

A Genealogical Analysis of Two Eastern Quebec Populations

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Abstract

This study is part of an ongoing research project that aims at analysing and comparing the demographic and genetic origins of Quebec's regional populations. Using the BALSAC-RETRO genealogical database, 160 genealogies from the Charlevoix and Rimouski regions were reconstructed; these ascendants go back over 12 generations. Various descriptive parameters were calculated for each group of genealogies. The origins and genetic contribution of nearly 2000 17th century founders identified in these genealogies are presented. These founders came mainly from north-western parts of France, and their contribution explains approximately 77% of the Charlevoix gene pool and 74% of the Rimouski gene pool. The first 10% of founders provide 63% and 79% of the total contribution to Rimouski and Charlevoix respectively.

Résumé

Cette étude s'inscrit dans un projet de recherche visant à analyser et à comparer les origines démographiques et génétiques des populations régionales du Québec. À partir du fichier généalogique BALSAC-RETRO, 160 généalogies provenant des régions de Charlevoix et de Rimouski ont été reconstruites; ces ascendances remontent à plus de 12 générations. Divers paramètres descriptifs ont été calculés pour chaque corpus de généalogies. Les origines et la contribution génétique de quelque 2000 fondateurs du 17^e siècle identifiés dans ces généalogies sont présentées. La plupart de ces fondateurs proviennent des régions du Nord-Ouest de la France et leur contribution explique à peu près 77% du pool génique de Charlevoix et 74% du pool génique de Rimouski. Les premiers 10% des fondateurs fournissent 63% et 79% de la contribution totale à Rimouski et Charlevoix, respectivement.

Key Words: genealogies, genetic contribution, Quebec population

Introduction

Research in the field of human genetics has experienced a tremendous development in recent years. The expansion of knowledge on the structure and function of the human genome (Dausset et al., 1990; Olson 1993; Dib et al. 1996; Collins et al. 1998) has led to a better definition and understanding of monogenic diseases (i.e., diseases caused by a mutation on a single gene). Moreover, research has now tackled the problem of more complex and common diseases such as diabetes, asthma, cancer and heart diseases (Lander and Schork, 1994; Long et al. 1997).

Human population genetics combines notions of human genetics and demography. It aims to describe and explain the genetic structure of a population with the study of its demographic history. Research in human population genetics can help to better understand the origins, transmission, diffusion and frequency of hereditary diseases, hence improving methods to identify at-risk populations (Scriber, 1991). In Quebec, the identification of the origins of some rare hereditary diseases has led to numerous studies in the last 20 years (for an extensive review, see Vézina, 1996). Recent results indicate that although it is quite difficult to pinpoint the exact origin of a given gene (Heyer et al., 1997), a few individuals show higher probabilities of being the

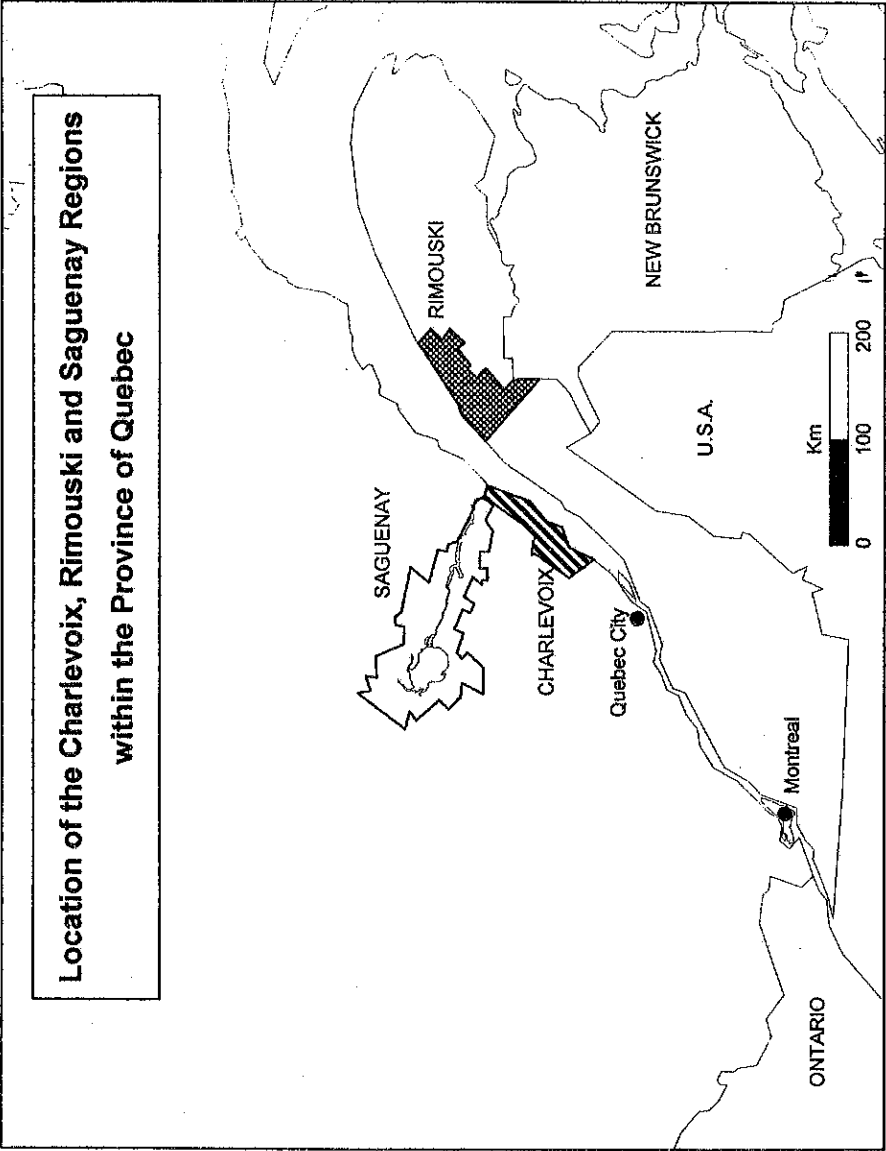
ones to have introduced specific deleterious genes in the Quebec population (Heyer, 1999).

