

2.3 Sex-linked inheritance

One of the preconditions for H-W equilibrium is random mating in the population. In such a population H-W equilibrium always occurs after one generation of random mating, provided that both males and females have the same gene frequency.

By sex-linked inheritance there is not necessarily the same gene frequency in males and females after random mating, since the two sexes get their genes from two different sources.

In mammals the males get all their sex-linked genes from their mothers, whereas the females get half their genes from their sires and half from their dams. The opposite is the case in birds, where the female is the heterogametic sex.

Example, the orange gene in cats: In a population the following number of the three genotypes were found. The orange gene (O) gives yellow coat colour. The genotype Oo gives a mixed colour, which is caused by random inactivation of the X-chromosome in XX individuals. The oo phenotype is non-yellow.

	females					males	
Genotype	OO	Oo	oo	Total	O	o	Total
Number	3	53	117	173	28	149	177
Frequency	0.02	0.31	0.67	= 1.00	0.16	0.84	= 1.00

In mammals the sex-linked gene is shown directly in the males. That is, the genotype frequency equals the gene frequency. Whereas in the females the calculation of the gene frequency is identical with the calculation for autosomal genes, as shown below.

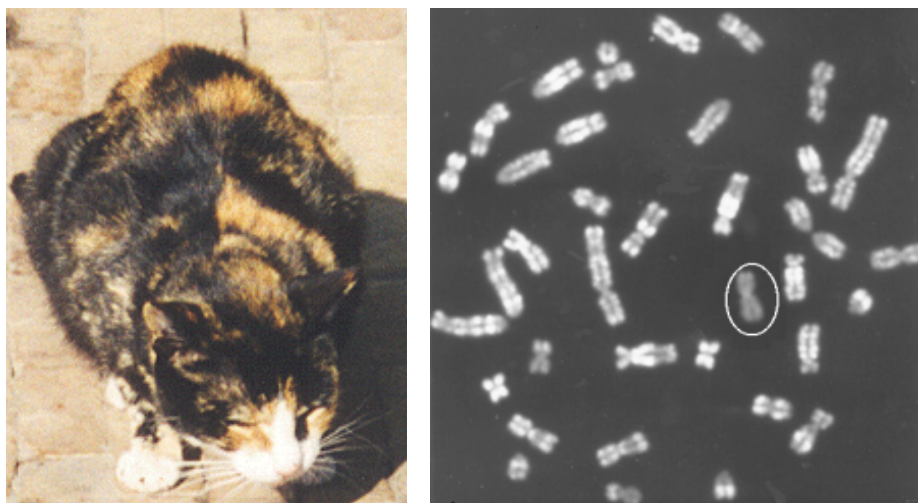
$$\begin{array}{lcl} \text{Frequency of } O & \text{calculated as } p = (2 \cdot 3 + 53) / (2 \cdot 173) = 0.17 \\ \text{do } o & \text{do } q = (2 \cdot 117 + 53) / (2 \cdot 173) = 0.83 \\ & \text{-----} \\ & 1.00 \end{array}$$

There are only minor differences between the frequency in the two sexes, as shown. If there is a difference it will be halved in each new generation with random mating, so in practice H-W equilibrium is reestablished after a few generations, if the population deviated from equilibrium in the first place. Phenotypically the heterozygotes are very special, since only one gene is active in each cell. The phenotypes occur because of a random blend of cells with an alternative activated gene.

From the chromosome preparation it can be seen that one of the X-chromosome is inactive (circled), figure 2.4 left. To the right is a heterozygotic female cat with the genotype Oo. It has yellow and black spots caused by alleles in the sex-linked orange locus. The white colour is caused by an autosomal gene for spots.

Figure 2.4

Heterozygote female cat with the genotype Oo in the sex-linked Orange locus. Photo Bodil Andersen, Bogense. It gives a blended coat colour of yellow and black with varying spot sizes. This is caused by random inactivation of one X-chromosome in female mammals. The inactive



swine X-chromosome is circled.

The inactive X-chromosome is detected by means of Acridine orange staining on cells grown for 6.5 hours with BrdU (Brom deoxy-Uridine) in the media. DNA-syntheses which occur after adding BrdU can be seen as weakly stained areas on the chromosomes. There is a circle around the inactive X chromosome. The active genes replicate early in the cell cycle and are therefore found in the white band.

The majority of cases with recessive sex-linked diseases occur in males. Haemophilia in human is one of the best known examples of sex-linked recessive inheritance, the frequency in boys being 100 times larger than the one in girls. This occurs when the gene frequency is 0.01, corresponding to the frequency in boys. Whereas the gene in the girls has to come from both father and mother, each with a probability of 0.01, this corresponds to a frequency in girls of 1 in 10'000.