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Problem 1:

SS: 99

Ss: 418

ss: 483

Total: 1000

SS observed frequency: $99/1000 = 0.099$

Ss observed frequency: $418/1000 = 0.418$

ss observed frequency: $483/1000 = 0.483$

$S = SS*2 + Ss \rightarrow S = 99*2 + 418 = 616 \rightarrow p = S/2N = 616/2000 = 0.308$

$s = ss*2 + Ss \rightarrow s = 483*2 + 418 = 1384 \rightarrow q = s/2N = 1384/2000 = 0.692$

SS expected frequency: $P = p^2 = 0.308^2 = 0.095$

Ss expected frequency: $H = 2pq = 2*0.308*0.692 = 0.426$

ss expected frequency: $Q = q^2 = 0.692^2 = 0.479$

$X^2 = (O-E)^2 / E$

$X^2_{SS} = (0.099 - 0.095)^2 / 0.095 = 0.168*10^{-3}$

$X^2_{Ss} = (0.418 - 0.426)^2 / 0.426 = 0.150*10^{-3}$

$X^2_{ss} = (0.483 - 0.479)^2 / 0.479 = 0.033*10^{-3}$

$Total = 0.168*10^{-3} + 0.150*10^{-3} + 0.033*10^{-3} = 0.351*10^{-3} = X^2_{total}$

df = number of genotypes - number of alleles

df = 3 - 2 = 1

$X^2_{0.05, 1} = 3.81$

$X^2_{0.05, 1}$ vs X^2_{total}

$0.351*10^{-3} < 3.81 \rightarrow$ The Null Hypothesis is true, so the population follows a Hardy-Weinberg equilibrium

Problem 2:

Recessive (aa)

Affected ratio = 1/10000

a/A frequencies?

Aa frequency?

$p = A$

$q = a$

$p + q = 1$

$p^2 = AA$

$2pq = Aa$

$q^2 = aa$

$q^2 = 1/10000 \rightarrow \sqrt{0.1*10^{-3}} = q = 0.01$

$p + q = 1 \rightarrow p = 1 - q \rightarrow p = 1 - 0.01 \rightarrow p = 0.99$

$p^2 + 2pq + q^2 = 1 \rightarrow 1 - p^2 + q^2 = 2pq \rightarrow 1 - 0.99^2 + 0.01^2 = 2pq = 0.0198 \rightarrow 0.0198*100 = 1.98\%$

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The frequency for the normal allele (A) is 0.99 and the frequency for diseased allele (a) is 0.01
The proportion of heterozygote individuals (Aa) is of 1.98%, almost 2 individuals out of every 100.

Problem 3:

12% men, X linked → 0.12=A

% women A? % aa if it was recessive?

For the case of the dominant trait

$$0.12=A=p$$

Male options: $A=p$, $a=q$

A in phenotype can only have probability of $p \rightarrow p=0.12 \rightarrow p+q=1 \rightarrow q=1-0.12 = 0.88=q$

Female options: $AA=p^2$, $Aa=2pq$, $aa=q^2$

A in phenotype can be p^2 or $2pq$, so $A_{ph} = p^2+2pq \rightarrow A_{ph}=0.12^2+(2*0.12*0.88) = 0.23$

23% of women will present this phenotype in the dominant case

In case of the recessive trait

$$0.12=a=q$$

Male options: $a=q$, $A=p$

a in phenotype can only have probability of $q \rightarrow q=0.12 \rightarrow p+q=1 \rightarrow p=1-0.12 = 0.88=p$

Female options:

Recessive trait will only show if $aa (q^2) \rightarrow q^2=0.12^2 = 0.0144$

1.4% of women will present this phenotype in the recessive case

Problem 4:

ss 58

Ss 42 (Stubble)

SS are dead

SS observed frequency: $0/100 = 0$

Ss observed frequency: $42/100 = 0.42$

ss observed frequency: $58/100 = 0.58$

$$S=0*2+42=42 \rightarrow p=S/2N=42/200=0.21$$

$$s=58*2+42=158 \rightarrow q=s/2N=158/200=0.79$$

The frequency of allele S is 0.21 and the frequency of allele s is 0.79.

