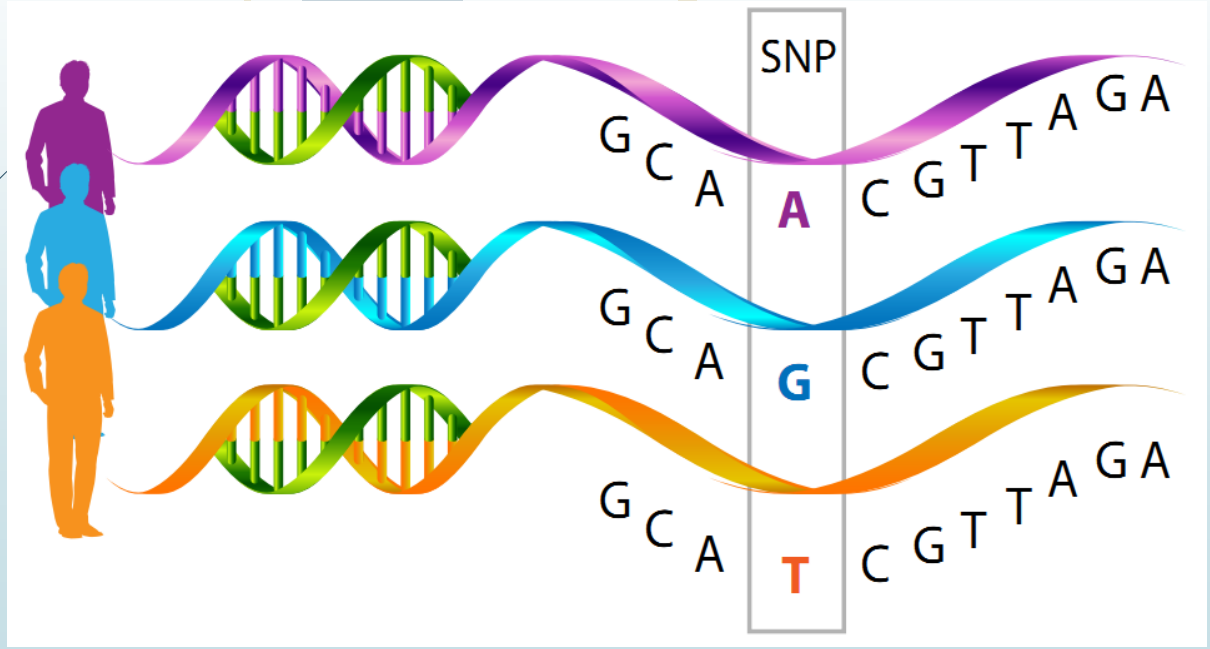
Chapter 3: Genetic Variation and Polymorphism

- Document 1: Mutations and the Environment
- Document 2: Mutation and Multiple Alleles
- Document 3: Polymorphic Genes in a Population
- Document 4: Detection of Genetic Polymorphism
- Document 5: Genetic Identity of Individuals

Document 3

Polymorphic Genes in a Population



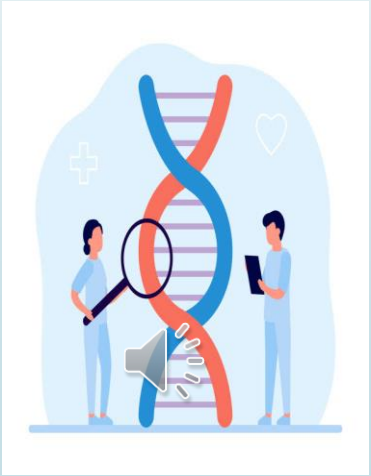
- A polymorphic gene is a gene which exists in multiple alleles within a population.
- A gene is considered polymorphic if many alleles are present in the population each with a frequency higher than 1% .
- The allele that codes for the most common phenotype is considered as the wild-type(normal) allele.

- **Examples on polymorphic genes:**

  → Gene coding for ABO blood group.

  → Major Histocompatibility Complex (MHC)

