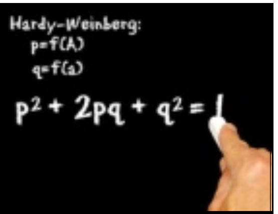


WHY

Population genetics and Hardy-Weinberg matter

Evolution

In one of the last classes we have covered:

What is the evidence for evolution?

Darwin observations

Evolution

In this class we will cover:

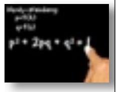
The Hardy-Weinberg Equilibrium

- Hardy–Weinberg and its assumptions
- Each assumption is a population genetic process
- How to evaluate the HWE in a sample
- What are the main factors that affect HWE

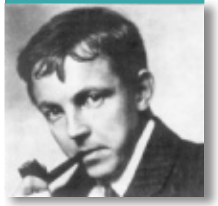
Genetic variability • Composição Genética • Hardy-Weinberg

HWE: establishes the relationship that can be used to predict *allele frequencies* given *genotype frequencies* or predict *genotype frequencies* given *allele frequencies*

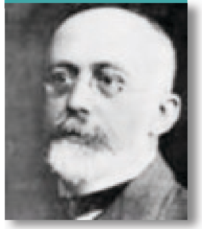
Genetic variability • Composição Genética • Hardy-Weinberg



Hardy, Godfrey Harold
(1877-1947)

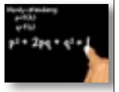


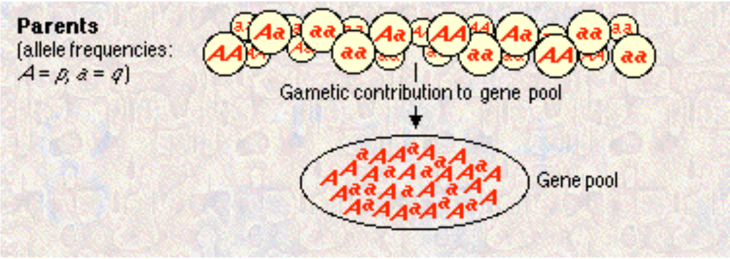
Weinberg, Wilhelm
(1862–1937)



Castle, William E.
(1930-1961)

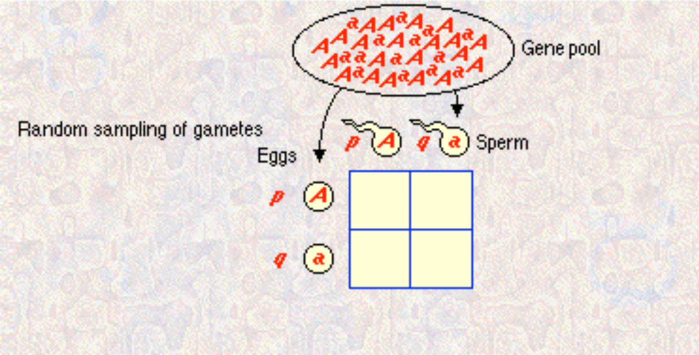
Genetic variability • Composição Genética • Hardy-Weinberg



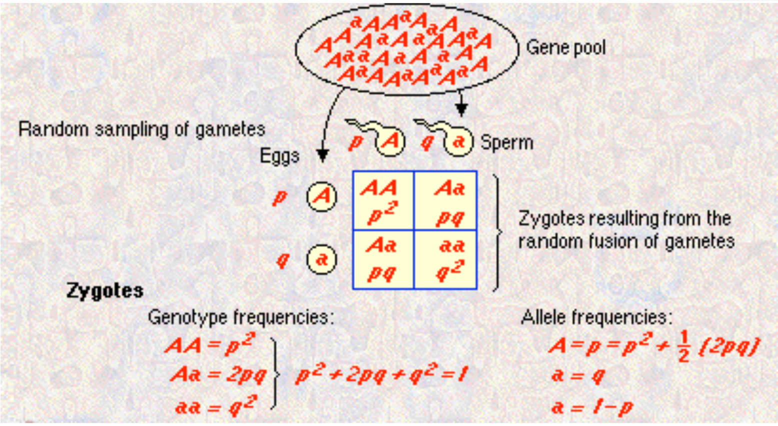


Parents
(allele frequencies: $A = p$, $a = q$)

Genetic variability • Composição Genética • Hardy-Weinberg



Genetic variability • Composição Genética • Hardy-Weinberg



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

Zygotes resulting from the random fusion of gametes

Zygotes

Genotype frequencies:

$$\left.\begin{aligned} AA &= p^2 \\ Aa &= 2pq \\ aa &= q^2 \end{aligned}\right\} p^2 + 2pq + q^2 = 1$$

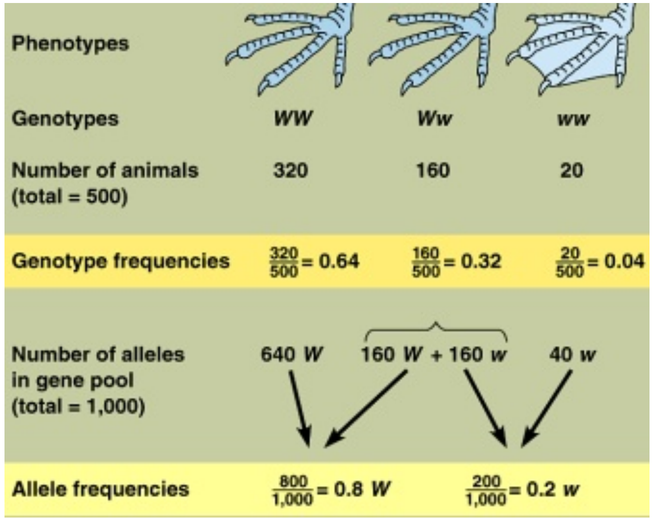
Allele frequencies:

