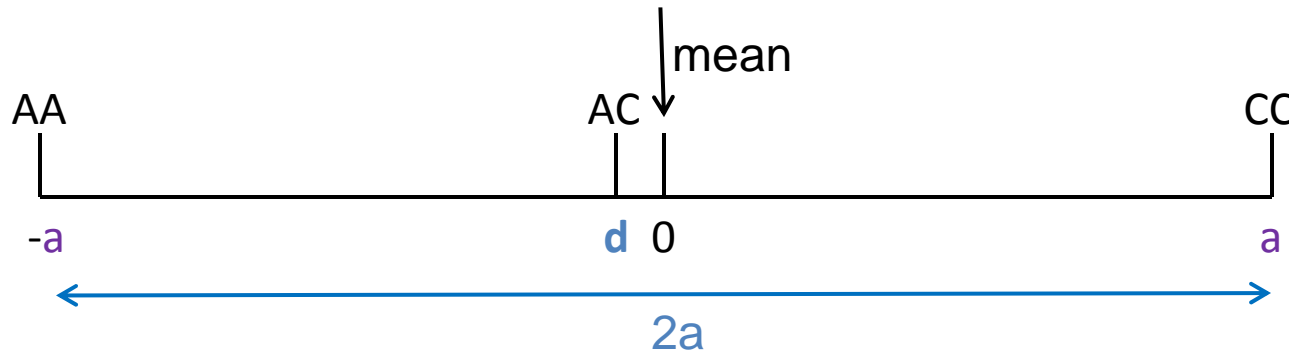
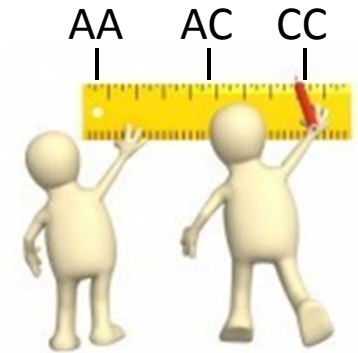


Mean IL-6R concentration of each genotype:

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:  $q=0.61$

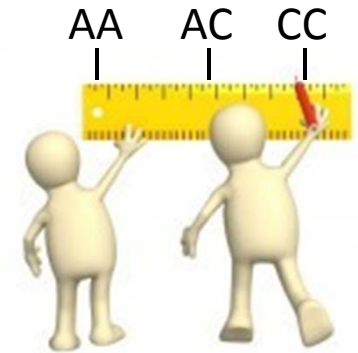
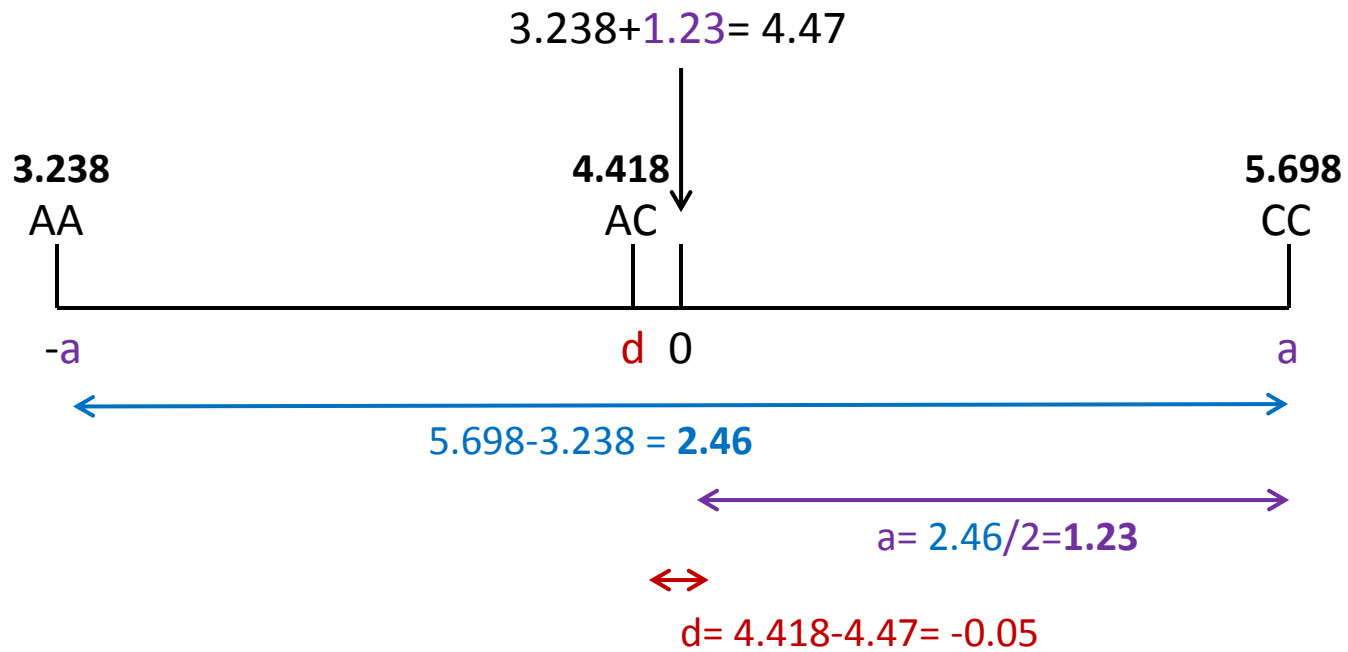


QUESTIONS (Falconer & MacKay; 1996: Introduction to quantitative genetics)

1. Calculate genotypic values ( $a$  and  $d$ ) (page 109)
2. Calculate the average effect of the alleles (page 113)
3. Calculate the genotype frequencies (page 7)
4. Calculate the mean IL6-R concentration in the population (page 110)
5. Calculate how much of the variance is explained by this SNP  
(*Variance = Sum of squared deviations from the mean*)
6. Calculate heritability

## QUESTIONS

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



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=  $q[a+d(q-p)] = 0.61[1.23-0.05(0.61-0.39)] = \mathbf{0.74}$

Average effect A=  $-p[a+d(q-p)]= -0.39[1.23-0.05(0.61-0.39)] = \mathbf{-0.48}$

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:  $q=0.61$

	<b>CC</b>		<b>CA</b>		<b>AA</b>
1. Genotype frequencies	$= p^2$	+	$2pq$	+	$q^2$
	$= 0.39^2$	+	$2*0.39*0.61$	+	$0.61^2$
	$= \mathbf{0.15}$	+	$\mathbf{0.48}$	+	$\mathbf{0.37}$
mean IL6R	5.698		4.418		3.238
2. Population mean	$p^2 * 5.698$	+	$2pq * 4.418$	+	$q^2 * 3.238$
	$\mathbf{0.15 * 5.698}$	+	$\mathbf{0.48 * 4.418}$	+	$\mathbf{0.37 * 3.238 = 4.17}$
	↓		↓		↓
3. Deviation from mean	$= 5.698 - \mathbf{4.17}$		$4.418 - \mathbf{4.17}$		$3.238 - \mathbf{4.17}$
	$= 1.52$		$0.24$		$-0.94$
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:  $q =0.61$

	<b>CC</b>	<b>CA</b>	<b>AA</b>
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)	$= p^2 * 2.32$	+	$2pq * 0.06$
	$= 0.15 * 2.32$	+	$0.48 * 0.06$
			+
			$q^2 * 0.88$
			$= 0.37 * 0.88 = 0.71$ (SD = 0.84)

Total variance of IL-6R concentration= 1.35

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

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

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

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