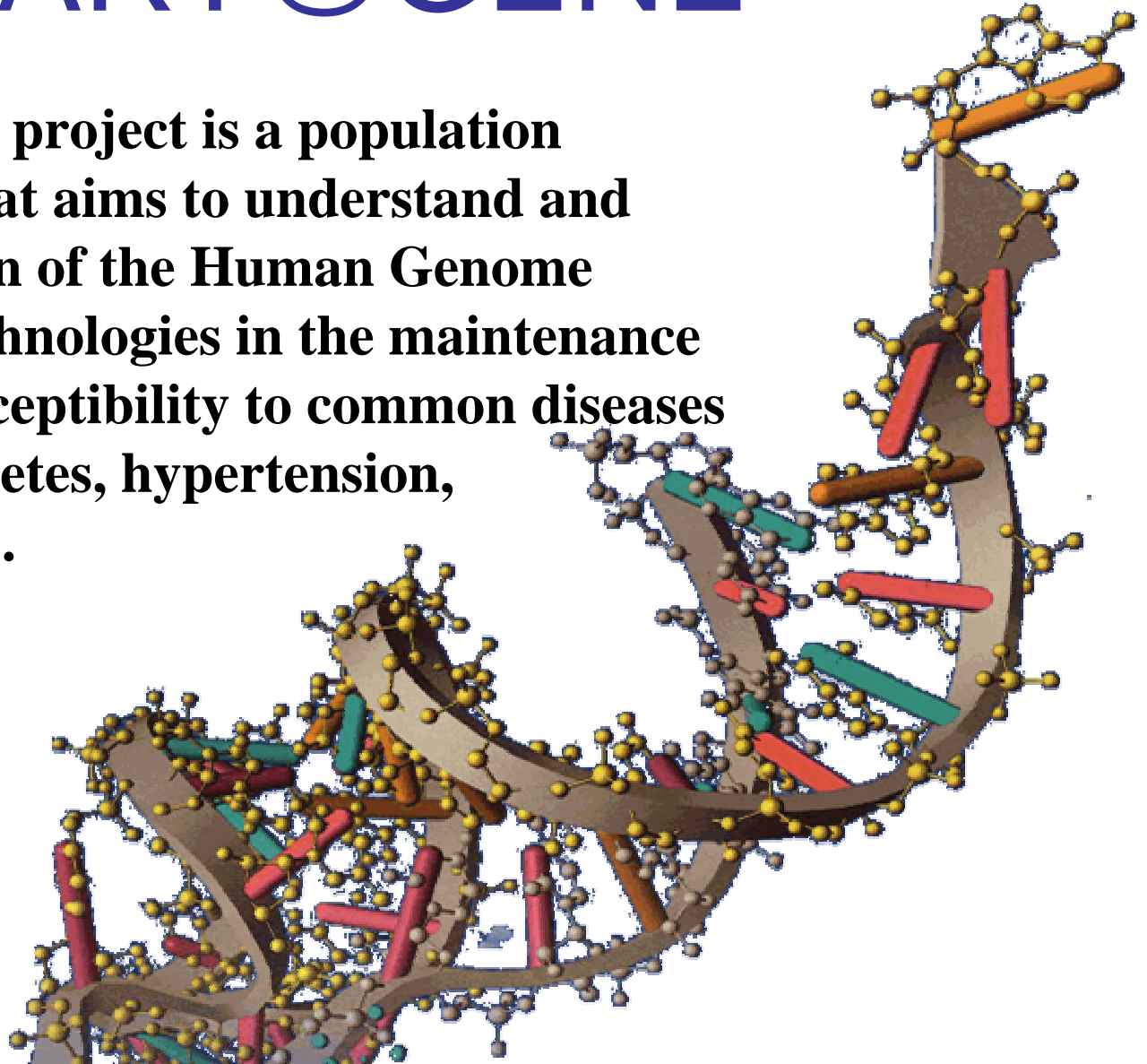


# CART@GENE

**The CART@GENE project is a population genomics project that aims to understand and guide the application of the Human Genome Project and new technologies in the maintenance of health and in susceptibility to common diseases such as cancer, diabetes, hypertension, arthritis, and others.**



# WHAT IS CART@GENE?

- A resource of health, environmental and genetic information to be widely used by the Quebec, Canadian and worldwide research communities interested in human genetics and public health.
- An unbiased, random sample of the heterogeneous Quebec population
- A double-coded, semi-longitudinal survey.
- 50,000 adults between 25-74 years of age representing  $\approx 1.2\%$  in that age group. (Phase A = 20,000 over 3 years).

# CART@GENE will:

- provide a public resource (e.g. CEPH Model) for the research community, through an Open Selection Process
- allow studies of genetic and environmental factors (and their interactions) that modify disease risk.
- serve for the validation for candidate gene studies.
- serve as a baseline study for the development of pharmacogenetics and public health programmes (education, promotion, prevention, policymaking and health services).

