Genealogical Analysis of Maternal and Paternal Lineages in the Quebec Population

MARC TREMBLAY[1] AND HÉLÈNE VÉZINA[1]

Abstract The Quebec population is one of the rare populations of its size for which genealogical information is available for an uninterrupted period of almost four centuries. This allows for in-depth studies on the formation and evolution of a young founder population. Using data from two major population registers, in this study we focus on the maternal and paternal lineages (i.e., strictly female or male genealogical lines) that can be traced back within the Quebec genealogies. Through the analysis of these lineages it is possible to characterize the founders who transmitted to the contemporary population their mitochondrial (for females) and Y-chromosome (for males) DNA. The basic material consists of 2,221 ascending genealogies of subjects who married in the Quebec population between 1945 and 1965. On average, more than nine generations of ancestors were identified among the lineages. Analyses of maternal and paternal lineages show that the number of paternal founders is higher and their origins and genetic contributions are more variable than that of maternal founders, leading to a larger effective population size and greater diversity of Y chromosomes than of mtDNA. This is explained for the most part by differential migratory patterns among male and female founders of the Quebec population. Comparisons of sex-specific genetic contributions with total genetic contribution showed a strong correlation between the two values, with some discrepancies related to sex ratio differences among the founders' first descendants.

Paternal lines of descent correspond to the patronymic lineage (transmission of surnames) and to the transmission line of Y chromosomes, whereas female lineages correspond to the transmission line of mitochondrial DNA, which is passed from mothers to their children. These two genetic systems are widely studied in population genetics because they allow for a sex-based comparison of differential reproductive and migratory patterns throughout human evolutionary history (Kumar et al. 2006; Oota et al. 2001; Ségurel et al. 2008; Wilder et al. 2004; Wilkins 2006). Comparisons of paternal and maternal lines of descent in various populations have shown quite different pictures in terms of origins and diversity

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of Y-chromosome and mitochondrial DNA [see, for example, Kittles et al. (1999) for Finland, Helgason et al. (2003) for Iceland, Malyarchuk et al. (2004) for Russia, Tomàs et al. (2006) for the Balearic islands, Hünemeier et al. (2007) for Brazil, and Ségurel et al. (2008) for Central Asia]. Study of these sex-specific lineages based on genealogical data is particularly interesting because it allows for identification of founders who have transmitted, up to the contemporary population, their mitochondrial and Y-chromosome DNA through their female and male descendants, respectively.

In the present study we focus on maternal and paternal lineages in the Quebec, Canada, population. The Quebec population is a fruitful ground for such investigation, because it is one of the rare populations of its size for which genealogical information is available for a period of almost four centuries (up to 17 generations in certain cases). Based on the availability of population registers and knowledge gained from previous studies, a vast research program aimed at describing the genealogical structure of the Quebec population was initiated by the Interdisciplinary Research Group on Demography and Genetic Epidemiology (2009). Recent studies have revealed interesting findings about the geographic origins and genetic contributions of the founding ancestors of the population (Tremblay et al. 2008; Vézina et al. 2006). For example, results have shown that about 80% of the contemporary gene pool of Quebec French Canadians comes from approximately 3,000 founders who settled in Nouvelle-France during the 17th century (Vézina et al. 2006). Some of these early founders are present in the genealogical tree of almost every individual in the population, whereas others appear only sporadically. This kind of founder effect is not unique to Quebec. The populations of Iceland and Finland, for instance, also experienced founder effects (Helgason et al. 2003; Kittles et al. 1999). One of the consequences of founder effects is an elevated frequency of some mutations responsible for rare inherited diseases. After their introduction, these mutations were transmitted from generation to generation along lines of descent that can be traced back through the genealogical trees.

In this study we address new questions about the contribution of the founders of the Quebec population. Although results have been published on the total genetic contribution of male and female founders, little is known about their specific maternal and paternal contributions. For instance, what proportion of these founders appears in the maternal and paternal lines? What are the generational lengths of the maternal and paternal lineages? Can we assume that the geographic origins of the maternal and paternal founders are similar to those of all female and male founders? And, most important, do the paternal and maternal lineages display the same level of diversity? Do male and female founders differ in their frequency distribution and in their genetic contribution to the contemporary population?

The Quebec Population

The province of Quebec is located in the eastern part of Canada (Figure 1). Approximately 82% of its 7.8 million inhabitants speak French. Most of these

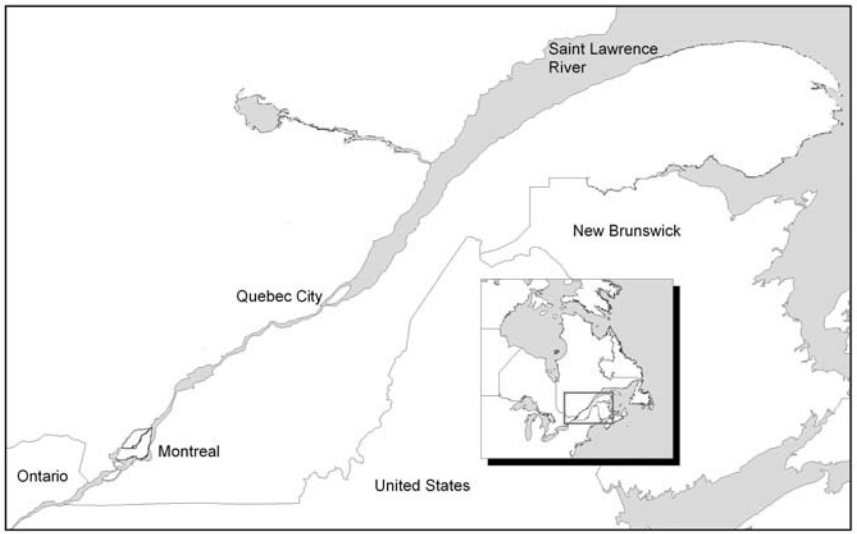


Figure 1. Location of the province of Quebec.

French Canadians are descendants of some 10,000 settlers who came from France during the 17th and 18th centuries (Charbonneau et al. 2000). The relative contribution of each of these founders to the Quebec population is highly variable depending on, among other factors, their period of arrival in North America (Heyer et al. 1997). Following the British conquest of 1759, the French Canadian population expanded in a context of relative isolation (mostly for geographic, linguistic, and religious reasons) and rapid growth, which contributed to the amplification of the consequences of the founder effect (Bouchard and De Braekeleer 1991; Henripin and Péron 1972), including an elevated frequency of some inherited disorders in this population (Laberge et al. 2005b; Scriver 2001). During the last part of the 18th century and for most of the 19th century, most immigrants to Quebec came from the British Isles and most of them settled in urban areas, such as Montreal and Quebec City (Beaujot and Kerr 2004; McInnis 2000). After the 19th century, the origins of the immigrants became more diversified, with many newcomers from southern and eastern Europe and, more recently, from Asia, Africa, South America, and the Caribbean (Girard 2008; Piché 2003). Although some of these newcomers have contributed to the French Canadian gene pool through marriage, barriers to these marriages have remained relatively important, especially outside the urban areas, for a long time. In a little more than three centuries, the Quebec population has increased by a factor of 520 (from 15,000 people at the end of the 17th century to 7.8 million today). Up to the beginning of the 20th century, this strong growth was explained mostly by high levels of reproduction among the descendants of the first pioneers (Charbonneau et al. 2000; Henripin and Péron 1972).

