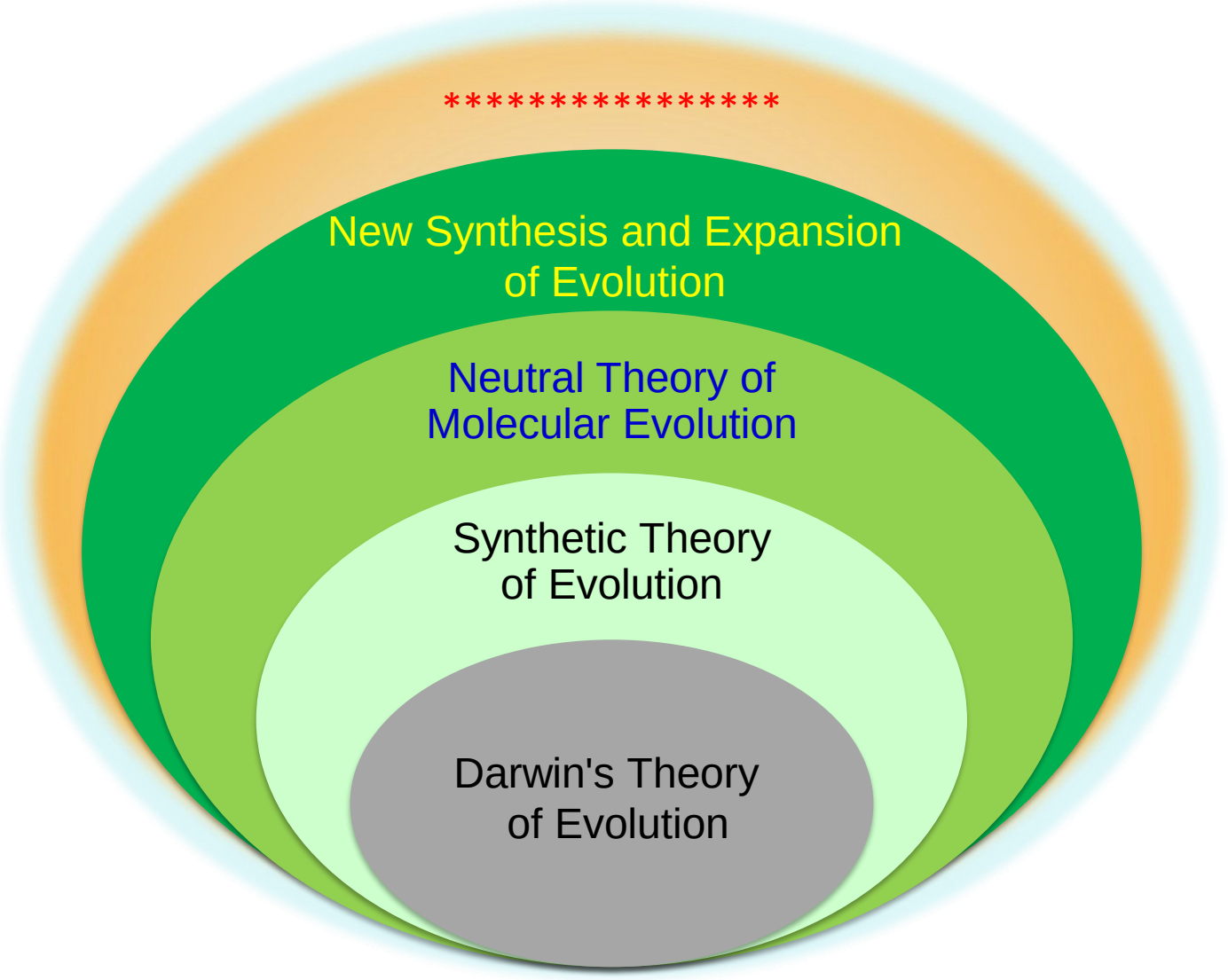
Evolution of Evolutionary Theories



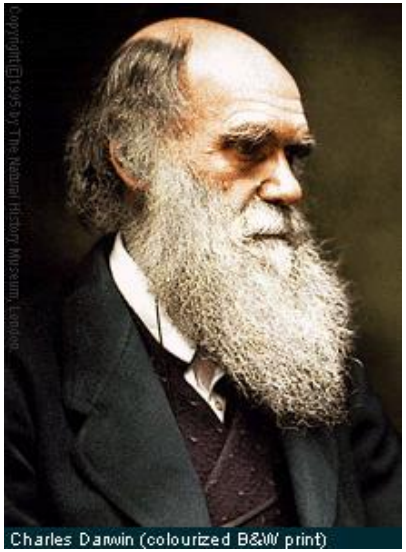
Survival of the Fittest

- **Selectionists/Darwinists:**

  Variation in nature

  – Created by mutation

  – Fixed and maintained by selection


Charles Darwin (colourized B&W print)

Survival of the Luckiest

- **Neutralists:**

  Variation in nature

  – Created by mutation + genetic recombin...

  – Fixed and maintained mainly by Drift

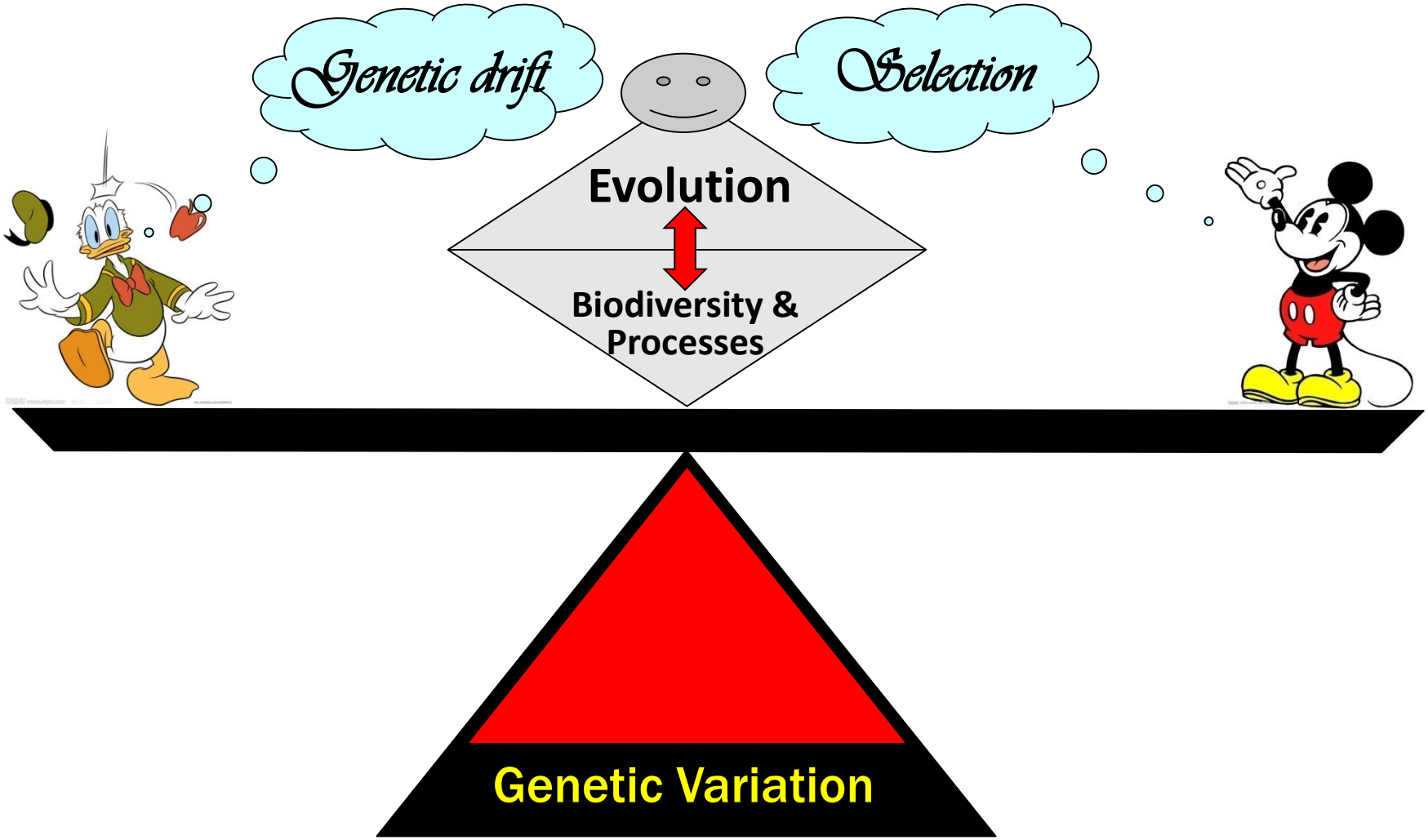

MOTOO KIMURA

The neutral theory of molecular evolution

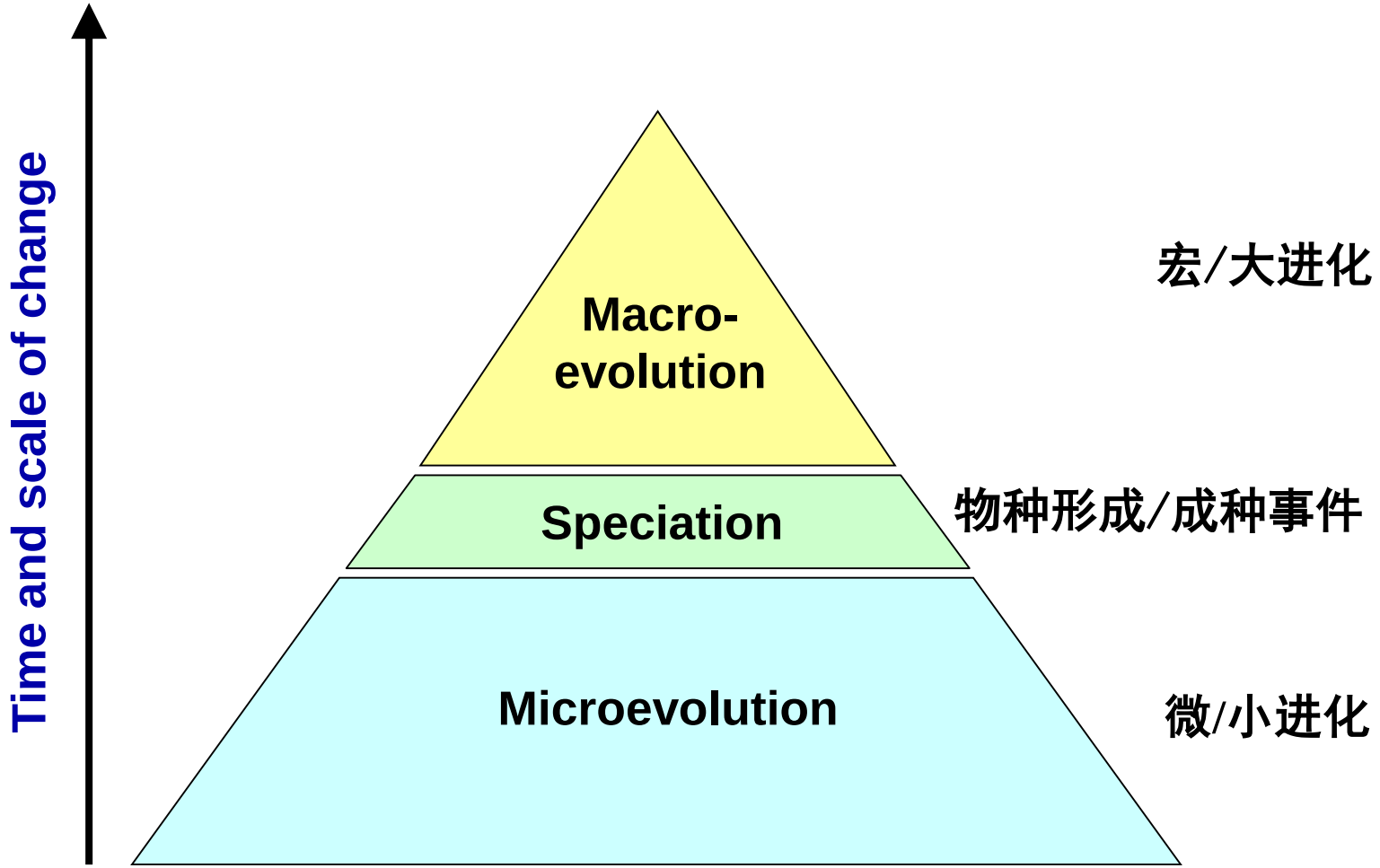
- **The neutral theory** claims that most observed variation (esp. at molecular level)— both polymorphism within species and divergence between species — is not due to natural selection driving the fixation of advantageous mutations but to random fixation of selectively neutral mutations which confer no fitness advantage.

