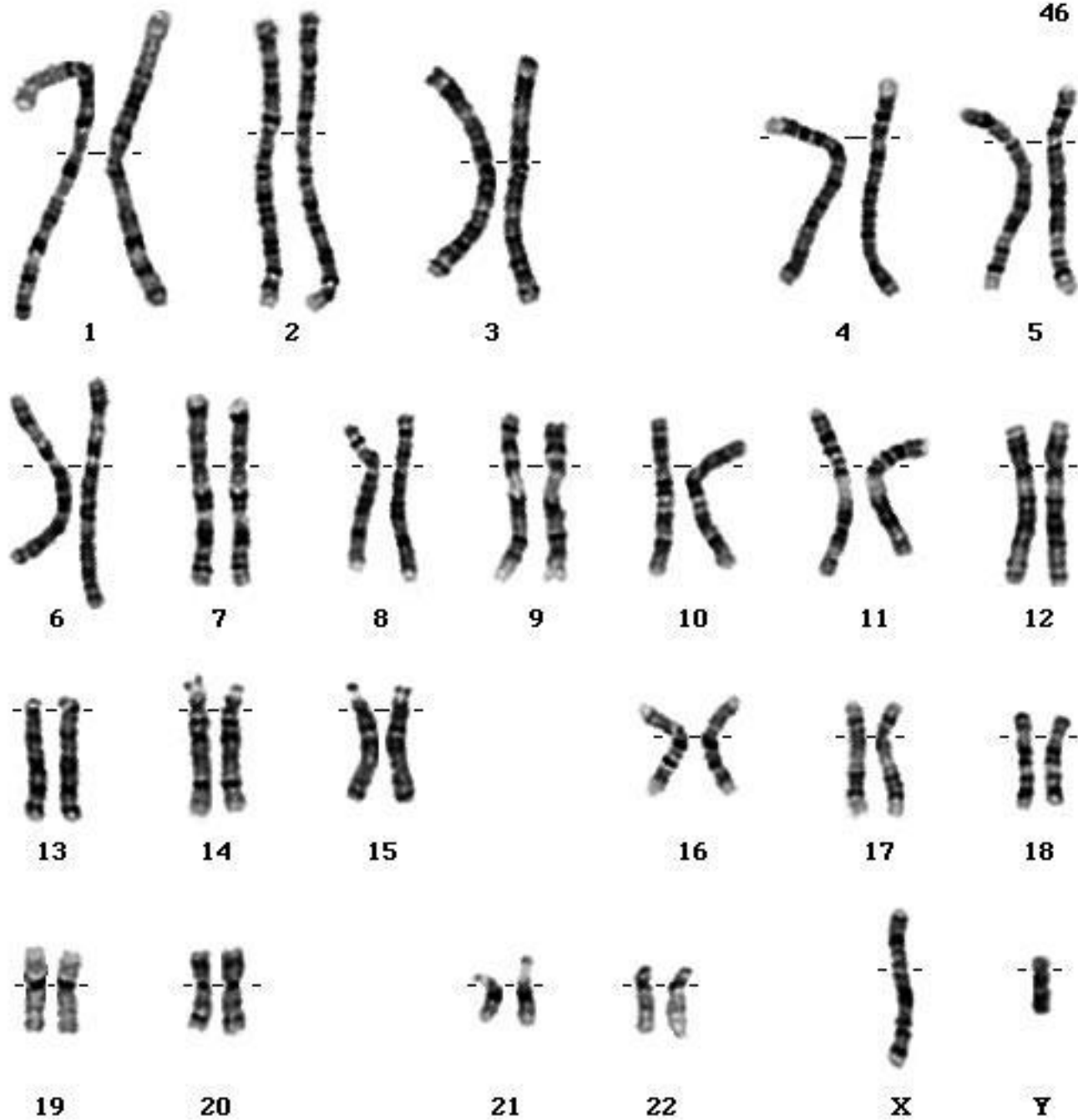


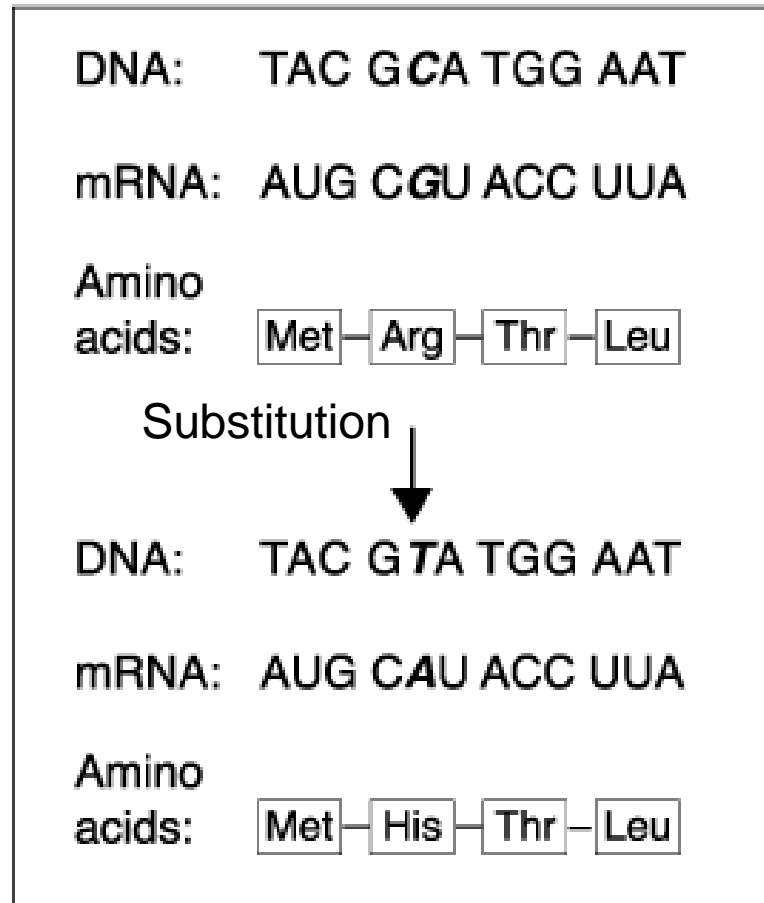
