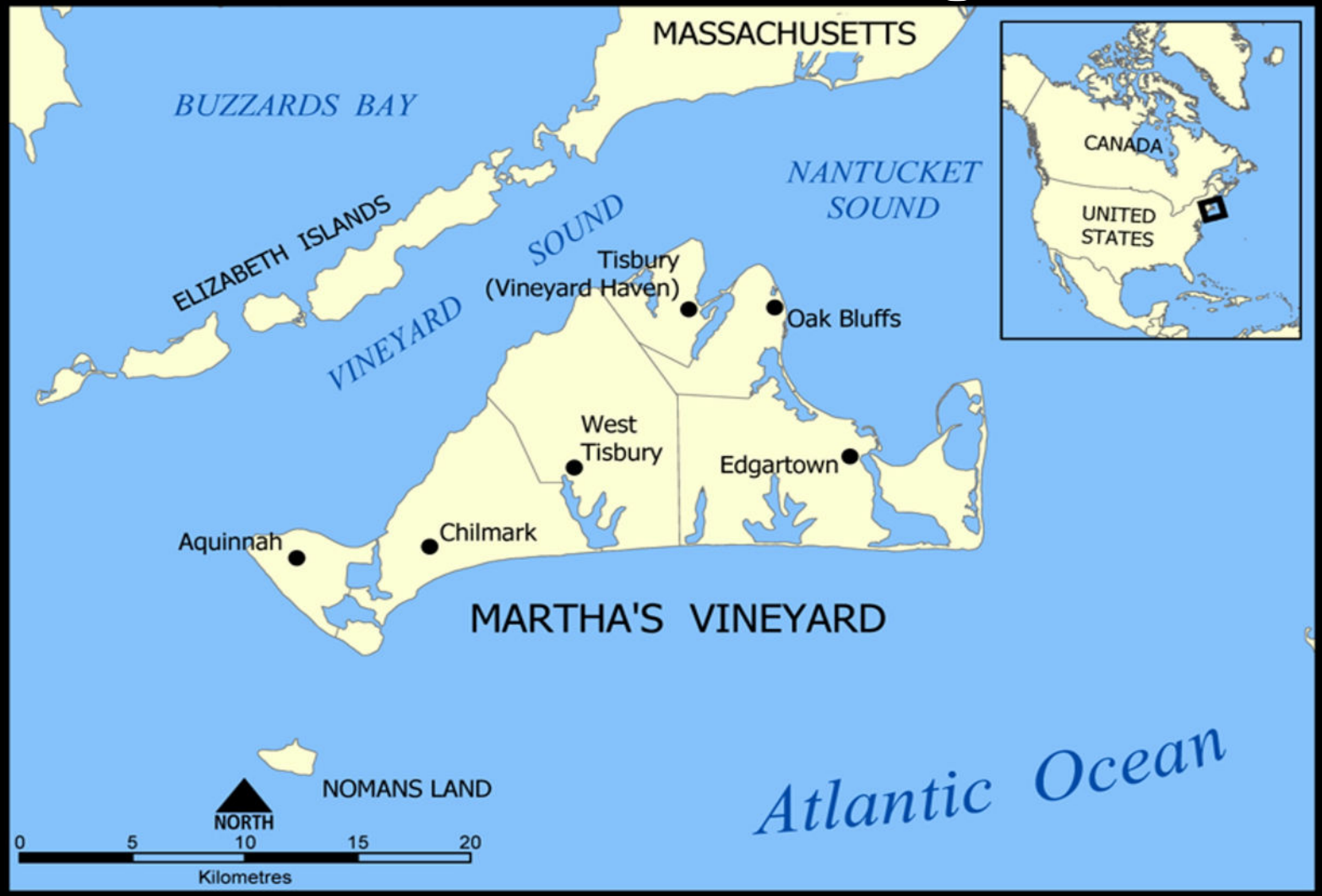
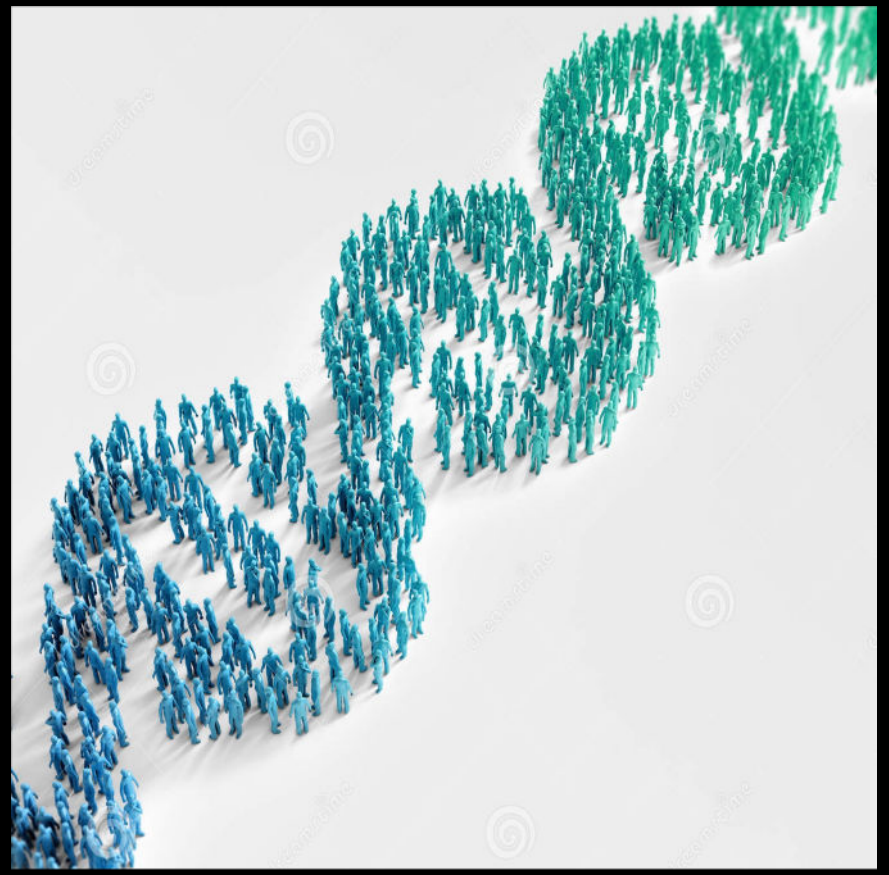
Deafness on Martha's Vineyard: A Real-World Case for Teaching Genetic Models



Objectives

- Connect Martha's Vineyard migration history to shifts in allele frequencies.
- Practice allele counting and Hardy–Weinberg math.
- Describe evolutionary mechanisms shaping Island hereditary deafness.
- Apply historical context to strengthen quantitative reasoning.



Handouts available for practice calculations

Case study: 200 years of deafness on Martha's Vineyard



You will take the role of ***CJB2* gene**

- Also known as Connexin-26

Before Martha's Vineyard

- The Weald, England
- Old Kent sign language
- Hereditary deafness in Lambert, Mayhew, Tilton, West, Skiffe families
  - Likely ***CJB2* gene**

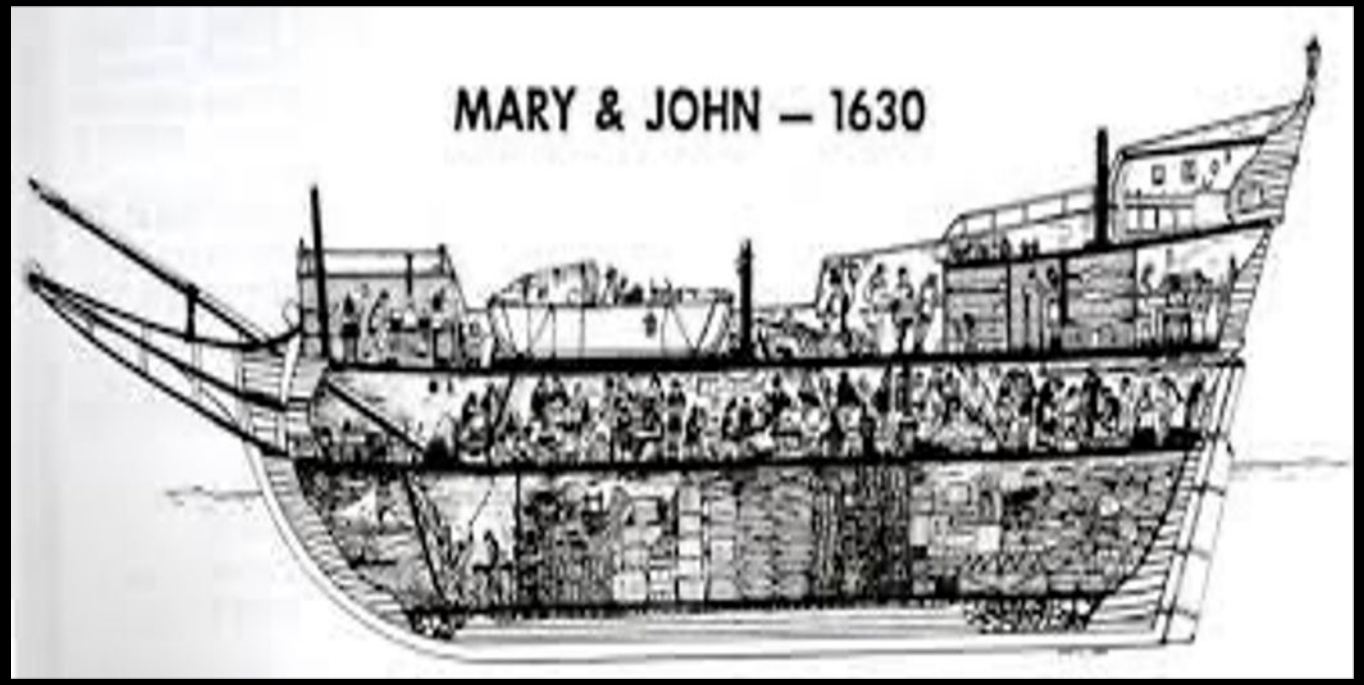




The Weald, England



England → Massachusetts → Martha's Vineyard





Jonathan Lambert – first known Deaf Islander

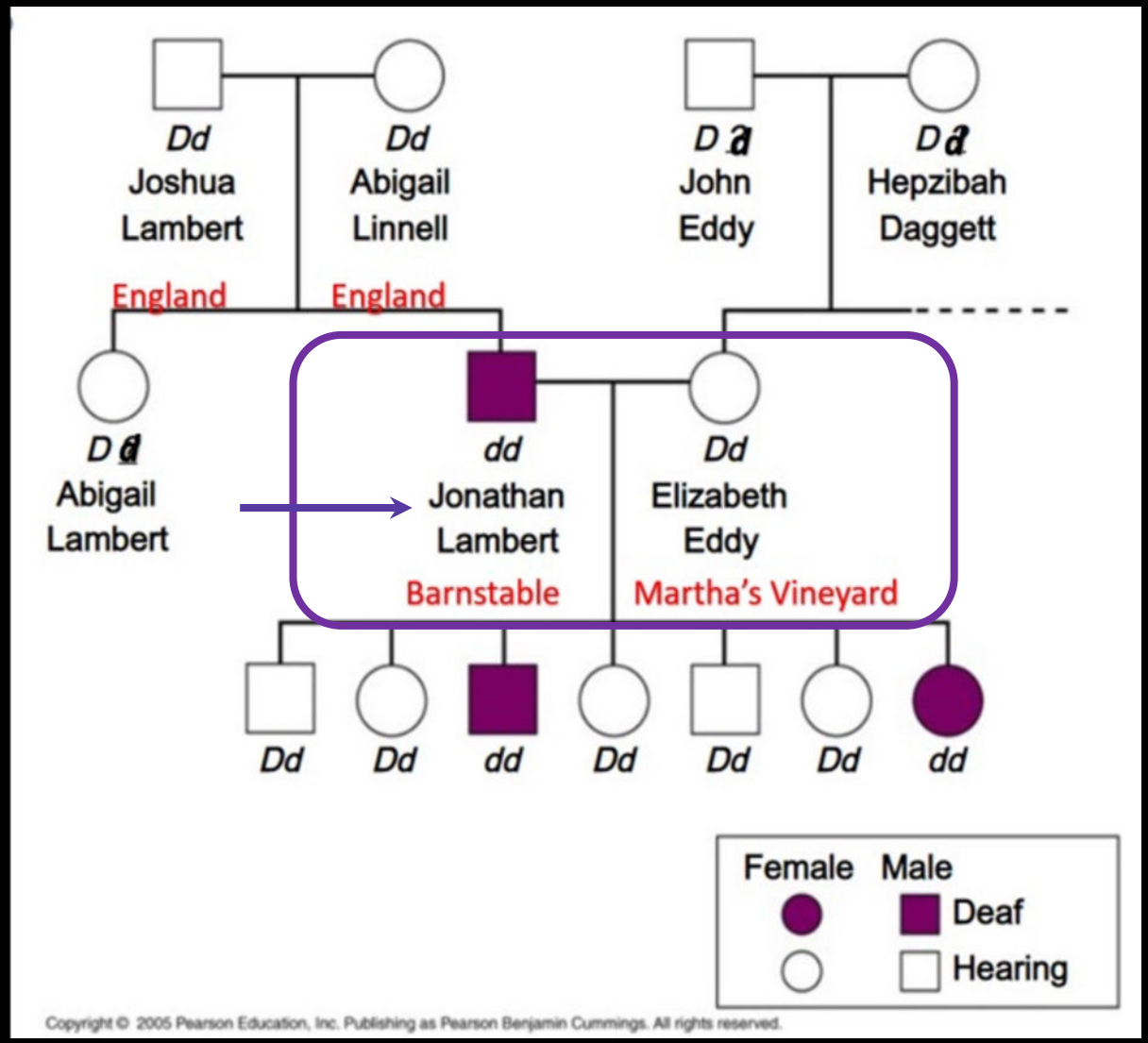
1683: Jonathan married Elizabeth Eddy of Martha's Vineyard

- 7 children: 2 deaf
- Likely ***CJB2* gene**

Introduced Old Kent Sign Language to Island

Carpenter & farmer

Home: Lambert's Cove



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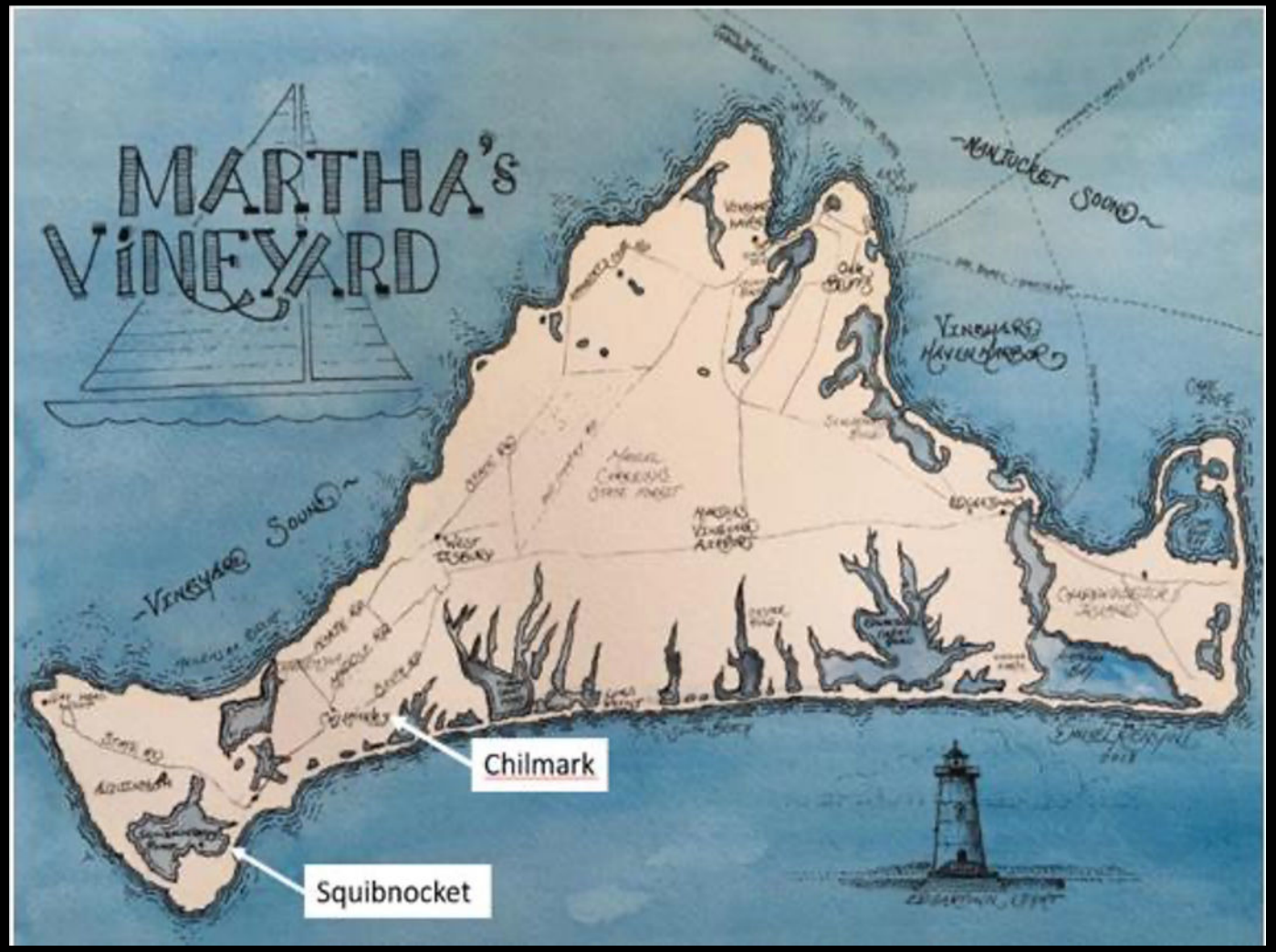
Deaf Islanders

