

Contemporary human evolution

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Learning objectives

Has evolution stopped for humans?
If not, how are humans still evolving?

"Is" and "Ought" are not
necessarily the same

Things that exist in nature:

Malaria

Parental care

Mass extinction

Mutualism

Caching food

Box 1 | **Why humans continue to evolve despite the many benefits of hygiene and modern medicine**

Within a decade of the publication of *On the Origin of Species*, the misconception developed that modern hygiene and medicine have caused natural selection to stop working on human populations⁵⁸. This was fuelled by another misconception: that selection operates only through differences in survival. We now know that natural selection on traits occurs whenever there is variation among individuals in fitness and in traits and when the variation in traits is correlated with the variation in fitness. A response to selection will then follow if some portion of the variation in the traits is heritable. A good proxy for fitness is lifetime reproductive success (LRS) or number of children per parent per lifetime. LRS has both a survival component — one must survive to reproduce — and a reproductive component. Good hygiene and medical care that reduce prenatal, infant and child mortality rates reduce the variation among individuals in the survival component but that does not eliminate natural selection, as substantial variation among individuals in the reproductive component remains. For example, consider an extreme case in which medical and public health measures were so good that everyone who was born survived to age 80. This would not eliminate natural selection, as individuals would still differ in their LRS and that variation would drive natural selection. The potential for natural selection only vanishes when all individuals have exactly the same reproductive success or when no trait is correlated with the variation in reproductive success that still exists. These states are unlikely ever to occur in any population.

The effect of culture on biology raises interesting issues. Birth control, assisted reproductive technology and the increased prevalence of late marriage and divorce complicate the evolutionary genetics of reproduction. These factors can be dealt with by regarding them as part of a changing environment that is changing selection intensities. A more fundamental solution awaits the development of methods of analyzing gene–culture co-evolution that can be applied to large, longitudinal human data sets.

| Trait | Sex | Selection | p | n | Population (century) | Refs |
|-------------------------|-----|-----------|-----|-------|----------------------|------|
| <i>Life history</i> | | | | | | |
| Age at first birth | F | – | *** | 306 | Finland (17th–19th) | 109 |
| | F | – | *** | 395 | Finland (18th–19th) | 17 |
| | F | – | *** | 2,227 | USA (20th) | 12 |
| | F | – | ** | 314 | Finland (20th) | 110 |
| | F | – | *** | 1,459 | Australia (20th) | 111 |
| | F | – | ** | 2,443 | USA (20th) | 112 |
| | M | – | ** | 395 | Finland (18th–19th) | 113 |
| | M | – | ** | 2,443 | USA (20th) | 112 |
| Interbirth interval | F | – | *** | 306 | Finland (17th–19th) | 109 |
| Age at last birth | F | + | *** | 306 | Finland (17th–19th) | 109 |
| | F | + | * | 314 | Finland (20th) | 110 |
| Age at menopause | F | +/s | ** | 2,227 | USA (20th) | 12 |
| | F | + | ** | 1,459 | Australia (20th) | 111 |
| Age at death | M | + | *** | 746 | USA (19th) | 114 |
| <i>Morphology</i> | | | | | | |
| Weight | F | + | ** | 1,278 | USA (20th) | 115 |
| | F | +/s | *** | 2,227 | USA (20th) | 12 |
| | M | s | *** | 2,616 | USA (19th–20th) | 116 |
| Height | F | + | * | 216 | Gambia (20th) | 43 |
| | F | –/s | ** | 3,552 | Great Britain (20th) | 100 |
| | F | – | ** | 1,278 | USA (20th) | 115 |
| | F | – | *** | 2,227 | USA (20th) | 12 |
| | M | s | *** | 2,616 | USA (19th–20th) | 116 |
| | M | + | * | 322 | USA (20th) | 44 |
| | M | + | *** | 3,201 | Poland (20th) | 117 |
| <i>Physiology</i> | | | | | | |
| Cholesterol | F | – | ** | 2,227 | USA (20th) | 12 |
| Systolic blood pressure | F | – | * | 2,227 | USA (20th) | 12 |
| Blood glucose | F | s | ** | 2,227 | USA (20th) | 12 |