# DNA Mutations

12-4

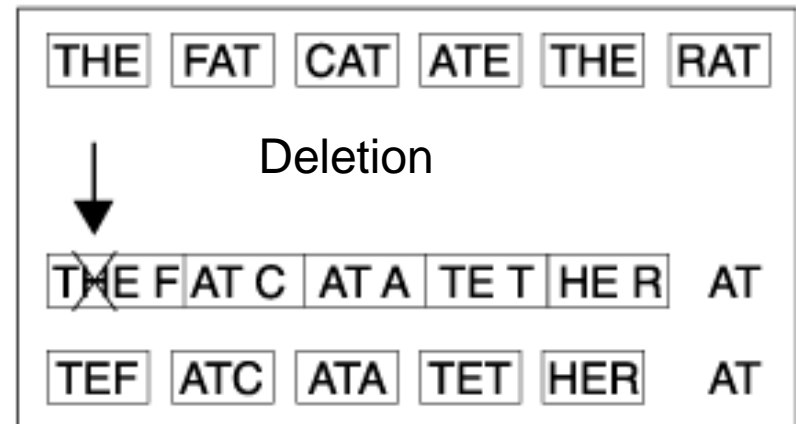
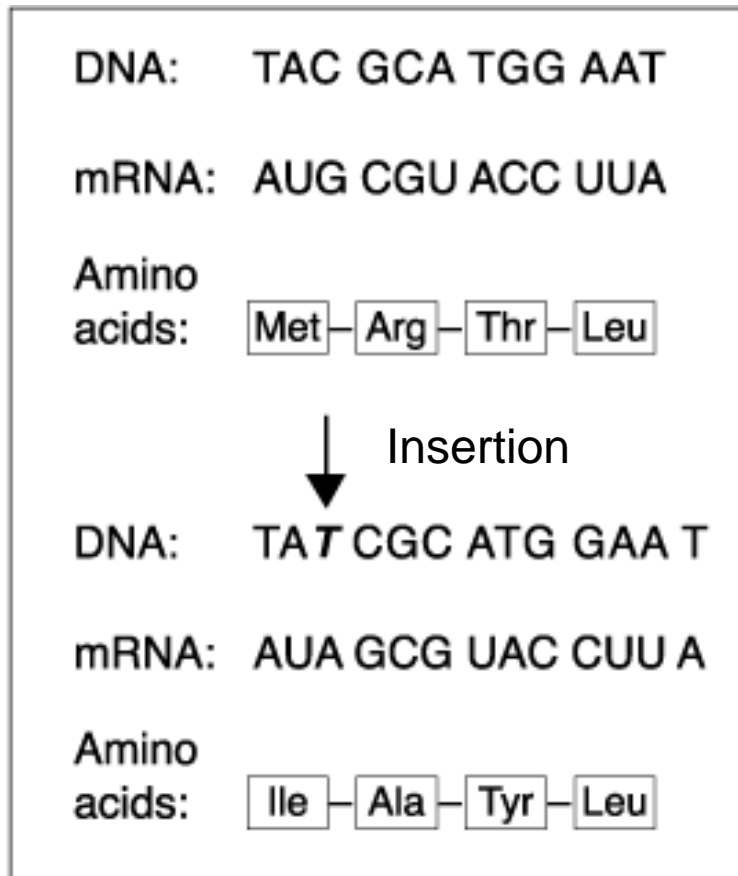


