BSc and MSc Degree Examinations 2018-9

Department:
BIOLOGY

Title of Exam:
Genes and genomes in populations and evolution

Time Allowed:
24 hours

Marking Scheme:
Total marks available for this paper: 100

The marks available for each question are indicated on the paper

Instructions:
All questions should be answered on this question paper using Arial font size 11 or larger. Work should be submitted via the Yorkshare VLE. When a line limit is given, this is for guidance only. You will not be penalised if your answer is outside this range.

For marker use only:

<table>
<thead>
<tr>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th>Total as %</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>2</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>3</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>4</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>5</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>6</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>7</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>8</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Page 1 of 13
1. It is known that 10% of the population of a tiny remote island is colour blind. This colour blindness is caused by deleterious autosomal recessive allele.

a) What is the expected frequency of heterozygote carriers on the island?

\[ \text{Frequency of aa} = 0.1 \]
\[ \text{Assuming HW, a allele frequency} = 0.1^{0.5} = 0.316 \]
\[ \text{Frequency of aA} = 2 \times 0.316 \times (1 - 0.316) = 0.432 \]

b) It is also known that 30% of the island’s population are carriers, but have normal colour vision. Suggest a reason for the difference from the expected number of carriers with normal colour vision. (indicative line limit: 4 - 6 lines)

There are fewer heterozygotes (0.30) than expected under HW (0.432)
The population is not randomly mating resulting in fewer heterozygotes. For example, perhaps people with normal colour vision (AA and Aa) to some extent avoid reproducing with colour blind people.

c) In the US: 0.003% of the population are colour blind. Assuming that both the US and island populations are each individually in Hardy-Weinberg equilibrium, test whether there are significant differences in genotype frequencies between the island and the US.

Part (c) was excluded from the marking because we did not provide population sizes, so the exam was marked out of 97 not 100 and rescaled.

d) Suggest reasons why the island might have such a high rate of colour blindness.

Severe bottleneck event resulting in a population in which genetic drift rather than selection was stronger (1 mark). Effective population size further reduced due to probable strong reproductive skew (1 mark). Further increases in recessive allele frequency due to inbreeding (1 mark).

Feedback: a) Almost all students answered this correctly, although full marks were not given unless the calculations were shown.

b) A mark was given for saying that the island population is not in HWE, and the heterozygotes are fewer than expected. Many students suggested population subdivision as a reason, and marks were awarded for this.

d) Most students suggested genetic drift, many suggested inbreeding, very few mentioned reproductive skew.
2. The stick insect *Timema cristinae* displays a genetically based colour polymorphism: a striped and an unstriped form and an intermediate form. The figure below shows the results of a mark-recapture experiment of the three morphs on the shrub *Ceanothus* (labelled host C in the figure) and on the shrub *Adenostoma* (labelled host A in the figure). The black circles represent the unstriped form, white circles the striped form and grey circles the intermediate form. The circles show the proportion recaptured plus 1 standard error.

![Mark-recapture experiment results](image)

a) Describe the results of the experiment. Based on this experiment, explain what distribution of the morphs you expect to find on the two shrub species in natural populations. (indicative line limit: 7 - 10 lines) 

The striped form is recaptured more frequently on *Adenostoma* than the other two morphs, whereas the unstriped form followed by the striped form are recaptured more frequently on *Ceanothus* (2 marks). Based on this, due to spatially heterogeneous selection, the striped morph is most likely to be found more frequently on *Adenostoma* than the unstriped and intermediate morph (1 mark), and the frequency of the unstriped form is likely highest and that of the intermediate form lowest on *Ceanothus* (1 mark).

b) Assuming that your prediction in (a) is correct, discuss which two processes might have lead to the distribution of morphs on the two shrubs. (indicative line limit: 8 - 12 lines)

The two most likely processes that could lead to this distribution are (i) selection (1 mark) or (ii) a single origin of the individuals on each shrub species (1 mark) with different morph frequencies in the original populations on each shrub (1 mark) that are now evolving independently on the two plant species (1 mark). Selection appears to be the more likely process as the mark-recapture data suggest that there is divergent selection on the two shrubs (1 mark), which might be due to the ability to feed on each shrub (1 mark) or predation (1 mark).
Other sensible answers that follow on from student’s answer in (a) will be given credit.

c) How would you distinguish between the two processes that you described in (b)? (indicative line limit: 6 - 10 lines) (5 marks)
To test whether the populations on each shrub have separate origins, phylogenetic analyses or clustering methods could be used (either is fine, maximum 1 mark) using neutral genetic markers (1 mark). Individuals from multiple populations (1 mark) from each shrub (1 mark) should be used for this. If local selection is responsible for the pattern, one would expect individuals from each shrub not all to be closely related (1 mark). Sensible answers that follow on from student’s answer in (b) will be given credit.

