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Answers

Barley and lesion mimic mutants

Answers to practice exam questions on p. 6

Martin Rowland

1 Allow 1000 Mb (decimal)

OR

1024 Mb (computing/binary)

2 (In eukaryotic cells) transcription produces pre-mRNA

(Pre-mRNA) contains introns that must be removed/spliced

The remaining exons can be joined in different sequences

Each sequence can code for a different protein/polypeptide

Each protein/polypeptide can be active in a unique metabolic pathway

Psoriasis: scratching the surface

Answers to practice exam questions on p. 35

Martin Rowland

1 3.44 million

2 <u>Supporting conclusion</u>

- Bimekizumab cleared psoriasis in 62% of patients but secukinumab only cleared psoriasis in 49% of patients.
- Bimekizumab inactivates interleukins IL-17A and IL-17F, whereas secukinumab only inactivates IL-17A.

  OR
- Bimekizumab causes greater inhibition of inflammation than secukinumab.

<u>Not supporting conclusion</u> (maximum of 3 marks)

- Only percentages are given, so no information about the number of patients / sample size.
- No statistical analysis given, so we do not know whether the difference is significant.
- No information about age of patients / severity of psoriasis / activity of patients / diet of patients.
- No information about concentration of monoclonal antibodies.

Evaluating experiments: wild gull chase

Suggested answers to questions on pp. 36–40

Kevin O'Dell

Question 1

The simplest explanation is that the blue/white colour variation is determined by a single gene, where the blue allele (+) is dominant to the white allele (–). Taking each row of Table 1 in turn:

Row 1: both parents are blue. The chicks fall into two distinct categories:

- In most nests all chicks are blue – if all the chicks are blue then at least one parent is homozygous blue (+/+).
- In some nests around a quarter of the chicks are white – if approximately one quarter of the chicks are white (–/–) then both parents must be heterozygous (+/–).

Row 2: one blue parent and one white parent. The chicks fall into two distinct categories:

- In some nests all chicks are blue – if the blue parent is homozygous (+/+) then all the chicks will be blue.
- In some nests around a half of the chicks are white – if the blue parent is heterozygous (+/–) then half the chicks will be blue (+/–) and half will be white (–/–).

Row 3: both parents are white. In all nests all the chicks are white – the parents must all be homozygous white (–/–), so all the offspring are homozygous white (–/–).

Question 2

All we really know at this stage are two things:

(1) The birds spend the summer in northern Canada, and the winter further south in the southern USA and Mexico.

(2) There is an east–west cline of colour variation, with white birds being more common in the east and blue birds more common in the west.

The most plausible explanation is that it has something to do with camouflage, with the white gulls being better camouflaged in a snowy environment and the blue gulls being better camouflaged when no snow is present. This of course assumes that these gulls have at least one predator.

This argument would be strengthened if the weather is indeed warmer in western Canada, meaning there might be less snow, so blue (Dull) gulls are at a selective advantage, and if there is more snow in the east, white (Hull) gulls are at a selective advantage. This hypothesis could be tested by looking at relative predation rates in western and eastern Canada. We might expect relatively more blue gulls to be predated in the snowy east and relatively more white gulls to be eaten in the absence of snow in the west.

Any answer that realistically considers points 1 and 2 would be acceptable.

Question 3

We are looking for a genetic variant (single nucleotide polymorphism or something more substantial) that is always found in blue birds but never in white birds, and vice versa. A good answer will explain more clearly an outline strategy of how this is done using sequence data alone and must consider the fact that the blue allele is dominant to the white allele. Thus if, for example, the polymorphism was caused by an SNP that was G (white allele) or A (blue allele), we would expect white birds to be GG and blue birds to be GA or AA.

Question 4

When a new mutation arises it is clearly only on one chromosome. Over time and many generations that single chromosome will undergo mutation and recombination, thus acquiring more variation around the newly arisen mutation. Therefore, the allele with more variation around it is the ancestral allele.