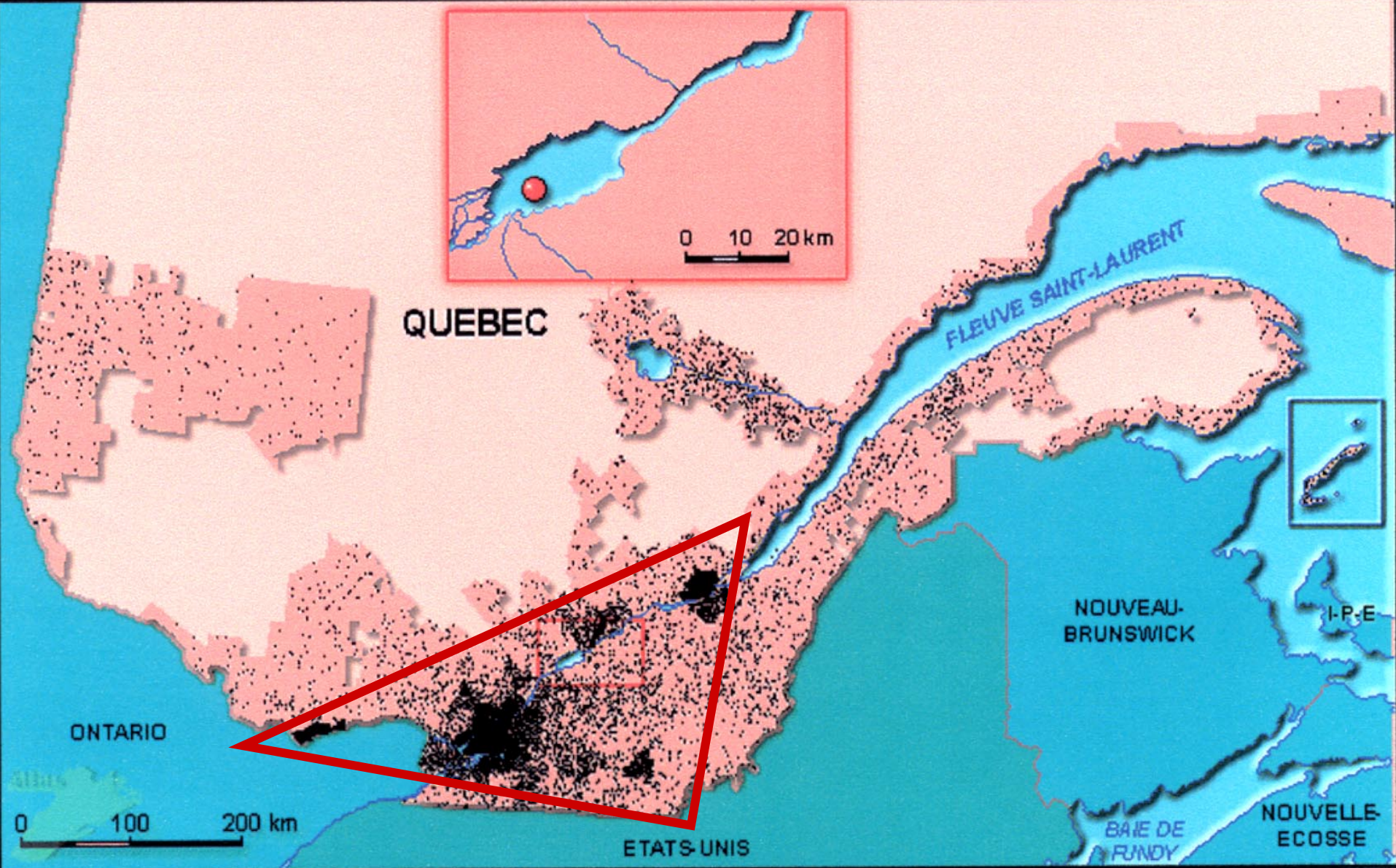
Advantages of doing a population genomics project in Quebec: Universal health care system, managed by a central body (RAMQ) and regional boards.

QuickTime™ and a  
Photo - JPEG decompressor  
are needed to see this picture.



1. Unique Health Insurance Number
2. Name and Surname at Birth
3. Date of birth and sex
4. Expiration date
5. Personal Identifier





