

Population Genetics



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Hardy-Weinberg Law

A: natural locks

a: no locks



	A(p)	a(q)
A(p)	AA(p ²)	Aa(pq)
a(q)	Aa(pq)	aa(q ²)

$$p^2 + 2pq + q^2 = 1$$

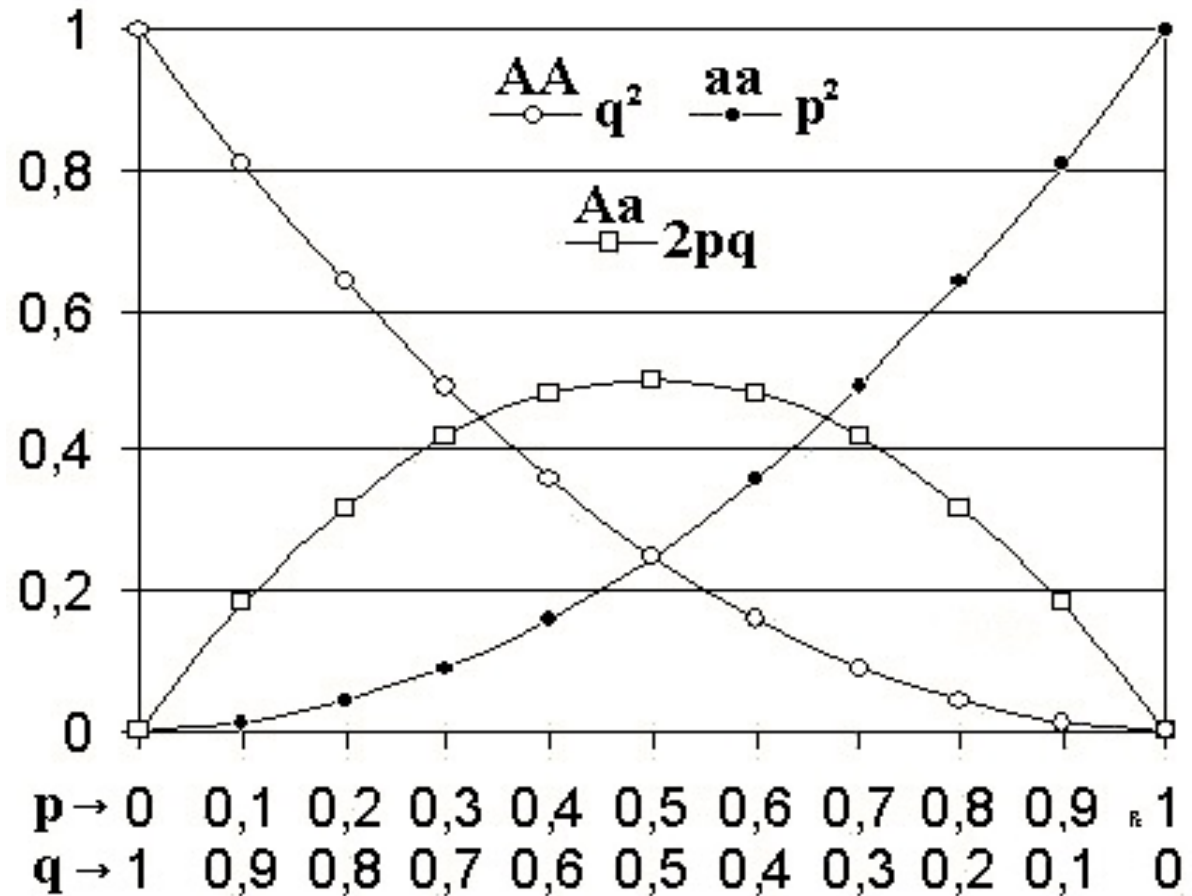
$$p + q = 1$$

Hardy-Weinberg Law

A: natural locks

a: no locks

