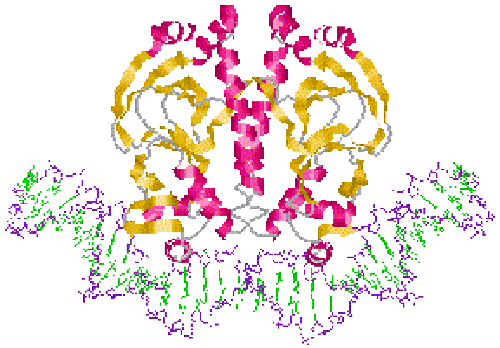
Population and evolutionary genetics



Prof. Dr. İsmail AKYOL
Prof. Dr. M. Ali YILDIZ
Prof. Dr. M. Muhip ÖZKAN
Ankara Üniversitesi

Outline of course

- Most populations and species harbor considerable genetic variation.
- This variation is reflected in the alleles distributed among populations of a species.
- The relationship between allele frequencies and genotype frequencies in an ideal population is described by the Hardy–Weinberg law.
- Selection, migration, and genetic drift can cause changes in allele frequency.
- Mutation creates new alleles in a population gene pool.
- Nonrandom mating changes population genotype frequency but not allele frequency.
- A reduction in gene flow between populations, accompanied by selection or genetic drift, can lead to reproductive isolation and speciation.
- Genetic differences between populations or species are used to reconstruct evolutionary history.

Genetic Variation Is Present in Most Populations and Species

FIGURE 26.1 The size difference between a Chihuahua and a Great Dane illustrates the high degree of genetic variation present in the dog genome.

Genetic Variation Is Present in Most Populations and Species

	Exon 3	Intron 3	Exon 4
Consensus *Adh* sequence:	C C C C	G G A A T	C T C C A* C T A G
Strain			
Wa-S	T T • A	C A • T A	A C • • • • • • •
Fl1-S	T T • A	C A • T A	A C • • • • • • •
Ja-S	• • • •	• • • • •	• • • T • T • C A
Fl-F	• • • •	• • • • •	• • G T C T C C •
Ja-F	• • A •	• • G • •	• • G T C T C C •

FIGURE 26.2 DNA sequence variation in parts of the *Drosophila Adh* gene in a sample of the 11 laboratory strains derived from the five natural populations. The dots represent nucleotides that are the same as the consensus sequence; letters represent nucleotide polymorphisms. An A/C polymorphism (A*) in codon 192 creates the two *Adh* alleles (F and S). All other polymorphisms are silent or noncoding.

The Hardy–Weinberg Law Describes Allele Frequencies and Genotype Frequencies in Population Gene Pools.

	Sperm	
	fr(A) = 0.7	fr(a) = 0.3
Eggs fr(A) = 0.7	fr(AA) = 0.7×0.7 = 0.49	fr(Aa) = 0.7×0.3 = 0.21
Eggs fr(a) = 0.3	fr(aA) = 0.3×0.7 = 0.21	fr(aa) = 0.3×0.3 = 0.09

FIGURE 26.3 Calculating genotype frequencies from allele frequencies. Gametes represent samples drawn from the gene pool to form the genotypes of the next generation. In this population, the frequency of the *A* allele is 0.7, and the frequency of the *a* allele is 0.3. The frequencies of the genotypes in the next generation are calculated as 0.49 for *AA*, 0.42 for *Aa*, and 0.09 for *aa*. Under the Hardy–Weinberg law, the frequencies of *A* and *a* remain constant from generation to generation.

