Rewarding Learning

eGUIDE//Biology

Biochemistry, Genetics and Evolutionary Trends

Unit A2 2 5.6 Population Genetics and Evolutionary Trends

This e-book is designed to complement other support materials and enhance the understanding of this unit for students at GCE level. The topics covered are in accordance with those topics present in the current specification.

Contents

Population Genetics

5.6

Learning Outcomes from A2 2 5.6

Students should be able to:

- Demonstrate knowledge and understanding of the concept of the gene pool as the total sum of the alleles in a population
- Demonstrate knowledge and understanding of the Hardy-Weinberg equation and its use, applying it to calculate allele, genotype and phenotype frequencies in an outbreeding population.
- Demonstrate knowledge and understanding of the source and maintenance of genetic variation
- Demonstrate knowledge and understanding of selection and its contribution to maintaining polymorphic populations and evolutionary change in populations
- Demonstrate knowledge and understanding of the concept of species and the process of speciation.

What is a population?

(Quote from Introduction to Quantitative Genetics by D. S. Falconer, 1960, Ronald Press.) "A population in the genetic sense, is not just a group of individuals, but a breeding group; and the genetics of a population is concerned not only with the genetic constitution of the individuals but also with the transmission of the genes from one generation to the next. In the transmission the genotypes of the parents are broken down and a new set of genotypes is constituted in the progeny, from the genes transmitted in the gametes. The genes carried by the population thus have continuity from generation to generation, but the genotypes in which they appear do not. The genetic constitution of a population, referring to the genes it carries, is described by the array of gene frequencies, that is by specification of the alleles present at every locus and the numbers or proportions of the different alleles at each locus."

Population genetics is the study of genetic variation in populations. It involves looking at the distribution and change in frequency of genes and alleles in a population over space and time. It is concerned with populations rather than individuals and is the cornerstone of the study of evolutionary biology and speciation (the formation of new species).

The collection of all of the alleles of all of the genes in a population is called the gene pool. Individuals in the population derive their alleles from other members of the gene pool (their parents) and pass their alleles onto their offspring who are also members of the gene pool. So, in effect, population genetics is the study of changes in the frequency of genes and alleles within the gene pool of a population, and how this changes from one generation to the next.

When the frequency of genes and alleles within the gene pool changes, this leads to evolution. A large gene pool has extensive genetic diversity and is better able to withstand

adverse changes and challenges in the environment. Organisms that inbreed have a low genetic diversity and are at risk if environmental conditions change, as they may not have the genetic variability to withstand new stresses. This could result in their extinction.

The Hardy-Weinberg Equation

The Hardy-Weinberg Equation is a model used to predict how gene frequencies are transmitted from generation to generation.
There are a number of factors which can affect the gene pool in a population. These are:

- population size
- genetic drift (when allele frequencies within a population change by chance)
- natural selection
- migration
- mutation
- non-random mating patterns.

Hardy, in England and Weinberg, in Germany concluded in 1906 (working independently), that the gene and genotype frequency in a gene population do not change from generation to generation provided the following assumptions apply.

1. There is a large population size so that it minimizes the effects of genetic drift.
2. There is no natural selection operating which could favour certain alleles/genotypic combinations.
3. There is no net migration into or out of a population.
4. There are no recurring mutations.
5. Mating is random – there are no favourable alleles for traits that will encourage individuals to mate specifically with others in the population over and above that proportionate to their genotypic ratios.

This situation is described as a genetic equilibrium.