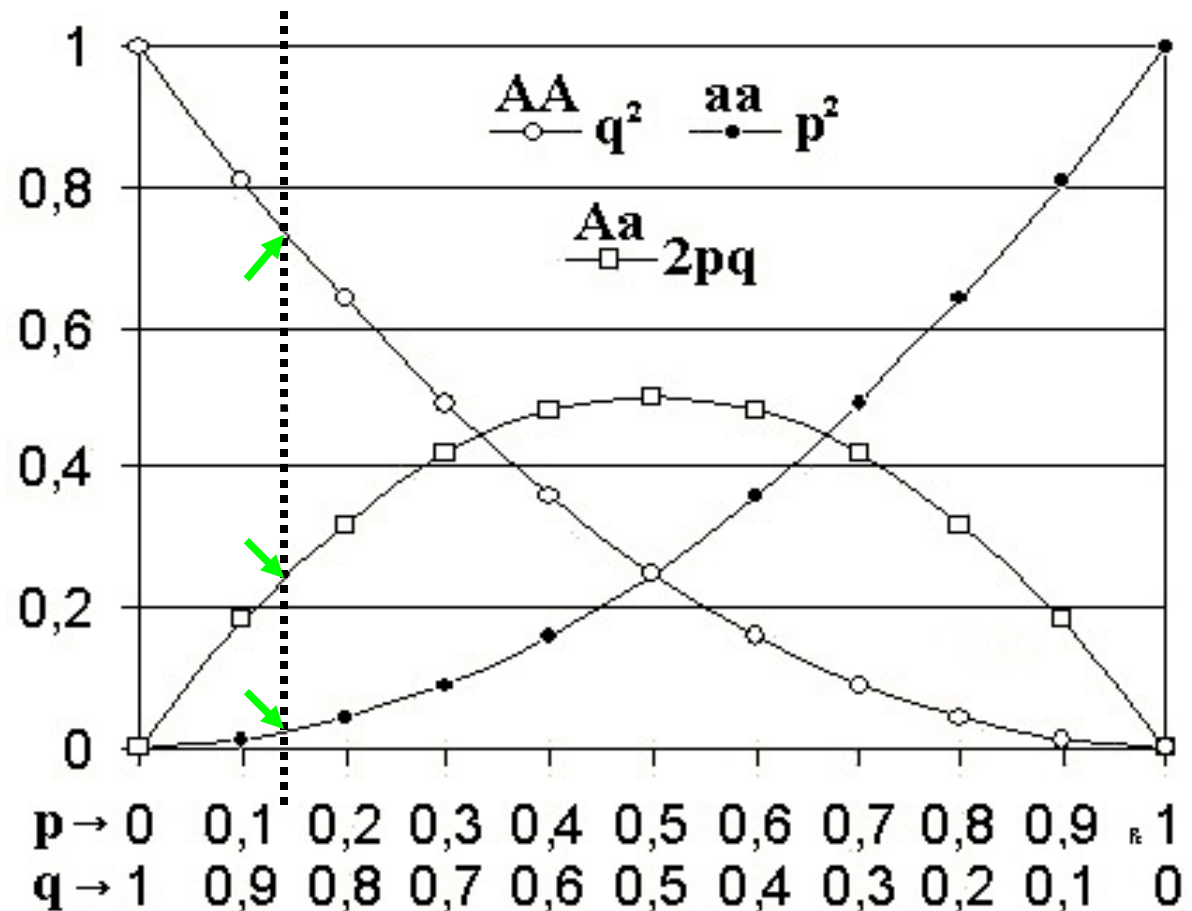
Population Genetics



Hardy-Weinberg Law

fitness (*dostosowanie*) = reproductive success

If fitness "A"
is equal to
fitness "a"
p and q keep
stable over
time



Hardy-Weinberg Law

CYP1B1/432

colorectal cancer cases

CC: (405) 0.31

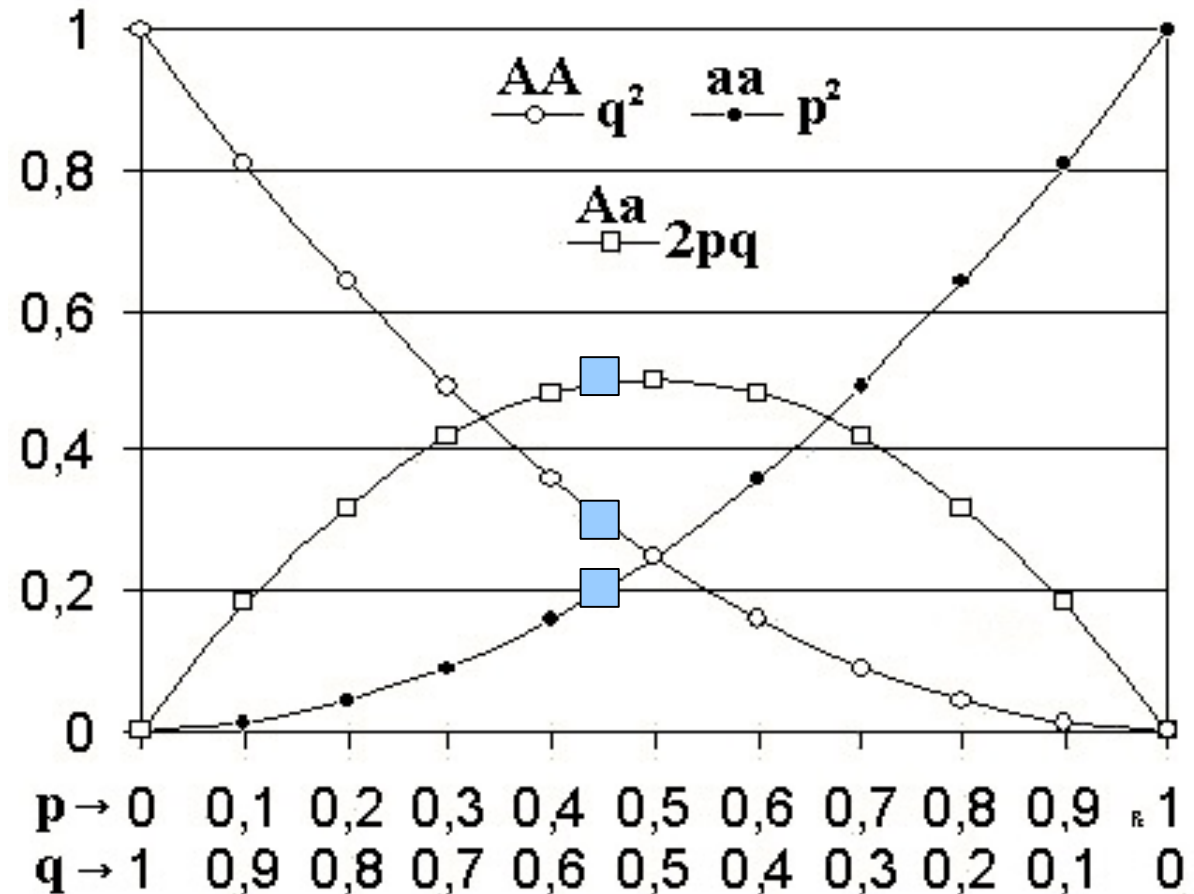
GG: (256) 0.20

CG: (645) 0.49