It is estimated that about two thirds of the present francophone Quebec's gene pool (i.e., all of the different genes that can be found in this population) came from founders who settled in Nouvelle-France in the 17th century (Charbonneau et al., 1993). It is also known that the nature and the frequency of inherited disorders (caused by deleterious genes) display important regional variations (Bouchard, 1989; Scriver, 1991; Bouchard and DeBraekeleer, 1992). These variations can be explained by the demographic dynamics (mainly differential fertility and migration) of the 17th century founders and their descendants (Bouchard et al., 1991). This study focuses on some consequences of that differential behaviour. To this effect, the genetic contribution of 17th century founders to the contemporary populations (i.e., the proportion of the contemporary gene pool that comes from these ancestors) of two regions of the eastern part of the province of Quebec, namely Charlevoix and Rimouski, were compared using data from the BALSAC-RETRO genealogical database¹. A main feature of this database is the possibility it offers to conduct longitudinal studies over several generations (up to 16 in some cases). Results from these studies help to evaluate the magnitude and the variability of founder effects in different Quebec regions. In turn, these evaluations are useful in other fields of human genetics studies (Heyer, 1995). For example, extensive genealogical information helps to enhance the efficiency of molecular analysis, which aims at the identification and localisation of deleterious genes responsible for monogenic or complex hereditary diseases (Wright et al., 1999). Genealogical analyses can also provide novel demographic estimations for the study of human evolution (Tremblay and Vézina, 2000). Results from such research can be applied to other populations where genealogical data is not as readily available nor as rich and complete as in Quebec.

Historical background

The Charlevoix region (pop. 30,000) is located on the north shore of the St. Lawrence River, north-east of Quebec City (Figure 1). French Canadian settlement in that region began at the end of the 17th century (Jetté et al., 1991). Between the middle of the 18th century and the middle of the 19th century, the average annual growth rate of the population was around 3%. During the second half of the 19th century, people from Charlevoix emigrated to the Saguenay and became the first settlers in this region (Roy et al., 1988; Gauvreau et al., 1991). It is estimated that between 1838 and 1911, nearly 14,000 people from Charlevoix settled in the Saguenay region (Gauvreau et al., 1991). Many others emigrated elsewhere in Quebec or the United States during the same period. Since the early 1960's, the Charlevoix population has practically ceased to expand.

Figure 1



Rimouski (pop. 55,000) is located further east on the south shore of the St. Lawrence River. The earliest settlements in the Rimouski region go back to the 18th century, but the population's expansion only began in the 19th century (Fortin et al., 1993). Between 1831 and 1881, the population grew at a rate of 2.5% per year. The next 50 years were characterised by an out-migration towards the United States and Western Quebec; between 1881 and 1891, the population of the region actually declined. Since WW2, the population growth has been mostly limited to the town of Rimouski. The population of the rural parts of the region has been declining since the early 1970's.

