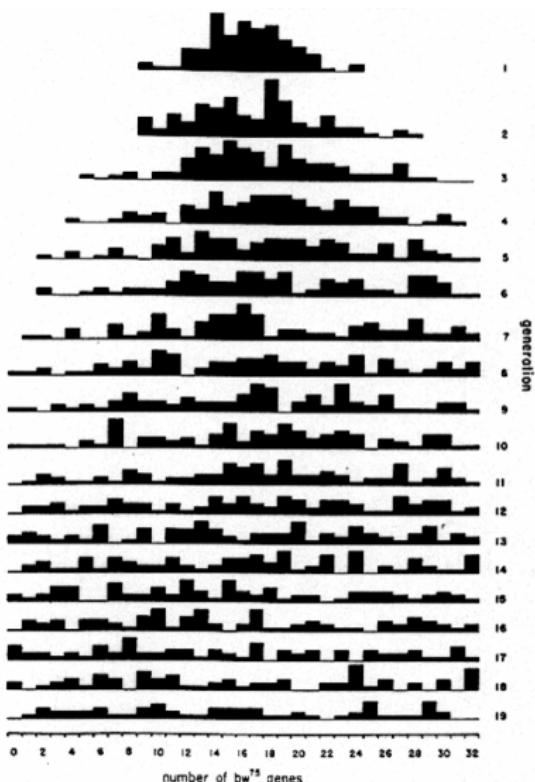
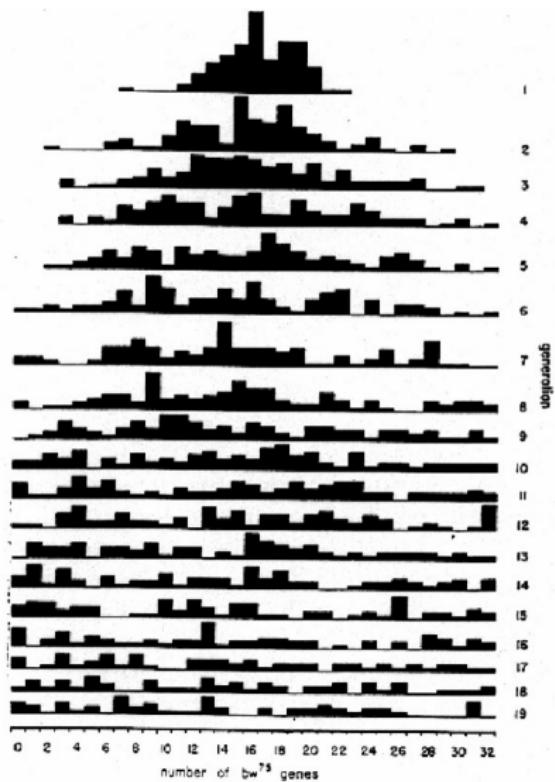