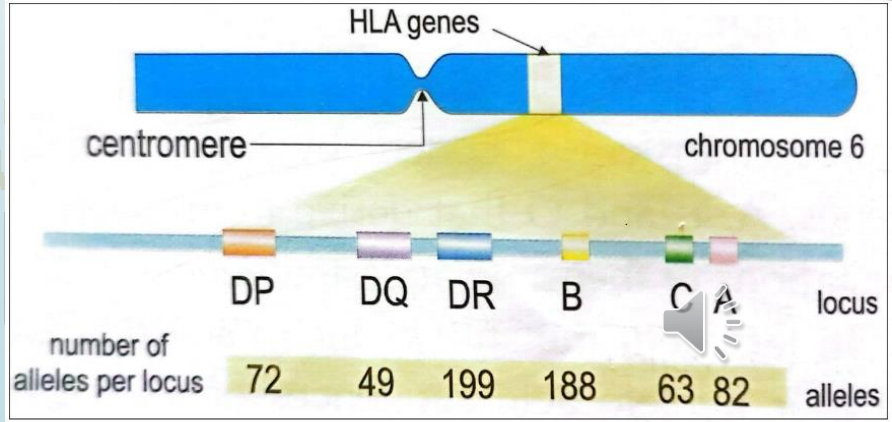
  → B-globin



I. Major Histocompatibility Complex (MHC)

- **MHC:** are set of 6 genes carried on chromosome number 6 in humans, which code for membrane proteins expressed on the surface of all nucleated cells of the body. These proteins act as biological markers which help the body to differentiate between self (what belongs to our body) and non-self (foreign) molecules.

- MHC is called HLA (Human Leukocyte Antigens).



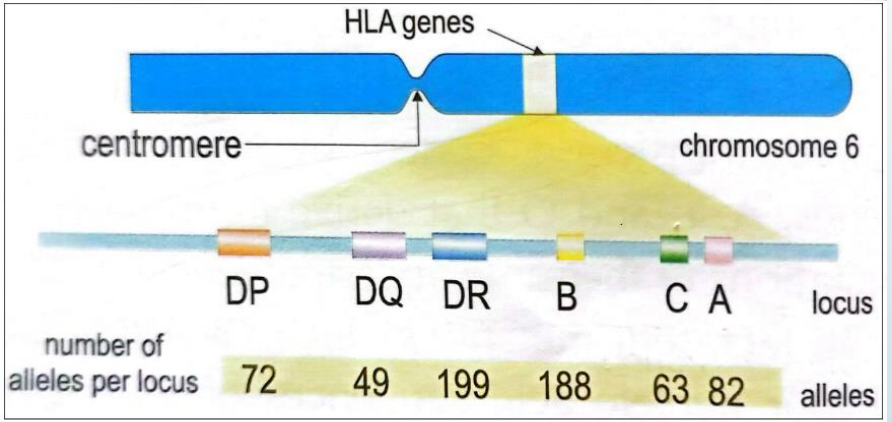
Doc.a MHC multiple alleles

***MHC genes coding for surface proteins are grouped in 6 loci called:**

DP, DQ, DR, B, C and A.

- Each of the 6 genes has a large number of alleles.

- Any individual will have two random alleles of each gene and both alleles are expressed (codominant), so it impossible for two people to have identical alleles in all six loci except for identical twins.



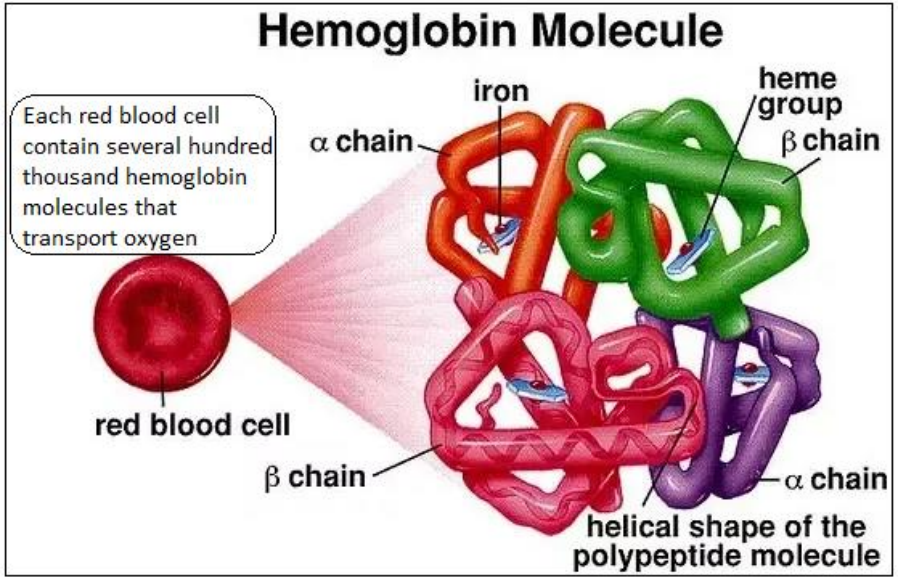
Doc.a MHC multiple alleles

→ *HLA or MHC are identical on all nucleated cells within the same body and between identical twins, but they differ from one individual to another.*

→ *Graft success depends on the similarity of HLA molecules between the donor and the recipient.*

