Retracing Prehistoric Population Events in Finland Using Simulation

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Abstract:
Due to Finland's low population density, geographical location and gradual settlement history, the country has been a genetic isolate. Genetic diversity is still slightly reduced among the Finns, especially in the eastern part of the country. This diversity reduction and the specific 'Finnish Disease Heritage' are explained by founder effects and bottlenecks and there is archaeological evidence for profound fluctuation of population sizes in the past. Here, we apply forward-time population simulations including archaeologically justified population size estimates to detect past demographic events. Our results indicate that simulation models beginning with serial founder effects have clearly reduced genetic diversity in early prehistory. Compared with our previous simulations, the gender-specific migration brings the simulated genetic diversity closer to the observed contemporary genetic diversity in Finland. Additionally, the simulation scenarios with temperate constant gene flow produce Y chromosomal diversity measures similar to those observed in present-day Finnish population.

Keywords:
Prehistoric Population, Population Simulation, Population Bottleneck, Stone Age, Early Metal Period, Finland

1. Introduction

The first pioneers settled in the Finnish region circa 11,000 BP following the retreat of the continental ice sheet after the last glaciation. According to archaeological finds, the region has been continuously inhabited ever since. Finland's prehistoric population events and settlement history have been studied by a number of archaeologists (e.g. Siiriäinen 1981; Meinander 1984; Nuñez, M. 1987; Huurre 1990, 2001; Lavento 1997, 1998, 2001; Carpelan 1999a, 1999b; Edgren 1999; Halinen 1999, 2005; Nuñez & Okkonen 1999; Mökkönen 2002, 2011; Pesonen 2002, 2005; Takala 2004; Rankama & Kankaanpää 2008; Saipio 2008; Tallavaara, Pesonen & Oinonen 2010; Tallavaara & Seppä 2012).

On the other hand, geneticists have carried out numerous studies concerning the population genetics and the origins of Finns (e.g. Nevanlinna 1972, Cavalli-Sforza et al. 1994; Sajantila et al. 1995, 1996; Lahermo et al. 1996,1998; Lahermo 1998; De la Chapelle & Wright 1998; De la Chapelle 1999; Kittles et al. 1998, 1999; Savontaus & Lahermo 1999; Norio 2000, 2004; Peltonen et al. 2000; Varilo et al. 2000, 2003; Kere 2001; Uimari et al. 2005, Lappalainen et al. 2006, 2008; Lappalainen 2009; Salmela et al. 2006, 2008; Service et al. 2006, Hedman et al. 2007; Hannelius et al. 2008; Jakkula et al. 2008; McEvoy et al. 2009; Palo et al. 2009). Finns are, when compared to West European populations, an outlier (Lao et al. 2008). Especially in the male-mediated Y chromosome diversity is reduced and there are significant differences between eastern and western Finland. The genetic diversity reduction and the specific 'Finnish Disease Heritage' (Norio 2003a, 2003b, 2003c), the enrichment of rare endemic genetic diseases in Finland, is explained by a profound founder effect or a bottleneck in the past.

Adequately preserved organic remains from the Mesolithic and Neolithic yielding DNA are practically non-existent in Finland due to the naturally acidic soil. Thus, we cannot obtain direct evidence of haplotypes or diversities in DNA of the ancient inhabitants. Therefore, we combine knowledge from two different fields of science to reconstruct prehistoric demographic events: population size estimates based on archaeological data and population genetic simulations. Disentangling past population events is a complex task that benefits from the synthesis of

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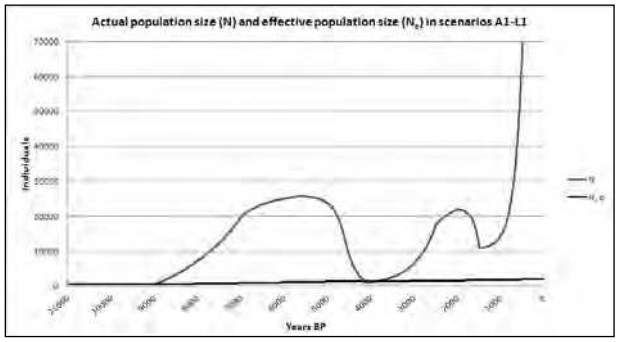

***Figure 1**. The actual total population size (N, dark grey) and theoretical effective population size (N_e, black). The effective population size has been calculated as the harmonic mean of the actual population size.*

archaeological and genetic data. In this contribution we build upon our previous work (Sundell et al. 2010) and evaluate the effects of an initial founder effect vs. serial founder effects, internal and gender-specific migrations as well as migration waves and a Neolithic population bottleneck on the Finnish gene pool.