# Geographic Population Structure

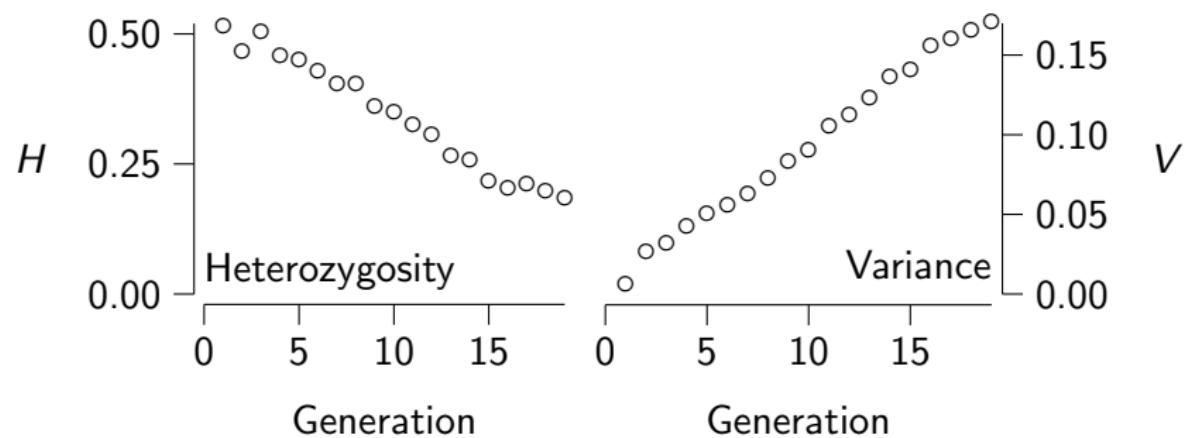
Alan R. Rogers

April 5, 2020

# Buri's (1956) data

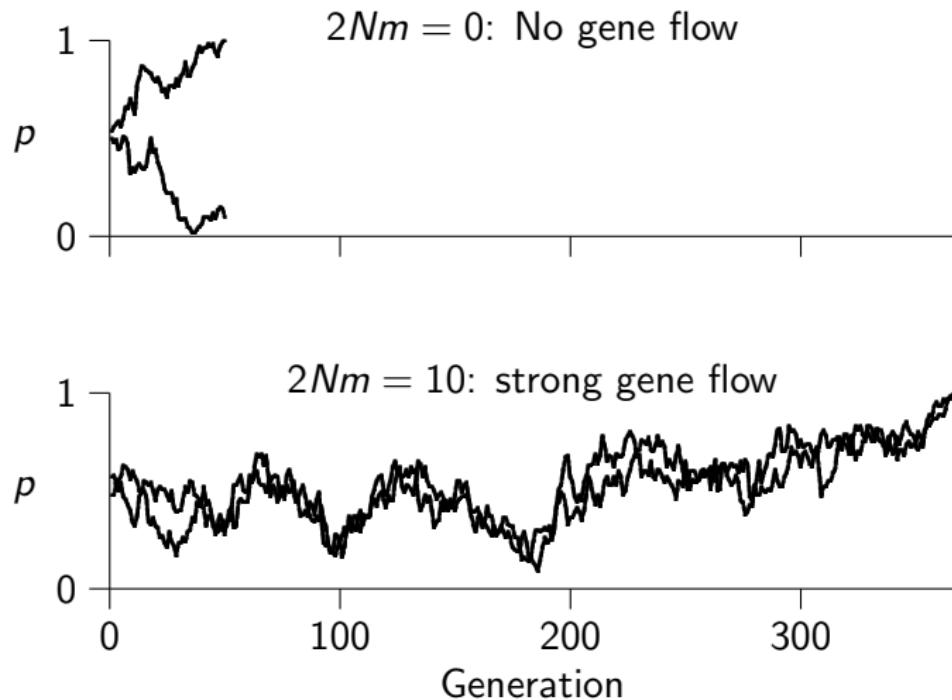


Drift reduces heterozygosity w/i groups while increasing variance among groups

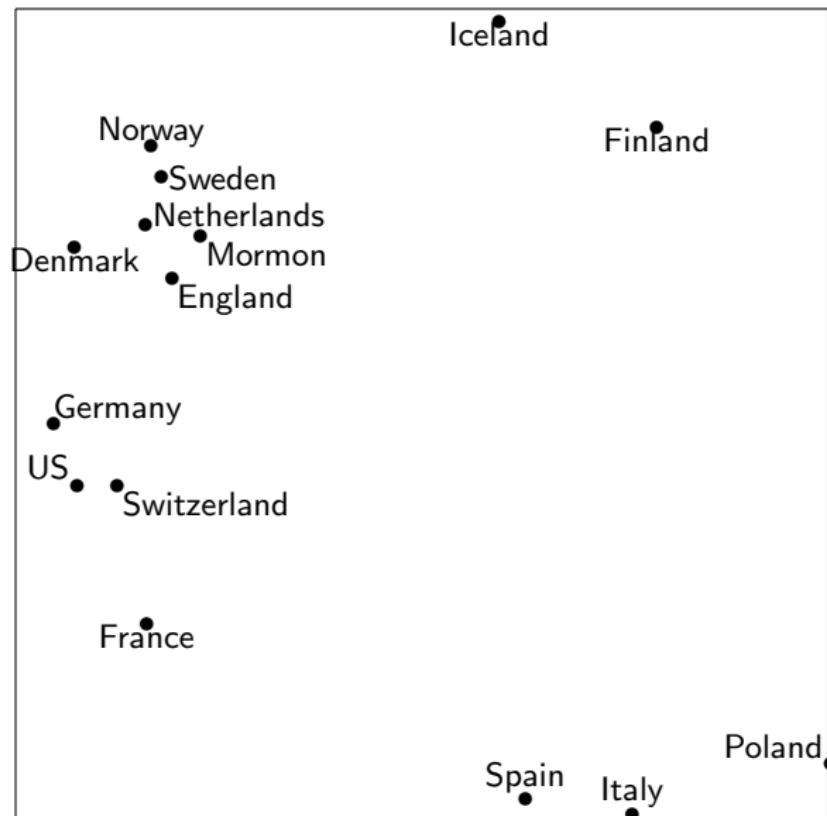


Data: Buri (1956)

## Gene flow (migration) reduces population differences

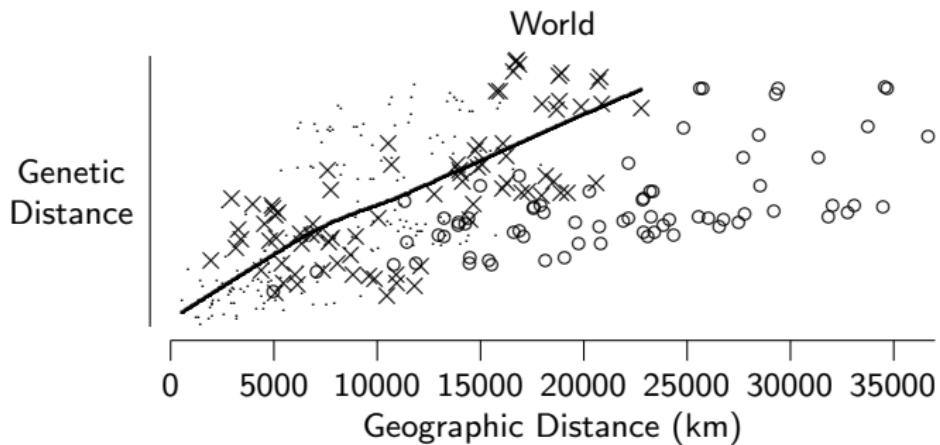
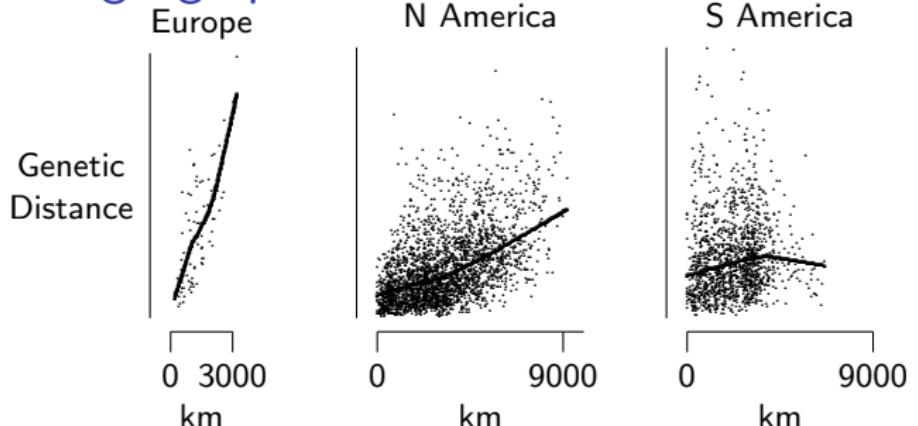


# PC map of European genetic distances



(McLellan et al  
1984)

# Genetic and geographic distance



× Australia and Pacific; ○ Americas

## Wahlund principle: subdivision reduces heterozygosity

	$A_1$	$A_1A_1$	$A_1A_2$	$A_2A_2$
Pop 1	4/16	1/16	6/16	9/16
Pop 2	12/16	9/16	6/16	1/16
Species	8/16	5/16	<b>6/16</b>	5/16
Hardy-Weinberg	8/16	4/16	<b>8/16</b>	4/16

Amount of reduction: 2/16

	$p_i - \bar{p}$	$(p_i - \bar{p})^2$
Pop 1	-4/16	1/16
Pop 2	4/16	1/16
Variance:		1/16

$$\mathcal{H}_S = \mathcal{H}_T - 2V \quad (\text{Wahlund 1928})$$

- ▶ Wahlund showed that heterozygosity is reduced by group differences in allele frequencies.
- ▶ Buri's experiment illustrates that
  - ▶ Drift reduces heterozygosity
  - ▶ Drift increases group differences
- ▶ We need a theory to connect these facts.
- ▶ Let us build one on top of what we have already.

## What we already know about heterozygosity

$$\begin{aligned} E[\mathcal{H}_t | p_t] &= 2p_t q_t && \text{(Hardy-Weinberg)} \\ E[\mathcal{H}_t | p_0] &= 2p_0 q_0 \left(1 - \frac{1}{2N}\right)^t && \text{(Ch. 2)} \end{aligned}$$

How can these both be true?

It must be true that

$$E[p_t q_t | p_0] = p_0 q_0 \left(1 - \frac{1}{2N}\right)^t$$

On the other hand, it is also true that

$$E[p_t q_t | p_0] = p_0 q_0 - V_t \quad (\text{Wahlund})$$

where  $V_t$  is the variance of  $p_t$  about  $p_0$ .

Rearranging gives the variance among groups

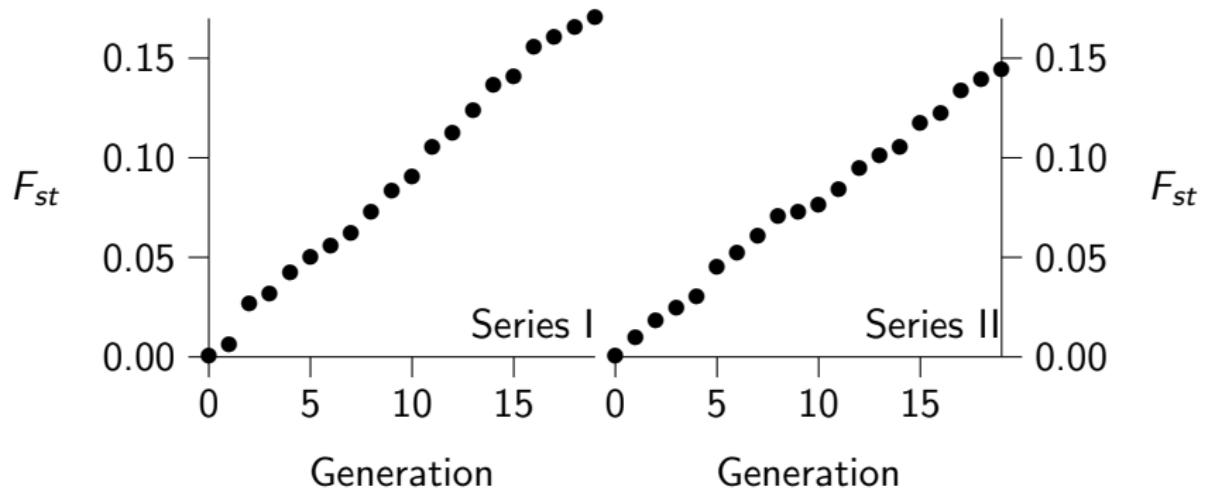
$$V = p_0 q_0 \left[ 1 - \left( 1 - \frac{1}{2N} \right)^t \right]$$

