Chap.3 Doc.3 Polymorphic genes in a population

Polymorphic genes are genes that possess many alleles in a population and occupy the same locus in chromosomes.

A gene is considered polymorphic if many alleles are present in the population, each with a frequency higher than 1%

Polymorphic genes

There are many examples of the polymorphic gene but we will state only three:

- ✓ The polymorphic gene of the ABO system:
  The wild allele, present on the long arm of chromosome 9, undergoes substitution mutations which result in two alleles: A and B, and it undergoes a deletion mutation which leads to the allele O. Each allele codes for a given antigen, the three antigens are formed of a substance H and a sugar is added to this substance due to the presence of the alleles A and B but the allele O does not lead to the addition of a sugar to H substance.
- ✓ The polymorphic gene of the HLA system:
  This gene, present on chromosome 6, is very polymorphic due to the presence of many alleles of the six genes: DP, DQ, DR, B, C, A. All of these genes code for membrane proteins expressed on the surface of nucleated cells (HLA molecules).
- ✓ The polymorphic gene of β globin:
  This gene codes for β globin, which is involved in the synthesis of hemoglobin. Thalassemia is the most well-known disease deriving from the mutation of this gene

Probing the activity p.63

1. Each of the six HLA loci (DP, DQ, DR, B, C, and A) contains two alleles in 2^n different combinations ("n" being the number of alleles at each locus). This makes it practically impossible for two individuals to have the exact combinations of alleles at all six HLA loci. On the other hand, identical twins have exactly the same genotype, and hence exactly the same alleles at each of the loci in the MHC complex.

2. A wild-type allele is the allele that codes for the most common phenotype in a population.

3. The polymorphic gene of B-globin gene is due to the presence of diverse alleles in the human population. These alleles are the result of different types of mutation by insertion, deletion or substitution of DNA nucleotides.

4. The severity of β-thalassemia depends on the site and extent of mutation of the β-globin gene. For example, a mutation that leads to the substitution of an amino acid by a characteristically similar amino acid (i.e. a basic amino acid lysine for another basic amino acid arginine) may lead to a mild aberration of the function of the β-globin protein, and hence the individual may be mildly thalassemic. On the other hand, a mutant β-globin protein may have resulted from a deletion of a long stretch of DNA from the β-globin gene. This may lead to a severely thalassemic individual. One must note as well the severity of thalassemia also depends whether the individual is the carrier of a single mutant allele or two.