

GENETIC VARIATION AND CHANGE

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Introduction – Genetic variation

Variation occurs throughout nature both within and between species. Variation between organisms is determined both by **inheritable factors** (e.g. DNA / genes / alleles) and **non-inheritable factors** (e.g. available nutrients / light / water / temperature). Humans, through **selective breeding**, have greatly increased the variation within domestic species, resulting in a large number of different *breeds* – there are now:

- over 80 different registered breeds of cat
- over 500 different breeds of dogs.



Short-haired tabby cat – this is the closest cat breed to original 'wild' type cat from which all other breeds have been selectively bred



Five of the 80 cat breeds ...

2 Achievement Standard 91157 (Biology 2.5)

All organisms are classified into different **species**, and given a *scientific name* – e.g. humans are *Homo sapiens*, domestic cats are *Felis catus*, domestic dogs are *Canis familiaris*. Organisms are placed into a species on the basis that *they are able to interbreed to produce fertile offspring*. Species are *reproductively isolated* from other species and therefore share a common **gene pool**. All cats (despite their very different breeds) share a common gene pool and all dogs (despite their very different breeds) share a common gene pool (which is different from that of cats) – therefore, cats and dogs cannot interbreed. However, at times, individuals from closely related species (e.g. members of cat families, dog families, horse families) may interbreed and produce offspring known as **hybrids**. These hybrids are typically *infertile*, e.g.:

donkey (male) × horse (female) → mule

lion (male) × tiger (female) → liger



A zeedonk – offspring from zebra × donkey

Hybrids typically occur in unnatural conditions (e.g. zoos, aquaria), where mating opportunities with individuals of the same species may be limited.

Within a species, forms may exist that, while not reproductively isolated, are sufficiently different to be identified as a **subspecies**.

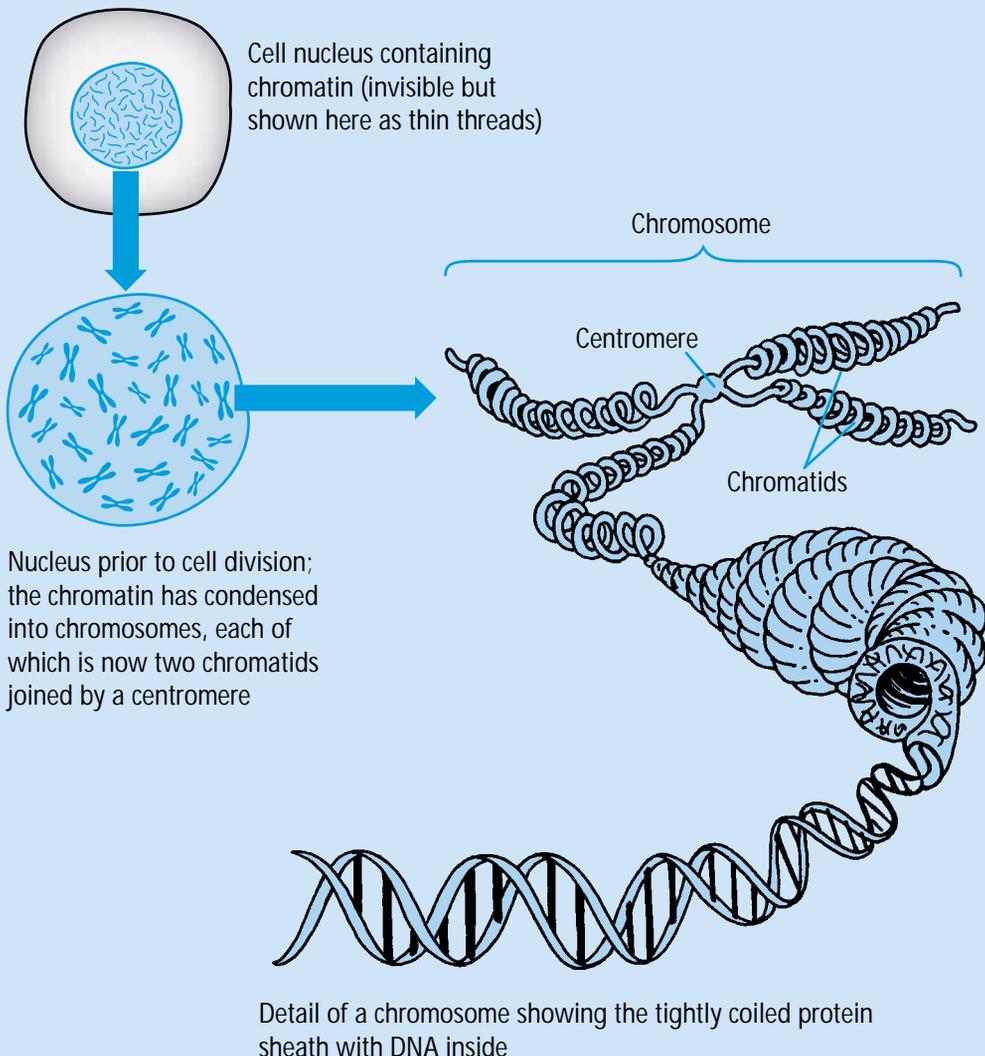
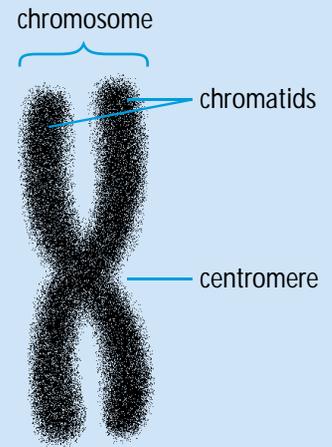
The amount of variation within a species is important to the survival of the species in times of environmental change. Individuals with favourable variations (**adaptations**) that assist them in surviving the change may then breed and their offspring may then inherit these favourable variations. The alleles that are responsible for these variations will *increase in frequency in the gene pool of the species* – biological **evolution** is occurring. The greater the range of alleles in the gene pool, the greater the **genetic diversity** in the species, and the greater the chances of survival of the species.

