AQA Biology A-level

Topic 7: Genetics, populations, evolution and ecosystems

Notes

Inheritance

Keywords:

There are a number of different key words that are required for this topic these are listed and described below.

Genotype – all of the alleles that an organism carries on its chromosomes.
Phenotype – observable characteristics of an organism which the result of the genotype as well as environmental factors.

Most organisms are diploid and therefore carry two alleles for each gene that they possess on each chromosome. Alleles can either be:

Dominant – only a **single allele** is required for the characteristic to be expressed in the phenotype. Whether the organism only has one dominant allele (**heterozygous dominant**) or has two dominate alleles (**homozygous dominant**) the trait will be expressed.
Recessive – the characteristic is only expressed if there is **no dominant allele** present. It therefore means that there must be two recessive alleles (**homozygous recessive**) present in order for the phenotype to be expressed.
Codominant – both alleles are expressed equally and contribute to the phenotype, with there either being a blend of the two characteristic or both characteristics being seen together.

The term **pure breeding** is often used and means that it is a **homozygous pair of alleles**.

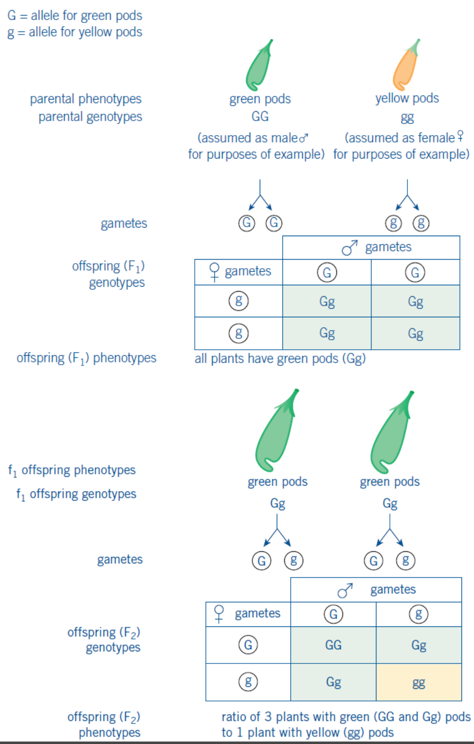
Remember a allele is an **alternative form of a gene** and the **locus** is the specific position of the gene on the chromosome. The two alleles (or more if there is more than two) for the gene are both found at the same loci on the homologous pairs of chromosomes.

Monohybrid inheritance

Monohybrid inheritance is when a phenotype or trait is controlled by a **single gene**. For instance, cystic fibrosis where the individuals with doubly recessive phenotype are affected. The diagram gives the example of crossing pea plants. You can see that there is a cross between a pure breeding green podded pea plant (dominant) and a yellow podded pea plant (recessive).

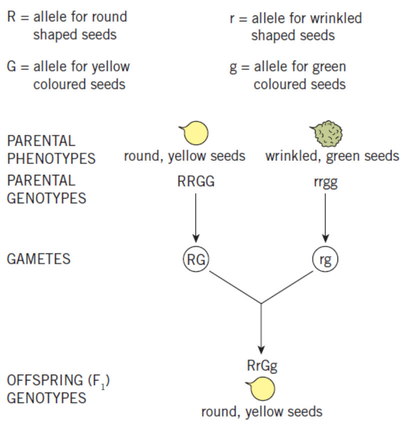
You can see that all the plants will have green pea pods because they all posses the dominant tree pod allele.

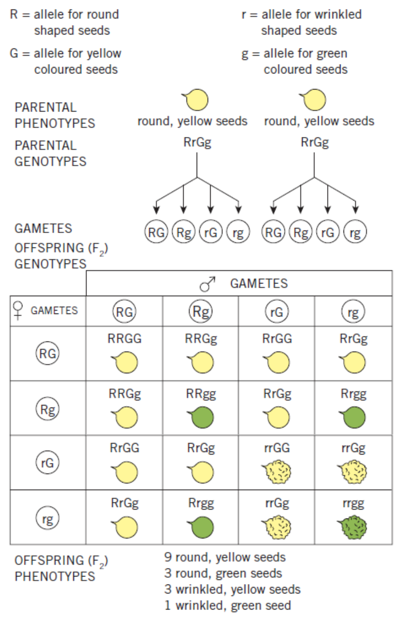
In the event of a second cross as seen in the second diagram a **3:1 ratio** would be observed. The reasons for this is that when the male and female gametes join the way they do so is random and therefore it is down to probability the alleles that are present. In real situations the 3:1 ratio isn't exact, but is close.