Question 5

When a new mutation arises it is clearly only on one chromosome. Over time and many generations that single chromosome will undergo mutation and recombination, thus acquiring more variation around the newly arisen mutation. The more similar gull DNA sequences are around the causal mutation (the G/A change at codon 85 in the *melanocortin-1 receptor* gene) the more recently that mutation arose. Therefore, if you know the mutation rate and the generation time, and can measure the amount of DNA variation around the newly arisen allele, it should be possible to estimate the date at which the mutation first arose.

Question 6

Perhaps the most plausible explanation might be that 11000 years ago marked the end of the last ice age, and we are currently experiencing global warming, both of which are contributing an increase in global temperatures. This is pushing the distribution of snow in northern Canada further north.

Therefore, if the advantage to white gulls of being white is linked to being camouflaged in a snowy environment, the loss of snow will result in the environment being more favourable to blue gulls.

Other answers are plausible.

Question 7

Genetic factors must be variation in other genes that we might call modifier genes. These might be genes that affect the pattern of gene expression, or the efficiency of the *MC1R* protein in a pattern-specific manner. Different explanations are possible, but must include variation in other genes in a pattern-specific fashion.

Non-genetic factors could be environmental factors that affect pigmentation pattern, especially at the critical time in development when the pigmentation is laid down. This could be temperature (as in Siamese cats) or food, or any other plausible, variable resource.

Question 8

The numbers of each *MC1R* colour genotype is 199 homozygous for the white allele (*val*/*val*), 203 heterozygous blue (*val*/*met*) and 598 homozygous for the blue allele (*met*/*met*). Since 1000 gulls were genotyped, we have data for 2000 alleles.

Calculating each allele frequency in turn:

White (*val*) allele frequency is:

$$\frac{(2 \times 199) + 203}{2000} = 0.3005$$

Blue (*met*) allele frequency is:

$$\frac{(2 \times 598) + 203}{2000} = 0.6995$$

Question 9

Let us define the frequency of the white allele as p and the frequency of the blue allele as q. We already know (from the answer to Question 8) that $p = 0.30$ and $q = 0.70$. Quite correctly, $p + q = 1$.

The HWE formula allows us to predict the frequency of each genotype:

homozygous white should be $p^2 = (0.30)^2 = 0.09$

heterozygous blue should be $2pq = 2 \times 0.30 \times 0.70 = 0.42$

homozygous blue should be $q^2 = (0.70)^2 = 0.49$

When added together these three frequencies equal 1.

The observed and predicted genotype frequencies are as follows:

	Observed	**Expected**
val/*val*	0.199	0.090
val/*met*	0.203	0.420
met/*met*	0.598	0.490

We can undertake a chi-squared test to investigate whether these differences are significant. We always do a chi-squared test on the original data – the absolute numbers of individuals – which in this case is a sample of 1000. Therefore:

***MC1R* genotype**	**Observed number**	**Expected number**	***O* – *E***	**(*O* – *E*)²**	$\frac{(O-E)^2}{E}$
val/val	199	90	109	11881	132
val/met	203	420	–217	47089	112
met/met	598	490	108	11664	24
Total	1000	1000	0		268

Therefore, chi-squared = 268

For 1 degree of freedom, the cut-off for 0.05% is 3.84. Therefore, the white and blue *MC1R* alleles in the Kanguq gull population are not in HWE.

Question 10

There is a lack of heterozygous gulls. There are various plausible reasons why. For example:

- Heterozygous gulls are at a selective disadvantage, because they have mixed blue and white plumage, which does not act as camouflage in any environment.
- Blue gulls prefer to mate with gulls that look like their parents, which are usually, but not always blue. Similarly, white gulls also prefer to mate with gulls that look like their parents, which are often, but not always, white. This is a kind of sexual selection called assortative mating.

Question 11

Experiments that will distinguish between the two hypotheses proposed will depend on what those hypotheses are. However, each experiment must be feasible, and written appropriately, with alternative outcomes and appropriate controls.