Sources of variation in a gene pool

Mutations and genetic variation

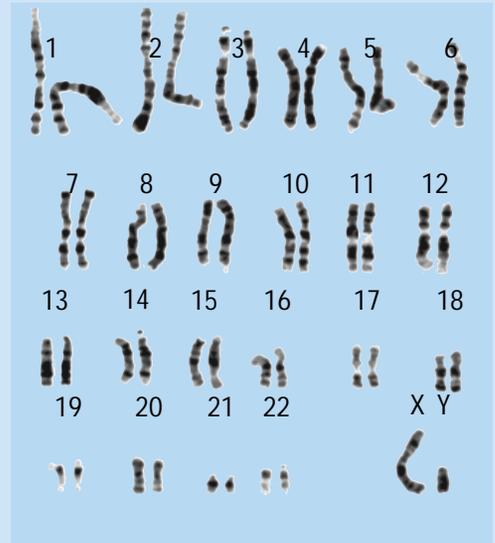
Chromosomes, genes, and alleles

The nucleus of cells contains the **chromosomes**. Different species may have different numbers of chromosomes (commonly 10–50) and their chromosomes may be different shapes and sizes. During normal cell activity, the chromosomes are long thin strands of *chromatin* and are invisible. Chromatin is made of **DNA** wrapped around proteins called histones and contained within a protein sheath. It is only prior to cell division (**mitosis** and **meiosis**) that the long strands of chromatin condense and coil tightly and can be stained and become visible as small, black, sausage-shaped chromosomes when viewed under a microscope. At this stage of the cell cycle, each chromosome occurs as two identical **chromatids** held together by a body called the **centromere**. The two chromatids are the result of DNA replication in the chromatin.



Chromosomes occur in *pairs*, with one member of each pair inherited from the male parent and the other from the female parent. These pairs are known as **homologous pairs** (or homologues). Each chromosome in a pair is identical in length and shape. Humans have 46 chromosomes existing as 23 pairs – 22 pairs are the autosomes which provide information for general body characteristics, while the remaining pair is the sex chromosomes, X and Y. Females are XX, while males are XY.

Sections of chromosomes (more specifically their DNA) may be identified as **genes** which determine characteristics. Homologous chromosomes *have the genes for the same characteristic at the same position on them* – the **locus**. **Alleles** are slightly different forms of a gene and determine traits – e.g. for the characteristic of tongue rolling, two different alleles exist, one determining the *ability* to roll one's tongue and the other determining the *inability* to roll one's tongue. Because chromosomes occur in pairs, alleles for a characteristic occur in pairs.



