

a behaviour that benefits all participants because all share at least some of the same genes. By helping defend, feed, or care for relatives, and individual improves the chances that some of its own genes will be passed on.

- Cooperative attack (hunting) behaviour. The orca benefit from this behaviour because it enables them to bring down a prey animal with relatively little effort/energy expenditure on the part of any one individual. If an individual orca had to hunt alone, its chance of success would be lower and it would expend more energy in chasing and securing its prey. A group attack ensures success with little effort.

### 75. The Role of Variation in Populations (page 103)

- Variation describes the diversity of phenotypes within a population.
  - Variation arises through mutation and sexual reproduction (meiosis and fertilisation).
- Variation is the raw material on which natural selection acts. Selection acts on the phenotype (the result of genotype and environment), selecting for or against particular allele combinations, enhancing or reducing the representation of these in the next generation. In this way, allele frequencies change from generation to generation (evolution).
- Fitness is a measure of the genetic contribution an individual makes to the next generation. It incorporates not only survival but reproductive success. Fitter individuals make a greater contribution to the next generation.
  - Selection pressure is the combination of factors in the environment that determine whether an organism will be more or less successful at surviving and reproducing (i.e. its fitness).

### 76. Mutations (page 104)

- A mutation is a change to the base sequence of an organism's DNA.
- Mutations that benefit the individual by improving fitness will be retained (selection will favour them remaining in the gene pool). Mutations that are detrimental to fitness will reduce survival and reproductive success and so become less common in subsequent generations. Eventually, they will be eliminated. **Teacher's note:** Deleterious alleles may be retained if they are carried as recessives at low frequency or if they offer heterozygote advantage (e.g. sickle cell trait).
- A silent mutation is a point mutation that has no phenotypic effect, i.e. it does not change the protein encoded. **Teacher's note:** A silent mutation may be 'silent' because (1) it does not change the amino acid sequence, (2) it results in an amino acid with the same properties, or (3) it occurs in introns, outside the protein-coding region.
  - Silent mutations can be carried without being subject to selection pressure. However, they do involve a change in the code and the base changes may be subject to different selection pressures and prove advantageous in a future environment. **Teacher's note:** Most 'silent' mutations to exonic DNA are now not regarded as truly silent as they can affect translational machinery and the efficiency of gene expression. Silent mutations can be neutral but are not necessarily so. They may affect the fitness of the individual (for better or worse) in a different suite of conditions.

### 77. Beneficial Mutations (page 105)

- There has not been enough time for the mutation to be passed through the human population by descent. The human population is also very large so many generations would be needed.
- In cattle-raising populations there would have been a strong selection pressure to retain the ability to digest milk into adulthood as it would have been a readily available source of nutrition in those populations.
  - Milk contains sugars and fats for energy, protein for growth and development, and calcium for bone deposition and development. Being able to drink milk provides a dietary

advantage over those who cannot drink milk.

### 78. Sickle Cell Mutation (page 106)

- The Hb<sup>S</sup> mutation results in a haemoglobin protein with reduced solubility at low oxygen tensions, so it precipitates out of solution when oxygen levels fall.
- Two copies of the Hb<sup>S</sup> mutation produces sickle cell disease. The red blood cells are deformed into a sickle shape, which causes clumping and prevents the cells moving freely through capillaries. Sick cells are destroyed leading to anaemia and there are many circulatory and organ problems. The condition is lethal.
  - Heterozygotes carrying one Hb<sup>S</sup> allele are carriers for the disease. They have enough normal red blood cells that they are physically unaffected by the mutation (unless in low oxygen environments such as at altitude).
  - The alleles are codominant. Both are equally expressed.

### 79. Heterozygous Advantage (page 107)

- Carriers of the Hb<sup>S</sup> allele are protected somewhat against malaria. The Hb<sup>S</sup> allele results in red blood cells with low potassium levels which kills the malaria parasite, *Plasmodium*.
- People who are heterozygous for the sickle cell gene are somewhat affected by sickle cell anaemia but have considerable resistance to malaria which is widespread in the region. This heterozygous advantage maintains the mutant allele at a relatively stable frequency in the population despite its deleterious effects. The stable coexistence of both the sickle cell allele and the normal allele represents a balanced polymorphism.
- Malaria is widely distributed throughout central Africa, the Mediterranean, Middle East, and tropical and semi-tropical Asia.
  - The distribution patterns for malaria and Hb<sup>S</sup> allele do not completely match up and really only coincide in Africa. The Hb<sup>S</sup> allele is only present in Africa.
- Hb<sup>E</sup> allele: Present in India and SE Asia.  
 Hb<sup>S</sup> allele: Present in some regions of Africa.  
 Hb<sup>C</sup> allele: Present only in NW Africa.

Thalassaemia: Present in southern and northern Africa, through southern Europe, the Middle East, and right through to SE Asia.

These genes persist in the population because they occur in regions where malaria exists. In each case, the mutation provides some protection against the malaria parasite and reduces the incidence or severity of the malaria infection.

### 80. Gene Duplication and Evolution (page 109)

- Gene duplication is the duplication (repetition) of a region of DNA that contains a gene or genes.
- Virtually every species shows genomic evidence of gene duplication, often more than once.
- Like mutations, only those duplications that provide an adaptive advantage will be retained. Disadvantageous duplications will be lost.
  - A gene duplication can remove adaptive conflict, providing two copies of a gene so that each can respond to differing selection pressures. In a new environment, one gene copy can respond to the new selection pressures present.
- It would be an advantage to have two genes performing the same function when there is a high demand for a gene product, e.g. protein (the more is better scenario). More pigment might be useful against high UV for example.
- Duplication of the ancestral globin gene was followed by (different) mutations to each copy, which produced the  $\alpha$  and  $\beta$  haemoglobin genes. This was important because it allowed the evolution of a multiunit protein using the  $\alpha$ - and  $\beta$ - haemoglobins. There were then transpositions of these genes to different chromosomes and further duplications and mutations to produce a large family of globin proteins, one family on chromosome 16 and one on chromosome 11.
- AFPIII protein and SAS protein have similar structures and