Table 3. Population Frequencies of LP Alleles C/T⁻¹³⁹¹

| Designation | Region or Population | Three-Letter Code | N | No. with Genotype | | | Allele Frequency (%) | | Prevalence of LP (% [SD]) | |
|-------------|----------------------|-------------------|-----|-------------------|----|----|----------------------|------|---------------------------|---------|
| | | | | CC | CT | TT | C | T | | |
| 1 | South Korea | SKo | 23 | 23 | 0 | 0 | 100 | 0 | 0 | (.00) |
| 2 | Han Chinese | Han | 100 | 100 | 0 | 0 | 100 | 0 | 0 | (.00) |
| 3 | Ob-Ugric speakers | ObU | 62 | 58 | 4 | 0 | 96.8 | 3.2 | 6 | (3.02) |
| 4 | Komi | Kom | 10 | 7 | 3 | 0 | 85 | 15 | 30 | (14.50) |
| 5 | Udmurts | Udm | 30 | 12 | 16 | 2 | 66.6 | 33.4 | 60 | (8.90) |
| 6 | Mokshas | Mok | 30 | 13 | 17 | 0 | 71.6 | 28.4 | 56.6 | (9.01) |
| 7 | Erzas | Erz | 30 | 17 | 10 | 3 | 73.3 | 26.7 | 43.3 | (9.05) |
| 8 | Saami | Saa | 30 | 20 | 10 | 0 | 83.3 | 16.7 | 33.3 | (8.60) |
| 9 | Finns, eastern | FiE | 77 | 18 | 35 | 24 | 46.1 | 53.9 | 76.6 | (4.75) |
| 10 | Finns, western | FiW | 154 | 25 | 68 | 61 | 38.3 | 61.7 | 83.7 | (2.98) |
| 11 | Daghestan Druss | DaD | 17 | 13 | 4 | 0 | 88.2 | 11.8 | 23.5 | (10.30) |
| 12 | Daghestan Nog | DaN | 20 | 15 | 5 | 0 | 87.5 | 12.5 | 25 | (9.70) |
| 13 | Daghestan mixed | DaM | 23 | 19 | 3 | 1 | 89.1 | 11.9 | 17.4 | (7.90) |
| 14 | Balti | Bal | 23 | 23 | 0 | 0 | 100 | 0 | 0 | (.00) |
| 15 | Burusho | Bur | 30 | 29 | 1 | 0 | 98.3 | 1.7 | 3.3 | (3.26) |
| 16 | Kashmiri | Kas | 20 | 15 | 5 | 0 | 87.5 | 12.5 | 25 | (9.68) |
| 17 | Kalash | Kal | 30 | 30 | 0 | 0 | 100 | 0 | 0 | (.00) |
| 18 | Pathan | Pat | 28 | 12 | 15 | 1 | 69.6 | 30.4 | 57.1 | (9.35) |
| 19 | Hazara | Haz | 14 | 13 | 1 | 0 | 96.4 | 3.6 | 7.1 | (6.86) |
| 20 | Baluch | Blu | 19 | 10 | 6 | 3 | 68.4 | 31.6 | 47.4 | (11.46) |
| 21 | Sindi | Sin | 28 | 10 | 13 | 5 | 58.9 | 41.1 | 64.3 | (9.11) |
| 22 | Brahui | Bra | 30 | 17 | 10 | 3 | 73.3 | 26.7 | 43.3 | (9.05) |
| 23 | Makrani Baluch | MaB | 29 | 19 | 10 | 0 | 82.8 | 17.2 | 34.5 | (8.83) |
| 24 | Mohannes | Moh | 29 | 16 | 10 | 3 | 72.4 | 27.6 | 44.8 | (9.23) |
| 25 | Parsi | Par | 29 | 21 | 8 | 0 | 86.2 | 13.8 | 27.6 | (8.30) |
| 26 | Iranians | Ira | 21 | 17 | 4 | 0 | 90.5 | 9.5 | 19 | (8.56) |
| 27 | Qashqai | Qas | 10 | 9 | 1 | 0 | 95 | 5 | 10 | (9.49) |
| 28 | Arabs | Ara | 51 | 42 | 8 | 1 | 90.2 | 9.8 | 17.6 | (5.33) |
| 29 | Southern Italy | SIIt | 100 | 89 | 11 | 0 | 94.5 | 5.5 | 11 | (3.13) |
| 30 | French | Fra | 17 | 6 | 9 | 1 | 61.7 | 38.3 | 58.8 | (11.94) |
| 31 | Basques | Bas | 85 | 7 | 44 | 34 | 34.1 | 65.9 | 91.7 | (2.99) |
| 32 | Utah | Uta | 92 | 7 | 33 | 52 | 25.5 | 74.5 | 92.4 | (2.76) |
| 33 | Somalia | Som | 79 | 74 | 5 | 0 | 96.8 | 3.2 | 6.3 | (2.73) |
| 34 | Fulani Sudanese | Ful | 44 | 13 | 20 | 11 | 52 | 48 | 70.4 | (6.88) |
| 35 | Saharawi | Sah | 57 | 29 | 26 | 2 | 73.7 | 26.3 | 49.1 | (6.62) |
| 36 | Morocco | Mor | 90 | 62 | 25 | 3 | 82.7 | 17.3 | 31.1 | (4.88) |
| 37 | African Americans | Aam | 50 | 44 | 3 | 3 | 91 | 9 | 12 | (4.60) |