Feedback: (a) The majority of students described the results from the figure well and most students also explained the expected distribution well. Some students interpreted the recapture rates in a different way from the model answers, namely that the mark recapture experiment was used to estimate population sizes, and hence low recapture rates meant larger populations. This was given full credit because it was not clear from the question that only marked individuals were recaptured and counted.
(b) Most students correctly identified divergent selection and explained this well. There were a large number of other processes mentioned, few were similar to the model answer. Credit was given for explanations that included gene flow between populations on different shrubs, genetic drift with a sensible explanation, and genetic correlations with sensible explanations. No credit was given to explanations that cited phenotypic plasticity, as the question stated that the colour polymorphism is genetic.
(c) There were a wide range of approaches cited in answers to this questions and most were given credit when they were well explained, including experiments that were designed to test for phenotypic plasticity. Some students gave very general answers that did not describe an experiment, these did not get any credit.

3. Wagyu beef is a popular type of meat from four Japanese cattle breeds. The meat is highly valued because of its characteristic marbling, which results in a rich flavour. Wagyu beef is very expensive and the supply is limited, and thus there has been much interest by cattle farmers globally to start their own Wagyu cattle herds. Until the late 1980s Wagyu cattle were exclusively reared in Japan, mostly under highly controlled indoor conditions. A small number of Wagyu embryos were introduced into Australia in the early 1990s. Australia is now the second largest producer of Wagyu beef globally. In Australia, Wagyu cattle are typically kept outdoors and their meat often does not show the desired marbling as it does in Japan.
An Australian Wagyu cattle farmer would like to improve the quality of the beef in the longer-term and is considering selecting his herd for improved marbling. He is seeking your advice on what to consider before embarking on the breeding programme.

a) Explain to the farmer the quantitative genetics principles and population genetic processes he should consider before starting his programme. Explain the alternatives to a breeding programme he could consider to improve the quality of the meat. (indicative line limit: 20 - 25 lines) (16 marks)

Written mostly in suitably non-technical language (1 mark):

How much phenotypic variation (variation in marbling) is in the herd (1 mark)? If there is none, then it is not possible to select parents with a higher degree of marbling (1 mark).

The farmer needs to consider heritability, the proportion of phenotypic variation that is due to genetic variation (1 mark). Heritability might be low because (i) there is no genetic variation (1 mark) or (ii) environmental variation is high (1 mark). In either case, selection is unlikely to lead to the desired response as the response to selection is proportional to the heritability of the trait (1 mark).

The farmer should also consider the farm environment. The outdoor environment in Australia is likely to be variable and potentially of poor quality (1 mark), especially compared to highly controlled indoor conditions in Japan (1 mark). This will lead to a decrease in heritability (1 mark).

The farmer should also consider the history of Wagyu cattle in Australia: only a small number of individuals were introduced (1 mark), so the population has gone through a bottleneck (1 mark). This means that even though the population is now large (1 mark), the effective population size and thus genetic variation will be low (1 mark) (and hence low heritability is expected).

Given the issues identified above, the farmer is likely faced with two problems: low genetic diversity and high environmental variance. He should thus consider importing animals from outside Australia to increase the genetic diversity of his herd (1 mark). He could also consider decreasing the environmental variance by moving to an indoor farming system (1 mark).

b) What kind of data should the farmer collect before starting his programme and why? (indicative line limit: 4 - 6 lines) (4 marks)

The farmer should keep records of the quality of the beef for each individual (1 mark), and of the relatedness of the individuals (1 mark). He will then be able to estimate heritability using parent-offspring regression (1 mark), and this will inform whether he is likely to see a response to selection (1 mark).

Feedback: (a) Any noticeable attempt for writing this in “non-technical language” was given a mark. The majority of students explained heritability well, and identified the variable outdoor environment as a reason why selection might not work very well. However, surprisingly few students considered the history of the cattle and the
founder effect, which is a major problem in this system. Answers discussing inbreeding were given credit. Many students suggested indoor systems and introducing new genetic material.

(b) Again, there was some diversity in the answers, some were similar to the model answers. Many students suggested other sources of information, e.g. more data on how the cattle are kept in Japan. Sensible suggestions were given credit.

