tests.

This means that people who should not have access to a drug might nonetheless take it and experience side effects that were not detected during clinical trials.

The reliability of online direct-to-consumer tests can also be questioned. Some have no recognized scientific validity. The sample may have been contaminated during collection. Sample quality may also be compromised by transport time and conditions.

Given that the implementation of PHC must be based on evidence and that the genetic tests available on the market must have demonstrated scientific validity and clinical utility, the Commission recommends:

Recommendation no. 1

That research funding and regulatory bodies, researchers and the Institut national d'excellence en santé et en services sociaux (INESSS) reaffirm the principle of clinical utility and scientific validity in clinical decision making to show how this information can improve the quality of life and longevity of patients.

Given that PHC could change the design of clinical trials and therefore involve risks for the population, the Commission recommends:

Recommendation no. 2

That the Minister of Health and Social Services request that its federal counterpart determine, in a transparent manner, efficacy thresholds and ensure that the same exacting criteria apply to new clinical trial designs;

That the Minister ask Health Canada to increase the post-market surveillance of drugs used in "personalized" health care in order to monitor their side effects.

Given the risks of the misinterpretation of genetic tests and their effects on citizens' physical and psychological health, the Commission recommends:

Recommendation no. 3

That the Minister of Health and Social Services, together with the professional orders concerned, inform the population of the limitations and reliability of genetic tests and the dangers associated with their improper use and misinterpretation;

That the professional orders ensure that their members are aware of these limitations and the potential health risks of genetic tests;

That the Office de la protection du consommateur [Office of consumer protection] examine advertising for direct-to-consumer tests sold in Québec to ensure that citizens are adequately protected.

II- The repercussions on the clinical relationship and medical practice

The second issue is the quality of the health professional–patient relationship which is a vital component in the provision of health care. This relationship does not involve simply sharing information, but is based on a genuine bond of trust and on obtaining information that will allow the health professional to exercise his clinical judgement (Van Winsberghe and Gastmans, 2009). Mutual understanding and collaboration between the patient and the professional are crucial to ensure the quality of care and allow the patient to be autonomous. This relationship therefore embodies values that must be upheld.

The changing clinical relationship

It is increasingly clear that the shift towards "personalized" medicine will change the clinical relationship. The increase in the number of care providers and the diversity of expertise in health care teams along with shared decision making between professionals and patients require a high degree of coordination. The physician's role is transformed and the clinical relationship becomes technical.

The growing complexity of medical practice

The advent of PHC will have significant repercussions on the work of health professionals, especially physicians. Professionals now have access to new diagnostic and screening tools that generate a large volume of data. In some cases, medical decision making will be improved, while also becoming more complex.

The competence of health professionals

PHC will require special expertise on the part of health professionals. They will have to master a large volume of complex data. They will also have to be able to transmit this information to the patient properly and possibly assist the latter in making a decision.

Genetic counselling

Advances in molecular genetics make it possible to screen for a growing number of hereditary diseases or conditions even though treatments are not always available. In fact, more genetic diseases can be screened for today than can be treated. Consequently, knowing without being able to do anything gives rise to ethical problems: appropriate genetic counselling is therefore essential.

An increasing number of genetic counsellors are consulted by future parents who ask them to include PHC tests in their practice. Sometimes these counsellors must deal with requests that go beyond their area of expertise. Training programs will have to be adapted to take this new reality into account.

Given that health professionals must be adequately trained in genetics to appropriately counsel, inform and treat patients with a disease with a genetic component, the Commission recommends:

Recommendation no. 4

That professional orders and universities in Québec include adequate genetics training in their curricula for future health professionals;

That professional orders offer their members continuing education in genetics;

That the Office des professions [Québec office of professions] regulate the profession of genetics counsellor.

The patient's increased responsibility for his health

The preventive approach associated with PHC may increase the importance given to the fact that individuals are responsible for their own health. While this may be true sometimes, it is not always so. Other health determinants involved in the correlation between a person's life situation and his health must be taken into consideration.

Conversely, since genetic health determinants are central to PHC, PHC could also have the effect of taking away responsibility. If people know they are – or are not – genetically predisposed to a particular disease, they might make poor choices, believing that they will ultimately have little impact on their future health.

