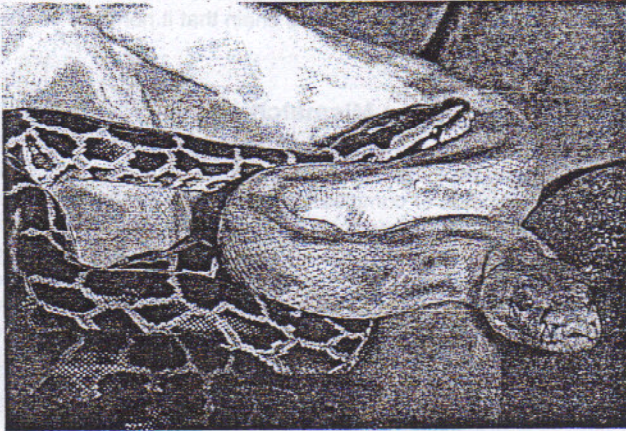


For Harm or Benefit?

It is not correct to assume that all mutations are harmful. There are many documented cases where mutations conferring a survival advantage have arisen in a population. These **beneficial mutations** occur mostly amongst viruses and bacteria, but some (such as pesticide resistance) occur in multicellular organisms (especially those, like insects, with short **generation times**).

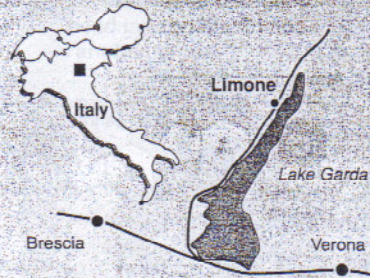


Harmful Mutations

There are many well documented examples of mutations with harmful effects. These include the mutations giving rise to **cystic fibrosis (CF)** and **sickle cell disease**. The sickle cell mutation involves a change to only one base in a DNA sequence, whereas a common CF mutation involves the loss of a single triplet. The malformed proteins resulting from these mutations cannot carry out their normal functions. **Albinism** is caused by a mutation in the gene producing an enzyme in the metabolic pathway to **melanin**. It occurs in a large number of animals. The example above shows an albino python. Albinos are uncommon in the wild because they tend to be more vulnerable to predation and damaging UV radiation.

Mutations that are neither harmful nor beneficial are called **neutral (silent) mutations**. Neutral mutations can be virtually impossible to detect because they are not associated with any observable change in phenotype. If there is no selective pressure against a mutation, it may be carried silently in the population and prove to be beneficial (or harmful) at some time in the future.

High blood cholesterol is implicated in the formation of fatty plaques in the coronary arteries and in the development of cardiovascular disease (CVD). The people of Limone show a higher (than usual) resistance to CVD.



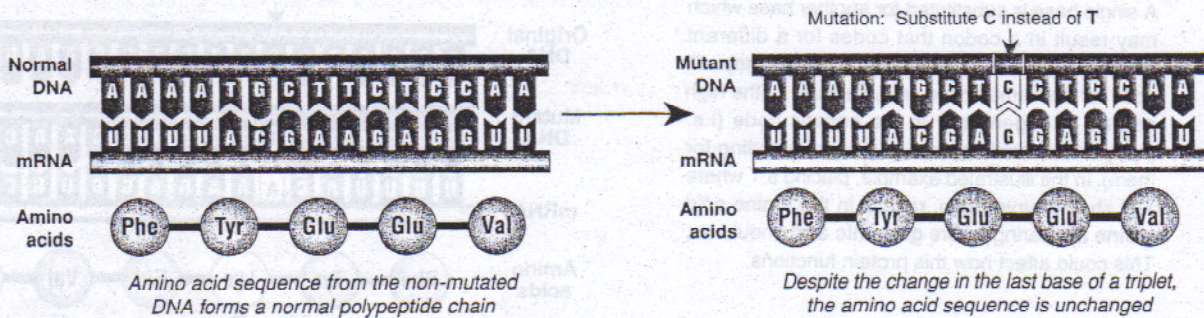
Beneficial Mutations

A well documented example of a beneficial mutation is the **tolerance to high cholesterol levels** in humans. In the small village of **Limone**, about 40 villagers have extraordinarily high levels of blood cholesterol, with no apparent harm to their coronary arteries. The village has 980 inhabitants and was largely isolated from the rest of the world until recently. The 40 villagers possess a mutation that alters a protein by **one amino acid** and makes it ten times more effective at mopping up excess cholesterol. Excess cholesterol is always disposed of, no matter what the dietary intake. All carriers of the mutation are related and are descended from one couple who arrived in Limone in 1636.

Neutral Mutations

Neutral mutations may be important in an evolutionary sense because they can be passed from generation to generation and may directly (or indirectly through subsequent mutation) be

subject to selection pressure in the future. The example below shows how a change to the DNA sequence can be silenced if there is no change to the resulting amino acid sequence.



1. (a) Explain the difference between neutral (silent), beneficial, and harmful mutations: _____

(b) Identify which of these mutations is the most common and suggest why: _____

2. Explain how the mutation that 40 of the villagers of Limone possess is beneficial under current environmental conditions: _____