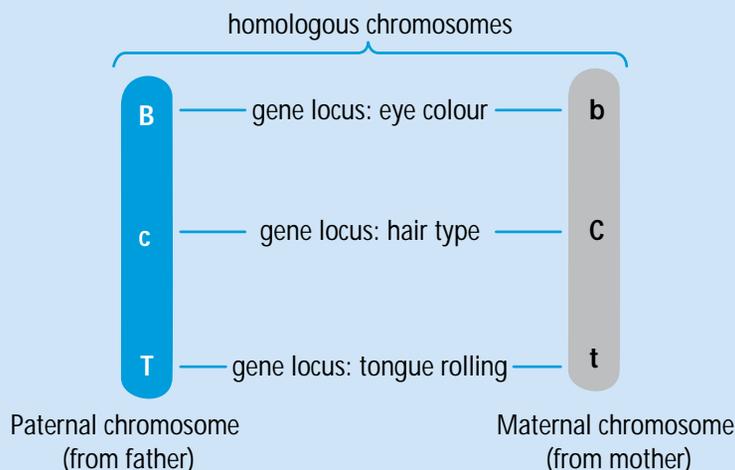
A karyogram showing the chromosomes in a male human



Non-tongue roller



Tongue roller



This is *not* a real example – i.e. eye colour, hair type and tongue rolling genes are *not* on the same chromosome.

The combination of alleles for a characteristic is known as the **genotype**; the visible expression of the genotype in the individual is the **phenotype**. Alleles are typically represented by letters – e.g. tongue rolling **T**, non-rolling **t**.

- ii. Describe the phenotype for each of the genotypes of the kittens.

- iii. Indicate, with reasons, whether any of the kittens has a lethal combination of alleles.

3. NCEA exemplar question

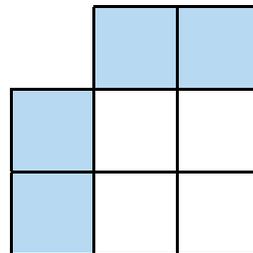
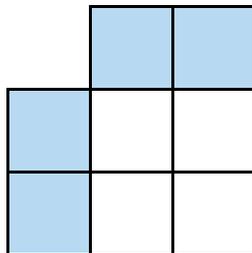
Dexter cattle are the smallest breed of European cattle, with a Dexter being about half the size of a typical Hereford. A mutated allele (**S**) shortens the legs; when two copies of this allele are inherited, the embryonic calves have extreme shortening of the legs as well as severe facial deformities; these calves – known as ‘bulldog’ calves – do not survive, usually being stillborn.

Breeders and farmers who want short-legged Dexters usually breed a short-legged Dexter to a normal-legged Dexter.

Discuss why breeding a short-legged Dexter to a normal-legged Dexter is the preferred breeding combination for the production of short-legged Dexter cattle. Use the Punnett squares to support your answer.



Short-legged Dexter cow



Dihybrid inheritance and genetic variation

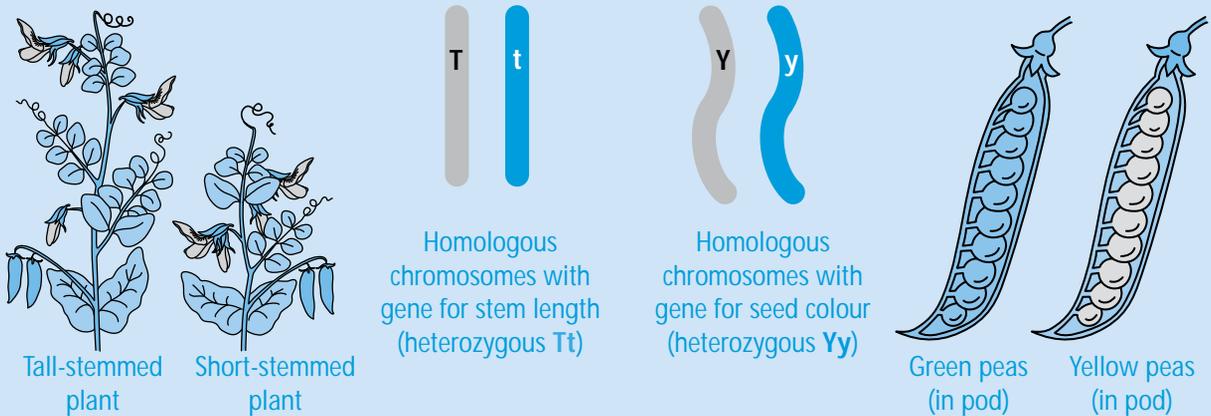
Dihybrid inheritance

Dihybrid inheritance is the inheritance of *two characteristics* controlled by *two genes*. The genes may have two or more different alleles. The genes may be on the same chromosome so are **linked**, or they may be on different chromosomes so are not linked.