$A = p = p^2 + \frac{1}{2}(2pq)$

$a = q$

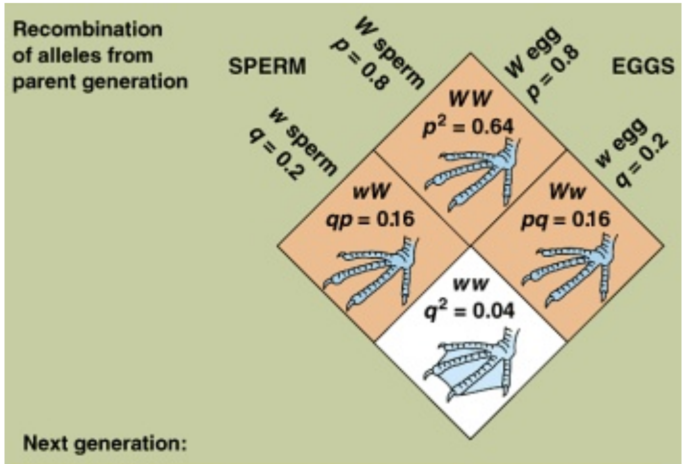
$a = 1 - p$

Genetic variability • Composição Genética • Hardy-Weinberg



Phenotypes			
Genotypes	*WW*	*Ww*	*ww*
Number of animals (total = 500)	320	160	20
Genotype frequencies	$\frac{320}{500} = 0.64$	$\frac{160}{500} = 0.32$	$\frac{20}{500} = 0.04$
Number of alleles in gene pool (total = 1,000)	640 *W*	160 *W* + 160 *w*	40 *w*
Allele frequencies	$\frac{800}{1{,}000} = 0.8\ W$		$\frac{200}{1{,}000} = 0.2\ w$

Genetic variability • Composição Genética • Hardy-Weinberg

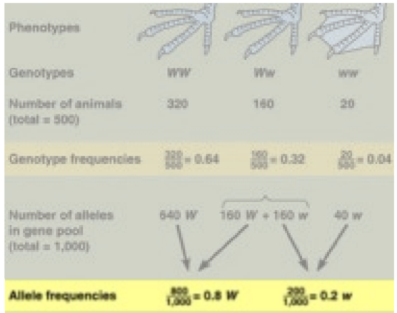


Next generation:

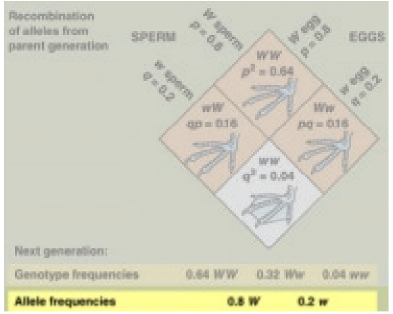
Genotype frequencies	0.64 *WW*	0.32 *Ww*	0.04 *ww*
Allele frequencies	0.8 *W*	0.2 *w*	

Genetic variability • Composição Genética • Hardy-Weinberg

First generation



Second generation



Alleles frequencies did not change!

Genetic variability • Composição Genética • Hardy-Weinberg

A single generation of reproduction where a set of conditions, or assumptions, are met will result in a population that meets Hardy–Weinberg expected genotype frequencies

Se p e q forem as frequências alélicas de **A** e **B** numa geração, as frequências genotípicas na geração seguinte serão:

p^2	q^2	$2pq$
$f(AA)$	$f(BB)$	$f(AB)$

[expansão do binómio $(p+q)^2$]

Para uma população com 3 alelos temos que a relação entre os dois tipos de frequências é descrita pelo polinómio: $(p+q+r)^2$:

AA	*AC*	*CC*	BB	AB	CB
p^2	$2pq$	q^2	r^2	$2pr$	$2qr$

- População modelo?

A reprodução é sexual, envolve gâmetas de tipos distintos, mas cuja contribuição em termos de sexos é igual (isto é a frequência de um alelo é igual nos dois sexos);

As gerações não se sobrepõem;

Os cruzamentos são casuais;

A população é de grandes dimensões;

A migração é negligível;

Não há mutação;

A selecção natural não afecta os alelos em questão.

Pressupostos	**Razão**
População grande	Muitas possibilidade de combinações alélicas
Sem selecção natural	Não há restrição nos cruzamentos pelo que todos os alelos têm igual probabilidade de integrar a geração seguinte
Sem mutação	Não há introdução de novos alelos
Sem migração	Não há entrada ou saída de indivíduos
Cruzamentos ao acaso	Qualquer combinação de alelos é igualmente provável.

Nenhum destes pressupostos é realista....

IMPORTANTE

Descreve a relação, numa população modelo, entre as frequências alélicas numa geração e as frequências genotípicas da geração seguinte: sendo f(A) e f(B) numa geração, as frequências genotípicas da geração seguinte serão

$f(AA)= f(A)^2$; $f(AB)= 2\,f(A)\,f(B)$ e $f(BB)= f(B)^2$

IMPORTANTE

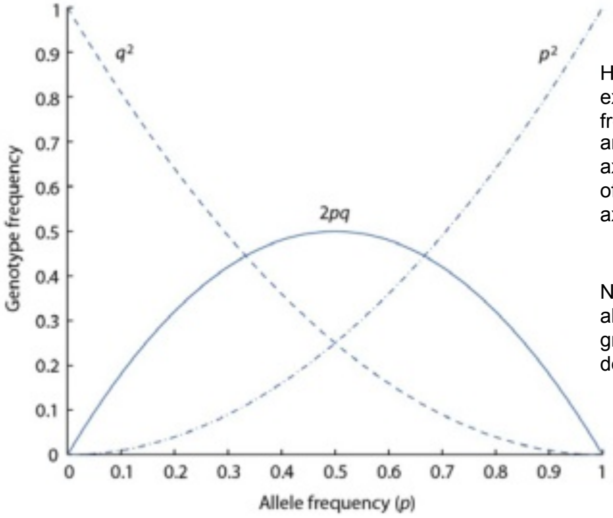
Descreve a relação, numa população modelo, entre as frequências alélicas numa geração e as frequências genotípicas da geração seguinte: sendo f(A) e f(B) numa geração, as frequências genotípicas da geração seguinte serão

$f(AA)= f(A)^2$; $f(AB)= 2\,f(A)\,f(B)$ e $f(BB)= f(B)^2$

As frequências alélicas entre gerações não se alteram.

