Recessive allele $l$ codes for lactose intolerance. People who are recessive homozygous ($ll$) cannot digest milk; dominant homozygotes ($LL$) and heterozygotes ($Ll$) have no problem digesting milk.

Among 200 people from Eastern Mongolia, 18 were unable to digest milk. Assuming that the population does not deviate from the Hardy-Weinberg equilibrium, estimate the frequency of each allele and each genotype:

- $f_r(l) = \frac{18}{200} = 0.09$
- $f_r(L) = 0.90$
- $f_r(ll) = 0.09$
- $f_r(Ll) = 2 \times 0.7 \times 0.3 = 0.42$
- $f_r(LL) = 0.42$

Genetic Drift

- Changes of allele frequencies due to sampling errors
- Generation 1: $f_r(G) = f_r(P) = 0.5$
- Generation 2: $f_r(G) = 0.65$, $f_r(P) = 0.45$

Founder effect

- One of the founders: Altagracia
- Heterozygous for a single base substitution in 5-alpha-reductase-2 autosomal gene
- 5-alpha-reductase-2 is an enzyme that converts testosterone into dehydrotestosterone necessary for testes development
- Low activity of this enzyme in homozygotes
- XY individuals develop as females until puberty

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 bottleneck
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

Probability of Allele Fixation and Population Size
Probability of allele fixation

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

#genes = 2N = 16

\[ P(\text{all A}) = \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} \ldots = \left(\frac{1}{2}\right)^{2N} = 0.000015 \]

\[ P(\text{all a}) = \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} \ldots = \left(\frac{1}{2}\right)^{2N} = 0.000015 \]

\[ P(\text{fixed}) = P(\text{all A}) + P(\text{all a}) = 2 \times \left(\frac{1}{2}\right)^{2N} = 2(0.5)^{16} \]

= 0.00003

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

\[ P(\text{all A}) = \left(\frac{94}{100}\right)^{16} = 0.37 \]

\[ P(\text{all a}) = \left(\frac{6}{100}\right)^{16} \approx 0.0000\ldots \]

\[ P(\text{fixed}) = 0.37 \]

Gene flow

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

Relative Genetic Contribution

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

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

Add weights:

\[ fr(A)_{\text{new}} = \frac{(3 \times 0.4 + 5 \times 0.9)}{8} = 0.71 \]

\[ fr(a)_{\text{new}} = 0.29 \]

Mutations

- Changes in the sequence of a DNA molecule; introduce new alleles into a population

Types of Mutations

- Silent (synonymous)
- Missense
- Nonsense (introduce stop codon)
- Indels (insertions/deletions) → frame shift

New codons: A T G A A G C A C G T