C: (1455) 0.56

G: (1157) 0.44

Chi-square test ✓



Hardy-Weinberg Law

CYP1B1/119

colorectal cancer cases

CC: (588) 0.45

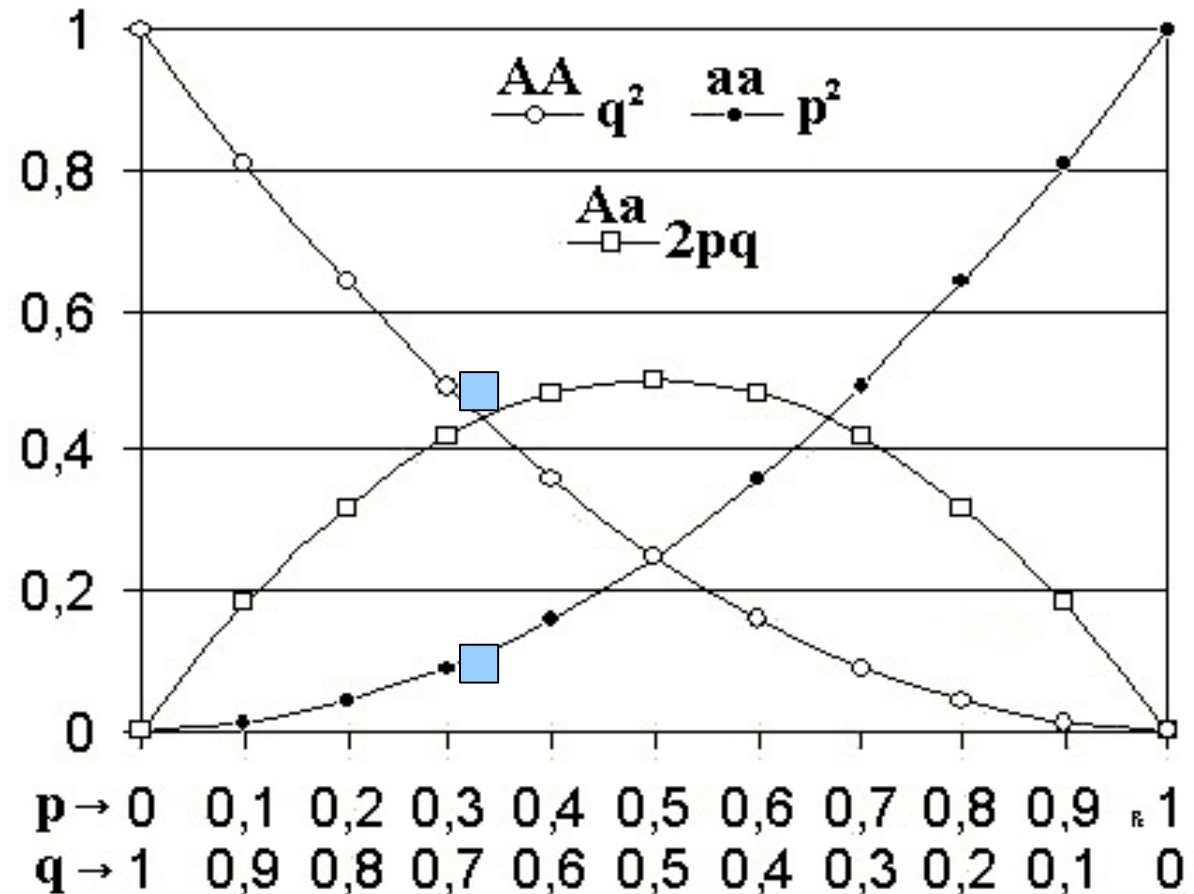
TT: (604) 0.46

CT: (113) 0.09

C: (1780) 0.68

T: (830) 0.32

Chi-square test **X**



Hardy-Weinberg Law

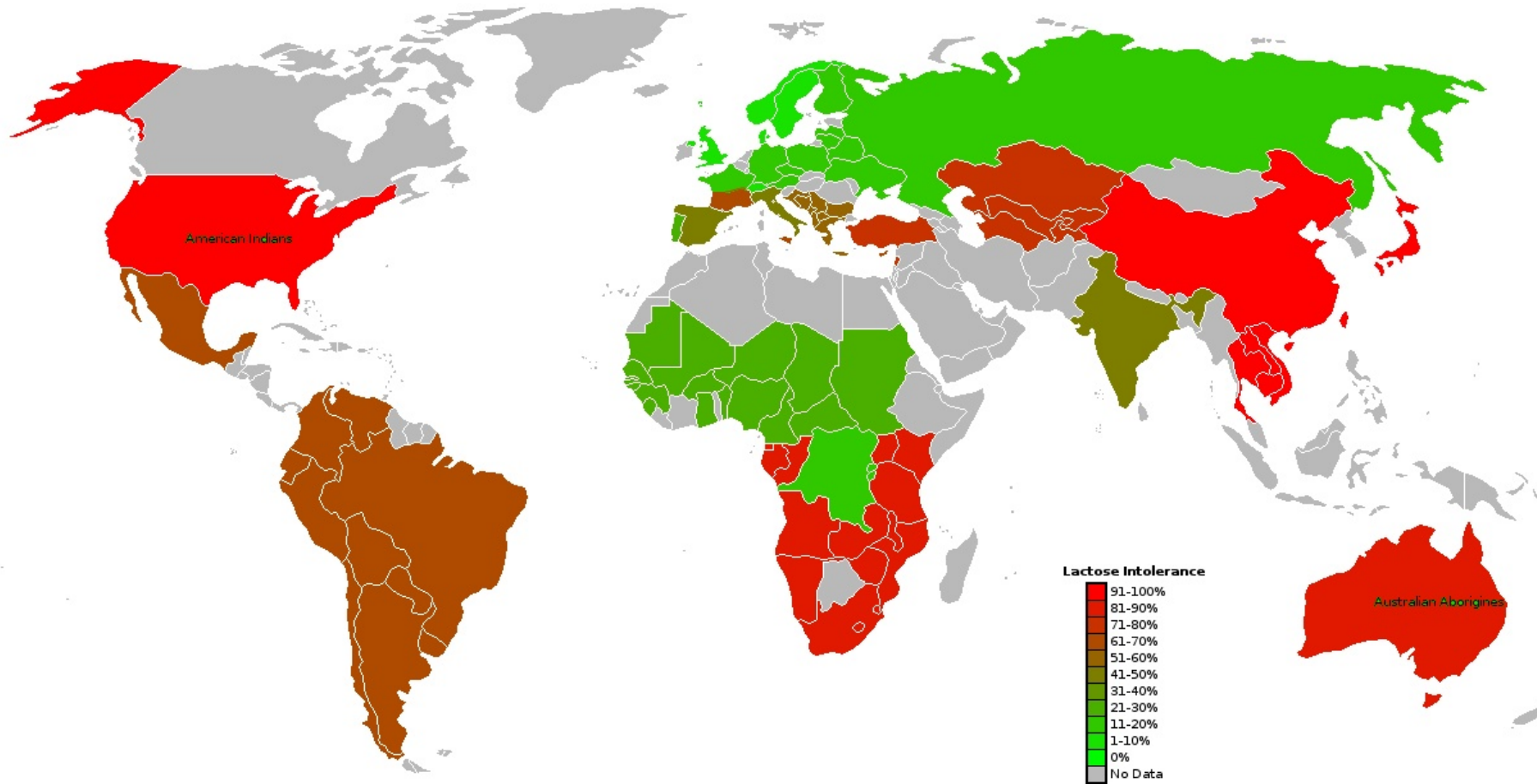
HW Law does not apply?