The patient as partner

Beyond individual responsibility, patient empowerment also means that the patient's role is evolving. From being minimally informed, he is becoming an active participant, an informed collaborator engaged in participatory medicine.

We must not, however, overlook the fact that there is an asymmetry between those making decisions, i.e., the patient and care providers, with respect to literacy, standard of living, etc. The patient-as-partner approach assumes that the patient is also an expert, which is not necessarily the case. In addition, many patients prefer to defer to their physician's good judgement. While the patient-as-partner approach is certainly interesting, it seems difficult to bring into widespread use.

The complex and sensitive nature of genetic information

The probabilistic nature of genetic information makes it complex to interpret. Thus there is not necessarily a direct link between the test result and the choice of treatment. Genetic information can also be sensitive, since it concerns traits that are specific to each individual. The information obtained can sometimes be used to predict the course of a person's physical condition and, consequently, have psychological and social repercussions.

Incidental findings

Some markers used in genetic tests to detect a specific gene in an individual may be associated with patient – or disease – characteristics other than those targeted by the test.

Familial genetic traits

Genetic testing can detect a hereditary mutation in a person that may have implications for other family members. Some patients prefer not to disclose this information to their relatives, which puts the physician in a delicate position where he must choose between respecting professional secrecy and his patient's privacy and his obligation to inform people about possible health risks.

Given that genetic counselling is not regulated, the Commission recommends:

Recommendation no. 5

That physicians and genetic counsellors encourage information sharing with family members, while taking into consideration that:

1) The patient's wishes and his right to know or not know information concerning him must be respected;
2) As is the practice under French bioethics legislation, should a patient not wish to inform his relatives, a letter must be written and given to family members by the physician to inform them that medical information of a familial nature may be of relevance to them and invite them to seek genetic counselling. The letter would not disclose the name of the person who had the test or the abnormality in question and the associated risks.

The issue of consent

PHC will exacerbate problems with consent, both in clinical practice and research. Since genetic testing may provide much more information than was initially sought, informed consent becomes all the more important. The patient must also be aware of the probabilistic nature of the information that will be obtained and that may make treatment decisions significantly more complex.

III- The social effects

The third issue to consider is that in addition to individual and relational consequences, PHC will probably have an impact on a societal level, in particular with respect to the risks of genetic discrimination and the phenomenon of new therapeutic orphans.

Genetic discrimination

The development of PHC could lead to situations of exclusion, such as discrimination, stigmatization and barriers to access to health care, which could increase social inequities. The Commission focused on three situations, namely, employment discrimination, insurance coverage by private insurers and immigration.

Employment

With respect to employment, it is feared that if an employer obtains genetic information about an applicant, the latter will be refused a job, a promotion, etc. Rights, duties and basic values such as access to employment, employee health protection and protection of the public would then be at stake. However, these fears appear to be hypothetical. Moreover, while Canadian legislation does not specifically protect people against genetic discrimination, it is written in sufficiently broad terms to encompass this type of discrimination.

Insurance coverage by private insurers

In Canada, insurers do not require genetic testing. However, advances in genomics could result in their wanting to know the genetic profile of their clients in order to determine insurance premiums. The Commission is afraid that this will lead to discrimination, with some people denied insurance coverage or charged excessive premiums based on their genetic characteristics.

In Québec, the main concerns with respect to private insurance relate to life insurance and disability insurance.

Given that every person should have access to basic insurance coverage, the Commission recommends:

Recommendation no. 6

That the Government of Québec negotiate an agreement with private insurers on the use of genetic tests based, in particular, on the model developed in the United Kingdom.

Immigration

Most countries are interested in the health of immigration applicants. Medical examinations are one of the steps applicants must follow to be admitted to Canada. Due to the sensitive nature of the health information collected, whose use may be seen as a possible source of discrimination or invasion of individual privacy, the utmost caution and compliance with principles of consent and confidentiality are required.

A possible increase in discrimination towards certain communities

Communities that are the subject of pharmacogenetics and pharmacogenomics research may be exposed to substantial risks should the findings establish a link between an ethnic or regional community and a high non-response rate to a particular treatment or susceptibility to a disease, for example. Communities or groups could then be discriminated against based on ethnoracial criteria.