In a population;
The frequency of the dominant allele A in a population equals p
The frequency of the recessive allele a in a population equals q

In a Punnett square where two heterozygotes cross, there is

	Female gametes		
Male gametes		p(A)	q(a)
	p(A)	pp or p^2	pq
	q(a)	pq	qq or q^2

So the genotype frequencies after one generation are

$$AA = p^2 \qquad Aa = 2pq \qquad aa = q^2$$

Since these genotypes cover all (100%) of the possible genotypes for this trait in the population (100% =1)

$$p^2 + 2pq + q^2 = 1$$

As there are only two alleles in the gene pool for this trait, then added together the frequencies p and q must make up 100% of the allele frequency

$$p + q = 1$$

So there are two equations that can be used to find out the frequency of each of the alleles and genotypes in a population:

- Firstly look for the frequency of a genotype in a population which is easy to identify
- As one cannot tell the difference between a homozygous dominant (AA) and a heterozygous (Aa) individual for a given trait, the starting point is the double recessive (aa)
- Knowing how many double recessive individuals there are in a population of known size means that the value for q^2 can be worked out
- $p + q = 1$, so the value for p can be worked out
- Once the values for p and q are known the allele frequencies are known, so the other genotype frequencies can be worked out using p^2 and 2pq

Watch 'Solving Hardy-Weinberg Problems' by Bozeman Science (11mins 7secs) which looks at the gene pool, the Hardy-Weinberg Equation and how to solve problems using the Hardy-Weinberg principles.
https://www.youtube.com/watch?v=xPkOAnK20kw

Hardy-Weinberg practice problems by BleierBiology (12mins 35secs) works through three practice questions on mice phenotypes and cystic fibrosis to help explain the concepts and processes involved.
https://www.youtube.com/watch?v=IVGEusDdJGk

Some questions on Hardy-Weinberg for practice are at the following links. Some have the answers attached or on a link so try them first and then check the answers.
http://www.sciencegeek.net/Biology/review/U6HardyWeinberg.htm
https://www.msu.edu/~moscarel/BS110/homework2Fall04_KEY.pdf
http://www.phschool.com/science/biology_place/labbench/lab8/quiz.html
http://www.houstonisd.org/cms/lib2/TX01001591/Centricity/Domain/5363/Hardy%20Weinberg%20Problem%20Set%20KEY.pdf

Question – CCEA May/June 2014 Q7

Cystic fibrosis is a condition caused by a fault in the CFTR protein, a trans-membrane protein responsible for pumping chloride ions out of cells.

If the CFTR protein is faulty, chloride ions may not be pumped out of cells. This results in the mucus immediately outside some cells (for example, cells lining the airways into the lungs) becoming thick and viscous as a consequence of reduced water content.
The symptoms of cystic fibrosis include clogged airways in the lungs and blocked enzyme ducts in the pancreas.

a) Using the information provided, suggest the role of chloride ions in maintaining a normal thin, watery mucus in the lung airways.
b) Around 70% of people with cystic fibrosis in northern Europe have the same gene mutation: three base pairs are missing, with the loss of a phenylalanine amino acid in the protein. The other 30% can have any of up to a thousand different types of

mutations, some with only a single base being affected.
The severity of the condition in individuals varies and depends on the degree of protein malformation, which in turn depends on the type and extent of mutation involved.
In relation to protein structure, explain the link between the type and extent of mutation and the severity of the cystic fibrosis,

c) About 1 in 2500 babies in Northern Europe is born with cystic fibrosis. The condition is caused by an autosomal recessive disorder.
Using the Hardy-Weinberg equation, calculate the percentage of people in Northern Europe who are heterozygous (carriers) for cystic fibrosis.

Source and Maintenance of Genetic Variation

The Hardy-Weinberg equation describes a population that exists in genetic equilibrium i.e. allele frequencies do not change from generation to generation.

However, for evolution to occur, the gene frequencies for a population have to change. A number of factors can play a role in changing gene frequencies such as:

- The ability to survive
- The ability to reproduce

For example, if a mutation occurs which enables an organism to be better adapted to its environment with the result that it can live longer and reproduce more efficiently (be fitter) than others in the population, those animals will survive and pass the mutant allele onto their offspring, who will also have a beneficial phenotype. In this way the allelic frequencies will change in the population, as the beneficial allele's frequency increases with successive populations, and the population will evolve.

