Part II- Sex linkage

Exercise 5

The frequency of one form of recessive X-linked colour-blindness is 5% among European males. What is the expected frequency of this form of colour blindness among females? What fraction of females would be heterozygous carriers? Note, consider le locus X with the 2 alleles X1 and X2, X2 making colour-blindness.

Frequency of X_2 = p = frequency of color-blind men (genotype X_2Y). q=1-p= 0.95.

Frequency of color-blind women (genotype X_2X_2) = $p^2 = 0{,}05^2 = 0.0025$,

Frequency of carriers of the recessive gene (X_2X_1) = 2pq = 2 x 0.05 x 0.95 = 0.095.

Frequency of women carriers: 0.0975, less than 1 %. That is much less color-blind women than men.

Exercise 6

Searle (1949) gives the frequencies of a number of genes in a sample of cats in London. The animals examined were sent to clinics for destruction, they were therefore not necessarily a random sample. Among the genes studied was the sex-linked gene called "Orange" (O). All three genotypes in females are recognizable, the heterozygote being "tortoiseshell" or "calico".

The data were tested against the Hardy-Weinberg expectations, to see particularly if there was any evidence of non-random mating. The first test was to see whether the gene frequency is the same in the two sexes. Then the genotypes in females were tested against the Hardy Weinberg law in the same way.

a- Test for the HWE.

	Number of individuals						
	Females				Males		
	++	+O	OO	Total	+	O	Total
Observed	277	54	4	335	311	42	353
Expected	276	56	3	335			

$$+ = \frac{2n_{++}}{2N} + \frac{1n_{+O}}{2N} = \frac{2 \times 277}{2 \times 335} + \frac{1 \times 54}{2 \times 335} \sim 0.827 + 0.080 = 0.907$$

$$O = \frac{2n_{OO}}{2N} + \frac{1n_{+O}}{2N} = \frac{2 \times 4}{2 \times 335} + \frac{1 \times 54}{2 \times 335} \sim 0.012 + 0.081 = 0.093$$

If HWE,

++= $(0.907)^2$=0.823 (x335 ~ 276 ind.), OO= $(0.093)^2$=0.009 (x335 ~ 3 ind.), +O=335-(276+3)=56

χ^2 = 0.0036+0.0714+0.3333= 0.408; d.f. = 3-2 =1; $0.5 < p\text{-value} < 0.9$: we can conclude that the observed genotype frequencies at this locus don't deviate significantly from the HWE.

b- Calculate the number of each allele for every sex and in the population.

	Number of alleles			Frequency of O-allele
	+	O	Total	q
in Females	608	62	670	0.0925
in Males	311	42	353	0.1190
Total	919	104	1023	

In females, $q_f = 62/670 = 0.0925$; In males, $q_m = 42/353 = 0.1190$.

c- Does the alleles frequency is different in males and in females?

HWE implies that the gene frequencies between sex are the same but let's test it. Expected values: total in sex x total allele / total alleles.

e.g.: number of alleles expected in females = (670 x 919) / 1023 ~ 602

	Number of alleles expected		
	+	O	Total
in Females	602	68	670
in Males	317	36	353
Total	919	104	1023

$$\chi^2 = \frac{(608-602)^2}{602} + \frac{(62-68)^2}{68} + \frac{(311-317)^2}{317} + \frac{(42-36)^2}{36} = 0.060 + 0.529 + 0.114 + 1$$

$\chi^2 = 1.7$; d.f. = 3-2 = 1, $0.5 < p\text{-value} < 0.9$: we can conclude that the allele frequencies in male don't differ to frequency in female.