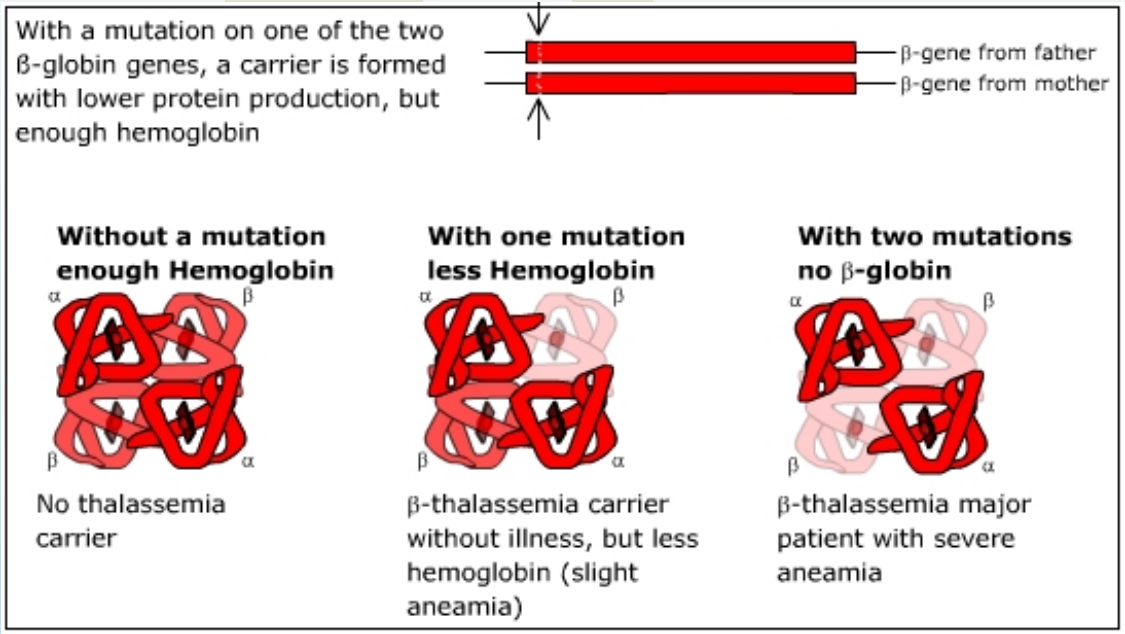
II. B- globin

- Hemoglobin is a protein molecule found in R.B.C which binds to iron.