Immigration slowed after 1710

- Island population ~400

- Deaf Islanders: 1 in 155
- Chilmark: 1 in 25
- Squibnocket: 1 in 4

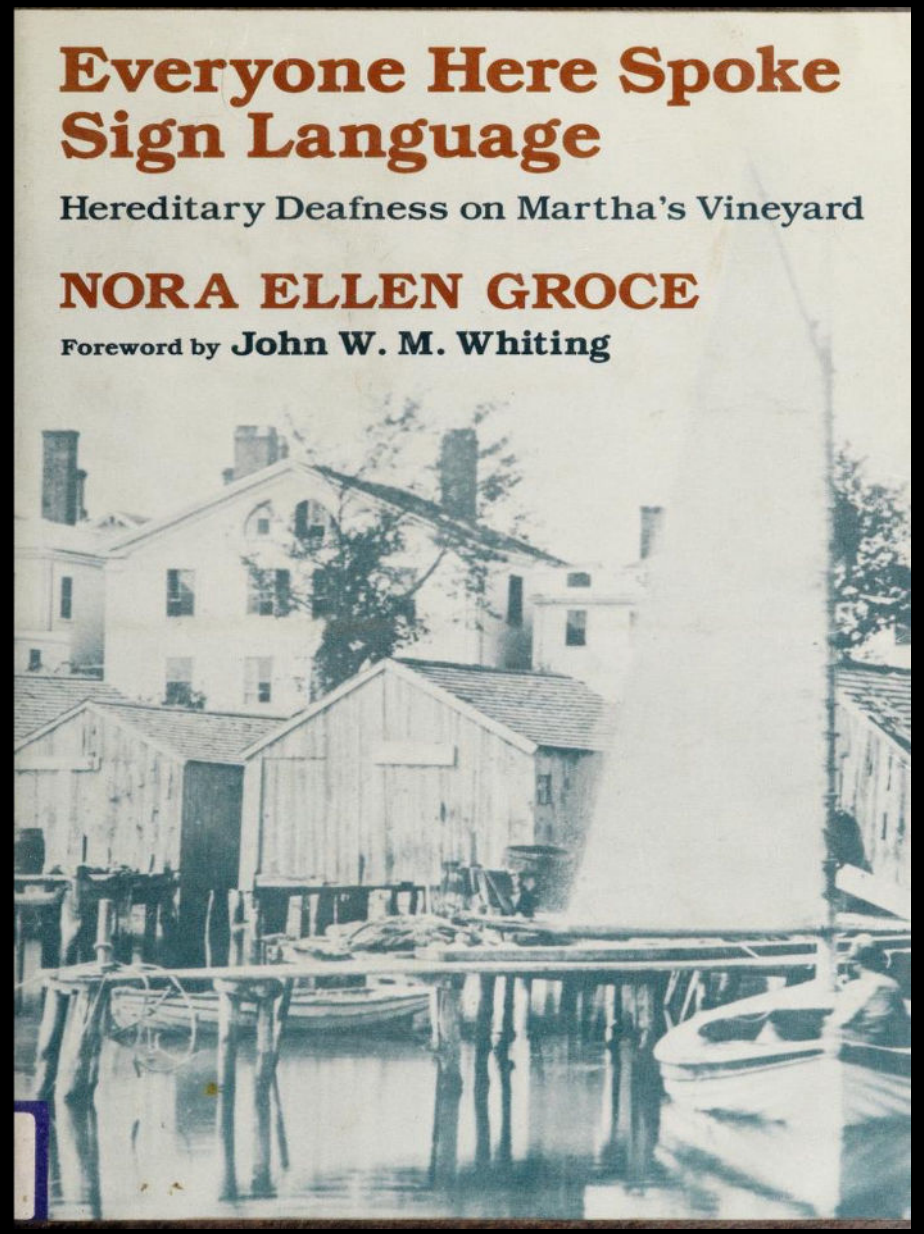
- Mainland comparison: 1 in 5700



All islanders fluent in Martha Vineyard Sign Language (MVSL)

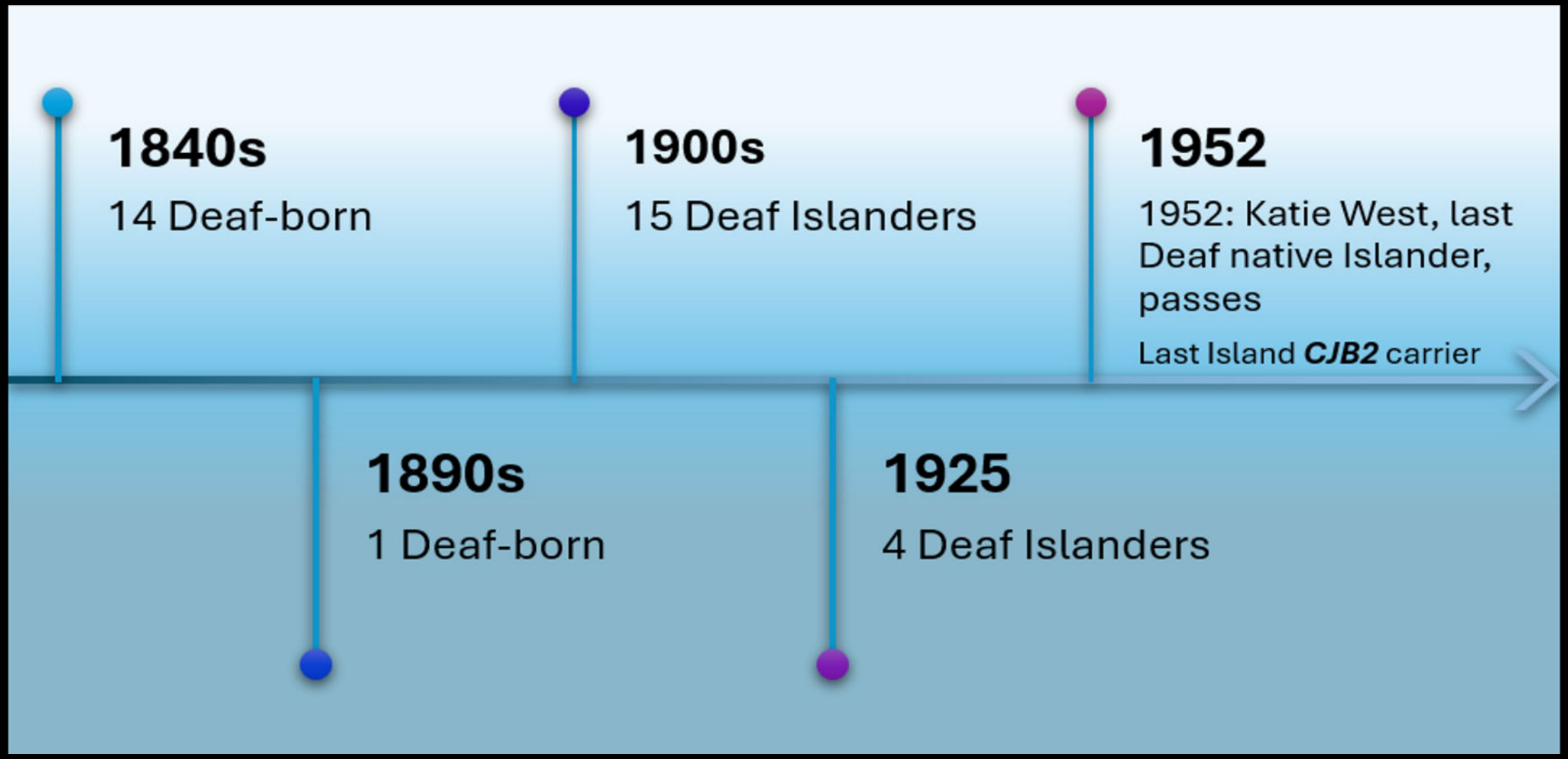
Being Deaf was normal

- Deaf owned farms and businesses, served in town government.
- Signing was a community life skill, like fishing.
- MVSL learned through families and daily interaction, no formal instruction.
- MVSL belonged to the Island, used by all residents.



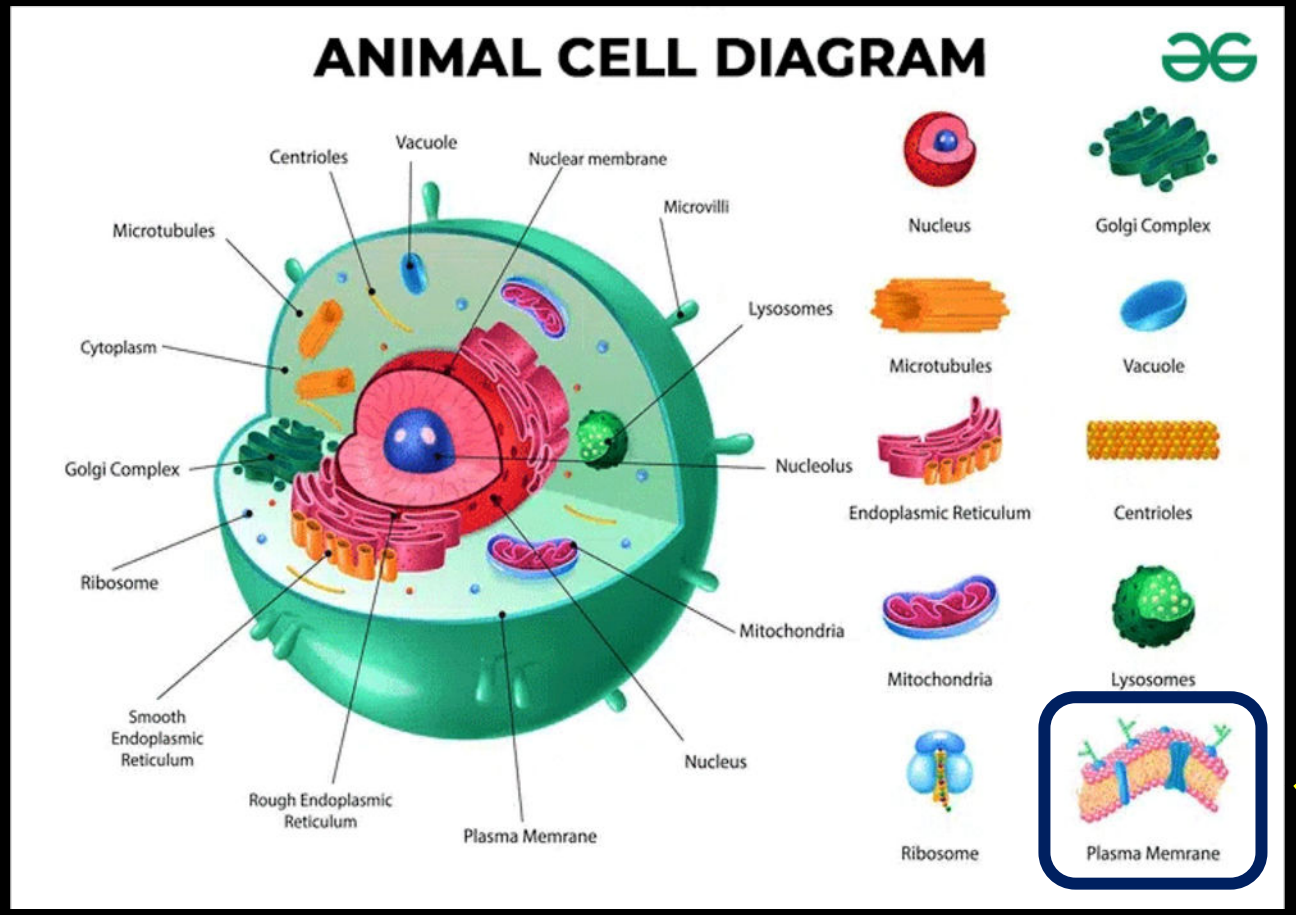
Due to time, MV language and culture evolution not included today. See me if interested

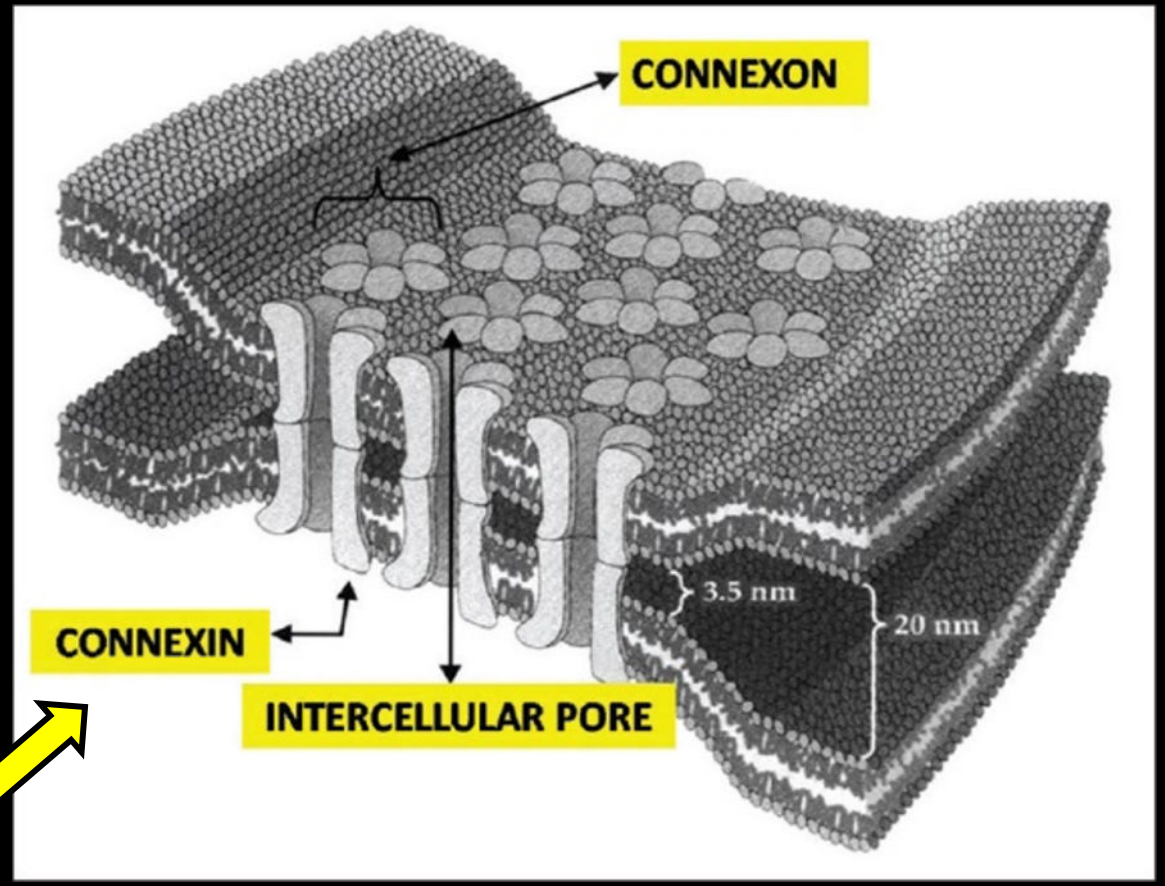
Extinction of Island Deafness and MVSL



CJB2 and hereditary deafness

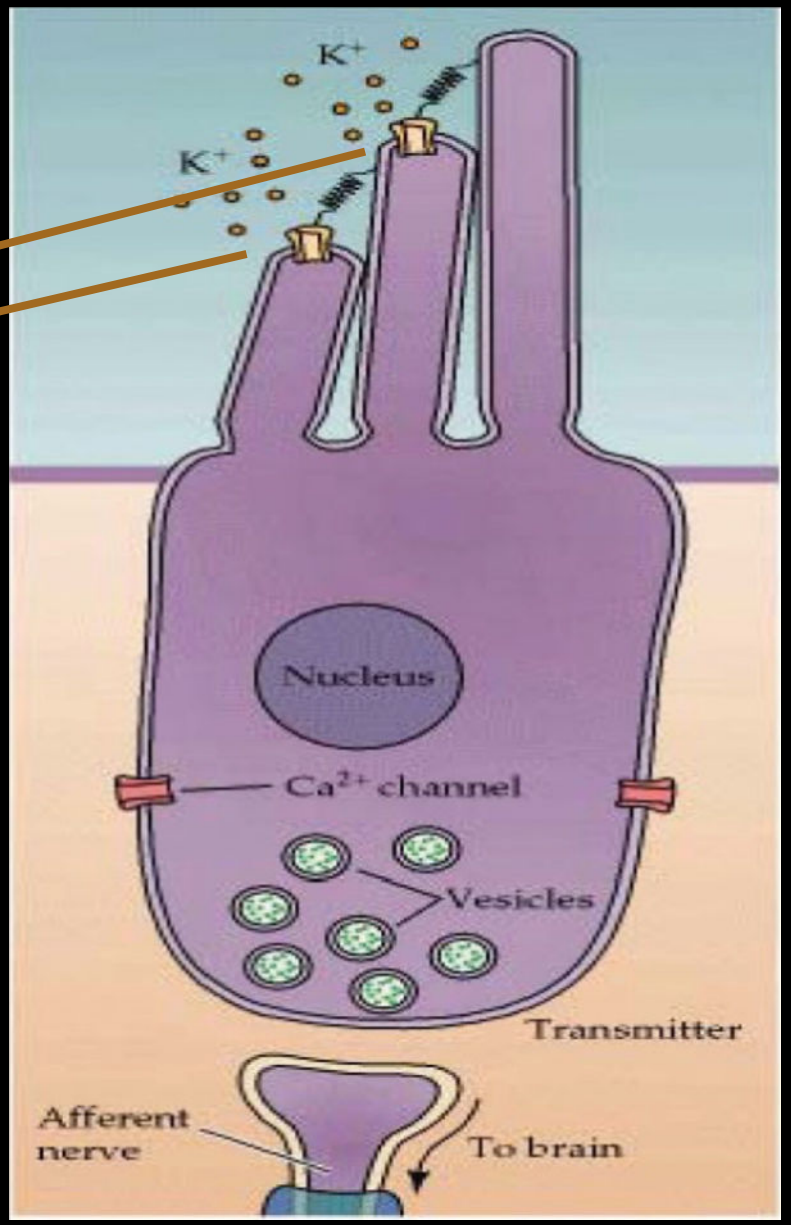
- Connexin-26 (*CJB2*) mutations: most common cause of autosomal recessive nonsyndromic deafness