1996



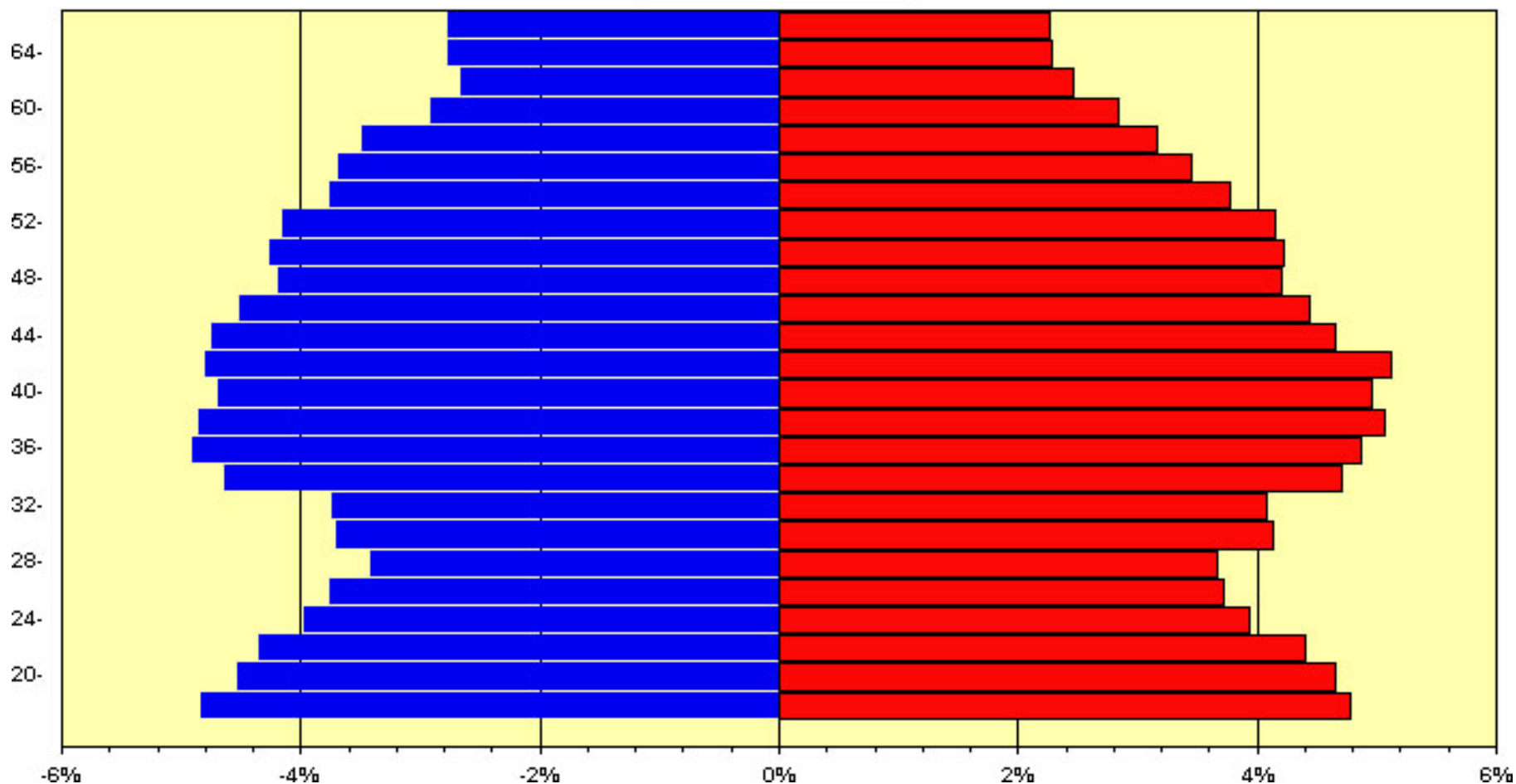
# GÉNOMIQUE DES POPULATIONS (CARTaGENE): RECRUTEMENT

RÉGIONS SOCIO SANITAIRES	N= (M+F)*	% d'inscrits	50,000 Ind.	8,000 Fam.
Gaspésie-Îles-de-la-Madeleine	104 111	1,44	720	116
Bas-Saint-Laurent	204 626	2,84	1420	227
Saguenay-Lac-Saint Jean	286 624	3,98	1990	318
Québec	632 592	8,78	4390	702
Chaudière-Appalaches	385 374	5,35	2675	428
Mauricie et Centre-du-Québec	479 015	6,65	3325	532
Estrie	282 709	3,92	1960	314
Montréal-Centre	1 774 405	24,62	12310	1970
Laval	336 339	4,67	2335	373
Laurentides	450 695	6,25	3125	500
Lanaudière	386 412	5,36	2680	429
Montréal-Est	1 281 909	17,79	8895	1423
Outaouais	308 313	4,28	2140	342
Abitibi-Témiscamingue	153 694	2,13	1065	171
Côte-Nord	102 565	1,42	710	114
Nord-du-Québec, Nunavik, Baie-James, etc	37 251	0,52	260	41
<b>ENSEMBLE DU QUÉBEC</b>	<b>7 206 634</b>			

\* Estimé de **5 566 020** personnes âgées de plus de 18 ans. Il y a **5 040 602** personnes entre 18 et 68 ans selon Statistiques Québec au 1 juillet 1999. Un recrutement de **50,000** personnes représente donc 1% de la population. Si on présume que 8-9,000 des individus recrutés peuvent construire un trio (les deux parents vivants), on peut estimer l'addition de 16-18,000 personnes au total, soit **66-68,000** ou **>1.3%** de toute la population adulte du Québec. La colonne 8,000 Fam. donne une idée de la distribution territoriale des trios.

## INDIVIDUAL RECRUITMENT

■ FEMALES ■ MALES





**Table 1: CARTaGENE Project Recruitment and Enrolment Phases**

Phase	Contact Target	Target Enrollment	Rationale
Phase A (ICI Project)	30,000 to 45,000	20,000	Participants near major university-hospital centres including: Montreal, Quebec City, Saguenay -Lac St-Jean, Sherbrooke
<div data-bbox="129 411 1195 1219"> <p>The figure is a map of the province of Quebec, Canada, showing administrative boundaries. Areas designated for Phase A recruitment are highlighted in green. These areas are concentrated in the Montreal region and include parts of the St-Lawrence River Basin. A red box on the map indicates a specific area of interest. Two inset maps are provided: one in the bottom left showing a detailed view of the Montreal area, and another in the bottom right showing a map of the St-Lawrence River Basin. A blue box with white text at the bottom center of the map area reads 'PHASE A Recruitment 20,005'.</p> </div> <p>Figure 1: Phase A Recruitment</p>			
Phase B	45,000 to 70,000	30,250	Western areas of Montreal, St-Lawrence River Basin, Outaouais River and Montérégie, Laurentide and other outlying regions.