The problem of therapeutic orphans

The use of "personalized" drugs could give rise to "therapeutic orphan" groups if, for example, existing treatments are found to be ineffective or unsafe for individuals with a particular genotype or if pharmaceutical companies consider the target population "too small" to produce a drug for ("orphan genotype").

Given the justice and equity issues raised by the development of PHC, the Commission recommends:

Recommendation no. 7

That the Minister of Health and Social Services request that its federal counterpart adopt new regulations similar to those that regulate orphan diseases to promote the development of drugs for therapeutic orphans;

That the Fonds de recherche du Québec [Québec research fund] and other funding bodies set aside a portion of their budget for the creation of programs targeting therapeutic orphans.

IV- Data management

The fourth issue concerns the significant volume of sensitive information that PHC will generate and that will circulate among many people. The management of this data raises ethical issues, in particular with respect to the protection of confidentiality, which will be difficult to guarantee. Other social protection measures will have to be taken to prevent stigmatization. Without being alarmist, greater transparency will be needed in this area and the public will have to be clearly informed about the limits of confidentiality protection for medical information.

The nature and volume of data

The volume of stored genetic data will grow exponentially as genetic testing and sequencing become common practice. The main risk associated with this type of technology is related to data protection.

The storage, sharing and exchange of personal information

In clinical practice

Data from genetic tests will, in all likelihood, be entered in the electronic health record that various stakeholders in the health network will have access to. Much of the information is coded and various procedures to ensure its security and confidentiality are being put in place to protect patient privacy.

However, with the deployment of the Québec Health Record (QHR), several types of records (in electronic or paper format) are kept together in the different health care facilities (family medicine groups [FMG], clinics, hospitals, pharmacies). PHC will add further data and exacerbate the tension between data sharing and protection of confidentiality.

In research

At present, genetic data and the material it is obtained from are stored in institutional databanks, that is, in hospitals where research is conducted and in banks managed by pharmaceutical companies. The centralization of data in population-based banks raises significant ethical concerns.

Data is currently being transferred between public and private institutions on a national and international level. Databanks created using public research funds are open source, unlike private databanks. This leads to a clash of values between research studies depending on whether they are publicly or privately funded.

Consent in research

Furthermore, difficulties ensuring subject anonymity and anticipating future research at the time of consent call into question the principles governing consent in research and make data management more complex.

The narrowing of the interface between clinical practice and research is of particular concern to the Commission. The Commission would like to remind clinicians that research records must be kept strictly separate from clinical records and that tests that have not yet been validated should never be documented in a patient's clinical record.

Given the sensitive nature of genetic data and the increasing volume of personal health information, the Commission recommends:

Recommendation no. 8

That the Commission d'accès à l'information (CAI) provide a proper framework for information sharing between clinical practice and research as well as for the interface between the two in order to maintain the confidentiality of data while allowing advances in the knowledge required to improve individual and population health.

V- The management of Québec's limited health care system resources

The final issue concerns the health care system which, with finite resources, must meet the needs and demands of a large number of people. It is therefore important to be judicious when allocating resources so that these needs can be met in the best way possible (CMPA, 2007). Suboptimal access to advanced technologies could mean that a segment of the population is deprived of basic care.

The Québec State's financial responsibility

The portion of the budget allocated to health is growing rapidly: it could account for nearly 70% of total government revenues in the next 20 years. The structural increase in costs is thought to be the most important factor in this increase. Consequently, to stabilize these costs, the system's efficiency must be improved.

The cost of "personalized" drugs

According to some experts, tailor-made drugs will be more expensive than traditional drugs despite the reduction in clinical trial costs (Rothstein and Epps, 2001). In this context, the conditions under which "personalized" drugs will be reimbursed by the public drug insurance plan will have to be defined.

Cost-effectiveness analyses

Considerations of justice and equity must be set alongside the criterion of cost effectiveness to ensure that everyone has access to the health care they need.

However, there will always be a tension between the collective interest and the individual interest. One way or another, limits will have to be set. Yet, whatever these limits are – irrespective of whether they significantly restrict access and reimbursement or make them available to everyone –, they will always reflect a special balance between these interests.