Material and Methods

BALSAC and BALSAC-RETRO databases

The main purpose of the BALSAC population register is to support research aimed at describing the formation and evolution of Quebec's population, with the use of information on individual and family life-courses (Bouchard, 1992; Bouchard et al., 1995). Numerous research projects using this register have been conducted since 1972. These projects cover various fields of research (history, social science, demography, human geography, anthropology, human population genetics and epidemiology, etc). Initially, the register was limited to the population of the Saguenay region. All parish records (birth, marriage and death certificates) dating from 1842 to 1971 have been computerised and linked. The database is currently being expanded to all regions of Quebec for the 19th and 20th centuries (marriage certificates only). As of November 1999, BALSAC contained nearly 1.5 million records, of which 690,000 have been linked (Bouchard, 1999).

BALSAC-RETRO is a peripheral database that was developed for specific projects through the use of genealogical records (Jomphe and Casgrain, 1997). At present, this database contains genealogical data on 244,000 individuals from all Quebec regions and goes back as far as the early 17th century.

Genealogical reconstruction

The starting points of the ascending genealogies (from now on called the "subjects") were chosen randomly for each region, using a classic random number generator, among the available data in the BALSAC register. For Charlevoix, the 80 subjects were drawn among all births that occurred in this region between 1950 and 1971. In the case of Rimouski, only marriage records dating between 1900 and 1948 were available in the BALSAC register at the time of the study. The Rimouski group was thus formed with 80 "fictional" children from each one of the 80 randomly selected couples².

Ascending genealogies were reconstructed using the information available in the BALSAC-RETRO database. Some published records were used, mainly for the first generations. Data from the Registre de Population du Québec Ancien (Programme de recherche en démographie historique, Université de Montréal) and the Jetté genealogical dictionary were also used for the period covering the 17th and 18th centuries (Légaré, 1988; Jetté, 1983). All genealogies were completed up to the first immigrants (who came from France, for the most part) or until the needed information (i.e. names and marriage date of parents) could no longer be found in the available sources.

Structure of the genealogies

The completeness index per generation, the average genealogical depth and a heterogeneity index (total and per generation) were calculated for each group of genealogies.

The completeness index (C_g) for a given generation level is the ratio of the number of known ancestors at that generation level to the maximum number of ancestors at that same level:

where: g = generation level (that of the subjects' parents being the first)

$$C_g = A_g / (N \cdot 2^g)$$

A_g = number of known ancestors at level g

N = number of genealogies

This index shows how complete is the genealogical information at each generation. For example, if $C_3 = 0.5$, then half of the subjects' great-grand-parents have been found in the database. The maximum C_g value of 1 means that at generation level g , all ancestors have been identified. By definition, C_{g+1} is always inferior or equal to C_g .

Summing C_g through all generation levels gives the average genealogical depth (D):

$$D = \sum_{g=1}^m (A_g) / (N \cdot 2^g)$$

where: m = maximum generation level

The average genealogical depth can be defined as a measure of the average generation level where the genealogies' branches stop (i.e. beyond which the

necessary information is not known). It gives a good idea of how far back the genealogies reach.

Since several ancestors appear more than once in the genealogies, it is useful to know how many of them are found at each generation level. A heterogeneity index (H_g) can then be calculated to indicate the proportion of distinct ancestors (DA_g) among all known ancestors (A_g):

$$H_g = DA_g/A_g$$

In this measure, each ancestor is counted only once, even if he or she appears at more than one generation level. Therefore, in a genealogical structure where some ancestors appear several times (either in a single genealogy or in many), the heterogeneity index will decrease rapidly as the generation level increases. At some level, where genealogical information becomes scarce, the H_g value often drops to 0 (meaning that all ancestors identified at that level have already appeared at lower levels).

Origin, frequency and genetic contribution of 17th century founders

The 17th century founders are those ancestors who married and immigrated in Canada before the year 1700³. According to Charbonneau et al. (1993), a total of approximately 5,000 immigrants settled in Canada between 1608 and 1700. It is estimated that 75% of them have descendants in the contemporary Quebec population. Many of these 17th century founders appear in the Charlevoix and Rimouski genealogies. Following a study by Heyer et al. (1997) based on genealogies from the Saguenay region, the geographical origins of the founders were investigated. These origins might represent either the place of birth, marriage, or emigration.