Esta relação estabelece-se de uma geração para a seguinte e perdura enquanto as condições da população modelo se mantiverem.

De Finetti diagram



Hardy–Weinberg expected genotype frequencies for *AA*, *Aa*, and *aa* genotypes (*y* axis) for any given value of the allele frequency (*x* axis).

Note that the value of the allele frequency not graphed can be determined by $q = 1 - p$.

What good is a model with so many restrictive assumptions?

Are all these assumptions likely to be met in actual populations?

The Hardy–Weinberg model is not necessarily meant to be an exact description of any actual population…

although actual populations often exhibit genotype frequencies predicted by Hardy– Weinberg

---

So….

X Mutation

X Drift

X Selection

X Migration

X Mutation

X Drift

X Selection

X Migration

First generation

↓

Second generation

Alleles frequencies did not change!

~~Alleles frequencies did not change!~~

First generation

↓

Second generation

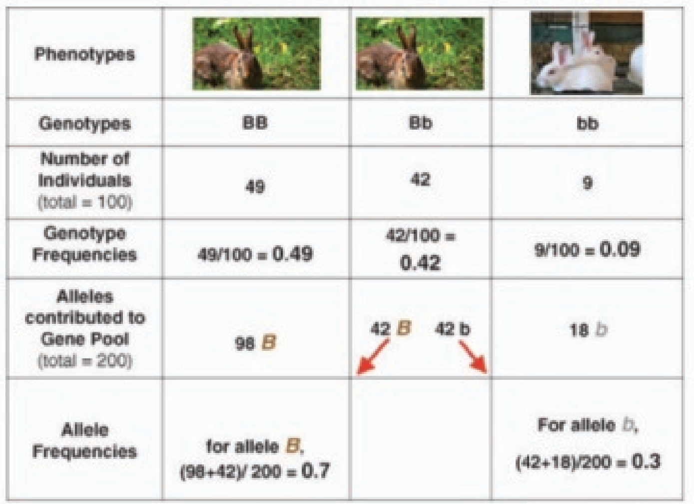
~~Alleles frequencies did not change!~~

First generation

↓

Second generation

- ? Mutation
- ? Drift
- ? Selection
- ? Migration



Phenotypes			
Genotypes	BB	Bb	bb
Number of Individuals (total = 100)	49	42	9
Genotype Frequencies	49/100 = 0.49	42/100 = 0.42	9/100 = 0.09
Alleles contributed to Gene Pool (total = 200)	98 B	42 B 42 b	18 b
Allele Frequencies	for allele B, (98+42)/ 200 = 0.7		For allele b, (42+18)/200 = 0.3

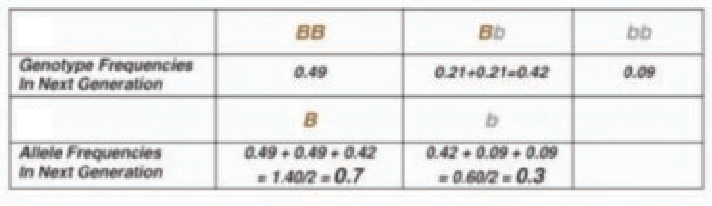
Punnett square

Probability of Alleles in Gametes: Sperm and Eggs	0.7 B	0.3 b
0.7 B	BB 0.7 X 0.7 = 0.49	Bb 0.7 X 0.3 = 0.21
0.3 b	Bb 0.7 X 0.3= 0.21	bb 0.3 X 0.3 = 0.09

Genetic variability • Composição Genética • Hardy-Weinberg

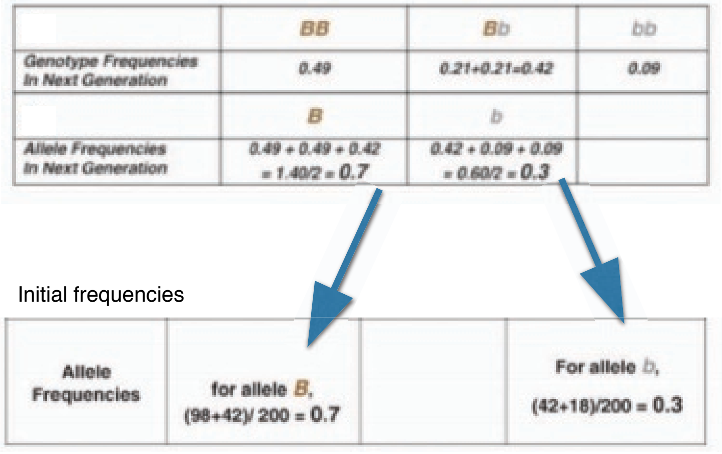
A	BB	Bb	bb
Genotype Frequencies In Next Generation	0.49	0.21+0.21=0.42	0.09

Genetic variability • Composição Genética • Hardy-Weinberg



	BB	Bb	bb
Genotype Frequencies In Next Generation	0.49	0.21+0.21=0.42	0.09
	B	b	
Allele Frequencies In Next Generation	0.49 + 0.49 + 0.42 = 1.40/2 = 0.7	0.42 + 0.09 + 0.09 = 0.60/2 = 0.3	

Genetic variability • Composição Genética • Hardy-Weinberg



Final frequencies

	BB	Bb	bb
Genotype Frequencies In Next Generation	0.49	0.21+0.21=0.42	0.09
	B	b	
Allele Frequencies In Next Generation	0.49 + 0.49 + 0.42 = 1.40/2 = 0.7	0.42 + 0.09 + 0.09 = 0.60/2 = 0.3	

Initial frequencies

Allele Frequencies	for allele B, (98+42)/ 200 = 0.7		For allele b, (42+18)/200 = 0.3

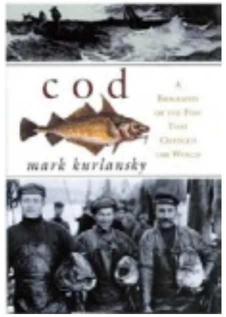
EXAMPLES

Does Hardy-Weinberg equilibrium ever exist in nature?