A good example of this is the peppered moth in England, which can be light coloured or dark in colour. It was found that prior to industrialisation in England, the light coloured moth was predominant. However, due to industrialisation, soot was deposited on the barks of trees, the darker coloured moths were better camoflagued and less clearly seen by predators on the darker tree trunks and therefore their numbers increased. As a result the dark allele's frequency increased at the expense of the light allele.

Two colour varieties of peppered moth Biston betularia on the bark of a tree. These two colour forms, light (above) and dark (beneath), are a famous example of evolution. In nature the light colour of this moth is most common. A light colour camouflages the moth on lichen-covered bark of trees, protecting the moth from bird predators. In 1840s England, industrial pollution turned trees a sooty black. The darker moth (occurring as a genetic mutation) was better camouflaged on sooty trees, and within 50 years became the commonest form. The light moth was seen and preyed upon by birds, an example of natural selection.

Go to this link to find out more about work on the peppered moth and learn about some of the criticisms and doubts about the scientific methodologies employed in studying this phenomenon.
http://www.millerandlevine.com/km/evol/Moths/moths.html

Mutations can occur which are neutral and have no effect on the viability of the population or its ability to reproduce, or it can be harmful and be selected against in the population and its frequency will fall with successive populations.

Non-random mating, sometimes called assertive mating, will affect genotype frequencies. In humans for example, non-random mating may occur for certain traits such as intelligence, height or physical stature, but not for other traits such as blood type. Individuals do tend to pair with people of a similar intellectual level, (although not always) and studies have found that married couples have higher correlation with regards to intelligence than siblings do.

Migration into or out of a population will alter gene frequencies by either bringing in more copies of an allele that is already present in the population or by introducing a new mutation of an allele through mating with a member of that population. This introduction of new alleles is called gene flow. Migration and subsequent sexual reproduction can therefore increase variation in gene pool/population.

Is there any selective advantage of a dominant allele over a recessive allele?
When looking at autosomal recessive diseases, the homozygous recessive individual, who will therefore exhibit the symptoms of the disease, may have reduced life expectancy. They will be (and perhaps for other reasons), be less likely to reproduce and pass their

defective alleles on to the next generation. As a result the frequency of this allele will decrease in subsequent generations as it is selected against.

However as the heterozygote always contains the recessive allele, it will not disappear in its entirety. The heterozygote may also have some selective advantages over the homozygous dominant individual. This is called heterozygote advantage, where carriers of the disease will be more likely to survive than those individuals without the disease causing recessive allele. In this way heterozygotes are important as reservoirs of genetic variation in populations. This can be seen in the resource below were people who are carriers of sickle cell anaemia, have a survival advantage (are in selection terms 'fitter') against malaria, the disease caused by Plasmodium infection. They will therefore live longer, reproduce and pass this the recessive allele on to future generations. It has been found that sickle cell mutation is, in fact, highly, selected in populations from areas of the world were malaria is very frequent, with sometimes 10-40% of the population carrying this mutation.

This free resource on 'Population Genetics, Selection, and Evolution' by HHMI's BioInteractive.org is a hands-on activity that uses simulations with beads to teach students about population genetics, the Hardy-Weinberg principle and how natural selection alters the frequency distribution of heritable traits. It looks at sickle cell anaemia and malaria as an example of heterozygote advantage. Teaching notes and student handouts are available as downloads. The teaching notes have a link to a short film (14mins 3secs) on 'Natural Selection in Humans' which can be used to help complete the activity.
http://www.hhmi.org/biointeractive/population-genetics-selection-and-evolution

The following link goes to 'Population Genetics and Evolution (Lab Eight)' which explores the effects of selection, heterozygote advantage and genetic drift on a population. It gives typical answers to the type of questions asked in examination situations.
https://sites.google.com/site/kathleenpettinato/apbiology

Mutations
Mutations can be an important source of genetic variation, for example in the peppered moth, by creating new alleles.

Mutations can occur as a spontaneous change or can be caused by environmental factors such as radiation, free radicals, UV rays from the sun, or chemicals such as those found in cigarettes. The factors that can lead to an increase in the frequency of mutations are described as mutagenic agents.