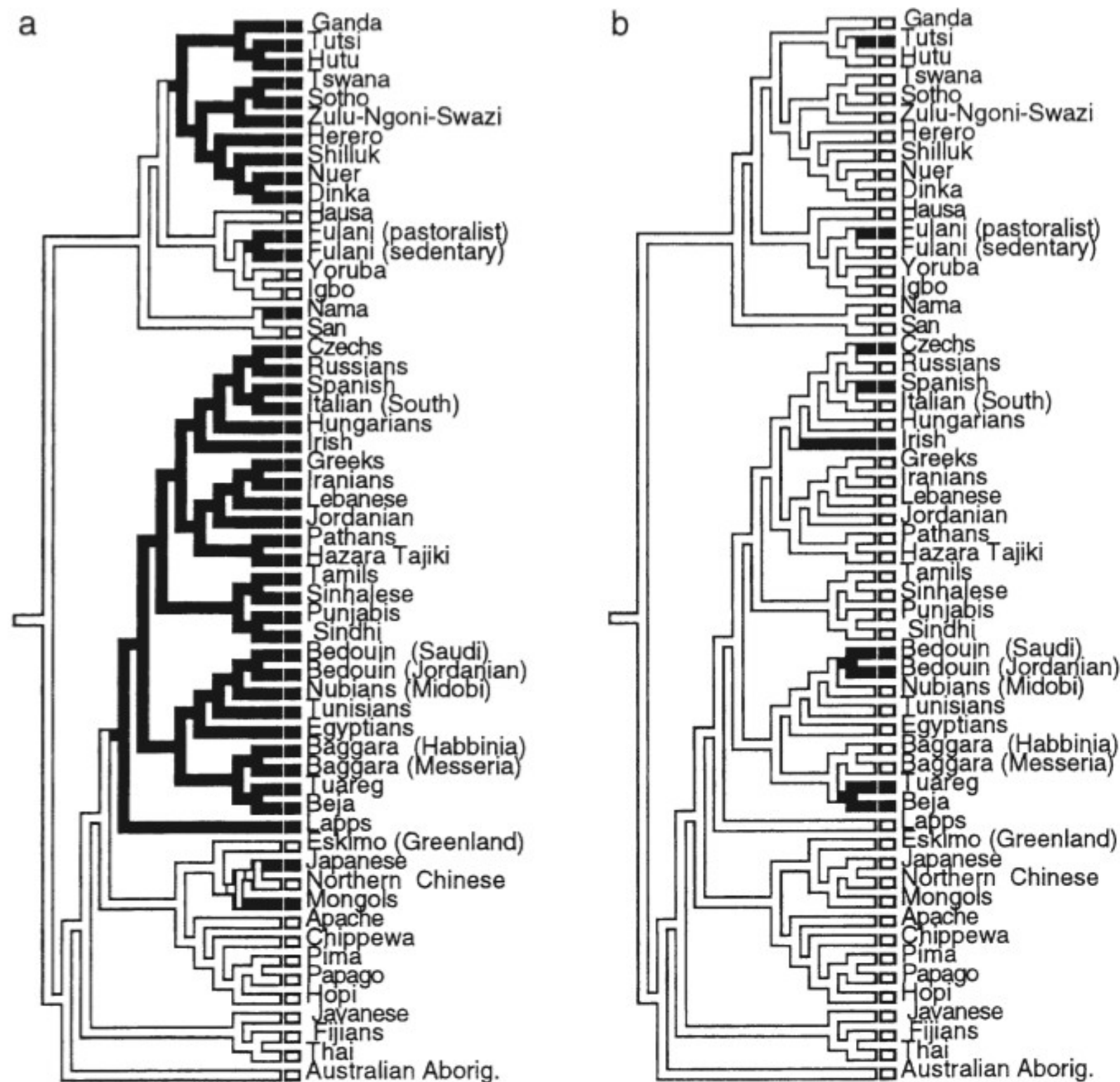
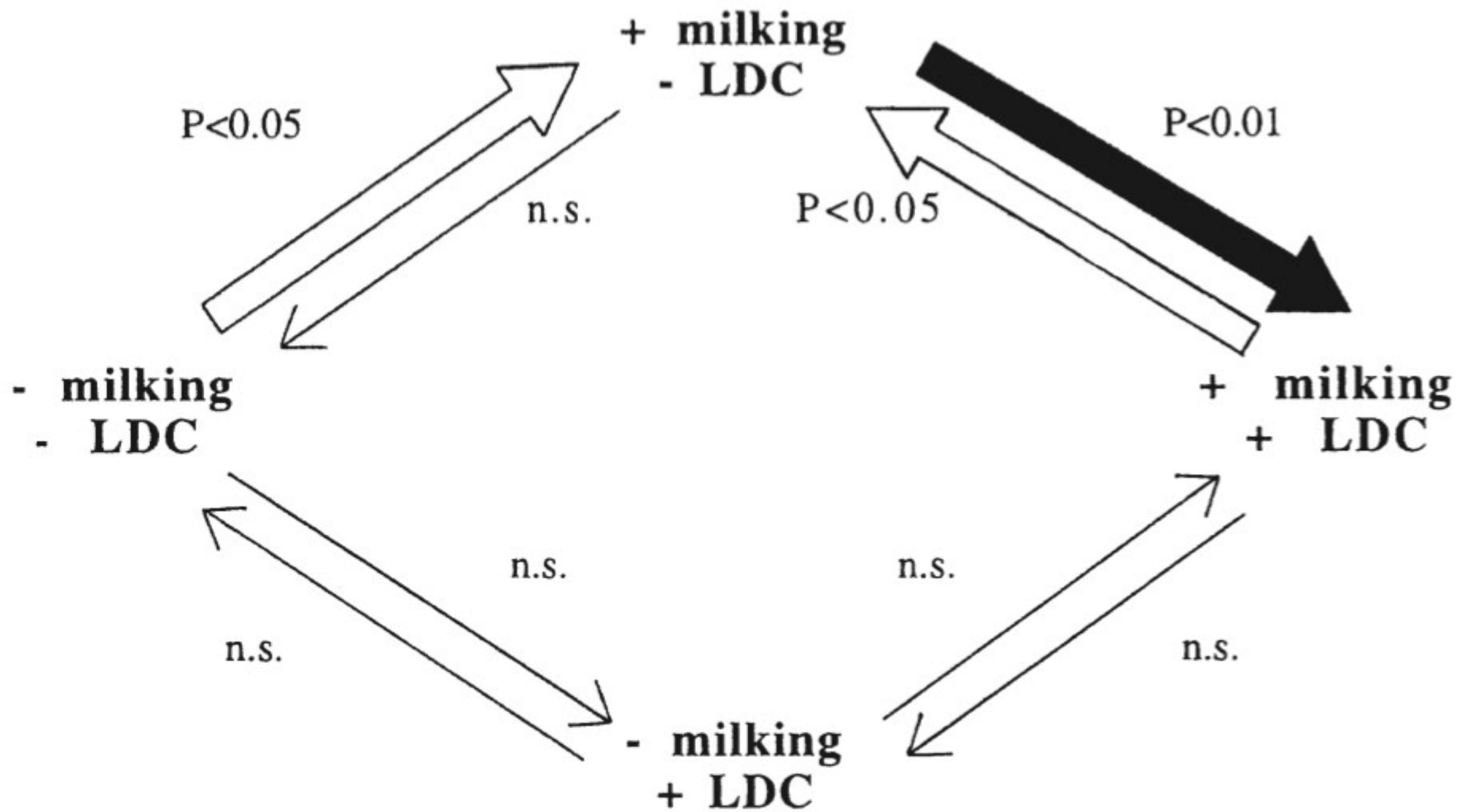