All links between the 17th century founders and the 160 subjects were identified in order to calculate the number of genealogies in which each founder appears as well as the founders' mean number of appearances per genealogy. The frequency and generation level of these appearances were used to calculate the founder's genetic contribution to each group of subjects. The genetic contribution (GC_f) of a given founder to a number of subjects is calculated as follows:

$$GC_f = \sum_{i=1}^s \sum_{j=1}^p (1/2)^{g_{ij}}$$

where: s = number of subjects genealogically linked with the founder (f)
 p = number of genealogical paths between the founder (f) and the subject
 g_{ij} = number of generations, in each path (j), between the founder (f) and the subject (i)

As an illustration, consider a subject and each of his four grand-parents. Each grand-parent is linked with the subject by only one path (if we assume, of course, that the subject's parents are not brother and sister). The genetic contribution of each grand-parent to this subject will thus be $(1/2)^2 = 0.25$.

The genetic contribution of a founder can be interpreted as the expected number of carriers, among the subjects, of a specific gene copy originating from this founder (Roberts, 1968; O'Brien et al., 1994; Heyer and Tremblay, 1995). Interestingly, genetic contribution allows the evaluation of the genetic consequences of the demographic behaviour of a founder and his descendants (Heyer, 1995). A founder with a relatively high genetic contribution will have a higher probability of having transmitted copies of his genes through his descendants to contemporary subjects. Comparison of the genetic contributions of each founder will thus give an indication of the variability of the demographic behaviour (mainly differential reproduction and migration) among the ancestors of the population under study.

Summing GC_f for all founders with the same geographical origin and dividing the result by the number of subjects gives the proportion of these subjects' gene pool which came from that origin. This measure provides useful information to evaluate the importance of a region's contribution to another region's population settlement or expansion.

Results

Charlevoix genealogies have an average depth of 10.8 generations, compared to 9.4 generations for the Rimouski genealogies (Table 1). The difference of 1.4 generation between the two groups is mainly due to the periods at which each set of genealogies were initiated (genealogies with a more recent start point reach a longer depth). The average time between the birth of Charlevoix subjects and the marriage of their parents is 6.6 years. Adding this value to the mean marriage year of Rimouski couples (1924) gives 1931 as an estimation for the mean birth year of Rimouski subjects, compared to 1961 for the Charlevoix subjects.

Charlevoix genealogies are fairly complete (over 0.90) up to the ninth generation. The completeness of Rimouski genealogies falls under 0.90 at the 8th generation. After the 12th generation (11th for Rimouski), the values are below 0.10. Still, some Charlevoix branches reach as far back as 16 generations (15 for Rimouski).

Table 1
Structural Characteristics of the Charlevoix and Rimouski Genealogies

generation level	Number of known ancestors (Ag)		Number of distinct ancestors (DAg)		Completeness (Cg)		Heterogeneity (Hg)	
	Charlevoix	Rimouski	Charlevoix	Rimouski	Charlevoix	Rimouski	Charlevoix	Rimouski
1	160	160	158	157	1.000	1.000	0.988	0.981
2	320	320	301	306	1.000	1.000	0.941	0.956
3	636	628	553	566	0.994	0.981	0.869	0.901
4	1,264	1,234	955	997	0.988	0.964	0.756	0.808
5	2,506	2,442	1,458	1,610	0.979	0.954	0.582	0.659
6	4,966	4,772	1,626	2,334	0.970	0.932	0.327	0.489
7	9,818	9,274	1,471	2,603	0.959	0.906	0.150	0.281
8	19,292	17,606	1,819	2,430	0.942	0.860	0.094	0.138
9	37,620	32,110	2,220	2,174	0.918	0.784	0.059	0.068
10	71,722	48,972	2,196	1,249	0.876	0.598	0.031	0.026
11	118,040	49,808	1,239	493	0.720	0.304	0.010	0.010
12	115,988	28,216	282	71	0.354	0.086	0.002	0.003
13	58,980	9,106	14	2	0.090	0.014	0.000	0.000
14	16,330	1,472	0	0	0.012	0.001	0.000	0.000
15	2,714	78	0	0	0.001	0.000	0.000	0.000
16	108	0	0	0	0.000	0.000	0.000	—
Total	460,464	206,198	14,292	14,992	10.80	9.38	0.031	0.073

Source: Interuniversity Institute for Population Research, BALSAC-RETRO database

The heterogeneity of both groups is relatively high until the 4th generation (0.76 for Charlevoix and 0.81 for Rimouski). Afterwards, the values rapidly decline which means that more and more ancestors repeatedly appear in the genealogies. Overall, the proportions of distinct ancestors among all known ancestors are only 3% and 7% respectively in the Charlevoix and Rimouski genealogies. This means that each ancestor appears 32 times on average in the Charlevoix genealogies and 14 times in the Rimouski genealogies. Ancestors with the highest number of appearances were found mainly among the 17th century founders.