# Point Mutations

- Substitution : one nucleotide is substituted for another

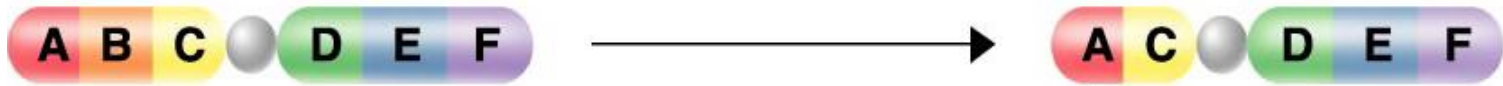


- Frameshift mutation: a nucleotide is deleted, or an extra one is inserted

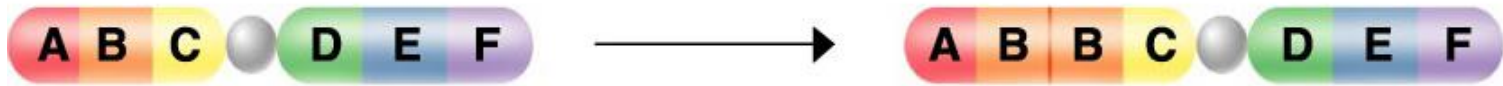


# Chromosomal Mutations

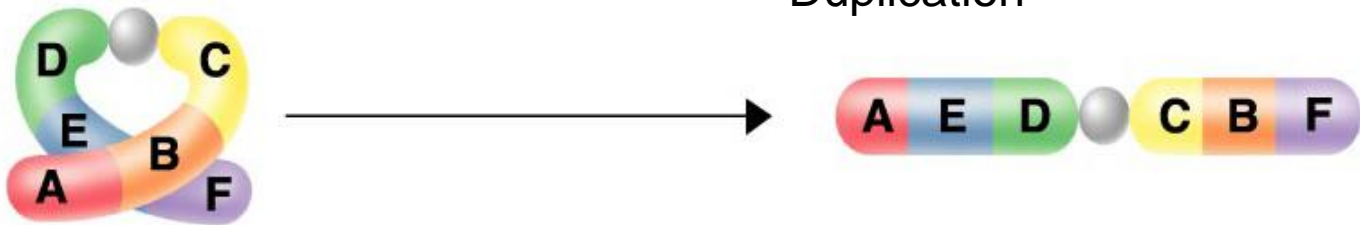
- Number or location of genes on a chromosome are changed.