Dihybrid inheritance

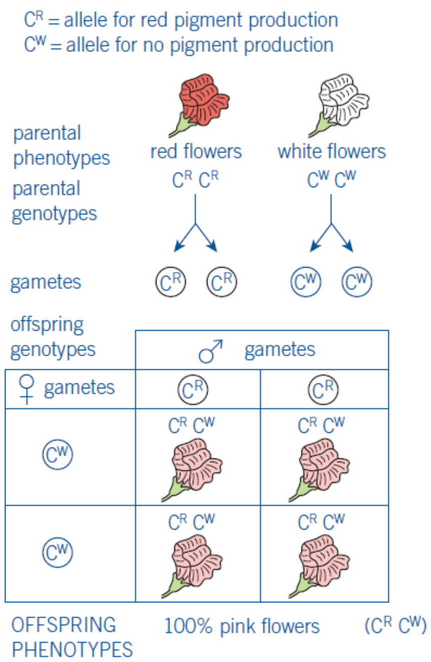
Dihybrid inheritance is when two characteristics are studied and is determined by **two different genes** that are present on two different chromosomes at the same time. For example in the diagram you can see the results from crossing round, yellow seeds (both dominant) with a plant pure breeding for wrinkled, green seeds (both recessive).

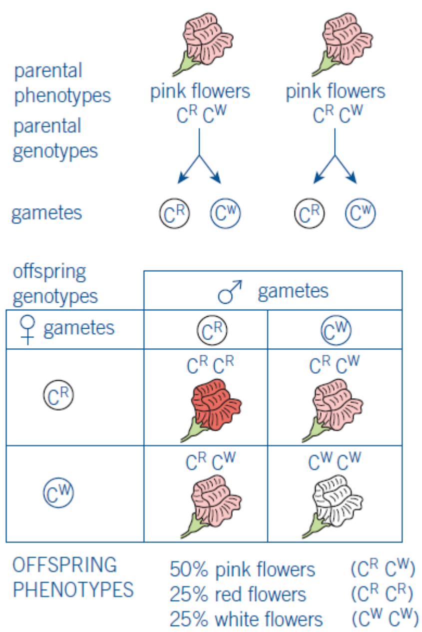




Codominance and multiple alleles

As discussed above **codominance** is when **two phenotypes are expressed** such as when birds have feathers that are both black and white. The diagrams below again detail an example using roses with one having red flowers and the other having white flowers.





A further key example of codominance is shown in human ABO blood groups. There are three alleles that are associated with the immunoglobulin gene (gene I). These lead to different antigens on their surface, these are:

1. Allele I^A - leads to the production of antigen A
2. Allele I^B - leads to the production of antigen B
3. Allele I^o - leads to the production of neither antigen A or B

There are to homologous chromosomes that cause this and therefore two loci. As a result I^A and I^B are codominant, whereas I^O is recessive to both of these. The table shows the possible genotypes that can arise.

Blood group	Possible genotypes
A	I^AI^A or I^AI^O
B	I^BI^B or I^BI^O
AB	I^AI^B
O	I^OI^O

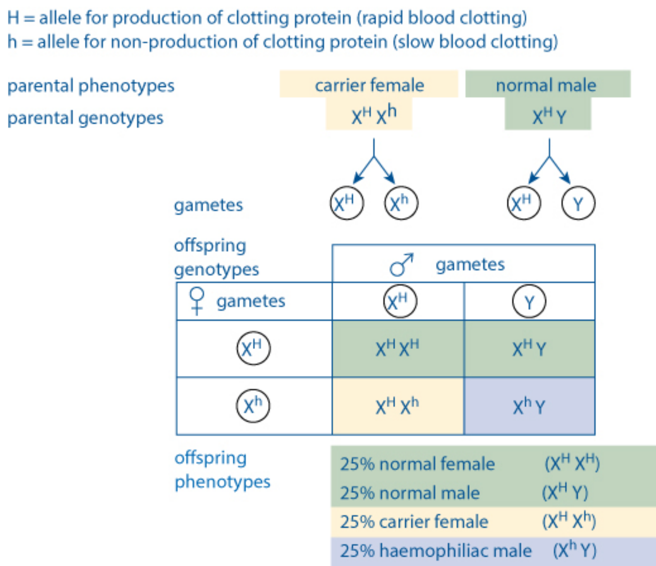
Sex linkage

Sex linkage is the expression of an allele dependent on the gender of the individual as the gene is located on a sex chromosome. Humans have 22 pairs of autosomal chromosomes that are not involved in sex determination, and one pair of sex chromosomes, called either X or Y.