Another characteristic of the Quebec population rests on the existence of databases containing extensive demographic and genealogical information, which allows for detailed studies of founder effects and population structure. Various research projects in population genetics and historical demography have been conducted using the Quebec genealogical data, sometimes in combination with medical or genetic information (Bouchard et al. 1988; Heyer and Tremblay 1995; Laberge et al. 2005b; Scriver 2001). Their general aim is to understand and explain the role of demographic dynamics and population history in the contemporary frequency and distribution of gene variants in the population. Work conducted on inherited disorders has also relied on this approach to study the genealogies of affected individuals and document the modalities of introduction and diffusion of mutant alleles involved in these disorders [see, for instance, Mathieu et al. (1990) and Yotova et al. (2005) on myotonic dystrophy, Laberge et al. (2005a) on Leber hereditary optic neuropathy, Vézina et al. (2005) on breast cancer, and Plante et al. (2008) on mucolipidosis II].

Materials and Methods

Databases. Material for this study was obtained from the 2009 BALSAC population register (http://www.uqac.ca/balsac) and the 2009 Registre de Population du Québec Ancien (http://www.genealogie.umontreal.ca). These databases contain demographic and genealogical information on the population of Quebec since the beginning of European settlement in the early 17th century. They were constructed with material extracted mostly from Catholic parish records (baptisms and marriage and death certificates). The Registre de Population du Québec Ancien covers the 17th and 18th centuries and contains approximately 700,000 linked records, whereas the BALSAC register (still under construction) covers the 19th and 20th centuries with more than 2.5 million records (Bouchard 2008).

Genealogical Sample. Genealogical data were retrieved from a sample of 2,221 ascending genealogies of subjects who married in the Quebec population between 1945 and 1965 (Tremblay et al. 2008; Vézina et al. 2006). Selection of subjects was done randomly and reflects the geographic distribution of the Quebec population as portrayed by the 1956 Canadian census, right in the middle of the baby boom period. The genealogies were reconstructed as far back as the available sources would permit. For the most part, the genealogical branches go back to the 17th century. Data available from these genealogies include spouse's name, year and place of marriage, and place of origin for the immigrants. All known genealogical links between individuals appearing in the genealogies were established.

The genealogical sample contains more than 5 million occurrences of ancestors. Because most subjects share a certain number of ancestors, many of these ancestors appear more than once in the genealogies. The average number of occurrences per ancestor is 33, but there is a high variability among ancestors.

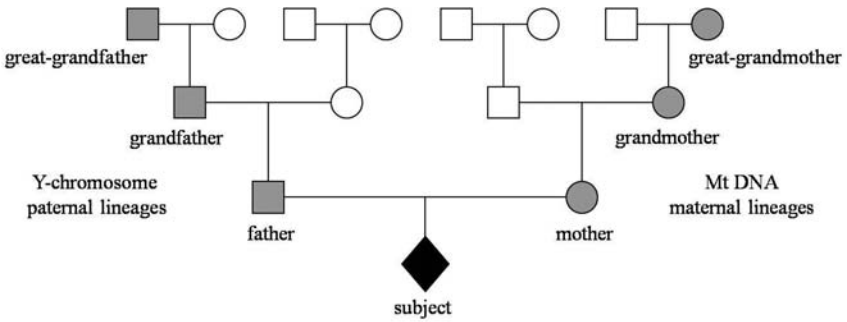


Figure 2. Maternal and paternal lineages: example with three generations of ancestors.

Nearly 15% of ancestors appear only once, whereas 1% of ancestors appear in more than 1,000 genealogies.

The completeness of the genealogies is a key indicator of the richness of the data, because it shows how far the genealogical information can be traced. The completeness represents the proportion of ancestors that have been identified at a given generation level, starting with the subject's parents (level 1). Summing all completeness values for each generation level gives the mean genealogical depth of the genealogies (Cazes and Cazes 1996). On average, the 2,221 genealogies have a depth of 9.3 generations, with some branches going back as far as 17 generations. Up to the seventh generation, the completeness of the genealogies is still more than 90%, meaning that at this level, 9 out of 10 ancestors have been identified. This genealogical depth is quite rare for such a large set of genealogies. After the ninth generation, the completeness drops rapidly because of the lack of information. These older generations correspond to most of the first immigrants who came from France during the 17th century (Tremblay et al. 2008; Vézina et al. 2006).

Maternal and Paternal Lineages. In all genealogical branches, founders are defined as the first ancestors who immigrated to Quebec. Among these, founders of maternal and paternal lineages are those who have been identified while following branches strictly through mothers or fathers. Figure 2 shows an example of maternal and paternal lineages, up to the third generation (great-grandparents). The length of maternal and paternal lineages was measured in terms of the number of generations from the subjects to the founders. If the great-grandparents of the subject in Figure 2 are both founders, then the length of these lineages is three generations.

Measurement of kinship between two subjects through maternal or paternal lineages was also defined in terms of the number of generations between the subjects and their common maternal or paternal ancestor (Figure 3). In this case,

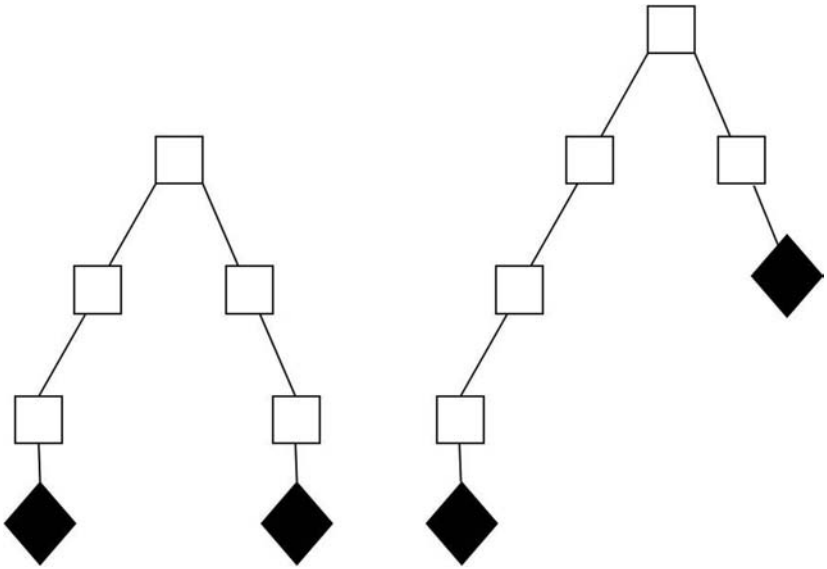


Figure 3. Examples of kinship links through paternal lineages.

kinship was calculated by adding the lengths of the lineages from the subjects to their common ancestor, minus 1. In other words, this corresponds to the number of ancestors that can be traced between the two subjects, following the paths connecting them to their common ancestor. Figure 3 shows two examples of kinship links with a value of 5.