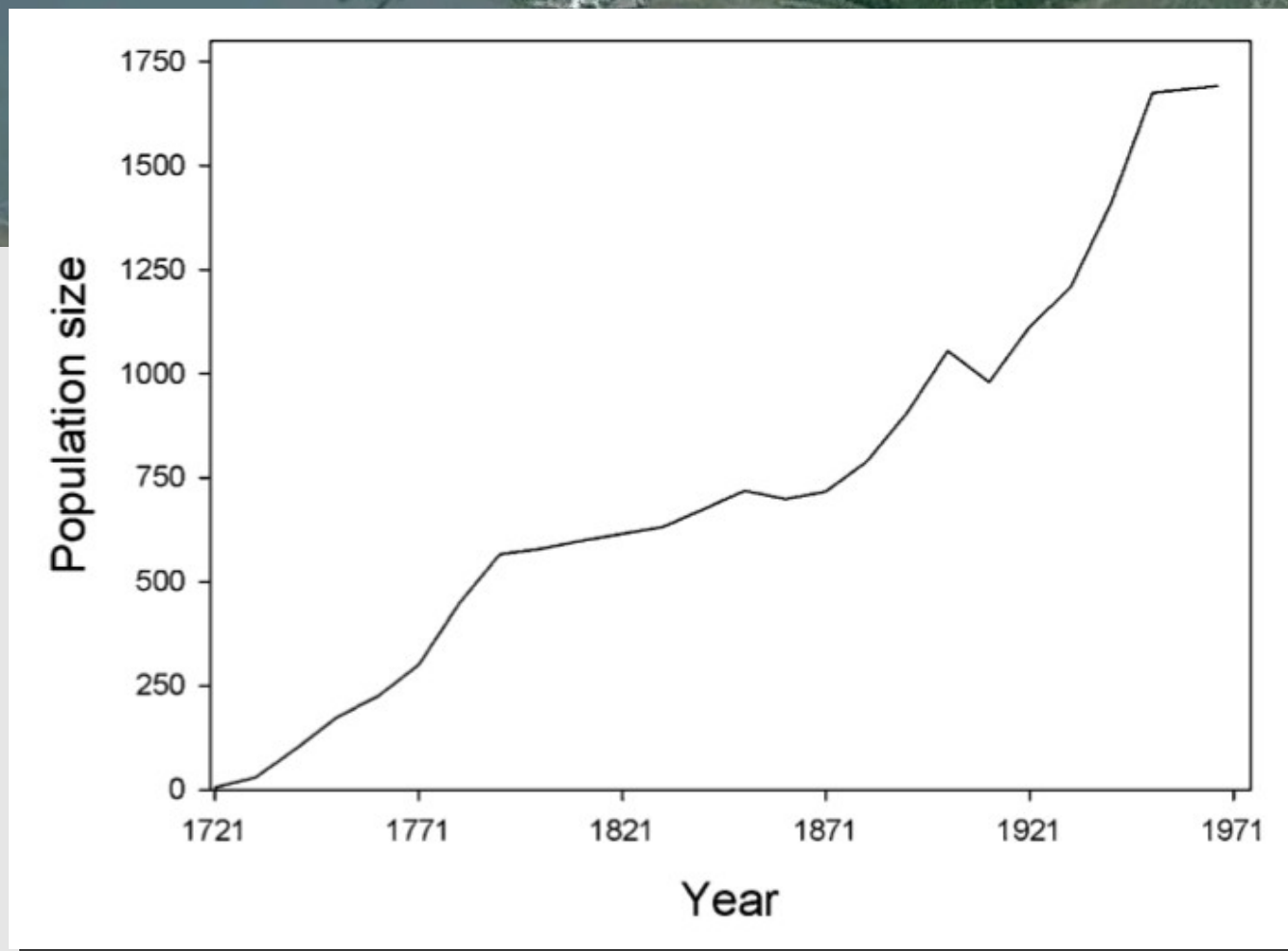
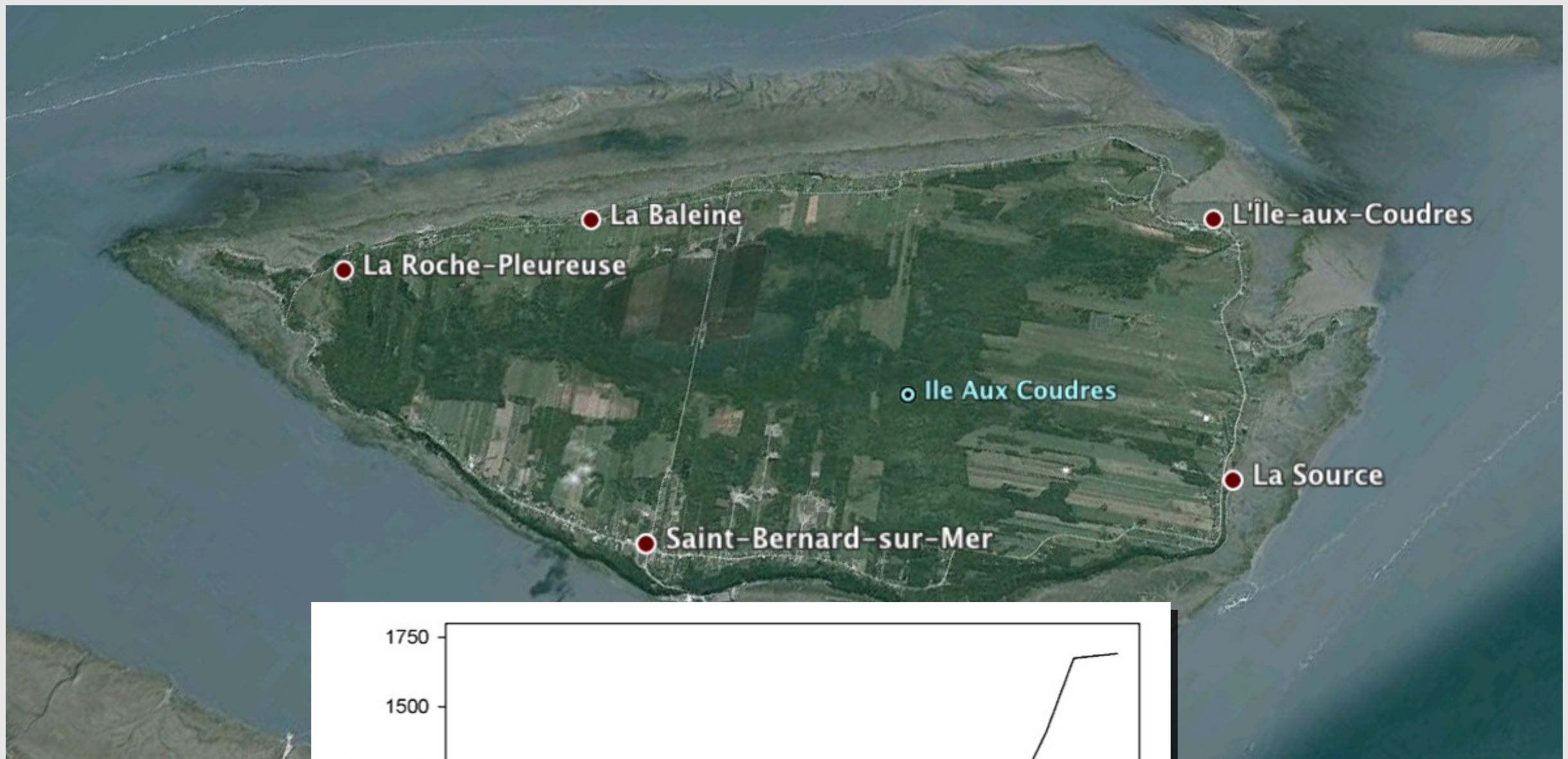