1.1 Archaeological background

When assessing population size the principal assumption is that the archaeological signal, evidenced by e.g. radiocarbon date distribution, correlates with the population size: the stronger the detected archaeological signal is, the larger the population that left the signal has been. This does not yield an absolute value of the population size at a given time but, instead, estimates the relative population sizes between consecutive time periods.

The archaeological records from all of the excavations conducted in Finland have been documented in the Registry of Ancient Monuments, a national database of the National Board of Antiquities (http://kulttuuriymparisto.nba.fi/netsovellus/rekisteriportaali/mjreki/read/asp/r_default.aspx).

According to the Registry there are (in June 2012) 10,150 dwelling sites on mainland Finland that belong to the Stone Age (ca. 8800-1800 calBC) while only 556 belong to the Early Metal Period (1800 calBC -1 AD). There seems to be a clear difference between the numbers of settlement sites in inland culture when turning from the Neolithic to the Early Metal Period (Lavento 1997, 2001; Saipio 2008). The Early Metal Period dwelling sites are usually smaller and spatially differently distributed than in the previous period. The small number of structures found and the smaller sizes of sites could suggest temporary sites, possibly associated with hunter-gathering population and their seasonal settlement model. According to the archaeological signal, the number of inhabitants severely declined from the periods when large villages were settled in Saimaa, ca. 2500-1800 calBC (Lavento 2001). Although it can be argued that Early Metal Period dwelling sites are more difficult to find, the taphonomic processes (Surovell and Brantingham 2007; Surovell et al. 2009) would, however, have more impact on older Stone Age sites and should cause a sampling bias favouring the Early Metal Period sites.

Despite the uncertainties in defining what constitutes a dwelling site, the difference between the numbers of Stone Age and Early Metal Period sites appears substantial. The above-mentioned facts may thus reflect a genuine decrease in population size, in genetic terms a bottleneck. The archaeological evidence (see above) suggests that this may have been especially notable in eastern Finland between the Late Neolithic and the Early Metal Period.

In addition to the discrepancy between the numbers of dwelling sites from preceding eras, archaeological evidence tells about other demographic processes which can be used as prior knowledge in the simulations, e.g. possible migration from neigbouring areas. The spread of Typical Comb Ware (TCW) culture into Finland ca. 4000-3500 calBC is apparent and brings significant changes into archaeological finds. Together with a novel style of ceramics, there is a notable rise in the frequency of new raw materials such as flint and amber as well as a new house building tradition (e.g. Vuorinen 1982; Meinander 1984; Carpelan 1999a; Halinen 1999; Pesonen 2002; Edgren 2007). Furthermore, the population reaches a maximum at that time (Tallavaara et al. 2010).

The population peak is also evident through a Bayesian spatial analysis of archaeological record from the time period (Kammonen et al. 2012). Moreover, the weaker archaeological signal following the population peak at 4000-3500 calBC indicates a subsequent decline of the population. This has been

rendered as proof of a plausible Neolithic population bottleneck (Lavento 2001, Sundell et al. 2010) in Finland. Additionally, the spread of Corded Ware (CW) culture (ca. 2900-2300 calBC) has influenced the repertory of archaeological finds in the country (e.g. Edgren 1999; Carpelan 1999a). Again, a new style in ceramics together with new artifacts and new settlement patterns as well as burial practices.

1.2 Population genetic simulations

Population genetic simulation tools can be used to study the effects of population demography on genetic diversity over centuries or even millennia. The data sets simulated under different population historic scenarios can be contrasted with the observed genetic variation at the studied loci and thus support or refute certain scenarios.

