Inferring ancient population sizes: An application of coalescent theory
Dem260 Math Demog
Spring 2020
Lecture 12

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Agenda for today

1. Coalesence when population size is changing
2. Inference from MRCA times
3. ($n > 2$ comparisons and relative branch lengths)
4. Demo: Using real mtDNA to estimate population sizes.

Caveat: We're still using N (not $2N$) as the number of haploids

Last time

- We defined the coalescent as the stochastic process going back in time to common ancestors
- For constant population size, we proved that time to coalescence for a sample pair is exponential.
- We showed (math and simulation) that $E(T) = N$.
- We showed that we could estimate N from observed mutations if we knew the mutation rate
- Last time was models, today mostly application to making inferences about the real history of human populations.

Coalesence when population is changing

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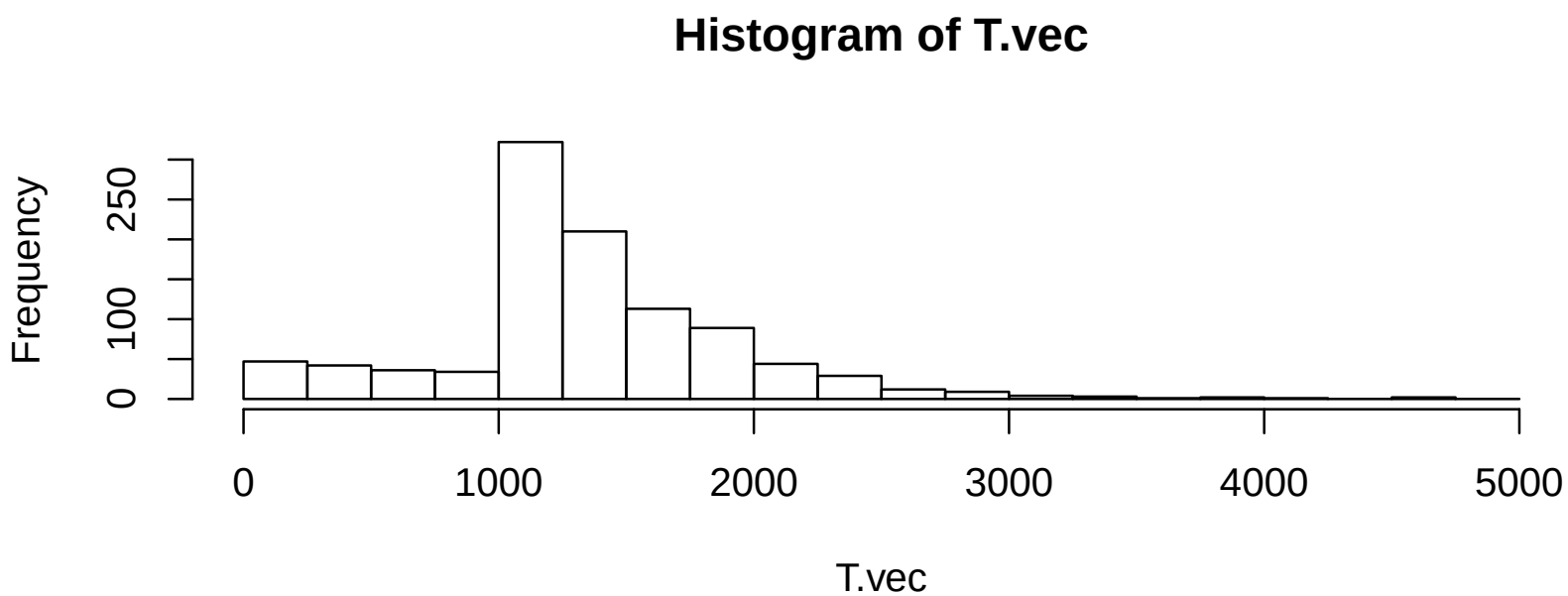
- If the population size changes over time $N(t)$, then hazards of coalescence in will change too: $h(t) = 1/N(t)$.

A small simulation

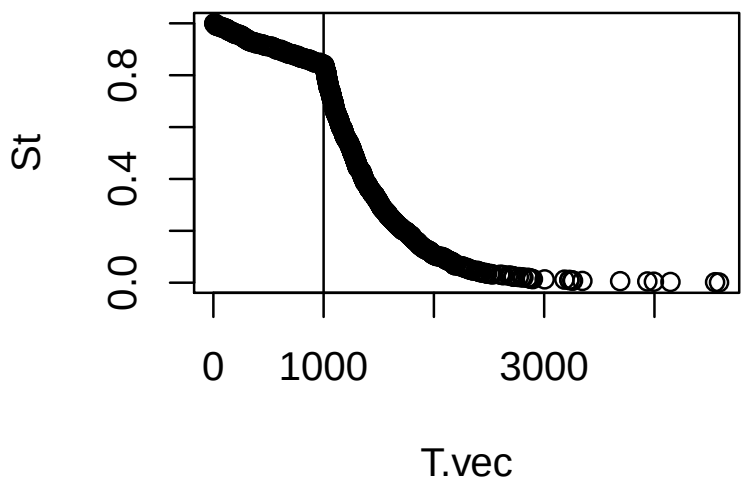
```
N_recent = 5000 ## population last T_thresh years
T_thresh = 1000
N_ancient = 500 ## earlier population
n = 1000 ## sampled individuals
set.seed(0.4886)
T1 <- rexp(n, rate = 1/N_recent) ## give everyone a chance to coalesce
T1[T1 > T_thresh] <- NA ## if they don't in 1st 1000 years, resample them
n2 <- sum(is.na(T1))
T2 <- T_thresh + rexp(n2, rate = 1/N_ancient) ## at ancient rate
T.vec <- c(T1, T2)
hist(T.vec, breaks = seq(0, 5000, 250))
```