XY - Males - have one of each type of sex chromosome.
XX - Females - have two X chromosomes.

Most sex linked traits are located on the X chromosomes with there being no equivalent locus on the Y chromosome. Therefore females will carry two alleles of sex linked gene, but males will only carry a single allele.

An example of a sex-linked diseases is haemophilia which is a disease in which the blood clots slowly and there may be slow and persistent internal bleeding, especially around the joints. It is caused by a change to the DNA sequence resulting in a faulty protein being created. The digram shows a cross of this disease.

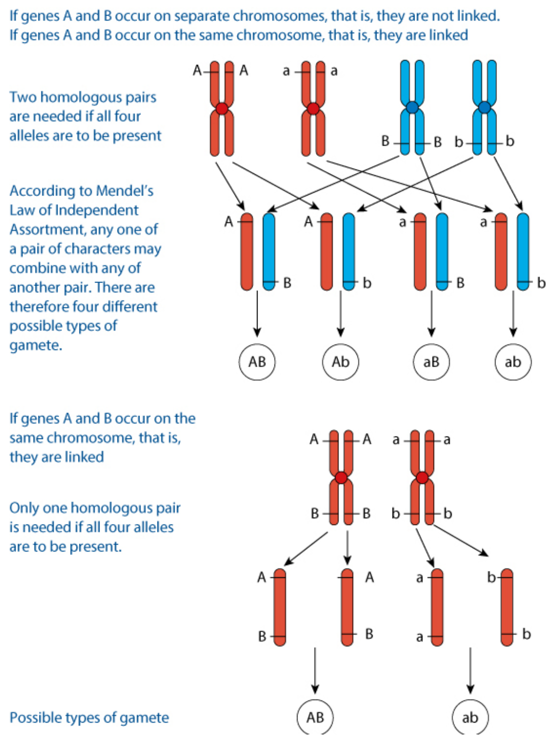


As a male can only obtain the Y chromosome from his father, it means that the X chromosome must come from his mother. As this disease is not found on the Y chromosome it means that this disease is always inherited from the mother in males. If the mother doesn't suffer from the disease but is heterozygous with the alleles then she is a carrier.

Autosomal linkage

In an **autosomal linkage** two or more genes are on the **same autosomal chromosome**. The digram shows a comparison as to whether A and B are on the same chromosomes or on different chromosomes.

From the diagram it can be seen that when A and B are on two different chromosomes then there are four possible combinations of alleles. However if they are on the same chromosomes then there are only two possible combinations of alleles.