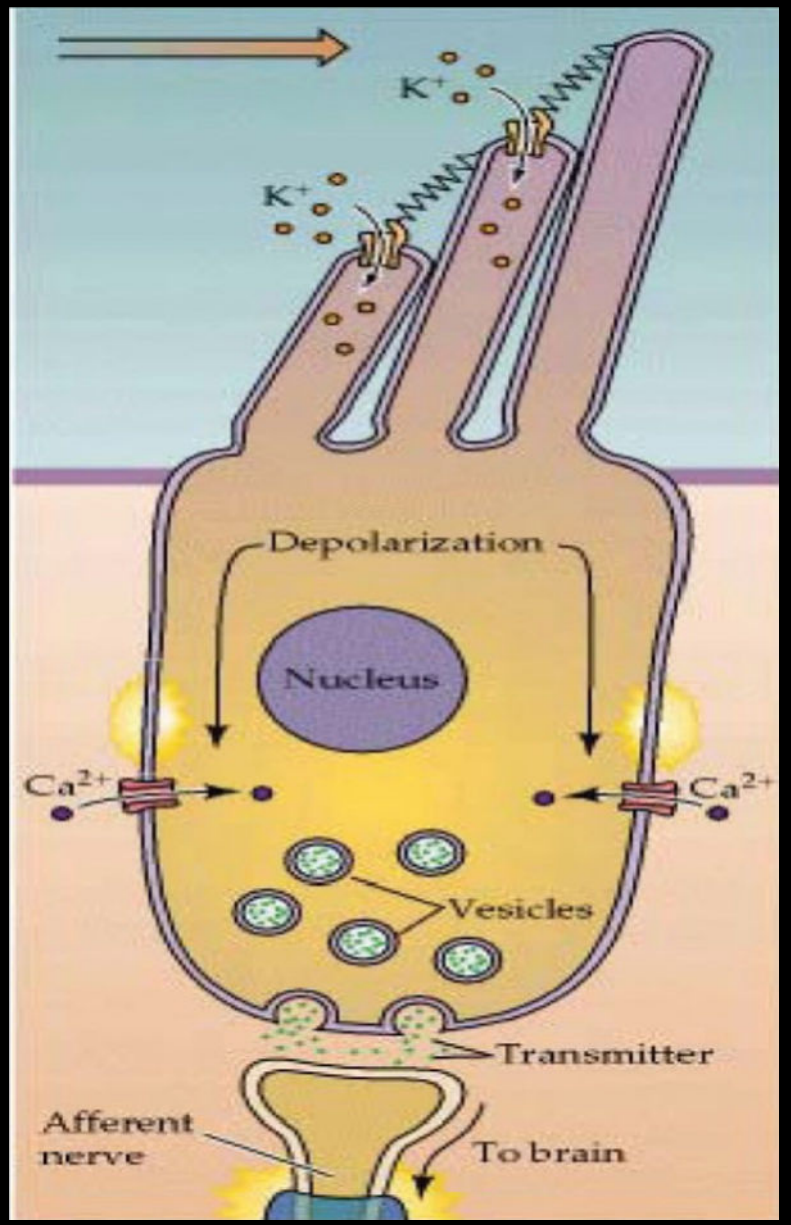




Connexon channels

Hair cell at rest





1) Sound waves bend hair cells

2) Bending opens connexon channels

3) Potassium (K^+) flows into hair cell

4) K^+ opens calcium (Ca^{2+}) channels → neurotransmitters → brain

Connexon mutations block K^+ flow → no signal → no hearing.

Martha’s Vineyard deafness and population genetics

What is population genetics?

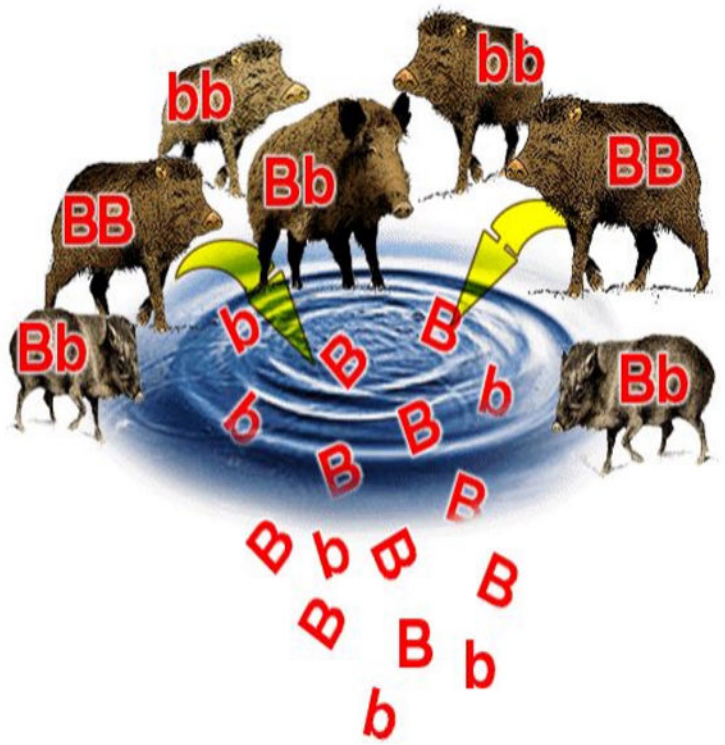
- Tracks allele frequency in a population
- Shows allele transmission to the next generation
- *Populations evolve, not individuals!*
  - Evolution = allele frequency change over time

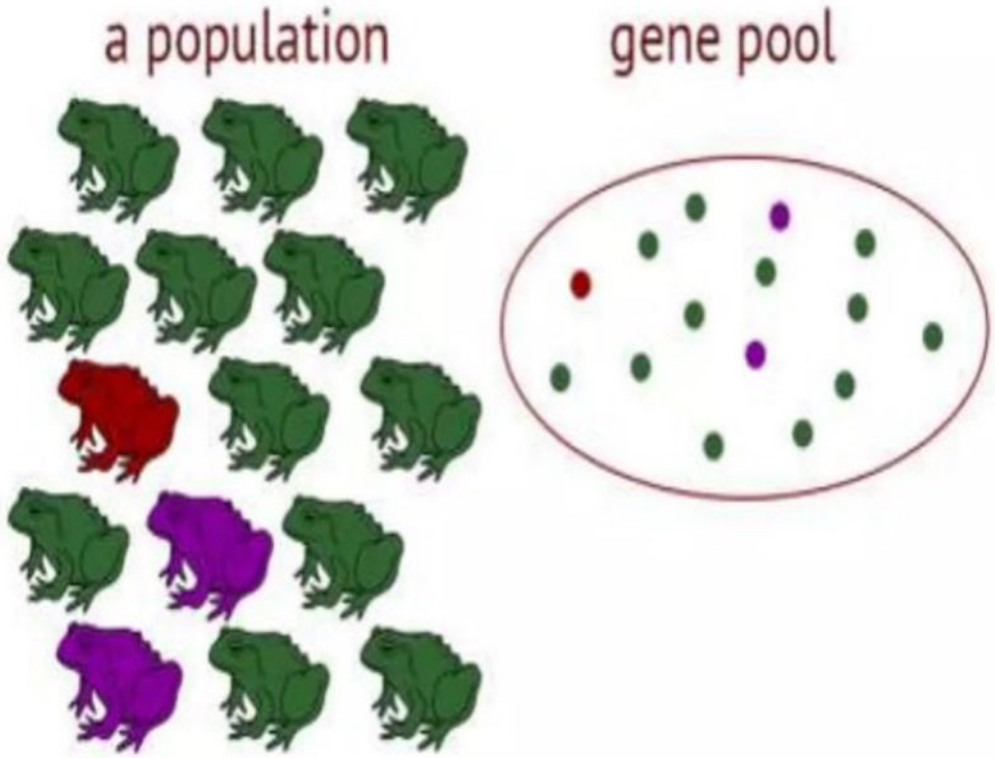


Key terms

Gene Pool

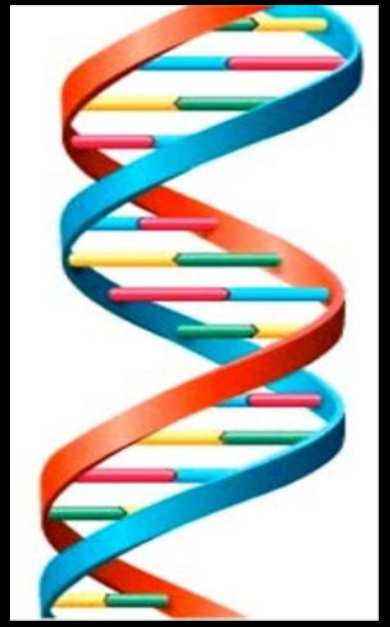
- Is the combination of all the genes in one population put together





Phenotype and Genotype

Genotype = genes an organism inherits



Phenotype = physical expression of genes

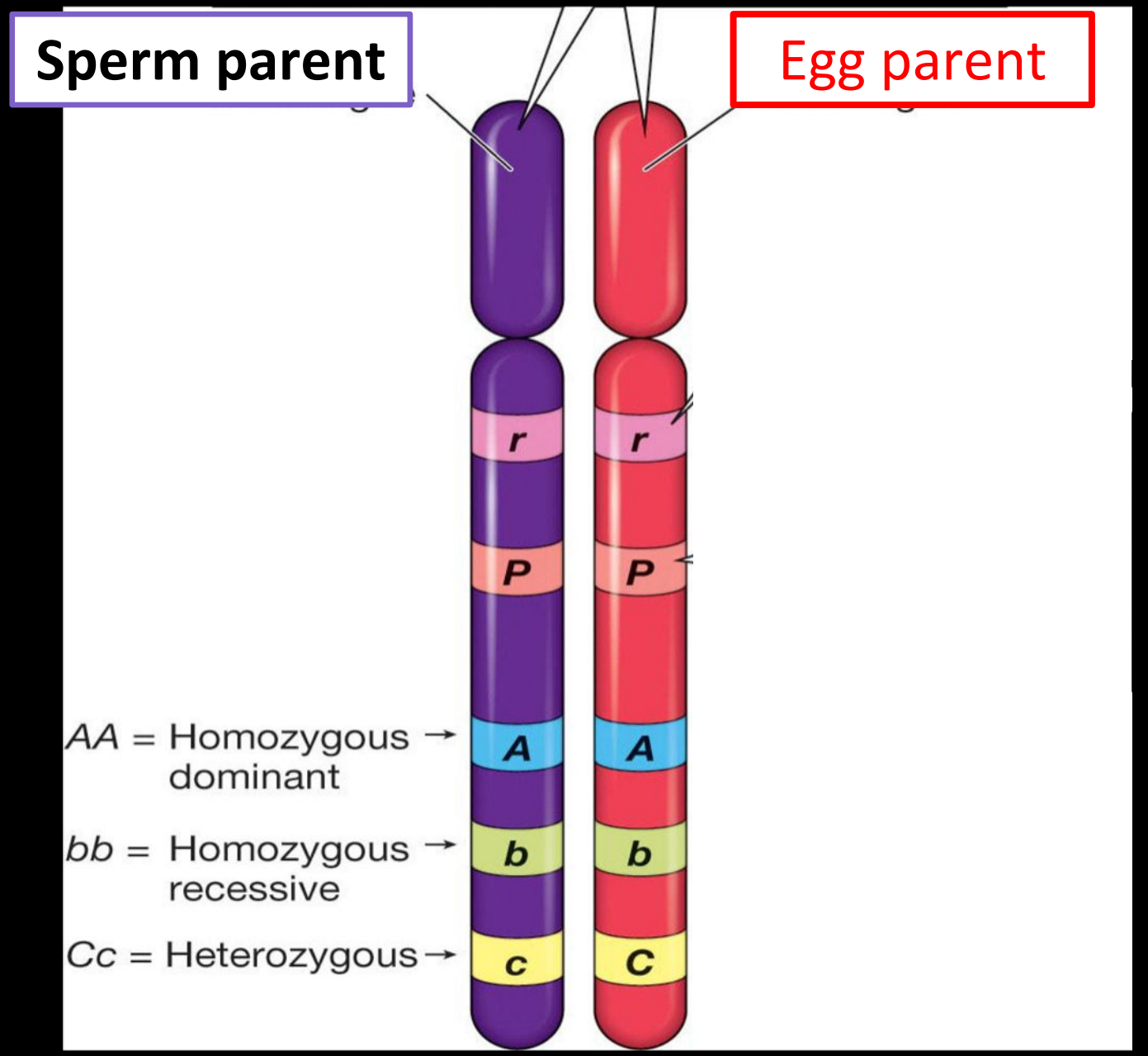


Allele: gene version that codes for a trait

Homozygous: Same alleles

- Homozygous dominant → **AA**
- Homozygous recessive → **bb**

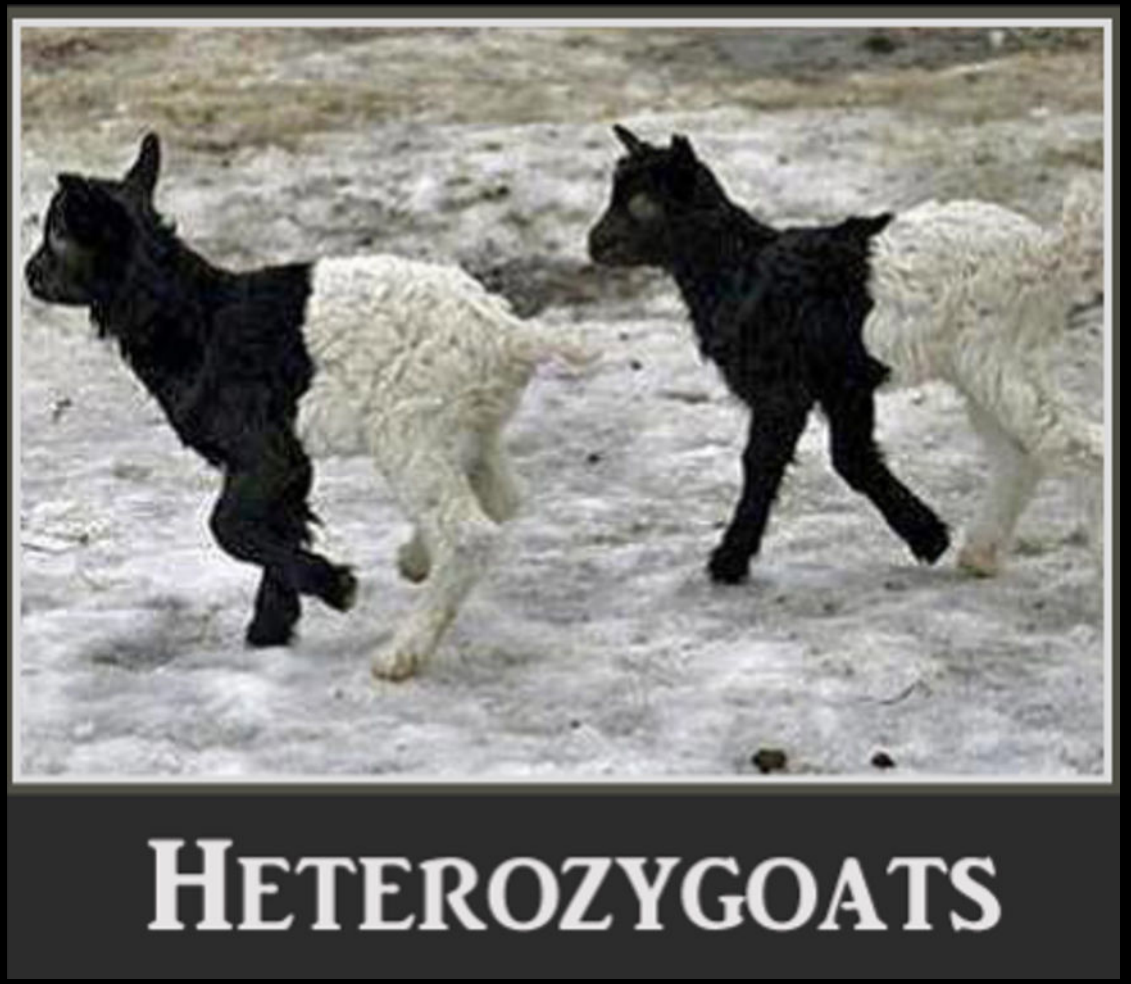
Heterozygous: Different alleles → **Cc**