- **The main power** of **neutral theory** is to provide a **theoretical expectation for genetic variation** in the absence of selection.

The Rise and Fall of Selectionism



进化在一定意义上就是世代间的变化



§5.2 Heredity and Variation of Plants

植物的遗传与变异

- **Heredity and Variation**
  遗传与变异
- **Variation Sources of Plants**
  个体变异的基础
- **Genetic Variation in a natural population**
  自然居群中的遗传变异
- **Variation and selection**
  变异与自然选择

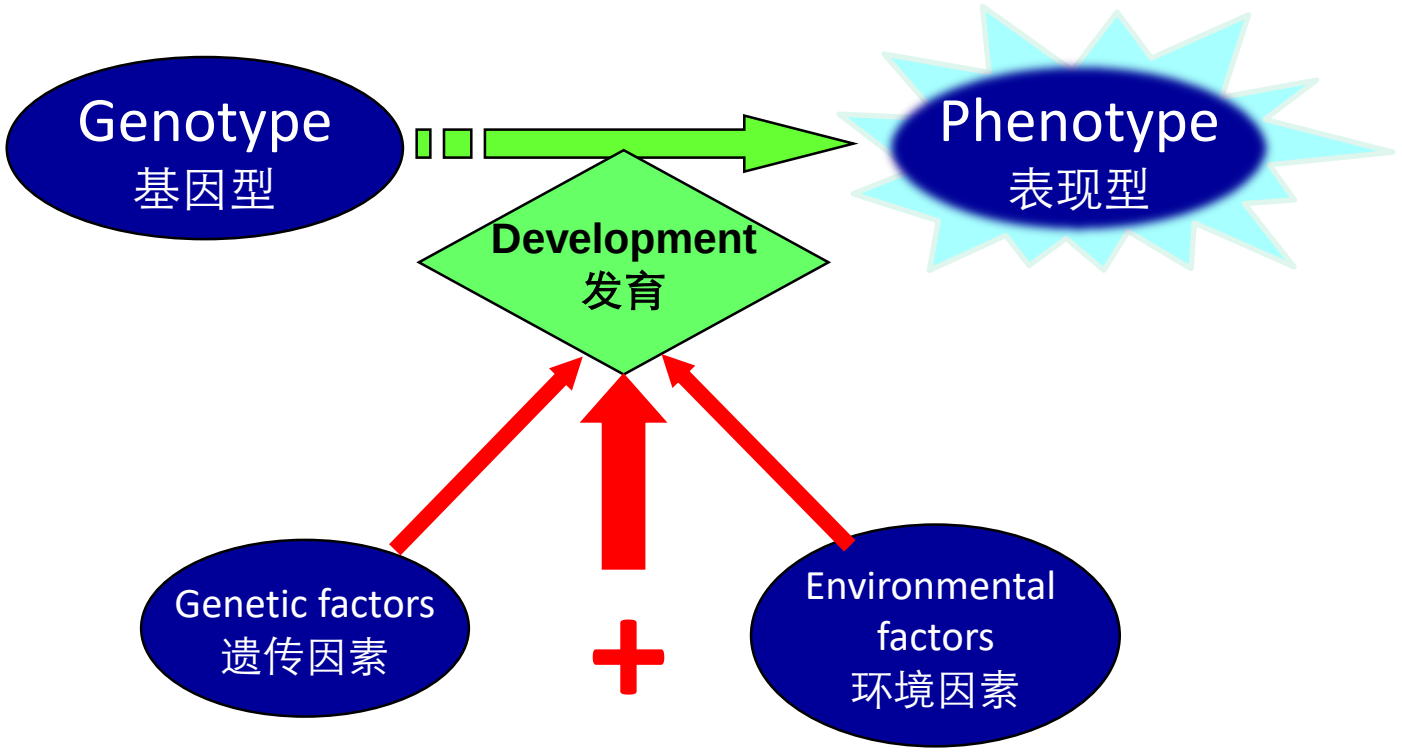
Heredity of Plants

- **Transmission of traits/genes from one generation to the next**
  - ➢ Asexual Reproduction
  - – offspring inherits all of its genes (DNA, chromosomes) from a single parent
  - ➢ Sexual Reproduction
  - – offspring inherits genetic material from two parents (1/2 from each)
- **Offspring differs somewhat from parents phenotypically and genetically**------this means *EVOLUTION*

Variation of Plants/植物的变异

变异——由遗传或环境因素所引起的不同层次，包括细胞间、生物个体间、不同居群间、物种及其以上分类阶元间的任何变化。

☞ Sources of phenotypic variation 表型变异的来源



Variation of Plants/植物的变异

变异——由遗传或环境因素所引起的不同层次，包括细胞间、生物个体间、不同居群间、物种及其以上分类阶元间的任何变化。

☞ Sources of phenotypic variation 表型变异的来源

- 环境因素引起的表型变异
  - Phenotypic plasticity/表型的可塑性
- 遗传因素引起的表型变异
  - Mutation/突变
  - Meiosis/Recombination of sexual reproduction /有性生殖过程中产生的遗传重组
- 发育过程引起的表型变异
  - Spatiotemporal deployment of developments /发育的时、空调整
  - Epigenetic change /表观遗传变化

Genetic Variation

- **Genetic Variation**: The **substrate** for natural selection and the **raw material** for Evolution/自然选择的底物和进化的原材料
  - If there is **no genetic variation** neither ***Natural Selection*** nor ***Genetic Drift*** would be able to change allele frequencies, because there would be nothing to change
- **Sources of Genetic Variations**
  - Allelic variation/等位基因的变异：***mutations***
  - Genotypic variation/基因型的变异：***meiosis* in sexual reproduction**

Genetic Variation

- **Mutations (changes in the coding and noncoding regions):**
  - Nucleotide substitutions, insertions, deletions/核苷酸的替换、插入和缺失
  - Gene duplications or deletions/基因重复和缺失
  - Exon Shuffling/外显子重排
  - Chromosomal duplications or deletions/染色体的重复和缺失
  - Deletions of large chromosomal regions /染色体大片段的缺失
  - Chromosomal inversions and translocations/染色体倒位和移位
  - Whole Genome Duplications (WGD) /整个基因组的重复/多倍化
- **Meiosis (sexual reproduction):** No novel alleles, only novel genotypes:
  - Genetic Recombination: Shuffling of combinations of alleles along a chromosome/染色体上基因的重排
  - Random Mating: Recombination of haploid chromosomes/同源染色体重组

Types of Mutations

- **At the Nucleotide Level (Point mutations/点突变):**
  - Single nucleotide substitutions (transitions/替换, transversions/颠换)
  - Nucleotide Insertion or/and deletion, "*INDEL*"
- **At the "Gene" Level:**
  - Gene Insertions (Gene duplications, transposons/转座子, horizontal gene transfer"*HGT*"/水平基因转移)
  - Gene Deletions (pseudogenization /假基因化, transposons)
  - Exon Shuffling/外显子重排
- **At the Chromosome Level:**
  - Chromosome duplications, deletions, inversions, fusions
- **At the Genome Level:**
  - autopolyploidization/同源多倍化
  - allopolyploidization/异源多倍化

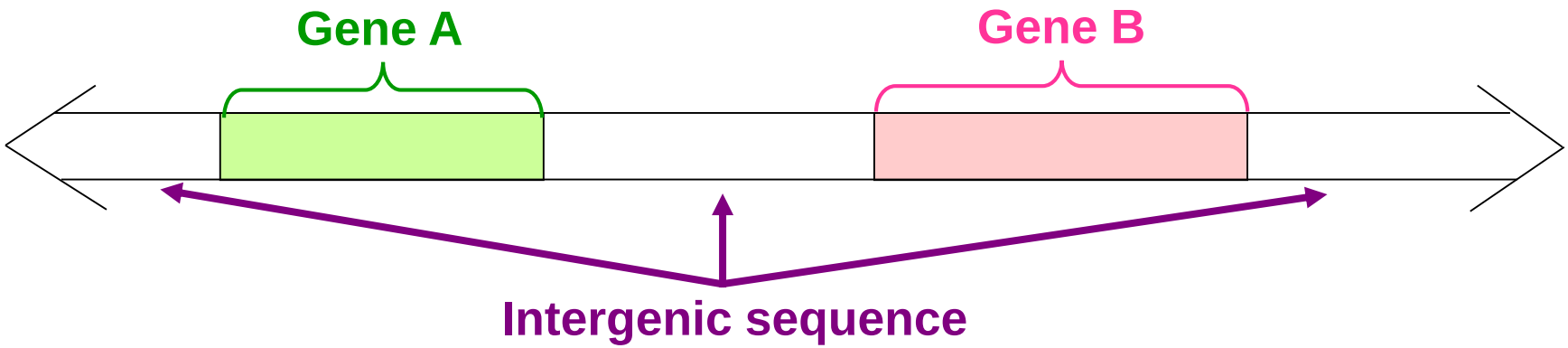
Mutations: Double-Edged Sword

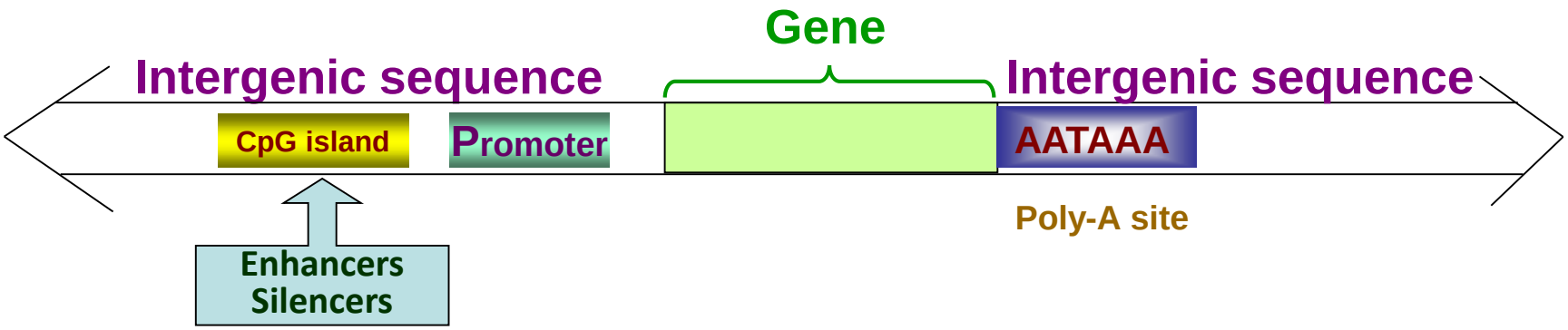
- **Most mutations** are **'neutral'** with no effect on fitness, as most of the genome is nonfunctional
- **Most mutations** that affect functional genes are **harmful**
- **A very small number** of mutations are **favorable**, selection for favorable mutations leads to adaptation.
- **Mildly deleterious mutations persist longer** in a population because it takes longer to select them out
- **Recessive mutations remain longer** in the population, because they are eliminated when homozygous, not when heterozygous; when they are heterozygotes, they are "masked" from selection

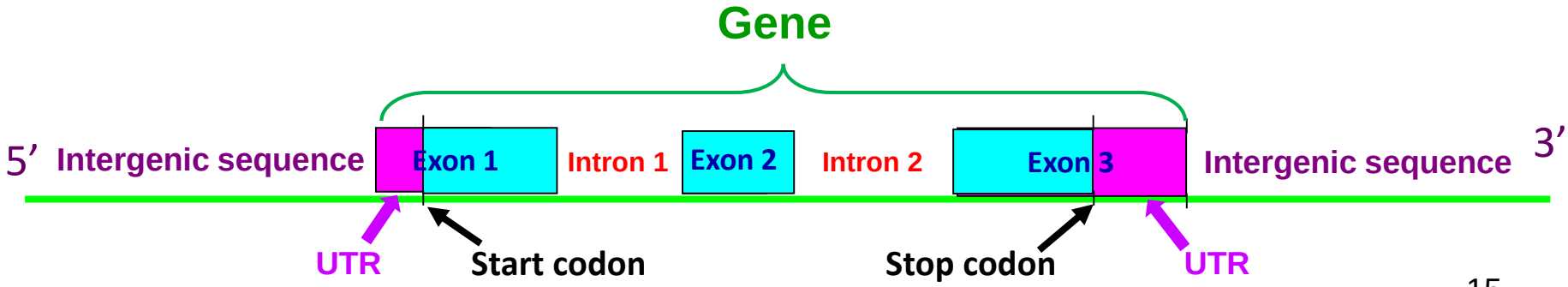
Most mutations have no Effect

- Most of the genome is non-coding sequence and has no function (up to 95%):
  - mutations here are “Neutral”
- Mutations that affect function are what matter within coding sequences of genes, or within regulatory sequences that affect the expression of genes