# Principles

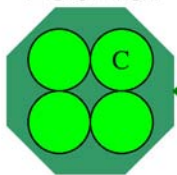
- Individuality
- Diversity
- Complexity
- Reciprocity
- Solidarity
- Security
- Accountability
- Equity
- Citizenry
- Universality

# IPEG

*Institute of Populations, Ethics and Governance\**

- Who:** Not-for Profit Corporation  
Board of public representatives
- Why:** Independent ethical governance of population tissue/ databases including:  
*Biobanks*  
*Orphan populations/banks*  
*Large cohorts*  
Public information on the ethical collection/uses of such tissues/databases
- Mandate:** Establish an ethical framework for collection/uses  
Periodic public information and periodic audits  
Ensure optimal uses for the public good  
Identify and manage conflicts of interests
- \* Three Founders (Not to be represented on the Board):  
*University of Montreal (representing Quebec Universities)*  
*FRSQ (Quebec Health Research Funding Agency)*  
*Genome Quebec*

P3G-ICI



IPEG

Institute of  
Populations and  
Genetics

SCIENTIFIC  
COMMITTEE

ETHICS  
COMMITTEE

ETHICS  
REVIEW  
(MHSS)

CARTaGENE

APPROVAL  
ACCESS  
COMMISSION

PUBLIC  
CONSULTATION

QHIB

RECRUITMENT

Age = 25-75  
N = 50,000

ASSENT  
CONSENT

SAMPLE

1.5% = 60-65,000

(Appointment)  
Participant

ETHICS  
(Audit)  
(Ongoing)

QUESTIONS  
Hx Health  
Hx Diseases

PHYSICAL  
& PHYSIO.  
MEASURES

GENEALOGIES  
CODED  
3-4 generations

DNA  
Cells  
Plasma

ENDO-  
PHENOTYPES  
(Biological tests)

DOUBLE-CODED GENETIC DATABASES

• **IPEG:** Complete governance of CARTaGENE

**DOUBLE-CODING - RAMQ:**

- ICD-9 Codes (public); medication codes
- Right to withdrawal
- Follow-up on health and lifestyle events
- Specific “new” questionnaires

**RECRUITMENT BY MRC:**

- (More community-based than FSA)
- Expertise from ISQ and Santé-Québec
- No more recruitment of regional “trios”