Homozygoats

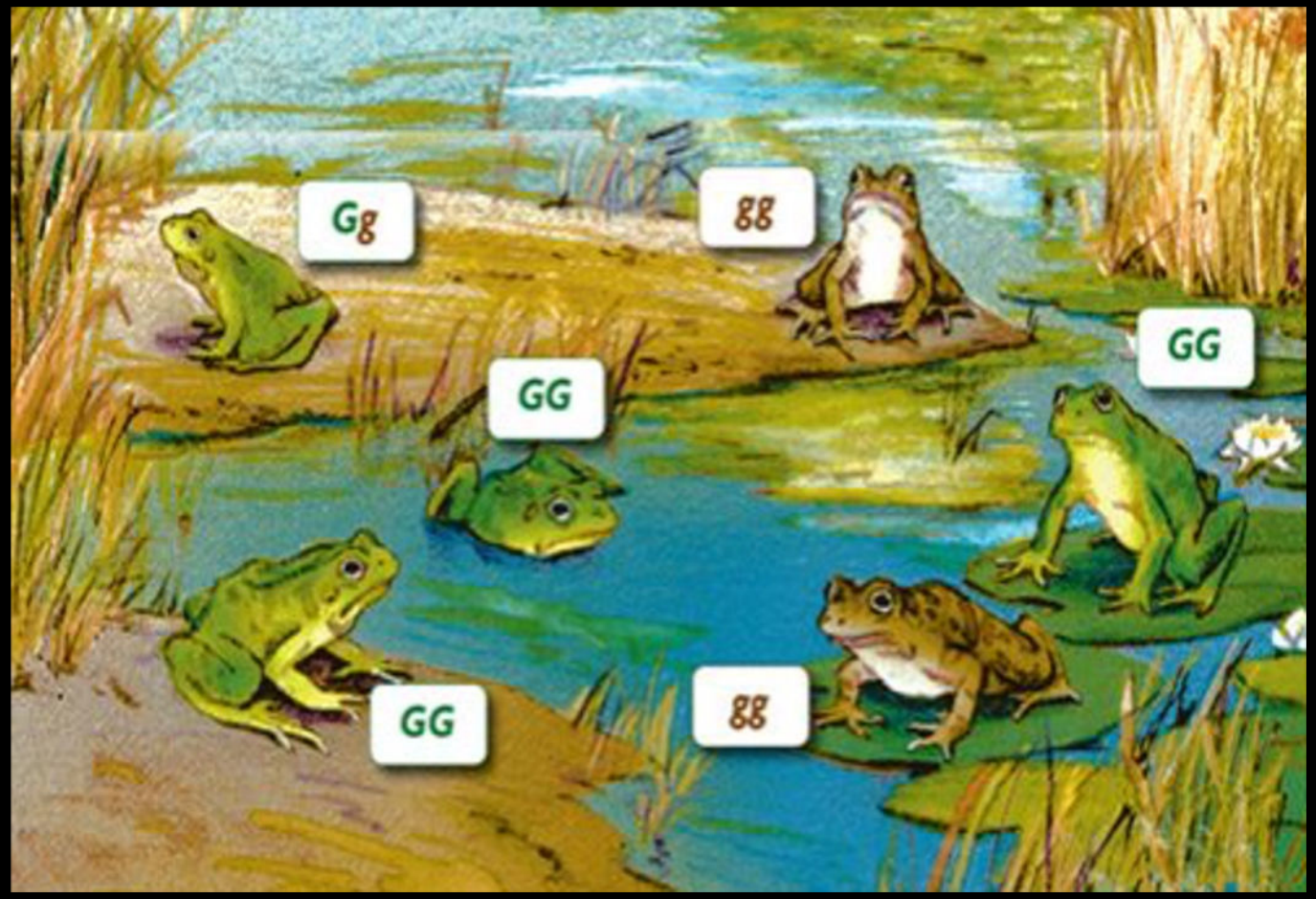
Homozygous: **DD** or **dd**



Heterozygous: **Dd**

Allele frequency

Gene pool: Allele frequency



G codes for green

7 Gs in gene pool

g codes for brown

5 gs in gene pool

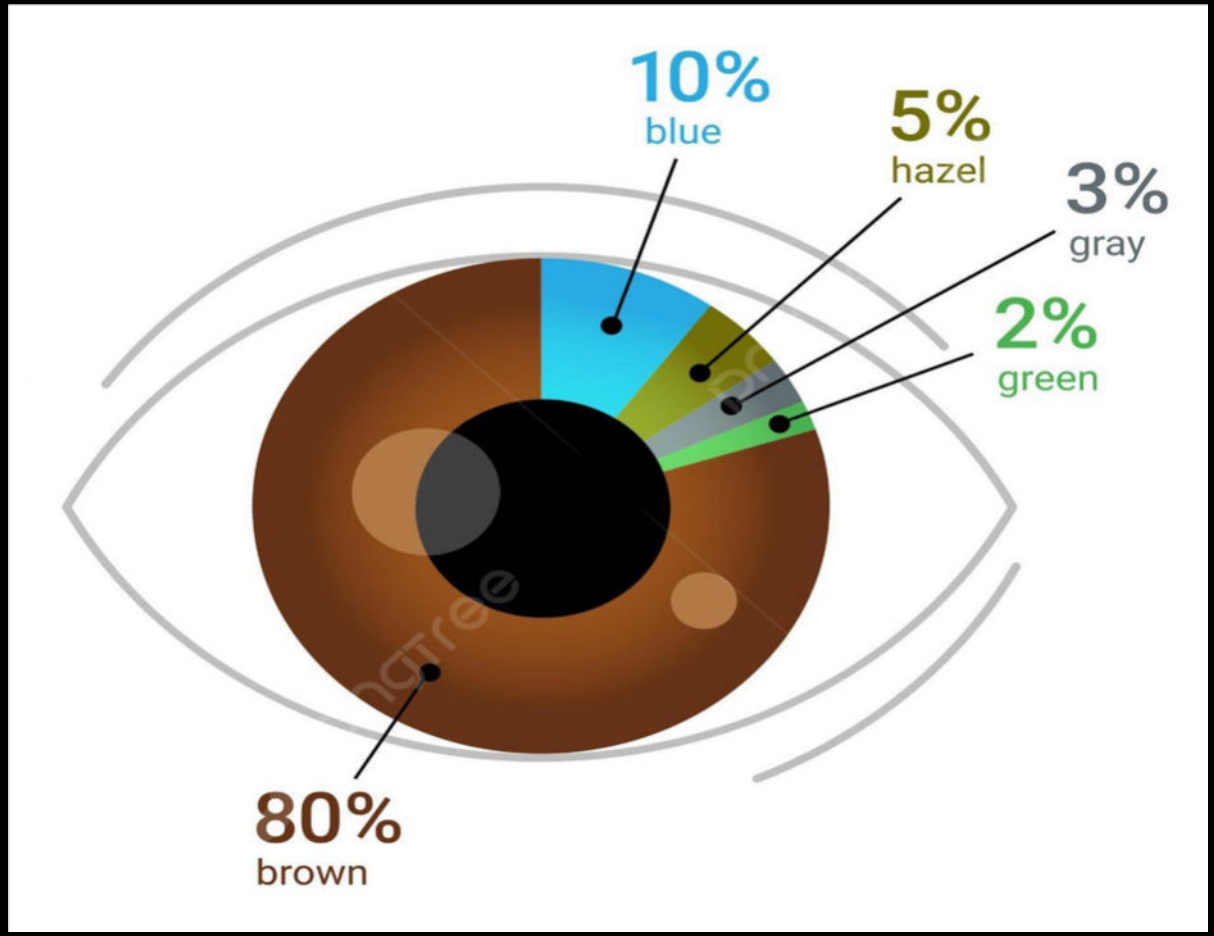
12 total alleles for skin color trait in gene pool

Frequency of allele $G = \frac{7}{12} =$ = 0.583 ≈ 58.3%

Frequency of allele $g = \frac{5}{12} =$ = 0.417 ≈ 41.7%

Allele frequency example: Eye color gene pool

- B = brown
- b = not brown



Allele frequency example: Class eye color

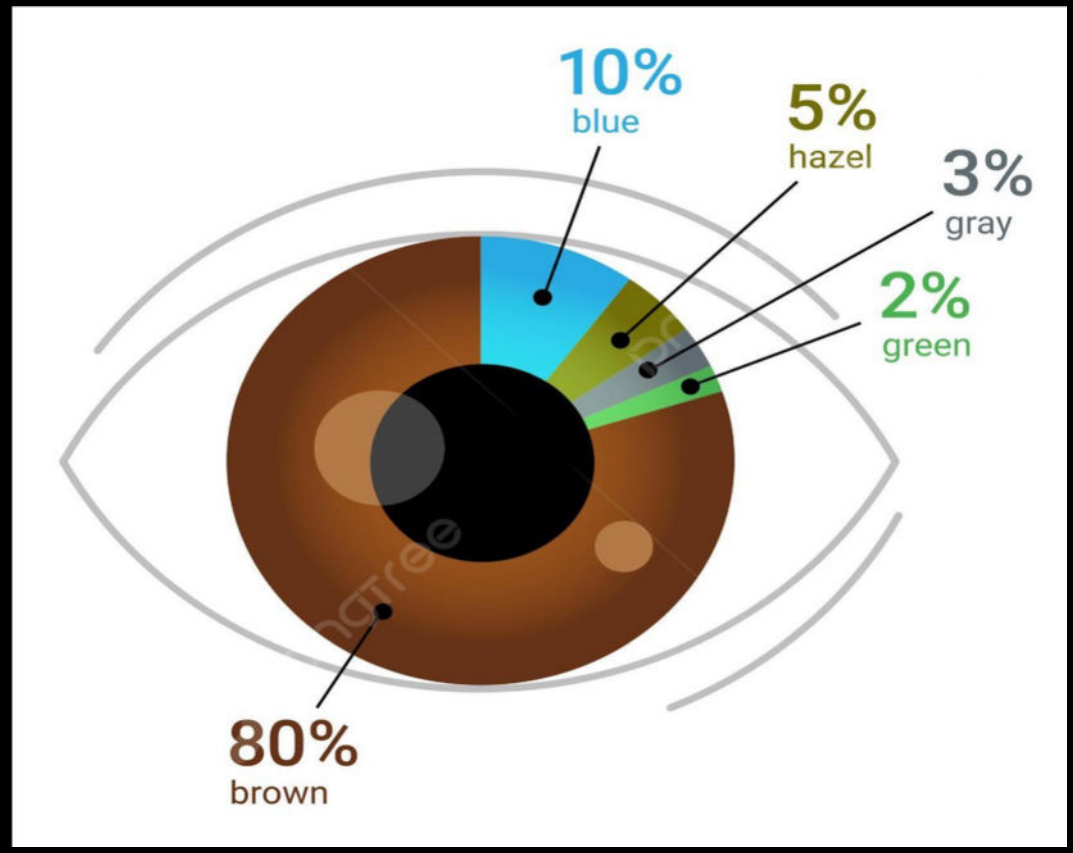
How many B? X

How many b? X

B + b alleles: X + X = X

Frequency of B X / X = X = X %

Frequency of b: X / X = X = X %



Allele frequency of D/d in Jonathan Lambert's family

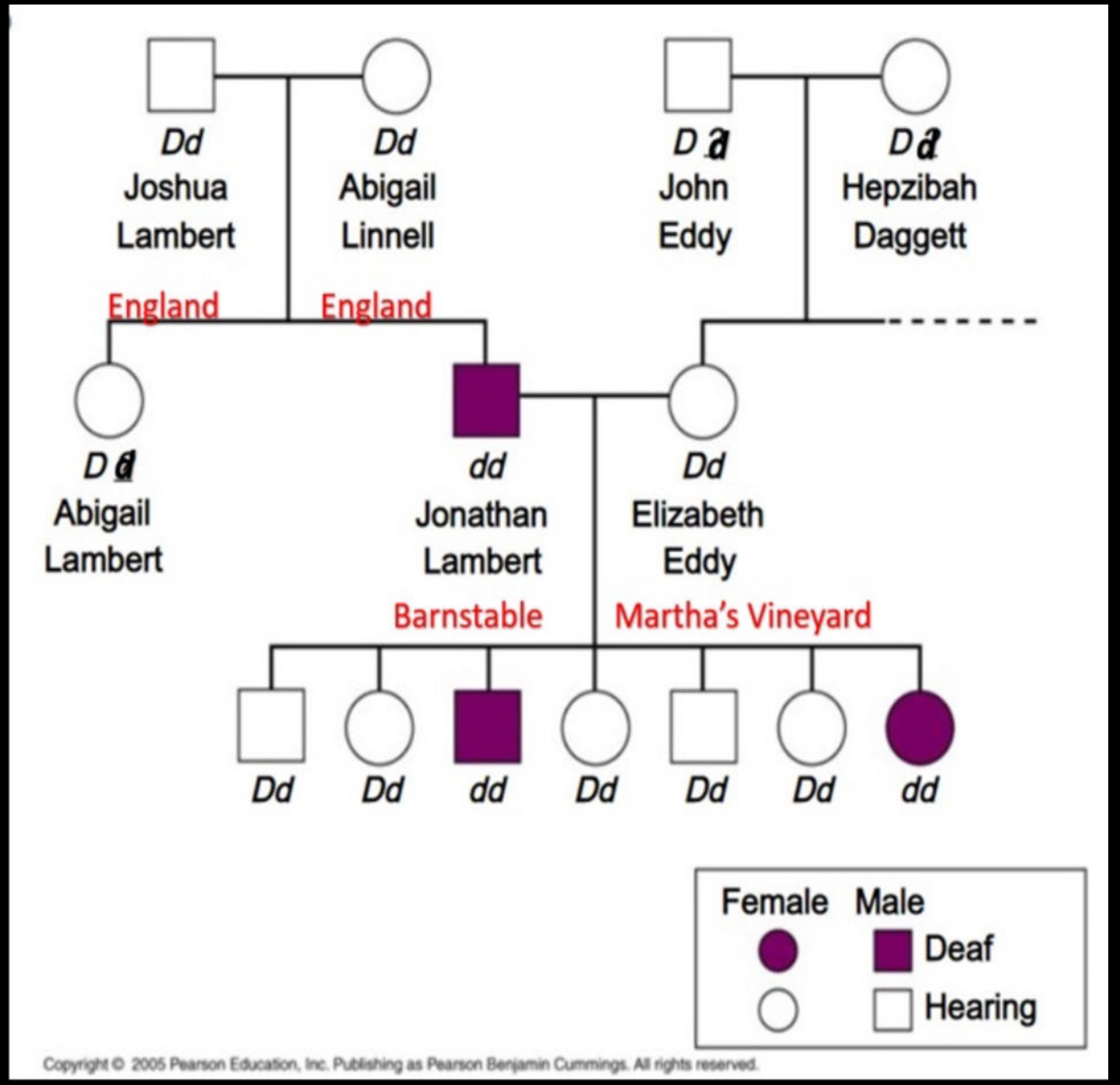
How many D? **11**

How many d? **17**

D + d allele total: **11 + 17 = 28**

Frequency of D: $^{11}/_{28} = 0.39 = 39\%$

Frequency of d: $^{17}/_{28} = 0.61 = 61\%$



What do you do if allele frequencies are unknown?

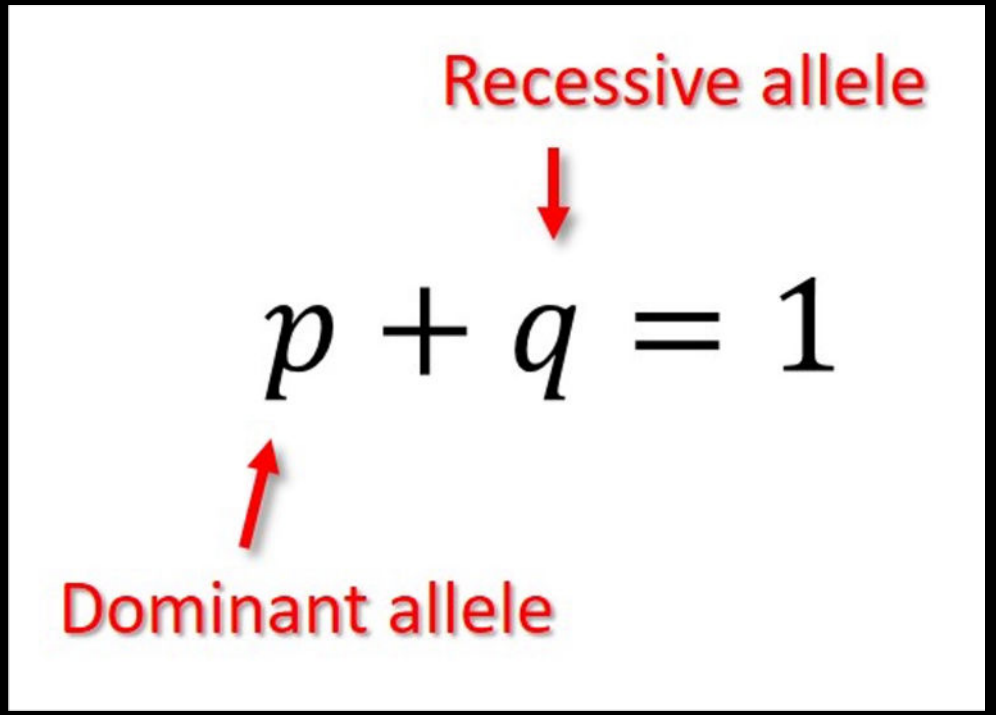
Hardy-Weinberg!