The Hardy–Weinberg Law and Its Assumptions

		Sperm	
		$fr(A) = p$	$fr(a) = q$
Eggs	$fr(A) = p$	$fr(AA) = p^2$	$fr(Aa) = pq$
	$fr(a) = q$	$fr(aA) = qp$	$fr(aa) = q^2$

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

Testing for Hardy–Weinberg Equilibrium in a Population

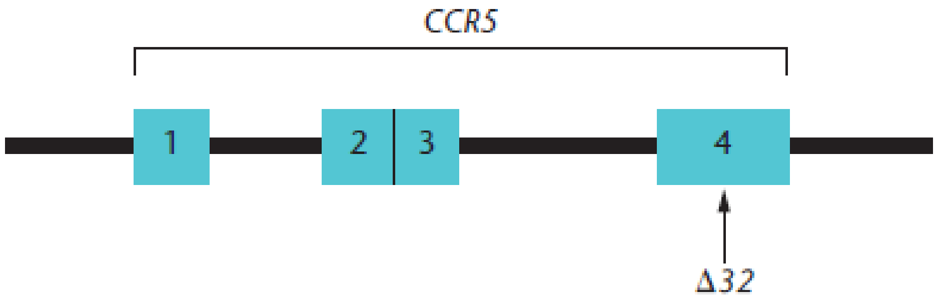


FIGURE 26.5 The organization of the *CCR5* gene in region 3p21.3 of human chromosome 3. The gene contains 4 exons and 2 introns (there is no intron between exons 2 and 3). The arrow shows the location of the 32-bp deletion in exon 4 that confers resistance to HIV-1 infection.

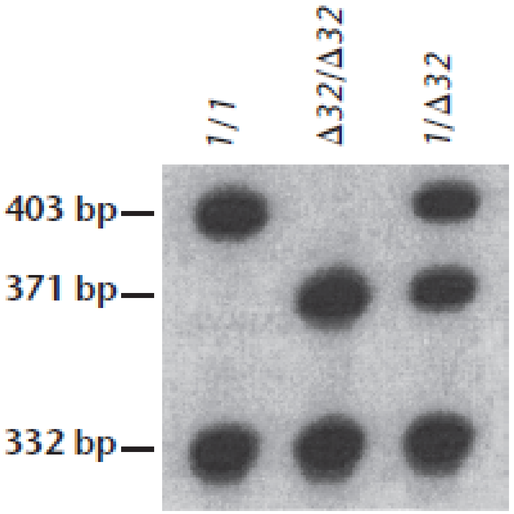


FIGURE 26.6 Allelic variation in the *CCR5* gene. Michel Samson and colleagues used polymerase chain reaction (PCR) to amplify a part of the *CCR5* gene containing the site of the 32-bp deletion, cut the resulting DNA fragments with a restriction enzyme, and ran the fragments on an electrophoresis gel. Each lane reveals the genotype of a single individual. The *1* allele produces a 332-bp fragment and a 403-bp fragment; the *Δ32* allele produces a 332-bp fragment and a 371-bp fragment. Heterozygotes produce three bands.

Testing for Hardy–Weinberg Equilibrium in a Population

TABLE 26.2 Methods of Determining Allele Frequencies from Data on Genotypes

(a) Counting Alleles	Genotype *1/1*	*1/Δ32*	*Δ32/Δ32*	Total
Number of individuals	79	20	1	100
Number of *1* alleles	158	20	0	178
Number of *Δ32* alleles	0	20	2	22
Total number of alleles	158	40	2	200

Frequency of *CCR5-1* in sample: 178/200 = 0.89 = 89%

Frequency of *CCR5-Δ32* in sample: 22/200 = 0.11 = 11%

(b) From Genotype Frequencies	Genotype *1/1*	*1/Δ32*	*Δ32/Δ32*	Total
Number of individuals	79	20	1	100
Genotype frequency	79/100 = 0.79	20/100 = 0.20	1/100 = 0.01	1.00

Frequency of CCR5-1 in sample: 0.79 + (0.5)0.20 = 0.89 = 89%

Frequency of *CCR5-Δ32* in sample: (0.5)0.20 + 0.01 = 0.11 = 11%

Calculating Frequencies for Multiple Alleles in Populations

$$p + q + r = 1$$

$$(p + q + r)^2 = p^2 + q^2 + r^2 + 2pq + 2pr + 2qr = 1$$

TABLE 26.3 Calculating Genotype Frequencies for Multiple Alleles in a Hardy–Weinberg Population Where the Frequency of Allele $I^A = 0.38$, Allele $I^B = 0.11$, and Allele $i = 0.51$