We also measured the number of subjects to which each founder is linked directly through either women-only or men-only lineages. This number corresponds to the maternal or paternal genetic contribution of the founder to the population (the 2,221 subjects). Dividing this genetic contribution by the total number of subjects gives the proportion of mtDNA or Y chromosomes that came from this founder. These proportions were used to calculate mtDNA and Y-chromosome diversity indexes in the population, which in turn were used to estimate the effective population size for each system, following a method adapted from Mourali-Chebil and Heyer (2006). We also calculated the genetic contributions of maternal and paternal founders according to their origin and period of marriage. The origin of the founders corresponds to their place of birth, their place of marriage, or emigration. Last, for each founder, we compared total genetic contribution and maternal or paternal genetic contribution to see whether there was a correlation between these two values. Total genetic contribution was calculated using the method described by Vézina et al. (2006) and Tremblay et al. (2008). All measures were performed using the S-Plus based GENLIB software package (Interdisciplinary Research Group on Demography and Genetic Epidemiology 2009).

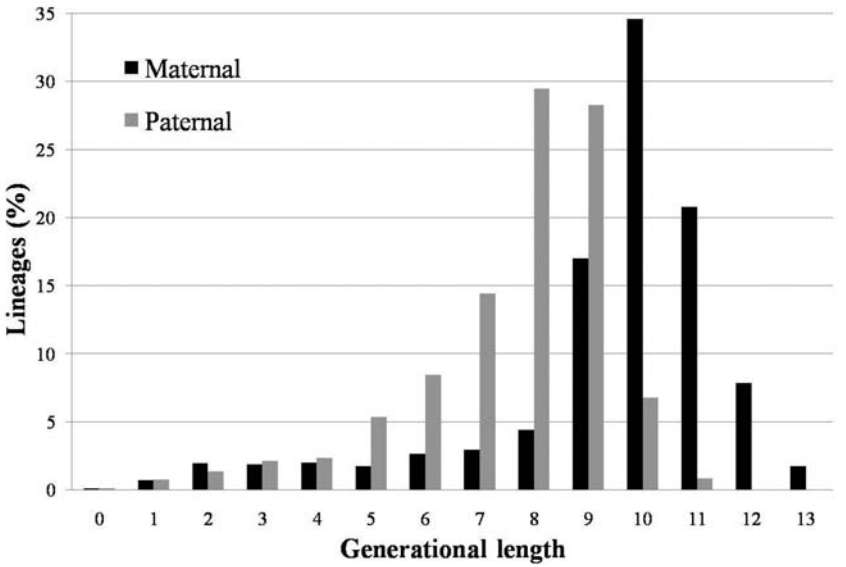


Figure 4. Distribution (%) of maternal and paternal lineages according to their generational length.

Results

Generational Length and Kinship Links. The distribution of the 2,221 maternal and paternal lineages according to their generational length is shown in Figure 4. In accordance with the completeness of the genealogies, most lineages reach more than seven generations. On average, though, maternal lineages (9.4 generations) are longer than paternal lineages (7.6 generations). Nearly two-thirds (64.8%) of maternal lineages have a length of 10 generations or more (with a maximum of 13 generations), compared to only 7.6% of paternal lineages (maximum of 11 generations). This is consistent with the fact that intergenerational intervals are usually longer between fathers and sons than between mothers and daughters (Fenner 2005; Helgason et al. 2003; Tremblay and Vézina 2000). On average, maternal intervals have a length of 29.5 years, compared to 33.9 years for paternal intervals (see Table 1). Hence for a similar time period there are more mother-daughter intervals than father-son intervals.

Most of the founders of maternal and paternal lineages are shared between two subjects or more. Indeed, 84% of the subjects have a common maternal ancestor with at least one other subject. This proportion is lower for paternal lineages (69%), indicating that paternal ancestry within the Quebec population is more diverse than maternal ancestry is. Again, the length of the kinship links varies greatly among and between maternal and paternal lineages (Figure 5). Most of the maternal kinship links (74%) contain 18–23 ancestors, whereas 76% of paternal

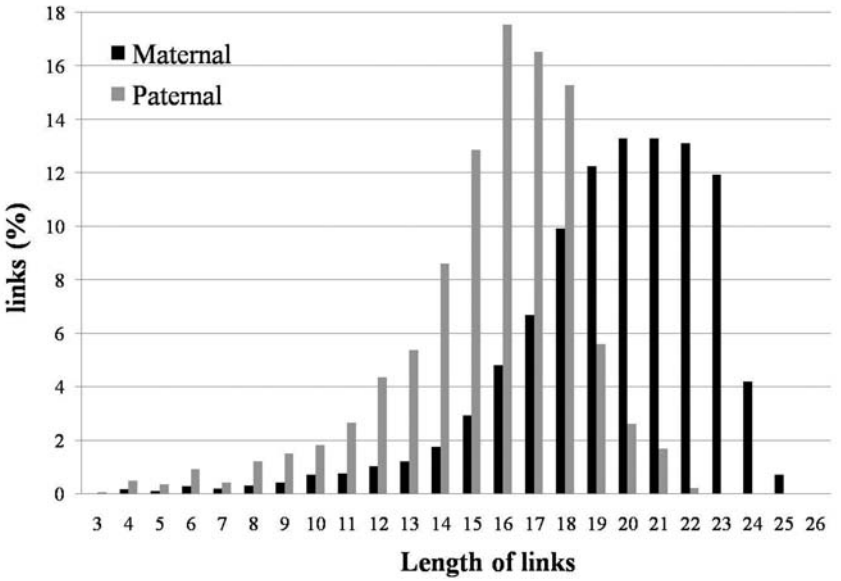


Figure 5. Distribution (%) of kinship links within maternal and paternal lineages.

kinship links have 14–19 ancestors. On average, 20 ancestors are found in the maternal kinship links and 16 are found on the paternal side.