Hardy-Weinberg:
Allele frequencies using phenotype

Dominant + recessive = total alleles

Tip: start with recessive phenotype (q)

Then calculate dominant phenotype (p)



Hardy-Weinberg: Lambert allele frequency

Recessive allele

D

$$p + q = 1$$

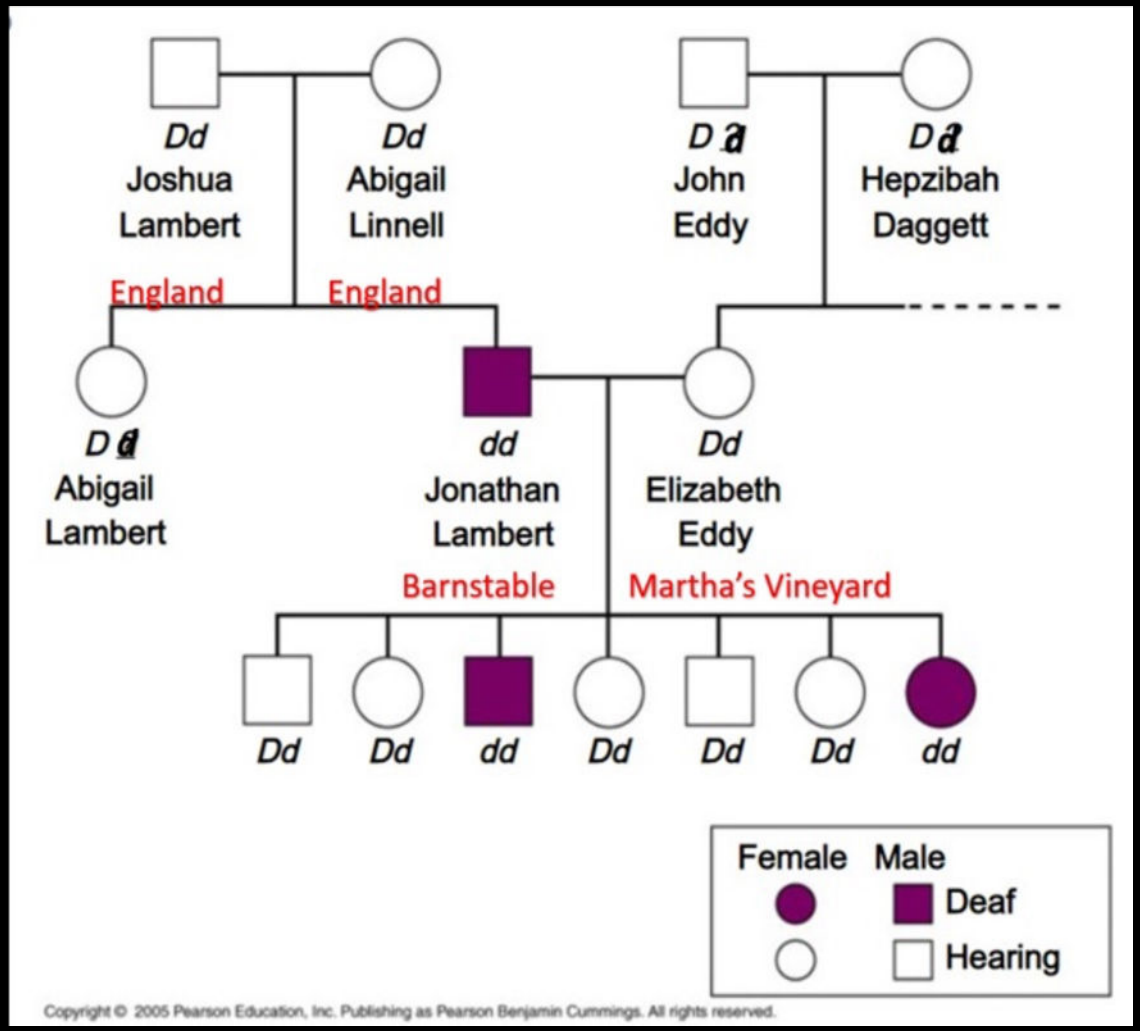
d

Dominant allele

Frequency of p = $^{11}/_{28} = 0.39$

Frequency of q = $^{17}/_{28} = 0.61$

p + q = 0.39 + 0.61 = 1.0



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Hardy-Weinberg: phenotype frequency

Blue colored lobsters are less common than orange. Martha's Vineyard fishermen noticed that 10% of the day's catch was blue.



What is orange color frequency?

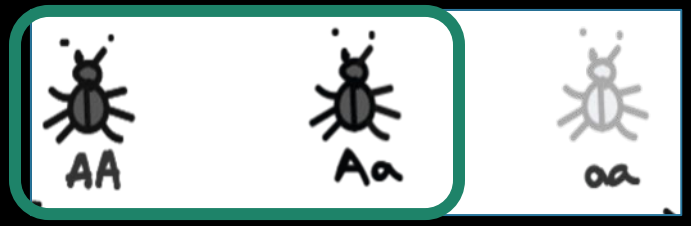
p: **0.90**

q: **0.10**

p + q = **p + 0.10** **= 1.0**

Phenotype to Genotype

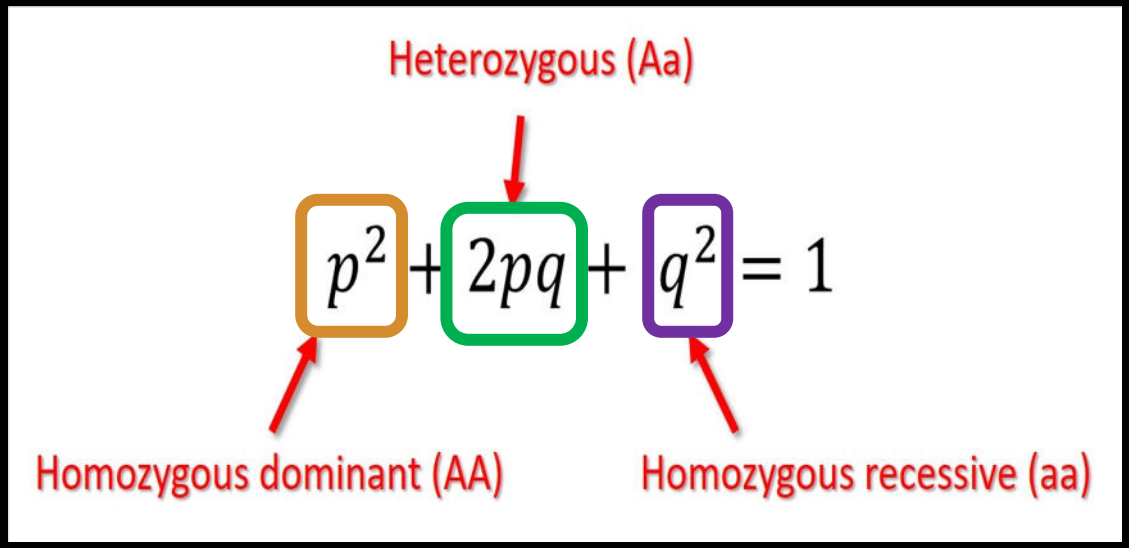
Heterozygotes have the same phenotype as homozygous dominant.



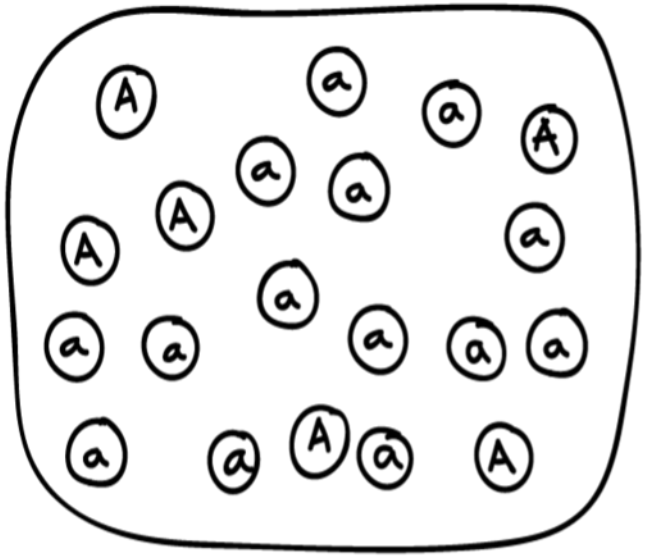
How calculate heterozygote genotypes?

Hardy-Weinberg formula converts *phenotype* counts into *genotype* frequencies

	p	q
p	pp = p^2	pq
q	pq	qq = q^2



BIG POOL o' GAMETES*



Frequency of A = 0.3
Frequency of a = 0.7

* Gametes = eggs & sperm

p = freq. of A
q = freq. of a

$p^2 + 2pq + q^2 = 1$ HARDY-WEINBERG EQUATION

Freq. of AA | Freq. of Aa | Freq. of aa

p = 0.3
q = 0.7



$p^2 = (0.3)^2 = 0.09$

$2pq = 2(0.3)(0.7) = 0.42$

$q^2 = (0.7)^2 = 0.49$

0.09 + 0.42 + 0.49 = 1

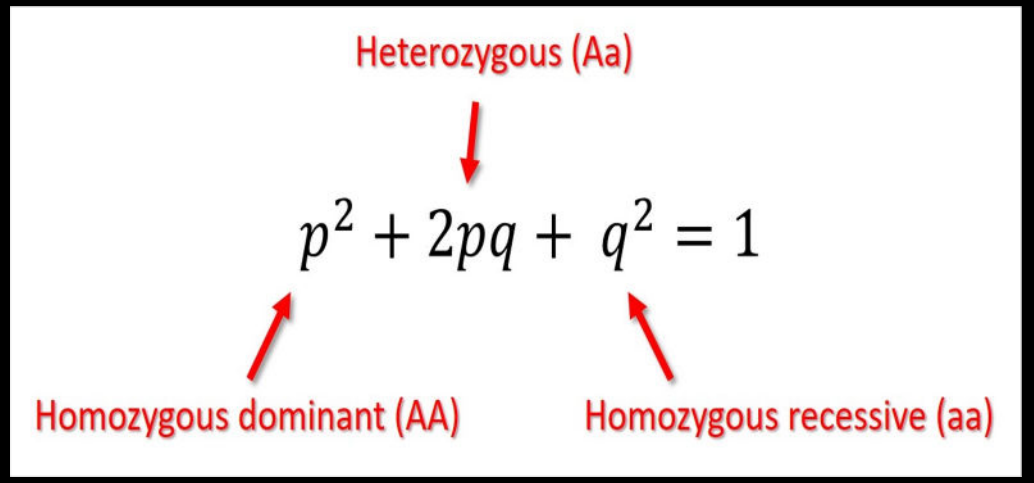
Population genetics and Hardy-Weinberg

Punnett square: *individual* genotypes

Hardy-Weinberg: *population* genotypes

- No change in allele frequency = *no evolution*
- No evolution = population in equilibrium

	p	**q**
p	**pp** = p^2	**pq**
q	**pq**	**qq** = q^2



Punnett, Hardy, and Weinberg

Punnett's dilemma: Why dominant alleles not replace recessive alleles?

Punnett asked his cricket partner Hardy.

Hardy showed allele frequencies stay constant under equilibrium.

Weinberg independently published the same principle that year.



Reginald Punnett
1875-1967
English biologist



Hardy
1877-1947
English mathematician

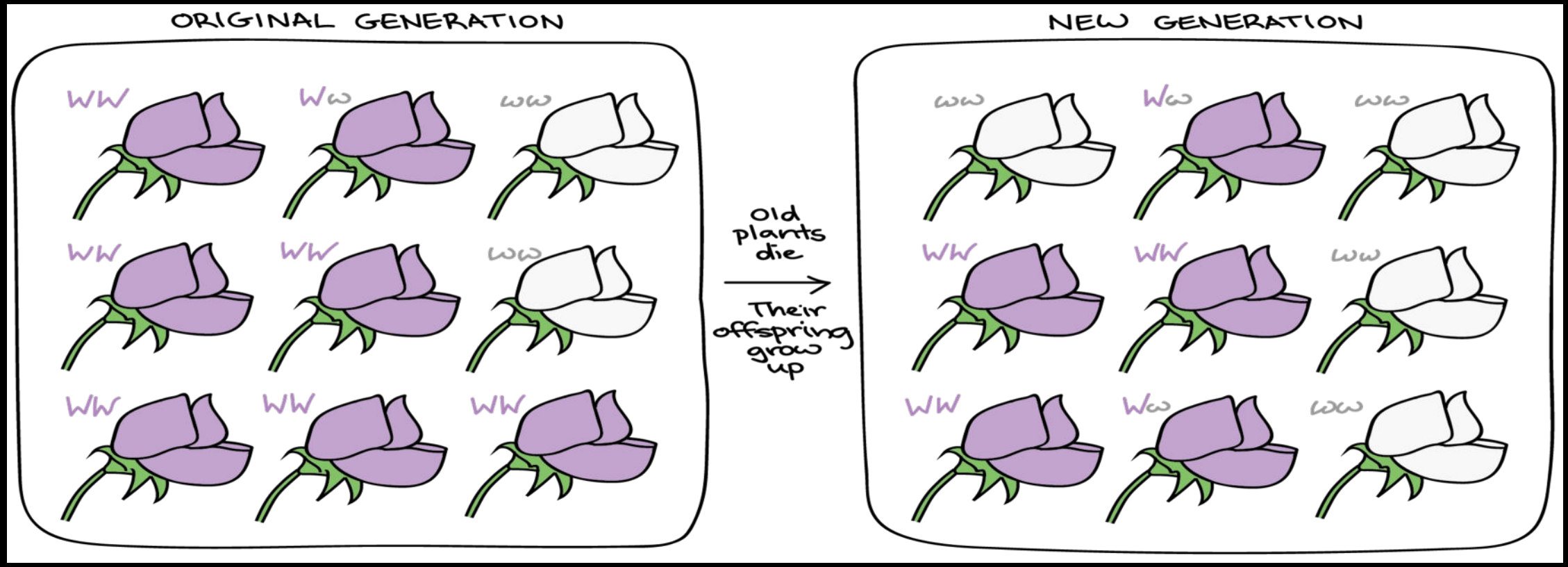


Weinberg
1862 – 1937
German Ob/Gyn

Genotype frequency calculations with Hardy-Weinberg

Freckles, a recessive trait, appears in 36% of Islanders. What is the ***genotype*** frequency?

1) Calculate recessive (q) *allele* frequency:	$q^2 = 0.36$ $\sqrt{q^2} = \sqrt{0.36}$ $q = 0.6$
2) Calculate dominant (p) *allele* frequency:	$p + q = 1$ $p + 0.6 = 1$ $p = 0.4$
3) Calculate (p) *genotype* frequency:	$p = 0.4$ $p^2 = (0.4)^2$ $p^2 = 0.16$
4) Solve $p^2 + 2pq + q^2 = 1$ $0.16 + 2(0.4)(0.6) + 0.36 = 1$ $0.16 + 0.48 + 0.36 = 1$	$p + q = 1$ $0.4 + 0.6 = 1$
16% no freckle gene, 48% heterozygote carriers, 36% with freckles	



Frequency of W = 13

Frequency of w = 5

p frequency (W) = $^{13}/_{18}$ = 0.72

q frequency (w) = $^{5}/_{18}$ = 0.28

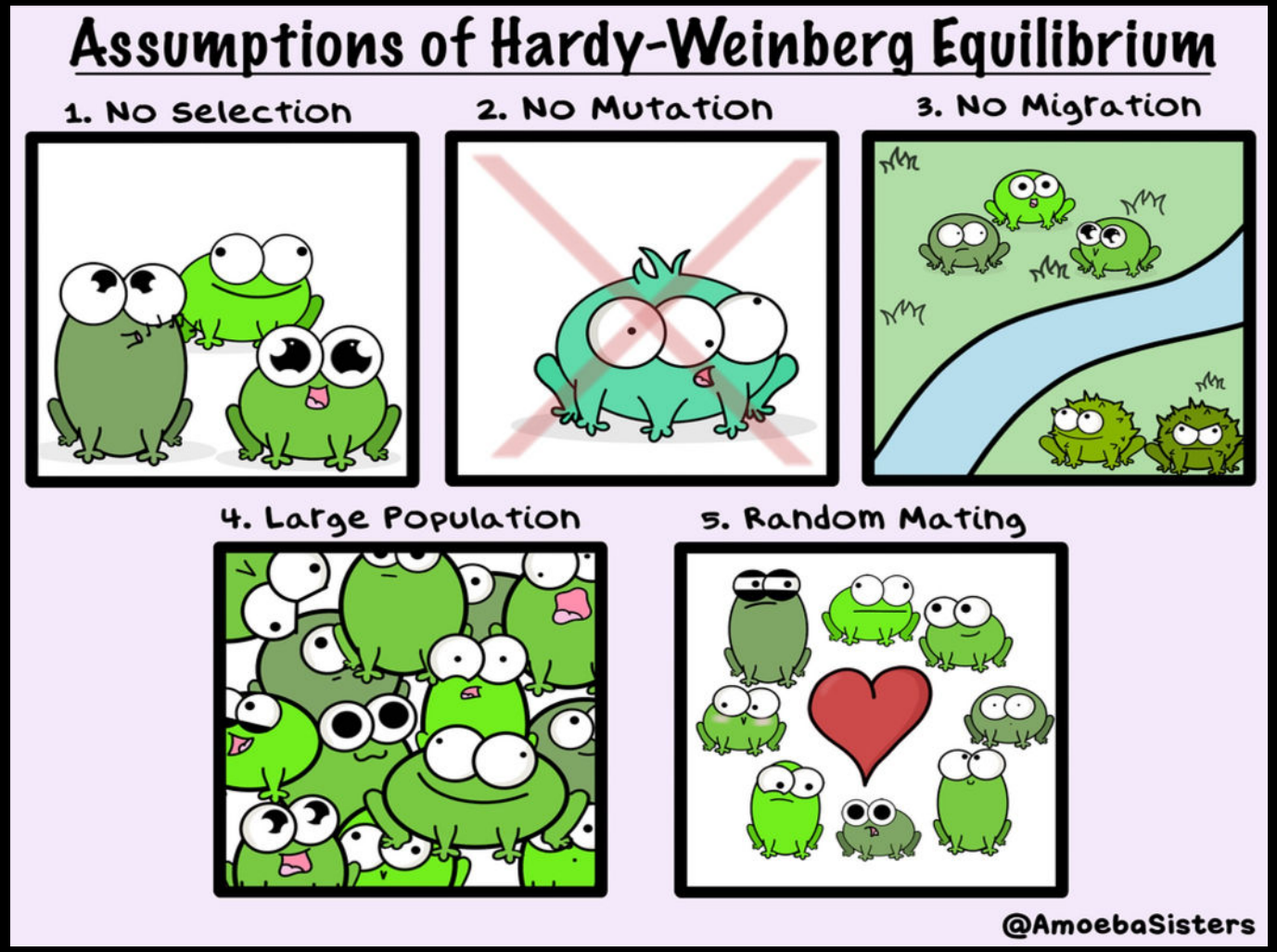
Frequency of W = 8

Frequency of w = 10

p frequency (W) = $^{8}/_{18}$ = 0.44

q frequency (w) = $^{10}/_{18}$ = 0.56

Hardy-Weinberg Equilibrium: Five Required Assumptions



1) No genetic mutations
2) No migration / gene flow
3) Large population size
4) Random mating
5) No natural selection

