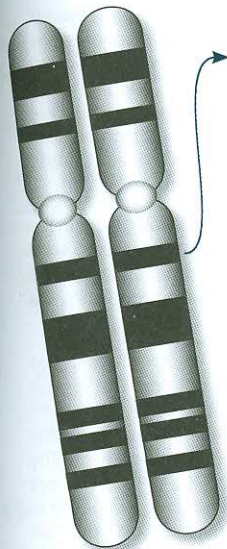


Mutations and Development

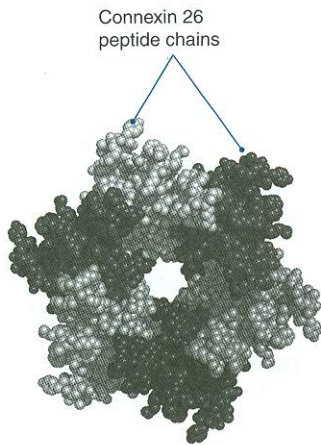
Mutation of the DNA molecule can lead to abnormal development in the individual. Every time a DNA molecule is copied (DNA replication), there is a chance that a base or series of bases will be copied incorrectly. Some changes in development can be mild and have little effect, others can be of greater significance. DNA replication has a low **error rate**, with only one mistake for

every billion base pairs copied. Errors that have no effect on the organism or its offspring are called **neutral mutations**. Other errors may create new **alleles**, some of which may be beneficial, although most will be detrimental to development. One example is the most common form of genetic hearing loss (called NSRD), which accounts for up to 50% of childhood deafness.

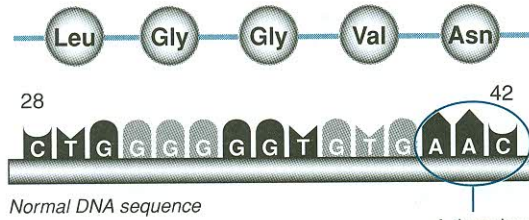


Chromosome 13

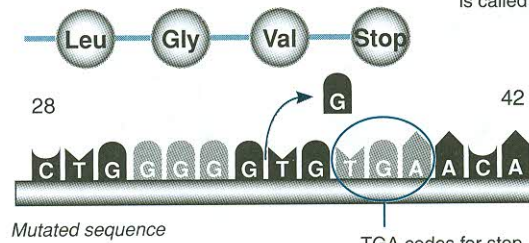
The gene that codes for the peptide chain connexin 26.



Six connexin 26 peptide chains join together to form a connection between cells. If these proteins are not made correctly, they are unable to form the final structure.

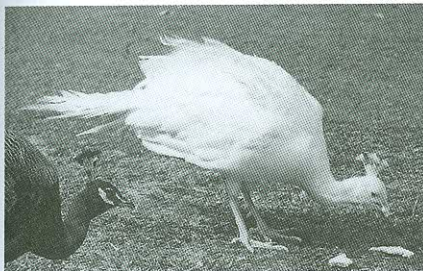


A three base sequence coding for an amino acid is called a **codon**



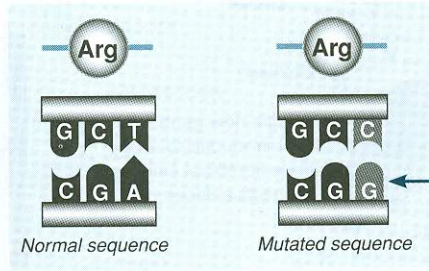
The most common mutation in this gene is **deletion of the 35th base (G)**. DNA is read in groups of three bases, so deleting the guanine alters the reading sequence. The result is a short peptide chain, which cannot function correctly and results in deafness.

The deletion of this one base causes the formation of a **recessive allele**. Deafness results when there are two recessive alleles.



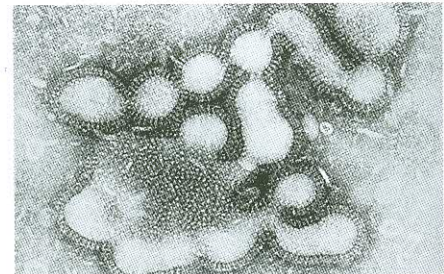
Harmful Mutations

Most mutations cause harmful effects, usually because they stop or alter the production of a protein (often an enzyme). Albinism (above) is one of the more common mutations in nature, and leaves an animal with no pigmentation.



Silent Mutations

Silent mutations do not change the amino acid sequence nor the final protein. In the genetic code, several codons may code for the same amino acid. Silent mutations are also **neutral** if they do not alter the fitness of the organism.



Beneficial Mutations

Sometimes mutations help the survival of an organism. In viruses (such as the *Influenzavirus* above) genes coding for the glycoprotein coat are constantly mutating, producing new strains that avoid detection by the host's immune system.

Regulation of Gene Expression

- How can changes in a DNA sequence occur? _____
- How can a mutation in a single base be as damaging as a mutation in a sequence of bases? _____
- Explain how mutation can be harmful or beneficial: _____