Example: Atlantic cod (*Gadus morhua*) in Nova Scotia

as a juvenile…

… and as an adult



EXAMPLES

Does Hardy-Weinberg equilibrium ever exist in nature?

Example: Atlantic cod (*Gadus morhua*) in Nova Scotia

A sample of 364 fish were scored for a single nucleotide locus

A1A1 = 109
A1A2 = 182
A2A2 = 73

EXAMPLES

A_1A_1 = 109
A_1A_2 = 182
A_2A_2 = 73

Total: 109+182+73=364

Passo 1: Calculem as frequências genotípicas

Frequência de A_1A_1

Frequência de A_1A_2

Frequência de A_2A_2

EXAMPLES

A_1A_1 = 109
A_1A_2 = 182
A_2A_2 = 73

Passo 1: Calculem as frequências genotípicas

Frequency de A_1A_1 = 109/364 = 0.2995

Frequency de A_1A_2 = 182/364 = 0.5000

Frequency de A_2A_2 = 73/364 = 0.2005

$f(A_1A_1) = 0.2995$
$f(A_1A_2) = 0.5000$
$f(A_2A_2) = 0.2005$

EXAMPLES

$f(A_1A_1) = 0.2995$
$f(A_1A_2) = 0.5000$
$f(A_2A_2) = 0.2005$

Passo 2: Calculem as frequências alélicas

Frequency de A_1

Frequency de A_2

EXAMPLES

$f(A_1A_1) = 0.2995$
$f(A_1A_2) = 0.5000$
$f(A_2A_2) = 0.2005$

Passo 2: Calculem as frequências alélicas

Frequency of A_1 = p = Freq (A_1A_1) + ½ Freq (A_1A_2)

Frequency of A_2 = q = Freq (A_2A_2) + ½ Freq (A_1A_2)

EXAMPLES

$f(A_1A_1) = 0.2995$
$f(A_1A_2) = 0.5000$
$f(A_2A_2) = 0.2005$

Passo 2: Calculem as frequências alélicas

Frequency of A_1 = p = Freq (A_1A_1) + ½ Freq (A_1A_2)
= 0.2995 + ½ (0.5000)
= 0.5495

EXAMPLES

$f(A_1A_1) = 0.2995$
$f(A_1A_2) = 0.5000$
$f(A_2A_2) = 0.2005$

Passo 2: Calculem as frequências alélicas

Frequency of A_1 = p = Freq (A_1A_1) + ½ Freq (A_1A_2)
= 0.2995 + ½ (0.5000)
= 0.5495

Frequency of A_2 = q = Freq (A_2A_2) + ½ Freq (A_1A_2)
= 0.2005 + ½ (0.5000)
= 0.4505

Check that p + q = 0.5495 + 0.4505 = 1

EXAMPLES

$f(A_1) = 0.5495$
$f(A_2) = 0.4505$

Passo 3: Estimar as frequências genotípicas em EHW

Expected No. of A_1A_1 = p_2 x N

Expected No. of A_1A_2 = 2pq x N

Expected No. of A_2A_2 = q_2 x N

EXAMPLES

$f(A_1) = 0.5495$
$f(A_2) = 0.4505$

Passo 3: Estimar as frequências genotípicas em EHW

Expected No. of $A_1A_1 = p^2 \times N$
$= (0.5495)^2 \times 364$
$= 109.9$

Expected No. of $A_1A_2 = 2pq \times N$
$= 2(0.5495)(0.4505) \times 364$
$= 180.2$

Expected No. of $A_2A_2 = q^2 \times N$
$= (0.4595)^2 \times 364$
$= 73.9$

EXAMPLES

$Nb_0(A_1A_1) = 109$
$Nb_0(A_1A_2) = 182$
$Nb_0(A_2A_2) = 73$

$Nb_E(A_1A_1) = 109.9$
$Nb_E(A_1A_2) = 180.2$
$Nb_E(A_2A_2) = 73.9$

Passo 4: Comparar frequências genotípicas observadas e esperadas em EHW

Genotype	Observed	Expected
A_1A_1	109	109.9
A_1A_2	182	180.2
A_2A_2	73	73.9

EXAMPLES

$Nb_0(A_1A_1) = 109$
$Nb_0(A_1A_2) = 182$
$Nb_0(A_2A_2) = 73$

$Nb_E(A_1A_1) = 109.9$
$Nb_E(A_1A_2) = 180.2$
$Nb_E(A_2A_2) = 73.9$

Passo 4: Comparar frequências genotípicas observadas e esperadas em EHW

Genotype	Observed	Expected
A_1A_1	109	109.9
A_1A_2	182	180.2
A_2A_2	73	73.9

$$\chi^2\text{ (calc)} = \Sigma \frac{(\text{Obs.} - \text{Exp.})^2}{\text{Exp.}} = 0.036$$

Graus de liberdade = número de classes (AA, AB,BB) - 1 - número de parâmetros estimados (p,q)= 1

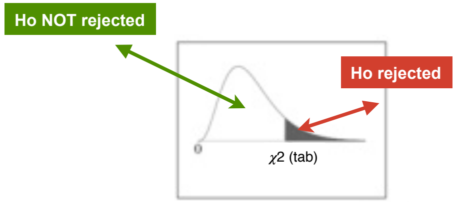
EXAMPLES

Hipótese nula H_0:

as proporções genotípicas observadas *não* diferem das proporções esperadas de acordo com a lei de HW