Violation of any H-W assumption: **Evolution!**

Hardy-Weinberg assumptions:

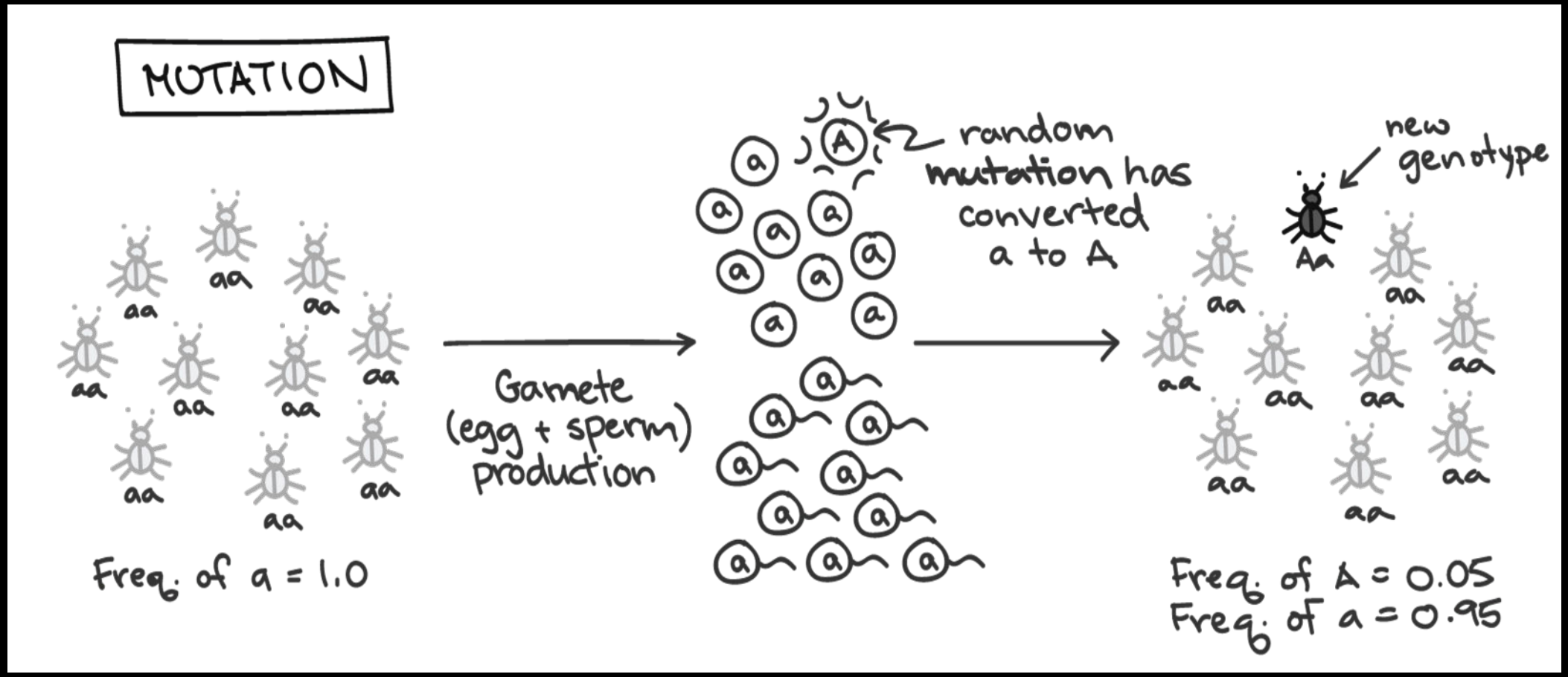
1) No genetic mutations
2) No migration / gene flow
3) Large population size
4) Random mating
5) No natural selection

Mechanisms of evolution:

1) Genetic mutations
2) Migration / gene flow
3) Small population size / genetic drift / founder effect
4) Non-random mating
5) Natural selection

1) Genetic mutation

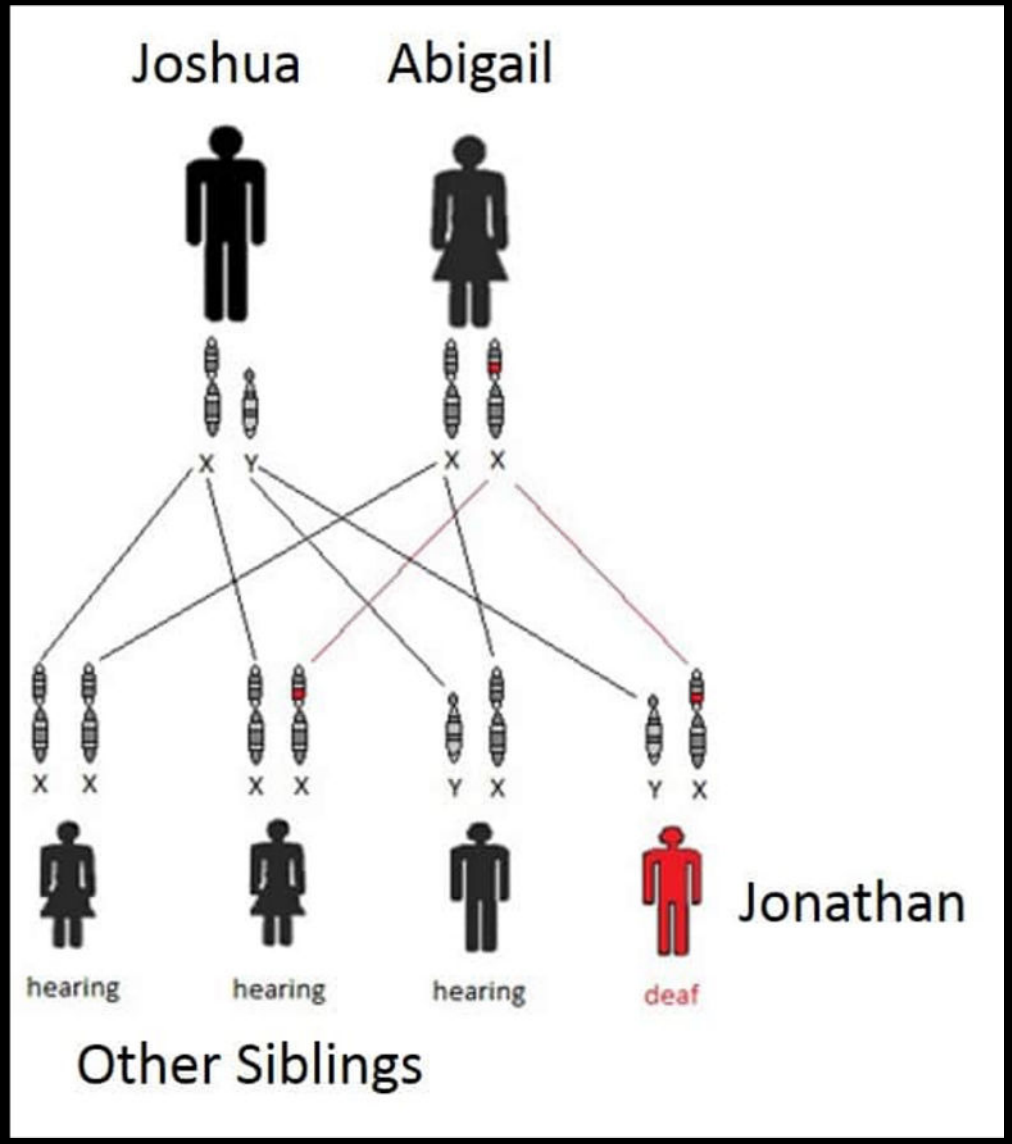
Mutations are an important source of genetic variation in a population.



Genetic mutation – MV application

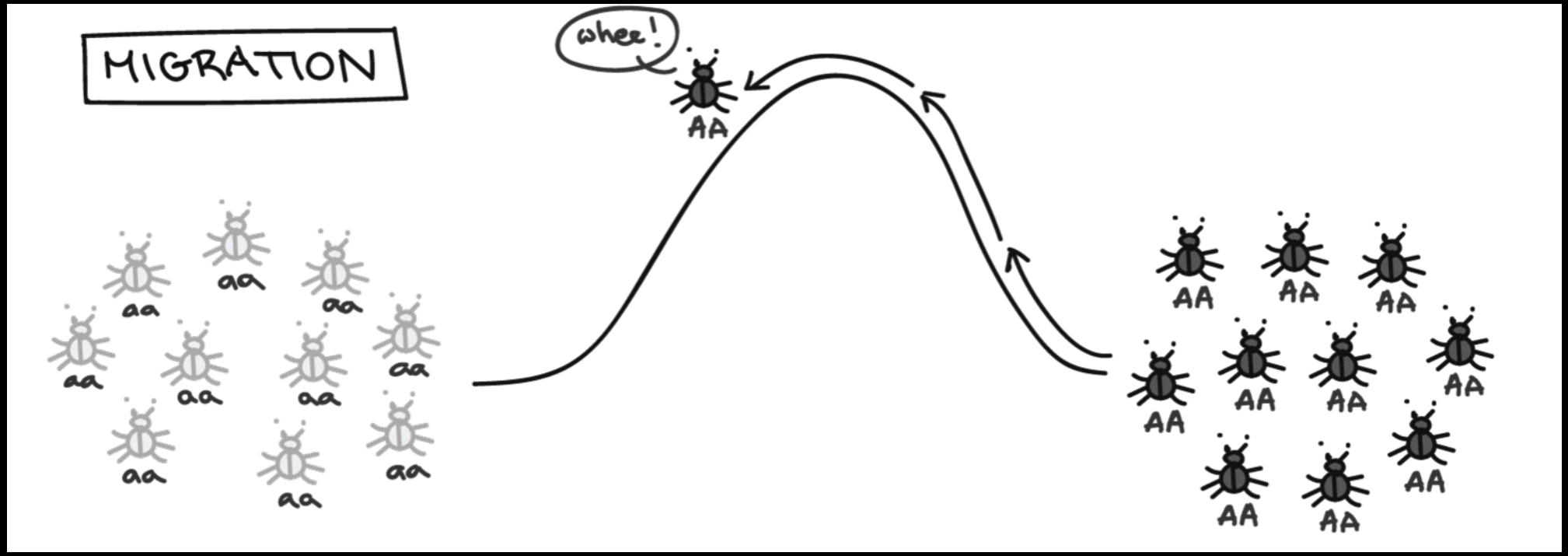
What is the genetic mutation in Martha's Vineyard gene pool?

CJB2 gene (Connexin-26)!



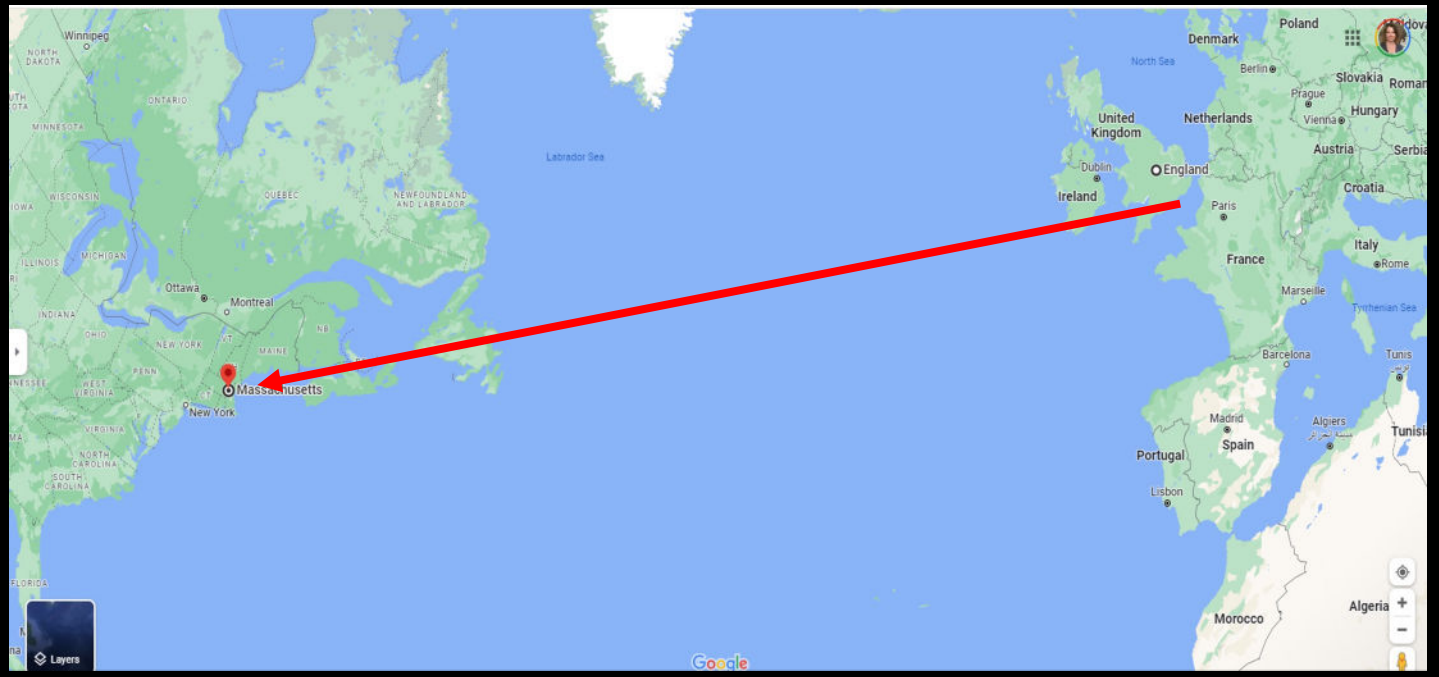
2) Migration / gene flow

- Gene flow = movement of alleles from one population to another.
- Increases genetic variation within a population.



Migration / Gene flow – MV application

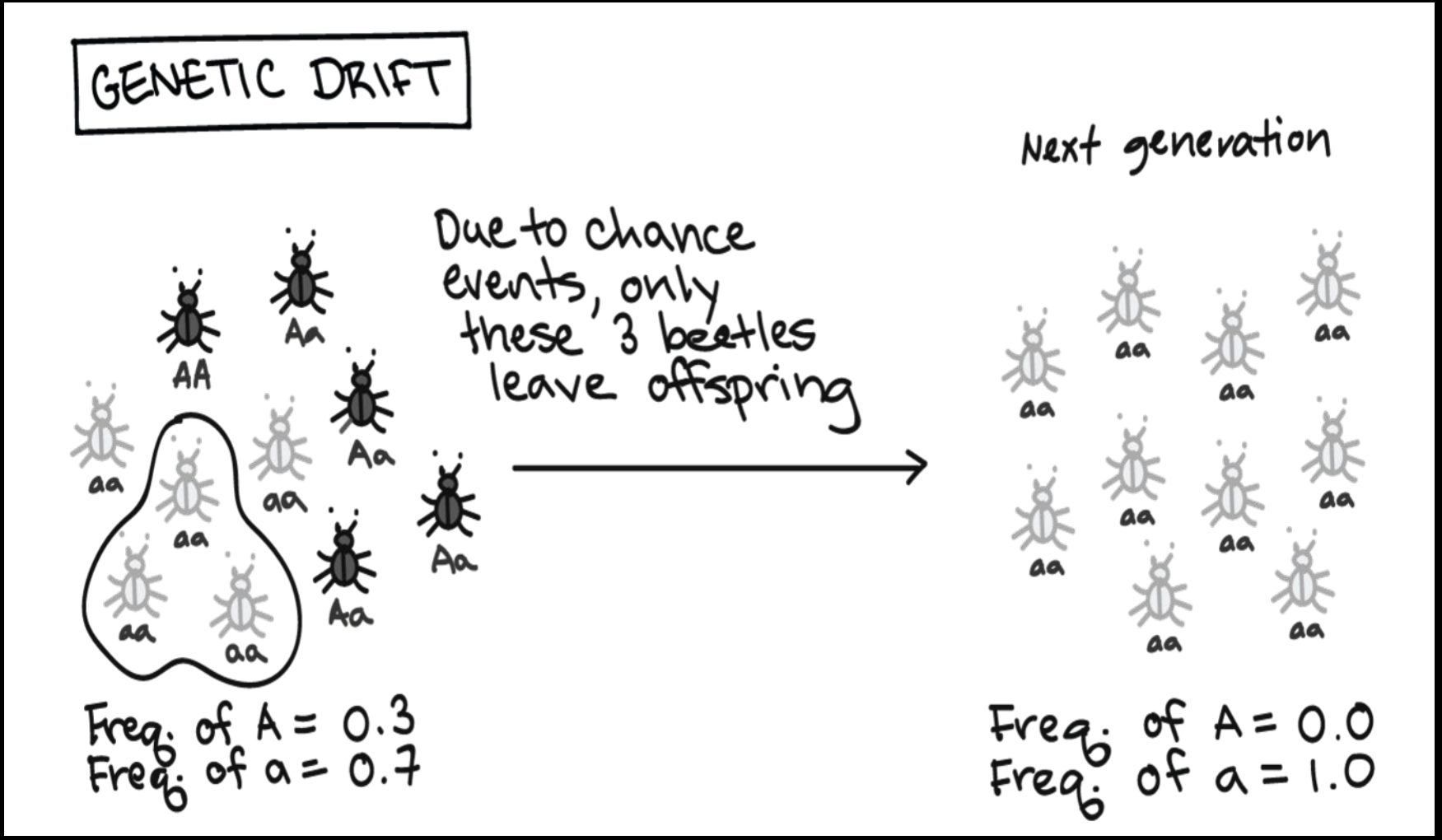
CJB2 gene entered the Martha's Vineyard gene pool from The Weald, England.