4. An orchid that does not provide a reward for its pollinators has a striking purple-yellow flower colour polymorphism throughout its geographic range. The polymorphism has a genetic basis. The following fitness has been observed in populations with different frequencies of the yellow morph:

The figure shows (a) male and (b) female reproductive success of the yellow morph relative to that of the purple morph as a function of the yellow morph frequency in the population. Pollinia removal is used as a measure of male reproductive success and pollinia deposition (open symbols, solid blue line) and fruit set (closed symbols, dashed red line) are measures of female reproductive success. The dashed vertical lines show the frequencies of the yellow morph at which the fitness of both morphs is equal for each trait.

a) Describe the pattern of male fitness shown in Figure (a). (indicative line limit: 4 - 6 lines) (2 marks)

Male relative fitness of the yellow flower morph decreases with increasing frequency of the yellow morph (1 mark). The relative fitness of the yellow morph is higher than that of the purple morph at frequencies between 0 and ~0.7, and lower than that of the purple morph at higher frequencies (1 mark).

b) Explain which process is likely maintaining flower colour variation in these populations. (indicative line limit: 3 - 4 lines) (1 mark)

The variation is most likely maintained by frequency-dependent selection because the relative fitness of a morph is dependent on its frequency in a population (1 mark).
c) Explain the expected change of the flower colour frequencies in a population with a current frequency of the yellow morph of 0.2. (indicative line limit: 3 - 4 lines) (2 marks)

The frequency of the yellow morph should increase in this population up to a frequency of between 0.6 and 0.7 (1 mark) and be maintained as a stable equilibrium at this frequency (1 mark).

d) Speculate how the interaction of the flowers with a pollinator might lead to the observed relative fitness of the two morphs and comment on the frequency at which both morphs have equal fitness. (Note that this was corrected by email during the 24 h assessment period). (indicative line limit: 8 - 12 lines) (5 marks)

The pollinators are presumably attracted to the flowers due to the colour and thus pollinate (1 mark). However, they receive no reward for visiting the flower and may thus opt for visiting a different type of flower (i.e. a different colour morph) (1 mark). The pollinator is likely to encounter the more common morph first, and is thus more likely to switch to the rarer morph, thus increasing this morph’s relative fitness (1 mark). With this mechanism, one would expect the equilibrium and equal relative fitnesses to be at a frequency of 0.5 (1 mark). The actual equilibrium frequency for the yellow morph is higher, which suggests an innate preference for yellow by the pollinators (1 mark).

e) Outline an experiment to test your prediction in (d). (indicative line limit: 8 - 12 lines) (5 marks)

The hypothesis to be tested is that a pollinator will alter its preference for flower colour after visiting flowers of a particular colour (1 mark).

The experimental set-up should be a type of choice experiment (1 mark). The outline of the experiment should include a description of the treatments, e.g. experience of either colour as the explanatory variable (1 mark) followed by choice between the two colours as the response variable (1 mark). The description should also include whether the experiment is lab or field based (1 mark).

Other sensible experiments that are consistent with the student’s answer given in (d) will be given credit.

Feedback: (a-c) The majority of students answered these questions well. Marks were sometimes lost when the point of equal fitness was not mentioned in (a) or (c). A small number of students did not identify frequency-dependent selection as the process.

(d) Apologies again for the mistake in the question. This has not affected the answers in a significant way. Quite a lot of students explained the process well and recognised that the lack of reward will lead to a pollinator avoiding a particular colour. Quite a few answers argue that rare colours would “stand out” more; these were given some credit if they were well explained, but not full marks. Many students provided a correct reason
why the equilibrium was not at 0.5, and students who identified the problem but not the answer were given 1 mark for this.

(e) No student suggested the experiment outlined in the model answer. Again, there was a wide variety of suggestions, which were given credit when they followed on logically from (d). There were some good suggestions that involved manipulating the flowers so that they would provide a reward. A common problem was that an experiment was suggested without a prediction or hypothesis.

5. The table below shows the extent of population structure in three plant species as measured using SNPs in the nuclear genome and the chloroplast genome.