Origins and Genetic Contribution of the Founders of Maternal and Paternal Lineages. We identified 768 maternal and 1,181 paternal founders in the 2,221 genealogies, indicating that, on average, maternal and paternal founders are linked to 2.9 and 1.9 subjects, respectively (Table 1). However, the distribution of founders according to the number of subjects to which they are linked is highly variable. Nearly 64% of paternal founders contribute to only one subject, compared to 53% of maternal founders. Most of the other founders (35% of paternal founders and 43% of maternal founders) are related to 2–10 subjects. Fifteen female founders transmitted their mtDNA to 15 subjects or more, and together they are linked to 17.4% of all lineages. Among them, the top two contributors are linked to nearly 5% of all subjects, indicating that 1 out of 20 subjects carries an mtDNA transmitted by one of these two women. In contrast, only 5 founders transmitted their Y chromosome to 15 subjects or more, and together they are linked to 4.9% of subjects. These proportions correspond to a diversity index of 0.99571 for mtDNA, compared to 0.99811 for Y-chromosome DNA, with effective population sizes of 3,140 and 7,304, respectively.

Founders with a low genetic contribution immigrated more recently on average than founders with a high genetic contribution, as shown by the average

Table 1. Distribution of Maternal and Paternal Founders According to Their Genetic Contribution, Their Mean Intergenerational Length, and Mean Number of Generations Between Them and the Subjects to Which They Are Linked

Genetic Contribution	*N*	*%*	*Mean Intergenerational Length (Years)*	*Mean Number of Generations*
Maternal founders				
1	410	53.4	30.1	6.3
2	120	15.6	29.5	9.2
3	74	9.6	29.6	9.5
4	51	6.6	29.2	9.8
5	28	3.6	29.2	10.1
6	14	1.8	29.1	10.2
7	10	1.3	29.8	9.7
8	9	1.2	30.1	10.2
9	9	1.2	29.3	10.0
10	12	1.6	29.3	10.3
11	6	0.8	29.6	10.5
12	6	0.8	29.1	10.1
13	3	0.4	29.8	10.3
14	1	0.1	29.6	10.1
15–19	6	0.8	29.3	10.5
20–29	5	0.7	28.8	10.8
30–49	2	0.3	29.0	11.8
50+	2	0.3	29.3	11.5
Total	768	100.0	29.5	9.4
Mean genetic contribution	2.9			
Diversity index	0.99571			
Effective size	3,140			
Paternal founders				
1	752	63.7	33.7	6.5
2	215	18.2	34.1	7.7
3	88	7.5	34.3	8.0
4	54	4.6	34.1	8.3
5	26	2.2	33.6	8.4
6	16	1.4	34.3	8.7
7	5	0.4	33.8	9.0
8	7	0.6	33.8	9.0
9	6	0.5	34.0	9.0
10	2	0.2	37.1	7.9
11	3	0.3	34.0	9.3
12	1	0.1	32.4	10.3
13	0	0.0	–	–
14	1	0.1	33.9	8.7
15–19	3	0.3	33.8	8.7
20–29	1	0.1	35.3	9.1
30–49	1	0.1	33.4	8.9
50+	0	0.0	–	–
Total	1,181	100.0	33.9	7.6
Mean genetic contribution	1.9			
Diversity index	0.99811			
Effective size	7,304			

Table 2. Distribution (%) of Founders According to Their Origin and Period of Marriage

Origin	*Before 1660*	*1660–1699*	*1700–1765*	*After 1765*	*All Periods*
Maternal founders					
France	23.1	43.5	1.9	3.5	72.0
British Isles	0.1	0.1	0.6	7.4	8.3
Other European	0.1	0.1	0.0	1.0	1.3
Acadia	0.0	0.3	9.3	4.4	14.0
Other American	0.1	0.7	1.0	2.5	4.4
All origins	23.5	44.8	12.8	18.8	100.0
Paternal founders					
France	14.0	44.5	19.3	3.8	81.7
British Isles	0.0	0.1	0.5	5.3	5.9
Other European	0.1	0.4	0.8	1.7	2.9
Acadia	0.0	0.0	4.9	2.6	7.5
Other American	0.0	0.0	0.7	1.2	1.9
All origins	14.1	45.0	26.3	14.6	100.0

number of generations separating the founders and the subjects. Correlation coefficients between the genetic contributions of maternal and paternal founders and the number of generations between them and the subjects are 0.4256 and 0.3118, respectively. On the other hand, the mean intergenerational length does not vary significantly according to genetic contribution ($r = –0.0626$ for maternal founders and $r = 0.0183$ for paternal founders).

The distributions of maternal and paternal founders according to their place of origin and period of marriage are shown in Table 2. As observed among all founders (Tremblay et al. 2008; Vézina et al. 2006), French origins are very important among founders of maternal and paternal lineages: 72.0% of maternal and 81.7% of paternal founders came from France. Most of these French founders were married before 1700, with a certain concentration between 1660 and 1699. This 40-year period corresponds to the peak of French pioneer immigration to Canada (Charbonneau et al. 2000). A greater proportion of paternal than maternal founders were married between 1700 and 1765, as a result of the military reinforcement that took place during the wars between the French and the British.

Founders from Acadia are the second most important group for both maternal and paternal founders. Their proportion among female founders is higher than that among male founders. Most of these Acadian founders are descendants of French pioneers who immigrated to Quebec after their deportation by the British authorities in 1755 (Dickinson 1994). Together, French and Acadian founders make up 86.0% of maternal founders and 89.2% of paternal founders. Founders from the British Isles and other European countries represent 9.6% of maternal founders and 8.8% of paternal founders.

Table 3. Genetic Contribution % of Founders According to Their Origin and Period of Marriage

Origin	*Before 1660*	*1660–1699*	*1700–1765*	*After 1765*	*All Periods*
Maternal genetic contribution					
France	45.5	40.8	0.9	1.1	88.3
British Isles	0.0	0.1	0.3	2.4	2.8
Other European	0.2	0.1	0.0	0.3	0.7
Acadia	0.0	0.2	4.6	1.6	6.5
Other American	0.0	0.3	0.6	0.8	1.7
All origins	45.9	41.4	6.4	6.3	100.0
Paternal genetic contribution					
France	28.0	44.8	13.1	2.1	88.0
British Isles	0.0	0.0	0.6	3.0	3.6
Other European	0.3	0.3	0.6	1.0	2.3
Acadia	0.0	0.0	3.5	1.5	5.0
Other American	0.0	0.0	0.5	0.6	1.1
All origins	28.3	45.1	18.3	8.3	100.0

We also examined variability of the genetic contributions of maternal and paternal founders according to the founders' origins and period of marriage (Table 3). The results show that proportionally the French genetic contribution is higher than the frequency of French founders. French founders account for 88.3% of maternal lineages and 88.0% of paternal lineages. All other groups of founders have a genetic contribution lower than their frequency. Therefore, on average, maternal and paternal founders from France have a higher genetic contribution than other founders do. Indeed, the mean genetic contribution of French maternal founders is equal to 3.8 subjects, compared to 1.6 subjects or less for the other maternal founders. The mean genetic contribution is lower among French male founders than among female founders (2.1 subjects), but it is still higher than among founders from other origins (less than 1.5 subjects). Also, earlier founders have on average a much higher genetic contribution than the more recent founders. Founders who married before 1660 have a proportional genetic contribution twice as high as their frequency among all founders. The two most prolific female founders are among these early French pioneers. They married in France in 1615 and 1620, respectively. On the other hand, founders who married after 1700 have a genetic contribution that is lower than their frequency, and this is especially noticeable among maternal founders.