A total of 1925 17th century founders (1206 males and 719 females) were identified in the 160 genealogies. Almost 69% of them (1135) were found in both groups. 376 of the 1511 Charlevoix founders (25%) did not appear in the Rimouski genealogies, whereas 414 of the 1549 Rimouski founders (27%) were absent from the Charlevoix genealogies. The difference between the numbers of male and female founders is due to the fact that marriages between male immigrants and Canadian-born females were much more frequent than marriages between female immigrants and Canadian-born males (Charbonneau et al., 1993).

Nearly all founders came from France (Table 2). Among these, most were born in the provinces located in the northern and western parts of the country. The provinces of Normandie, Île-de-France, Aunis and Poitou provided 58.3% (Charlevoix) and 58.1% (Rimouski) of all founders. There are some important differences between the origins of male and female founders: the proportions of founders coming from Bretagne, Normandie, Poitou and Saintonge were significantly higher for males, whereas the proportions of female founders coming from Île-de-France and Aunis were greater. These findings are similar with those obtained by Heyer et al. (1997) for the Saguenay region. Since 69% of the founders were common to the two groups, there are no important differences between the origins of Charlevoix and Rimouski founders.

Most founders did not appear in a large number of genealogies (Table 3). For both regions, approximately 60% of founders appeared in 10% or less of the genealogies. This proportion was somewhat higher (68%) for the Saguenay region (Heyer et al., 1997). At the other end of the scale, nearly 8% of Charlevoix founders appeared in more than 80% of the genealogies, compared to less than 5% for Rimouski. The vast majority of founders appeared only once or twice in the genealogies; the average number of appearances per genealogy is less than 2 for 83% of Charlevoix founders and 86% of Rimouski founders. Nonetheless, it is worth noting that a few founders turned up quite frequently, especially in the Charlevoix genealogies where 39 founders (2.6%) had an average of ten appearances or more per genealogy, 17 of them reaching an average of over 20 appearances. Only 6 founders (0.4%) had an average number of appearances higher than 10 (but lower than 20) in the Rimouski genealogies.

Table 2
Distribution (%) of 17th Century Founders according to their Geographical Origin

Region	Female founders		Male founders		All founders	
	Charlevoix	Rimouski	Charlevoix	Rimouski	Charlevoix	Rimouski
North-west of France:	25.2	26.2	37.6	38.7	32.8	34.0
<i>Arjou</i>	0.7	1.2	1.8	2.3	1.4	1.9
<i>Bretagne</i>	1.4	1.5	4.3	4.5	3.2	3.4
<i>Maine</i>	2.3	2.0	3.2	3.1	2.8	2.7
<i>Normandie</i>	15.2	15.9	21.7	21.3	19.2	19.2
<i>Perche</i>	4.9	4.9	5.5	6.0	5.3	5.6
<i>Touraine</i>	0.7	0.7	1.1	1.5	0.9	1.2
North-centre of France:	35.1	35.2	12.5	11.6	21.2	20.5
<i>Artois</i>	0.2	0.0	0.3	0.2	0.3	0.1
<i>Beauce</i>	1.2	1.9	0.9	0.7	1.0	1.2
<i>Brie</i>	1.2	0.7	0.5	0.7	0.8	0.6
<i>Île-de-France</i>	27.7	28.2	8.1	7.6	15.6	15.5
<i>Orléanais</i>	2.4	2.5	1.1	0.9	1.6	1.5
<i>Picardie</i>	2.4	1.9	1.6	1.5	1.9	1.6
Centre-west of France:	26.3	25.8	33.4	33.6	30.7	30.6
<i>Angoumois</i>	1.2	1.3	3.5	2.7	2.6	2.2
<i>Aunis</i>	15.3	14.2	9.8	9.7	11.9	11.4
<i>Poitou</i>	6.3	6.6	14.8	15.4	11.6	12.0
<i>Saintonge</i>	3.5	3.7	5.3	5.8	4.6	5.0
Other region of France	5.9	5.7	11.3	11.2	9.2	9.2
France, unspecified	4.9	4.2	3.3	3.0	3.9	3.5
Other origins	1.7	1.9	1.1	1.2	1.3	1.4
Unknown origin	0.9	1.0	0.8	0.7	0.9	0.8
Total	100.0	100.0	100.0	100.0	100.0	100.0

Source: Interuniversity Institute for Population Research, BALSAC-RETRO database

Table 3
Distribution (%) of Founders according to the Percentage of
Genealogies in which they appear and their Average Number of
Appearances per Genealogy