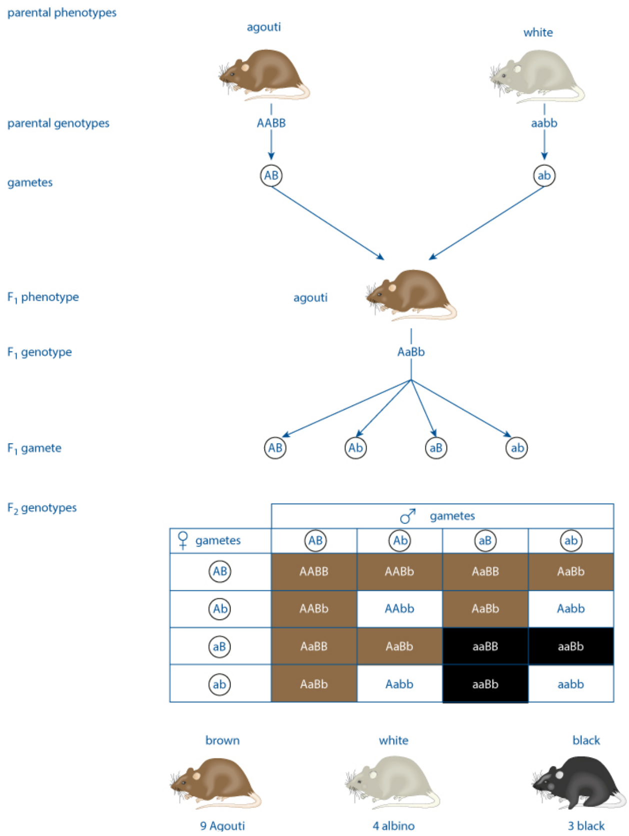


Epistasis

In **epistasis** the interaction of different loci on the gene, **one gene locus affects the other gene locus**. One gene loci can either **mask** or **suppress** the expression of another gene locus. An example is two genes in mice that control fur colour. Genotypes AA or Aa have black bands in their fur, while genotype aa has solid black fur. Gene B controls the expression of gene A. Genotypes BB or Bb will allow expression of gene A, but genotype bb will not. Mice with genotype bb are all white, called albino. This idea is illustrated in the diagram.

- **Recessive epistasis** occurs when the presence of a recessive allele prevents the expression of another allele at a second locus. Recessive epistasis gives the ratio of **9:3:4**.

- **Dominant epistasis** is when a dominant allele at one locus completely masks the alleles at a second locus. Dominant epistasis gives a ratio of **12:3:1**.



Chi-Squared test

The chi squared test is a statistical test which can be used to establish whether the difference between observed and expected results is small enough to occur purely due to chance. It can be used to test the null hypothesis. A null hypothesis is one in which the results of a scientific investigation will produce no statistical significance e.g. there is no difference in the number of times a flipped coin will land on heads and the number of times it will land on tails.

Certain criteria for this must be met, these are:

- The sample size must be sufficiently large enough, that is over 20.
- Used only for data that falls into discrete categories.
- Only raw counts and not percentages, rates, etc can be used

The formula for this test is shown below:

$$X^2 = \sum \frac{(\text{observed - expected})^2}{\text{expected}}$$

The value obtained is compared to the critical value. In chi-square the critical value is p=0.05. Where the value obtained is equal to or greater than the critical value, the null hypothesis is accepted as the difference due to chance is not significant. To read off the value though the degrees of freedom need to be known. These are simply the number of categories minus one.

Whereas in a case where the value is less than the critical value, the null hypothesis is rejected meaning that the difference between observed and expected results is not due to chance, as is significant.

Populations

A population is a group of organisms of the same species occupying a particular space at a particular time that can potentially interbreed. The total number of alleles that are present in a population is known as the gene pool whilst the allelic frequency expresses as a decimal or percentage the proportion of a certain allele in a gene pool.

The Hardy-Weinberg Equation can be used to estimate the frequency of alleles in a population and to see whether a change in allele frequency is occurring in a population over time. In order to do this the following points are assumed:

- no mutations occur to create new alleles.
- there is no movement of alleles into or out of the population by migration.
- the population is large.
- there is no selection, so every allele has an equal chance of being passed to the next generation.
- mating is random.

The formulae for the **Hardy-Weinburg principle** are shown below:

$p + q = 1.0$ **AND** **$p^2 + 2pq + q^2 = 1$**

Where:

p = the frequency of the **dominant** allele (represented by A)

q = the frequency of the **recessive** allele (represented by a)

p^2 = frequency of AA (**homozygous dominant**)

2pq = frequency of Aa (**heterozygous**)

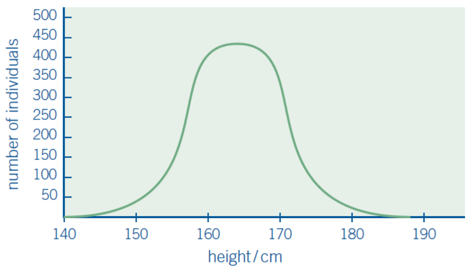
q^2 = frequency of aa (**homozygous recessive**)

Variation in phenotype

There are variations in the alleles of genes for members of the same species due to a number of factors such as:

1. **Random fertilisation** - the gametes that are carrying different alleles will join together randomly.
2. **Meiosis** - meiosis is the nuclear division that creates gametes and means that the alleles will be assorted in the gametes at random.
3. **Mutation** - the mutation of an allele can go on to lead to the creation of another new allele which can then be passed to the next generation

There may also be **environmental influences**, with the majority of phenotypic traits being influences by environmental factors. An example is two plants that possess the same alleles for the flower colour. However an environmental factor of one plant growing in a soil that lacks a certain mineral may mean the pigment in one will not develop so will have a slightly different flower colour. If a large enough sample is taken out of this trait then a **normal distribution** will be shown as seen in the digram showing this for height in adults.



