## PROBLEMS

1) Two alleles: S (p) and s (q)

1000 people

99 SS

418 Ss

483 ss

Allele and genotype freq?

Population is in HWE?

| Genot. | # ind | Genot. freq | # S alleles | # s alleles |
|---|---|---|---|---|
| SS | 99 | P = 0.099 | 198 | 0 |
| Ss | 418 | H = 0.418 | 418 | 418 |
| ss | 483 | Q = 0.483 | 0 | 966 |
| | 1000 | | | |

**Allele freq:**

$$p = \frac{(2 \cdot 99) + 418}{2 \cdot 1000} = 0.308$$

$$q = \frac{(2 \cdot 483) + 418}{2 \cdot 1000} = 0.692$$

Test if pop is in HWE

| Genot. | Obs. | Exp | $\chi^2$ |
|---|---|---|---|
| SS | 99 | $p^2 \cdot N = 94.86$ | 0.18 |
| Ss | 418 | $2pq \cdot N = 426.27$ | 0.16 |
| ss | 483 | $q^2 \cdot N = 478.86$ | 0.03 |
| | | | 0.37 |

$\chi^2$ test

$df = 3-2 = 1$ $\quad \chi^2_{0.05,1} = 3.84$

$0.37 < 3.84$ $\quad$ Accept $H_0$ (equal) → Pop. in HWE

2) Aa Allele freq.? Proportion heteroz. indiv. in human pop?

$p + q = 1$ $\quad$ PKU → autosomal recessive

$p^2$ = AA homoz. dom.
$2pq$ = Aa heteroz.
$q^2$ = aa homoz. recess.

Then, homozygous recessive ($q^2$) will have the disease
Heterozygous ($2pq$) will be carriers
Homozygous dominant ($p^2$) will not have the disease and will not be carriers.

1/10 000 persons w/ dis.

↳ $q^2 = 1/10\,000 \Rightarrow q = \sqrt{1/10\,000} = \boxed{0.01 = q}$

$p + 0.01 = 1 \rightarrow \boxed{p = 0.99}$

$p^2 + (2pq) + q^2 = 1$ → Prop. heteroz. indiv.

$2 \cdot 0.99 \cdot 0.01 = 0.0198 = \underline{1.98\%}$

3) 12% men → Dom trait (X-linked) → A = 0.12

% women exp. to present the phenotype? % if trait was recess.?

Males → A = p, a = q → 1 possib. of A
Females → AA = $p^2$, Aa = $2pq$, aa = $q^2$

2 possib. of A → $p^2 + 2pq$

→ For men with dom. trait:

$p = 0.12 \qquad p + q = 1 \rightarrow q = 0.88$

Then, for women:

$p^2 + 2pq = (0.12)^2 + 2(0.12)(0.88) = 0.2256$

$q^2 = (0.88)^2 = 0.7744$

We expect 22.56 % of women presenting this dominant phenotype

→ For men with recess. trait:

$q = 0.12 \qquad p + q = 1 \rightarrow p = 0.88$

Then, for women

$q^2 = (0.12)^2 = 0.0144$

We expect 1.44 % of women presenting this recessive phenotype

$p^2 + 2pq = (0.88)^2 + 2(0.88)(0.12) = 0.9856$

4) 58 flies → normal bristles → s
42 flies → Stubble bristles → S (Dominant)

SS → flies with Stubble phenotype
Ss → flies with Stubble phenotype (carriers of recess. wild-type phenotype)
ss → flies with wild-type phenotype

| Genotype | # indiv. |
|---|---|
| SS | 0 |
| Ss | 42 |
| ss | 58 |
| | 100 |

Allele frequencies

$p(S) = \frac{42}{2 \cdot 100} = 0.21$

$q(s) = \frac{(2 \cdot 58) + 42}{2 \cdot 100} = 0.79$

To know if population is in H-W eq.

| Genot. | Obs. | Exp. | $\chi^2$ | |
|---|---|---|---|---|
| SS | 0 | $p^2 \cdot N = 4.41$ | 4.41 | 7.06 |
| Ss | 42 | $2pq \cdot N = 33.18$ | 2.34 | |
| ss | 58 | $q^2 \cdot N = 62.41$ | 0.31 | |

$\chi^2$ test

df = 3-2 = 1 $\qquad \chi^2_{0.05,1} = 3.84$

$7.06 > 3.84$

We reject the null hypothesis ($H_0$) of H-W equilibrium. The observed genotype frequencies are significantly different from the expected genotype frequencies under H-W equilibrium.