Gene Structure ——— DNA: A, C, G, T







Genetic variation of individuals

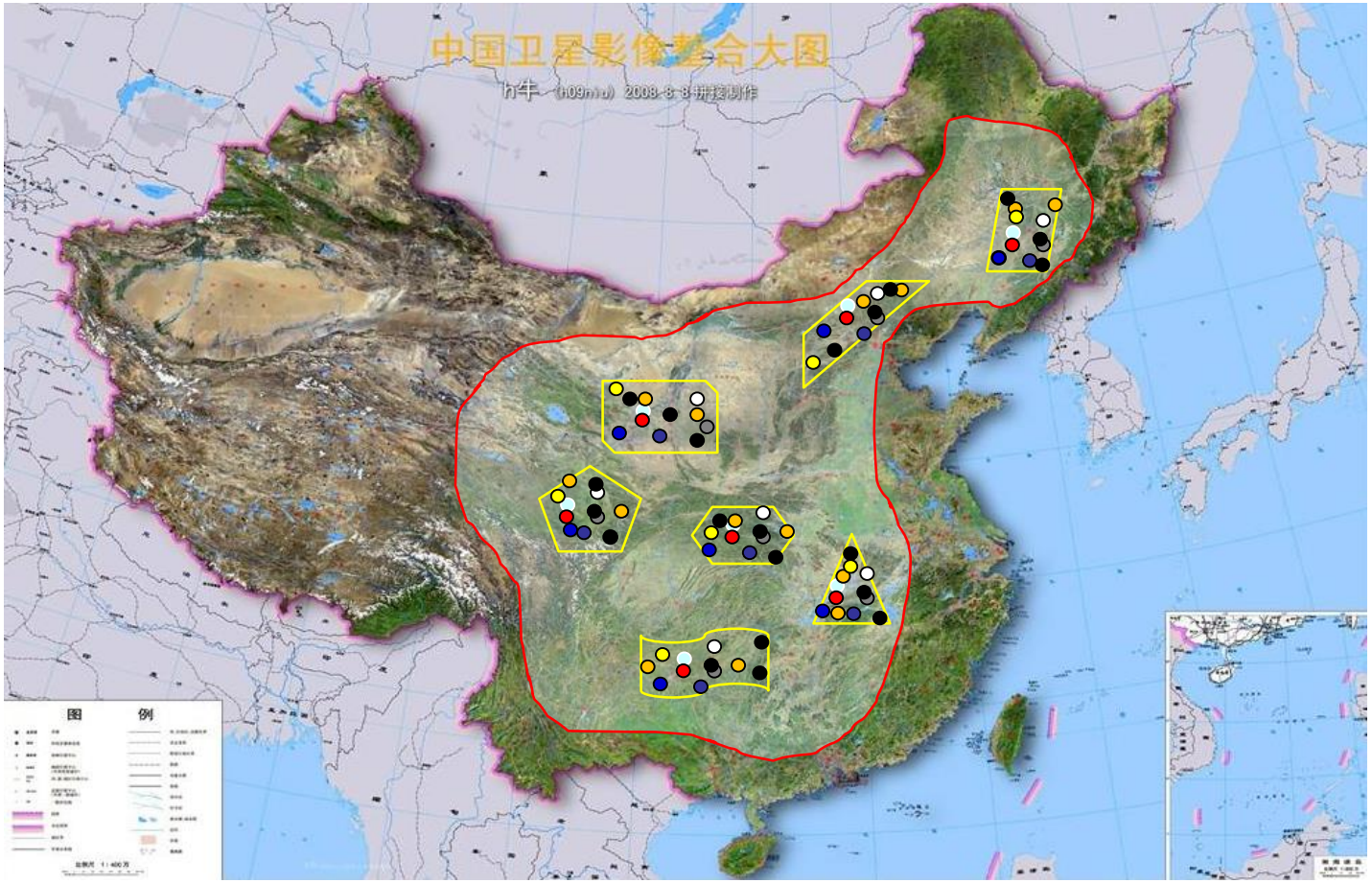
?

Species

The “hierarchical” structure of a species

物种构成的嵌套式结构

- **Species/种、物种------ group of populations whose individuals can potentially interbreed**




What happens in a population?
What's the "population"?

§5.3 Population genetic structure and Natural selection

植物居群的遗传结构与自然选择

- **Concept of 'Population'**
  居群/种群/群体的概念
- **Population genetic structure**
  居群的遗传结构
- **Factors shaping 'population genetic structure**
  影响居群遗传结构的因素
- **Natural selection**
  自然选择

Population /居群/种群/群体

Population/居群—— A population is a group of interbreeding individuals (belonging to the same species) that exist together in time and space /指分布在同一地区、同一物种的一群互交能育的个体群。

- **Population ------** this definition has two components:
  - ☞ A genetic one------ individuals belong to the same species/ gene pool
  - ☞ A spatial one ----- individuals live in the same area
- **A population ------** a gene pool:
  - ☞ meaning the total aggregate of genes in a population at any one time
  - ☞ consisting of all gene loci in all individuals of a population
- **The nature of natural populations** ------ *dynamic* in many dimensions:
  - ☞ *over time* they change in size, density and location
  - ☞ *over space* they can fragment into several subpopulations and join with others.

The Smallest Unit of Evolution

- One **common misconception** about evolution is that individual organisms evolve, in the Darwinian sense, during their lifetimes
- Natural selection acts on individuals, but populations evolve, so the unit of evolution is **POPULATION**
- Genetic variations in populations contribute to evolution
- **Population genetics** provides a foundation for studying evolution

Population Genetics & The Modern Synthesis of Evolution

- **Population genetics**
  - Is the study of how populations change genetically over time
  - Reconciled Darwin's and Mendel's ideas
- **The modern synthesis**
  - Integrates Mendelian genetics with the Darwinian theory of evolution by natural selection
  - Focuses on populations as units of evolution

Population Genetics & Microevolution

- **Population genetics**
  - Is the study of how populations change genetically over time
  - Reconciled Darwin's and Mendel's ideas
- **Microevolution**
  - Is change in the genetic makeup of a population from generation to generation

Microevolution

------The Evolution of Populations

Microevolution *usually* refers to the Changes occurring at population level within a species (change over short time scales---generations)

Genetic variation and structure
In natural populations

Microevolution

------The Evolution of Populations

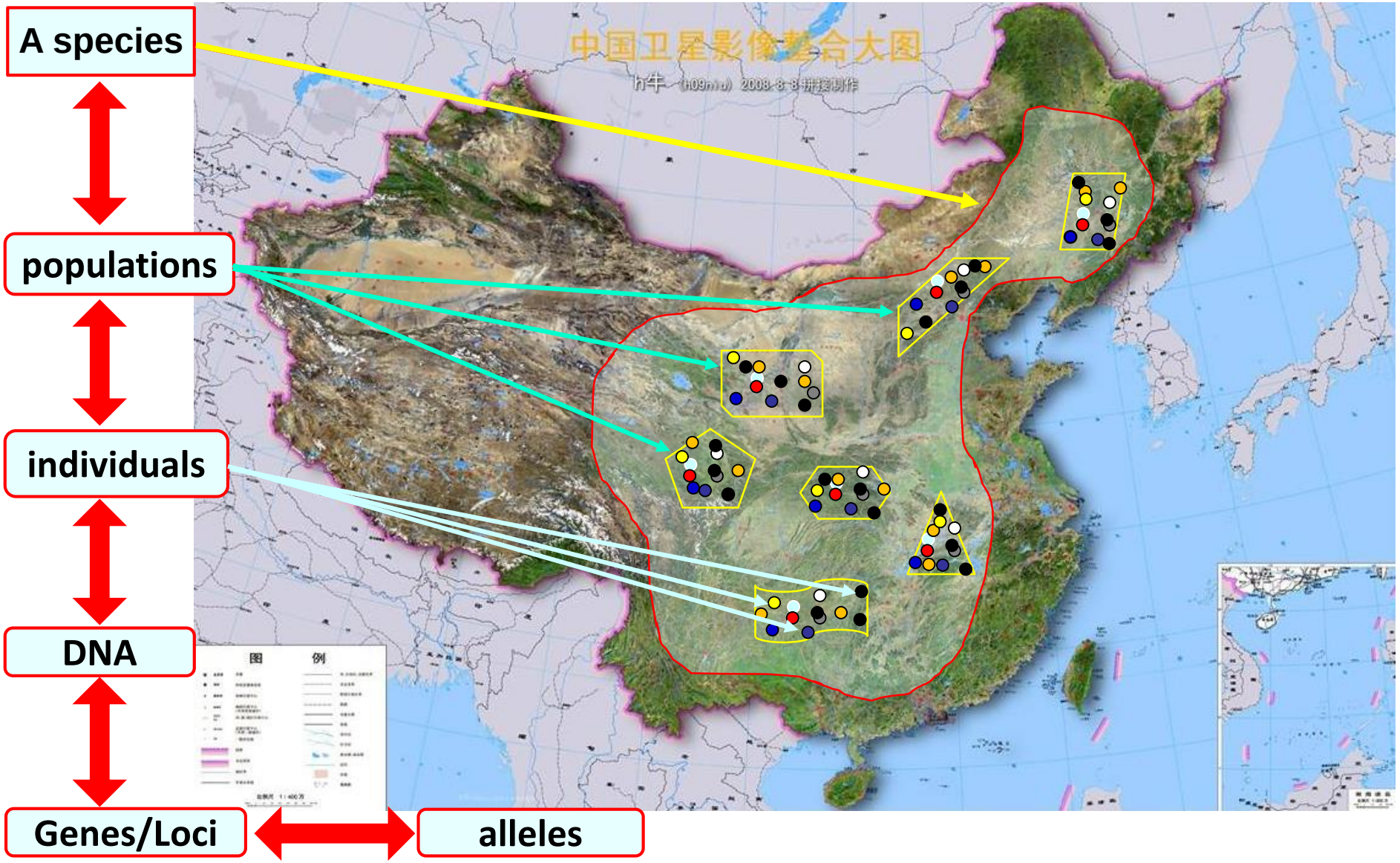
- **Microevolution** is a change in allele frequencies of a population over generations.
- Natural selection acts on **individuals**, but only **populations** evolve.
- **Population** is the smallest unit of evolution
- **Genetic variations** in a population contribute to the fitness difference among individuals.
- **Genetic variations** in populations of a species contribute to evolution.

Firstly,

To review important concepts and definitions of Microevolution

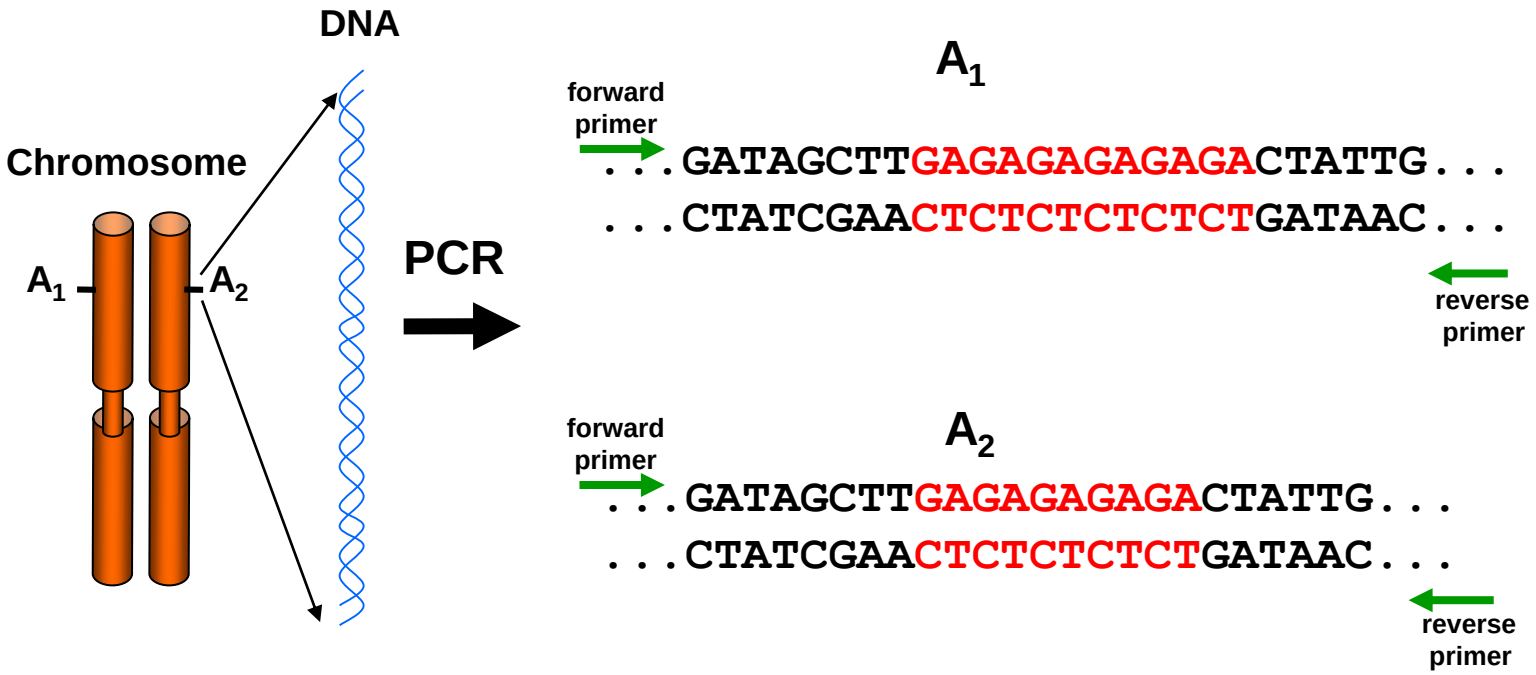
- **Species**/种、物种------ group of populations whose individuals can potentially interbreed
- **Population**/居群------ potentially interbreeding group of individuals occupying a geographic area (or localized group of individuals of the same species).
- **Locus (loci)** /位点、基因位点------ the place where a particular gene resides on a chromosome, i.e. a point in the genome, identified by a marker.
- **Allele** /等位基因------ an alternative form of a gene. One of the different forms of a gene that can exist at a single locus.