Deletion



Duplication

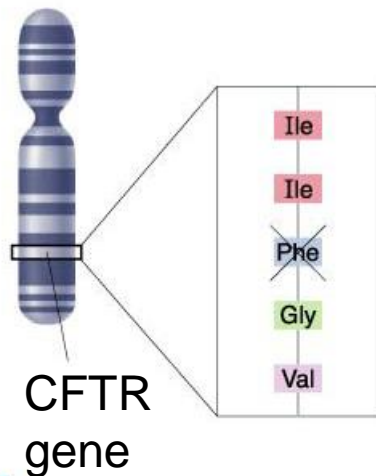


Inversion

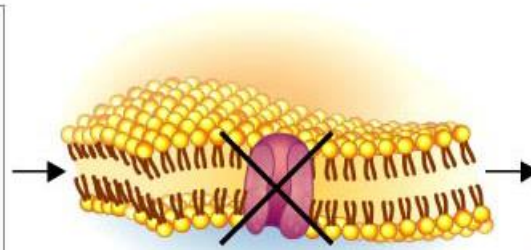


Translocation

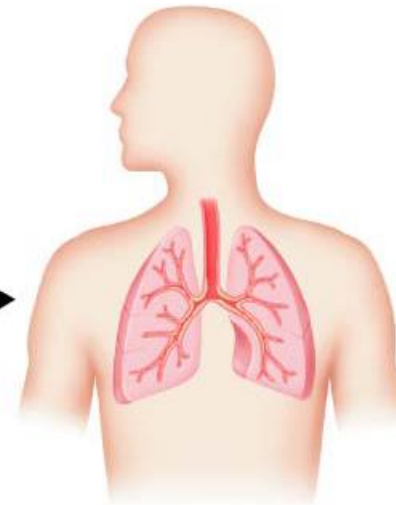
## Chromosome # 7



**A** The most common allele that causes cystic fibrosis is missing 3 DNA bases. As a result, the amino acid phenylalanine is missing from the CFTR protein.