Figure 5. (a) Tree used for the maximum-likelihood analysis, using DISCRETE, based on the F_{ST} genetic tree. Black indicates milking populations, and white indicates nonmilking populations. (b) Tree used for the maximum-likelihood analysis, using DISCRETE. Black indicates high LDC populations, and white indicates low LDC populations, following the bimodal distribution of this trait seen in Figure 7.



LDC = lactose digestion capacity = can process lactose



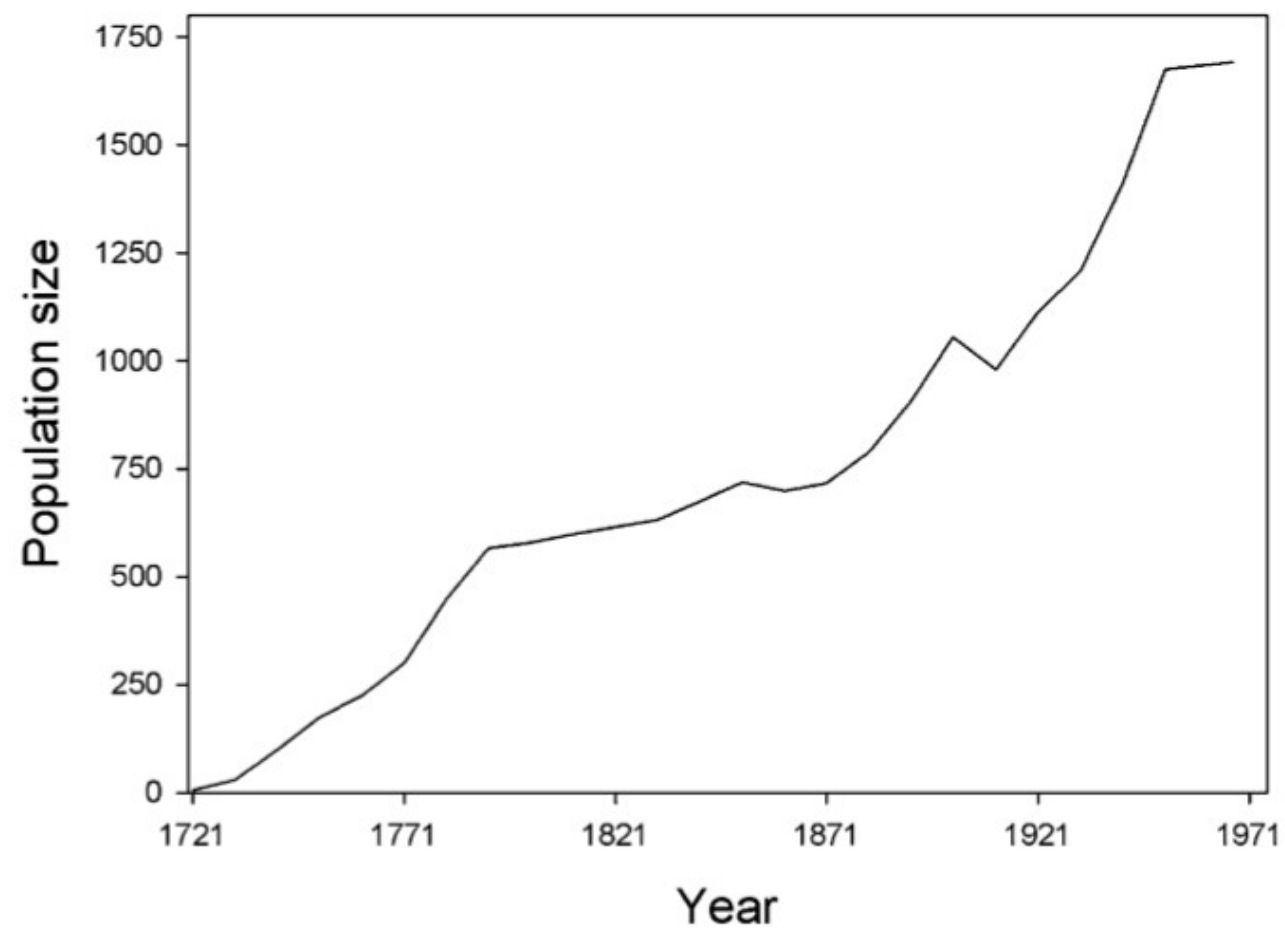


Table 1. Average phenotypic values (\pm SD) for female life-history traits in the preindustrial human population of île aux Coudres

| Trait | Migration dataset* | Subfecundity dataset | Women included under the subfecundity hypothesis only |
|------------------------------------------------------------|-----------------------|-----------------------|-------------------------------------------------------|
| Marriage–first birth interval (mo) | 13.9 \pm 6.2 (360) | 17.8 \pm 22.0 (564) | 25.7 \pm 34.6 (204) |
| Age at first birth (y) | 23.4 \pm 3.9 (363) | 23.8 \pm 4.3 (572) | 24.5 \pm 4.9 (209) |
| Age at last birth (y) | 38.7 \pm 6.7 (363) | 36.1 \pm 7.3 (572) | 31.6 \pm 6.1 (209) |
| Longevity (y) | 56.9 \pm 22.2 (252) | 58.2 \pm 21.6 (301) | 65.1 \pm 17.0 (49) |
| Fertility (completed family size) | 8.6 \pm 3.9 (363) | 7.0 \pm 4.1 (572) | 4.3 \pm 2.9 (209) |
| Lifetime reproductive success (offspring living to age 15) | 7.0 \pm 3.4 (363) | 5.1 \pm 3.5 (363) | 3.5 \pm 2.6 (209) |