In our simulation, each individual carries a mitochondrial genome and, if male, also a Y chromosome. These genomes are affected by similar evolutionary forces as natural chromosomes: they are transmitted from generation to generation, they are prone to mutate and their frequencies drift by chance. However, when contrasted to autosomal chromosomes, these molecules are haploid and uniparentally inherited, and as a consequence have lower molecular effective sizes. Hence they respond more rapidly to changes in population demography than autosomes. In addition, characteristic demographic processes e.g. population growth and decline, population subdivision and migration, can be incorporated into the models.

Our simulations begin at 11,000 BP when the first postglacial pioneers settled the country. The population growth is simulated according to the strength of the archaeological signal (Tallavaara et al. 2010). We employ two archaeologically justified bottlenecks: one at 4100-3800 BP and another at 1500-1300 BP. We split the population into geographic sub-populations, add gender-specific internal migration as well as migration waves and constant gene flow from neighbouring populations, compatible with archaeological phenomena.

2. Materials and Methods

We used simuPOP (Peng & Kimmel 2005), a forward-time population genetics simulation environment. Forward simulations can be used to create virtual populations which are then simulated through entire simplified model histories. SimuPOP consists of a number of components from which users assemble a suitable simulator. These components are operated through Python script files.

The simulation moves forward in ten-year-steps. The simulated population is age-structured i.e. generations overlap. Reproductive ages are set at 20-60 years for males and 20-40 years for females. The maximum lifespan is 60 years and the natural mortality rate is 15 % per ten years. The mating of individuals as well as the selection of those individuals who die is based on random sampling. Each mating produces 1 to N offspring according to a zero-truncated Poisson distribution ($\lambda = 2$). Offspring is produced until it, together with the surviving part of the population, reaches the population size at the next simulation step.

The simulations focus on mitochondrial and Y chromosomal variation. Mitochondrial DNA (mtDNA) is maternally inherited and simulated as a 631 bp DNA sequence, corresponding to HVS-I and HVS-II of the mtDNA control region. Any nucleotide at any position can mutate creating new variation. The mutation rates (μ) follow the 2-parameter model of Kimura (1980), distinguishing between transitions (μ = 0.00000128/locus/10 yrs) and transversions (μ = 0.00000064/locus/10 yrs). The Y chromosome is simulated as a haplotype consisting of alleles from linked STR loci, each allele expressing the numbers of short tandem repeats per locus. The mutation model is stepwise, thus new variation is introduced by mutations that either decrease or increase the number of repeats in a locus. The mutation rate is 0.0007/locus/10 years. Our aim is to observe how different simulation scenarios affect the genetic diversity and whether any of the models produce diversity measures similar to those perceived in present-day Finns.

The population is simulated through its entire history (Figs 1 and 2). The simulation starts at 11,000 BP with a small initial pioneer population of 250 females and 250 males that is divided into two subpopulations: Saami and Other Finland. This is run for 200 steps, or 2000 years, to create a more natural, highly interrelated small founder population. We employ two different scenarios for

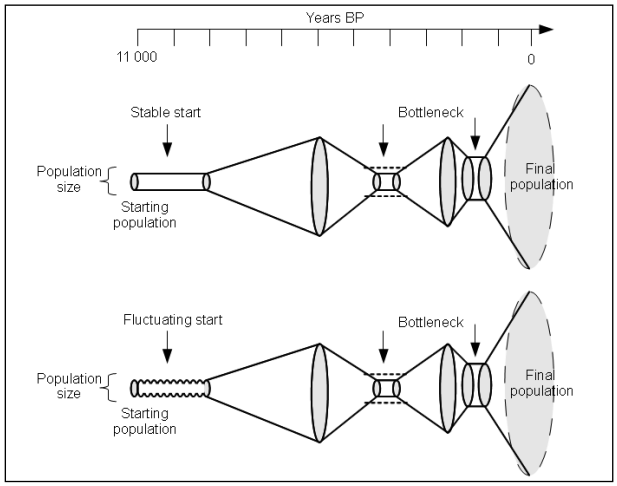

***Figure 2**. The general demographic model. The width of the cone represents the relative population size (not to scale) at the time before present depicted on the x axis.*

this initial phase: a stable starting population is compared with a fluctuating start i.e. serial founder effects. In the stable starting population models, the population size remains constant at 500 individuals (Saami 250, Other Finland 250), whereas in the serial founder effect models, the population fluctuates between 240-630 (Saami 120-315, Other Finland 120-315 respectively). Both subpopulations have the same size change rate, with population minima reached ca. every 200 years.

