Chapter 11
Gene Expression

11-1
Control of Gene Expression

**Gene Expression** - the activation of a gene to form a protein
-a gene is “on” or expressed when it is transcribed.
-cells do not always need to produce all of the proteins

**Genome**
-an individual’s complete genetic material

Most gene expression occurs in two steps - transcription and then translation

**Prokaryotic Gene Expression**

*Jacob and Monod* studied gene expression in *E. coli*
In 1965 they won the Nobel Prize.
They studied the metabolism of lactose.

Regulatory Elements of a Gene (**Operon**)
1) **Structural Genes** - genes that code for an actual protein. Jacob and Monod studied three structural genes that code for three enzymes that metabolize lactose.
2) **Promoter** - where RNA polymerase attaches to DNA - initiates transcription.
3) **Operator** - DNA segment where a protein may bind to block transcription. It controls the ability of RNA polymerase to attach to DNA.

**Operon** - a series of genes that code for specific proteins and the regulatory elements that control these genes.

**Lac Operon**

*Lactose = Lac*
This gene was only expressed (transcribed) when lactose was present.
**Repression**

**Repressor Proteins**
Produced by a regulatory gene
The protein binds to the operator, blocking transcription.
Lac Operon- without lactose, a repressor protein binds to the Lac operon.

**Activation**
In the lac operon, lactose is an **inducer**; it binds to the repressor protein and removes it.
So when lactose is present, transcription occurs.

Eukaryotes
- each cell contains a complete set of genes, but only certain genes are used in each cell.
Gene expression is much more complex in eukaryotes.
After mitosis or meiosis certain sections of DNA will uncoil,
-only the uncoiled chromatin- the **Euchromatin** is copied.

DNA contains
- **Exons**- expressed (coding) segments.
- **Introns**- non-coding segments
Both are transcribed, but the introns are removed before mRNA leaves the nucleus.
Introns are not fully understood. They may
  - help regulate RNA the is transcribed
  - influence gene expression; act as “switches” to turn on or off protein production.

Eukaryotes can modify RNA after transcription (in the nucleus) and before translation (in the cytoplasm)

1. pre-mRNA –contains introns and exons
2. mRNA- introns are removed, only exons remain (coding segments)
3. exons are spliced together (with spliceosomes)
4. mRNA leaves the nucleus → translation
Transcription Factors- regulatory proteins. They can bind to enhancers and RNA polymerase to allow transcription
Enhancers- segments of DNA that facilitate transcription. They must be activated for transcription to occur.

Cell Differentiation- the development of cells with specialized functions
Morphogenesis- the development of an organisms form.

Homeotic Genes- regulatory genes that control body formation, they determine anatomy, where structures will form.” Master genes” They produce regulatory proteins that increase or decrease cell division in areas.
Homeobox- a sequence of DNA that controls a specific region of the body. Regulates development.
Homeotic genes contain a homeobox.
Proto-oncogenes normally code for proteins that control cell growth. A mutation may cause it to become an oncogene.

Oncogenes- A gene that causes cancer or uncontrolled cell growth.

Cancer
Tumor- abnormal cell growth
   a) Benign- not cancerous, does not spread.
   b) Malignant- cancerous, cells invade other tissues. Metastasis=spread

Tumor Suppressor Genes
Normally prevent uncontrolled growth
May mutate, so they cannot suppress cancer.

Cancer cells behave differently
- they continue to grow even when crowded
- they do not need to be in contact with other cells- allows them to spread.

Causes of Cancer
- exposure to carcinogens increase chances of cancer.
Mutagens- carcinogens that cause mutations
Chapter 12
Inheritance Patterns and Human Genetics

12-1
Chromosomes and Inheritance
Thomas Hunt Morgan -1900’s
-worked with fruit flies- \textit{Drosophila melanogaster}
-determined that females were XX and males were XY
-50-50 chance of being male or female
-discovered sex chromosomes and autosomes

The Y chromosome- contains a gene- SRY- \textbf{Sex-determining Region Y}. This gene codes for a protein that causes the gonads of an embryo to develop as testes.

Sex Linkage
Because of its larger size, more genes are on the X chromosome than on the Y chromosome.
\textbf{X-linked genes}- genes on the X chromosome
\textbf{Y-linked genes}- genes on the Y chromosome
\textbf{Sex-linked trait}- a trait that is coded for by an allele on a sex chromosome.