The “hierarchical” structure of a species



Locus and Allele

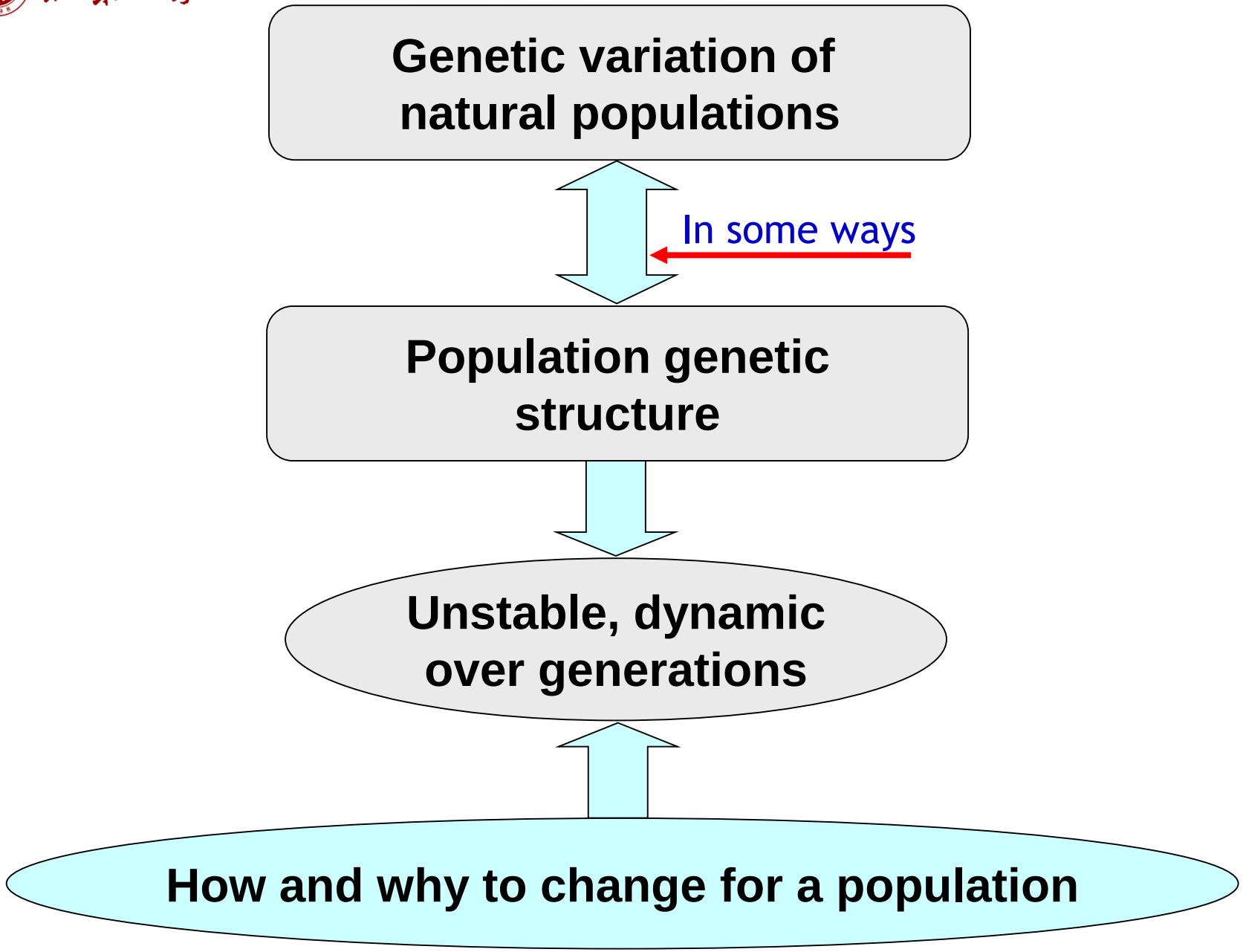
- A locus is a particular DNA sequence located on a chromosome.
- Alleles are variant DNA sequences at a locus.

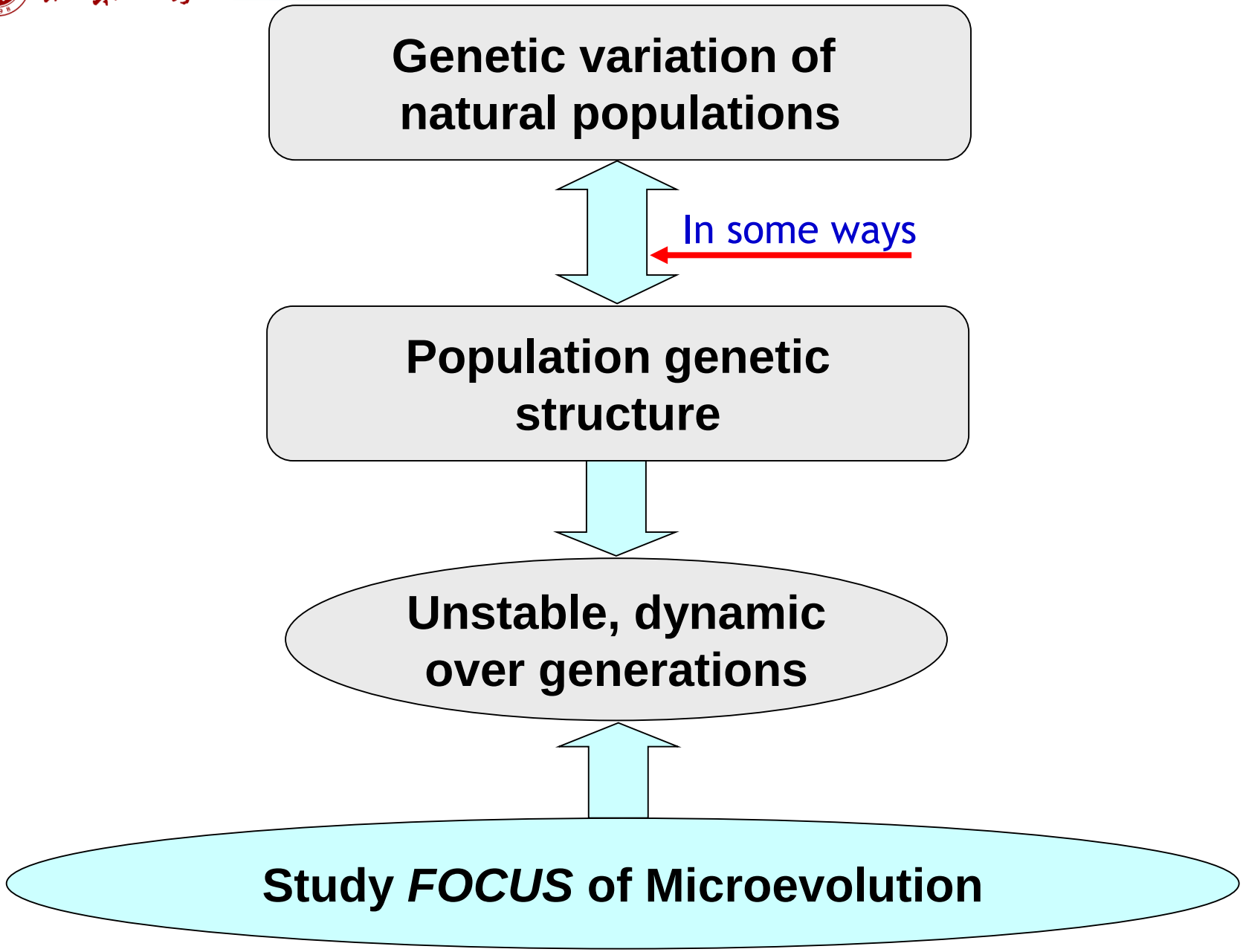


***A locus may or may not represent a gene.**

To review important concepts and definitions of Microevolution

- **Genome**/基因组 ------ A single copy of all genes in an organism, so a diploid individual would have two complete genomes.
- **Genotype**/基因型 ------ The specific allelic composition of a cell, either of the entire cell or more commonly for a certain gene or a set of genes.
- **Phenotype**/表型、表现型 ------ The observable morphological forms, structure, function or behavior of a living organism.
- **Homozygote**/纯合子 ------ have 2 identical alleles for a given trait (dominant or recessive) (e.g. *AA* or *aa*)
- **Heterozygote**/杂合子 ------ have 2 different alleles for a given trait (e.g. *Aa* or *aA*)


Genetic variation of natural populations
In some ways
Population genetic structure
Unstable, dynamic over generations
How and why to change for a population


Genetic variation of natural populations
In some ways
Population genetic structure
Unstable, dynamic over generations
Study *FOCUS* of Microevolution

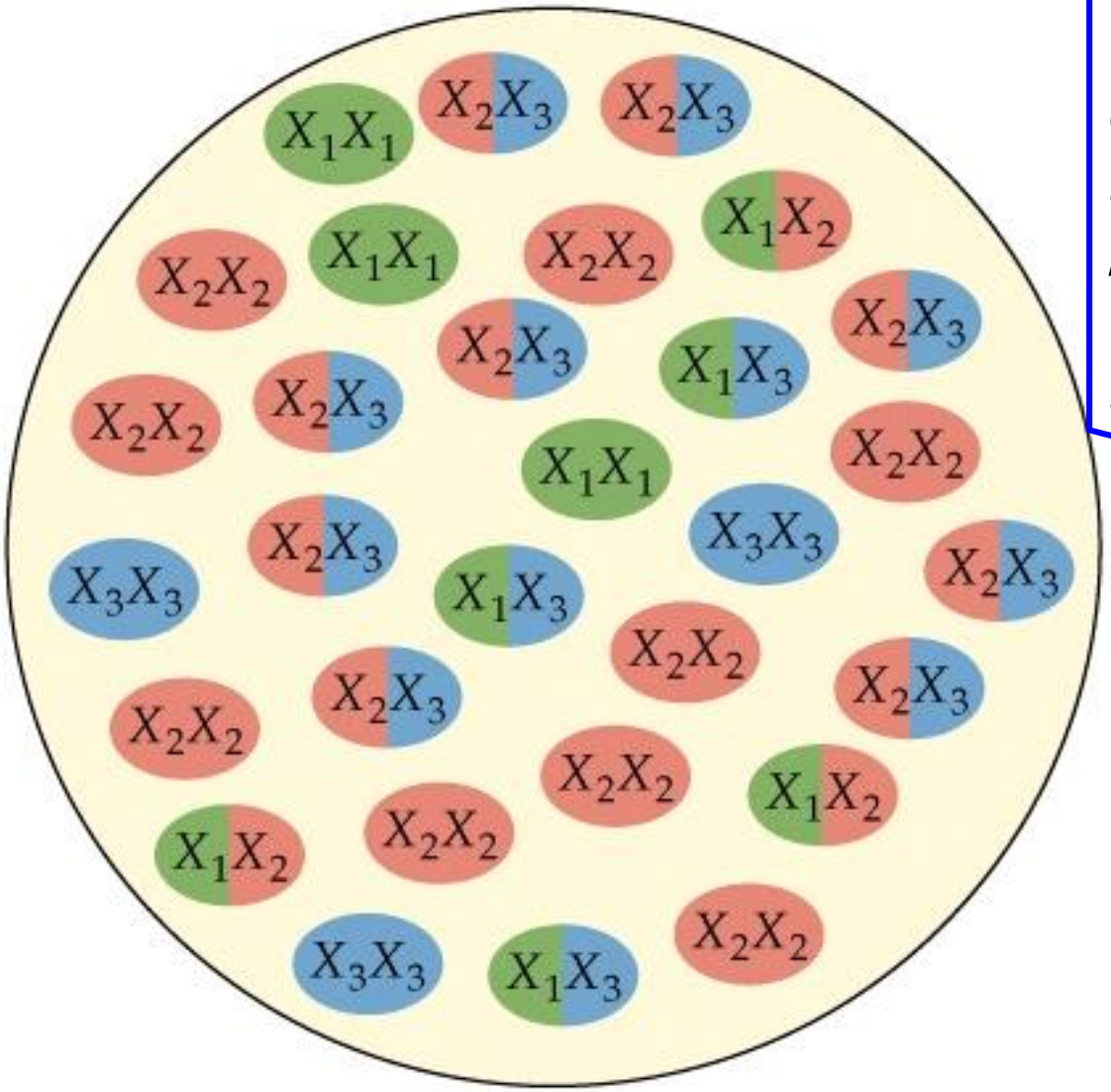
Genetic composition of a population
居群的遗传组成

Allele frequency/基等位因频率 : the proportion of a specific allele out of all the alleles in the all the individuals of a population/指居群中某一等位基因占该位点上全部等位基因的比率。

Genotype frequency/基因型频率 ： the proportion of a specific genotype out of all the genotypes in all the individuals of a population/指居群中某一特定基因型所占的比率。

Allele Frequency /等位基因频率

------ the frequency of a particular allele in the population.