- Selection (natural or not) is taking place. Including selective mating (inbreeding, sexual selection)

Selection

Tolerance to lactose

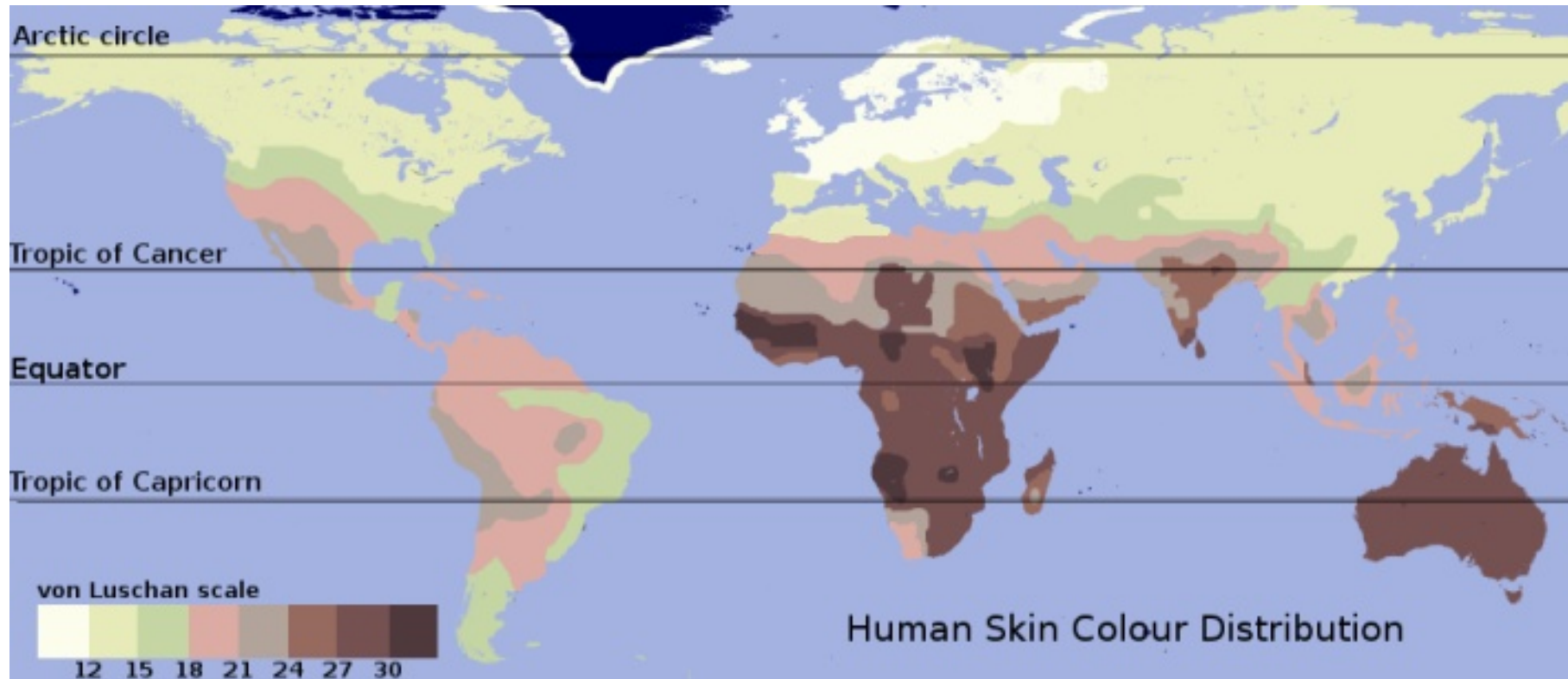
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Selection