Group	Percentage of Genealogies			Average Number of Appearances pr Genealogy		
	]0,10]	]10,80]	]80,100]	]0,2]	]2,10]	10+
Charlevoix	60.8	31.5	7.7	83.2	14.2	2.6
Rimouski	59.1	36.2	4.7	85.7	13.9	0.4

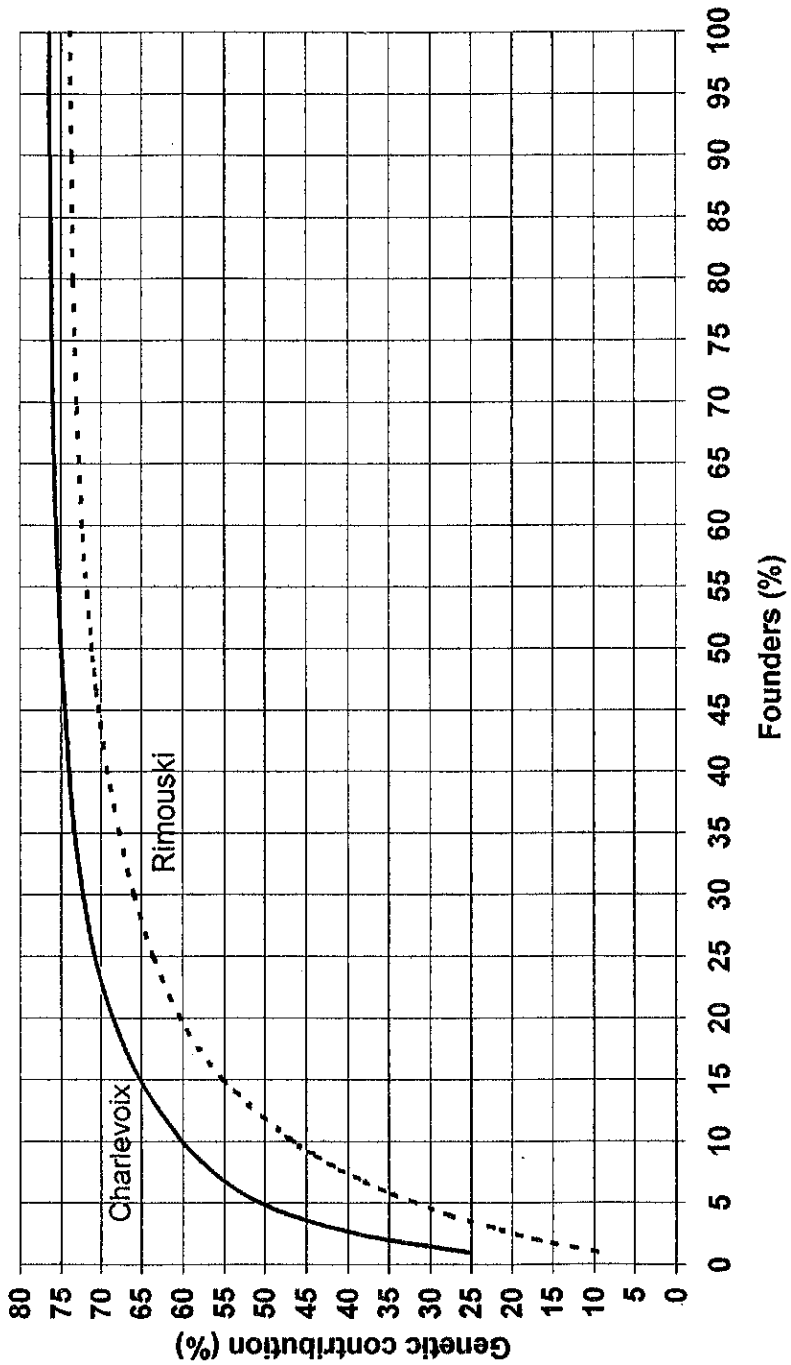
Source: Interuniversity Institute for Population Research, BALSAC-RETRO database

The total genetic contribution of the 17th century founders reached 61.2 in the Charlevoix genealogies and 59.1 in the Rimouski genealogies. In other words, these founders explain 76.5% and 73.9% of the Charlevoix and Rimouski gene pool respectively (Figure 2). A value of 82% was obtained by Heyer et al. (1997) for the Saguenay.