A example:

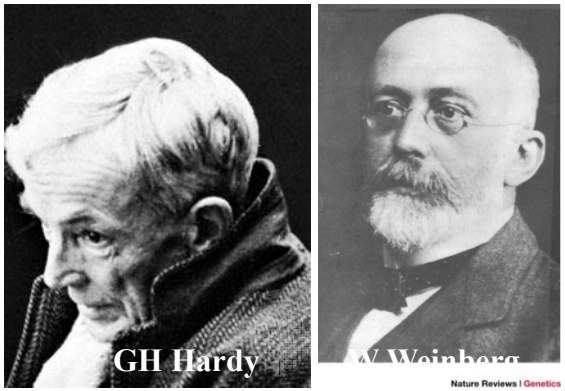
Calculating the frequency of a particular allele or a genotype of the locus /gene (X) with 3 alleles in a population with 30 individuals

Nature of the Population
—— Hardy-Weinberg Equilibrium

A population will be in Hardy-Weinberg Equilibrium if the following five assumptions are met.

1. Infinite population size
2. Random mating
3. No gene flow, i.e. no movement of individuals from population to population
4. No mutation (no biochemical changes in DNA that produce new alleles.)
5. No natural selection, i.e. the different genotypes have equal fitness.

Hardy-Weinberg Equilibrium



Godfrey H. Hardy (English mathematician) and **Wilhelm Weinberg** (German physiologist) independently enunciated in 1908 what is now known as the Hardy-Weinberg equilibrium.

Hard-Weinberg equilibrium ------- The gene and genotype frequencies do not change in a large Mendelian population with random mating and no mutation, selection, or gene flow. Considering a locus with two alleles *A* and *a* with frequencies *p* and *q* The relationship between the allele and genotype frequencies can be expressed as following:

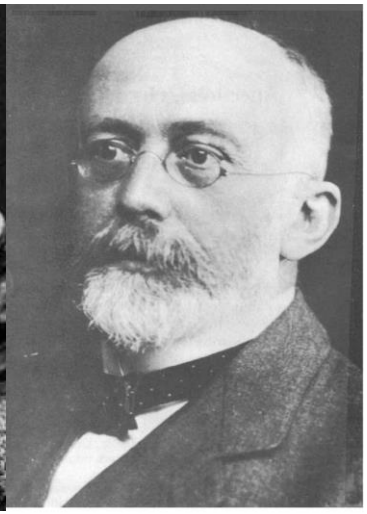
$$(p + q)^2 = p^2 + 2pq + q^2 = 1$$

★在一个无限大、随机交配的居群中，如果没有突变、迁移和选择因素的影响，居群的基因频率和基因型频率在世代间保持不变。

Hard-Weinberg Theorem/Equilibrium

Hardy-Weinberg 定律/平衡

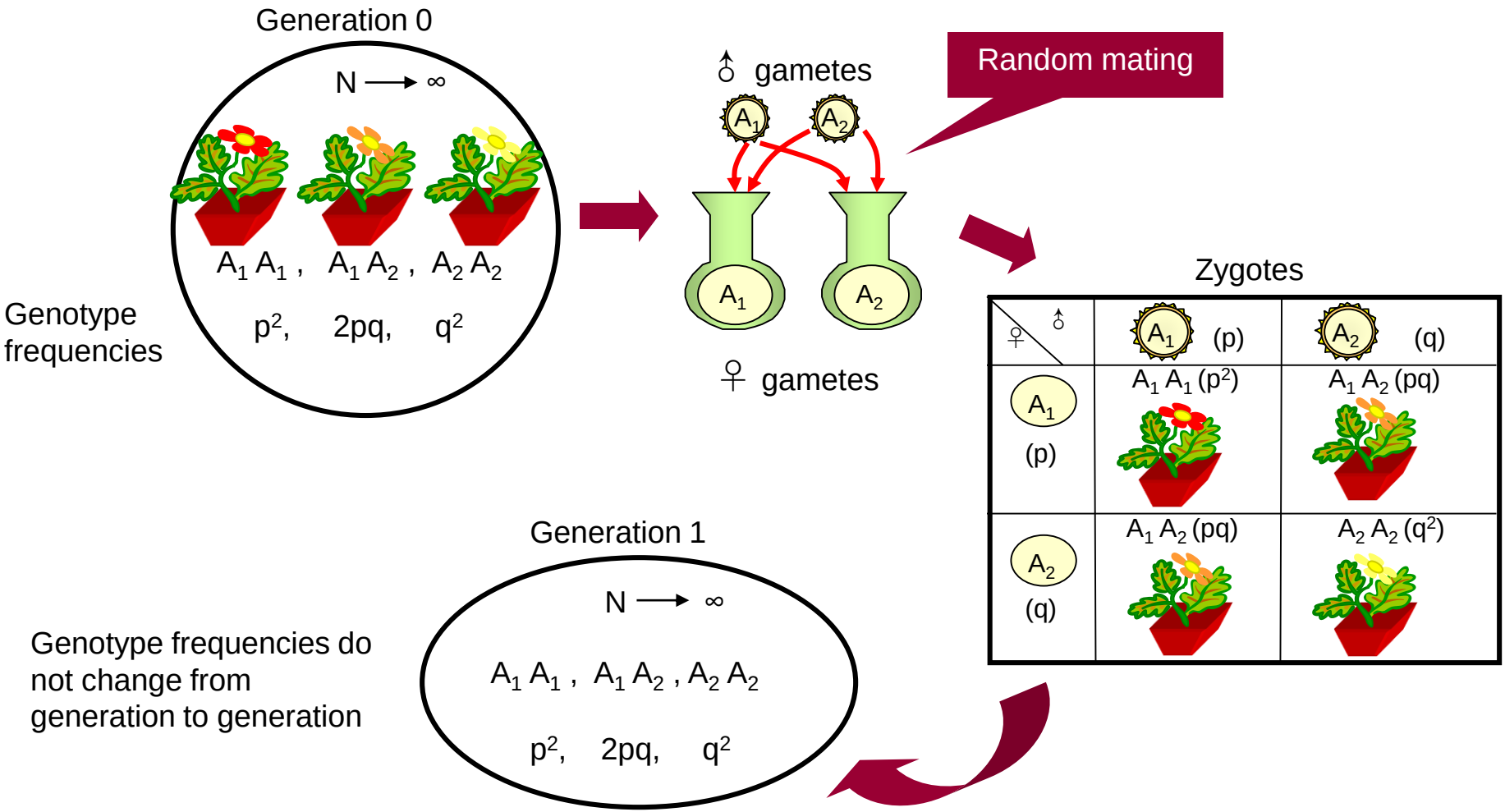
此定律表述了居群在一定条件下的自然属性，是一个统计学意义上的定律。



★在一个无限大、随机交配的居群中，如果没有突变、迁移和选择因素的影响，居群的基因频率和基因型频率在世代间保持不变

没有进化

Demonstrating the H-W principle



Hardy-Weinberg equilibrium

A population at equilibrium obeys the Hardy-Weinberg rule:

Assume two alleles *A* and *a*

The frequency of *A* equals a value *p*

The frequency of *a* equals a value *q*

The frequency of the three possible genotypes is given by:

$$p^2(AA) + 2pq(Aa) + q^2(aa) = 1$$

Hardy-Weinberg rule

Assign $p = 0.5$ and $q = 0.5$:

	AA	*Aa*	*aa*
Fraction	1/4	2/4	1/4
Frequency	0.25	0.5	0.25

	A	*a*
A	*AA*	*Aa*
a	*Aa*	*aa*

Hardy-Weinberg rule

Consider a situation where the frequencies are not identical

Assign p = 0.7 and q = 0.3:

AA	$p \times p = (0.7)(0.7) =$	**0.49**
Aa	$2pq = 2(0.7)(0.3) =$	**0.42**
aa	$q \times q = (0.3)(0.3) =$	**0.09**
		1

A population of 1000

Consider a population of 1000 with this frequency of alleles and each individual generates two gametes:

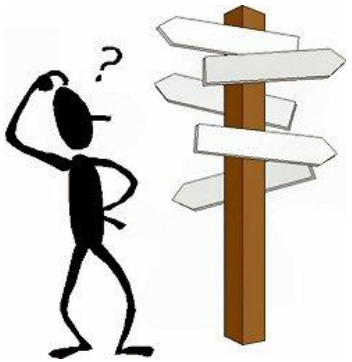
0.49(1000) = 490 *AA*	980 *A*	
0.42(1000) = 420 *Aa*	420 *A*	+ 420 *a*
0.09(1000) = 90 *aa*		180 *a*
	1400 *A*	+ 600 *a*
	0.7	0.3

The same allele frequency

What's H-W Equilibrium good for?

As allele and genotype frequencies will not change as long as the assumptions are met, then we can:

- Predict genotype frequencies given allele frequencies
- Predict the genotype frequencies in generation 2 If we know the allele frequencies in generation 1.



What does Hardy-Weinberg equilibrium mean?

Hardy-Weinberg Equilibrium is defined as the situation in which no evolution is occurring.

No Evolution !!

Assumptions of Hardy-Weinberg equilibrium essentially never occur in nature, so Hardy-Weinberg equilibrium is an ideal case!!

