Gene Mutation

- A mutation is a change in DNA sequence.
- Mutagen is a chemical that causes mutation.
- Spontaneous mutations happen naturally and randomly, usu. linked to normal processes.
- Caused by incorrect pairing of bases
- Induced mutations result from the influence of an external factor, either natural or artificial.
  - Ex. – exposure to radiation, cosmic rays, in rocks, drugs
- Natural Protection Against Mutation
  - Mutation in third codon spot may not matter
  - Change in a.a. may not change protein enough
  - May need only one good copy of gene to be alright
  - Some mutations need special triggers.
- DNA Repair
- Types of Mutations
  - Somatic mutations occur nonsex cells.
  - Germ-line mutations occur in gametes and are inherited.
  - Point mutations is a change in a single DNA base.
    - Ex. ATCGGCTA becomes ATGGGCTA
  - Missense mutations change a codon from one to another. Changes protein.
  - A nonsense mutation changes a codon into a stop codon. Results in premature termination of translation.
  - A silent mutation changes a codon but does not result in a change in the amino acid.
  - A frameshift mutation occurs when bases are added or deleted, except multiples of three.
    - Ex. ATC GTA CTA GCA becomes ATC GAC TAG CA…
  - Deletion = loss of base(s)
  - Insertion = added bases(s)
  - Duplication - Extra triplet repeat sequences are responsible for some conditions.
  - Transposons - Segments of DNA that move

Chromosomes

- Strands of DNA wrapped around balls of proteins called histones.
- "X" is two strands connected at a centromere ready for cell division.
- telomere - repetitive sequence at the ends of chromosomes that protect the ends of the chromosome
- Distinguished by size and staining
- Humans' have 22 pairs of autosomal and 2 sex = 46 total
- Karyotype
  - Sized-ordered chart of chromosomes
- Method for sampling cells
  - Amniocentesis – sampling fetal cells shed into amniotic fluid with a needle through the abdominal wall. Done at ~15-16 weeks
  - Chorionic Villus Sampling – cells taken from placenta
  - Cell Free DNA Testing - small pieces of DNA from pregnancy circulates in pregnant woman's blood ~10 weeks
- Chromosomal Shorthand
  - Normal male – 46, XY
  - Normal female – 46,XX
  - Missing an X – 45, XO
  - An extra chromsome, 47, XX +17

- Polyploidy - extra set of chromosomes
- Aneuploidy - missing or having an extra single chromosome
  - Trisomy – having three of one type of chromosome
    - Ex. Down's - Trisomy 21
  - Monosomy – having only one of a type

- Ex. Turner Syndrome(45 XO)
- Nondisjunction – problem in making gamete during meiosis
  - XO – Turner's
  - XXY Klinefelters
  - XXX – Triplo – X
  - XYY – Jacob's , Supermales
- Deletion
  - Part of chromosome is missing.
  - Ex. Cri-du-Chat – XY, del(5)
- Duplication
  - Extra chromosome part
- Translocation
  - Part of different chromosome is attached
- Inversions
  - Part of the chromosome is flipped around