The need to make choices and set priorities

With the advent of PHC, decision makers will have to make concrete choices regarding the allocation of health care system resources. This will require reflection as to what constitutes an ethically optimal reconciliation of the values and interests at stake in order to guide decision making on access to drugs and pharmacogenomic tests and their coverage. In this regard, there are two opposing visions:

- There are inequities in the health care system. Everyone must have access to a minimum level of health care. The high cost of PHC could exacerbate inequities and the pressure on health care budgets;
- PHC could lead to savings in the billions of dollars spent on health care for the treatment of adverse drug reactions, promoting better management of the health care system for the common good. According to this vision, PHC must be funded.

Making choices

In public health care systems, the volume-based approach is often preferred, for it is considered more egalitarian than tariff setting. However, it has certain drawbacks. Firstly, it usually leads to waiting lists. Secondly, the possibility that some people may circumvent these waiting lists by turning to the private sector or going abroad to obtain care may lead to two-tier medicine. Lastly, the volume-based approach, if it is to be transparent and not susceptible to manipulation, necessarily relies on bureaucracy, that is, on the formulation and administration of strict, specific and impersonal rules that determine how resources will be allocated to different needs.

The management of PHC using a volume-based approach could very well give rise to more bureaucracy. In particular, we will have to determine which level of care will apply to which probability threshold, and which procedures will be covered at disease probability thresholds of 30, 50 and 70%. This bureaucratic stranglehold on health care would then lead to the paradox of so-called "personalized" medicine, where access to care is nonetheless wholly determined by fixed and impersonal rules.

Making choices raises a certain number of governance issues which create barriers to the implementation of effective health care innovations. The introduction of PHC will increase pressure on the system's financial viability if these problems are not resolved.

Given the State's limited financial resources and the lack of data to determine and anticipate actual case costs, the Commission recommends:

Recommendation no. 9

That the Government of Québec adopt a strategy for implementing "personalized" health care that will maximize its health benefits, which means finding a way to carefully assess the costs;

That access to care be recognized as a guiding principle of this strategy.

Glossary

Allele: Each of the different possible forms of the same gene. Alleles occupy the same locus on a pair of homologous chromosomes. They govern different aspects of the same character, whose expression depends on the relationship of dominant and recessive alleles (CEST, 2010).

Analytical validity: An indicator of a test's ability to measure the desired property or characteristic (INESSS, *Assessment Process and Criteria*).

Biomarker: A characteristic that is objectively measured and evaluated as an indicator of normal biological processes, pathogenic processes, or pharmacologic responses to a therapeutic intervention (National Institutes of Health, cited by AFSSAPS).

Clinical utility: The degree to which the results positively or negatively affect the patient (INESSS, Assessment Process and Criteria).

Clinical validity: A measure of how accurate a test is in identifying or predicting a clinical disorder (INESSS, *Assessment Process and Criteria*).

Cohort studies: Studies in which subjects are selected based on their exposure to risk factors for a disease or following treatment. Subjects are followed over a long period and compared with a group that has not been exposed to the risk factor or specific treatment [translated from the French] (Consortium national de formation en santé, 2011).

Companion test: Test intended to assist physicians in making treatment decisions for their patients. It does so by elucidating the efficacy and/or safety of a specific drug or class of drugs for a targeted patient group or subgroup. There are two main groups of companion diagnostics that include:

- tests that were developed after the drug was brought to market;
- tests that are developed at the same time as the drug is developed (AMGEN).

Evidence: A sufficient volume of sound data obtained from well-designed studies. It is a source of information that aids professionals in their clinical decision-making process. Clinical decisions are based on multiple data that must take the clinical context and the patient into account. In evidence-based medical practice, clinical decisions are based on the best available scientific evidence, clinical experience and patient consultation in order to choose the best option for the latter [translated from the French] (Sackett et al., 2000, cited by the Consortium national de formation en santé, 2011).

Genetic profile (an individual's genetic print or genotype): A genetic profile is a series of pairs of numbers called alleles for each marker or genetic system tested. A person's genetic print is unique and remains unchanged throughout his lifetime, so it can be used to identify an individual in a much more reliable way than a fingerprint) (LabGenetics website).