EXAMPLES

χ2 e probabilidade



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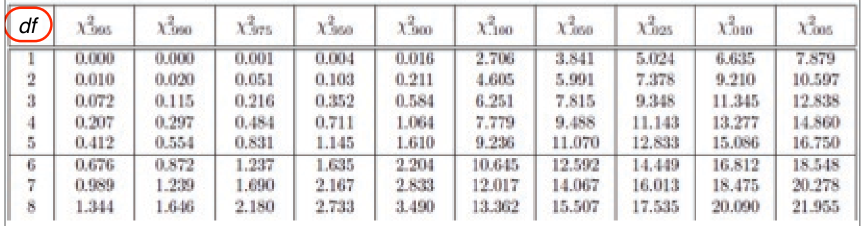
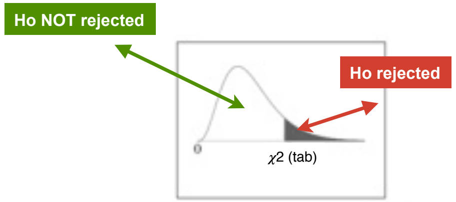
Ho rejected



df	$\chi^2_{.995}$	$\chi^2_{.990}$	$\chi^2_{.975}$	$\chi^2_{.950}$	$\chi^2_{.900}$	$\chi^2_{.100}$	$\chi^2_{.050}$	$\chi^2_{.025}$	$\chi^2_{.010}$	$\chi^2_{.005}$
1	0.000	0.000	0.001	0.004	0.016	2.706	3.841	5.024	6.635	7.879
2	0.010	0.020	0.051	0.103	0.211	4.605	5.991	7.378	9.210	10.597
3	0.072	0.115	0.216	0.352	0.584	6.251	7.815	9.348	11.345	12.838
4	0.207	0.297	0.484	0.711	1.064	7.779	9.488	11.143	13.277	14.860
5	0.412	0.554	0.831	1.145	1.610	9.236	11.070	12.833	15.086	16.750
6	0.676	0.872	1.237	1.635	2.204	10.645	12.592	14.449	16.812	18.548
7	0.989	1.239	1.690	2.167	2.833	12.017	14.067	16.013	18.475	20.278
8	1.344	1.646	2.180	2.733	3.490	13.362	15.507	17.535	20.090	21.955

EXAMPLES

χ2 e probabilidade



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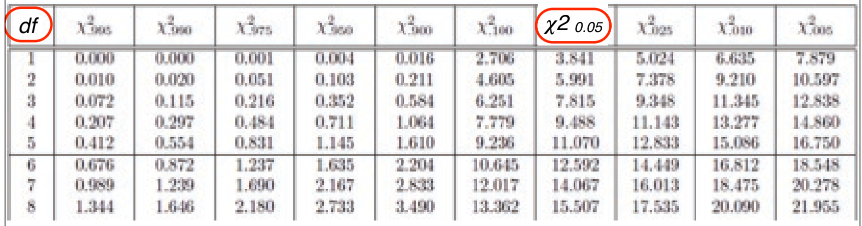

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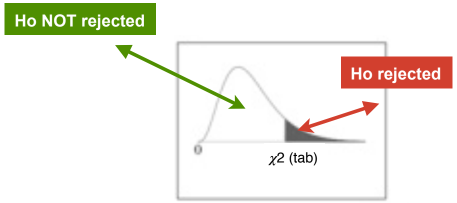
8	1.344	1.646	2.180	2.733	3.490	13.362	15.507	17.535	20.090	21.955

EXAMPLES

χ2 e probabilidade



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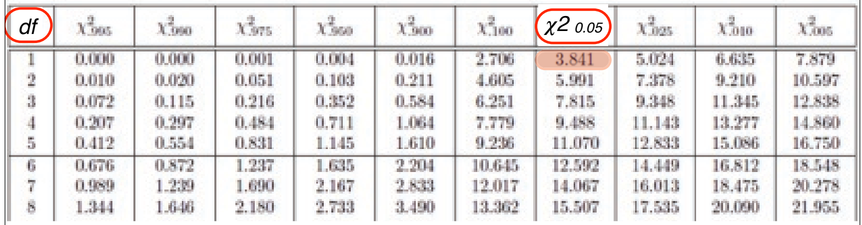

df	$\chi^2_{.995}$	$\chi^2_{.990}$	$\chi^2_{.975}$	$\chi^2_{.950}$	$\chi^2_{.900}$	$\chi^2_{.100}$	$\chi2_{0.05}$	$\chi^2_{.025}$	$\chi^2_{.010}$	$\chi^2_{.005}$
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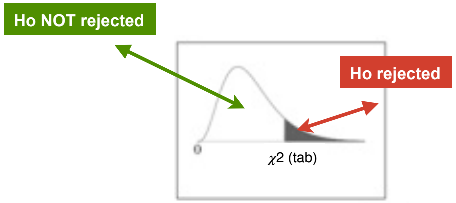
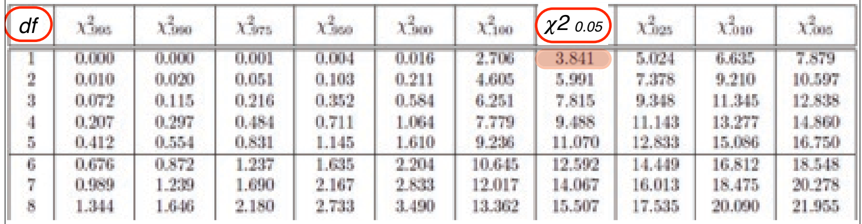
EXAMPLES