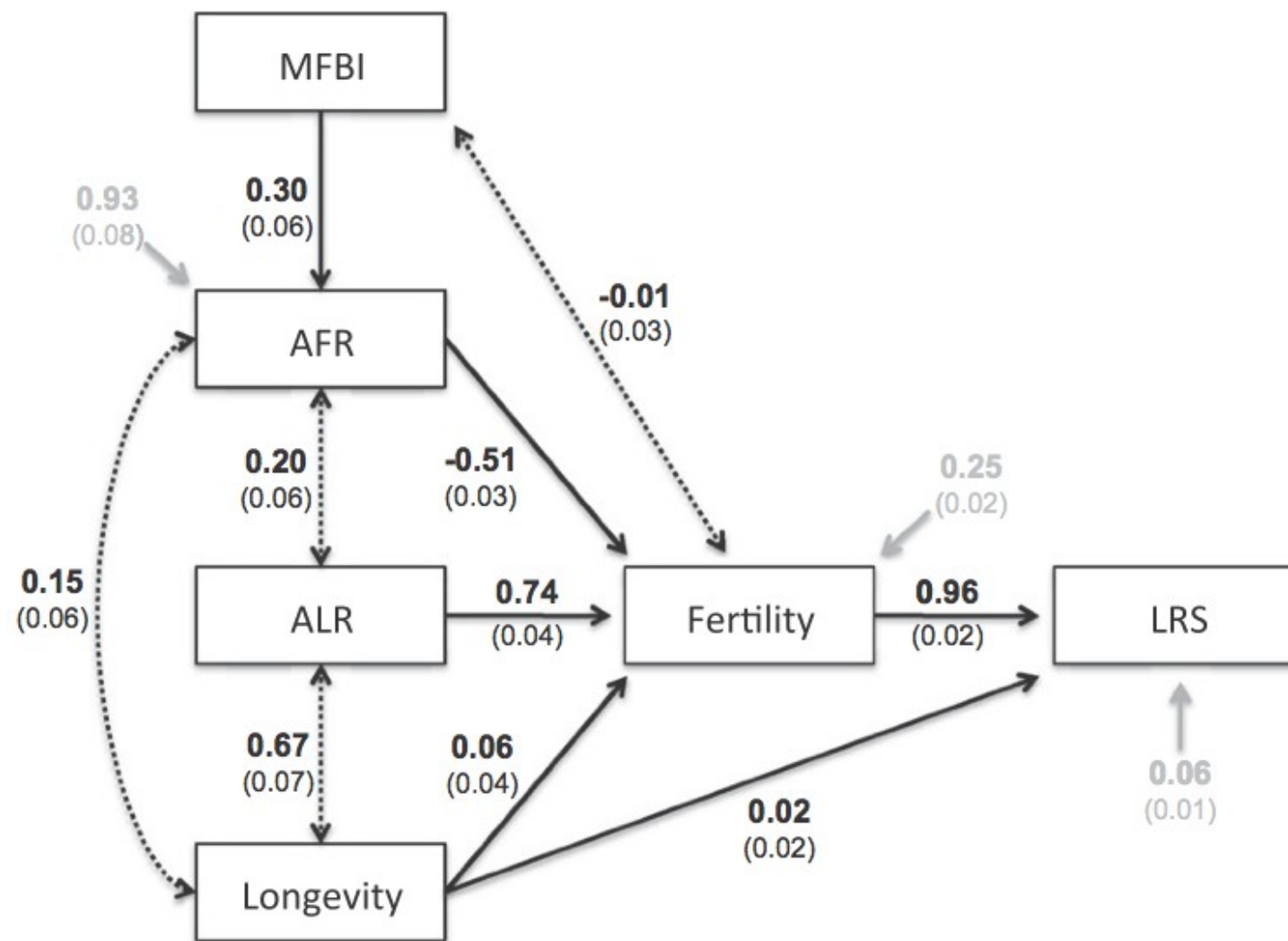
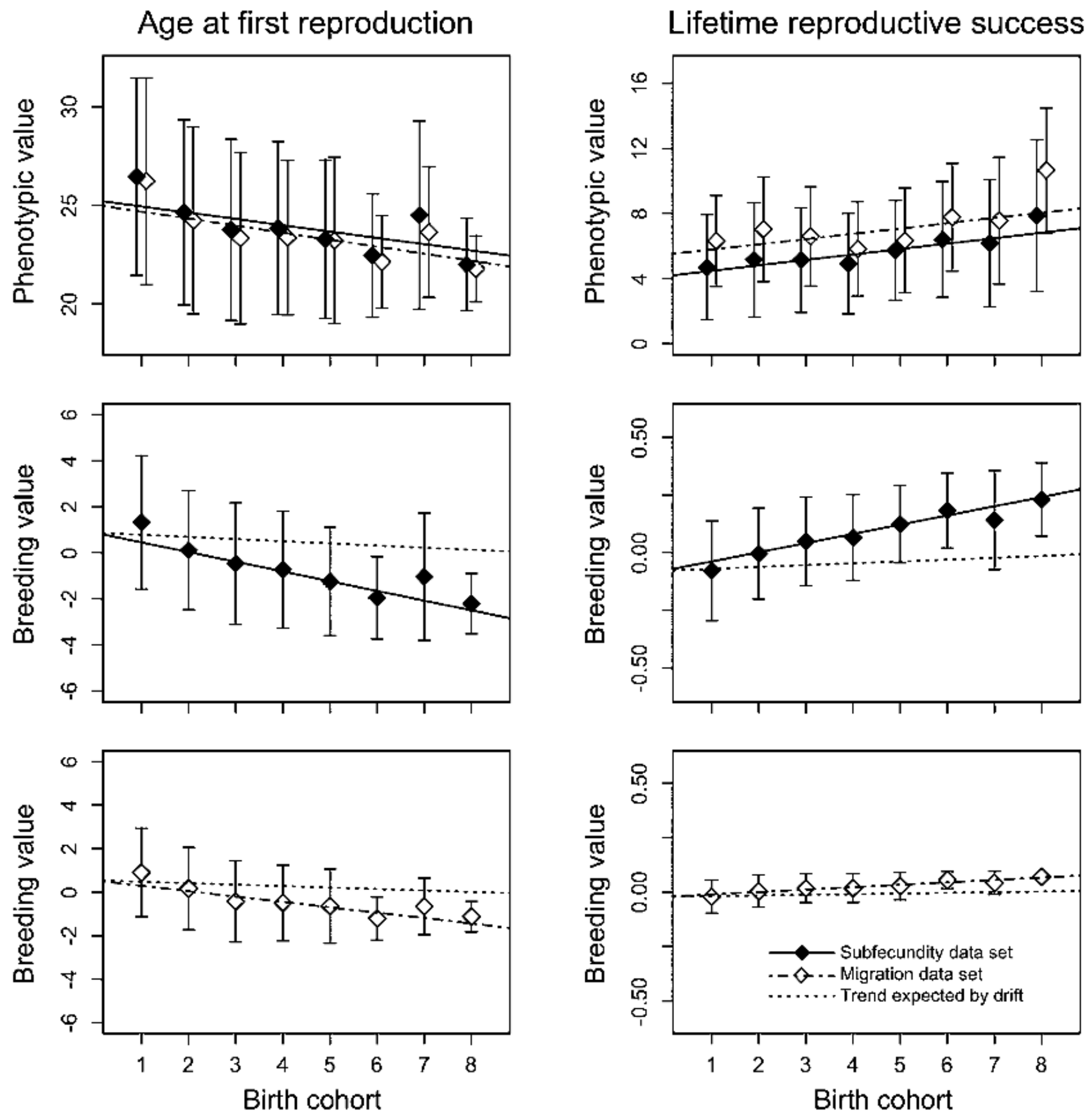