Five Agents of Microevolution

1. Finite population size

Genetic drift = random changes in allele frequency

Bottleneck

Founder effect

Inbreeding ➡ inbreeding depression

2. **Nonrandom mating** within a population

3. **Gene flow** can change a gene pool due to the movement of genes into or out of a population

e.g. migration

4. **Mutation** changes alleles

5. **Natural selection** leads to differential reproductive success

Population Size 居群的大小

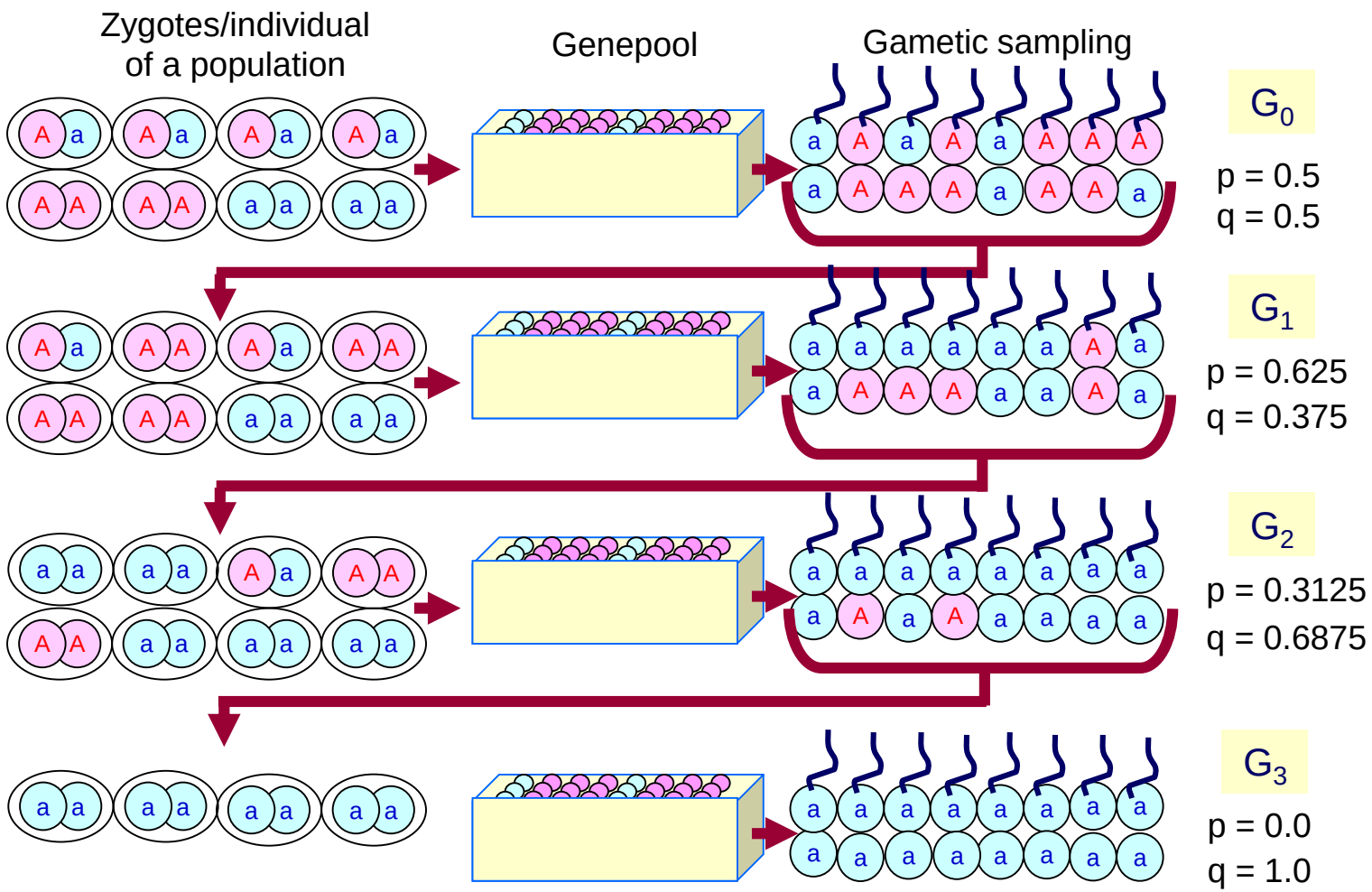
Hardy-Weinberg 定律是一个统计定律，只有在个体数量很大的居群中基因库才能稳定。因此，要保持居群处于 Hardy-Weinberg 平衡状态，有效居群要足够的大。

★ 有效居群的大小(Ne, effective population size)：
指居群中参与繁殖下一代的个体部分，通常它比实际居群要小得多。

Genetic Drift 遗传漂变

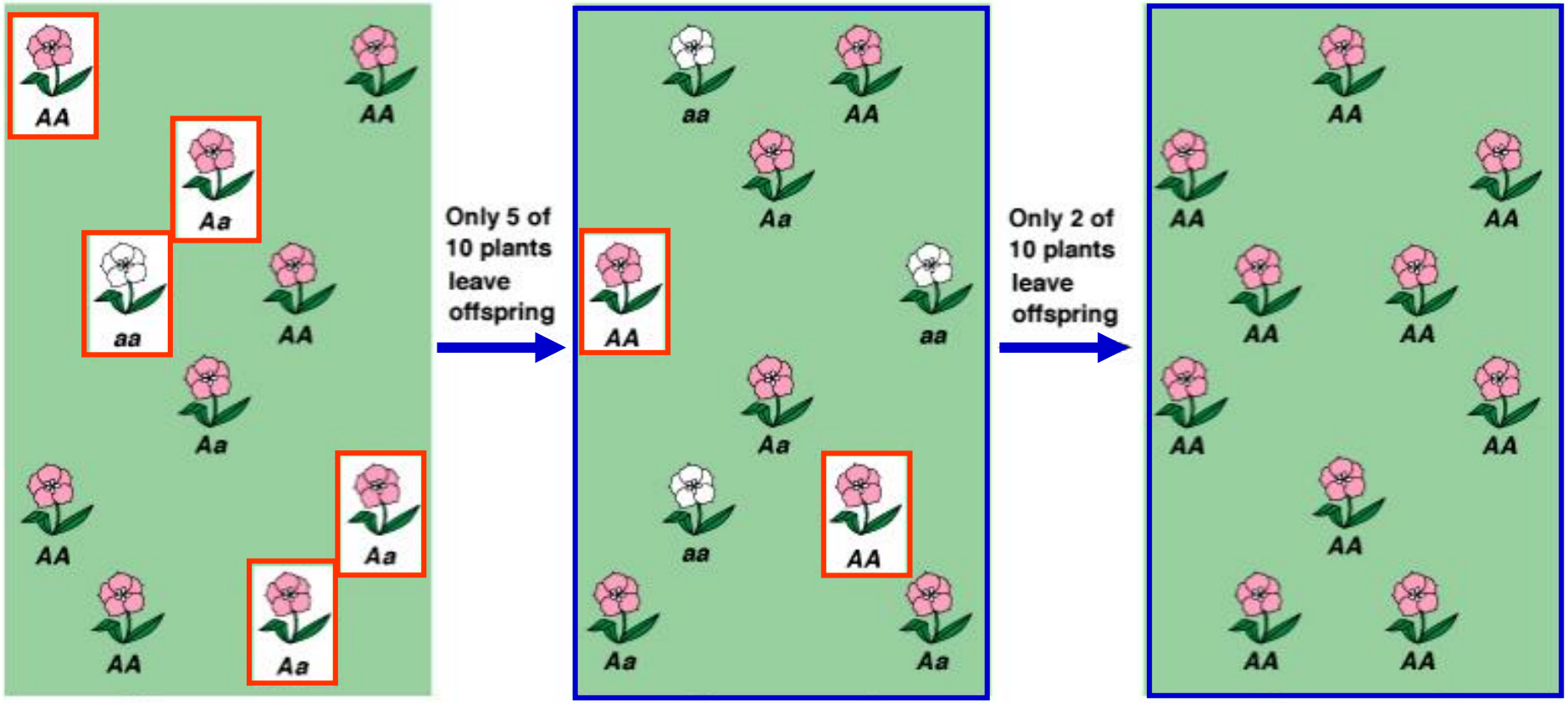
- **Concept of Genetic drift** —— it is the random change in allele frequency that results from the sampling of gametes from generation to generation by random chance.
- **It occurs if a population is not infinite in size.** Since no population is really infinitely large, there is always some genetic drift occurring; however, the effect is very small in large populations. The effect of genetic drift is larger and larger in smaller and smaller populations.