The total population size is set to grow exponentially from 9000 BP (simulation step 200) onward until the next bottleneck occurs. Immediately after 7000 BP (simulation step 400), and at a size of 15,000 individuals, the Other Finland subpopulation is divided into two: the South-western (SW) subpopulation and the North-eastern (NE) subpopulation. The respective sizes for these subpopulations are 5,000 and 10,000. Rather than a drastic population event taking place during a single simulation step, this split event is necessary to later enable internal migration. In simulation scenarios with internal gender-specific migration, the SW and NE subpopulations are allowed to exchange individuals.

We include two migration waves from neighbouring populations, the TCW migration wave at around 6000 BP (simulation steps 499, 500 and 501) and the CW migration wave at around 5200 BP (simulation steps 579, 580 and 581). Total population is set to reach a maximum of 25,000 shortly before 5750 BP (simulation step 525), the Stone Age population peak. The exponential growth slows down before this and the population size remains approximately the same for a brief period after this time point. After 5750 BP the population begins to gradually decline over 1600 years towards the Stone Age bottleneck. After the bottleneck the population recovers, followed by a less severe second bottleneck at 1500-1300 BP. During the last 1300 years the population size is set to grow to 1,000,000 individuals, the final population size.

2.1 The genetic diversity in the background populations

A few migration waves have been alleged to have entered Finland from neighbouring populations (see e.g. Carpelan 1999a, Lavento 2001). In addition to these, there has probably been minor, more or less constant, gene flow into the country. To allow for these phenomena, we formed three background populations: Archaic European, Archaic Scandinavian and Background Saami. From these, individuals migrate into the Finnish subpopulations with rates described below. We have selected actual ancient mtDNA haplotypes to form these two archaic background populations (Haak et al. 2005; Rudbeck et al. 2005; Melchior et al. 2007, 2008; Bramanti et al. 2009; Malmström et al. 2009; Skoglund et al. 2012). Since there is no information of ancient Saami DNA, both the Saami subpopulation and the Background Saami population were initialized with an approximation of present day Saami haplotype composition.

The Archaic European background population includes 50,000 individuals, the Archaic Scandinavian 25,000 individuals and the Background Saami 5,000 individuals. The sizes here are suggestive as we merely wish to evaluate if gene flow from other populations would have dramatic effects on the variation of Finnish subpopulations. Two sizes of migration waves were tested (see Table 1, scenarios E1-L2): temperate and small. In the temperate migration waves, the Typical Comb Ware migration wave replaces ca. 8% of the north-eastern subpopulation with Archaic European and the Corded Ware migration wave replaces ca. 3% of the south-western subpopulation with Archaic European. With smaller migration waves the

percentages are divided by four, i.e. approx. 2% and 0.8%, respectively. The volumes of migration were chosen based on our previous work (Sundell et al. 2010).

In addition to specific migration waves, temperate constant gene flow replaces 0.01% of the population with Archaic European every 10 years during the entire simulation. Additionally, 0.005% of the Saami and north-eastern subpopulation is replaced with Background Saami gene flow every 10 years and after 3500 BP 0.05% of the south-western subpopulation is replaced with Archaic Scandinavian every 10 years. Lower constant gene flow is one tenth of the above rates, i.e. 0.001%, 0.0005% and 0.005%, respectively (see Table 1, scenarios E1-L2). In our previous study (Sundell et al. 2010) we used higher gene flow rates and came to the conclusion that lower gene flow rates should be explored. The rates used there apparently forced the simulated populations' diversity to change towards the background populations'.