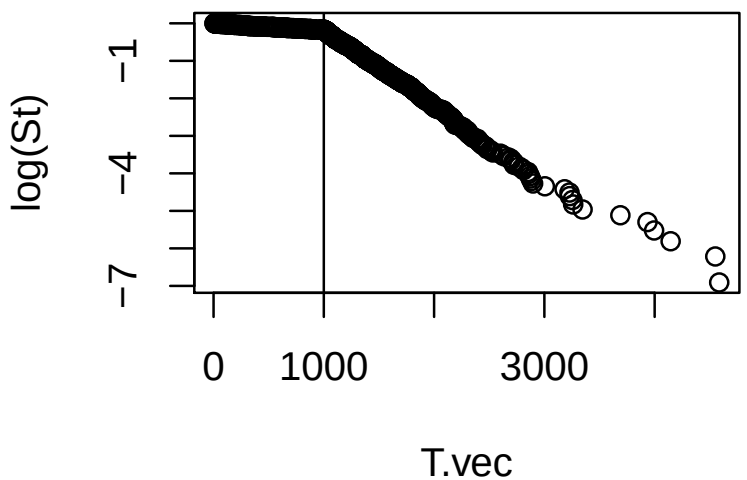


Q: How could we estimate population sizes from this histogram?

A small simulation, Demography Returns!

```
T.vec <- sort(T.vec)
St = (n:1)/n
par(mfrow = c(1,2))
plot(T.vec, St); abline(v = T_thresh); plot(T.vec, log(St)); abl
```





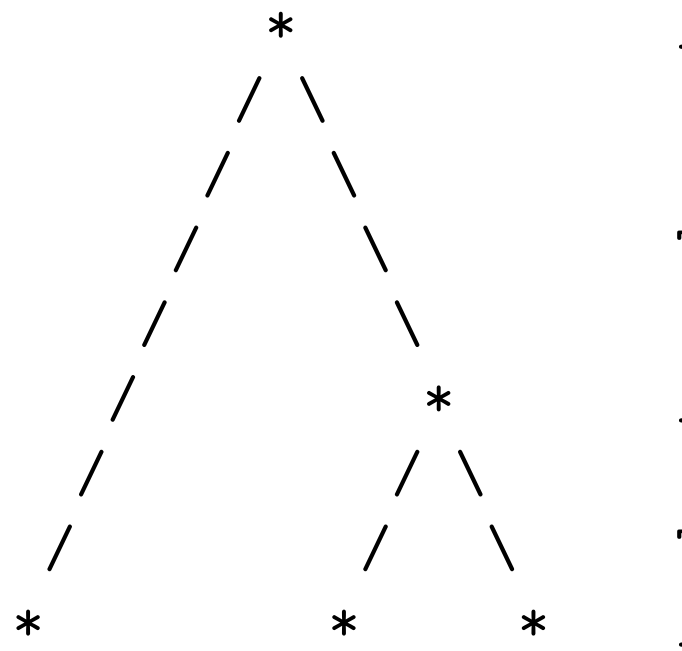
Q: How can we estimate hazards from this histogram?

Our approach

- Say we have i pairs of haploids
- We then compute how many pairwise differences there are, but instead of computing $\bar{k}$, we keep the distributional information k_i.
- Each k_i implies a T_i
- We then have a set of "death times" (coalescence times), can build a life table, estimate the hazards, and infer $N(t)$.

Detour to length of branches

A sample of 3: Note we're using N (not $2N$)



```
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T(2) : E(T(2)) = N

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T(3) : E(T(3)) = N * 2/[3*2]
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 ...

-------
T(n) : E(T(n)) = N * 2/[n * (n-1)]
```

Intuition: when we have more individuals, there's more chance that some pair of them will coalesce.

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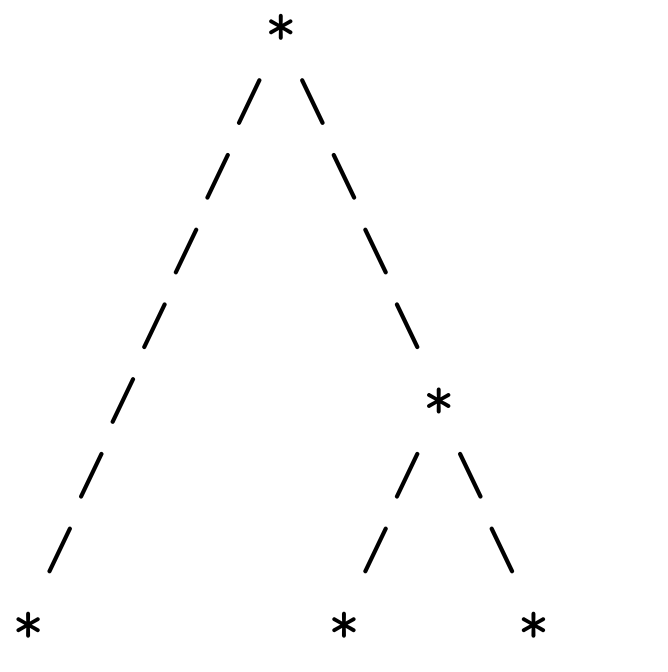

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Question: If we sample 4, how much of time to TMRCA do we have 4 branches, 3 branches, and 2 branches?

Last part: estimating actual historical pop sizes

Let's go to .Rmd in RStudio