

Unit 6 DNA ppt 3

Gene Expression and Mutations

Chapter 8.6 & 8.7 pg 248-255

- Which genes are transcribed on the chromosomes are carefully regulated at many points.
- Watch this!
- https://www.youtube.com/watch?v=OEWOZS_JTgk

How can organisms be different from each other if their DNA is made of the same nucleotides?

- Two individuals DNA are different because of the order of nitrogen bases
- The more closely related two organisms are, the more alike the order of nucleotides in their DNA will be.

Check this out!

<http://news.sciencemag.org/plants-animals/2012/06/bonobos-join-chimps-closest-relatives>

<http://futurehumanevolution.com/meet-our-top-10-evolutionary-relatives>



Gene Expression

- ✓ All cells within an organism have the same DNA and genes.
- ✓ What makes cells different from each other is that different genes are turned on and turned off in different cells.
 - ✓ Ex: Pigment in eyes or skin
 - ✓ Ex: Keratin in nails or hair



Regulation of gene expression (the making of protein)

- Your cells are NOT all the same.
- They differ because different sets of genes (segments of DNA) are turned ON or OFF at many different points .
- The Key is the START of TRANSCRIPTION.
- Several TRANSCRIPTION factors (Like DNA binding protein) can help the RNA polymerase “find the gene to transcribe”
- Most Eukaryotic cells have a nucleotide PROMOTER called TATAAAA box This signals polymerase to bind

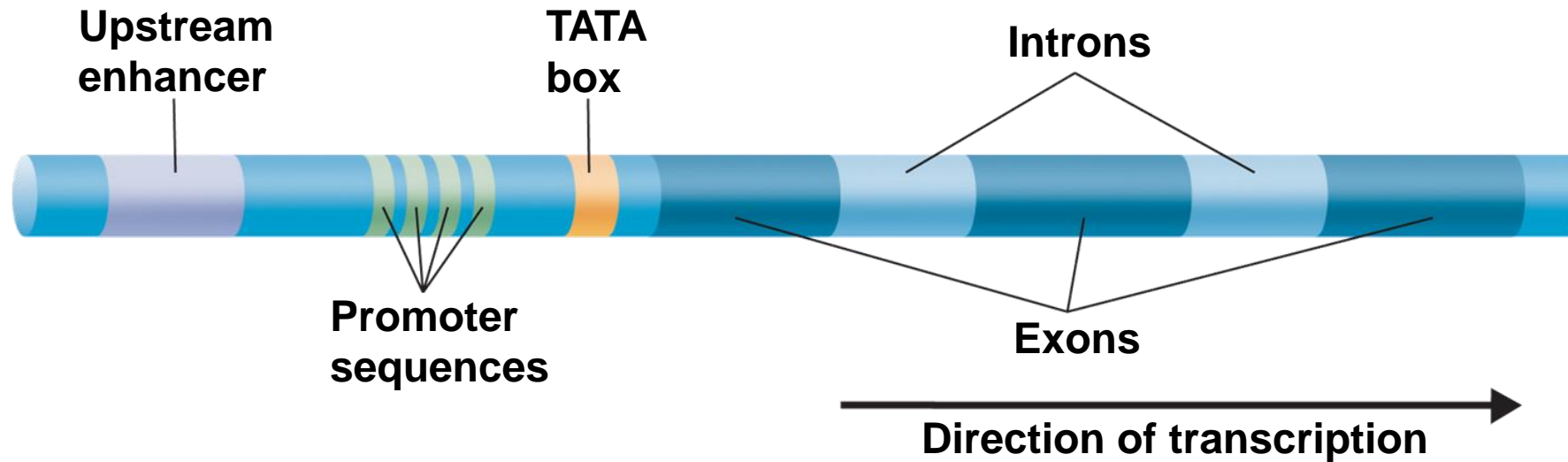
Genes are regulated in a variety of ways by enhancer sequences.

Many proteins can bind to different enhancer sequences.

Some DNA-binding proteins enhance transcription by:

- opening up tightly packed chromatin
- helping to attract RNA polymerase
- blocking access to genes.

The TATA box seems to help position RNA polymerase.