Genetic Drift

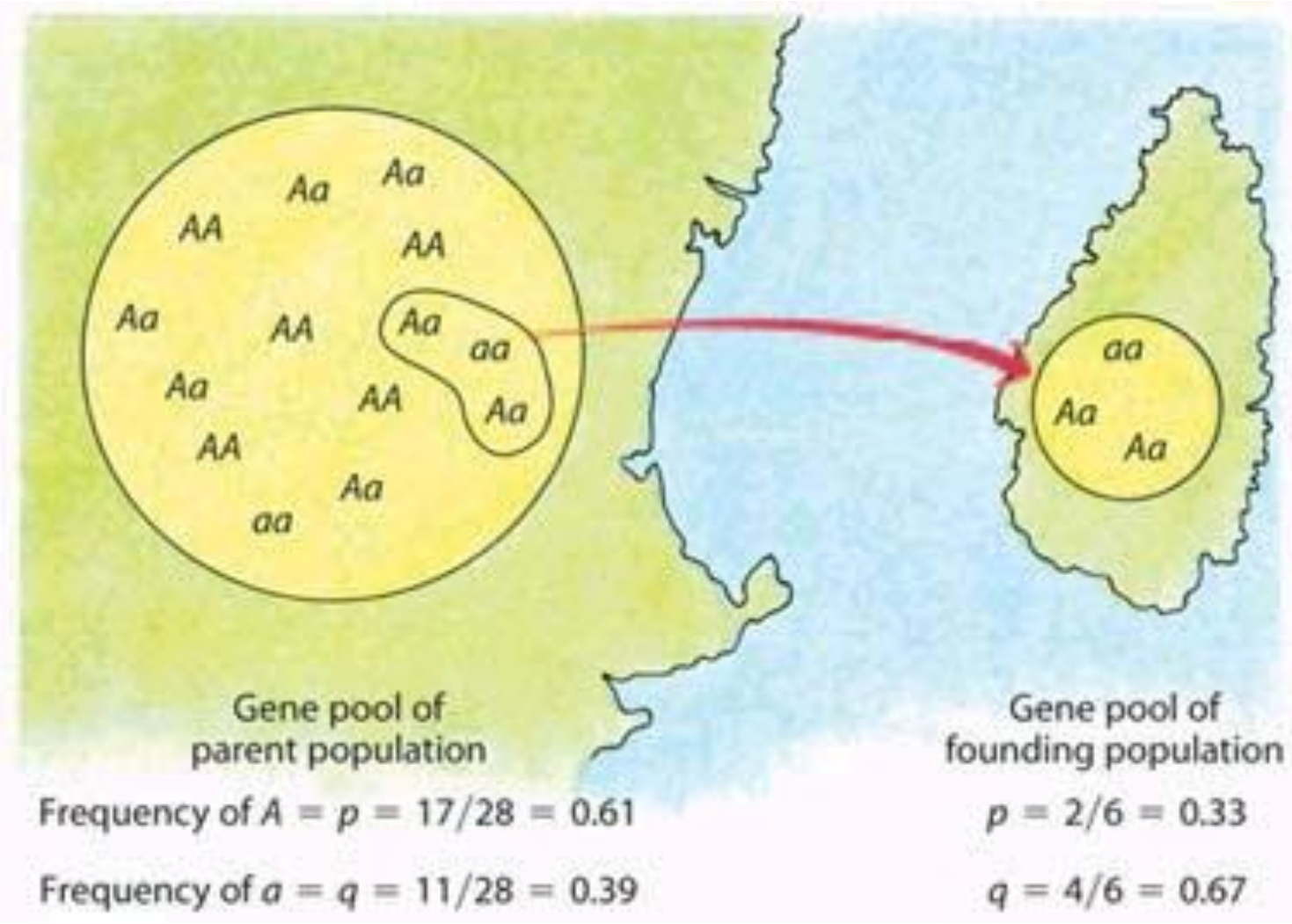


Genetic Drift 遗传漂变

- 遗传漂变是指由于随机因素使得小居群/种群中的基因频率在世代之间出现随机变化（增或减）的现象。



Founder Effect 奠基者（建立者）效应



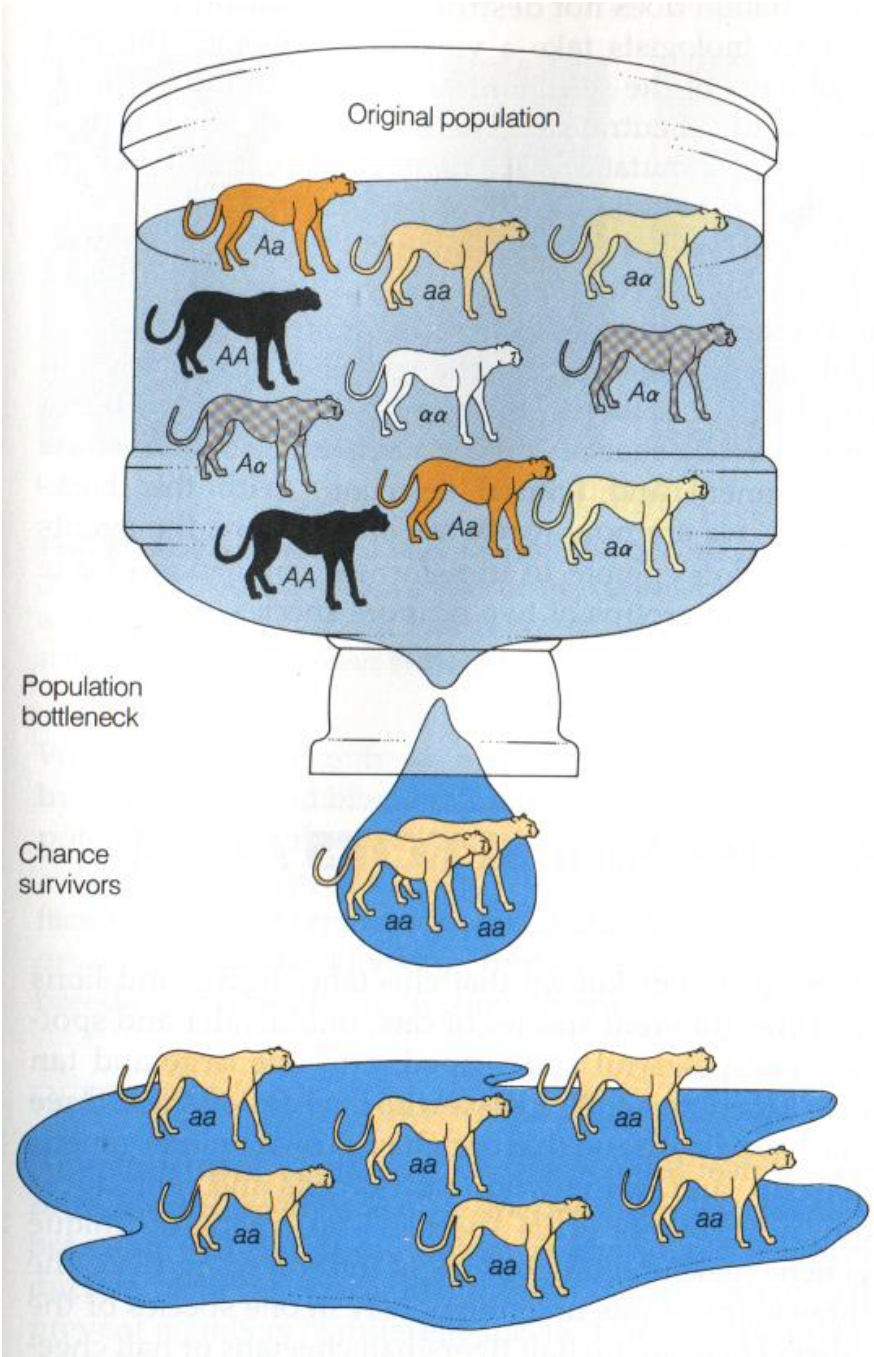
Founder effect

- Change in allele frequencies when a new population arises from only a few individuals.

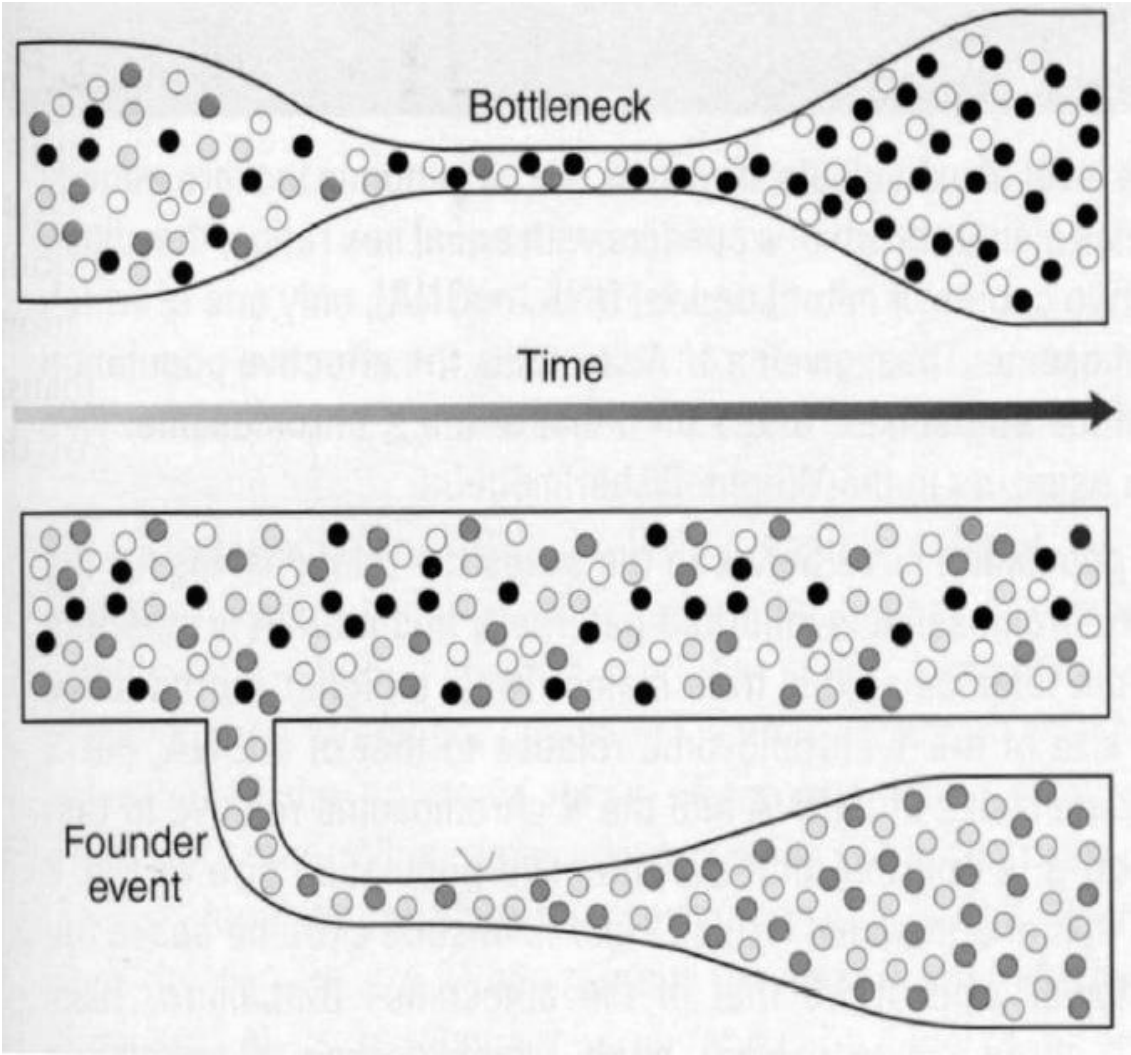
e.g., only a few birds move to an island.

Bottleneck Effect

瓶颈效应



Founder *vs* Bottleneck



Fonder Effect and Bottleneck Effect

奠基者（建立者）效应和瓶颈效应

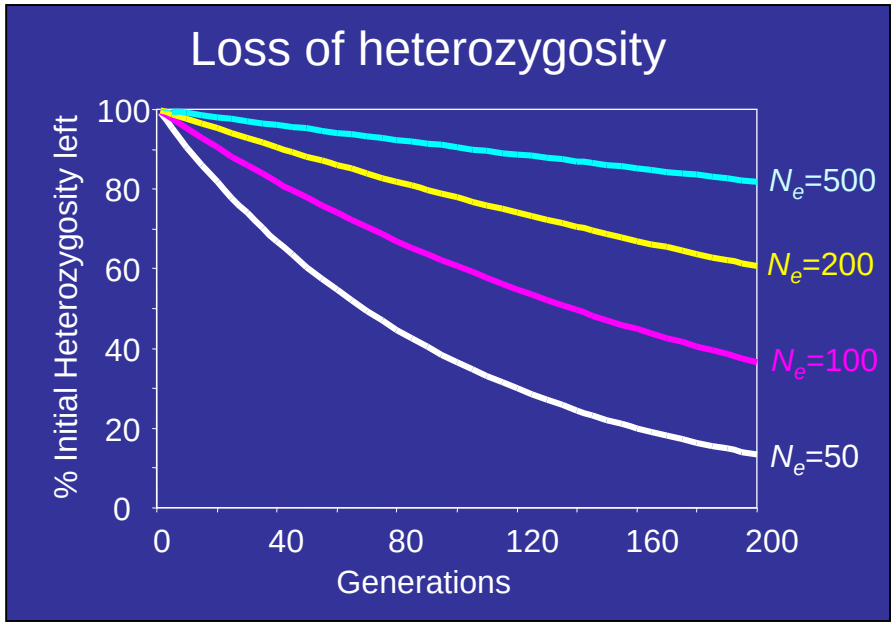
这是由少数个体的基因频率决定了它们后代基因频率的效应；也可以说是一种极端遗传漂变的结果，这种结果是由为数不多的几个个体建立一个新居群时产生的。

What are the genetic problems in small populations?

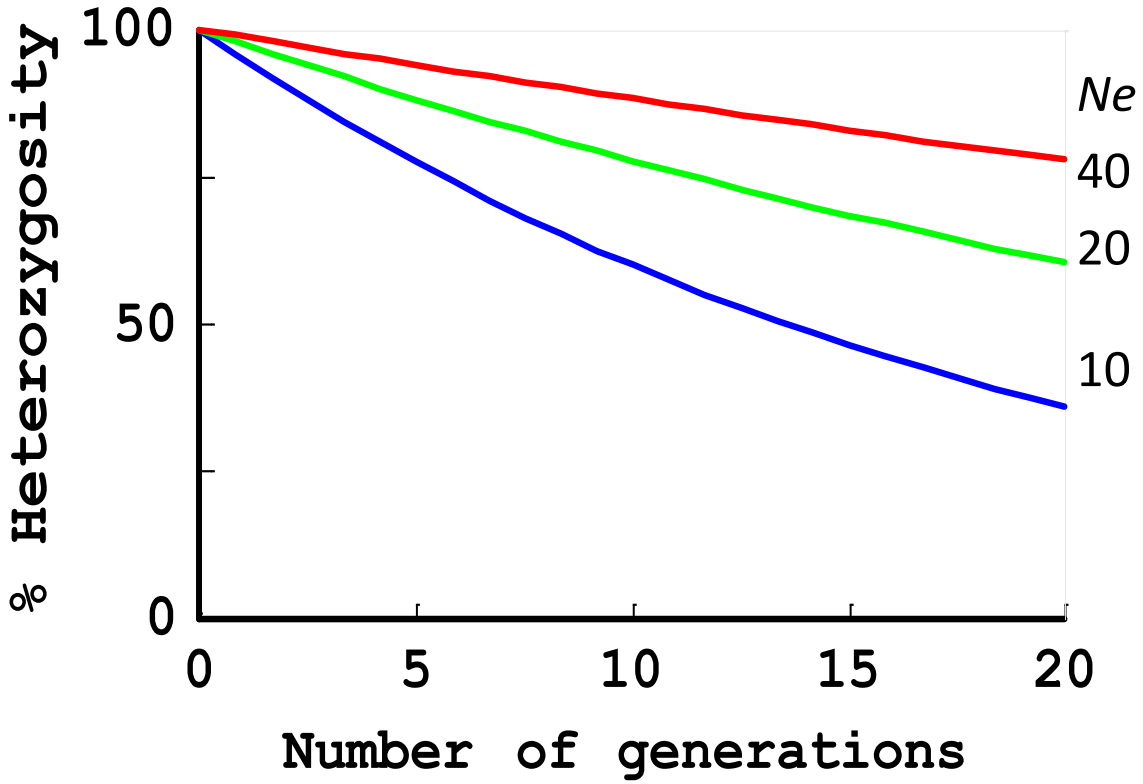
- Inbreeding and inbreeding depression
- Genetic drift and the loss of genetic variation
- Accumulation of deleterious mutations and loss of fitness

Inbreeding in small populations

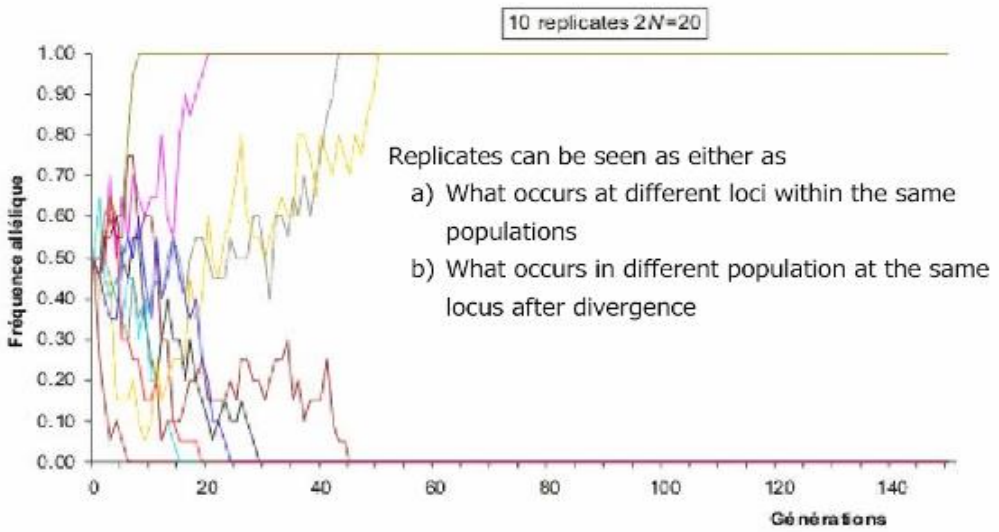
- **Unavoidable**
  - Relatives meet by chance
- **Inbreeding**
  - Increases over time
  - Simultaneous loss of heterozygosity
- **Inbreeding depression**
  - Recessive deleterious alleles may become homozygous