Figure 6 shows the relative genetic contribution of French founders according to their province of origin. These provinces correspond to the French administrative regions during the 17th and 18th centuries (up to the French Revolution). Paternal founders (Figure 6b) came from all regions of France, but maternal founders (Figure 6a) did not, pointing again to a higher diversity in paternal lineages. In both cases, however, a concentration of founders from the northern and western provinces is perceptible. Île-de-France, Normandie, Perche, Poitou,

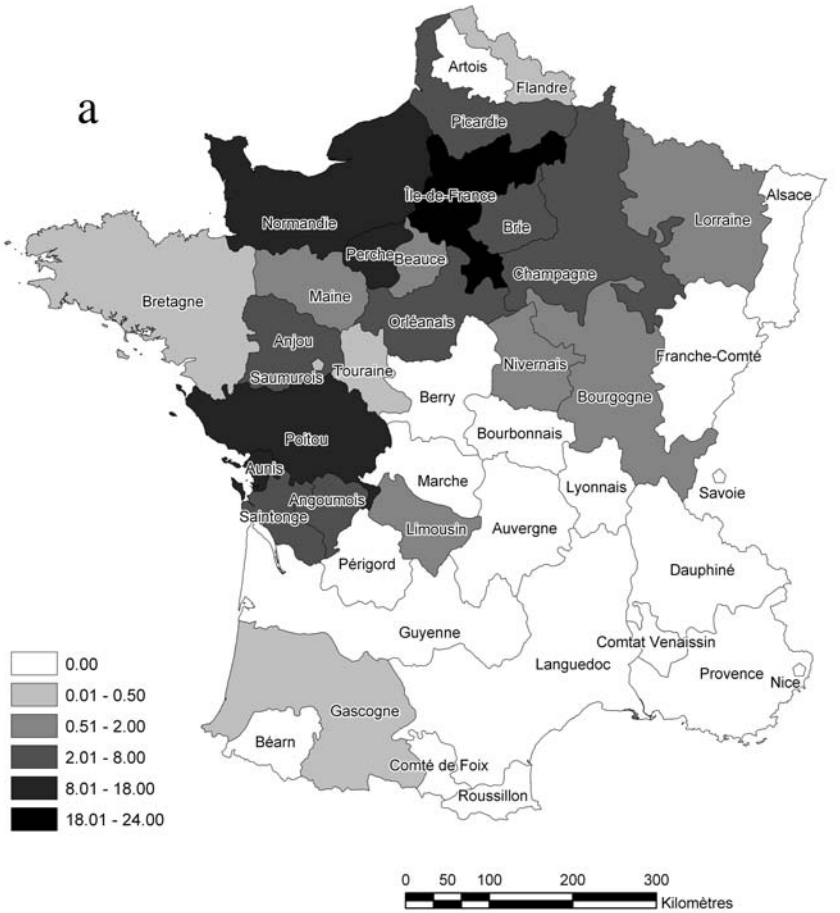
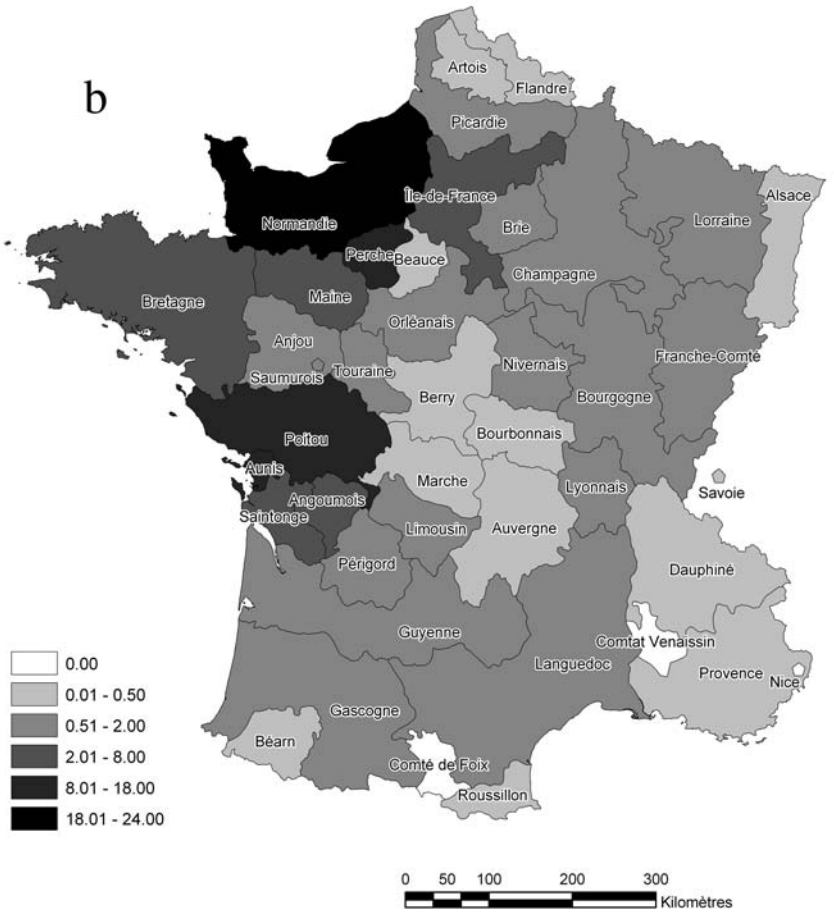


Figure 6. Genetic contribution (%) of French maternal and paternal founders according to their province of origin. (a) Maternal founders; (b) paternal founders (facing page).

and Aunis (a small region on the western coast) are among the most important provinces in terms of maternal and paternal genetic contributions, accounting for 77% and 61%, respectively, of the French genetic contribution. The province of Perche (south of Normandie) shows a larger genetic contribution than expected, ranking third among paternal founders and fourth among maternal founders. This small province provided relatively few immigrants to Canada, but most of them came early and had many descendants (Charbonneau et al. 1993). Nearly 90% of the Perche maternal and paternal founders were married before 1660, which is by far the largest proportion among all origins. The top maternal and paternal contributors are both from this region.



To verify whether those female and male founders with a high contribution to maternal and paternal lineages also have a high genetic contribution through all genealogical paths, we compared both types of contributions (Figure 7). The proportions of total genetic contribution and maternal or paternal genetic contribution of each founder are shown using the same genealogical sample. Comparison of the two sets of values indicates that the most prolific maternal (Figure 7a) and paternal (Figure 7b) founders also have a strong genetic contribution to the population, with a few exceptions ($r = 0.7383$ for maternal founders and $r = 0.6318$ for paternal founders). Some founders with a high contribution to maternal or paternal lines show a relatively low total genetic contribution, whereas other founders who appear less frequently among maternal or paternal lines display a total genetic contribution that is much higher than expected.