χ2 e probabilidade



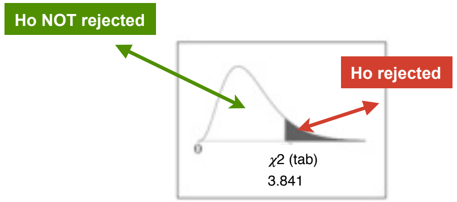
Ho NOT rejected

Ho rejected



df	$\chi^2_{.995}$	$\chi^2_{.990}$	$\chi^2_{.975}$	$\chi^2_{.950}$	$\chi^2_{.900}$	$\chi^2_{.100}$	$\chi2_{0.05}$	$\chi^2_{.025}$	$\chi^2_{.010}$	$\chi^2_{.005}$
1	0.000	0.000	0.001	0.004	0.016	2.706	3.841	5.024	6.635	7.879
2	0.010	0.020	0.051	0.103	0.211	4.605	5.991	7.378	9.210	10.597
3	0.072	0.115	0.216	0.352	0.584	6.251	7.815	9.348	11.345	12.838
4	0.207	0.297	0.484	0.711	1.064	7.779	9.488	11.143	13.277	14.860
5	0.412	0.554	0.831	1.145	1.610	9.236	11.070	12.833	15.086	16.750
6	0.676	0.872	1.237	1.635	2.204	10.645	12.592	14.449	16.812	18.548
7	0.989	1.239	1.690	2.167	2.833	12.017	14.067	16.013	18.475	20.278
8	1.344	1.646	2.180	2.733	3.490	13.362	15.507	17.535	20.090	21.955

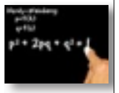
Conclusion ??

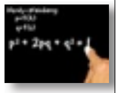


$\chi 2_{\text{(calc)}} < \chi 2_{\text{(tab)}}$ = null hypothesis NOT rejected

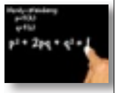
$\chi 2_{\text{(calc)}} > \chi 2_{\text{(tab)}}$ = null hypothesis rejected

---



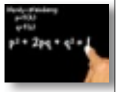


AA = 320
AB = 570
BB = 320
SUM= 1210



AA = 320
AB = 570
BB = 320
SUM= 1210

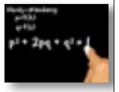
f(AA) = 320/1210
f(AB) = 570/1210
f(BB) = 320/1210



AA = 320
AB = 570
BB = 320
SUM= 1210

f(AA) = 150/800
f(AB) = 500/800
f(BB) = 150/800

f(AA) = 0.25
f(AB) = 0.50
f(BB) = 0.25



f(AA) = 0.25
f(AB) = 0.50
f(BB) = 0.25

f(A) =
f(B) =

f(AA) = 0.25
f(AB) = 0.50
f(BB) = 0.25

f(A) = (320+570/2)/1210 Contagem de indivíduos
f(B) = (640+570)/2420 Contagem de alelos

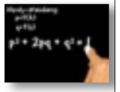
f(A) = 0.25+0.50/2 = 0.50
f(B) = 0.25+0.50/2 = 0.50

A partir das frequências

f(AA) = 0.1875
f(AB) = 0.625
f(BB) = 0.188

f(A) = 0.5
f(B) = 0.5

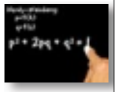
Genetic variability • Composição Genética • **Hardy-Weinberg**



f(AA) = 0.25 f(A) = 0.5
f(AB) = 0.50 f(B) = 0.5
f(BB) = 0.25

	Obs	Esp
AA	320	0.5^2*1210 = 302.5
AB	570	2*0.5*0.5*1210 = 605
BB	320	0.5^2*1210 = 302.5

Genetic variability • Composição Genética • **Hardy-Weinberg**



	Obs	Esp
AA	320	0.5^2*1210 = 302.5
AB	570	2*0.5*0.5*1210 = 605
BB	320	0.5^2*1210 = 302.5

$$\chi^2 = \Sigma \frac{(\text{Obs.} - \text{Exp.})^2}{\text{Exp.}}$$

$$\chi^2 = \Sigma \frac{(320 - 302.5)^2}{302.5} + \frac{(570 - 605)^2}{605} + \frac{(320 - 302.5)^2}{302.5}$$

$$= 1.012 + 2.025 + 1.012 = 4.05$$

Genetic variability • Composição Genética • **Hardy-Weinberg**

	Obs	Esp
AA	150	0.5^2*800 = 200
AB	500	2*0.5*0.5*800 = 400
BB	150	0.5^2*800 = 200

$\chi 2$ (calc) = 4.050
$\chi 2$ (tab) = 3.841

Conclusion?

EXAMPLES

EXAMPLES

A terrible crime has been committed.

Left at the crime scene was:

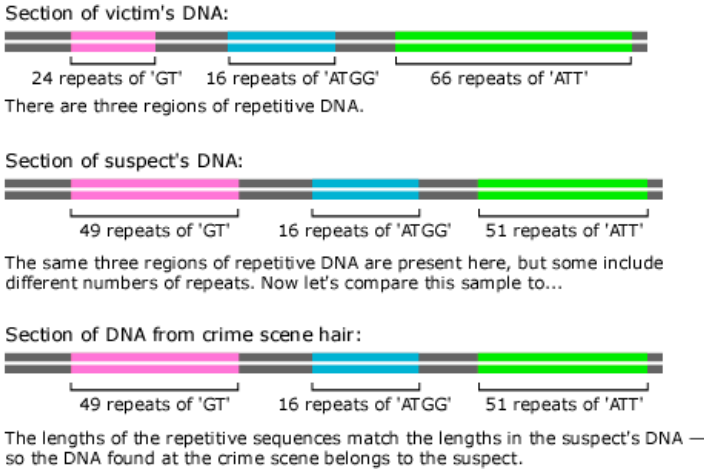
a *biological sample*

that law-enforcement authorities used to obtain a *multilocus* genotype or DNA profile.

EXAMPLES

A suspect in the crime has been identified and subpoenaed to provide a tissue sample for DNA profiling.

EXAMPLES



Section of victim's DNA:

There are three regions of repetitive DNA.

Section of suspect's DNA:

The same three regions of repetitive DNA are present here, but some include different numbers of repeats. Now let's compare this sample to...