Different types of mutation include:
Gene mutations
- base deletion
- base substitution

Watch these videos for a summary of the different types of gene-level mutation. Concentrate on the substitution and deletion sections.
http://www.nature.com/scitable/topicpage/dna-is-constantly-changing-through-the-process-6524898
This resource from The Concord Consortium has an interactive model that allows you to explore how changing a DNA sequence of bases can change the sequence of amino acids in a protein.
http://concord.org/stem-resources/mutations

Chromosome mutations
- deletion
- duplication
- inversion
- translocation

Watch 'Chromosomal mutations' by Kevin Glass (4 mins 20secs) which illustrates the four types of chromosome mutations – deletions, duplications, inversions and translocations.
https://www.youtube.com/watch?v=XAGxp9j5rtc

'Mutations' by Bozeman science (7 mins 2 secs) looks at the common causes of mutations and the different types of gene and chromosomal mutations.
https://www.youtube.com/watch?v=eDbK0cxKKsk

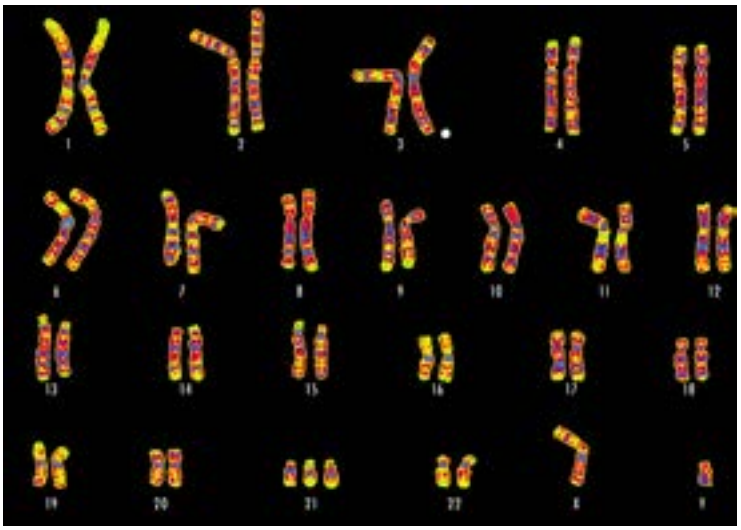
Changes in chromosome number
Aneuploidy refers to a chromosome mutation that causes individuals to have an abnormal number of chromosomes.

It occurs as a result of chromosome breakage or nondisjunction happening in the course of mitosis or meiosis, when homologous chromosomes are separating.

Nondisjunction occurs when homologous chromosomes do not separate properly producing individuals with an extra chromosome or missing chromosomes.
Down's syndrome occurs in humans where the individual, as a result of non-disjunction in an autosomal chromosome, has an extra chromosome 21.

If non-disjunction happens in sex chromosomes, this can result in conditions such as:
- Klinefelter syndrome – males have one or more extra X chromosomes (for example XXY)
- Turner syndrome – females have only one X chromosome (XO)

A chromosome mutation which results in individuals having more than one set of haploid chromosomes is called polyploidy.



Male Down's syndrome karyotype. A karyotype is the complete set of chromosomes of an organism. Humans usually have 46 chromosomes in total, arranged into 23 pairs. Down's syndrome, also called trisomy 21, is caused by the presence of three copies of chromosome 21. Down's syndrome results in mental retardation and flattened facial features. The sex chromosomes, which determine the individual's gender, are at bottom right. Males have an X and a Y sex chromosome, females have two X sex chromosomes.

Selection – polymorphic populations and evolutionary change

The environment in which an organism exists, as we have seen, exerts pressure or stresses on the population, which can lead to natural selection.

It is a phenomenon of populations, that they exhibit polymorphic traits (poly=many ; morph= form), i.e. there are a number of different traits in a population, some of which may enable an organism to live longer, reproduce more and pass their alleles onto greater numbers of offspring in a given environment– we sometimes refer to this as 'survival of the fittest'. This is the basis of natural selection.