Genotype	Genotype Frequency	Phenotype	Phenotype Frequency
I^AI^A	$p^2 = (0.38)^2 = 0.14$	A	0.53
I^Ai	$2pr = 2(0.38)(0.51) = 0.39$		
I^BI^B	$q^2 = (0.11)^2 = 0.01$	B	0.12
I^Bi	$2qr = 2(0.11)(0.51) = 0.11$		
I^AI^B	$2pr = 2(0.38)(0.11) = 0.084$	AB	0.08
ii	$r^2 = (0.51)^2 = 0.26$	O	0.26

Calculating Heterozygote Frequency

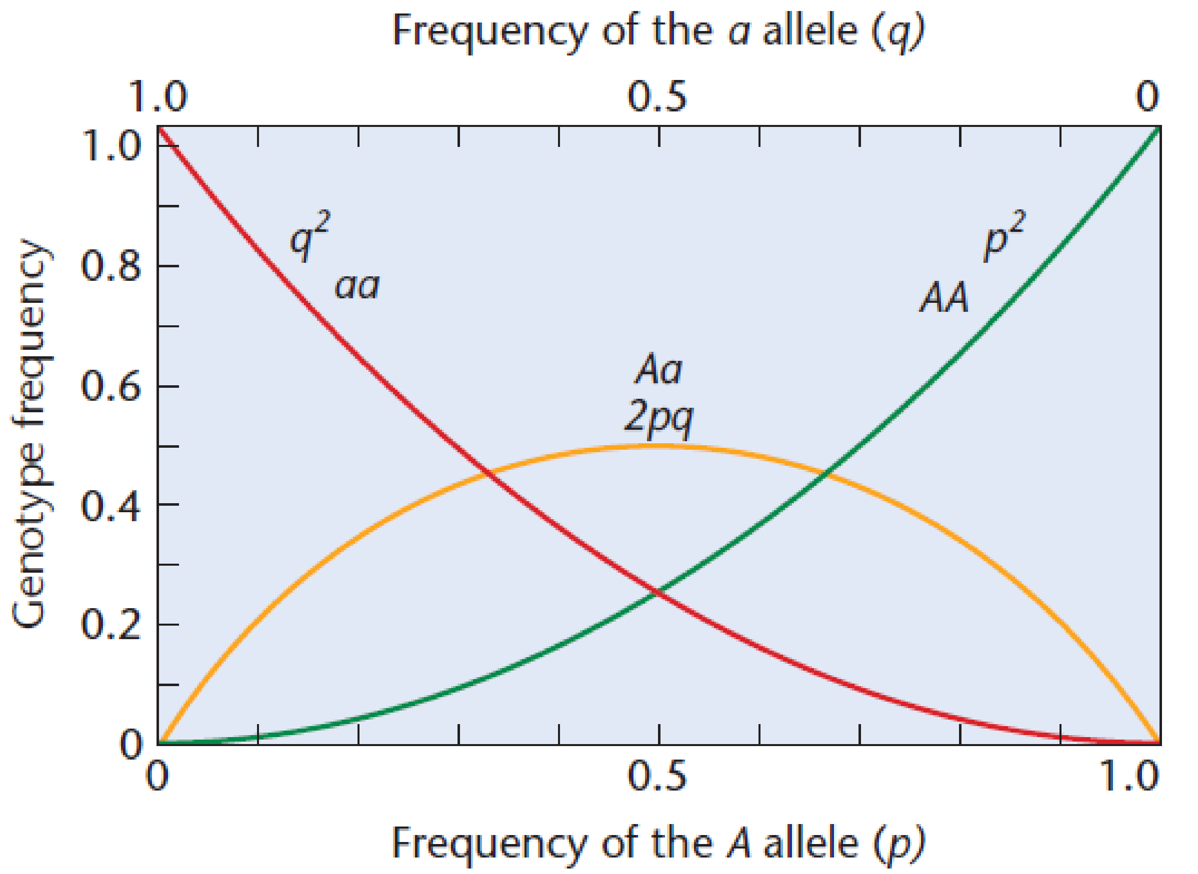


FIGURE 26.7 The relationship between genotype and allele frequencies derived from the Hardy–Weinberg equation.