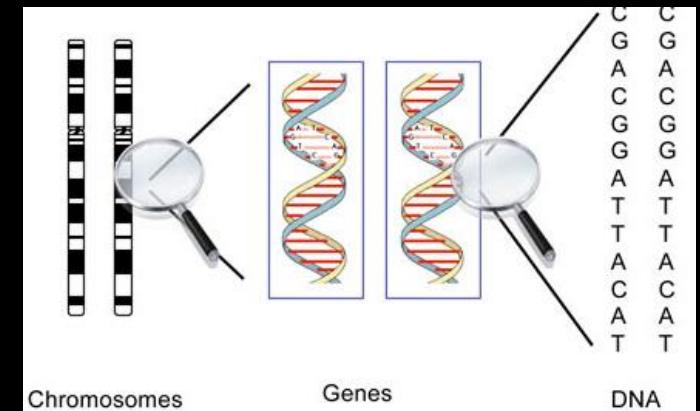


GENETIC VARIATION Not all genes that code for a certain protein are perfectly identical

- ✓ There are some variations in genes
 - ❑ ~every 1350th bp (average)
 - ❑ Known as polymorphisms “different forms”
 - ❑ Different forms of genes are called alleles

✓ Effects

- ❑ How well the protein works. EX: Melanin
- ❑ How the protein interacts with another protein or substrate. EX: Enzymes



MUTATIONS



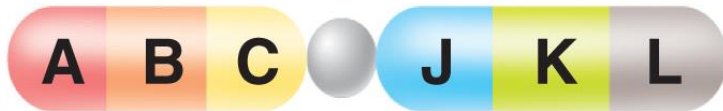
Original chromosome



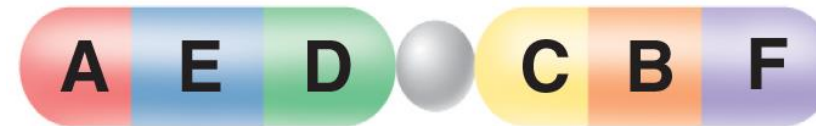
Deletion



Duplication



Translocation



Inversion



 What are mutations?

Mutations are changes in the genetic material



2 Kinds of Mutations



- Mutations that produce **changes in a single gene are known as gene mutations.**
- Mutations that produce **changes in whole chromosomes are known as chromosomal mutations.**



Gene Mutations



- Gene mutations involving a change in one or a few nucleotides are known as **point mutations** because they occur at a single point in the DNA sequence.
- Point mutations include substitutions, insertions, and deletions.



Substitutions usually affect no more than a single amino acid.

DNA: TAC GCA TGG AAT

mRNA: AUG CGU ACC UUA

Amino acids:

Met — Arg — Thr — Leu

↓ Substitution

DNA: TAC GTA TGG AAT

mRNA: AUG CAU ACC UUA

Amino acids:

Met — His — Thr — Leu



The effects of **insertions or deletions** are more dramatic.

The addition or deletion of a nucleotide causes a shift in the grouping of codons.

Changes like these are called **frameshift mutations**.



Frameshift mutations may change every amino acid that follows the point of the mutation.

Frameshift mutations can alter a protein so much that it is unable to perform its normal functions.



In an **insertion**,
an extra base is
inserted into a base
sequence.

DNA: TAC GCA TGG AAT

mRNA: AUG CGU ACC UUA

Amino
acids:

Met – Arg – Thr – Leu



Insertion

DNA: TAT CGC ATG GAA T

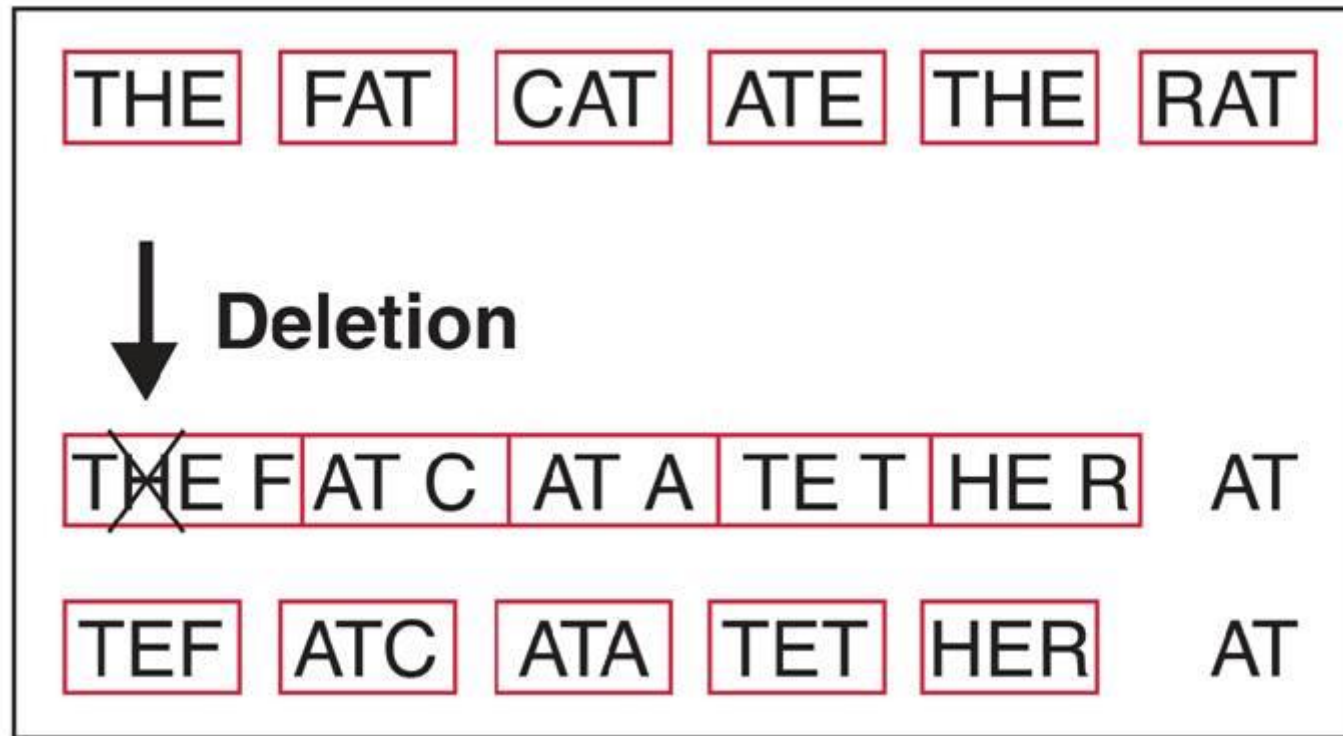
mRNA: AUA GCG UAC CUU A

Amino
acids:

Ile – Ala – Tyr – Leu



In a deletion, the loss of a single base is deleted and the reading frame is shifted.





Chromosomal Mutations

- Chromosomal mutations involve changes in the number or structure of chromosomes.
- Chromosomal mutations include deletions, duplication, inversions, translocations, and non disjunction.



Deletions involve the loss of all or part of a chromosome.



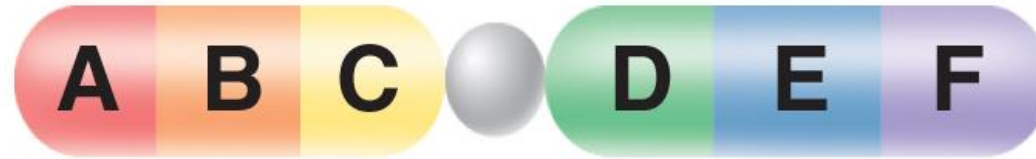
Original chromosome



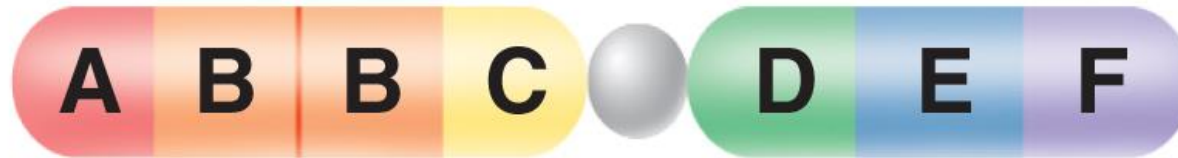
Deletion



- Duplications produce extra copies of parts of a chromosome.



Original chromosome



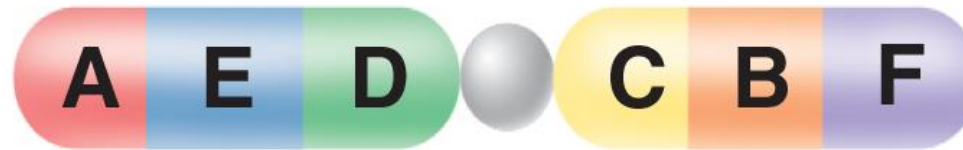
Duplication



Inversions reverse the direction of parts of chromosomes.



Original chromosome



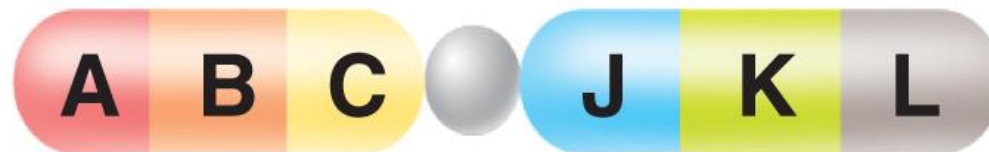
Inversion



Translocations occurs when part of one chromosome breaks off and attaches to another.



Original chromosome



Translocation



Significance of Mutations

- Many mutations have little or no effect on gene expression.
- Some mutations are the cause of genetic disorders.



Beneficial mutations may produce proteins with new or altered activities that can be useful.