The principles of monohybrid inheritance patterns for *complete* or *incomplete* or *co-dominance* or *multiple alleles* all apply to dihybrid inheritance when the genes are *not* linked. However, *only* complete dominance in dihybrid inheritance is assessed in AS 91159 (Bio 2.7).

Dihybrid inheritance (no linkage)

In pea plants, tall stem (**T**) is dominant to short stem (**t**); yellow seeds (**Y**) is dominant to green seeds (**y**). The genes for stem height and seed colour are on separate chromosomes.



P generation: cross a pea plant pure-breeding for tall stem and yellow seed (**TTYy**) with a plant pure-breeding for short stem and green seeds (**ttyy**).

Each gamete must receive an allele for height as well as an allele for seed colour. The combination of alleles is random – a result of independent assortment in meiosis. The genotypes are homozygous for both plants, so the tall yellow plant can only produce gametes with **T** and **Y** alleles and the short green plant can only produce gametes with **t** and **y** alleles. Therefore, the P cross to produce the F1 generation is:

TTYy × **ttyy**

	ty
TY	TtYy

The F1 offspring are all heterozygous **TtYy**, and all are tall plants with yellow seeds.

These heterozygous **TtYy** plants can produce four different gametes:

- **T** with **Y** to give **TY** gamete

5. NCEA exemplar question

In the sweet pea plant, *Lathyrus odoratus*, the allele for purple (**P**) flower colour is dominant over the allele for red (**p**) flower colour. A second gene determines the shape of the pollen. Long (**L**) pollen is dominant over round (**ℓ**) pollen.

- a. A purple, long-pollen plant (**PPLL**) was crossed with a red, round-pollen plant.
- i. Give the genotype of the F1 generation.

- ii. Work through a Punnett square of a cross between two of these F1 plants to give the possible genotypes of the F2 generation and the expected ratio of their phenotypes.

- b. However, when the F1 plants were crossed, the observed phenotypic ratio differed from the expected ratio – see table alongside.

	Observed phenotype ratio
Purple, long (PpLℓ)	12
Purple, round (Ppℓℓ)	1
Red, long (ppLℓ)	1
Red, round (ppℓℓ)	2

It was concluded that the gene for colour and the gene for pollen shape were not independently assorting as expected, therefore the genes must be linked.

Discuss why the expected phenotype ratio you calculated in **a. ii.** is different from the observed ratio given in the table. In your discussion, you need to:

- describe *linkage* and explain why linked genes do not assort independently
- explain how crossing over produces recombination
- explain how crossing over resulted in the low occurrence of purple, round-pollen and red, long-pollen phenotypes.

Write your answer on your own paper.



Changes to the allele frequency in a gene pool

The **gene pool** is the *total number of alleles present in a population* (although we can also refer to 'the gene pool of a species').

Genetic change refers to the *changes in frequency of alleles* in the gene pool of a population. The frequency of the alleles in the gene pool results from the processes of:

- mutation
- natural selection
- migration
- genetic drift.

The frequency of an allele in a gene pool is calculated using:

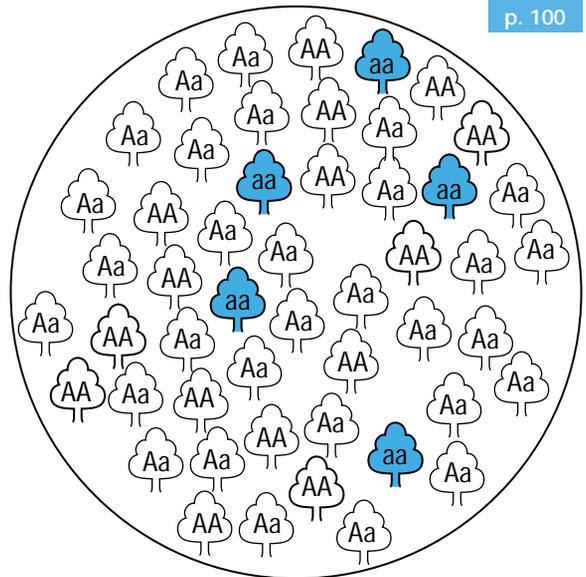
$$\text{frequency of an allele} = \frac{\text{occurrence of an allele}}{\text{total number of alleles}}$$

Mutations are the *source of new alleles* in the gene pool.