Genetic test: Laboratory test conducted with a view to obtaining information on specific aspects of genetic status. There are different types of genetic tests:

- diagnostic genetic tests: they are intended to diagnose a genetic disorder in a person who already has symptoms. The results may help to make choices about how to treat or manage health problems;
- predictive genetic tests: they are performed on individuals who show no symptoms as yet. They are geared to detecting genetic changes which suggest a risk of developing a disorder later in life;
- carrier tests: they are used to identify people who "carry" a mutated allele of a gene which is associated with a specific disease (e.g. cystic fibrosis). Carriers may show no signs of the disease. However, there is a risk that their children will be affected;
- pharmacogenomic tests: see definition below (Council of Europe, 2012).

Genome test: In comparison to a genetic test, a genome test examines the whole genome; it examines the entire genome of an individual or a tumour rather than targeting a single gene or a number of genes.

Genomics: Genomics can be defined as a scientific discipline that studies the genome, that is, all the genetic information (the genes and other DNA sequences) contained in an individual's chromosomes [translated from the French] (CEST, 2012).

Genotype: All the genetic material carried by an individual and which constitutes his hereditary material (CEST, 2010).

Pharmacogenetic test: A pharmacogenetic test detects the presence or absence of genetic polymorphisms (variations) that influence drug response [translated from the French] (Buclin et al., 2008).

Pharmacogenetics or pharmacogenomics: According to Health Canada, these two terms are often used interchangeably and specifically refer to the role of genetics in pharmaceutical research (Health Canada). However, pharmacogenomics is considered to be a natural development or extension of advances in pharmacogenetic research insofar as pharmacogenomics is the study of the way in which genetic variation across the human genome affects drug responsiveness [translated from the French] (Meyer, 2002, cited by CEST, 2012).

Pharmacogenomic test: The opposite of a pharmacogenetic test, a pharmacogenomic test assesses the effect of a drug on an individual's genome [translated from the French] (Bourel and Ardaillou, 2006).

Pharmacovigilance: Activity that involves recording and assessing reactions to the administration of drugs in order to ensure their safe use under normal conditions of use [translated from the French] (Grand dictionnaire terminologique).

Randomized studies: Studies in which subjects that are likely to benefit from a treatment are selected before being randomly assigned to either of two groups. The first group is given the study treatment while the second is given the usual treatment or a placebo [translated from the French] (Consortium national de formation en santé, 2011).

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QUEBEC NETWORK FOR PERSONALIZED HEALTHCARE – QNPHC: http://qnphc.org/.

Appendix 1: Restrictions to the use of genetic information by insurers for life insurance underwriting[2]

Table 3: Comparative analysis of restrictions to the use of genetic information by insurers

COUNTRY	MORATORIUM	LEGISLATION	GUIDELINES	OTHER	DRAFT
AUSTRIA	No	Yes	No		
AUSTRALIA	Partial Exp.: 2005	No	Yes	A bill on genetic privacy was introduced in 1998 but has not yet been accepted.	A joint inquiry on the protection of genetic information has recently been conducted by the Australian Law Reform Commission.
BELGIUM	No	Yes	No		
BULGARIA	No	No	No	Cellule vide Ratified the Oviedo convention 01/08/03.	A constitutional amendment prohibiting genetic discrimination has been introduced in the National Assembly (summer 2002).
CANADA	Partial	No	Yes		
CHILE	No	No	Yes		
CYPRUS	No	No	No	Ratified the Oviedo convention 01/07/02.	
CZECH REPUBLIC	No	No	No	Ratified the Oviedo convention 01/10/01.	
CROATIA	No	No	No	Ratified the Oviedo convention 28/11/03.	
DENMARK	No	Yes	Yes	Ratified the Oviedo convention 01/12/99.	
ESTONIA	No	Yes	No	Ratified the Oviedo convention 01/06/02.	
FINLAND	Yes Unlimited amount Exp.: none	No	Yes		