**SELF-QUESTIONNAIRES:**

- Logistic Help: 1-800- phone line (5/7 days)
- Mainly socio-demographic data

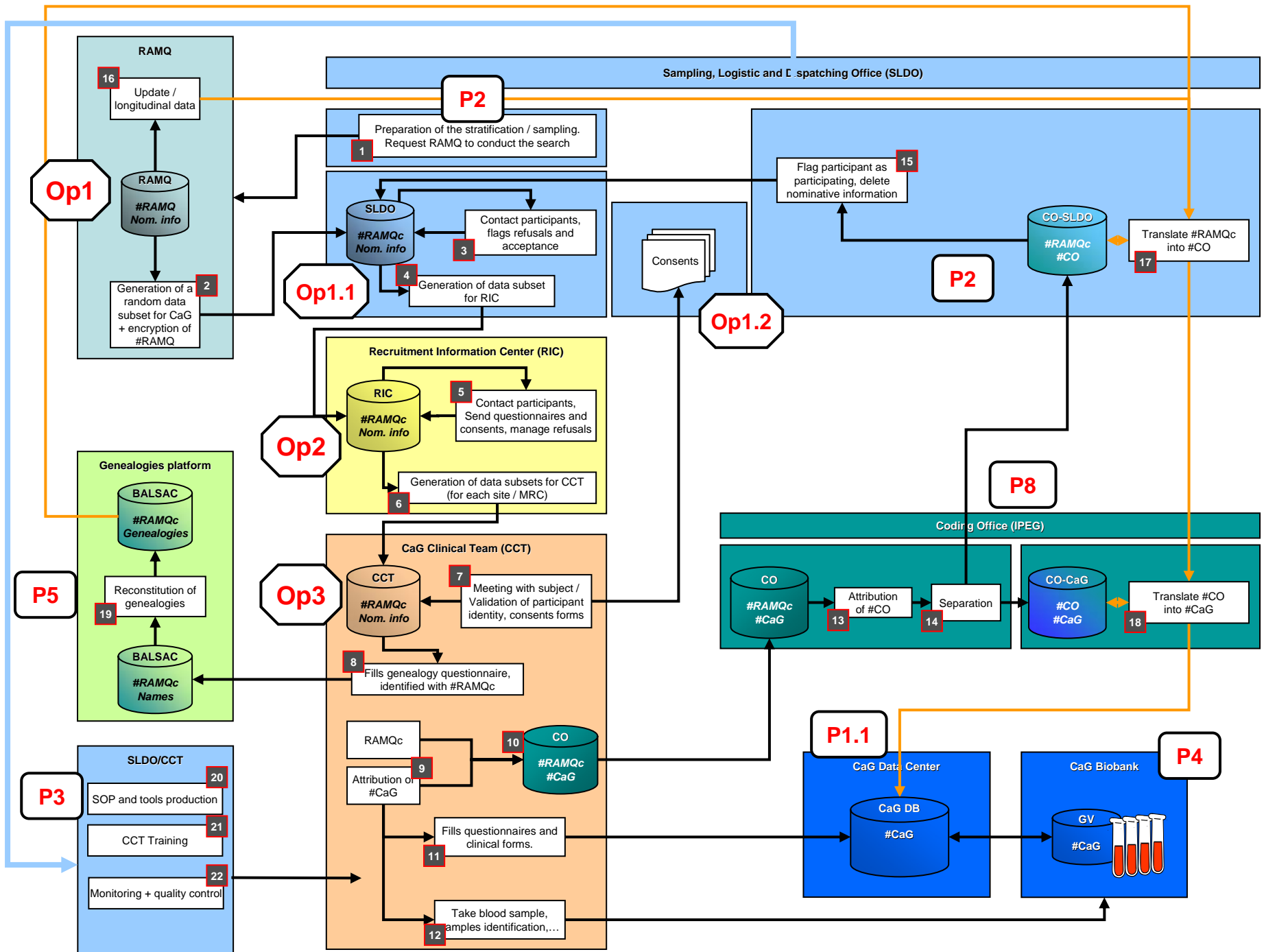
**CONSENT: (for 50 years)**

- Research on genetic diversity in relation to health or disease and the human evolution.
- Transfer of “public” codes such as ICD-9 & medication; recontact/follow-up on health and lifestyle events (v.g. death); and, for genealogical reconstruction.

**IMMEDIATE RESULTS:**

- Physical, physiological and biological
- Given to participant and/or transmitted to participant’s personal M.D.