Evolution and natural selection

The **niche** of a species is **its role within the environment.** Species which share the same niche compete with each other and a better adapted species survive. The idea that better adapted species survive is the basis of **natural selection.**

Many organisms have a **unsustainably large number of offspring**, for example fish may lay thousands of eggs which cannot all survive due to limited resources. However Darwin suggested that the reason may offspring are produced is so that there is **greater competition** within the species (**intraspecific competition**) and therefore only those that have the alleles best suited to the environment survive long enough to grow and reproduce passing the alleles on to the next generation.

The **variation in genotypes and phenotypes** within a population increases the chance than a species will survive in a habitat that is changing.

The process of evolution via natural selection is recapped and summarised below:

- There's a **variety of phenotypes** within a population.
- An **environmental change occurs** and as a result of that the **selection pressure changes.**
- Some individuals possess **advantageous alleles which give them a selective advantage and allow them to survive and reproduce.**
- The **advantageous alleles are passed on to their offspring.**
- Over time, **the frequency of alleles in a population changes** and this leads to evolution.

Types of Selection

Selection is the process by which individuals that are better adapted to their environment are more likely to survive and breed. This means that they can pass on their advantageous alleles. Every living organism is subject to selection determined by the conditions which they are living in. There are two key types of selection, these are:

Directional Selection - directional selection occurs when the environmental conditions change and the phenotypes best suited to the **new conditions** are more likely to survive. As a result these individuals will breed and produce offspring. Overtime the mean of the population will move in the direction of these individuals. An example of this is bacteria being resistant to antibiotics. A single bacteria will have had a mutation that meant it was not killed by penicillin as it could produce the enzyme penicillinase. As a result it was able to grow and populate, and the frequency of the allele that enabled penicillinase production increased in the population. Therefore the population moved to have greater penicillin resistance.

Stabilising Selection - In stabilising selection the phenotypes with **successful characteristics** are preserved and those of greater diversity are **reduced**. This selection doesn't occur due to changes in the environment. If the environment stays the same then the individuals closest to the mean are favoured because they have the alleles that have given them the survival advantage. The furthest from the mean are selected against. A example is new born babies weights. Those that have a birth weight of around 3kg are more likely to survive than those at the extremes.

Disruptive Selection - this is the opposite of stabilising selection and in this case **both extremes** of the normal distribution are favoured over the mean. An example of this is where certain large mammals are adapted to surviving long period without food due to increase body fat, whilst small mammals of the same species survive due to their decreased need for food. As a result intermediate sized animals are selected against.

Speciation

Speciation is the process by which new species arise after a **population becomes separated** and **cannot interbreed.**

For instance, **allopatric speciation** is caused by a **physical barrier**. As the two groups become separated and reproductively isolated the result is that the **gene flow is reduced**. Each group experiences a different selection pressure as the environment they live in is different. Over time, the frequency of alleles changes through **natural selection** and the two parts of the population **can no longer interbreed and become separate species**.

An example is the **camel** and the **llama** which both came from a common ancestor that live on the landmasses that are now Africa and South America before continental drift separated these two continents.

Another type of speciation is **sympatric speciation** where new species evolve from a **single ancestral species** when **inhabiting the same geographic region**, for example as a result of a **chromosomal error during cell division** which leads to **reproductive isolation**. Other reasons include mutations that mean courtship behaviour isn't recognised and anatomical changes that mean the penis cannot enter the vagina in mammals.

Populations in ecosystems

An **ecosystem** includes all the organisms **living** in a particular area known as the **community** as well as all the **non-living** elements of that particular environment. The **distribution** and **abundance** of organisms in a **habitat** is controlled by both **biotic** (living) factors e.g. predators, disease and **abiotic** (non-living factors) such as light levels and temperature. Each species has a particular role in its habitat called its **niche** which consists of its biotic and abiotic interactions with the environment.