Section of DNA from crime scene hair:

The lengths of the repetitive sequences match the lengths in the suspect's DNA — so the DNA found at the crime scene belongs to the suspect.

EXAMPLES

The DNA profiles from the suspect and from the crime scene are identical.

EXAMPLES

The DNA profiles from the suspect and from the crime scene are identical.

Can we unequivocally say the suspect is the **guilty** party?

EXAMPLES

Can anybody else share the same DNA profile?

Two matching profiles = same person??

EXAMPLES

Let’s see….

EXAMPLES

What is the probability that two individuals share the same profile??

HW prediction will help, but….

in using HW we are implicitly accepting that all of the assumptions of Hardy–Weinberg are approximately met.

EXAMPLES

Caution:

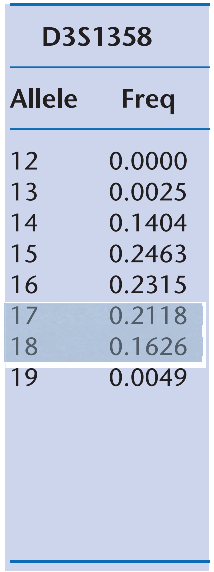
For this estimation to be done we need to assume that human populations are in HW

EXAMPLES

The DNA profile is shown in the table below.

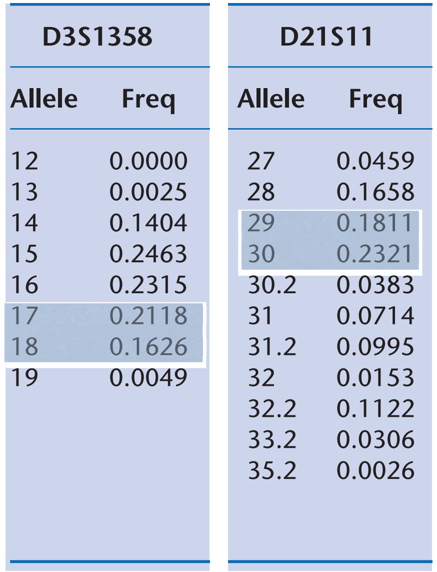
Locus	**D3S1358**	**D21S11**	**D18S51**
Genotype	17, 18	29, 30	18, 18

EXAMPLES



D3S1358	
Allele	**Freq**
12	0.0000
13	0.0025
14	0.1404
15	0.2463
16	0.2315
17	0.2118
18	0.1626
19	0.0049

EXAMPLES



D3S1358		**D21S11**	
Allele	**Freq**	**Allele**	**Freq**
12	0.0000	27	0.0459
13	0.0025	28	0.1658
14	0.1404	29	0.1811
15	0.2463	30	0.2321
16	0.2315	30.2	0.0383
17	0.2118	31	0.0714
18	0.1626	31.2	0.0995
19	0.0049	32	0.0153
		32.2	0.1122
		33.2	0.0306
		35.2	0.0026

EXAMPLES

D3S1358	
Allele	**Freq**
12	0.0000
13	0.0025
14	0.1404
15	0.2463
16	0.2315
17	0.2118
18	0.1626
19	0.0049

D21S11	
Allele	**Freq**
27	0.0459
28	0.1658
29	0.1811
30	0.2321
30.2	0.0383
31	0.0714
31.2	0.0995
32	0.0153
32.2	0.1122
33.2	0.0306
35.2	0.0026

D18S51	
Allele	**Freq**
<11	0.0128
11	0.0128
12	0.1276
13	0.1224
14	0.1735
15	0.1276
16	0.1071
17	0.1556
18	0.0918
19	0.0357
20	0.0255
21	0.0051
22	0.0026

EXAMPLES

D3S1358

17-allele frequency = 0.2118

18-allele frequency = 0.1626

Genotype frequency
= 2(0.2118)(0.1626) = 0.0689 or 6.89%

D21S11

29-allele frequency = 0.1811

30-allele frequency = 0.2321

Genotype frequency
= 2(0.1811)(0.2321) = 0.0841 or 8.41%

D18S51

18-allele frequency = 0.0918

Genotype frequency
= 0.0918^2 = 0.0084 or 0.84%

D3S1358 = 0.0689 or 6.89%

D21S11 = 0.0841 or 8.41%

D18S51 = 0.0084 or 0.84%

EXAMPLES

Product rule = the probability of observing multiple independent events is just the product of each individual event

EXAMPLES

D3S1358 = 0.0689

D21S11 = 0.0841

D18S51 = 0.0084

0.0689 X 0.0841 X 0.0084 = 0.000049 or 0.0049%

0.0049 — 100
1 — x
x= 100/0.0049

1/0.000049 = 20,408

EXAMPLES

1 in 20,408

Portugal ≃10^6 people

Probability of 500 individuals will share this genotype

NOT A RARE GENOTYPE!

EXAMPLES

Only when DNA profiles do not match, called an “*exclusion*,” can a suspect be unambiguously and absolutely excluded as the source of a biological sample at a crime scene

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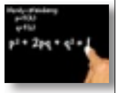
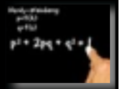
Genetic variability • Composição Genética • Hardy-Weinberg



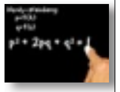
Se uma população não se encontra em EHW, então quais as razões:

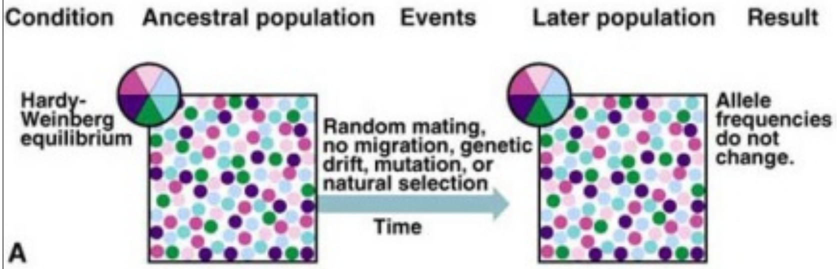
- **Selecção?**
- **Migração?**
- **Subdivisão?**
- **Deriva genética?**
- **Mutação?**