Skin color

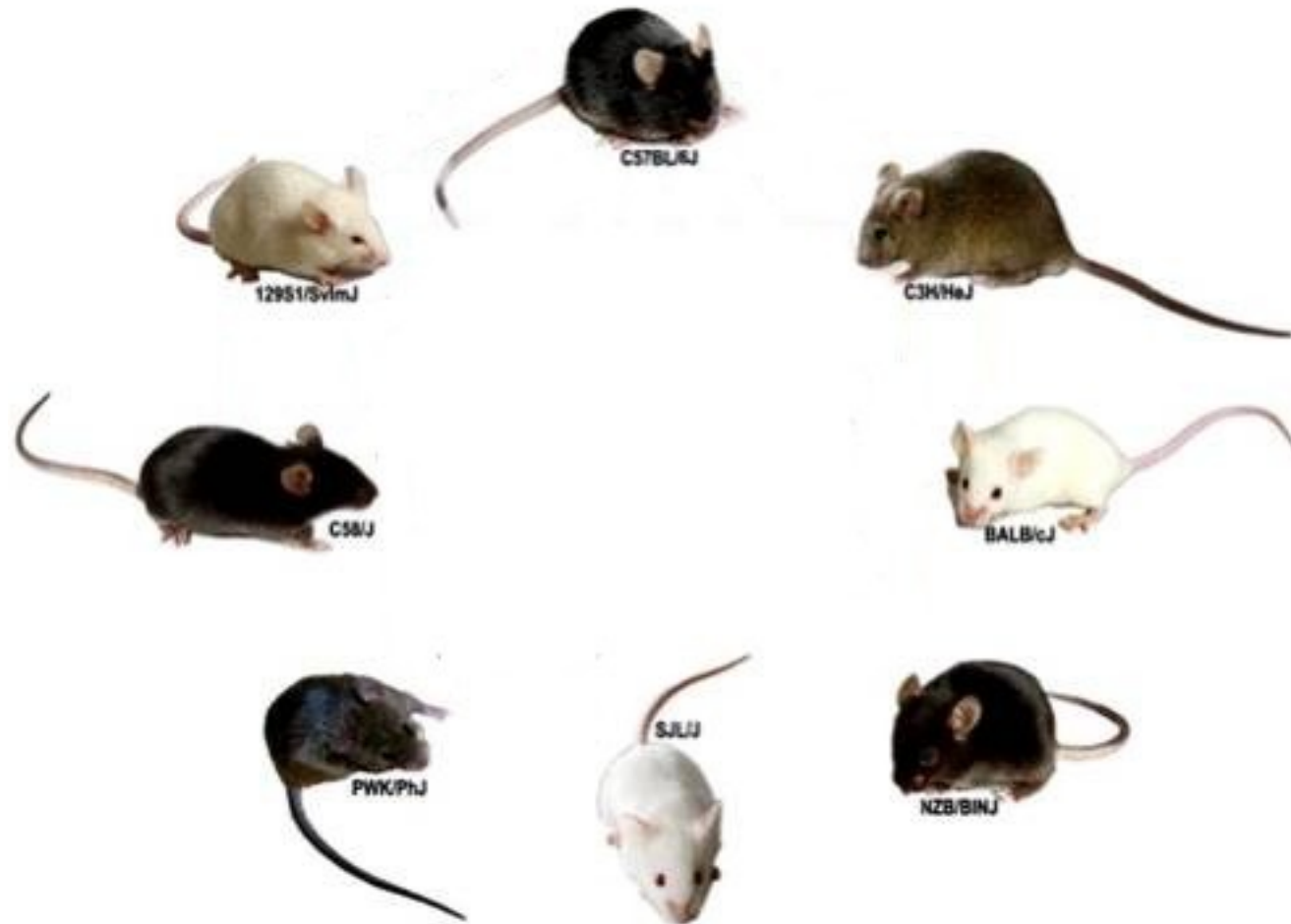
Population Genetics



Inbreeding

Experimental plants / animals: inbred strains

Population Genetics

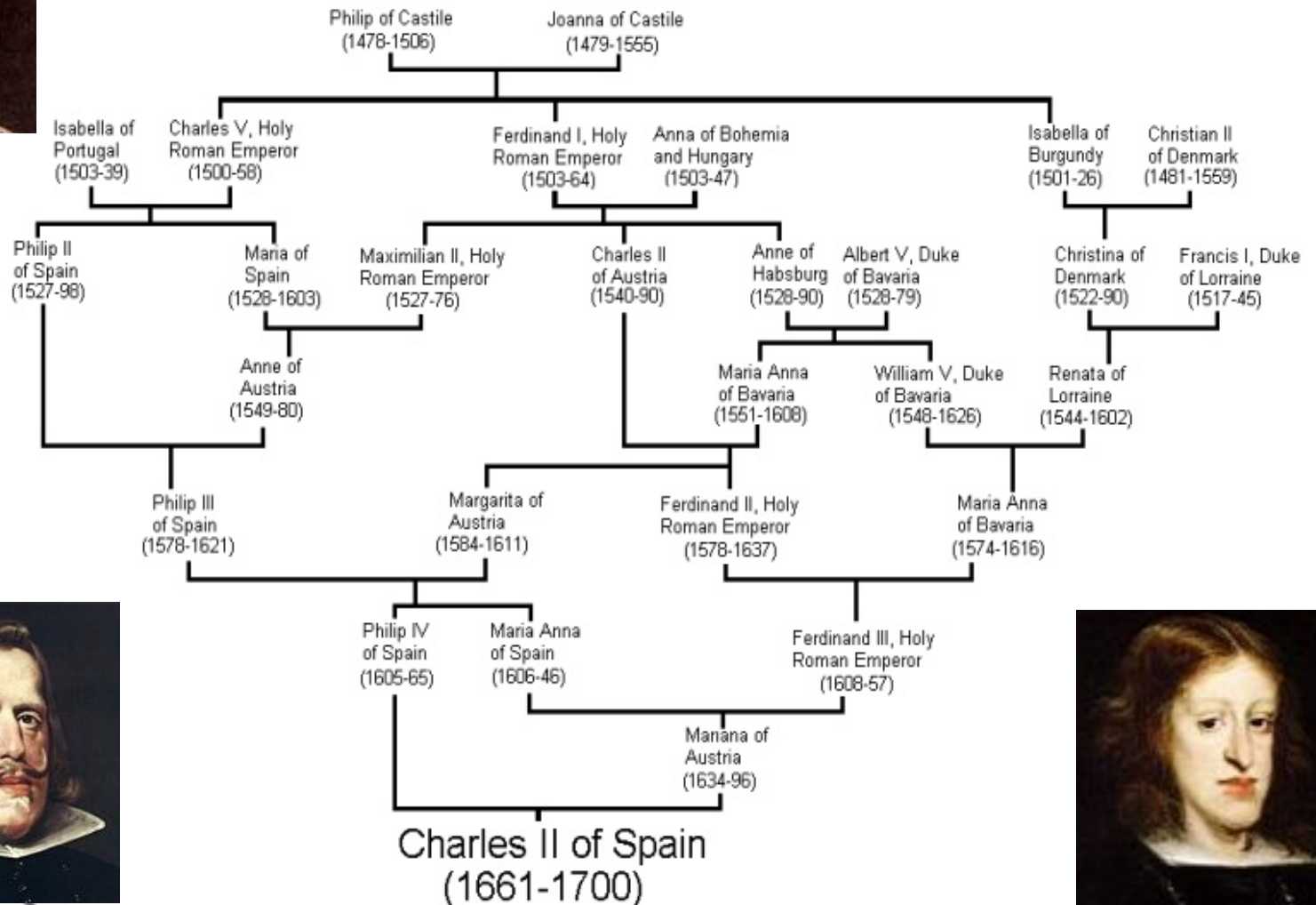


Inbreeding

Population Genetics