<table>
<thead>
<tr>
<th>Species</th>
<th>Nuclear $F_{ST}$</th>
<th>Chloroplast $F_{ST}$</th>
<th>Pollination</th>
<th>Seed dispersal</th>
</tr>
</thead>
<tbody>
<tr>
<td><em>Sorbus aucuparia</em></td>
<td>0.025</td>
<td>0.29</td>
<td>insect</td>
<td>birds</td>
</tr>
<tr>
<td><em>Quercus petraea</em></td>
<td>0.024</td>
<td>0.90</td>
<td>wind</td>
<td>small mammals and birds</td>
</tr>
<tr>
<td><em>Eucalyptus nitens</em></td>
<td>0.30</td>
<td>0.62</td>
<td>insect</td>
<td>wind</td>
</tr>
</tbody>
</table>

a) What is population genetic structure, and what causes it to arise? (indicative line limit: 7 - 10 lines) (4 marks)
Population structure refers to the situation when a population is not panmictic resulting in the population not being genetically homogenous (1 mark).
Population structure arises because individuals have limited dispersal ability (1 mark) causing different parts of the population to start diverging from one another due to drift and/or differences in selection pressure (1 mark). Barriers to migration such as mountains or rivers (dependant on ecology of organism) will cause population structure (1 mark).

b) How and why are the measures of population structure different between the nuclear and chloroplast genomes of the same species? (indicative line limit: 13 - 17 lines) (6 marks)
Chloroplast $F_{ST}$ is greater than nuclear $F_{ST}$ in all three species. (1 mark)
A seed is formed from the union of pollen (male) and egg (female). While the egg contains both nuclear DNA and other organelle DNA (including chloroplast DNA), pollen only contains nuclear DNA (1 mark). Nuclear $F_{ST}$ is based on nuclear SNPs and so consists of equal contributions from the pollen and egg, while chloroplast $F_{ST}$ is based on chloroplast SNPs towards which only the egg contributed (1 mark). High population structure indicates low migration/dispersal. In plants, dispersal takes place via movement of the seed and in cross-fertilising species also via movement of pollen. Therefore, the
chloroplast $F_{ST}$ is an indicator of migration distance mediated by seed dispersal (1 mark). The nuclear $F_{ST}$ is an indicator of migration distance mediated by seed as well as pollen movement (1 mark). As pollen, being smaller and often dispersed by wind or pollinators, in general moves greater distances than seeds, this leads to greater population structure detected (1 mark).

c) Compare the different $F_{ST}$ measures within and between the three plant species, and discuss these in view of the different pollination and seed dispersal strategies used by each. (indicative line limit: 17 - 22 lines) (6 marks)

Unclear how easily one can compare $F_{ST}$ values between the different species unless they were all carried out at the same geographic scale (1 mark). Chloroplast $F_{ST}$ is an indicator of migration distance mediated by seed dispersal, so one might expect Quercus > Sorbus > Eucalyptus because seed dispersal distance might be expected to be small mammal & bird < bird < wind (1 mark). However, the wind dispersed Eucalyptus has an intermediate Chloroplast $F_{ST}$. Nuclear $F_{ST}$ is more difficult to predict as it is an indicator of migration distance mediated by seed as well as pollen movement. Again Eucalyptus is perhaps odd as its pollen is insect dispersed and seeds wind dispersed so should have the lowest population structure, yet has the highest $F_{ST}$ (1 mark). More useful is probably the ratio (chloroplast $F_{ST}$)/(nuclear $F_{ST}$) which represents the relative values for each species (1 mark). These values are Quercus 37.5, Sorbus 11.6, Eucalyptus 2.1 and are perhaps more consistent with the different pollination and seed dispersal strategies. For example, the ratio is highest for Quercus which has long migration of pollen (wind pollination) and relative low seed dispersal (small mammals and birds) (1 mark). The ratio is smallest for Eucalyptus where pollen and seed dispersal are more likely to be of similar order as these are mediated by insects and wind respectively (1 mark).

Feedback: a) This part was generally answered well by most students.
b) Most students scored poorly in this section as only a few students identified that the seed transmits both nuclear DNA and chloroplast DNA, while the pollen only transmits nuclear DNA.
c) Getting high marks in this part was contingent on realising how seed and pollen differ in what DNA they transmit. Marks were awarded to many students who made sensible suggestions about differences in nuclear population structure. But very few students discussed differences between chloroplast and nuclear population structure. A mark was awarded for descriptions of the differences in population structure.