Genetic variability • Composição Genética • Hardy-Weinberg



Genetic variability • Composição Genética • Hardy-Weinberg





Condition | Ancestral population | Events | Later population | Result

Hardy-Weinberg equilibrium

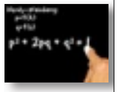
Random mating, no migration, genetic drift, mutation, or natural selection

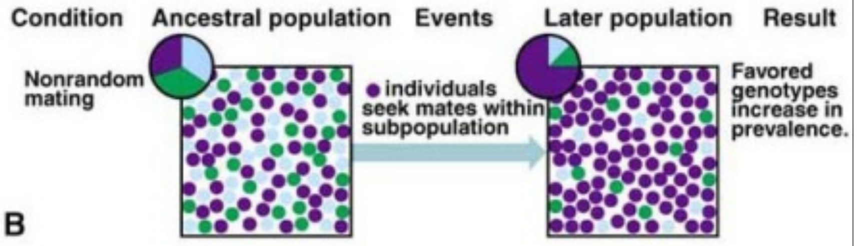
Time

Allele frequencies do not change.

A

Genetic variability • Composição Genética • Hardy-Weinberg





Condition | Ancestral population | Events | Later population | Result

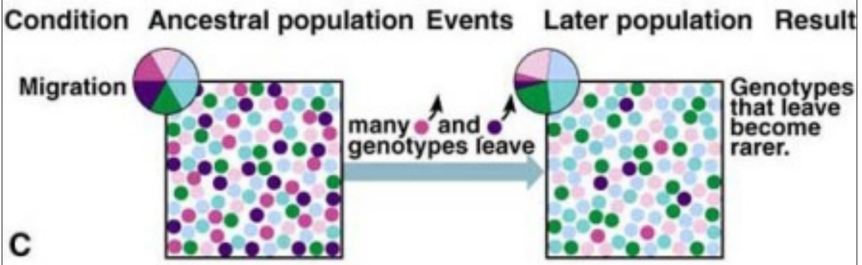
Nonrandom mating

● individuals seek mates within subpopulation

Favored genotypes increase in prevalence.

B

Genetic variability • Composição Genética • Hardy-Weinberg



Condition | Ancestral population | Events | Later population | Result

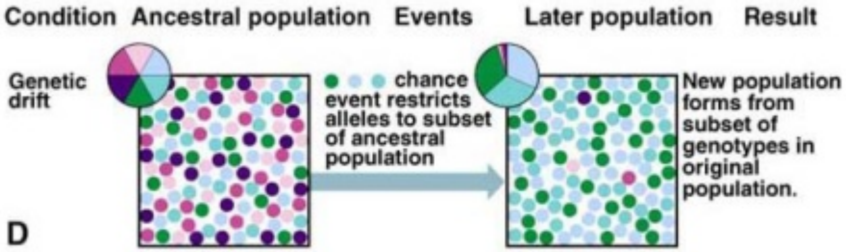
Migration

many ● and ● genotypes leave

Genotypes that leave become rarer.

C

Genetic variability • Composição Genética • Hardy-Weinberg



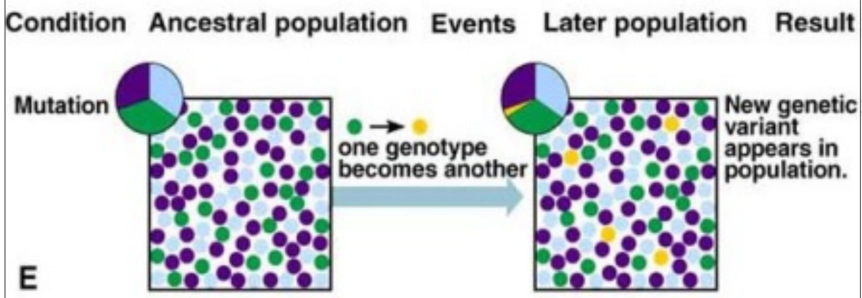
Condition | Ancestral population | Events | Later population | Result

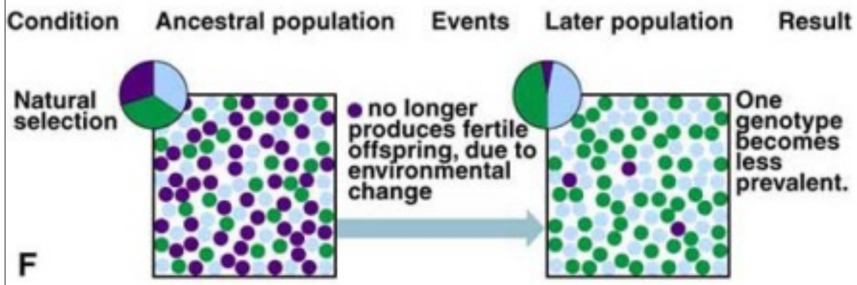
Genetic drift

● ● ● chance event restricts alleles to subset of ancestral population

New population forms from subset of genotypes in original population.

D





H-W is an important tool for population genetics.

If assumptions are met, we can use it to estimate allele and genotype frequencies that would otherwise be difficult to measure.

If assumptions are not met (can be tested statistically), then we know that some outside force is perturbing allele or genotype frequencies.

Take-home message

The **all-important** concepts of allele frequency and genotype frequency are central to the **Hardy-Weinberg principle**, which states that in the absence of perturbing factors, allele and genotype frequencies in a bi-allelic system remain constant over generations, with genotype frequencies at a ratio of p2:2pq:q2, in which p and q represent allele frequencies.

In this class we have covered:

The Hardy-Weinberg principle

- Main concepts
- How to evaluate the HWE in a sample
- What are the main factors that affect HWE