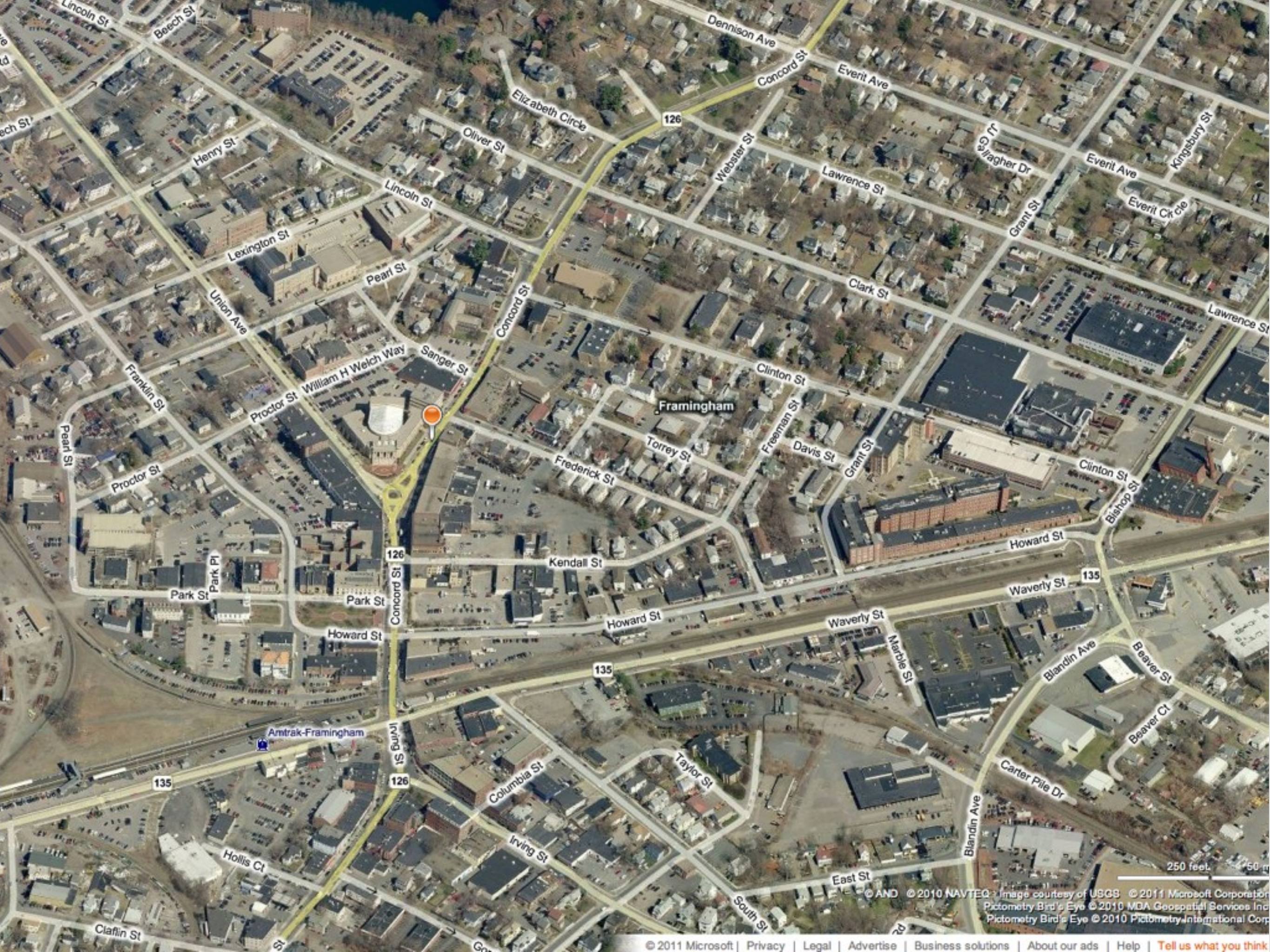
Fig. 1. Path diagram describing the selection exerted on female life-history traits at île aux Coudres. Solid one-way arrows show presumed causal relationships between variables, and dashed two-way arrows are noncausal correlations. Values (\pm SEM) next to solid arrows are standardized regression coefficients (direct effects for selection gradients), and values next to dashed arrows are correlation coefficients. Values (\pm SEM) and arrows in gray are for unmeasured causes (residual variance) of endogenous variables. Direct paths are those passing through causal relationships only (e.g., AFR \rightarrow fertility \rightarrow LRS), whereas indirect paths pass through at least one correlational relationship (e.g., AFR \leftrightarrow ALR \rightarrow fertility \rightarrow LRS). Life-history traits are: AFR, age of the woman at first reproduction; ALR, age of the woman at last reproduction; fertility, completed family size; longevity, woman's lifespan; LRS, lifetime reproductive success; MFBI, marriage–first birth interval. Results are for the subfecundity dataset ($n = 283$; *Materials and Methods*); the migration dataset led to similar path coefficients (Fig. S2).

Table 2. Genetic parameters and response to natural selection in woman's age at first reproduction and lifetime reproductive success at île aux Coudres between 1800 and 1939

| Dataset | Response variable | Heritability | | Shared familial environment effects | | Genetic correlation between AFR and LRS | | Genetic response | |
|--------------|-------------------|--------------|-----------|-------------------------------------|-----------|-----------------------------------------|----------------|------------------|-------------------------|
| | | Mode | Interval | Mode | Interval | Mode | Interval | Trend PBVs | Prob. drift \geq obs. |
| Subfecundity | AFR | 0.55 | 0.30–0.90 | 0.01 | 0.00–0.15 | –0.81 | –0.97 to –0.48 | –2.95 | 0.009 |
| | LRS | 0.04 | 0.00–0.43 | 0.00 | 0.00–0.07 | — | — | +0.28 | 0.009 |
| Migration | AFR | 0.30 | 0.08–0.73 | 0.01 | 0.00–0.12 | –0.81 | –0.99 to 0.16 | –1.74 | 0.058 |
| | LRS | <0.01 | 0.00–0.12 | 0.00 | 0.00–0.02 | — | — | +0.08 | 0.144 |

For heritability, shared familial environment effects, and genetic correlation, the mode of the posterior distribution (i.e., the point estimate of the parameter) and the 95% Bayesian posterior interval of highest density are reported separately for each dataset. The genetic correlation involves both traits and is only shown once for each dataset. The genetic response is the difference in mean PBVs between the first and last women's birth cohorts computed from the slope of the regression of PBVs on eight 20-y cohorts (means are over all women of a cohort and 1,000 MCMC samples). The trend in PBVs is in years for AFR and on the latent scale (Poisson model) for LRS. "Prob. drift \geq obs" indicates the probability of observing a trend as strong or stronger due to random genetic drift alone (two-tailed test).