Linked genes
Two or more genes found on the same chromosome
Linked gene tend to be inherited together
Linkage groups- a set of linked genes.

Crossing Over
Provides new combinations of genes that are not found in the parents

\textbf{Chromosome Mapping- page 238}
The farther apart two genes are, the more likely they will be separated by crossing over.
A chromosome map shows the linear sequence of genes on a chromosome.
Ex. Two genes that cross over 15\% of the time, are 15 \textbf{Map Units} apart.

Mutations
\textbf{Mutation}- a change in the nucleotide sequence
Mutations can affect the entire chromosome or a single nucleotide.
\textbf{Germ Cell Mutation}- occurs in gametes—affects the offspring, but not the organism itself.
\textbf{Somatic Mutation}- affects cells of the organism, but it is not passed on. Ex. Skin cancer, leukemia.

Mutations may be lethal, neutral or even beneficial.
Chromosome Mutations- page 239
Deletion- loss of a piece of a chromosome
Inversion- a section of the chromosome reverses.
Translocation- a piece breaks off and attaches to another chromosome

Nondisjunction
A chromosome does not separate from its homologue during meiosis.
One gamete has an extra chromosome, and one gamete is missing a chromosome.
Down syndrome is an example of nondisjunction – Trisomy 21

Gene Mutations P. 240
Point Mutations- substitution, addition or deletion of a single nucleotide.
   A) Substitution- replacing a nucleotide with a new base
      -if it codes for the same amino acids \( \rightarrow \) no problem
      -if it codes for a different amino acid \( \rightarrow \) wrong protein! Big problem!
      Ex. Sickle Cell anemia

   B) Insertion- adding one or more nucleotides
   C) Deletion- removing one or more nucleotides
Both cause Frame Shift Mutations
Ex. The fat cat ate one rat.
   The ftc ata teo ner at. (deleted first “a”)
**12-2**

**Human Genetics**

**Pedigrees—page 241**
-A diagram that shows how a trait is inherited over several generations.
Can show **Patterns of Inheritance**.

![Pedigree Diagram]

**Carriers**: have one copy of a recessive allele, but do not show the trait- Bb (Half filled in on pedigree.)

**Polygenic Traits**
Most traits are controlled by several genes
Ex. Skin color, height, hand, foot, nose size…

**Complex Characters**
Characteristics are influenced by both the environment and by genes.
Environmental factors influence many traits.
Ex. Height- influenced by diet and disease
   Skin color – influenced by exposure to the sun.

**Multiple Alleles**
Traits controlled by three or more alleles
Ex. Blood Type: I^A^, I^B^, i
Page 244

**Incomplete Dominance**
The heterozygous form is in between. In Caucasians, curly hair (HH), wavy hair (Hh), and straight hair (hh).

**X-Linked Traits**

<table>
<thead>
<tr>
<th>X</th>
<th>Y</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Normal F</td>
</tr>
<tr>
<td>2</td>
<td>Carrier F</td>
</tr>
<tr>
<td>3</td>
<td>Normal M</td>
</tr>
<tr>
<td>4</td>
<td>Hemo. M</td>
</tr>
</tbody>
</table>

Colorblindness (red-green)
Hemophilia→XX, X^h^X, X^h^X^h^, XY, X^h^Y
(Queen Victoria’s Family)
Duchenne muscular dystrophy
Much more common in males, they only have one X.

Sex Influenced traits
Expression is influenced by the sex hormones
Ex. Pattern baldness—B is dominant in males, but recessive in females.
Male- BB or BB’= baldness
Females- only BB is bald

Single Allele Traits
Controlled by a single allele of a gene
A) Dominant alleles- Huntington’s Disease(HD), dwarfism
B) Recessive allele-albinism, cystic fibrosis, PKU

Pedigree for Cystic Fibrosis

A Genetic Marker- a section of DNA associated with a particular gene. Does not always mean you will get the disease.

Genetic Screening
Ammniocentesis- uses amniotic fluid to form a karyotype. (at 14-16 weeks)
Chorionic Villi Sampling (CVS)- uses tissue from the fetus to form a karyotype (at 8-10 weeks)
Genetic Counseling
Uses data from genetic screening and the family’s pedigree.

Treating genetic Disease
PKU- Phenylketonuria
Screening is done at birth. If positive, the baby is put on a special diet that does not contain any of the amino acid Phenylalanine.

Gene therapy
Placing a healthy copy of a gene into the cells of a person with a defective gene. Used to treat Cystic Fibrosis (CF)