Allele frequency

The diagram shows a small population of trees where allele **A** produces sharply pointed leaves while allele **a** produces rounded leaves.

1. Calculate the frequency of the **A** allele and the **a** allele in the gene pool of this population.



2. Suppose that the sharply pointed leaves in the tree population arose from a gene mutation in an individual tree resulting in the **A** allele. Explain how the allele entered the gene pool and suggest reasons why it has become the more common allele in the gene pool.

Migration

Migration is the movement of individuals from one population to another.

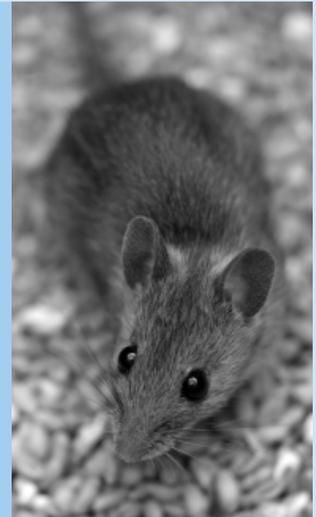
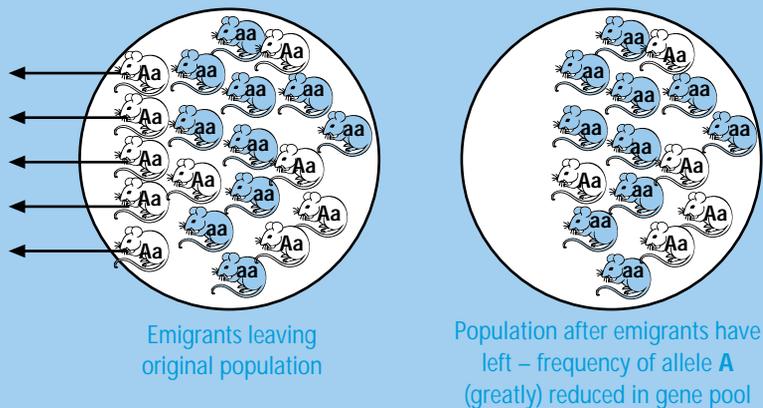
immigration = individuals migrate *into* a population

emigration = individuals migrate *out of* a population

Both processes allow for **gene flow** between populations. *Gene flow may change the frequency and/or the range of alleles in populations.*

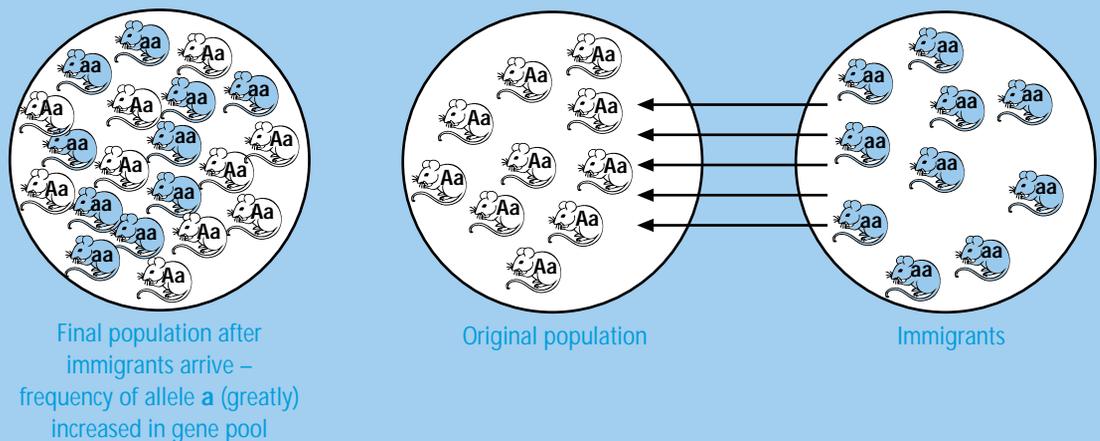
Emigration may remove alleles from a population, *reducing a population's genetic diversity.*

Example: Mice emigration



Immigration may add new alleles to a population, *increasing a population's genetic diversity.*

Example: Mice immigration



If populations are large, migration may have little or no effect on allele frequency. However, if populations are small, migration may have a marked effect on allele frequency.