3) Small population size / genetic drift

Change in allele frequency caused by a chance event.



Two types:

- Founder effect
- Bottleneck effect

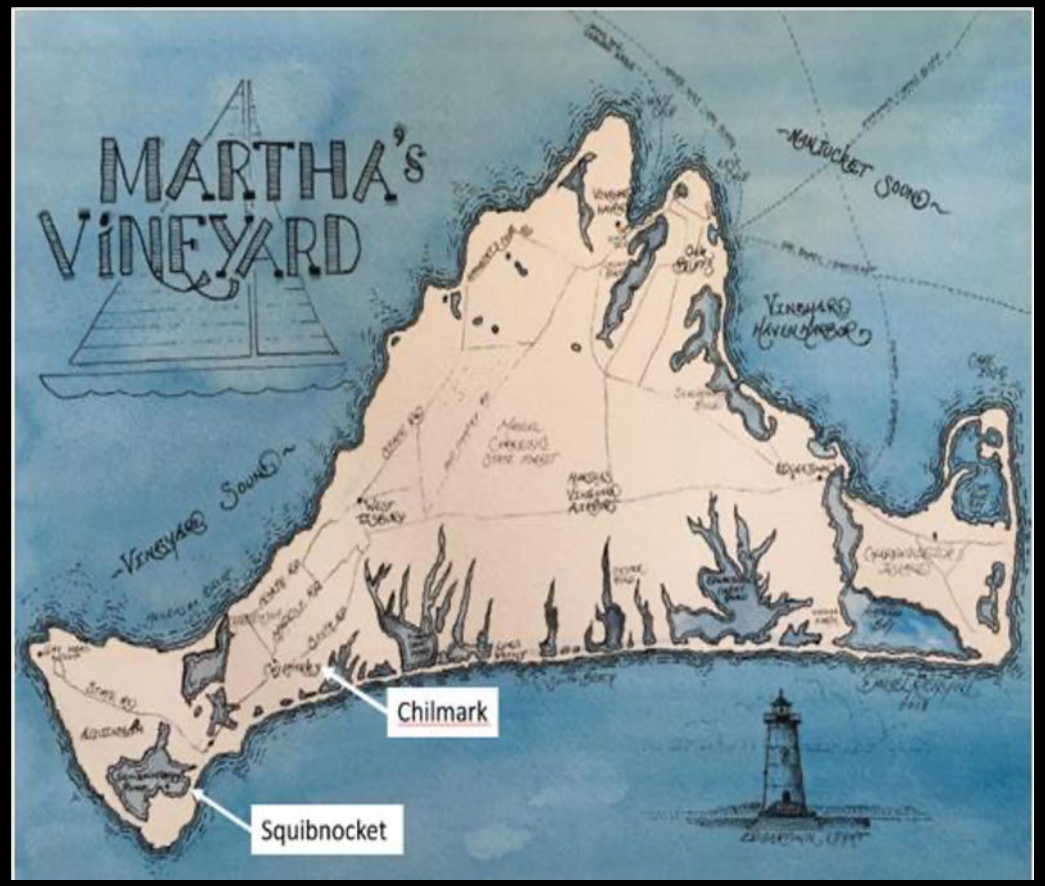
Genetic drift – MV application

Intermarriage in small gene pool for ~200 years.

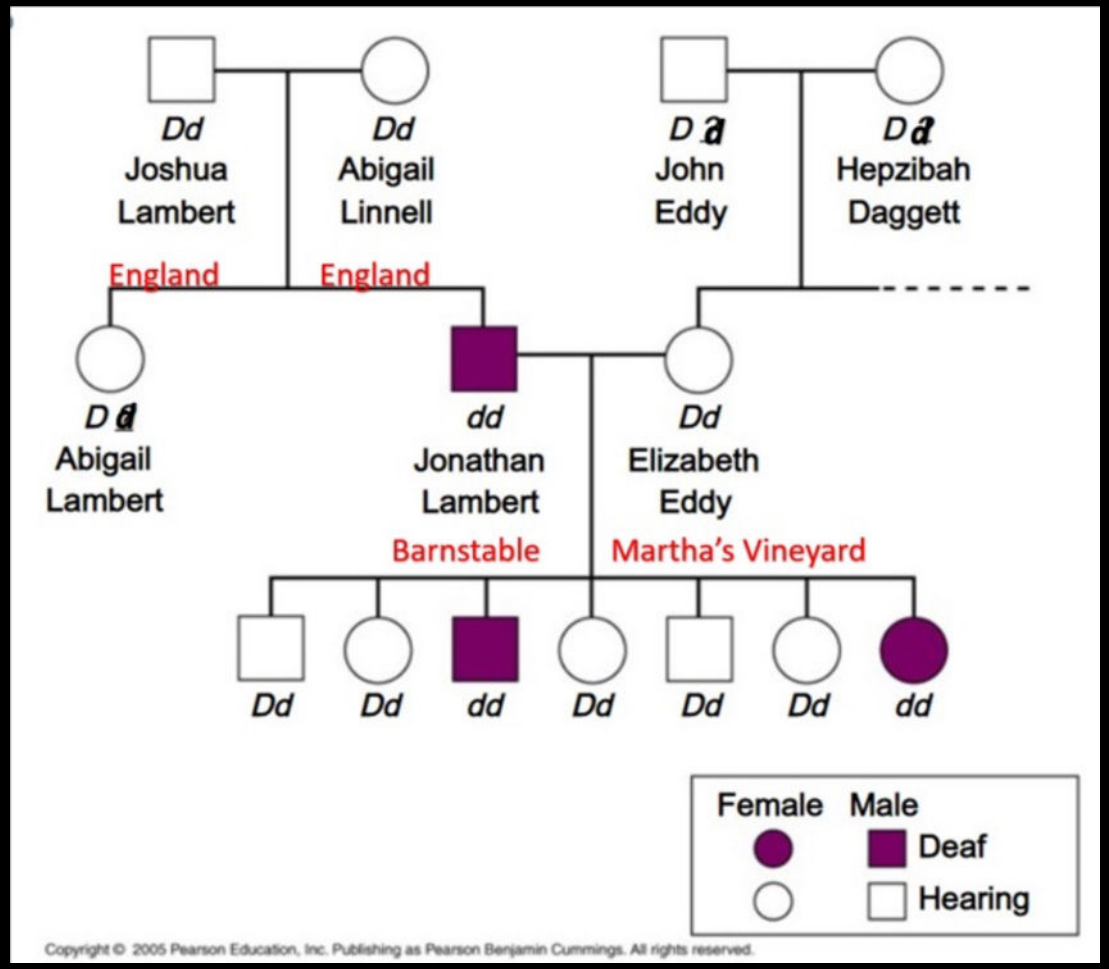
CJB2 drift + inbreeding increased hereditary deafness.

- Island prevalence: 1 in 155
- Chilmark: 1 in 25
- Squibnocket: 1 in 4

- Mainland comparison: 1 in 5700



Genetic drift: Founder effect – MV application



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Lambert, Mayhew, Tilton, West, Skiffe families among founders

- Carried the *CJB2* gene

- Island population ~400

3) Genetic drift: Founder effect in Amish

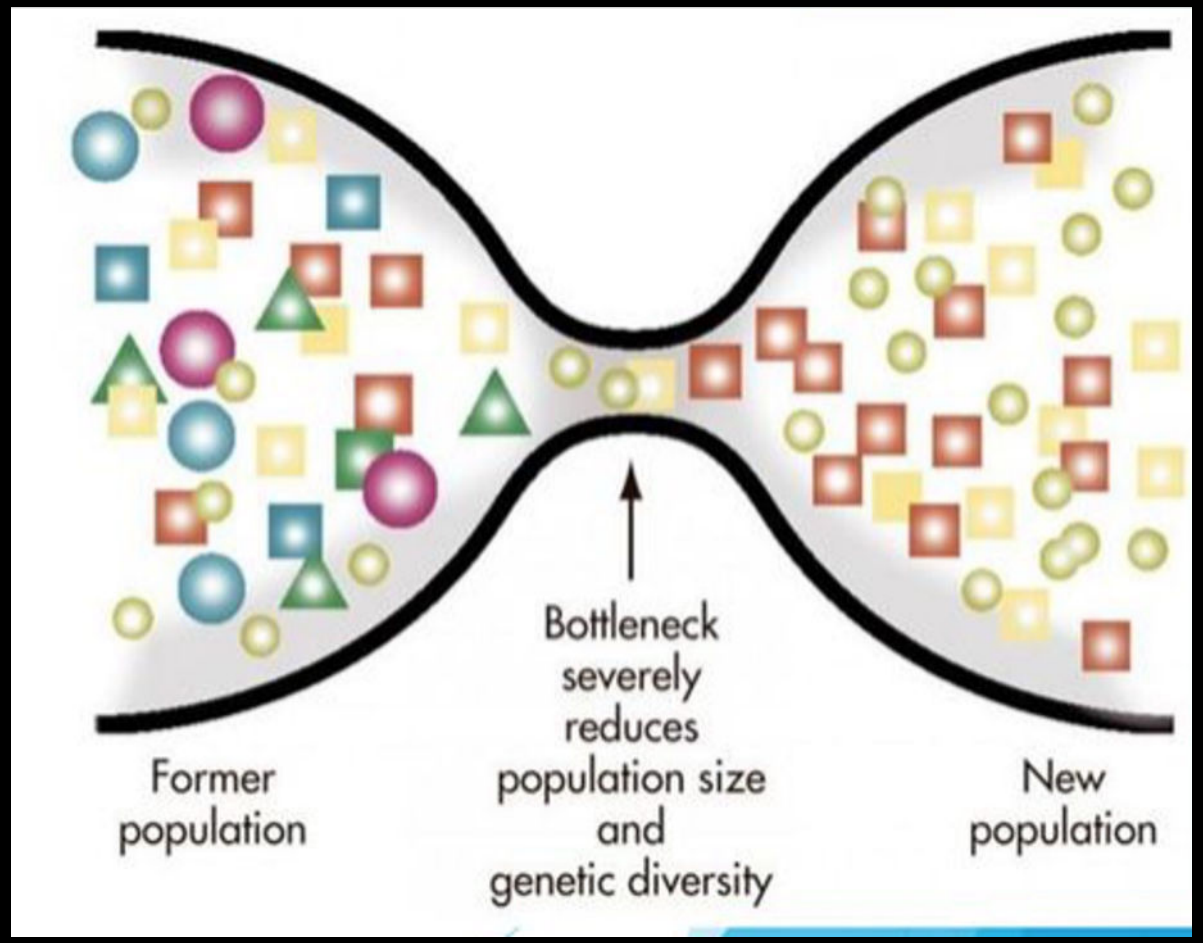
- Amish community in Lancaster County, PA descends from ~30 founders from Switzerland.

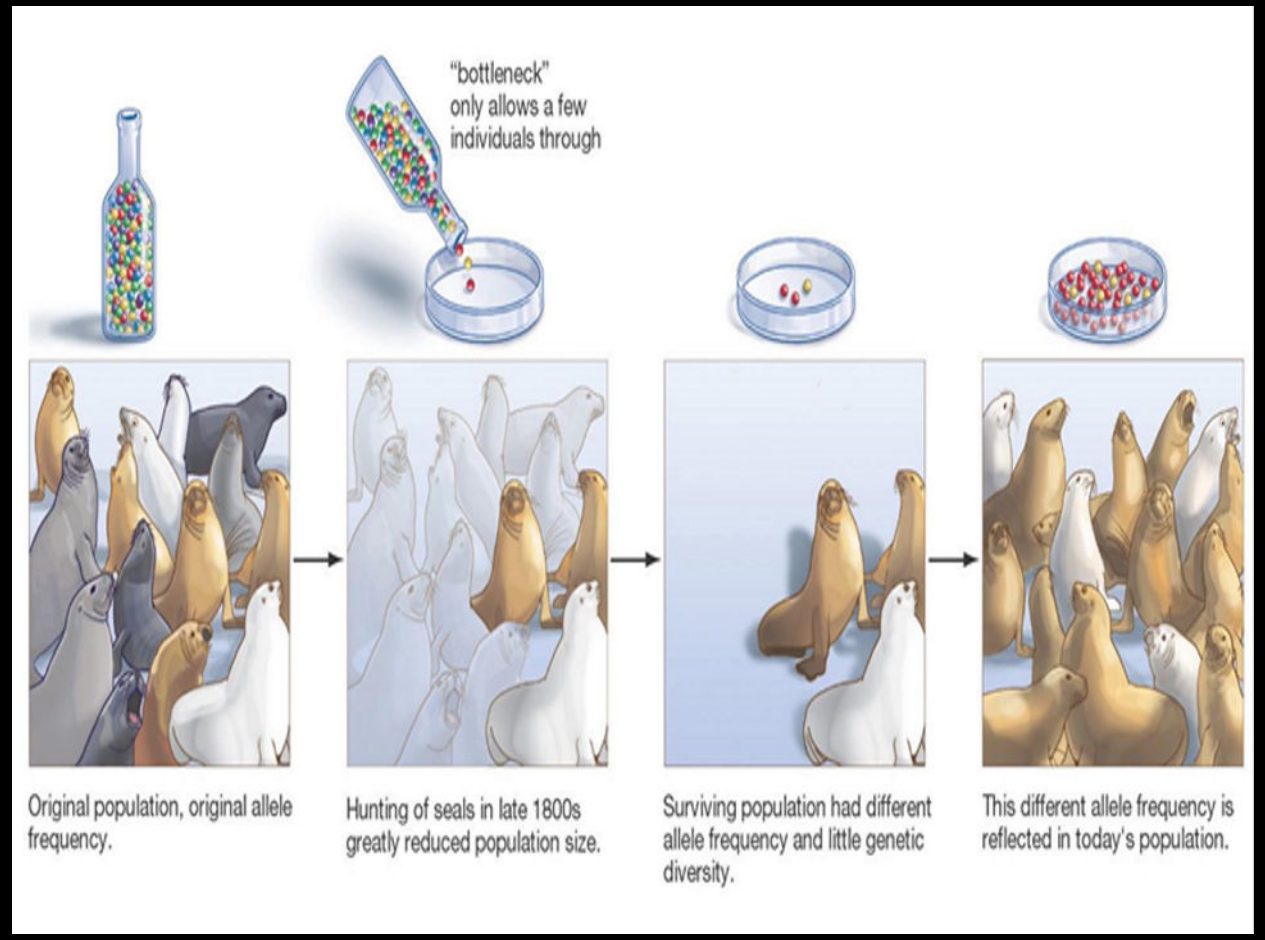
- One founder carried Ellis-van Crevald gene.
  - Polydactly
  - Short height
  - Heart defects

- 1:200 Amish are homozygous recessive.