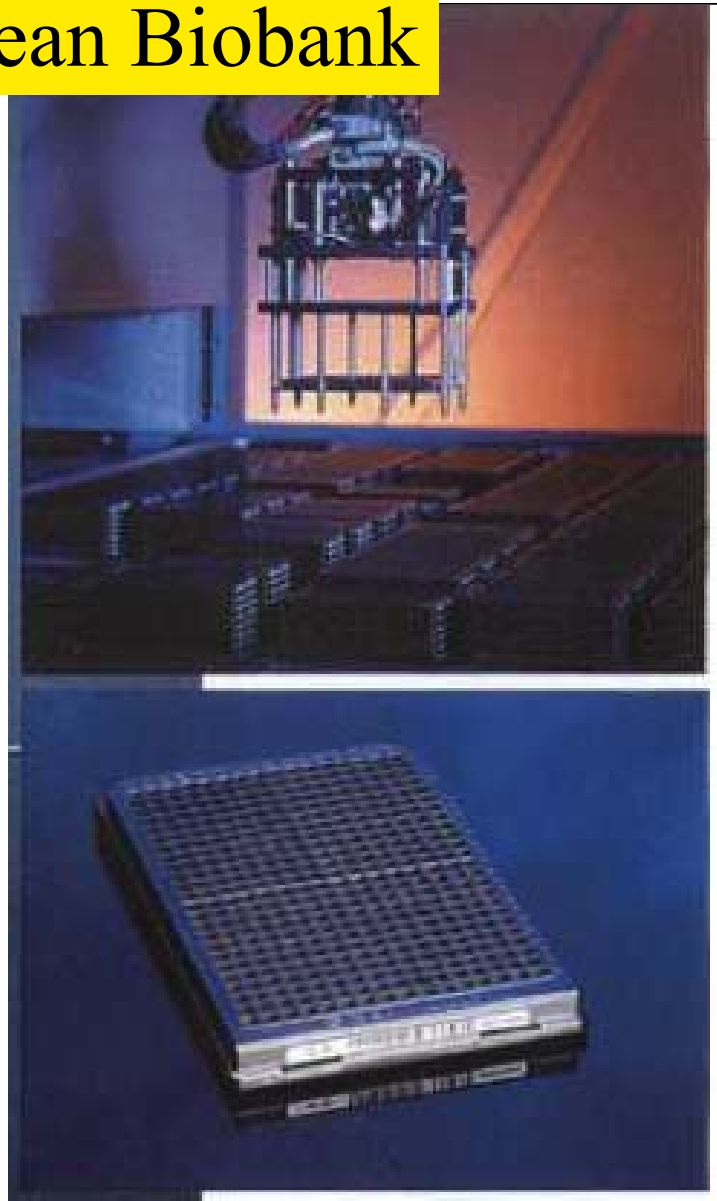




# Saguenay-Lac-St-Jean Biobank

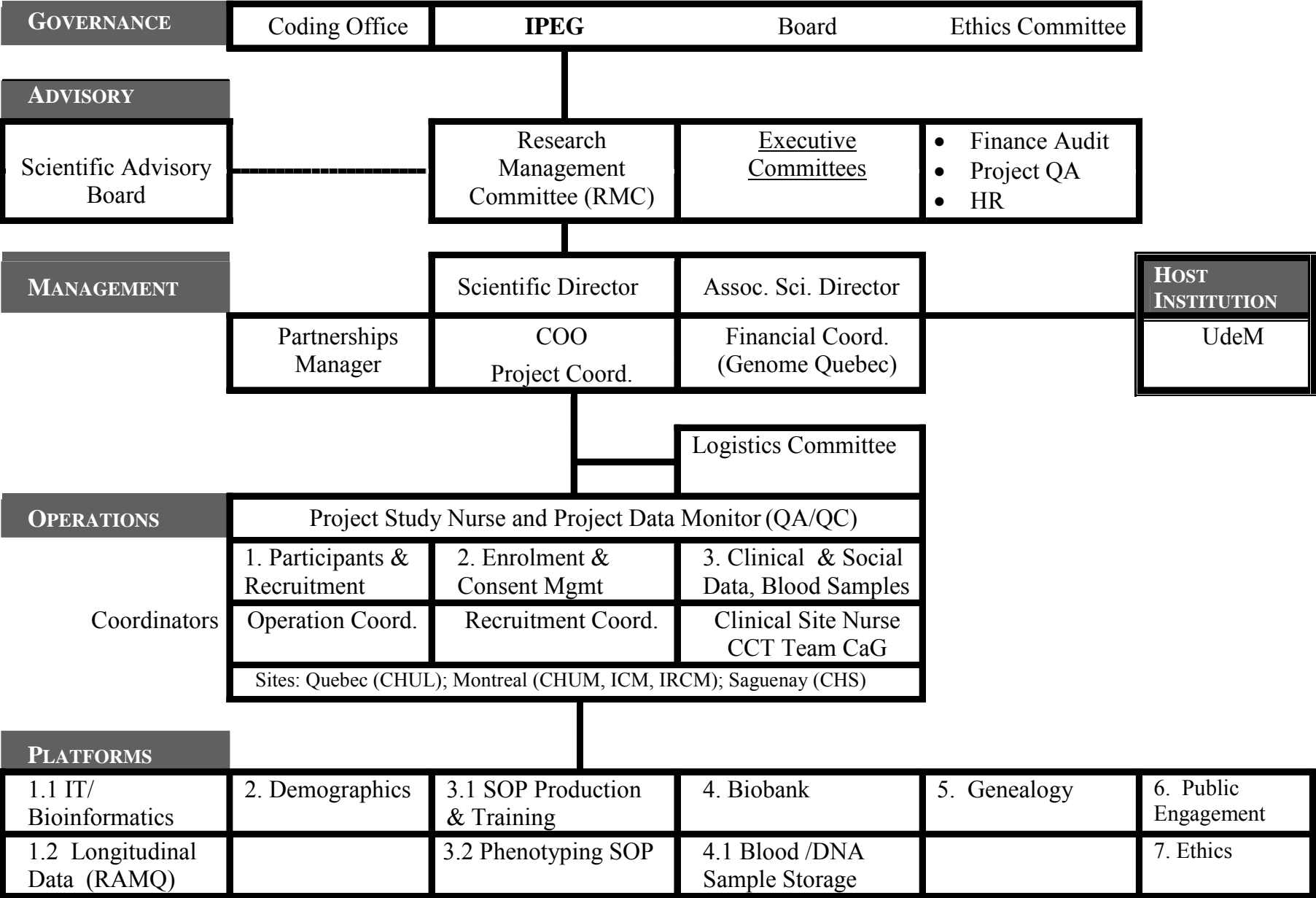


Figure 1: GenVault: DNA automated storage and retrieval system



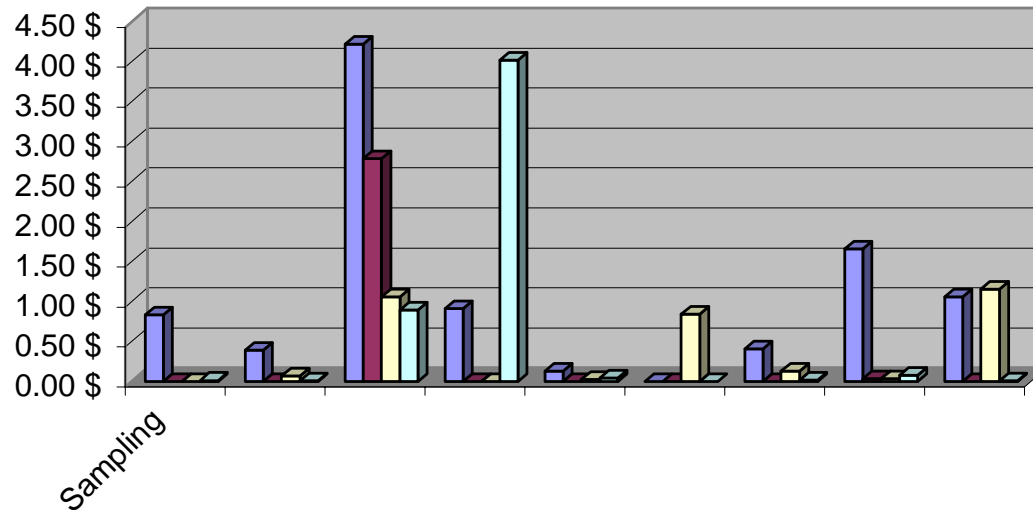
# Quebec Health Survey

- First “use” of CARTaGENE
- Early emphasis on CDV, diabetes and obesity related phenotypes:
  - anthropometric measures, blood pressure
  - fasting and 2-hour glucose
  - bio-impedance, measures of arterial stiffness
  - transcutaneous cholesterol measures
  - (automated report to patient and his physician)
  - blood phenotypes: lipid profile, ApoB, homocystinemia, HS-CRP, glycated hemoglobin, vitamin B-complex
  - genotyping of 100 SNPs in apolipoproteins, PPARs, LDLR, apoE, LPL, ACE, etc.