126

135

135

126

126

Framingham

Amtrak-Framingham

250 feet 50 m

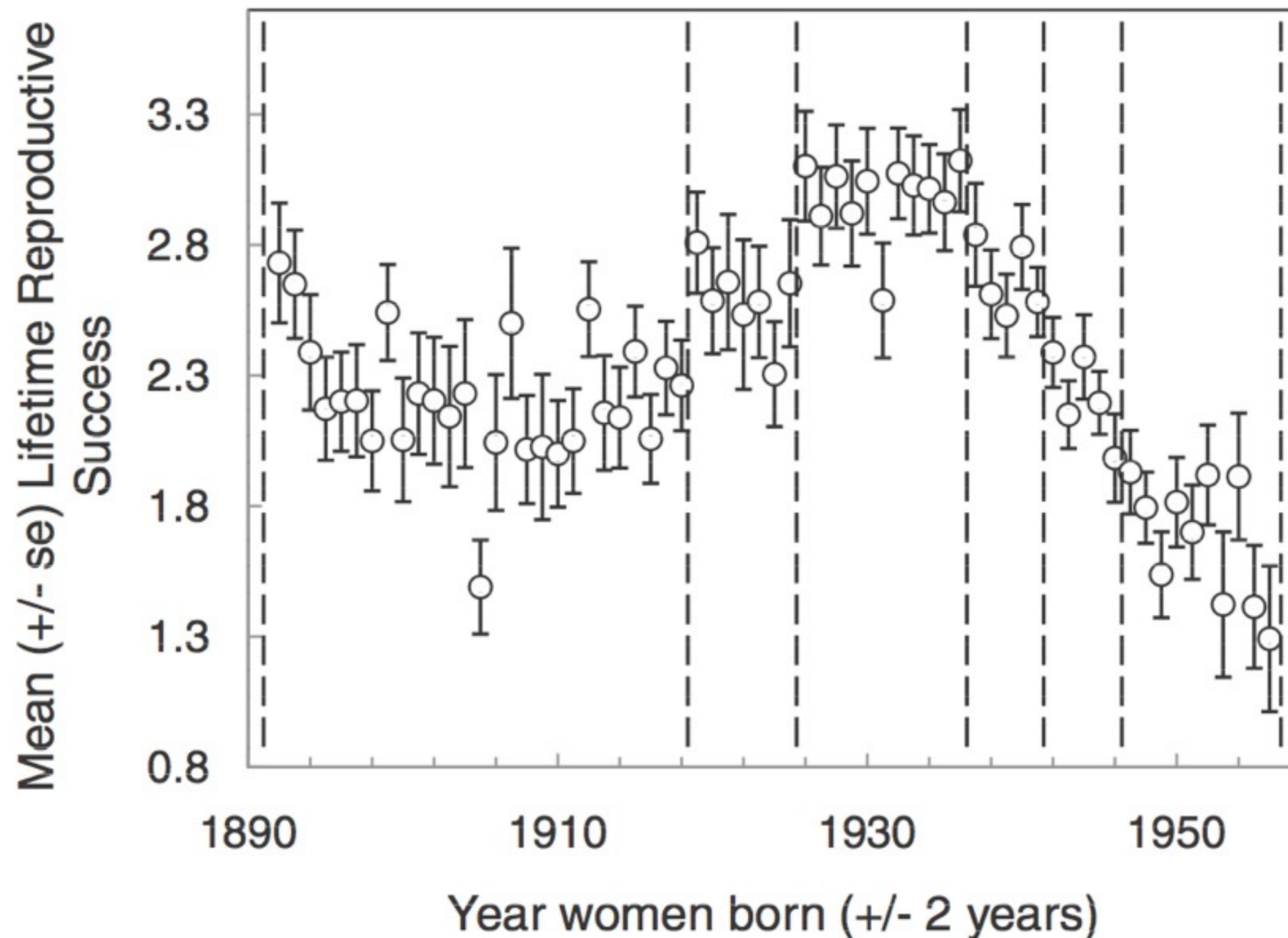


Fig. 2. LRS, by year of birth, for women in the FHS. To deal with secular demographic change, we divided the data into six periods and divided the relative reproductive success of each woman by the mean reproductive success of the women in her group.

Table 5. Projected evolutionary change, untransformed values

| | TC, mg/100 mL | WT, kg | HT, cm | SBP,mmHg | DBP, mmHg | GLU, mg/100 mL | Age at menopause | Age at first birth |
|------------|---------------|-------------|--------------|--------------|-------------|----------------|------------------|--------------------|
| Generation | | | | | | | | |
| 0 | 223.9 | 64.7 | 160.2 | 127.6 | 78.5 | 89.1 | 48.9 | 26.18 |
| 5 | 219.8 ± 1.58 | 65.4 ± 0.58 | 159.3 ± 0.41 | 126.3 ± 0.71 | 78.6 ± 0.34 | 88.4 ± 0.52 | 49.3 ± 0.23 | 25.96 ± 0.23 |
| 10 | 215.9 ± 3.35 | 65.6 ± 1.33 | 158.1 ± 0.90 | 125.2 ± 1.47 | 78.7 ± 0.73 | 88.1 ± 1.03 | 49.7 ± 0.51 | 25.74 ± 0.48 |
| % | 3.6 | 1.4 | 1.3 | 1.9 | 0.3 | 1.1 | 1.6 | 1.7 |
| Haldanes | 0.016 | 0.007 | 0.032 | 0.010 | 0.002 | 0.004 | 0.020 | 0.010 |

The averages for generation 0 are those observed for the Framingham original and offspring cohorts between the ages of 20 and 60 over the years 1955–2003. Values for later cohorts are the means that would be expected given the average conditions over the period of observation and evolutionary change. %, percent change in traits from generation 0 to 10. Haldanes: rate of evolution in SD per generation.

Natural selection is acting slowly and gradually on traits of medical importance and on life history traits in the FHS population. Selection varied in intensity, becoming generally less intense over time, but not in direction, and it has only operated consistently over the entire period to reduce age at first birth. Predictions for one generation are fairly reliable, but whether selection will be consistent and sustained enough to bring about significant genetic change can only be answered with longer periods of observation of more traits relevant to human health.

These results suggest slow evolutionary change. It is noteworthy, although not surprising, that both age at first birth and age at menopause appear to be changing so as to lengthen the reproductive period, which is consistent with previous findings (3). Because fertility is the driving force behind evolution in modern populations, we might have found larger effects of evolution on the levels of sex hormones and related traits had they been measured. The impact of fertility on selection could prove especially important now that many couples that would otherwise remain childless can produce offspring with medical assistance.

| Trait | Sex | Selection | <i>p</i> | <i>n</i> | Population (century) |
|---------------------|---------|-----------|----------|----------|----------------------|
| Education | F | – | ** | 2,443 | USA (20th) |
| | F and M | – | * | 1,906 | USA (20th) |
| | M | – | * | 2,443 | USA (20th) |
| Income | F | – | ** | 2 443 | USA (20th) |
| | F | – | * | 1,278 | USA (20th) |
| | F | – | * | 14,000 | Sweden (20th) |
| | F | – | ** | 5,576 | Great Britain (20th) |
| | F and M | – | * | 1,906 | USA (20th) |
| | M | +/– | ** | 10,436 | USA (20th) |
| | M | + | ** | 2,443 | USA (20th) |
| | M | + | ** | 5,576 | Great Britain (20th) |
| | M | + | *** | 14,000 | Sweden (20th) |
| Wealth | M | + | *** | 164 | Kenya (20th) |
| | M | + | * | 746 | USA (19th) |
| | M | + | *** | 302 | Sweden (19th) |
| Occupational status | F and M | – | * | 1,906 | USA (20th) |
| Rank | M | + | *** | 746 | USA (19th) |
| | M | + | * | 322 | USA (20th) |
| Hunting ability | M | + | * | 18 | Botswana (20th) |
| Intelligence | F and M | – | *** | 1,906 | USA (20th) |