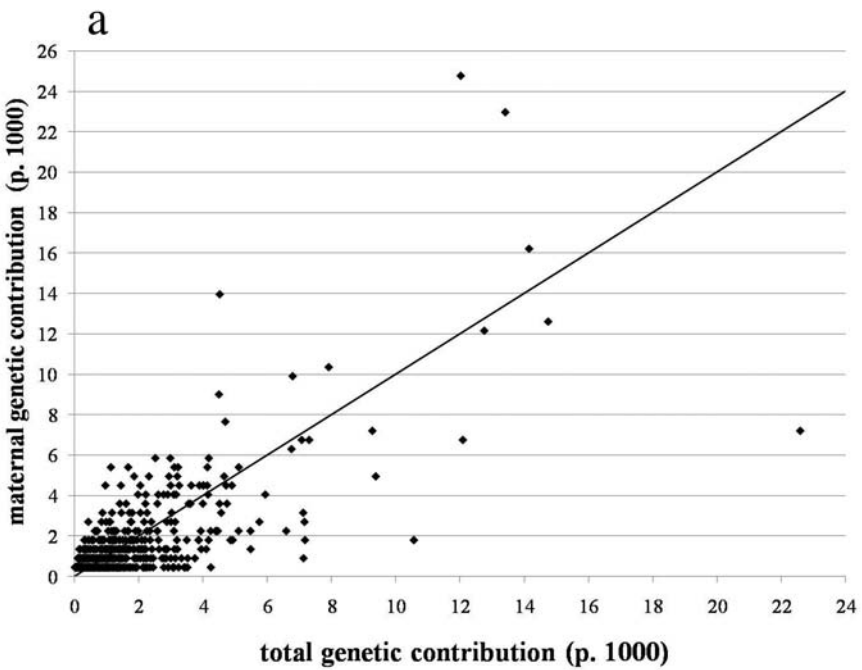
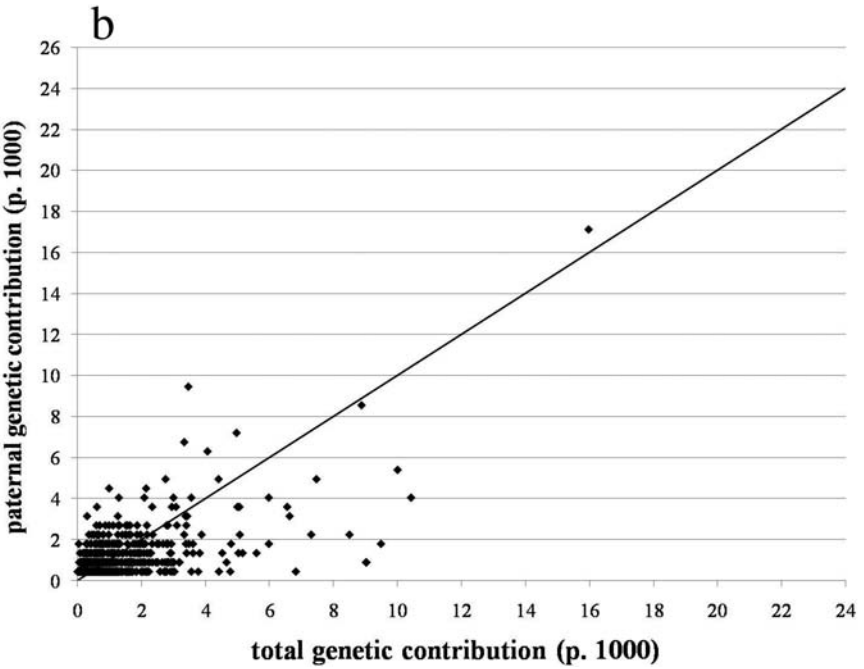


Figure 7. Comparison of maternal and paternal genetic contributions of founders with their total genetic contribution. (a) Maternal founders; (b) paternal founders (facing page).

Discussion

In this study the immigrant founders of maternal and paternal lineages in a Quebec population sample were identified and compared to detect sex-specific characteristics in terms of period of arrival, place of origin, and genetic contribution to the contemporary population. Results show that the number of paternal founders is higher than the number of maternal founders and that the origins and genetic contributions of paternal founders are more variable than that of maternal founders, leading to a larger effective population size and greater diversity of Y chromosomes compared with mtDNA. In both cases, most of the founders originated from France, but paternal founders came from all regions of France, whereas the origins of maternal founders are located in a smaller number of provinces. Because French founders settled earlier in Canada, it gave them an opportunity to leave more descendants, and this is reflected in their genetic contribution being higher than their actual frequency among founders.

Completeness of the genealogical material has proved to be a key factor for this type of analysis, especially when kinship links between the subjects are



concerned (Chaix et al. 2004). With a completeness of more than 90%, common ancestors are extremely rare before the sixth generation. After the ninth generation, nearly all subjects have a common ancestor with at least one other subject (Tremblay et al. 2008; Vézina et al. 2006). Therefore most kinship links become detectable only if the genealogical information is available for at least seven or eight generations. These distant kinship links appeared to be more frequent in maternal lineages than in paternal lineages, indicating again a higher diversity among paternal founders.

One factor that can explain the higher number of kinship links among maternal lineages is that, because of shorter intergenerational intervals, these lineages are longer in terms of number of generations than paternal lineages, allowing for more kinship links to appear along the genealogical branches. Similar results were observed in the Icelandic population (Helgason et al. 2003). The lower diversity of maternal lineages can also reflect a system of matrilocality, as observed in other populations (Thomas et al. 2002; Tomàs et al. 2006). However, in the Quebec population, this lower diversity among maternal lineages is mainly explained by differential migratory and matrimonial behavior at the beginning of settlement. Indeed, many more men than women were recruited into the colony, and thus there were more marriages between immigrant men

and nonimmigrant women than between immigrant women and nonimmigrant men (Charbonneau et al. 2000). As a result, the total number of male founders is three times higher than the number of female founders (Vézina et al. 2006). Among these male founders, approximately 23% were found in the paternal lineages, compared to 45% of the female founders. In a study on the genetic contribution of female founders in the Saguenay population, Heyer (1995b) found that 60% of all female founders with at least one descendant in the contemporary population transmitted at least one copy of their mtDNA. In this case, the mean genealogical depth (<4 generations) was much lower than that of our maternal lineages (9.4 generations), but this shorter genealogical length did not necessarily imply a higher probability of survival of lineages (Heyer 1995b). In a recent study on ancient mtDNA haplotypes in the Icelandic population, Helgason et al. (2009) found that approximately 40% of mtDNA from early medieval female founders (>30 generations) was still present in the contemporary population. However, our results must be taken with precaution because the number of lineages under study (2,221) is smaller than the total number of founders (6,808) identified through all genealogical branches. Although it is safe to assume that all important founders must have been identified through these genealogies, a larger genealogical sample would be necessary to provide more information on that matter.