A few founders accounted for a large portion of the gene pool. These founders are mainly those who appeared in most of the genealogies, but there are exceptions. Specifically, some founders appeared in many genealogies yet had a low genetic contribution, while some other founders who appeared in few genealogies showed a high genetic contribution. The Rimouski region showed a significantly lower concentration of genetic contribution in comparison with Charlevoix: only 10% of the Rimouski gene pool is explained by the top 1% contributors, compared to 25% of the Charlevoix gene pool. It takes only 5% of the founders to reach half of the Charlevoix gene pool, while 12% of the founders are needed to reach that proportion for Rimouski. The distributions appearing in Figure 2 clearly show that for both regions, most of the founders' total genetic contribution is explained by less than half of these founders.

The fact that many founders are common to the two groups does not imply that the genetic contribution of each of these founders is the same in each group. All together, the common founders' genetic contribution is 60.1 for Charlevoix and 57.2 for Rimouski; this contribution represents 98% and 97% of the total genetic contribution to Charlevoix and Rimouski respectively. Taken separately, the

Figure 2
Cumulative proportion of genetic contribution by
cumulative proportion of founders



genetic contribution of 379 founders is at least 25% higher for Charlevoix than for Rimouski; their total contribution is 47.8 for Charlevoix and 9.1 for Rimouski. Conversely, 654 founders have a genetic contribution that is at least 25% higher for Rimouski than for Charlevoix, with a total contribution of 44.0 for Rimouski and 7.9 for Charlevoix.

Founders from north-western France provided a large part (45% for Charlevoix and 42% for Rimouski) of the founders' total genetic contribution (Table 4). These proportions are higher than the north-western founders' relative weight among all founders (33% and 34% respectively); the frequency of genealogical links between these founders and the subjects is thus higher than average. The situation is reversed for the founders from the north-centre and centre-west regions, although some subgroups show otherwise. Founders from the Perche province had a remarkably high contribution, particularly for Charlevoix subjects (almost four times their relative weight). In fact, Perche founders had the highest average genetic contribution in both populations (0.159 for Charlevoix and 0.089 for Rimouski). Other higher-than-average contributions include those from Maine (0.097), Orléanais (0.066) and Angoumois (0.066) founders to Charlevoix, as well as Other origins (0.47), Normandie (0.043) and Orléanais (0.043) founders to Rimouski.

In the study by Heyer et al. (1997), 58 founders were identified as appearing at least once in 95% or more of the genealogies of at least one of seven Saguenay groups (a total of 772 subjects). Half of them were found in all groups. These 58 founders are the most important Saguenay founders: their genetic contribution accounts for 44% of the Saguenay gene pool. The contributions of these top Saguenay founders to the Charlevoix and Rimouski populations were compared (Table 5). Results clearly show that the contribution is much higher for Charlevoix than Rimouski. On average, the 58 founders appeared in 98% of the Charlevoix genealogies and 70% of the Rimouski genealogies. Nearly all founders appeared in 95% or more of the Charlevoix genealogies, but only a third of them were found in 95% or more of the Rimouski genealogies. The 58 founders' genetic contribution to Charlevoix reached 42%, which is similar to the Saguenay, but their contribution to Rimouski was much lower (15%). The average genetic contribution is 14 times higher than that of all Charlevoix founders and 5 times higher than that of all Rimouski founders. As for the top 29 founders, all of them showed up at least once in all Charlevoix genealogies. These 29 founders accounted for nearly one third of the Charlevoix gene pool and had an average genetic contribution which is 20 times higher than the average for all Charlevoix founders.

Table 4
Genetic Contribution of 17th Century Founders according to their Geographical Origin

Region	Total		%		Per Founder	
	Charlevoix	Rimouski	Charlevoix	Rimouski	Charlevoix	Rimouski
North-west of France:	27.78	24.83	45.4	42.0	0.056	0.047
<i>Anjou</i>	0.20	0.87	0.3	1.5	0.010	0.030
<i>Bretagne</i>	0.89	1.53	1.5	2.6	0.019	0.029
<i>Maine</i>	4.15	1.64	6.8	2.8	0.097	0.039
<i>Normandie</i>	9.76	12.72	15.9	21.5	0.034	0.043
<i>Perche</i>	12.71	7.67	20.8	13.0	0.159	0.089
<i>Touraine</i>	0.07	0.40	0.1	0.7	0.005	0.022
North-centre of France:	10.08	10.63	16.5	18.0	0.032	0.033
<i>Artois</i>	0.00	0.02	0.0	0.0	0.000	0.010
<i>Beauce</i>	0.39	0.52	0.6	0.9	0.026	0.029
<i>Brie</i>	0.50	0.27	0.8	0.5	0.042	0.027
<i>Île-de-France</i>	6.09	7.90	10.0	13.4	0.026	0.033
<i>Orléanais</i>	1.58	1.02	2.6	1.7	0.066	0.043
<i>Picardie</i>	1.52	0.90	2.5	1.5	0.052	0.036
Centre-west of France:	18.25	16.11	29.8	27.3	0.039	0.034
<i>Angoumois</i>	2.63	1.05	4.3	1.8	0.066	0.031
<i>Aunis</i>	9.06	6.91	14.8	11.7	0.050	0.039
<i>Poitou</i>	5.16	5.91	8.4	10.0	0.029	0.032
<i>Saintonge</i>	1.40	2.24	2.3	3.8	0.020	0.029
Other region of France	1.70	3.98	2.8	6.7	0.012	0.028
France, unspecified	2.24	2.19	3.7	3.7	0.038	0.041
Other origins	0.94	1.01	1.5	1.7	0.047	0.046
Unknown origin	0.21	0.35	0.3	0.6	0.016	0.027
Total	61.20	59.10	100.0	100.0	0.041	0.038