# CARTaGENE COST BREAKDOWN



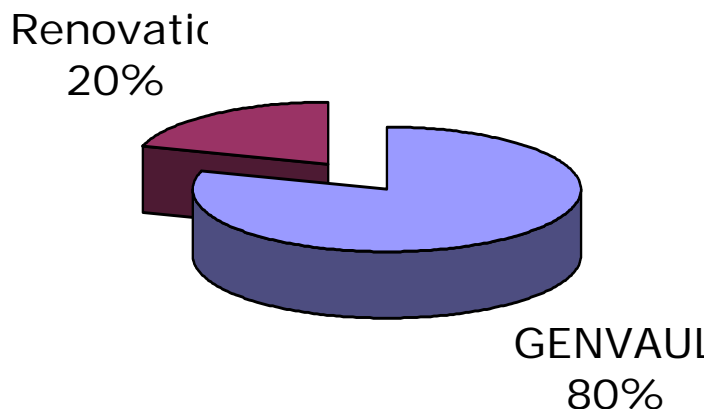
**Phase A: Budget: 20 770 080 \$**

**Phase B: Estimate: 20 M**

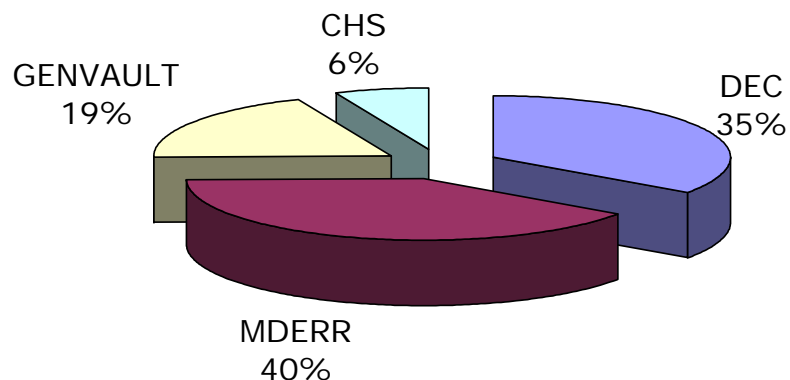
# Saguenay DNA Bank

to serve CARTaGENE Phase A, B and other projects

## Components

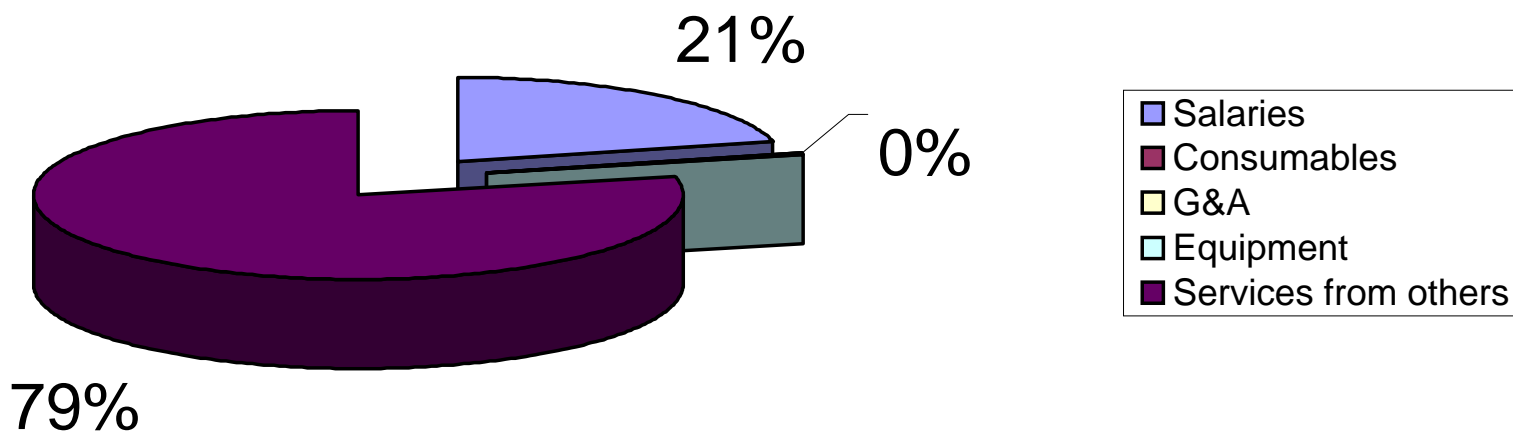


## Potential Funding Sources



**Total: \$ 5 771 572**

# Quebec Health Survey Costs



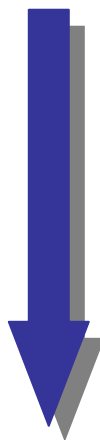
Budget: 1 579 436 \$

**Potential Funding Source: MSSS**

# QUANTITATIVE STUDY: Two-phase telephone survey

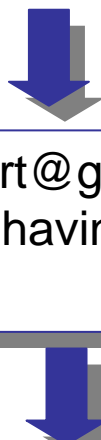
## PHASE 1

- 1,800 interviews held between Nov. 7 and Dec. 7, 2003



## PHASE 2

Mailout of Cart@gene documents to 1,394 people (77,5%) having priorly agreed to receive them



803 interviews (68%) held between Nov.19 and Dec.19

## FINDING

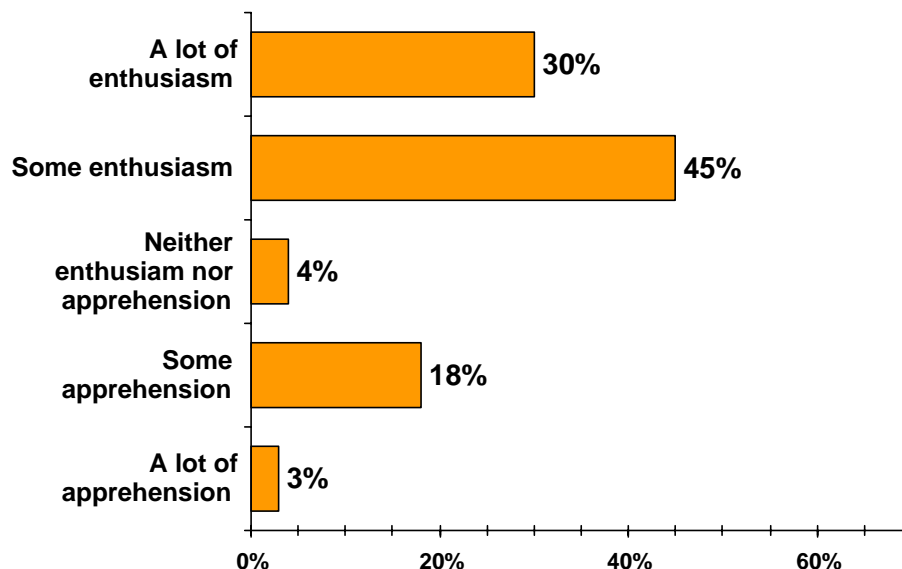


The high proportion of people accepting to receive Cart@gene info (77,5%) & the high rate of participation in the second phase (68%) reveal the interest in the project.



# Cart@gene accorded "qualified" support

Respondents had the following reactions to the project:



Close to half of respondents showed "some enthusiasm".

# Estimation of the rate of participation

- **Factorial correspondence analysis:** to identify and quantify different segments of individuals who differentiate themselves in regard to their particular responses among all the questions of the survey.

Segment	Proportion of the Quebec population	Rate of participation (inside the segment)	Rate of participation (in relation to the targeted population)