2 Source: Lemmens, Joly and Knoppers (2004).

COUNTRY	MORATORIUM	LEGISLATION	GUIDELINES	OTHER	DRAFT
FRANCE	Yes Unlimited amount Exp.: 2004	Yes	Yes		
GERMANY	Yes Limited amount Exp.: 2006	No	Yes		A parliamentary commission has declared that insurers should not use genetic test results.
GEORGIA	No	Yes	No	Ratified the Oviedo convention 01/03/01.	
GREECE	Partial	No	Yes	Ratified the Oviedo convention 01/12/99.	
HUNGARY	No	No	Yes	Ratified the Oviedo convention 01/05/02	
ICELAND	No	No	No	A bill has been presented but has not been enacted.	
INDIA	No	No	Yes		
IRELAND	Yes Unlimited amount Some conditions are excluded Exp:2005	No	No		
ISRAËL	No	Yes	No		
ITALY	No	No	Yes		Guidelines for genetic testing to be adopted shortly by the ministry of health.
JAPAN	No	No	Yes		The Association of Life insurance Medicine of Japan has a code of practice in preparation. Ministry of health to issue guidelines. Insurers are not allowed to ask for family history information.
LITHUANIA	No	No	No	Ratified the Oviedo convention 17/10/02.	
LUXEMBOURG	No	Yes	Yes		
MOLDOVA	No	No	No	Ratified the Oviedo convention 26/11/02.	
NETHERLAND	No	Yes	Yes		
NEW ZEALAND	Partial	No	Yes		
NORWAY	No	Yes	Yes		

COUNTRY	MORATORIUM	LEGISLATION	GUIDELINES	OTHER	DRAFT
PORTUGAL	No	No	Yes	Ratified the Oviedo convention 13/08/01. Article 13 of the Portuguese Constitution could prevent insurers to discriminate on the basis of genetic test results.	A task force established by the ministry of health has prepared key guidelines addressing genetic testing.
ROMANIA	No	No	No	Ratified the Oviedo convention 01/08/01.	
SAN MARINO	No	No	No	Ratified the Oviedo convention 01/12/99.	
SINGAPORE	No	No	Yes		
SLOVAKIA	No	Yes	No	Ratified the Oviedo convention 01/12/99.	
SLOVENIA	No	No	No	Ratified the Oviedo convention 01/12/99.	A Bill is expected to be drawn up in the near future addressing human genetics.
SOUTH AFRICA	Partial	No	Yes		Insurers are not allowed to ask for family history information.
SOUTH KOREA	No	No	No		A Bill prohibiting discrimination in insurance and employment has passed through the national assembly, presidential sanction is planned for February 2005.
SWEDEN	Yes Unlimited amount Exp.: Dec. 2004	Yes	Yes		
SWITZERLAND	Yes	Yes	Yes		
TAIWAN	No	No	No		Insurers are not allowed to ask for family history information.
TURKEY	Yes	No	No		
UNITED KINGDOM	Yes Limited amount Some conditions are excluded Exp.: 2006	No	Yes		
UNITED STATES (federal government)	No	No	No		Several bills addressing genetics and insurance have been drafted, but none enacted.

Appendix 2: The Commission's consultation activities

Round tables

Round table on the scientific aspects of PHC, Montréal, March 6, 2013

EXPERTS

Michal Blazejczyk
Head of Information Technology
Pharmacogenomics Centre

Dr. Pavel Hamet
Professor of Medicine
Canada Research Chair in Predictive Genomics
Chief of the Division of Gene Medicine, CHUM
Member of the Commission de l'éthique en science et en technologie

Dr. Michèle Marchand
Physician and philosopher
Collège des médecins

Dr. Yves Robert
Secretary
Collège des médecins

Céline Lafontaine
Associate Professor
Department of Sociology
Université de Montréal

MEMBERS OF THE STEERING COMMITTEE

Françoise Guénette (by telephone)
Freelance Journalist

Bernard Sinclair-Desgagné
Full Professor
Chair of International Economics and Governance
HEC Montréal

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Notary
Secretary General of the Commission de l'éthique en science et technologie

Édith Deleury
Professor Emeritus
Faculty of Law, Université Laval
President of the Commission de l'éthique en science et en technologie