6. The endangered Cross River Gorilla (Gorilla gorilla diehli) has a fragmented habitat and its population size is estimated to be about 250-300. Describe a conservation
strategy based on population genetic principles that could be used to reduce the likelihood of extinction. (indicative line limit: 17 - 22 lines) (12 marks)

The smaller a population is, the stronger the effects of genetic drift (1 mark). Genetic drift in small populations will result in the rapid loss of genetic diversity (1 mark). While the census population is 250-300, the effective population size will be much lower than this, especially as the population is fragmented (1 mark). Genetic drift will act faster in each of the population fragments. Genetic diversity is in general thought to be important for maintaining the adaptability and evolutionary potential of a species (1 mark). Therefore, the main aim should be to increase population size quickly, and to allow the mixing of fragmented populations, perhaps by creating wildlife corridors or translocating individuals (2 mark). The small populations are also at risk of suffering from inbreeding depression (1 mark). The population structure of the species could be assessed using variable markers such as a panel microsatellites (1 mark). This would allow one to assess the connectivity of populations, and also establish which populations are particularly genetically depauperate (and most at risk), which are reservoirs of high genetic diversity (and could act as sources of genetically diverse individuals), and whether any populations are more genetically distinct (and therefore merit greater protection) (2 mark). It may be necessary to genetically rescue the population by introducing gorillas from a different subspecies (1 mark). If so, it would be important to choose a population that is most similar to the Cross River gorilla to avoid introducing maladaptive alleles (1 mark). Despite these measures, it is important to understanding why the Cross River gorilla has become endangered in the first place (hunting? Habitat degradation? Lack of food?) (1 mark). Unless these issues are tackled the genetic measures alone are unlikely to be effective. [credit will be give for any other sensible answers]

Feedback: Most students discussed the problem of inbreeding and also suggested wildlife corridors and translocations. Surprisingly not that many wrote about genetic drift and why it is beneficial to have lots of genetic variation. Only a very small number of students mentioned effective population sizes and characterising the population structure using genetic markers.

7. The silver pheasant is a forest dwelling bird found in southeast Asia. Figure A shows 21 locations from which these birds were sampled for genetic analysis. Ellipses show rough distributions of birds in these areas. Colours of the ellipses correspond to the five clades shown in Figure B. The main rivers in this area are shown as dark blue lines. Figure B shows a phylogeny constructed using mitochondrial sequence from the sampled birds. The five main clades are highlighted with different colours.
a) Evaluate the extent to which rivers and other physical barriers have been important in creating the population genetic structure seen in the mtDNA data (Figure B). (indicative line limit: 8 - 10 lines) (5 marks)

The Hainan Island population is differentiated from the mainland populations (1 mark). The Red river appears to act as a barrier between the green and yellow populations. (1 mark). The Yangtze River may act as a barrier between the red and green clades, but these populations are also geographically distant, so the genetic divergence may simply be a result of isolation by distance. (1 mark) The Mekong and Salween rivers do not appear act as barriers to dispersal (1 mark). Populations 20 and 21 are very close to one another, yet appear genetically distinct (1 mark).

The pheasant samples were also genotyped using 11 microsatellite markers. These genotypes were used in the program STRUCTURE to find the most likely number of genetic groups among the sampled individuals. Figure C below shows the probability that each individual belongs to each of four groups (green, orange, blue and grey). The top bar show which mitochondrial clade (Figure b) each individual belongs to.
b) Describe the differences in population structure seen with mtDNA and nuclear markers. (indicative line limit: 4 - 6 lines) (3 marks)

The red mtDNA clade is not evident from the nuclear data. (1 mark)
The nuclear data shows evidence of genetic admixture between the green and orange mtDNA clades, and between the orange and grey mtDNA clades (1 mark).
Half of the orange mtDNA clade is more likely to be part of the grey nuclear clade (1 mark).

(c) Discuss how these differences could have arisen. (indicative line limit: 5 - 7 lines) (3 marks)

The differences could have arisen from the fact that mtDNA is maternally inherited (and therefore only reflects female population structure) (1 mark), and has a quarter the effective population size of the nuclear genome (resulting in faster coalescence) (1 mark). The effects of genetic admixture may be more evident in the recombining nuclear genome than in the non-recombining mtDNA (1 mark).

Feedback: a) Most students were able to identify the river and sea barriers. Fewer students mentioned lack of a barrier caused by the Salween and Mekong rivers, or that populations 20 and 21 were very genetically different yet close together. Only a very small number of students suggested that geographic distance rather than the Yangtze river might be causing the Sichuan divergence.
b) Most students noted the lack of the red mtDNA clade and also the lack of distinction between the yellow and grey clades (but most did not interpret this as genetic flow between the populations).
c) Many students did not did not answer the question of how the differences between mtDNA and nucDNA could have arisen. Instead these students were answering how differences between populations could have arisen. In these cases no marks were awarded as this is essentially the same as the answer to part a).