5) 4 alleles $\longrightarrow$ p = 0.43, q = 0.37, r = 0.18, s = 0.02

6-9 repeats TTA

If we have 4 alleles, we will have 10 genotypes:

$$\text{Genotype number} = \frac{4(4+1)}{2} = 10$$

Genotypes: AA, AB, AC, AD, BB, BC, BD, CC, CD, DD

Frequency: $p^2 + 2pq + 2pr + 2ps + q^2 + 2qr + 2qs + r^2 + 2rs + s^2$

Expected frequency of heterozygotes

$$\frac{\text{Het}}{\text{Total}} = \frac{2pq + 2pr + 2ps + 2qr + 2qs + 2rs}{p^2 + 2pq + 2pr + 2ps + q^2 + 2qr + 2qs + r^2 + 2rs + s^2}$$

$$= \frac{0.6454}{1} = \boxed{0.6454}$$

6) 4% → recessive phenotype

gene → Autosomal

Allele frequencies

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

$q^2 = \sqrt{0.04} = 0.2$

$p = 1 - 0.2 = 0.8$

Prob. descendant presents the recess. trait (two indiv. with dom.)

| | A | a |
|---|---|---|
| A | AA | Aa |
| a | Aa | aa |

$P(aa) = \frac{1}{2} \cdot \frac{1}{2} = \frac{1}{4}$

Final probability → 25%

7) Two alleles ($A_1$, $A_2$)
93 mice

Genotype freq:

$A_1A_1$: 0.226
$A_1A_2$: 0.4
$A_2A_2$: 0.374

Allele freq.

$p = 0.226 + \frac{1}{2}(0.4)$

$= 0.426$

$q = 0.374 + \frac{1}{2}(0.4)$

$= 0.574$

Population is in H-W eq.?

| Genotype | Obs. | Exp. | $\chi^2$ |
|---|---|---|---|
| $A_1A_1$ | 0.226 | $p^2 = 0.18$ | 0.012 |
| $A_1A_2$ | 0.4 | $2pq = 0.49$ | 0.016 |
| $A_2A_2$ | 0.374 | $q^2 = 0.33$ | 0.006 |
| | | | 0.034 |

$\chi^2$ test

$df = 3 - 2 = 1$

$\chi^2_{0.05,1} = 3.84$

$0.034 < 3.84$

We can conclude that the pop is in H-W eq.

8) 35% white mice → aa

$q = \sqrt{0.35} = 0.59$

$p = 1 - q = 0.41$

} Allele freq.

$2pq = 2(0.59)(0.41) = 0.484$

$p^2 = (0.41)^2 = 0.168$

$q^2 = (0.59)^2 = 0.348 \simeq 0.35$

} Genotype freq

9) Genotype freq. and allele freq?

Fem. - Brown color (BB, Bb, bb) → (0.81, 0.18, 0.01)

Mal. - 3 bulls (1 BB, 2 Bb)

- Possible combinations in the offspring:

  - BB x BB → BB
  - BB x Bb → 1/2 BB and 1/2 Bb
  - BB x bb → Bb
  - Bb x Bb → 1/4 BB, 1/2 Bb and 1/4 bb
  - Bb x bb → 1/2 Bb and 1/2 bb
  - bb x bb → bb

- Genotypes freq. (original pop.)

  - BB = 3/6 = 0.5
  - Bb = 2/6 = 0.33
  - bb = 1/6 = 0.17

- Alleles freq. (offspring)

  - B = 0.5 + 1/2 · 0.33 = 0.67
  - b = 0.17 + 1/2 · 0.33 = 0.33

- Genotypes freq. (offspring)

  - BB = $p^2 = (0.67)^2 = 0.45$
  - Bb = $2pq = 2(0.67)(0.33) = 0.44$
  - bb = $q^2 = (0.33)^2 = 0.11$

To determine if the next generation will be in H-W eq.

| Genot. | Obs. | Exp. | $\chi^2$ |
|---|---|---|---|
| BB | 0.5 | 0.45 | 0.005 |
| Bb | 0.33 | 0.44 | 0.03 |
| bb | 0.17 | 0.11 | 0.03 |
| | | | 0.07 |

Based on p and q alleles (0.67, 0.33)

$df = 3 - 2 = 1$

$\chi^2$ test

$\chi^2_{0.05,1} = 3.84$

$0.07 < 3.84$

Next generation will be in H-W eq.

10) 20 people (10 F, 10 M) → 40 total alleles

- 2 carry c allele (heterozygous, Cc)

$^2/_{40} = 0.05 = q$, $p = 0.95$

$q^2 = (0.05)^2 = 0.0025$

| | |
|---|---|
| 18 | CC |
| 2 | Cc |
| 0 | cc |
| 20 | ind. |

The incidence of cystic fibrosis on the island would be 0.25%, which means that 1 in 400 individuals on the island would have CF.