SECRETARIAT OF THE COMMISSION

Geneviève Trépanier
Ethics Consultant and Meeting Secretary

Round tables on the ethical issues associated with PHC, Montréal, June 18, August 29, December 4 and 11, 2013

EXPERTS

Johanne Castonguay
Assistant Vice-President
CIRANO

Hubert Doucet
Retired Professor
Bioethics – Université de Montréal

Yann Joly
Assistant Professor
Department of Human Genetics and the Bioethics Unit
Université McGill
Associate Researcher
CRDP – Université de Montréal

Céline Lafontaine
Associate Professor
Department of Sociology
Université de Montréal

Thérèse Leroux
Full Professor and Researcher
CRDP – Université de Montréal

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Professor Emeritus
Faculty of Law, Université Laval
President of the Commission de l'éthique en science et en technologie

SECRETARIAT OF THE COMMISSION

Geneviève Trépanier
Ethics Consultant and Meeting Secretary

Person heard before the Commission, August 26, 2013

Dr. Hubert Marcoux
Associate Professor, Department of Family Medicine and Emergency Medicine, Faculty of Medicine, Université Laval
Ethicist

Public consultations

The following speakers participated in four public consultations on PHC:

- **Dr. Pavel Hamet**: *Development of personalized health care* (in Montréal and Québec City)
- **Dr. Daniel Gaudet**: *Personalized medicine: Applications, limitations and challenges for disease prevention and management* (à Rimouski)
- **Mélanie Bourassa Forcier**: *Personalized health care (PHC): Legal and economic issues*
- **Professor Hubert Doucet**: *Personalized medicine and ethics*
- **Freelance journalist Françoise Guénette** led the discussions.

Montréal
Auditorium, Centre d'archives de Montréal, May 22, 2012, 7 p.m. to 9 p.m.

Québec City
La Margelle, Cégep de Sainte-Foy, May 29, 2012, 7 p.m. to 9 p.m.
Amphithéâtre Hydro-Québec, Université Laval, in collaboration with the AELIÉS Public Chair, November 21, 2012, 7 p.m. to 9 p.m. (*Health version 3.0: between ethics and genetics)*.

Rimouski
Amphitheatre, Cégep de Rimouski, June 12, 2012, 7 p.m. to 9 p.m.

The 2013 Acfas symposium

In collaboration with INESSS, the Commission presented an activity entitled *Personalized health care: overview, challenges and issues during the 81st Acfas symposium at Université Laval on May 9, 2013*.

SYMPOSIUM ORGANIZERS

Geneviève Trépanier, Commission de l'éthique en science et en technologie

André Jean, INESSS

FACILATOR

Bruno Leclerc, Université du Québec à Rimouski

WELCOMING ADDRESS

Mireille Mathieu, INESSS

SPEAKERS

Dr. Pavel Hamet, Université de Montréal: *The development of personalized health care*

Anne-Marie Savard, Université de Sherbrooke: *Personalized health care: legal and economic issues*

André Néron, Faculty Office for Patient Partner Expertise, Université de Montréal: *Personalized health care and the patient as partner*

Hubert Doucet, Université de Montréal: *Personalized medicine: medicine for the person?*

Dr. Stéphane P. Ahern, Université de Montréal: *Personalized health care and the evaluation of drugs: prolegomena to a debate*

Dan Cooper, INESSS: *The issues associated with the pharmacoeconomic evaluation of innovative drugs: when clinical practice, pharmacoeconomics and ethics come together!*

Paule DeBlois, RSSPQ: *Personalized health care: how can we prepare for it?*

Reiner Banken, INESSS: *Innovation, evaluation, deliberation and personalized health care*

Bernard Keating, Université Laval: *Accompanying the paradigm shift*

CLOSING ADDRESS

Édith Deleury, Commission de l'éthique en science et en technologie

"The challenges of customization" science bar

In collaboration with the Commission and with the financial support of the CIHR, UQAM's Cœur des sciences organized a science bar on October 9, 2013.