Polyploidy is the condition in which an organism has extra sets of chromosomes.

Polyploidy is common in plants but rare in animals.



Polyploid plants not only have larger cells but the plants themselves are often larger. This has led to the deliberate creation of polyploid varieties of such plants as watermelons, marigolds, and snapdragons.

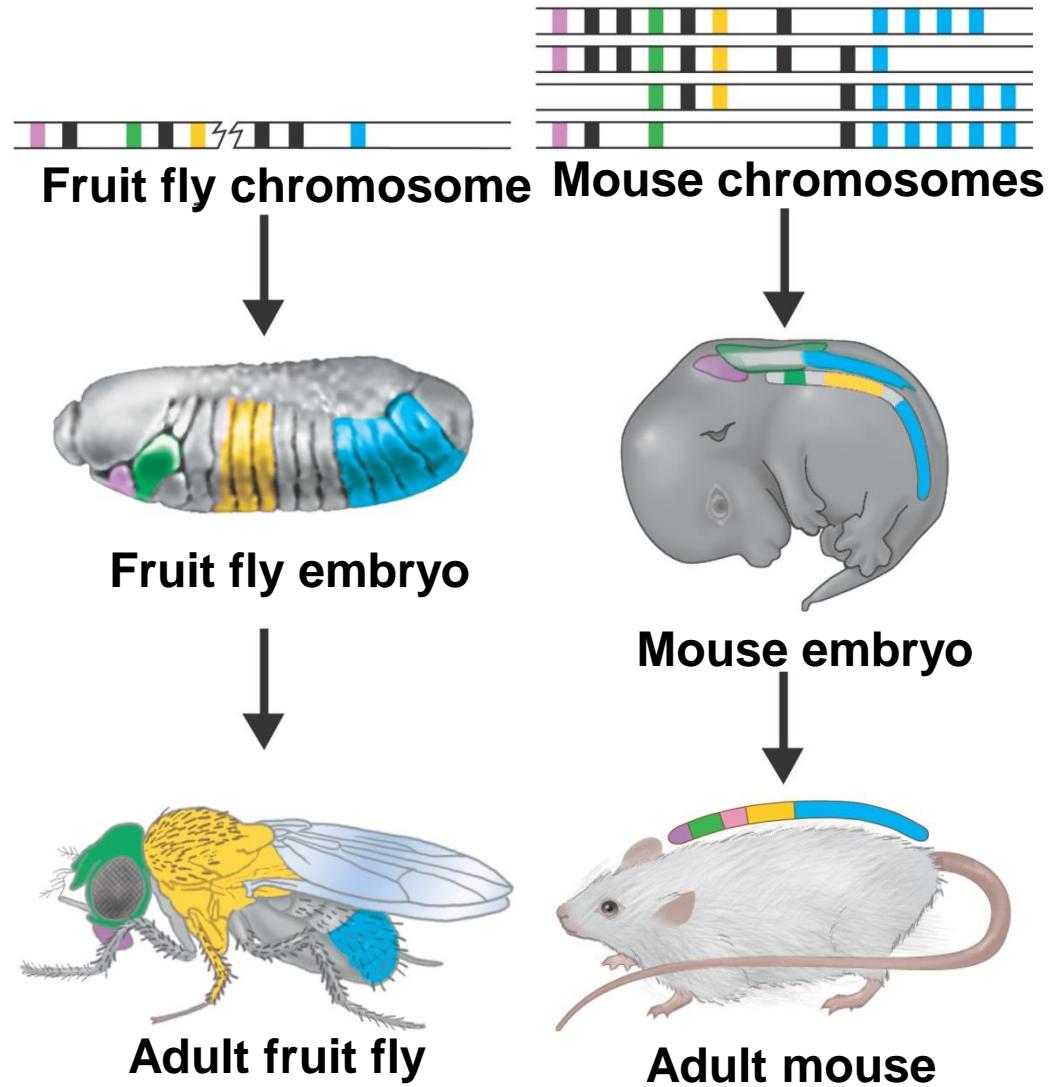
Development and Differentiation

- As cells grow and divide, they undergo **differentiation**, meaning they become specialized in structure and function.
- **Hox genes** control the differentiation of cells and tissues in the embryo.

Careful control of expression in hox genes is essential for normal development.

All hox genes are descended from the genes of common ancestors.

Hox Genes





Vocabulary for ppt 3 Gene expression and mutation

Nitrogen Bases

Gene expression

Transcription factor

TATA box

Polymorphism

Alleles

Mutation

Gene mutation

Point mutation

Substitution

Insertion

Deletion

Frameshift Mutation

Chromosomal Mutations

Inversion

Translocation

Polyploidy

Differentiation

Hox genes