A relatively small number of early founders have a high genetic contribution to the contemporary population, which is expected in a population that has experienced founder effects. Founder effects and their consequences have also been investigated in Iceland (Helgason et al. 2003), Ireland (Moore et al. 2006), and the Saguenay region of Quebec (Heyer 1995a) and among the Amish of Lancaster County in Pennsylvania (Pollin et al. 2007), although with variable results regarding the impact of these effects, partly because of important differences in the data and methodology used. For example, Moore et al. (2006) found that a single ancestor from the early Middle Ages explains 20% of the contemporary paternal lineages of northwestern Ireland. In the Amish of Pennsylvania, the founder effect is much more recent: 21 male founders born between the late 17th century and the early 20th century account for 99% of the paternal lineages of this community (Pollin et al. 2007). In the Quebec population, nearly 78% of all lineages can be traced back to some 1,100 founders who came during the 17th century. A quarter of female lineages are explained by only 29 founders, whereas 71 founders explain the same proportion of male lineages. Such differences lead to lower diversity and effective population size for mtDNA compared to Y-chromosome DNA. In this study, the ratio of estimated effective population sizes is 2.3 in favor of paternal lineages.

Comparisons between sex-specific genetic contributions and total genetic contribution showed a relatively strong correlation between the two measures, although there are some discrepancies. These discrepancies can be explained for the most part by variations of the sex ratio among the founders' first descendants. For instance, male founders with more boys than girls among their children and

grandchildren will have a greater chance of appearing more frequently along paternal lines of descent than among other lines. Conversely, male founders who had more girls than boys among their first descendants will likely have a lower paternal contribution compared with their total genetic contribution.

Analysis of geographic origins has shown that most male and female founders came from France. These French founders account for 88% of the paternal and maternal genetic contributions in the Quebec population, a proportion similar to that of the total genetic contribution from French founders (Vézina et al. 2006). However, one must keep in mind that our genealogical sample was built from Catholic records, thus excluding individuals from other religious confessions. According to Henripin and Péron (1972), about 88% of the Quebec population was Catholic during the period that the subjects married. Hence 12% of the population is not represented in our sample, but this does not mean that all founders from other religious groups do not appear in the genealogical data. Indeed, such founders may have been traced if some of their descendants married in the Catholic Church, which is most certainly the case for the earliest founders. Nevertheless, the contribution of non-Catholic founders (such as British and German Protestants) remains underestimated in our results.

Differences in the diversity and origins of male and female lineages were detected in other American populations that have experienced European colonization, such as Cuba (Mendizabal et al. 2008), Brazil (Carvalho-Silva et al. 2001), and Colombia (Carvajal-Carmona et al. 2000). In these populations native women rather than native men contributed largely to the gene pool. In Quebec, however, the native population was not demographically important when European colonization took place during the 17th century, and there seems to have been little admixture between the two populations (Charbonneau et al. 2000). Thus the genetic contribution of native Americans to the contemporary Quebec population is low, at least according to parish registers and genealogical data (less than 1% of maternal and paternal lineages). Ongoing research on the regional structure of the Quebec gene pool based on an approach combining genealogical and molecular information should bring new insights into this matter (Moreau et al. 2009).

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Literature Cited

Beaujot, R., and D. Kerr. 2004. *Population Change in Canada*, 2nd ed. Oxford, U.K.: Oxford University Press.

Bouchard, G. 2008. *Projet BALSAC: Rapport Annuel 2007–2008*. Chicoutimi, Canada: Projet BALSAC.

Bouchard, G., and M. DeBraekeleer. 1991. *Histoire d'un génome: Population et génétique dans l'est du Québec*. Québec, Canada: Presses de l'Université du Québec.

Bouchard, G., C. Laberge, and C. R. Scriver. 1988. Reproduction démographique et transmission génétique dans le nord-est de la province de Québec (18e–20es.). *Eur. J. Popul. Rev. Eur. Demogr.* 4:39–67.

Carvajal-Carmona, L. G., I. D. Soto, N. Pineda et al. 2000. Strong Amerind/white sex bias and a possible Sephardic contribution among the founders of a population in northwest Colombia. *Am. J. Hum. Genet.* 67:1287–1295.

Carvalho-Silva, D. R., F. R. Santos, J. Rocha et al. 2001. The phylogeography of Brazilian Y-chromosome lineages. *Am. J. Hum. Genet.* 68:281–286.

Cazes, M. H., and P. Cazes. 1996. Comment mesurer la profondeur généalogique d'une ascendance? *Population* 51(1):117–140.

Chaix, R., F. Austerlitz, T. Khegay et al. 2004. The genetic or mythical ancestry of descent groups: Lessons from the Y chromosome. *Am. J. Hum. Genet.* 75:1113–1116.

Charbonneau, H., B. Desjardins, A. Guillemette et al. 1993. *The First French Canadians: Pioneers in the St. Lawrence Valley.* Newark, DE: University of Delaware Press and Associated University Presses.

Charbonneau, H., B. Desjardins, J. Légaré et al. 2000. The population of the St.-Lawrence Valley, 1608–1760. In *A Population History of North America*, M. R. Haines and R. H. Steckel, eds. Cambridge, U.K.: Cambridge University Press, 99–142.

Dickinson, J. A. 1994. Les réfugiés acadiens au Canada, 1755–1775. *Canad. Stud.* 37:51–61.

Fenner, J. N. 2005. Cross-cultural estimation of the human generation interval for use in genetics-based population divergence studies. *Am. J. Phys. Anthropol.* 128(2):415–423.

Girard, C. 2008. *Le bilan démographique du Québec*, 2008 ed. Québec, Canada: Institut de la Statistique du Québec.

Helgason, A., B. Hrafnkelsson, J. R. Gulcher et al. 2003. A population-wide coalescent analysis of Icelandic matrilineal and patrilineal genealogies: Evidence for a faster evolutionary rate of mtDNA lineages than Y chromosomes. *Am. J. Hum. Genet.* 72:1370–1388.

Helgason, A., C. Lalueza-Fox, S. Ghosh et al. 2009. Sequences from first settlers reveal rapid evolution in Icelandic mtDNA pool. *PLoS Genet.* 5(1):e1000343.

Henripin, J., and Y. Péron. 1972. The demographic transition of the Province of Quebec. In *Population and Social Change*, D. V. Glass and R. Revelle, eds. London: Edward Arnold, 213–231.