Technically, each background population evolves separately for 12,000 years. A snapshot is saved every 2000 years. The snapshot populations are then used as pools for migration in the bottleneck simulations over a period of 2000 years each, so that an approximation of evolving background populations is reached without having to simulate them in parallel with the Finnish subpopulations.

2.2 Simulation scenarios

The simulations were carried out with 24 separate scenarios (A1-L2). The 24 scenarios include all possible permutations of the following parameters: population growth first 2000 years, bottleneck size, internal migration, migration waves and constant gene flow (Fig. 3).

2.3 Population genetic analysis

We employed two basic indicators of genetic diversity: the number of haplotypes (A) and haplotype diversity (Ĥ) in a sample. The first is simply a direct count of the number of different haplotypes (i.e. differing in at least one nucleotide position or microsatellite locus). Ĥ (Nei 1987) is based on haplotype frequencies in a population and measures the probability of observing different haplotypes

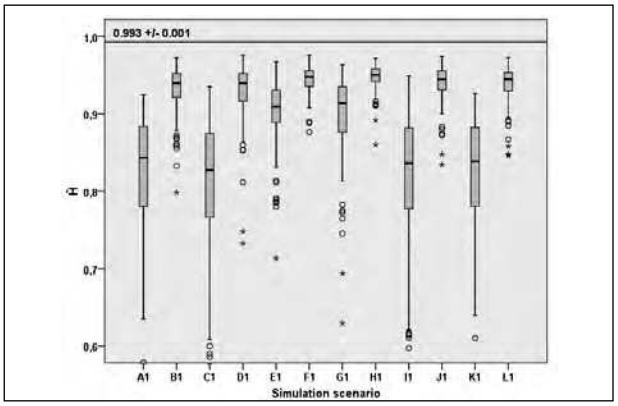

***Figure 3**. Mitochondrial haplotype diversity (Ĥ) in final population (0 BP).*

when sampling two random chromosomes or, as in this case, haploid individuals, from a population. Haplotype diversity was calculated with the formula $\hat{H} = n(1-\sum x_i^2)/(n-1)$ where n is the number of individuals and x_i the haplotype frequency of the *i*th haplotype. When Ĥ is low it is likely that two randomly drawn chromosomes are identical, and vice versa.

3. Results

First, compared with our previous simulations, gender-specific internal migration brings the simulated mitochondrial genetic diversity

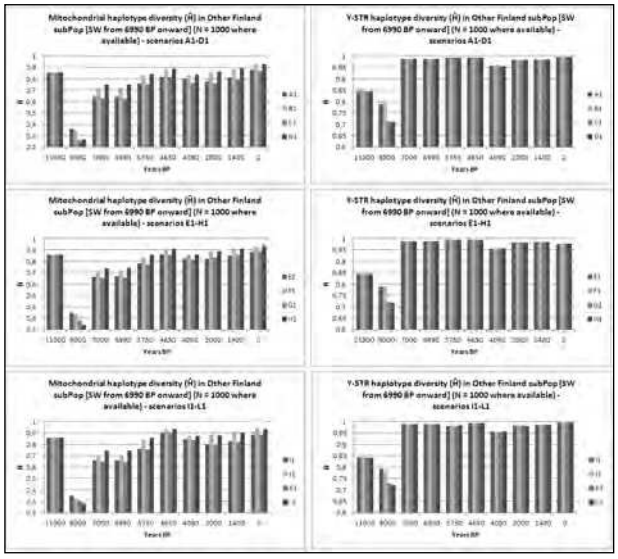

***Figure 4**. Mitochondrial (left) and Y-STR (right) genetic diversity in the Other Finland/SW subpopulation at checkpoints in all models with bottleneck size of 1000 individuals.*