Variation in population size

There are a number of factors that determine the **size of a population**. Initially a population may grow quickly due to there being **no limiting factors**, however there are many **abiotic** and **biotic** factors that will affect the size and rate of population growth. The abiotic factors that affect population growth are:

1. **Temperature** - each species has a different optimum temperature that it is best able to survive at, the further away from this the fewer individuals that are able survive.
2. **Light** - this is a basic necessity of light, with the rate of photosynthesis increasing as light intensity increases.
3. **pH** - this can have an impact on the action of enzymes with each enzyme having an optimum pH that it can work at. Where the appropriate pH exists there is a larger proportion of organisms.
4. **Water and humidity** - in instances where water is scarce only small populations of adapted species will exist. **Humidity** affects transportation in plants and therefore only those that are adapted to environments where transpiration is high will survive.

An ecosystem supports a certain size of population of a species, called the **carrying capacity**.

Competition

There are a number of different factors that members of the same species compete for. This type of competition is called **intraspecific competition** with factors affecting it being as follows:

- Food
- Water
- Mates
- Shelter
- Minerals
- Light

An example of intraspecific competition is **male robins**. These maintain a large territory to support their families, however in winter when food is short they move into other males territories to obtain enough food to survive.

Interspecific competition on the other hand is where members of different species compete with one another for the same resources. This occurs most commonly when different species occupy the **same niche**. An example is **red and grey squirrels** in the UK.

Predation

Predation is when one species (the prey) is caught and eaten by another species (the predator). The populations of the predator and prey will both affect one another. This is called a **predator-prey relationship** and occurs as follows:

1. When the prey is eaten by the predator the population of the prey falls.
2. This results in the predator population growing, however means that more prey is consumed.
3. Therefore the population of prey reduces and there is increased competition for the the prey between the predators.
4. The lack of food for the predators means that the population falls meaning that less prey is eaten.
5. This allows the population of the prey to recover and therefore the cycle occurs over in a oscillating manner.

The size of a population can be estimated using randomly placed **quadrats**, or quadrats along a **belt transect**, for **slow moving or non-motile organisms**. The abundance of different species can be measured in two main ways these are:

1. **Percentage cover** - suitable for plants or algae whose individuals numbers are difficult to count.
2. **Frequency** - expressed as a decimal or percentage, and is the number of times an organism appears in the sampling area.

For fast moving or hidden animals a **mark-release-recapture** method can be used. The process of mark-release recapture occurs are follows:

1. Known number of species is captured and marked in a way that doesn’t reduce their chance of survival.
2. These marked organisms are then released again into the same area they were caught.
3. After a suitable length of time another known number of organisms are captured, with the number of these that are marked being recored.

The population can then be estimated using the following equation:

$$\text{estimated population size} = \frac{\text{total number of individuals in the first sample} \times \text{total number of individuals in the second sample}}{\text{number of marked individuals recaptured}}$$

This method though relies upon the following assumptions:

- the proportion of marked to unmarked individuals in the second sample is the same as the proportion of marked to unmarked individuals in the population as a whole.
- The marked individuals released from the first sample distribute evenly amongst the remainder of the population and have sufficient time to do so.
- There is a definite boundary to the population so there is no immigration into or emigration out of the population.
- There are few, if any, births and deaths within the population.
- The method of making is not toxic and does not reduce the chance of survival for the population.
- The marks or labels do not rub off during the investigation.

Succession

Ecosystems are very dynamic, for instance succession is the change of one community of organisms into the other. Primary succession occurs when area previously devoid of life is colonised by communities of organisms, for example after the eruption of a volcano which lead to formation of a rock surface. The area is first colonised by the pioneer species such as lichens which are adapted to survive in such harsh conditions. As organisms die, they are decomposed by microorganisms thus adding humus, this in turn leads to formation of soil which makes the environment more suitable for more complex organisms. Over time, the soil becomes richer in minerals thus enabling larger plants such as shrubs to survive. Eventually, a climax community is established which is the final stage of succession, a self-sustaining and stable community of organisms.

Secondary succession occurs in a previously colonised area in which an existing community has been cleared. This type of succession can occur after events such as forest fires. As a soil layer is already present, succession begins at a later stage.

Conservation

Conservation is the human management of the Earth's resources and typically involves the managing of succession. For example the controlled burning of land is done in order to stop the formation of a climax community.