ANSWERS

Introduction – genetic variation (page 3)

1. a. A group of organisms that can freely interbreed to form fertile offspring; are reproductively isolated from other groups and share a common gene pool.
- b.
- | Common name | Scientific name |
|------------------------|------------------------------|
| kiore / Polynesian rat | <i>Rattus exulans</i> |
| brown kiwi | <i>Apteryx mantelli</i> |
| kakapo | <i>Strigops habroptilus</i> |
| kauri | <i>Agathis australis</i> |
| silver fern / ponga | <i>Cyathea dealbata</i> |
| rimu | <i>Dracodium cupressinum</i> |
- c. Dogs and wolves are very closely related with dogs having evolved from wolves, likely to be as a result of early humans domesticating wolves. Over thousands of years, dogs have become very different from wolves, and now they appear to be different species based on behavioural and structural differences. However, dogs and wolves can still interbreed, although this does not normally happen naturally in the wild. Therefore, based on the species criteria for reproductive isolation as well as its evolution, many researchers believe that it is more accurate to make dogs a subspecies of wolf (*Canis lupus familiaris*) rather than a separate species (*Canis familiaris*).
2. a. Inheritable variations are determined by genetic factors (DNA/ genes/alleles) which are inherited from the parents and will be passed on to the offspring, while non-inheritable variations are determined by environmental factors and are therefore not inherited from the parents nor can they be passed on to the offspring.
- b. i. Colour of skin (and hair) as a result of exposure to sun, height as a result of dietary factors, certain cancers as a result of exposure to mutagens.
- ii. Colour of hair and eyes, type of hair, blood groups, height, structure of body organs, etc.; all our essential structures and how they function are determined by our genes.
- c. Most likely to be from inherited variations as, while siblings have the same parents, they get a different assortment of alleles (from independent assortment in meiosis and the random nature of fertilisation) which means that, while there will be similarities, they will also vary from one another (and their parents). As siblings share a common environment most of the time, they are likely to display similar effects of environmental factors (e.g. dietary).
- d. i. Shape of leaves (oval or triangular), type of leaf margin (smooth or toothed), shape of 'teeth' on leaves (smooth or pointed).

ii.

Common name	Scientific name
red beech	<i>Fuscospora fusca</i>
silver beech	<i>Lophozonia menziesii</i>
mountain beech	<i>Fuscospora cliffortioides</i>
hard beech	<i>Fuscospora truncata</i>
black beech	<i>Fuscospora solandri</i>

- iii. Silver beech (*Lophozonia menziesii*) as it is a different genus (*Lophozonia*) from the other four beeches, which are all in the genus *Fuscospora*.
- iv. Temperature is the most likely environmental factor. Temperature decreases as altitude increases, therefore trees living higher up mountains will live in a colder environment therefore are likely to grow slower and be smaller than trees in the warmer lower altitudes over same time frame, regardless of genes for height.
3. a. An individual resulting from the breeding of members of two different species; is typically infertile.
- The term *hybrid* may also be used for the offspring from two different breeds (e.g. domestic dogs – spaniel × fox terrier) within a species.
- b. Wolphin is a hybrid as mother is a dolphin and father is a false killer whale – two different species. Breeding occurred in artificial conditions (within confines of a large aquarium) and there were no other member of the species for either to breed with.
- c. Wolphin has one set of chromosomes (and genes) from one parent, and another, different set of chromosomes (and genes) from the other parent. If most genes are expressed more or less equally then the offspring will be intermediate between both parents.
- Diploid chromosome number (2n) of *both* bottlenose dolphins and false killer whales is 44 – explains why wolphin possible.
4. a. Genetic diversity refers to the range of different alleles in the gene pool of a population (or a species).
- b. The greater the diversity in a gene pool, the greater the range of phenotypes in the population. This increases the chances of adaptations being present in the population that can cope with a change in the environment. Individuals with successful adaptations will survive a change and breed and pass on (the alleles for) these adaptations.
- c. If genetic diversity becomes very low then it reduces the chances that members of the population may have the adaptations needed to survive a change in the environment such that the population (and/or species) will not survive (i.e. becomes extinct).

Chromosomes, genes, and alleles (page 9)

1. a. *Chromatin* refers to the very long thin threads of DNA (and protein) in the nucleus of the cell in the stage of the cell cycle

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