Scenario	Population size first 2000 years	Bottleneck size at 4100-3800 BP	Internal migration between subpopulations	Migration waves (TCW and CW)	Constant gene flow
A1	stable	1000	no	-	-
A2	stable	200	no	-	-
B1	stable	1000	yes	-	-
B2	stable	200	yes	-	-
C1	fluctuating	1000	no	-	-
C2	fluctuating	200	no	-	-
D1	fluctuating	1000	yes	-	-
D2	fluctuating	200	yes	-	-
E1	stable	1000	no	small	temperate
E2	stable	200	no	small	temperate
F1	stable	1000	yes	small	temperate
F2	stable	200	yes	small	temperate
G1	fluctuating	1000	no	small	temperate
G2	fluctuating	200	no	small	temperate
H1	fluctuating	1000	yes	small	temperate
H2	fluctuating	200	yes	small	temperate
I1	stable	1000	no	temperate	small
I2	stable	200	no	temperate	small
J1	stable	1000	yes	temperate	small
J2	stable	200	yes	temperate	small
K1	fluctuating	1000	no	temperate	small
K2	fluctuating	200	no	temperate	small
L	fluctuating	1000	yes	temperate	small
L2	fluctuating	200	yes	temperate	small

Table 1. *Top. A reference matrix of different simulation scenarios (A1-L2). In addition to different simulation starts and bottleneck sizes, the other parameters are internal and gender specific migration between subpopulations, migration waves from Typical Comb Ware (TCW) and Corded Ware (CW) and constant gene flow from neighbouring populations.*

	Checkpoint	Time BP	Simulation steps
1	Starting point	11,000	0
2	Population starts to grow	9,000	200
3	Sampling before the population split	7,000	400
4	One step after the population split	6,990	401
5	Population peak	5,750	525
6	Before the first bottleneck	4,650	635
7	Immediately after the population minimum	4,090	691
8	Second population peak	2,000	900
9	In the middle of the second bottleneck	1,400	960
10	Present time, final generation	0	1099

Table 2. *Bottom. Simulation checkpoints. In every checkpoint, a random sample of 1000 individuals is extracted from the population extant at the time. If total population size at this point is smaller than 1000, all individuals in the population are included in the sample.*

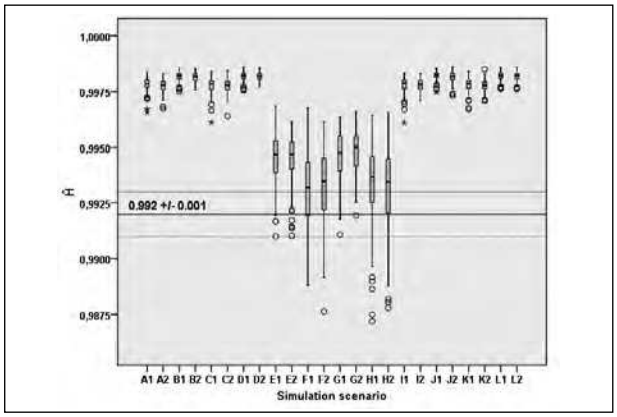

***Figure 5**. Y-STR haplotype diversity (Ĥ) in final population (0 BP) in a sample of 907 individuals. The reference line represents the value observed in the present (Palo* et al. *2009).*

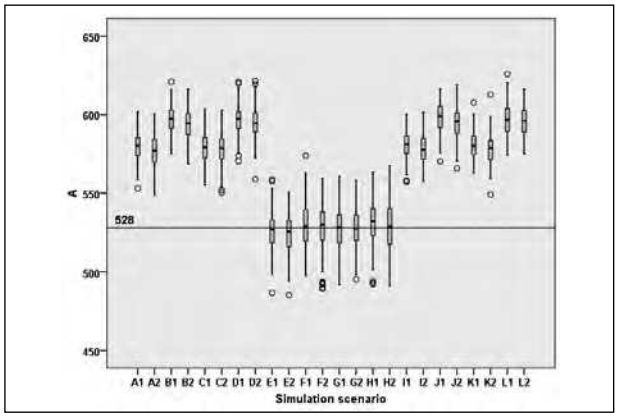

***Figure 6**. Number of Y-STR haplotypes (A) in final population (0 BP) in a sample of 907 individuals. The reference line represents the value observed in the present (Palo* et al. *2009).*

on the subpopulation level closer to the observed contemporary genetic diversity in Finland (Fig. 3).

Second, our results indicate that simulation models beginning with serial founder effects clearly reduce genetic diversity at the first checkpoint after the initial phase (Fig. 4).