Source: Interuniversity Institute for Population Research, BALSAC-RETRO database

Table 5
Top Saguenay Founders' Contribution to Charlevoix and Rimouski

	Founders Appearing In 95-100% of the Genealogies of at least One Sagueny Group (N=58)		Founders Appearing in 95-100% of all Sagueny Groups (N=29)	
	Charlevoix	Rimouski	Charlevoix	Rimouski
Average % of Genealogies Covered	98	70	100	82
% of Founders Appearing in at least 95% of the Genealogies	95	34	100	48
Total Genetic Contribution (%)	41.7	14.8	30.1	9.4
Average Genetic Contribution	0.575	0.204	0.831	0.260

Source: Interuniversity Institute for Population Research, BALSAC-RETRO database

For comparison purposes, all founders appearing in at least 95% of Charlevoix and Rimouski genealogies were also separately identified. A total of 87 founders was obtained for Charlevoix; this number is relatively high compared to the 58 Saguenay founders. Those founders who appeared in 95% or more of the Charlevoix genealogies but in less than 95% of the Saguenay genealogies were nevertheless quite common in the latter (not less than 78% of at least one Saguenay group's genealogies). For Rimouski, 22 such founders were identified, and only 2 of them appeared in less than 95% of the Saguenay (63%) and Charlevoix (46%) genealogies.

Conclusion

One of the consequences of the fact that a large part of the contemporary Quebec population originates from a relatively small number of 17th century founders is that some regional populations might share a large proportion of their gene pool. This idea has been suggested in previous studies, in particular in the case of the Charlevoix and Saguenay populations for which thorough historical demographic analysis has been made (Pouyez and Lavoie, 1983; Bouchard and DeBraekeleer, 1991). The presence of some rare hereditary diseases in eastern Quebec also provided evidence for a geographical concentration of certain genes in the population (Laberge, 1969; Scriver, 1991).

Differential migratory movements among the founders' descendants, along with differential reproduction, constitute the keys that allow to explain the evolution of the Quebec gene pool. The genetic impact of the Charlevoix emigration to the Saguenay region in the middle of the 19th century is clear: these populations appear to share a large portion of the same gene pool. The concentration of the gene pool seems, however, a little higher in the Saguenay population than in the Charlevoix population. This can be explained in part by the fact that a relatively small number of people from Charlevoix contributed for a large portion of the Saguenay gene pool (founder effect). Later migratory movements may have somehow differently influenced the evolution of each population. The Rimouski gene pool seems less homogeneous, although the contribution of some 17th century founders is also quite high in this region. Hence, hereditary diseases found in the Rimouski population might have originated from 17th century founders, but these founders may differ from those of the Saguenay or Charlevoix populations. Indeed, some important founders appeared more frequently in the Rimouski genealogies than in those of Charlevoix. Further analysis on the migratory patterns of the founders' descendants could provide useful information regarding this matter. Also, genealogical studies of other Quebec regions could enhance our knowledge on the heterogeneity of the Quebec gene pool and the potential spreading of certain hereditary diseases in the regional populations.

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Endnotes:

1. Preliminary results of this research were presented at the European Research Conference on "Inherited disorders and their genes in different European populations", Acquafredda di Maratea (Italy), 6-11 February 1998.
2. For further details regarding the selection and reconstruction of these genealogies, see Gagnon (1999).
3. Genealogical reconstructions do not go beyond the first immigrants. For this reason, kinship between 17th century founders was not taken into consideration (some founders may be brothers and sisters, cousins, etc.).

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