WELCOMING ADDRESS

Sophie Malavoy, Cœur des sciences

FACILITATOR

Valérie Borde, Scientific journalist and member of the Commission de l'éthique en science et en technologie

PARTICIPANTS

Dr. François Rousseau, Holder of the FRSQ/MSSS/CHUQ Research Chair in the Evaluation of Technologies and Practices in Laboratory Medicine, Université Laval

Paule De Blois, MBA, Executive Director, Quebec Network for Personalized Health Care (QNPHC)

Dr. Jacques Simard, Holder of the Canada Research Chair in Oncogenetics, Université Laval, and leader of the CIHR team on the familial risk of breast cancer

Yann Joly, Lawyer Emeritus (AdE), Barreau du Québec and Professor, Department of Human Genetics, Faculty of Medicine, McGill University and Associate Researcher, Centre de recherche en droit public, Université de Montréal

Édith Deleury, Chair of the Commission de l'éthique en science et en technologie and Professor Emeritus, Faculty of Law, Université Laval

The Commission-Jeunesse 2013

All documents related to the Commission-Jeunesse 2013 can be downloaded from the following address [available in French only]: http://www.ethique.gouv.qc.ca/fr/commissions-jeunesse/2013-soins-de-sante-personnalises.html

The following people did a critical review of the first version of the position statement

André Jean
Secretary General and Senior Ethics and Governance Consultant Institut national d'excellence en santé et en services sociaux

Dr. François Rousseau[3]
Medical biochemist specialized in molecular genetics and Professor, Department of Molecular Biology, Medical Biochemistry and Pathology, Faculty of Medicine, Université Laval

The Commission would like to thank all those who collaborated in its reflection and contributed to the content of its position statement on "personalized" health care.

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3 François Rousseau was awarded a grant by Genome Canada (Génome Québec, CIHR and partners) to conduct a study on personalized medicine. In connection with this grant, a number of private companies working in the field (QIAGEN, Life Technologies, Illumina, Ariosa Dx, Perkin Elmer) made in-kind donations (equipment, reagents) totalling approximately 1.6 million dollars.

The members of the steering committee

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Françoise Guénette
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SECRETARY GENERAL

Nicole Beaudry

4 Pavel Hamet is also President of the personalized medicine company Prognomix. The Commission would like to note that the latter did not participate in the steering committee for the position statement and provided only information of a scientific nature. He did not, at any time, influence the Commission's position on "personalized" health care.

A "personalized" health care approach in medicine would allow the diagnosis, treatment and prevention of diseases with a genetic component to be tailored to each individual. However, contrary to what the term personalized suggests, the person is not at the centre of this new approach, but rather his genetic make-up.

Although it gives hope to people who are sick or may have – or pass on – a serious disease with a genetic component, "personalized" health care involves ethical issues that must be taken into consideration. For instance, it may have a significant effect on the quality of the clinical relationship and medical practice. It also raises concerns about the possibilities of genetic discrimination by employers and insurance companies. Furthermore, since PHC will generate a large volume of data that will circulate among many people, it is important to ensure the confidentiality of this information. Lastly, these technological advances will put considerable pressure on the limited financial resources of Québec's health care system.

To be ethically acceptable, "personalized" health care must not interfere with the population's access to basic health care to meet priority needs.

In the position statement "Personalized" health care: prudence and guidance, the Commission de l'éthique en science et en technologie reports on the vision that emerged from the consultations it held and which is intended to ensure that the development of "personalized" health care will be of benefit to all.

Taking note of the challenges facing decision makers and stakeholders, the Commission makes a series of recommendations concerning various subjects, namely, the importance of evidence, the training of health professionals, the importance of the clinical validity and utility of approved genetic tests, the sharing of information with family members, the importance of pharmacovigilance and post-market surveillance of new drugs, the transfer of knowledge to the population, the use of genetic tests by insurance companies and employers, access to basic health care for all and the confidentiality of genetic data.

The full position statement (in French) as well as this English summary and the Commission's other publications are available at the following address: www.ethique.gouv.qc.ca

The mission of the Commission de l'éthique en science et en technologie is, on the one hand, to inform, raise awareness, receive opinions, stimulate reflection and organize debates on the ethical issues surrounding developments in science and technology and, on the other hand, to propose guidelines to assist stakeholders in their decision making.

Commission de l'éthique en science et en technologie
Québec