Finally, the simulation scenarios with temperate constant gene flow produce Y chromosomal diversity measures similar to those observed in present-day Finnish population (Figs 5 and 6).

4. Discussion

The ultimate aim of these simulations is to determine which population scenarios are most likely to be true by assessing whether any of the models produce diversity measures similar to those perceived in present-day Finns. Previously we simulated Finland as one large population without substructure. In this refined model we divided the population into subpopulations, which, in addition to being more realistic, also enables gender-specific internal migration. Population subdivision also allows genetic drift to operate more realistically. In comparison with our previous study, the gender-specific migration brings the simulated mitochondrial genetic diversity closer to the observed contemporary genetic diversity in Finland. Y chromosomes in these simulations mutate faster, which obscures the effect of migration.

Our results indicate that simulation models beginning with serial founder effects clearly reduce genetic diversity at the first checkpoint after the initial phase. In later phases, however, other factors seem to obscure this effect. Therefore we cannot distinguish in this study whether models beginning with or without serial founder effects fit observed current genetic diversity better.

In our previous study (Sundell et al. 2010) we used higher gene flow rates and came to the conclusion that lower gene flow rates should be explored. The rates used there apparently forced the simulated populations' diversity to change towards the background populations'. The simulation scenarios with temperate constant gene flow produce Y chromosomal diversity measures similar to those observed in present-day Finnish population. Indeed, all the models with temperate constant gene flow result in diversity measures compatible with Finnish genetic diversity, indicating that the background gene flow is actually a more important factor than initial bottleneck size, at least with the bottleneck sizes we used.

3.1 Subsistence

The deglaciation of the region took several thousand years, during which there were major changes in the subsistence conditions of pioneer settlers. Population size fluctuation during the pioneering stage caused by serial founder effects can be argued to more realistically model the conditions

in the region during this phase of settlement. It is highly plausible that some small groups migrating into the country have not persisted. The reproduction rate has most likely been extremely low at times, barely sufficient for inhabitation to continue. In the Boreal climate zone livelihood preconditions are limited and small changes in climatic conditions can already be significant in determining the success of different subsistence models.

3.2 Limitations of simulations

Simulations are a useful tool in studying population processes unreachable by other means. They may help us to find the demographic model capturing the important features that shaped genetic diversity. However, simulations have their limitations. Simulation models can never be an exact replication of the complex reality, but instead, are more useful as evidence that a certain model may depict reality better than another. However, one must also note that the similarities between the modelled and observed diversities do not directly prove causality. Therefore, simulations can be considered to perform best when refuting some hypotheses. Furthermore, different models created by different simulation settings may replicate the observed patterns of diversity equally well, thus leaving us with several possibilities. In addition, with an increase in parameters, simulation can become very complex, i.e. modelling with too many variable parameters may obscure otherwise justified conclusions.

Nevertheless, simulations are beneficial in evaluating the credibility of different scenarios. One of the principal benefits of a simulation model is that you can begin with a simple approximation of a process and gradually refine the model as your understanding of the process improves. This allows achieving good approximations of very complex models. Additionally, simulations allow randomness and are an error-tolerant approach enabling repeatability. As a result, models with several interrelating parameters will always benefit from using simulation. Despite the weaknesses entailed in simulations, the importance of acquiring credible models by simulating is evident. As statistician George E. P. Box put it when exploring the relationships between interdependent variables: "Essentially, all models are wrong, but some are useful" (Box and Draper 1987).

3.3 Future prospects

Incorporation of autosomal loci is one of our future goals. Adding them would bridge the gap between the effects of maternal and paternal inheritance. Furthermore, studying population admixture in more detail would also be highly interesting. Genetic consequences of population admixture, given that the source populations are somewhat different, can be observed in allele frequencies e.g. mtDNA and Y chromosome distributions and marker-to-marker linkage disequilibrium (LD) patterns. Interestingly, new research on natural and sexual selection in a historical Finnish population has been published very recently (Courtiol et al. 2012); it was shown that even in historical times, males have had higher variance in reproductive success than females. This could be added to our mating model to evaluate how much it would affect the Y chromosomal diversity.

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