Examples of some of the pressures in the environment are:
- The presence of predators.
- Competition for food
- Changing environmental conditions

Some members of the population may be better adapted to deal with such environmental pressures than others, and so will, with time, become more common in a population.
We are going to look at two types of natural selection – stabilising selection and directional selection.

Stabilising selection – individuals with intermediate (modal) traits have a selective advantage as they have the highest 'fitness' for that environment. This type of selection reduces variation as extremes of traits are selected against.

Directional selection – individuals that display a more extreme form of a trait possessing a selective advantage. They exhibit a greater 'fitness' than those with intermediate traits. This causes the allele frequency for that trait to continuously shift in one direction (the direction of that trait).

We represent these types of selection as shifts in the normal distribution curve for the population.

Watch this short animation (2mins 21 secs) Types of Natural Selection, by Teacher's Pet which looks at examples of stabilizing, directional (and disruptive) selection.
https://www.youtube.com/watch?v=64JUJdZdDQo

This online resource entitled "Stabilizing, Directional, and Diversifying Selection" from Boundless Biology gives a description and an example of each type of selection.
https://www.boundless.com/biology/textbooks/boundless-biology-textbook/the-evolution-of-populations-19/adaptive-evolution-132/stabilizing-directional-and-diversifying-selection-535-11742/

Task
Use the information in the animation by Teacher's Pet and webpages by Boundless Biology to produce annotated diagrams/graphs of stabilizing and directional selection. Try to find other examples of stabilising and directional selection and add these to your notes.

Here are some multiple choice questions by Varsity Tutors on natural selection for revision of this topic. The answers are given with a short explanation in each care.
http://www.varsitytutors.com/ap_biology-help/understanding-types-of-selection

Speciation

A species is a group of individuals of common ancestry that closely resemble each other, and are normally capable of interbreeding to produce fertile offspring.

When organisms in a species interbreed, genes can flow through the population of that species.

But how does what has been learnt so far help to explain how new species are formed – a phenomenon called speciation?

Natural selection has been looked at along with the process of how allele frequencies change. In some cases, a whole population can undergo such changes in allele frequencies that it diverges entirely from other populations of the same species. When it does so, and becomes reproductively distinct from the other populations, it can be determined that a new species has been formed.

This link at 'Understanding Evolution' by Berkeley Education, looks at a simplified model of speciation by geographic isolation to explain the process.
http://evolution.berkeley.edu/evolibrary/article/0_0_0/evo_42

This is described as allopatric speciation – new species are formed through geographic isolation with the result that, due to differing environmental and therefore selective pressures, the population (which has become separated) has diverged genetically from other populations of the species which have remained in the original environment.
The next link to the same resource looks at how reduction of the gene flow, even in the absence of a physical geographic barrier, can encourage speciation.
http://evolution.berkeley.edu/evolibrary/article/0_0_0/evo_43

The resource goes on to look at how geographical isolation can begin the process of speciation, but that if the populations are not then reproductively isolated and can still interbreed, then speciation will not occur, as, if the populations are able to mix again and breed feely, the gene flow between the populations will be re-established.
http://evolution.berkeley.edu/evolibrary/article/0_0_0/evo_44

So what are the potential barriers to reproduction, which would account for reproductive isolation between species in speciation? Here are some examples.

- Each species may have its own courtship displays.
- Each may have its own breeding season. Members of the two species do not then have the opportunity to interbreed.
- Interbreeding is prevented by barriers to the formation of the zygote for example the sperm unable to bind to the egg in animals, or the female reproductive organ of a plant is structured in such a way as to prevent the wrong pollinator from landing.
- The two species may be unable to interbreed successfully because of failure of the egg to become fertilised ,or when fertilised, to develop into a viable organism.