**B** Normal CFTR is a chloride ion channel in cell membranes. Abnormal CFTR cannot be transported to the cell membrane.

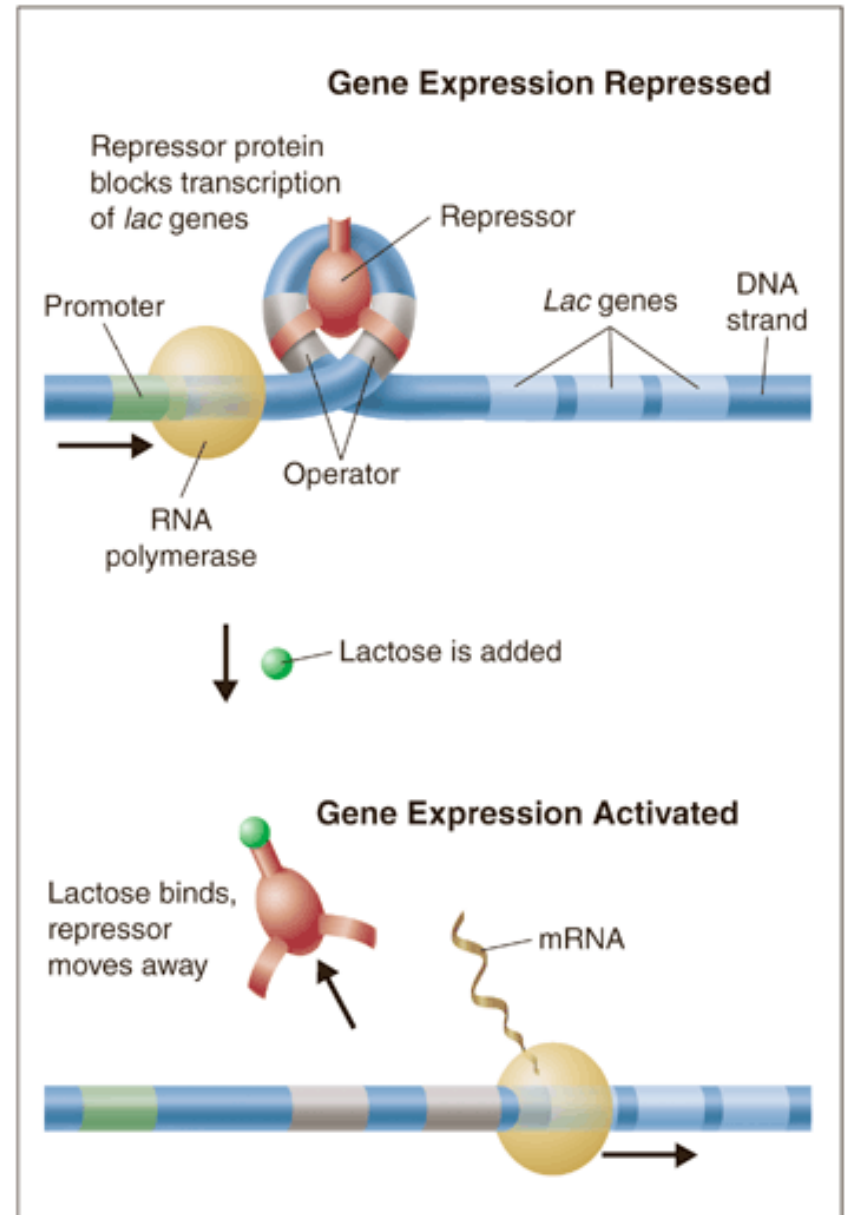


**C** The cells in the person's airways are unable to transport chloride ions. As a result, the airways become clogged with a thick mucus.



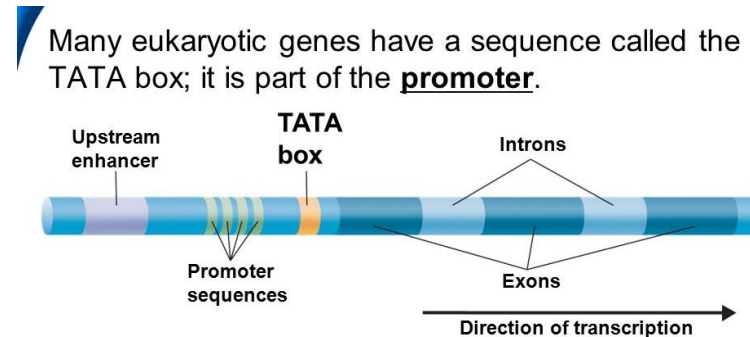
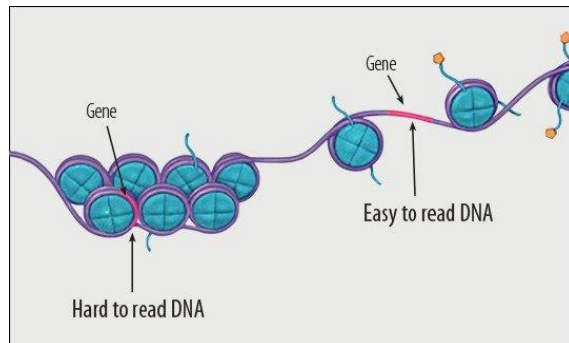
# Gene Expression

- How genes are turned on and off
- Prokaryotes – involves enhancers, repressors and operons
- Repressors block transcription
- Enhancers increase transcription