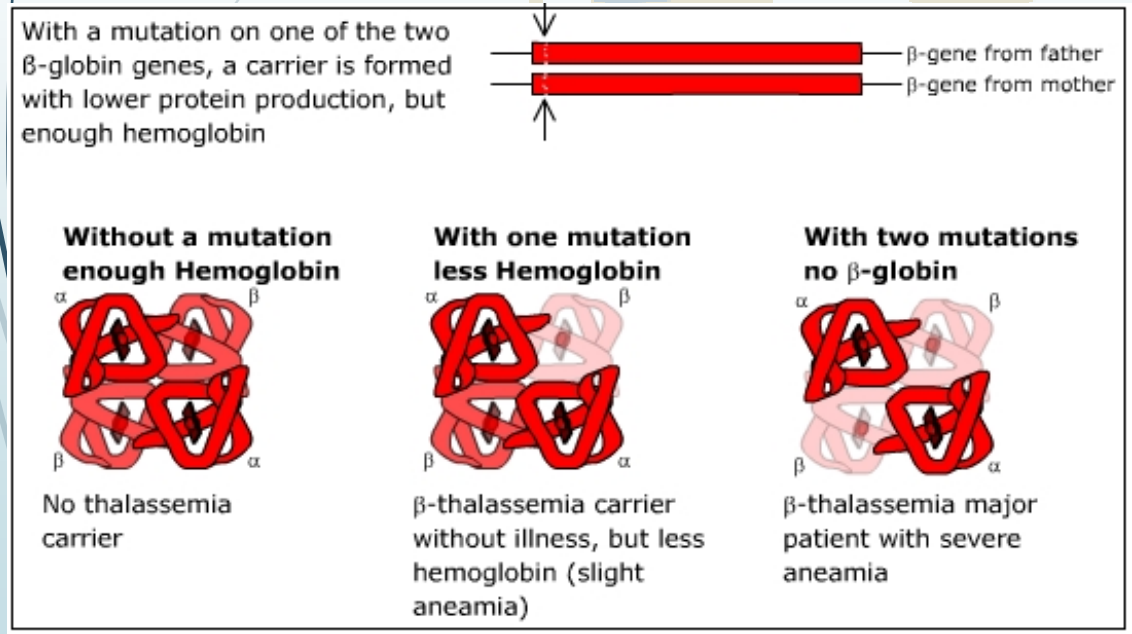


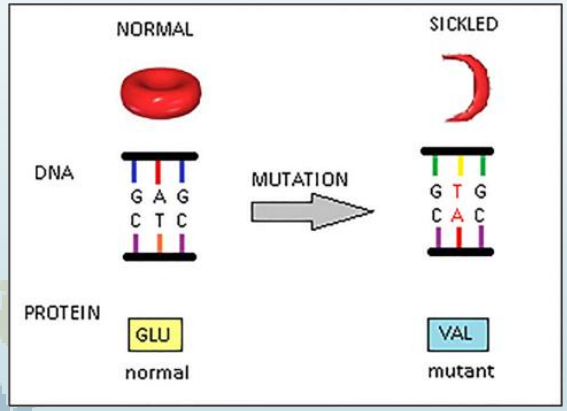
- B-globin gene is a gene which codes for B-globin polypeptide found in hemoglobin.
- Multiple alleles exist for this gene, many code for normal functioning B-globin polypeptide.

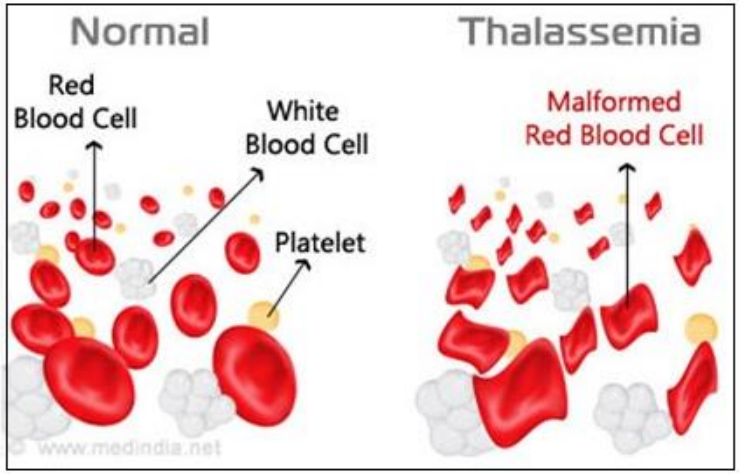
- Some mutations in the gene result in abnormal B-polypeptide leading to a dysfunctional hemoglobin molecule that can't bind iron properly which causes many diseases such as sickle cell anemia or B-thalassemia.



- The phenotypic consequence of a mutation depends on its type and location on the gene.
- The severity of the mutation in B-thalassemia depends on whether one or both alleles are affected.






Normal
Thalassemia
Red Blood Cell
White Blood Cell
Platelet
Malformed Red Blood Cell
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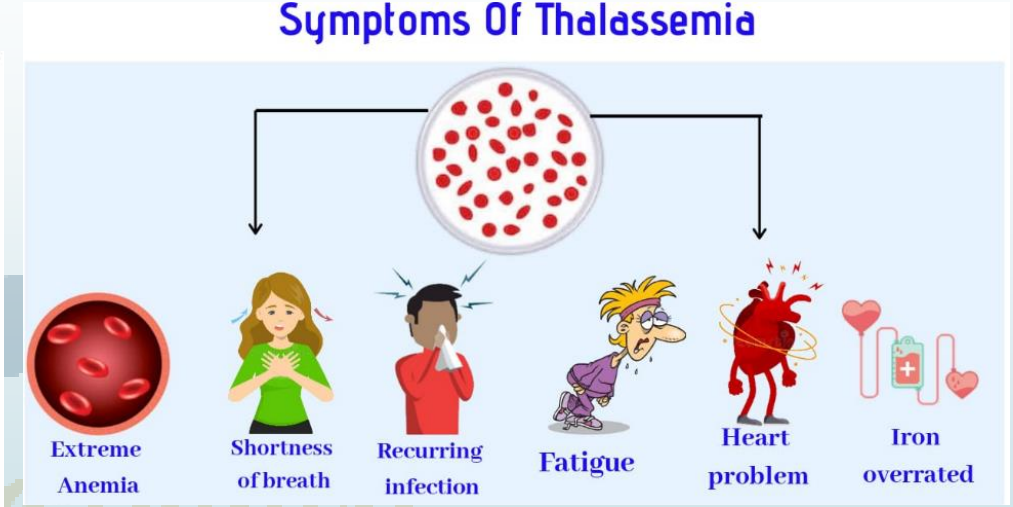
Symptoms Of Thalassemia
Extreme Anemia
Shortness of breath
Recurring infection
Fatigue
Heart problem
Iron overrated