Genetic drift: Bottleneck effect





Does not apply to MV

4) Non-random mating







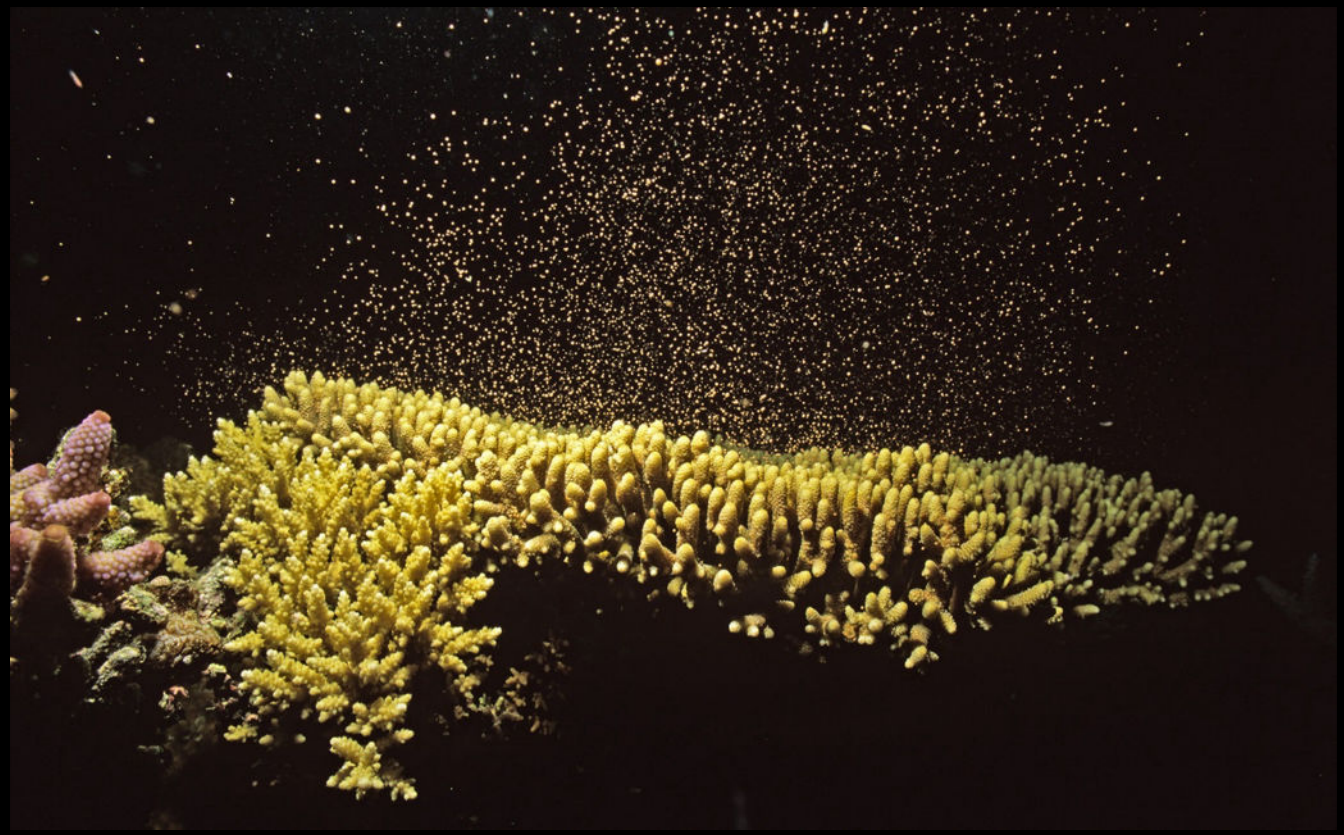


Random mating

Egg and sperm are released externally, left to fertilize by chance.

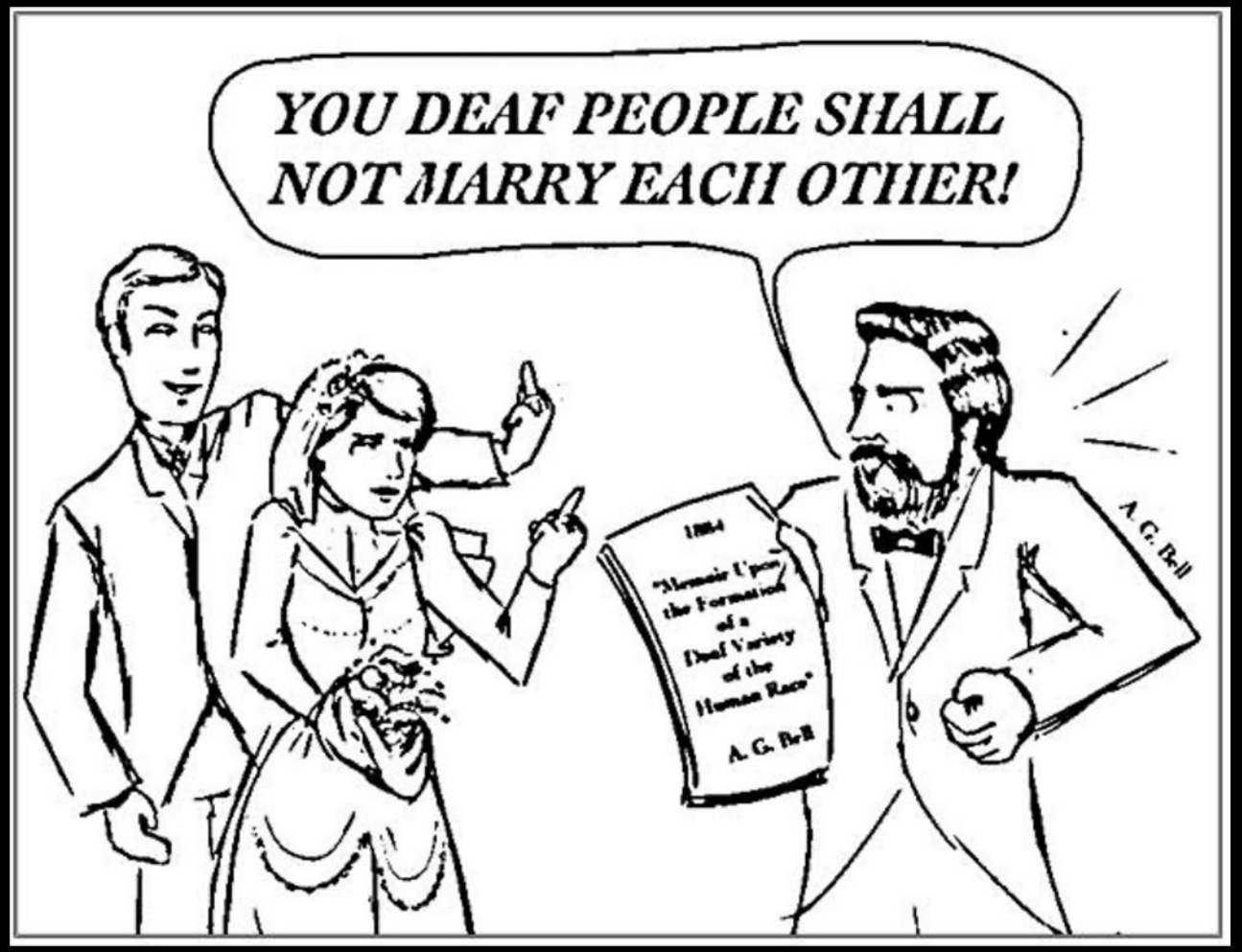
- Some types of pollen
- Coral spawning

NOT COMMON

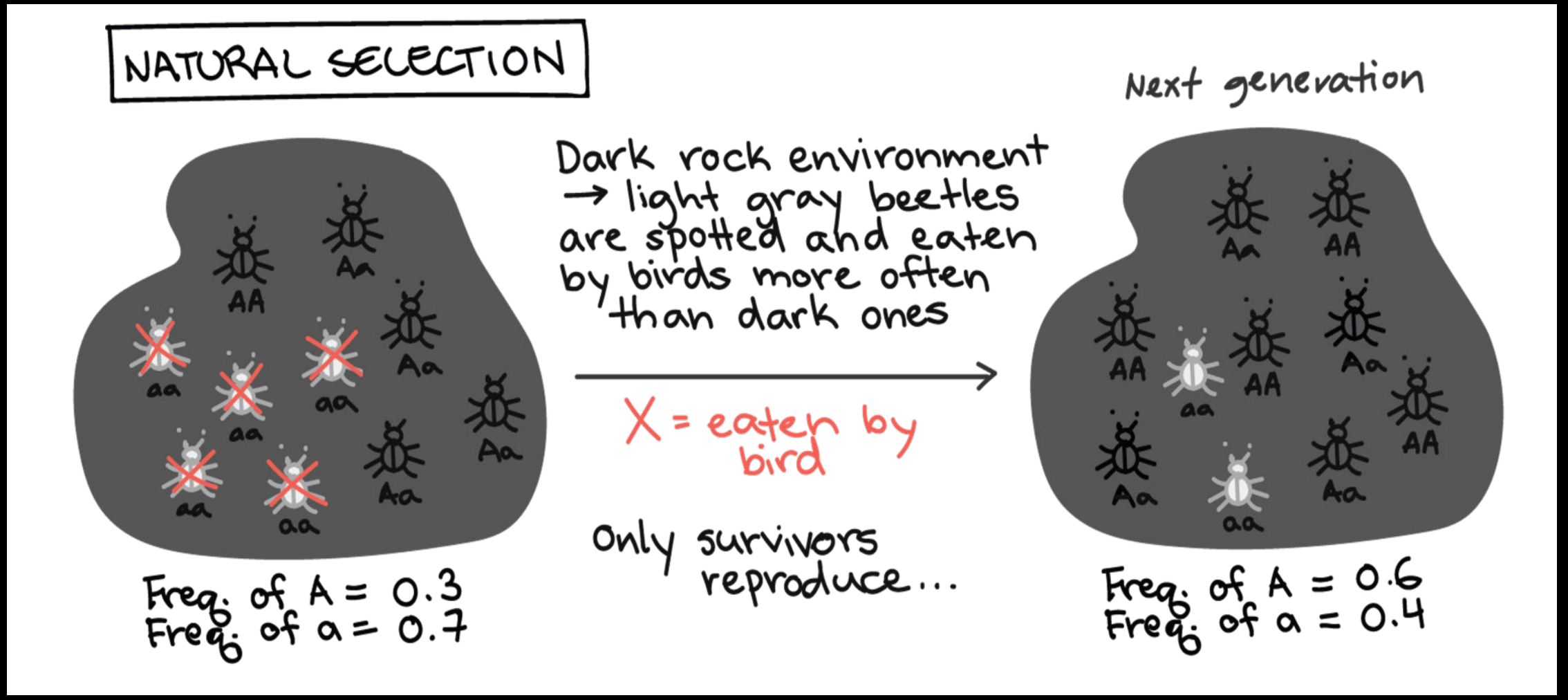


Non-random mating – MV application

Islanders were free to marry whoever they chose, regardless of deafness.

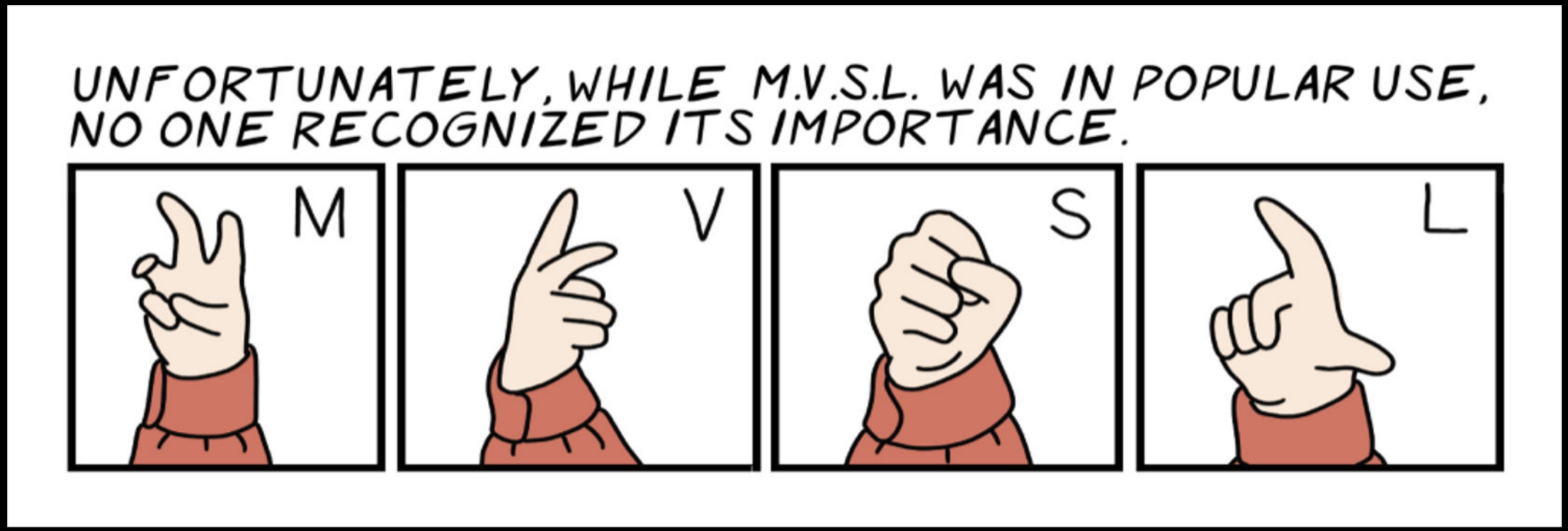


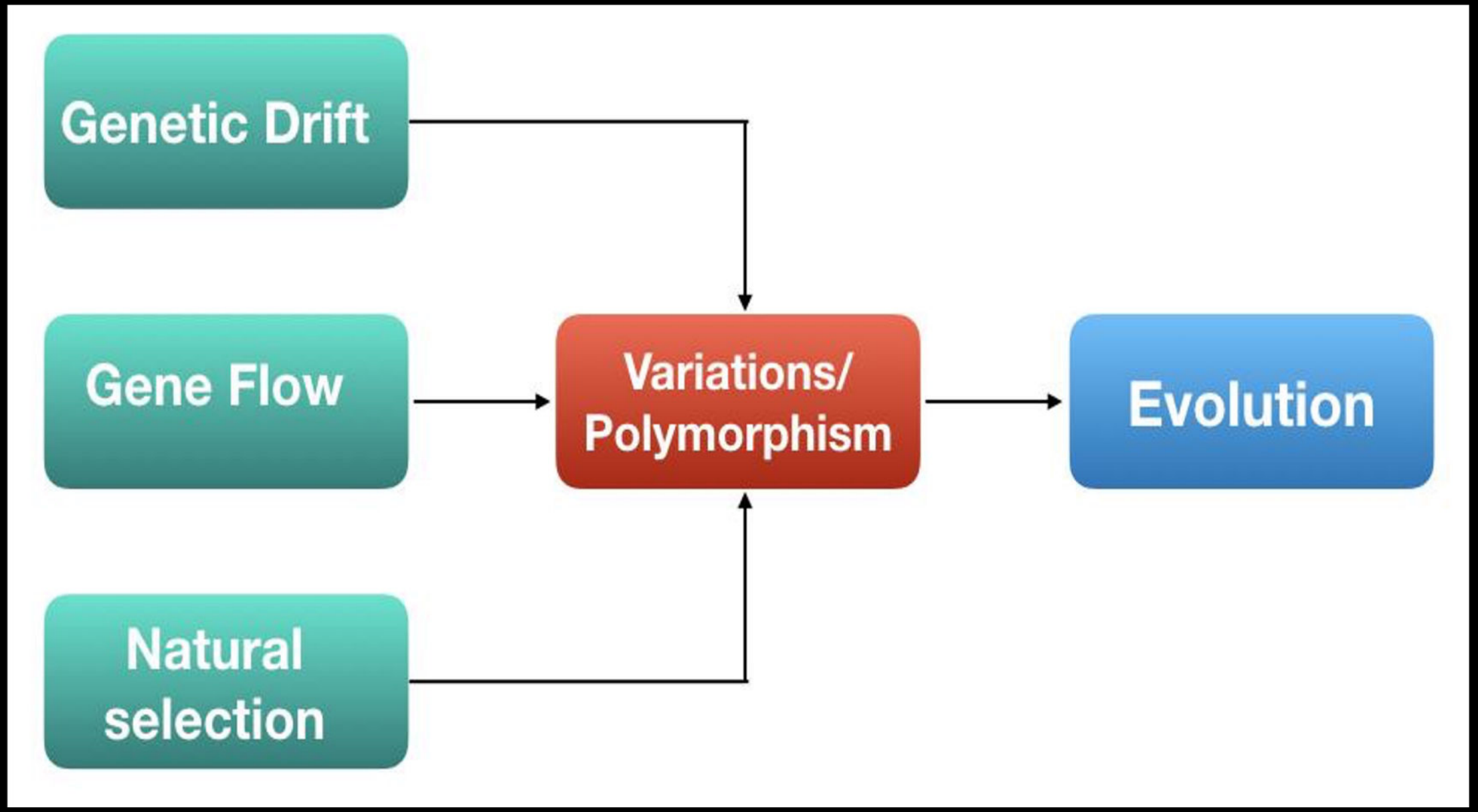
5) Natural Selection



Natural Selection – MV application

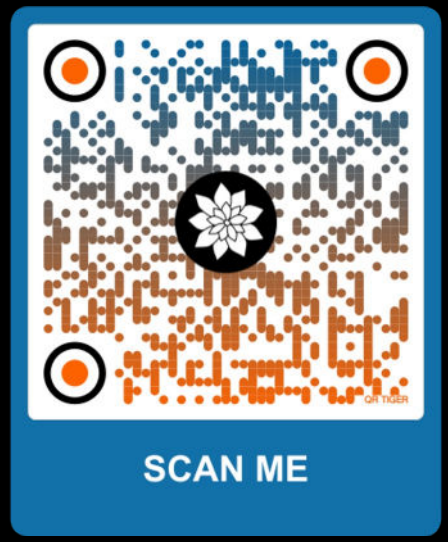
Deafness selected against, not by nature, but by culture.



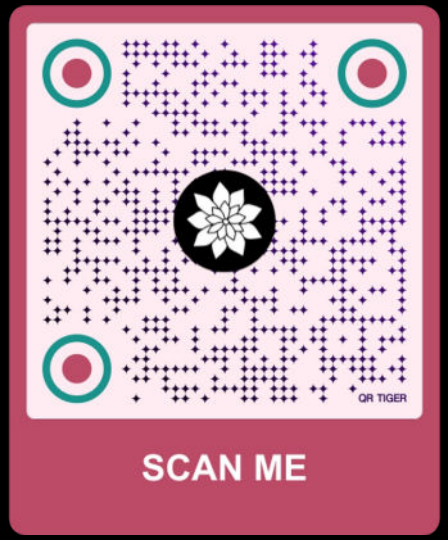

Genetic Drift
Gene Flow
Natural selection
Variations/ Polymorphism
Evolution

Thank you!

Full lesson plan with graphs + interview with Deaf genetic counselor will be available on QUBES BioGraphl in 2027: https://qubeshub.org/community/groups/biographi/materials



Additional biology + math educational resources: https://qubeshub.org/publications/browse



Dr. Elizabeth Ayers, RDMS RVT
Elizabeth.Ayers@rit.edu