The Ancestry of King Charles II of Spain (1661-1700)



Inbreeding

Inbreeding has different degrees (e.g. small, isolated groups)

Effect:

↑ Homogeneous



Homogeneity

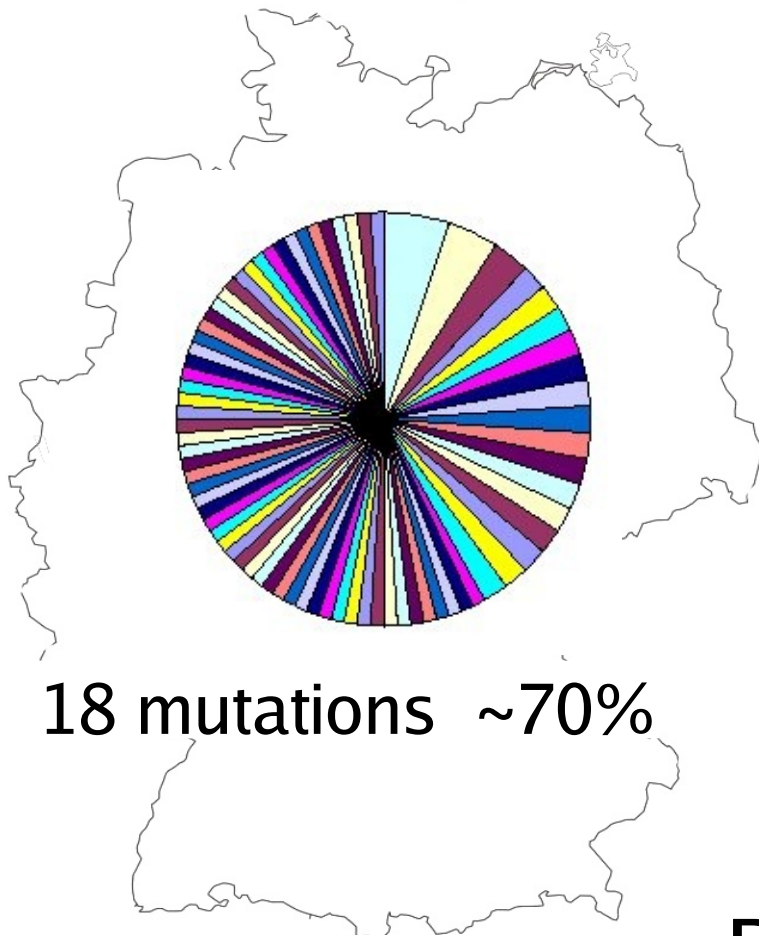
... and Poland?