This page goes to the evidence for speciation – looking at geographic patterns and experimental results.
http://evolution.berkeley.edu/evolibrary/article/0_0_0/evo_45

'Speciation' an animation by eChalk (2mins 1sec) explains allopatric speciation in a simple and fun way. This may be useful as an introduction to the topic of speciation.

https://www.youtube.com/watch?v=Q2vsG77PZ80

'Speciation: An Illustrated Introduction' by LabofOrnithology (8min 25sec) is a more detailed resource and uses speciation in birds to explore the concepts of geographic and reproductive isolation.
https://www.youtube.com/watch?v=8yvEDqrc3XE

'Clippy Island', is an activity pack designed to reinforce the concepts of natural selection, evolution and speciation. It was originally aimed at KS4 students but would work well as a tool for A level to consolidate work on this topic.
https://www.tes.com/teaching-resource/activity-guide-for-evolution-6330565

CCEA Past paper question May/June 2014 Q4

1. A consequence of sexual reproduction is variation in offspring.
   a) Apart from mutations, identify **three** processes that contribute to variation in a sexually – reproducing organism
   b) Babies produce the enzyme lactase to digest lactose, the disaccharide in milk. However, as they grow into adulthood, some people lose the ability to produce lactase and so cannot digest lactose.
   The ability to produce lactase into adulthood varies with different populations and is linked with milk consumption. In populations which do not keep cows to produce milk (for example in Asia), it is rare for adults to produce lactase. Conversely, in populations which keep dairy cattle (for example in Europe), there are high frequencies of adults capable of producing lactase.
   Lactase production is determined by a single gene with two alleles, one allele coding for lactase production, while the other enzyme fails to code for an effective enzyme. DNA analysis of human skeletal remains shows that the allele for lactase production was absent in adults until 3000 to 8000 years ago, when it was apparently introduced following a mutation. Other investigations indicate that the human populations began using cows as a source of milk 8000 to 9000 years ago.
      i) In terms of its usage in the passage above, explain what is meant by the term 'population'.
      ii) In terms of selection and evolutionary change in populations, explain the initial absence of the enzyme lactase in human adults and the subsequent development of some populations in which lactase is present in many of the adults.

CCEA Past paper question May/June 2015 Q4 (part of)

2. Sickle cell anaemia results in red blood cells becoming more rigid. Many red blood cells in individuals with sickle cell anaemia are therefore less flexible than those in unaffected individuals.
   (a) Suggest the effect that sickle cell anaemia has on blood flow in the capillaries of an affected individual.
   (b) People with two copies of the mutated allele have sickle cell anaemia. These individuals have very restricted oxygen-carrying capacity and have reduced life expectancy.
   People with one normal allele and one mutated allele (i.e. heterozygotes) are said to have *sickle cell trait*. These heterozygotes have less efficient oxygen-carrying capacity but can carry out activities that do not require high energy levels.
   Evidence shows that heterozygotes have some protection against malaria. Malaria is a disease caused by a parasite which carries out part of its life cycle within the red blood cells. The red blood cells in individuals carrying at least one sickle cell allele are not easily penetrated by the parasite. The parasite is transmitted from person to person by mosquito bites. Mosquitoes are particularly common in hot climates, such as much of central Africa, but are unable to live in colder climates.
   Explain why the frequency of the sickle cell allele remains at high levels in parts of Africa yet is very low in northern Europe.

Other useful resources

This powerpoint on 'Introduction to Population and Evolutionary Genetics' is a useful teaching aid.
https://www.ndsu.edu/pubweb/~mcclean/plsc431/overheads/popgen/popgen1.htm

'Population Genetics: When Darwin Met Mendel – Crash Course Biology #18'by Crash Course (11mins 3secs) talks about population genetics, and explains the evolution of populations over time by combining the principles of Mendel and Darwin, and by means of the Hardy-Weinberg equation.
https://www.youtube.com/watch?v=WhFKPaRnTdQ

Some of the terminology used in these flash cards are useful for revision purposes.
https://quizlet.com/20029412/evolution-flash-cards/