1- The uninterested	13%	22%	3%
2- The educated enthusiasts	26%	65%	17%
3- The wait and seers	24%	50%	10%
4- The apprehensive resisters	18%	10%	2%
5- The converted	19%	80%	16%

**Estimation of the rate of participation:**

**48%**

The estimation of the rate of participation is based on our current state of knowledge (project to date). With a wide, efficient and targeted communication strategy, the goal of achieving a higher participation rate seems feasible.

# Team Leaders

• Laberge Claude	Genetics	Laval U
• Gaudet Daniel	Genetics Saguenay	
• Knoppers Bartha Maria	Ethics	Montreal U
• Hudson Thomas J	Genomics	McGill
• Labuda Damian	Genetics	Montreal U
• Couture Patrick	Medicine	Laval U
• Hamet Pavel	Genetics	Montreal U
• Bouchard Gérard	Genealogy	Chicoutimi U
• Bergeron Pierre	Stats	Stat Inst.
• Vézina Hélène	Demography	Chicoutimi U
• Godard Béatrice	Ethics	Montreal U
• Arsenault Steve	Informatics	Chicoutimi
• Francois Rousseau	Biochem	Laval U
• Julian Little	Epidemiology	Ottawa U

# CARTaGENE schedule

- > 3 years of planning (2002-2005)
  - Statements of Principles of the RMGA:
    - Sampling/Access
    - Populations
  - Public consultation of the public: qualitative and quantitative phases.
  - Funding applications
- 
- Spring 2005: Authorization from Quebec's Access Commission and from a "national" REB.
  - Fall 2005: Trial phase to recruit participants according to CaG standard procedures.
  - 2006-2008: 20,000 participants
  - 2008-2011: 30,000 participants