**Mutations:**
- distinguish between somatic and germ mutations
- explain the different types of mutations
- explain how “jumping genes” influence diversity
- identify diseases caused by chromosomal mutations
  (Downs, Turner, Kleinfelters, Jacobs, XXX)
- describe the role of a genetics counsellor

**D: a permanent change made to one section of DNA**

Causes the gene to incorrectly tell the cell how to make a protein, so that the protein order of amino acids may be changed. It may still function, perhaps it doesn’t work, or can’t be made at all.
(Sense, mis-sense, and nonsense mutations)

*caused by a “mutagen”.... “induced mutations”*

*External ex’s....chemical exposure, radiation, or viruses*

*two kinds...*
- Physical mutagens (tears apart DNA strands)...radiation causing dimer changes in adjacent pyrimidine (C and/or T) bases.....kinks DNA structure.....inability to read or copy
- Chemical mutagens (molecules entering cell nucleus and interacting with or inserts itself into DNA)
Mutations in body cells (somatic mutations) cannot be inherited, they affect the cell they are in, and any daughter cells made from that mutated parent cell. (affecting a small local area of the body)

Germ mutations affect sex cells and are inheritable if they are involved in forming a zygote.

Most mutations have little effect..unless the genes controlling cell division change, division happens uncontrollably......cancerous tumor. (genes that mutate to cause cancer are called “oncogenes”)

Cancer Treatment :

Radiation therapy....
- blasting x-ray or gamma radiation at the affected part several times per week, interfering with mitosis....slowing or stopping tumor growth. For site specific cancers (breast, cervix, larynx, skin)
- may cause skin irritation, fatigue, hair loss, sterility

Chemotherapy.....
- combinations of drugs with / without radiation therapy, for body wide cancers (like leukemia)
- drugs interfere with mitosis in healthy and unhealthy cells
- causes hair loss, nausea, diarrhea, for the duration of the treatment
Treatments for Mutations....

Genetic counsellor -
If there’s a family history of a problem, might want to consult this person BEFORE another pregnancy!
A pro who learns about family genetics through interviews, blood tests, and connecting with geneticists. Can make pedigrees and predict probability of a child with a disorder.

Embryo screening -
Sperm and egg combined in a lab to make a number of zygotes. Each is analyzed, one without the disorder can be implanted into the “mom”

Prenatal testing and karyotyping -
Amniocentesis
CVS
Fetoscopy

Detecting Genetic Markers -
Characteristics of a creature that tell us about its genome.

Linked marker -
A known base sequence close to a disorder causing gene

Gene marker -
Base sequence in a gene itself

Found by introducing a radioactively tagged complementary base sequence....see if it attaches
Treatment of Genetic Disorders (page 609)

1. Genetic Screening....
   Detecting a disorder with a test at birth. Ex. PKU, early intervention to prevent complications later.

2. Surgery....
   Structural problems addressed using surgery. ex. cleft palate, reconstructions, tumor removal, etc.

3. Environmental controls...
   Minimize the effects symptoms of a disorder have on a person by avoiding environmental triggers...ex. diet changes, sun protection, etc.
4. Gene Therapy...

Normal, or altered, genes are inserted into the defective cells of an organism.

Sample of defective cells, cultured, virus carrying copies of a functioning gene attacks the cells, some incorporate the new gene, insert them into the patient, some take, function slowly returns.

Related Ethical Concerns....
- who decides what’s a “disorder” ?
- should we design people without flaws ?
- is it available for all, or just those who can pay ?
- could trigger a strong immune response, potentially fatal
- if not in the correct location, or
expressed incorrectly, results could be worse than the condition
- could interfere with other genes, or be influenced by them

Somatic gene therapy...
  body cells altered

Germ Line Therapy...
  changing sex cells
“Chromosomal mutations”...
changes in the physical structure of an entire chromosome.

Types....

Deletions -
part of a chromosome breaks off and is lost. Ex. Cri du chat syndrome.

Inversions -
a gene breaks off, but is reinserted backwards, changing the way it is read, and the product it makes. Ex. Autism

Duplication -
a gene is repeated many times over, making the chromosome thin and fragile in one spot...prone to breakage, “fragile X” syndrome results.
Translocation -
a chromosome part breaks off and attaches in a different spot, or even in a different chromosome

ex.
14-8 cancer
14-21 Down’s Syndrome
9-22 leukemia
Nondisjunction -  
Problem(s) separating chromosomes / chromatids during meiosis, resulting in too many or too few chromosomes in gamete and embryo...most miscarry early.

Monosomy  
Trisomy

Ex. Trisomy 21... Down’s  
Only 1 X.......Turner’s  
Extra X......Kleinfelter’s  
Extra Y......Jacob’s

Transposable genes (“transposons” or “jumping genes”) -  
short DNA sequences that can move about between chromosomes, increasing diversity.

cut out and spliced in by the enzyme transposase  
Discovered by Babara McClintock, 1957....nobel prize....boosts diversity and helps drive evolution of species  
Ex. Random coloration of Indian corn
“Point mutations”...
Changes that affect short sequences of bases.

2 Types.....

Substitution mutations...
One base replaced by another, change to the resulting protein causes silent, mis-sense, and nonsense mutations.

Frame shift mutations...
Base insertion or deletion results in the code being read in new combinations of 3. Different amino acid sequence results....different protein results.
Related jobs studying mutations.....

Oncologist:

cytogeneticist:
http://www.prospects.ac.uk/clinical_cytogeneticist_job_description.htm

medical geneticist:
http://en.wikipedia.org/wiki/Medical_genetics

genetic engineer: