Recessive allele **l** codes for lactose intolerance. People who are recessive homozygots (ll) can not digest milk; dominant homozygots (LL) and heterozygotes (Ll) have no problem digesting milk.

Among **200** people from Eastern Mongolia, **18** were unable to digest milk.

fr (ll) = 18/200 = 0.09

Assuming that the population does not deviate from the Hardy-Weinberg equilibrium, *estimate* the frequency of each allele and each genotype:

p+q=1

fr(l) = **q = sqrt (q^2) = 0.3** fr(L) = **p =1 – q = 1 – 0.3 = 0.7**

fr(ll) = **q^2**

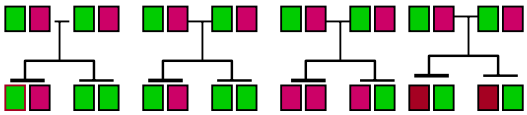
fr(Ll) = **2pq =2 x 0.7 x 0.3 = 0.42**

fr(LL) = **p^2 = 0.49**

Genetic Drift

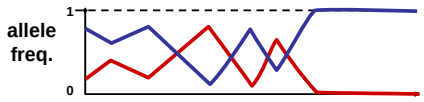
Genetic drift **–changes of allele frequencies due to sampling errors**

generation 1: fr(G) = fr (P) = 0.5



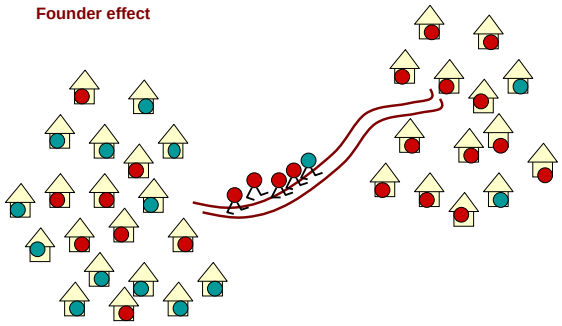
generation 2: fr(G) = 0.65 **fr (P) =** 0.45

Genetic Drift: allele frequency change



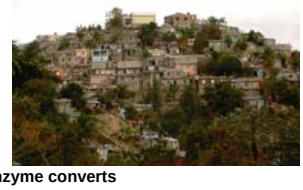
- **Drift has no direction**
- **Evolutionary change accumulates with time**
- **Causes loss of genetic variation in a population**
- **Increase of genetic difference between populations**
- **Two special forms of drift: founder effect and bottle neck**

Founder effect



Founder effect in Salinas village, Dominican Republic

one of the founders: Altagracia – heterozygous for a single base substitution in 5-alpha-reductase-2 autosomal gene

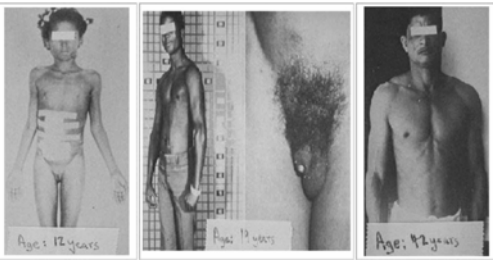


5-alpha-reductase-2 is an that **enzyme converts testosterone into dehydrotestosteron necessary for testes development**

low activity of this enzyme in homozygotes

XY individuals develop as females until puberty

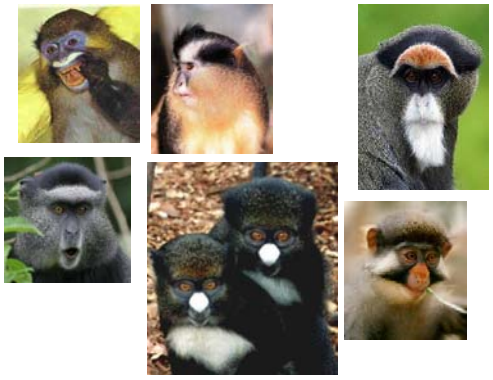
Guevedoces = “penis at 12”



Ectrodactyly, aka lobster claw syndrome

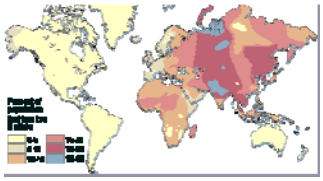
Vadoma -- a tribe in the west of Zimbabwe

Habitat fragmentation and founder effect in Guenons

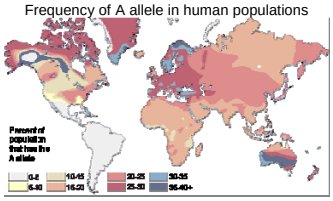


Blood groups

Frequency of B allele in human populations

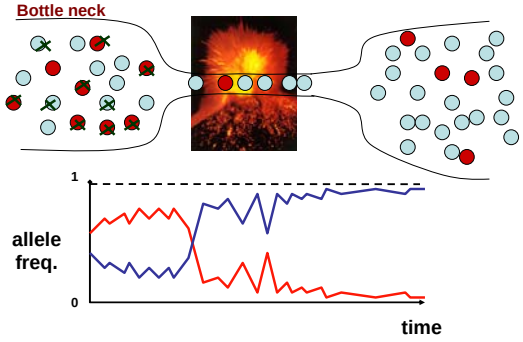


Frequency of A allele in human populations

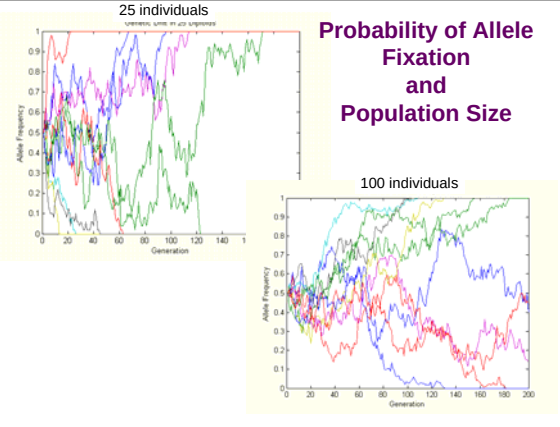


Bottle Neck - a type of genetic drift caused by a sudden change of population size

Bottle neck



Probability of Allele Fixation and Population Size



Probability of allele fixation

fr(A) = 0.5, fr (a) = 0.5 N=8

What is the probability (P) that all alleles in the next generation are A?

#genes = 2N =16

P(all A) = ½ x ½ x ½ …. = $(½)^{2N}$ =0.000015

P(all a) = ½ x ½ x ½ …. = $(½)^{2N}$=0.000015

P(fixed) = P(all A)+P(all a) = 2 x $(½)^{2N}$ = $2(0.5)^{16}$ =0.00003

fr (A) = 0.94, fr (a) = 0.06, N=8

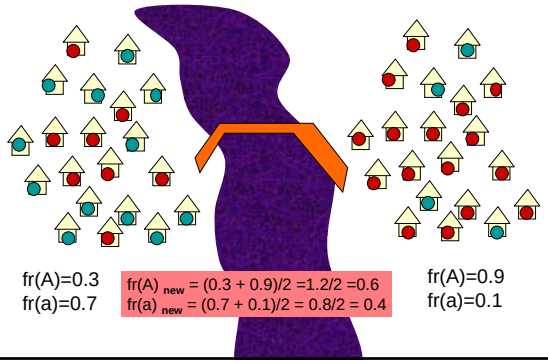
P(all A) = $[fr(A)]^{2N}$ =0.94^{16} =0.37

P(all a) = $[fr(a)]^{2N}$ =0.06^{16} =0.0000….

P(fixed) = 0.37

Gene flow

Gene flow **– is the transfer of alleles from one population to another**



fr(A)=0.3
fr(a)=0.7

$fr(A)_{new}$ = (0.3 + 0.9)/2 =1.2/2 =0.6
$fr(a)_{new}$ = (0.7 + 0.1)/2 = 0.8/2 = 0.4

fr(A)=0.9
fr(a)=0.1

Gene flow

- **Increases genetic variation within a population**
- **Decreases of genetic difference between populations**

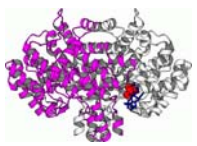
Relative Genetic Contribution

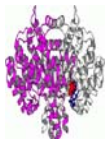
N=3,000	N=5,000
fr(A)=0.4	fr(A)=0.9
fr(a)=0.6	fr(a)=0.1

Add weights:

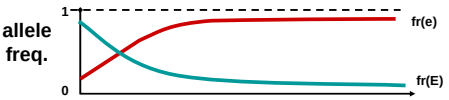
$fr(A)_{new}$ = (3x0.4 + 5x0.9)/(3+5) =(1.2+4.5)/8 =0.71
$fr(a)_{new}$ = 0.29

Mutations **– – changes in the sequence of a DNA molecule; introduce new alleles into a population**





E – normal enzyme mutation → ← reverse mutation **e – defective enzyme**



If not selected against, mutations will accumulate

Types of Mutations

somatic **gametic**

Only gametic mutations affect allele frequency in a population

A T T A C G C A C G T

Met stop

1. **silent (synonymous)**
2. **missense**
3. **nonsence – introduce stop codon**
4. **indels (insertions/deletions) → frame shift**

new codons: A T G A A G C A C G T