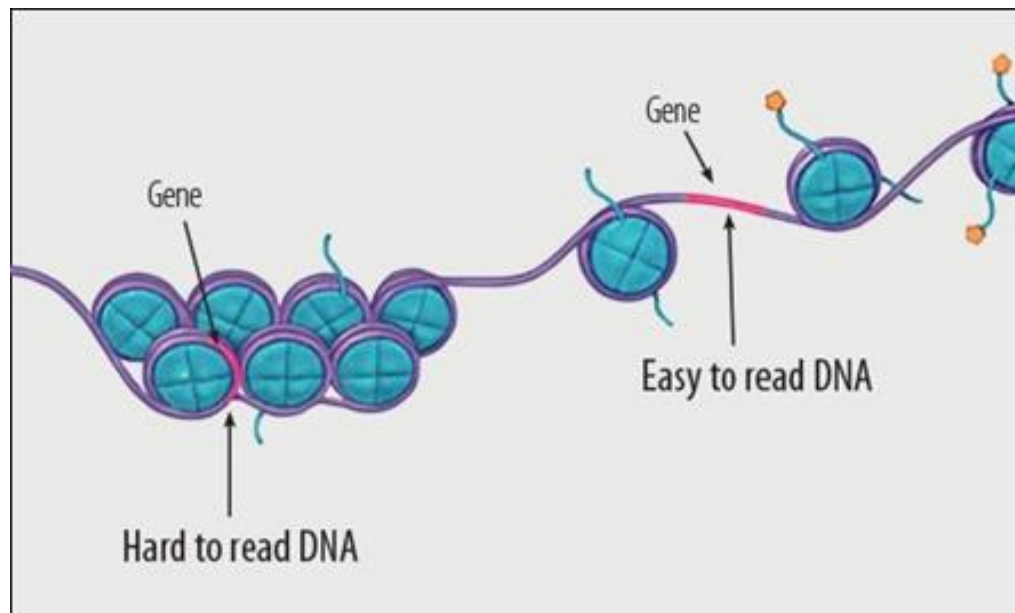


# Gene expression in Eukaryotes

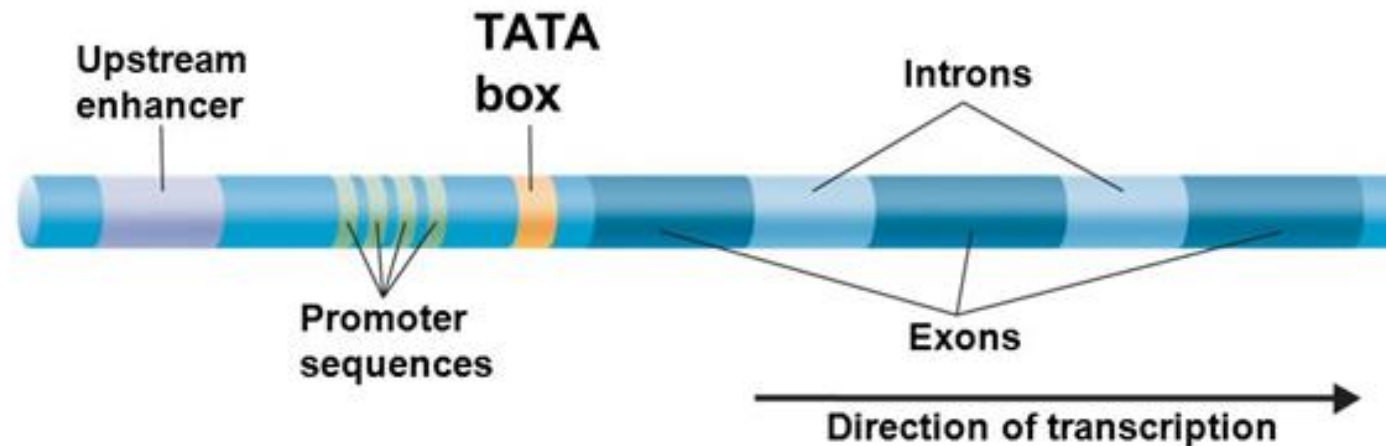
- more complicated in eukaryotic cells
- Each cell contains entire genetic code
- Cells are specialized
- Specific genes are expressed in certain cells
- Introns influence gene expression
- So do outside factors -- epigenetics







Many eukaryotic genes have a sequence called the TATA box; it is part of the **promoter**.



# Epigenetics

