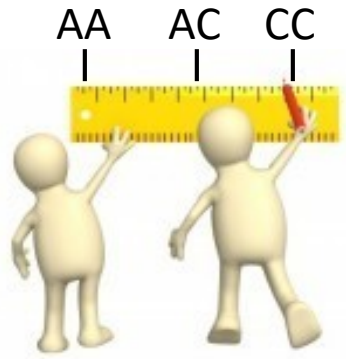
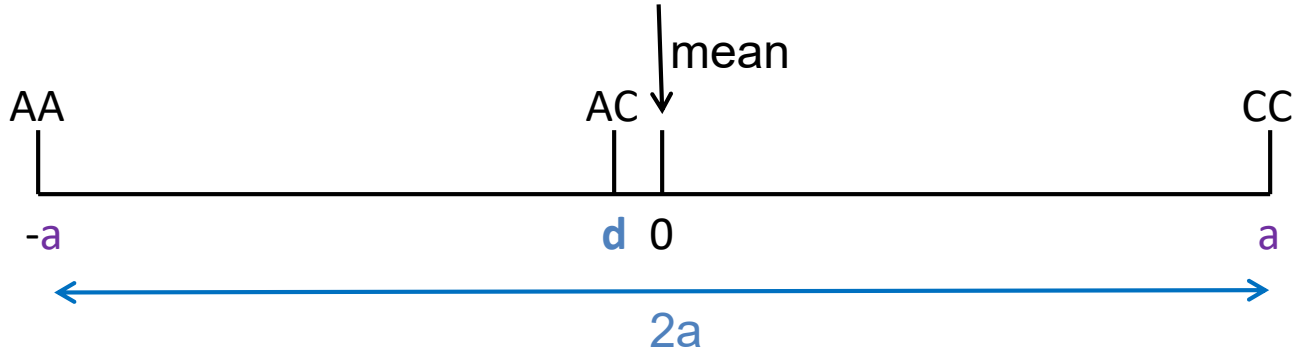
Mean IL-6R concentration of each genotype:

CC: 5.698 / CA: 4.418 / AA: 3.238 (10^{-8} g/mL)

Total Variance of IL-6R concentration=1.35

Frequencies: C, frequency: p=0.39 / A, frequency: q =0.61





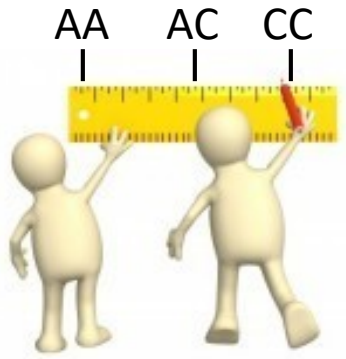
QUESTIONS (Falconer & MacKay; 1996: Introduction to quantitative genetics)

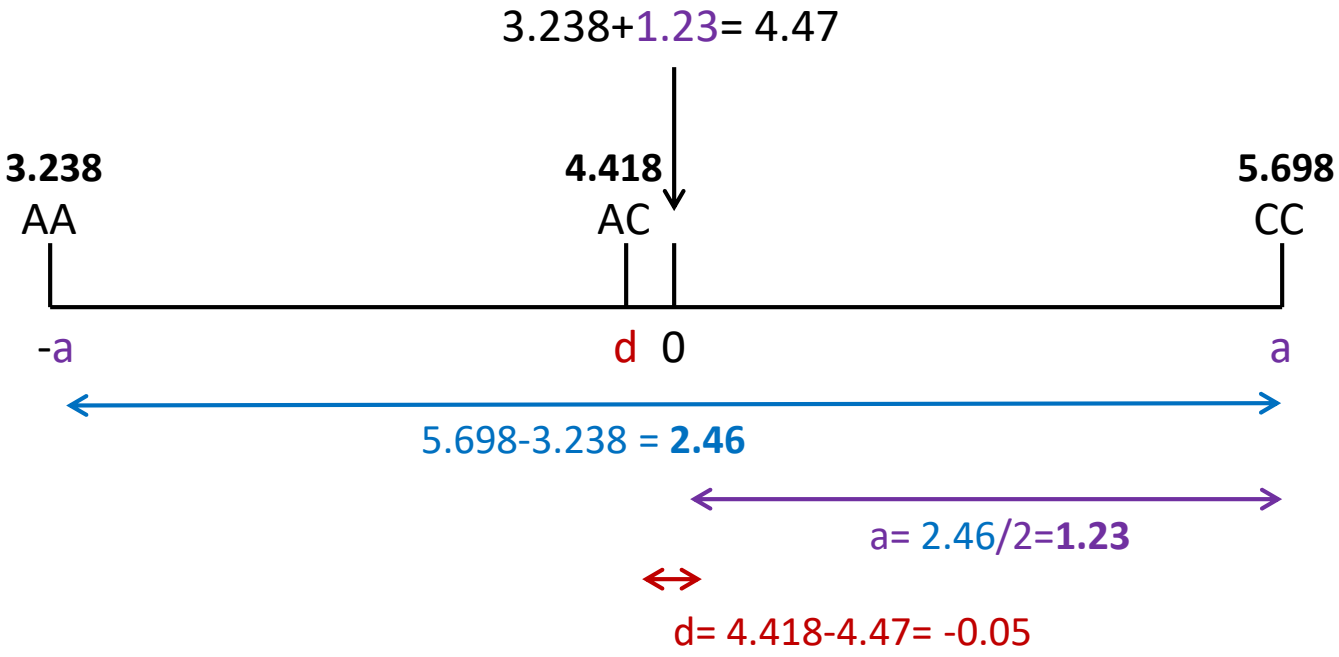
1. Calculate genotypic values (a and d) (page 109)
2. Calculate the genotype frequencies (page 7)
3. Calculate the mean IL6-R concentration in the population (page 110)
4. Calculate how much of the variance is explained by this SNP
   (*Variance= Sum of squared deviations from the mean*)