Heyer, E. 1995a. Genetic consequences of differential demographic behavior in the Saguenay region, Québec. *Am. J. Phys. Anthropol.* 98:1–11.

Heyer, E. 1995b. Mitochondrial and nuclear genetic contribution of female founders to a contemporary population in northeast Quebec. *Am. J. Hum. Genet.* 56:1450–1455.

Heyer, E., and M. Tremblay. 1995. Variability of the genetic contribution of Quebec population founders associated to some deleterious genes. *Am. J. Hum. Genet.* 56:970–978.

Heyer, E., M. Tremblay, and B. Desjardins. 1997. Seventeenth-century European origins of hereditary diseases in the Saguenay population (Quebec, Canada). *Hum. Biol.* 69:209–225.

Hünemeier, T., C. Carvalho, A. R. Marrero et al. 2007. Niger-Congo speaking populations and the formation of the Brazilian gene pool: mtDNA and Y-chromosome data. *Am. J. Phys. Anthropol.* 133(2):854–867.

Interdisciplinary Research Group on Demography and Genetic Epidemiology. 2009. *GRIG: Groupe de recherche interdisciplinaire en démographie et épidémiologie génétique*. http://www.uqac.ca/grig.

Kittles, R. A., A. W. Bergen, M. Urbanek et al. 1999. Autosomal, mitochondrial, and Y-chromosome DNA variation in Finland: Evidence for a male-specific bottleneck. *Am. J. Phys. Anthropol.* 108:381–399.

Kumar, V., B. T. Langstiehl, K. V. Madhavi et al. 2006. Global patterns in human mitochondrial DNA and Y-chromosome variation caused by spatial instability of the local cultural processes. *PLoS Genet.* 2(4):e53.

Laberge, A.-M., M. Jomphe, L. Houde et al. 2005a. A "Fille du Roy" introduced the T14484C Leber hereditary optic neuropathy mutation in French Canadians. *Am. J. Hum. Genet.* 77:313–317.

Laberge, A.-M., J. Michaud, A. Richter et al. 2005b. Population history and its impact on medical genetics in Quebec. *Clin. Genet.* 68:287–301.

Malyarchuk, B., M. Derenko, T. Grzybowski et al. 2004. Differentiation of mitochondrial DNA and Y chromosomes in Russian populations. *Hum. Biol.* 76(6):877–900.

Mathieu, J., M. De Braekeleer, and C. Prévost. 1990. Genealogical reconstruction of myotonic dystrophy in the Saguenay-Lac-Saint-Jean area (Québec, Canada). *Neurology* 40:839–842.

McInnis, M. 2000. The population of Canada in the nineteenth century. In *A Population History of North America*, M. R. Haines and R. H. Steckel, eds. Cambridge, U.K.: Cambridge University Press, 371–432.

Mendizabal, I., K. Sandoval, G. Berniell-Lee et al. 2008. Genetic origin, admixture, and asymmetry in maternal and paternal human lineages in Cuba. *BMC Evol. Biol.* 8:213.

Moore, L. T., B. McEvoy, E. Cape et al. 2006. A Y-chromosome signature of hegemony in Gaelic Ireland. *Am. J. Hum. Genet.* 78:334–338.

Moreau, C., H. Vézina, V. Yotova et al. 2009. Genetic heterogeneity in regional populations of Quebec: Parental lineages in the Gaspé Peninsula. *Am. J. Phys. Anthropol.* 139(4):512–522.

Mourali-Chebil, S., and E. Heyer. 2006. Evolution of inbreeding coefficients and effective size in the population of Saguenay Lac-St.-Jean (Québec). *Hum. Biol.* 78(4):495–508.

Oota, H., W. Settheetham-Ishida, D. Tiwawech et al. 2001. Human mtDNA and Y-chromosome variation is correlated with matrilocal versus patrilocal residence. *Nat. Genet.* 29:20–21.

Piché, V. 2003. Un siècle d'immigration au Québec: De la peur à l'ouverture. In *La démographie québécoise: Enjeux du XXI^e siècle*, V. Piché and C. Le Bourdais, eds. Montréal, Canada: Les Presses de l'Université de Montréal, 225–263.

Plante, M., S. Claveau, P. Lepage et al. 2008. Mucolipidosis II: A single causal mutation in the N-acetylglucosamine-1-phosphotransferase gene (*GNPTAB*) in a French Canadian founder population. *Clin. Genet.* 73:236–244.

Pollin, T. I., D. J. McBride, R. Agarwala et al. 2007. Investigations of the Y chromosome, male founder structure and Y-STR mutation rates in the Old Order Amish. *Hum. Hered.* 65:91–104.

Scriver, C. R. 2001. Human genetics: Lessons from Quebec populations. *Annu. Rev. Genom. Hum. Genet.* 2:69–101.

Ségurel, L., B. Martínez-Cruz, L. Quintana-Murci et al. 2008. Sex-specific genetic structure and social organization in Central Asia: Insights from a multilocus study. *PLoS Genet.* 4(9):e1000200.

Thomas, M. G., M. E. Weale, A. L. Jones et al. 2002. Founding mothers of Jewish communities: Geographically separated Jewish groups were independently founded by very few female ancestors. *Am. J. Hum Genet.* 70(6):1411–1420.

Tomàs, C., G. Jiménez, A. Picornell et al. 2006. Differential maternal and paternal contributions to the genetic pool of Ibiza Island, Balearic Archipelago. *Am. J. Phys. Anthropol.* 129(2):268–278.

Tremblay, M., and H. Vézina. 2000. New estimates of intergenerational time intervals for the calculation of age and origins of mutations. *Am. J. Hum Genet.* 66:651–658.

Tremblay, M., H. Vézina, B. Desjardins et al. 2008. Distant kinship and founder effects in the Quebec population. In *Kinship and Demographic Behavior in the Past*, T. Bengtsson and G. P. Mineau, eds. New York: Springer, 259–277.

Vézina, H., F. Durocher, M. Dumont et al. 2005. Molecular and genealogical characterization of the R1443X *BRCA1* mutation in high-risk French-Canadian breast/ovarian cancer families. *Hum. Genet.* 117(2–3):119–132.

Vézina, H., M. Tremblay, B. Desjardins et al. 2006. Origines et contributions génétiques des fondatrices et des fondateurs de la population québécoise. *Cah. Quebec. Demogr.* 34(2):235–258.

Wilder, J. A., Z. Mobasher, and M. F. Hammer. 2004. Genetic evidence for unequal effective population sizes of human females and males. *Mol. Biol. Evol.* 21:2047–2057.

Wilkins, J. F. 2006. Unraveling male and female histories from human genetic data. *Curr. Opin. Genet. Devel.* 16:611–617.

Yotova, V., D. Labuda, E. Zietkiewicz et al. 2005. Anatomy of a founder effect: Myotonic dystrophy in northeastern Quebec. *Hum Genet.* 117:177–187.