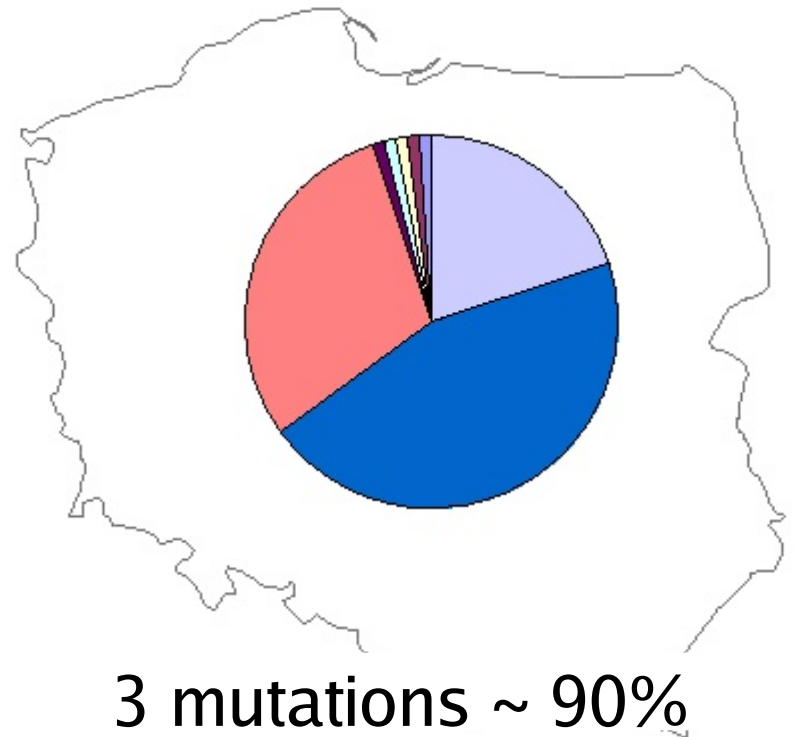
Homogeneity

Population Genetics

Germany



Poland



BRCA1(+) carriers

Hardy-Weinberg Law

HW Law does not apply?

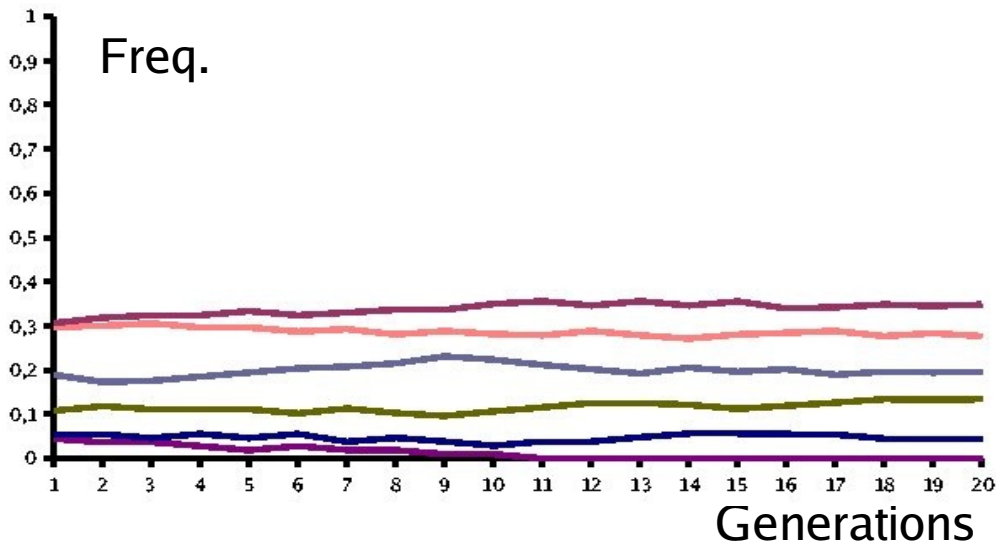
- Selection (natural or not) is taking place. Including selective mating (inbreeding, sexual selection)
- Allelic drift operating in small populations (genetic bottleneck)

Allelic drift

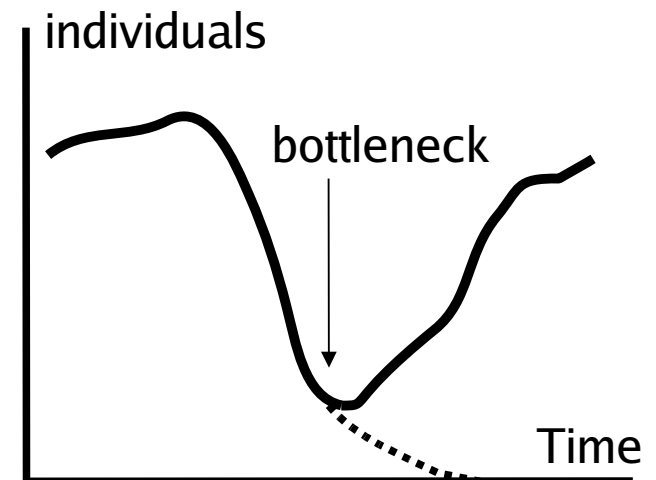
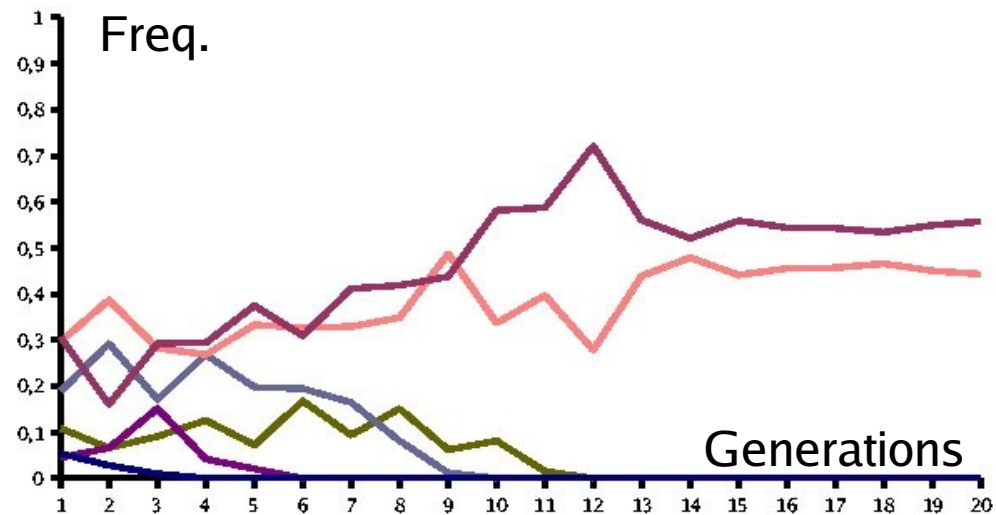
- Allelic distribution changes over time due to random events
- Reduction of heterogeneity
- Effects depend on the population size