We usually normalize this expression by dividing both sides by  $p_0 q_0$ . The result is called  $F_{ST}$ :

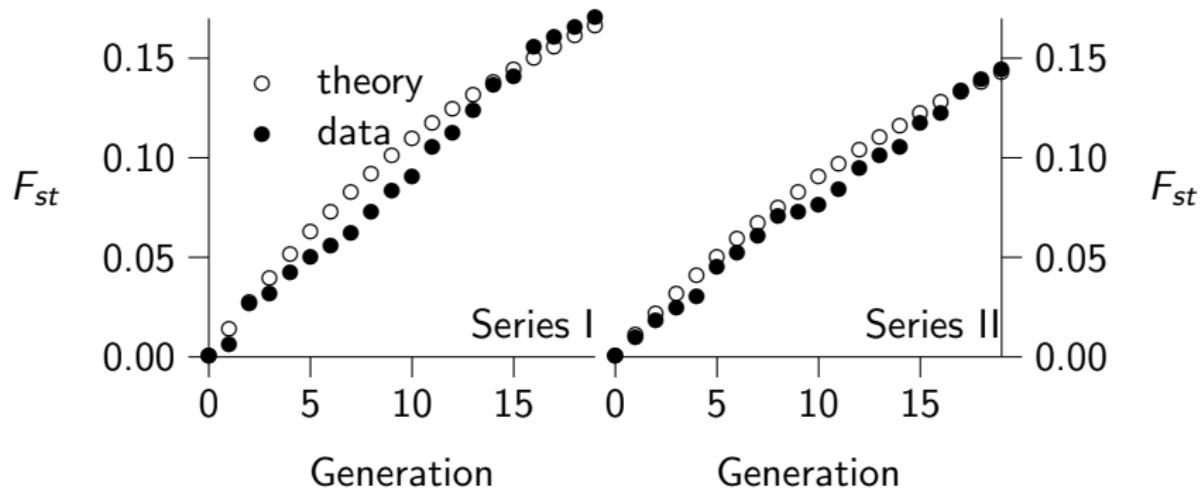
$$F_{ST} = \frac{V}{p_0 q_0} = 1 - \left( 1 - \frac{1}{2N} \right)^t$$

In data analysis, we take  $p_0 \approx \bar{p}$ , the current population mean.

Data from Buri (1956)



Data from Buri (1956)



## Application to differences among human races

Theory assumes *no* migration. Seems unlikely, but let's see where it leads.

For major human populations,  $F_{ST} \approx 0.1$ . Modern humans have been in Europe and E Asia for 50,000 years (2000 generations).

Plug this into

$$F_{ST} = 1 - \left(1 - \frac{1}{2N_e}\right)^t$$

and solve for  $N_e$ .

Answer:  $N_e \approx 10,000$ : agrees with genetic diversity within populations.

## The role of migration

- ▶ Drift increases group differences.
- ▶ Migration ( $m$ ) reduces them
- ▶ Eventually, an equilibrium is reached.

$$F_{ST} = \frac{1}{4N_e m + 1}$$

Depends only on  $N_e m$ . Small if  $N_e m > 1$ .

Human value ( $\approx 1/9$ ) implies  $N_e m \approx 2$ .

## Summary

- ▶ Drift increases variance among groups and reduces that within them.
- ▶ Wahlund's principle:  $\mathcal{H}_S = \mathcal{H}_T - 2V$
- ▶  $F_{ST}$  measures between-group variance relative to total variance.
- ▶ If major human populations have been isolated for 50,000 y, then  $N_e \approx 10,000$ .
- ▶ If they are at migration-drift equilibrium, then pairs of populations exchange  $\sim 2$  migrants per generation.