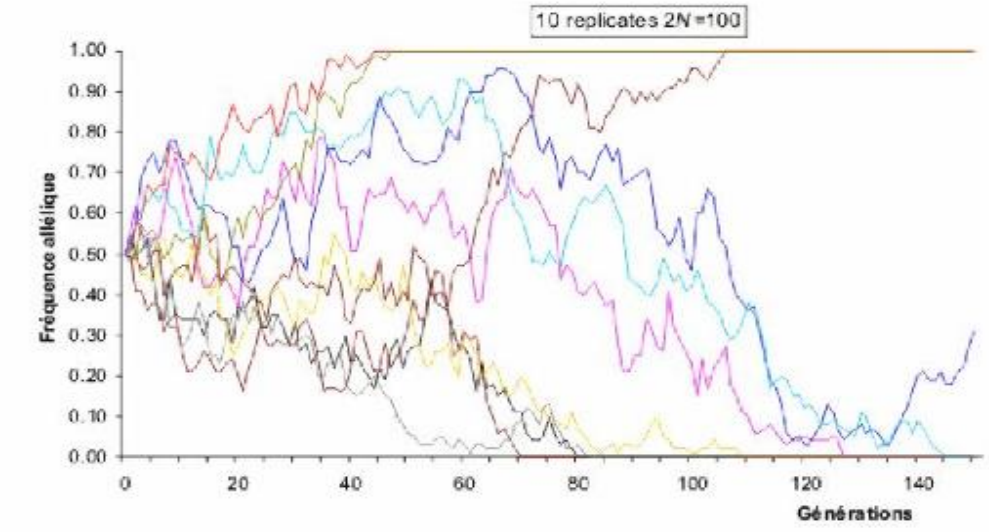
Genetic drift can reduce the genetic variation in small populations



Random fixation of an allele in small populations



In small populations, alleles can be rapidly lost by genetic drift



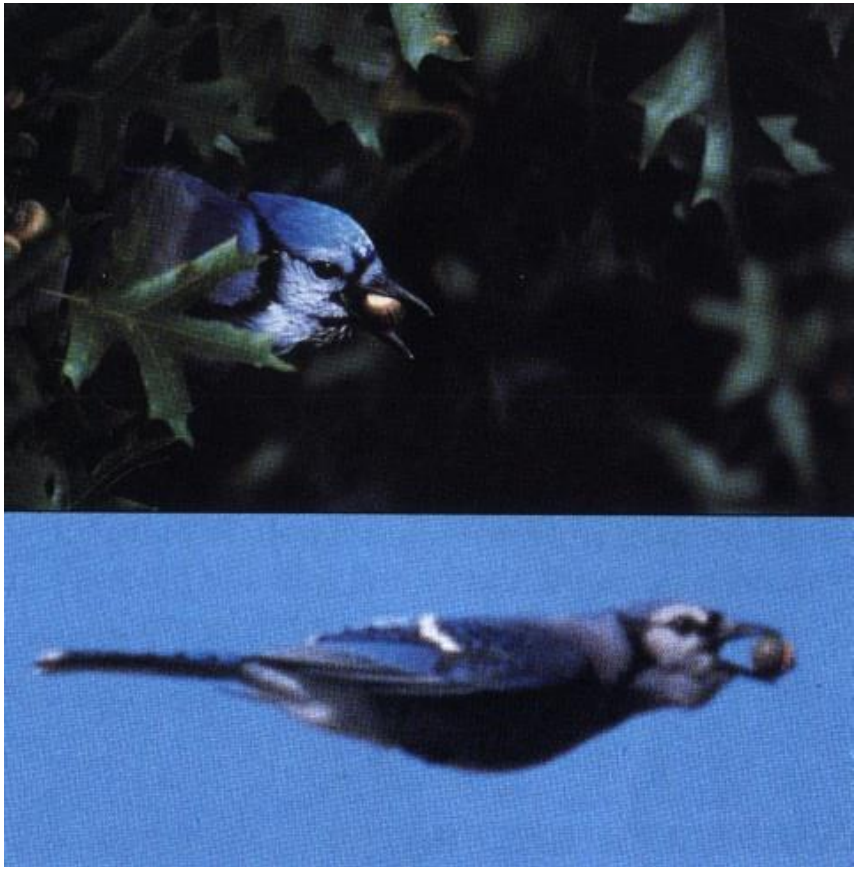
Random Mating 随机交配

随机交配是维持居群基因频率（即Hardy-Weinberg 平衡）不变的必要条件。其实在生物（植物或动物）居群中真正的随机交配是不存在的。

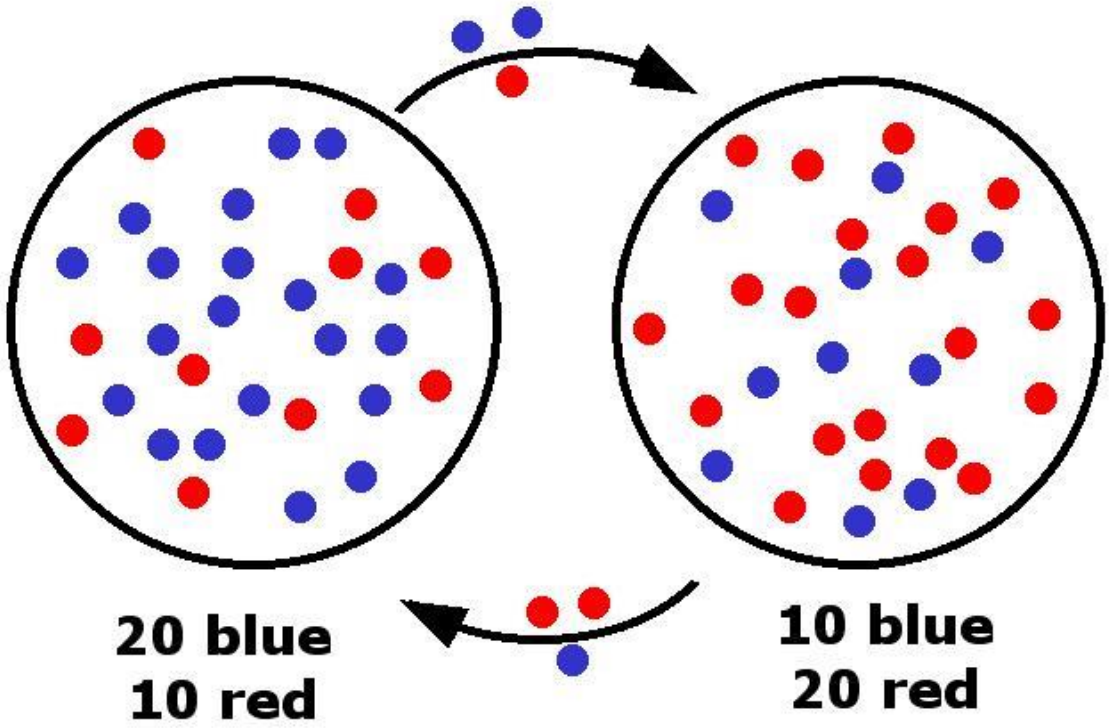
例如，植物居群中传粉媒介活动能力和范围的限制、自交不亲和等。

Gene Flow/Migration and Mutation

基因流/迁移和突变



Gene flow



Natural Selection

Individuals vary in their ability to survive and reproduce in a given population

- Results from differences in mortality (survivorship) as well as differences in fecundity (number of offspring).
- Some individuals produce more offspring than others (due at least in part to their genetics).
- More offspring means more of that individual's genes passed on to future generations.

Modes of Natural Selection
自然选择的类型

- 定向性选择 directional selection

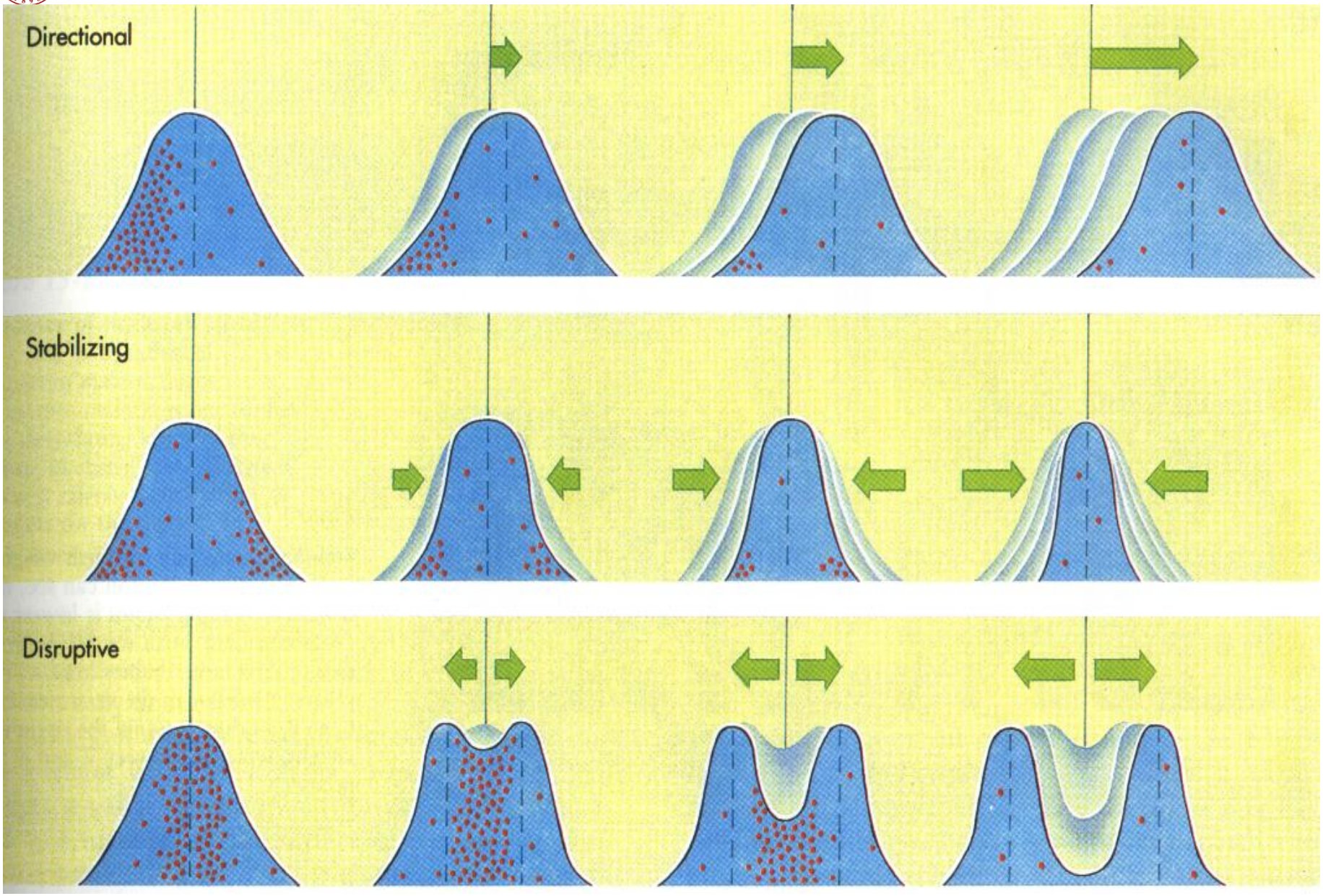
  one phenotype is the fittest in a population

- 稳定性选择 stabilizing selection

  an intermediate phenotype is the fittest in a population

- 歧化选择/分离选择 disruptive selection

  two or more phenotypes are fitter than the intermediates between them in a population


Directional
Stabilizing
Disruptive

The power of artificial selection

Brasssica oleracea

Natural Selection does not produce perfection

- Adaptation limited by historical and developmental constraints.
- Selection can only 'edit' existing variations.

Questions and Discussion

1. Evolutionary unit ?
2. Nature of the *Population* ?
3. What does *Hardy-Weinberg Equilibrium* mean ?
4. What's the main evolutionary forces at *Micro-* and *Macro-evolution*?
5. What is the unit of *Natural selection*? Is it the same as that of *Evolution??*
6. When faced with a change in environmental condition, what will or can *A Species* do?
7. Could a kind of *Selection* make things perfect? Why won't or will our lungs evolve to deal with air pollution?