SS expected frequency: $P=p^2=0.21^2=0.044$

Ss expected frequency: $H=2pq=2*0.21*0.79=0.332$

ss expected frequency: $Q=q^2=0.79^2=0.624$

$$X^2 = (O-E)^2 / E$$

$$X^2_{SS} = (0-0.044)^2 / 0.044 = 0.044$$

$$X^2_{Ss} = (0.42-0.332)^2 / 0.332 = 0.023$$

$$X^2_{ss} = (0.58-0.624)^2 / 0.624 = 0.003$$

$$\text{Total} = 0.003+0.023+0.044 = 0.07 = X^2_{\text{total}}$$

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df=number of genotypes-number of alleles

$$df=3 - 2=1$$

$$X^2_{0.05, 1} = 3.81$$

$$X^2_{0.05, 1} \text{ vs } X^2_{\text{total}}$$

$0.07 < 3.81 \rightarrow$ The Null Hypothesis is true, so the population follows a Hardy-Weinberg equilibrium

Problem 5:

4 alleles

6-9 TTA each

$$p=0.43$$

$$q=0.37$$

$$r=0.18$$

$$s=0.02$$

$$\text{Genotype number} = (\text{alleles} * (\text{alleles} + 1)) / 2 \rightarrow 4 * (4 + 1) / 2 = 20 / 2 = 10$$

10 genotypes are possible

The frequency of heterozygotes can be calculated with

$$\text{total} = p^2 + 2pq + 2pr + 2ps + q^2 + 2qr + 2qs + r^2 + 2rs + s^2 = 1$$

$$\text{htz} = 2pq + 2pr + 2ps + 2qr + 2qs + 2rs$$

$$f = \text{htz} / \text{total} \rightarrow f = 0.65 / 1 = 0.65$$

The expected frequency for an heterozygote is of 0.65

Problem 6:

$$0.04 \text{ recessive (aa)} \rightarrow q^2=0.04$$

(alle

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

$$p=1-q \rightarrow p=1-0.2 = p=0.8$$

The frequency for the recessive allele is of 0.2, the frequency of the dominant allele is of 0.8.

	A	a
A	AA	Aa
a	Aa	aa

The probability of a descendant of 2 carriers presenting the recessive trait is of 0.25, if the progenitors were not carriers (and dominant) it would be of 0%.

Problem 7:

93 mice

alleles A1 and A2

Observed:

$$A1A1=0.226$$

$$A1A2=0.4$$

$$A2A2=0.374$$

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$$p = A_1A_1 + (A_1A_2) / 2 = 0.226 + 0.4 / 2 = 0.426$$
$$q = 1 - p = 1 - 0.426 = 0.574$$

Expected:

$$P = p^2 \rightarrow P = 0.18$$

$$2pq = 0.49$$

$$q^2 = 0.33$$

$$X^2 = (O - E)^2 / E$$

$$X^2_{A_1A_1} = (0.226 - 0.18)^2 / 0.18 = 0.012$$

$$X^2_{A_1A_2} = (0.4 - 0.49)^2 / 0.49 = 0.016$$

$$X^2_{A_2A_2} = (0.374 - 0.33)^2 / 0.33 = 0.006$$

$$\text{Total} = 0.012 + 0.016 + 0.006 = 0.034 = X^2_{\text{total}}$$

df = number of genotypes - number of alleles

$$df = 3 - 2 = 1$$

$$X^2_{0.05, 1} = 3.81$$

$$X^2_{0.05, 1} \text{ vs } X^2_{\text{total}}$$

$0.034 < 3.81 \rightarrow$ The Null Hypothesis is true, so the population follows a Hardy-Weinberg equilibrium

Problem 8:

35% white mice $\rightarrow aa$

$$q^2 = 0.35 \rightarrow \sqrt{0.35} = q = 0.59$$

$$p = 1 - q \rightarrow p = 1 - 0.59 = 0.41$$

The frequency for the recessive allele is of 0.59, the frequency of the dominant allele is of 0.41.

$$p^2 = 0.41^2 = 0.168$$

$$q^2 = 0.35$$

$$2pq = 2 * 0.59 * 0.41 = 0.484$$

The frequency for the recessive homozygote is of 0.35, the frequency of the dominant homozygote is of 0.168 and the frequency for the heterozygote is of 0.484.

Problem 9:

Cows

$$BB = 0.81$$

$$Bb = 0.18$$

$$bb = 0.01$$

Bulls

$$BB = 0.33$$

$$Bb = 0.66$$

??

Problem 10:

20 individuals \rightarrow 40 alleles

2 Cc \rightarrow 2 c alleles

18 CC

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$$2/40 = 0.05 = q$$

$$p = 1 - q \rightarrow p = 1 - 0.05 = p = 0.95$$

$$q^2 = 0.05^2 = 0.0025 \rightarrow \text{proportion of cc individuals}$$

The incidence of cystic fibrosis in the island will be of 0.25% (1 out of 400 individuals will be cc)