extra: Calculate the average effect of the alleles (page 113)

QUESTIONS

1. Calculate genotypic values (a and d) (Falconer, page 109)





1. Calculate the genotype frequencies
2. Calculate the mean IL6-R concentration in the population (page 110)
3. Calculate how much of the variance is explained by this SNP

Minor allele: C, frequency: p=0.39

Major Allele: A, frequency: q =0.61

	CC		**CA**		**AA**
1.Genotype frequencies	= p^2	+	2pq	+	q^2
	= 0.39^2	+	2*0.39*0.61	+	0.61^2
	= **0.15**	**+**	**0.48**	**+**	**0.37**
mean IL6R	5.698		4.418		3.238

2. Population mean $p^2 * 5.698 + 2pq * 4.418 + q^2 * 3.238$

0.15 * 5.698 + 0.48 * 4.418 + 0.37 * 3.238 = **4.17**

3. Deviation from mean = 5.698-4.17 | 4.418 - 4.17 | 3.238 - 4.17

= 1.52 | 0.24 | -0.94

4. Squared deviation = 1.52^2 =2.32 | 0.24^2 =0.06 | -0.94^2 =0.88

1. Calculate the genotype frequencies
2. Calculate the mean IL6-R concentration in the population (page 110)
3. Calculate how much of the variance is explained by this SNP

Minor allele: C, frequency: p=0.39

Major Allele: A, frequency: q =0.61

	CC	**CA**	**AA**
3. Deviation from mean =	5.698-4.17	4.418 - 4.17	3.238 - 4.17
	= 1.52	0.24	-0.94
4. Squared deviation	= 1.52^2 =2.32	0.24^2 =0.06	-0.94^2 =0.88
5. Variance (SNP)	= p^2 * 2.32	+ 2pq * 0.06	+ q^2 * 0.88
	= 0.15* 2.32	+ 0.48* 0.06	+ 0.37*0.88 = **0.71** (SD = 0.84)

Total variance of IL-6R concentration= 1.35

% variance explained by SNP= 0.71/1.35 = 53%

Variation: $2pq[a+d(q-p)]^2 + (2pqd)^2$

$$\begin{aligned} a &= 0.5 \times (\text{sIL-6R}_{CC} - \text{sIL-6R}_{AA}) \\ &= 0.5 \times (5.698 - 3.238) = 1.23 \end{aligned}$$

$$\begin{aligned} d &= \text{sIL-6R}_{AC} - (\text{sIL-6R}_{AA} + a) \\ &= 4.418 - (3.238 + 1.23) = -0.05 \end{aligned}$$

$$\begin{aligned} V_A &= 2pq\,[a + d\,(q - p)]^2 \\ &= 2 \times 0.39 \times 0.61 \\ &\quad \times [1.23 - 0.05 \times (0.61 - 0.39)]^2 \\ &= 0.71 \end{aligned}$$

$$\begin{aligned} V_D = (2pqd)^2 &= (2 \times 0.39 \times 0.61 \times -0.05)^2 \\ &= 5.66 \times 10^{-4} \end{aligned}$$

Average effect

(associated with genes and not with genotypes)

The average effect of a gene (allele) is the mean deviation from the population mean of individuals which received that gene from one parent, the gene received from the other parent having come *at random* from the population.

Falconer (p112):The concept of average effect is not easy to grasp.

Average effect is related to genotypic values a and d

$q\,[a + d\,(q - p)] = \alpha_1$

$-p\,[a + d\,(q - p)] = \alpha_2$

Average effect of gene substitution is $\alpha_1 - \alpha_2 = \alpha$. This is the difference between the average effect of the 2 alleles:

$\alpha = a + d(q-p)$

2. Calculate the average effect of the alleles (page 113)

Minor allele: C, frequency: p= 0.39
Major Allele: A, frequency: q=0.61

a=1.23, d= -0.05

Average effect C= q[a+d(q-p)] = 0.61[1.23-0.05(0.61-0.39)] **= 0.74**

Average effect A= -p[a+d(q-p)]= -0.39[1.23-0.05(0.61-0.39)] **= -0.48**