Allelic drift & bottlenecks

- For large populations

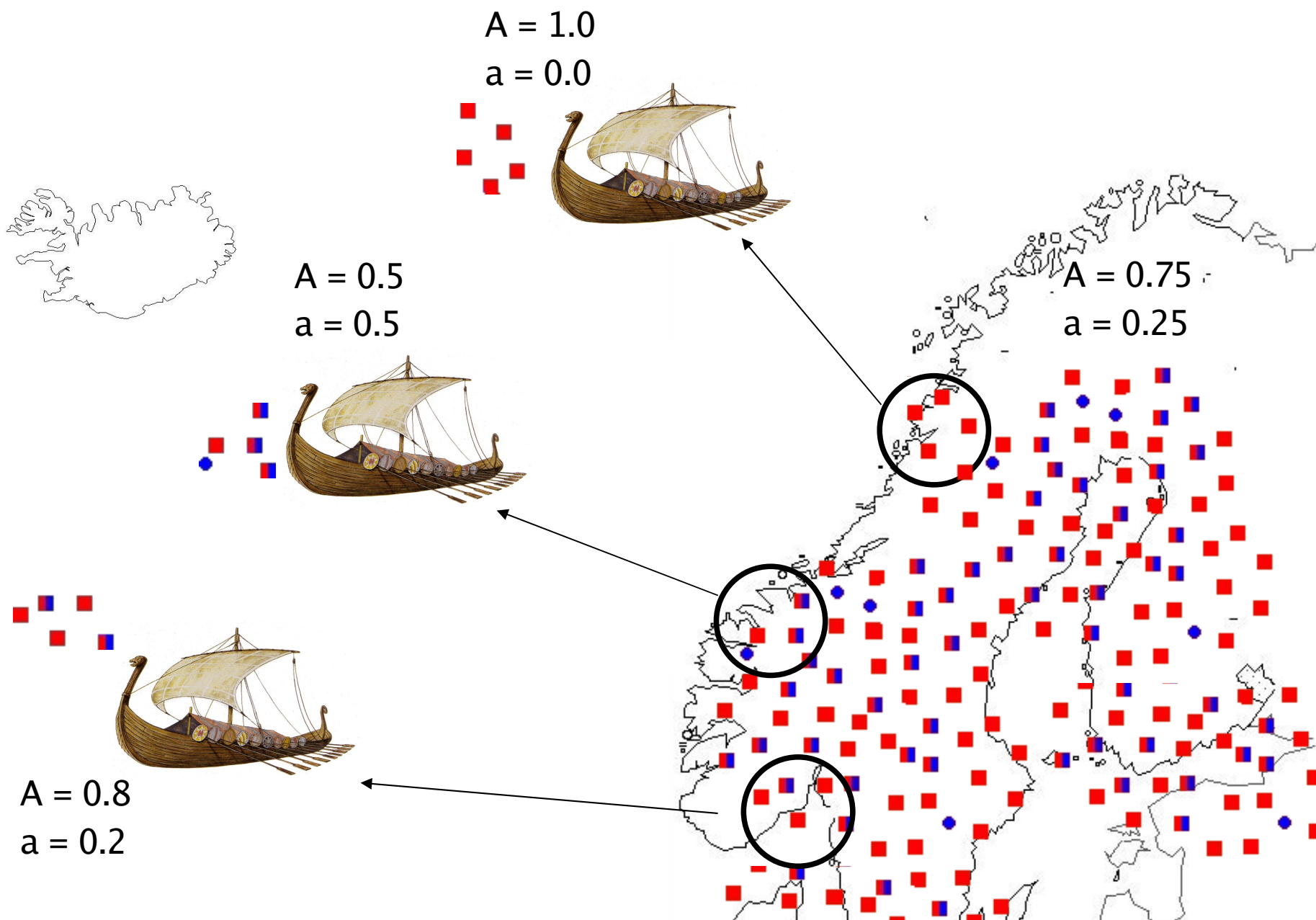


- For small population sizes: **genetic bottleneck**



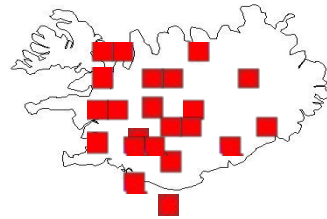
Allelic drift

Population Genetics



Founder effect

$A = 1.0$
 $a = 0.0$



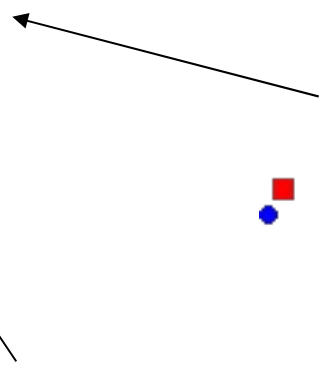
$A = 1.0$
 $a = 0.0$



$A = 0.5$
 $a = 0.5$



$A = 0.8$
 $a = 0.2$



Founder effect

BRCA1

Mutation	Ethnic groups
5382insC	Ashkenazi Jewish, Slavic , Hungarian
300T→G	Slavic , Hungarian
4153delA	Slavic
3171ins5	Scandinavian
2595delA	Scandinavian
1806C→T	Scandinavian
1201del11	Scandinavian

Summary

Mutation & recombination



Immigration



Directional Selection



Inbreeding



Assortative mating



Drift (small populations)



Balancing Selection



less

more



Genetic variability

Population Genetics