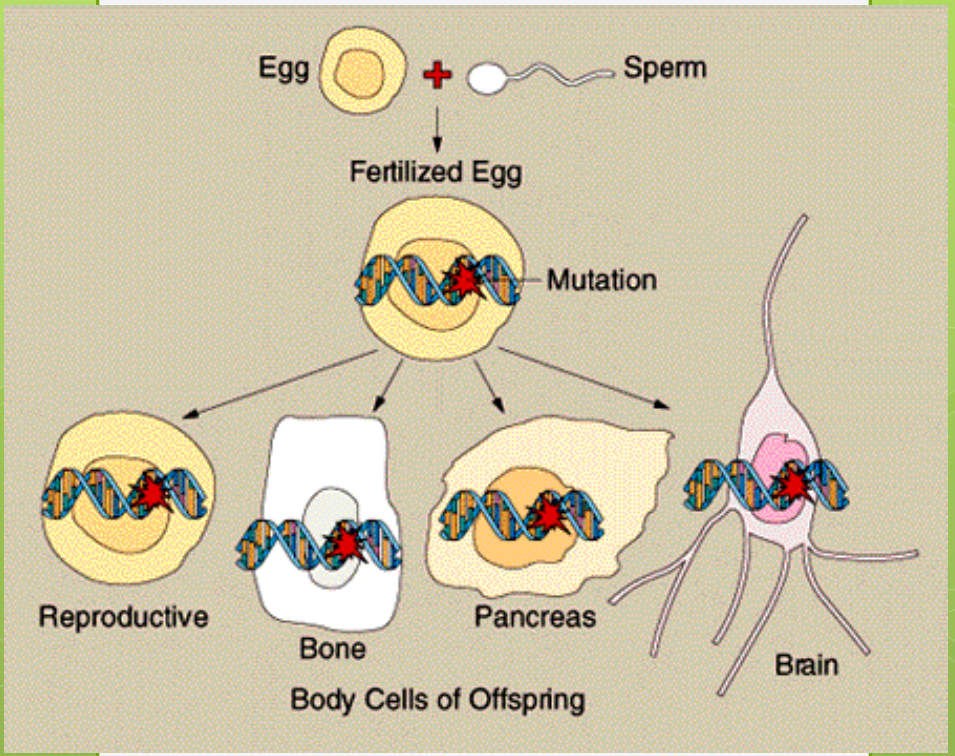
Mutations Quick Questions and Notes (#1)

QQ#1: What do you know about mutations?



mutation basics…

- Definition: a change in the genetic material of a cell
  - Note: not all mutations are bad
- Can occur in 2 types of cells:
  - Germ Mutations: occur in germ-line (sex) cells
  - Somatic mutations: occur in somatic cells
- Some are inherited and some are developed during embryonic development.

2 types of mutations:

- **Gene mutations:**
  - involve individual genes within a chromosome
- **Chromosomal mutations:**
  - involve segments of chromosomes, whole chromosomes, and even entire sets of chromosomes

Gene Mutations

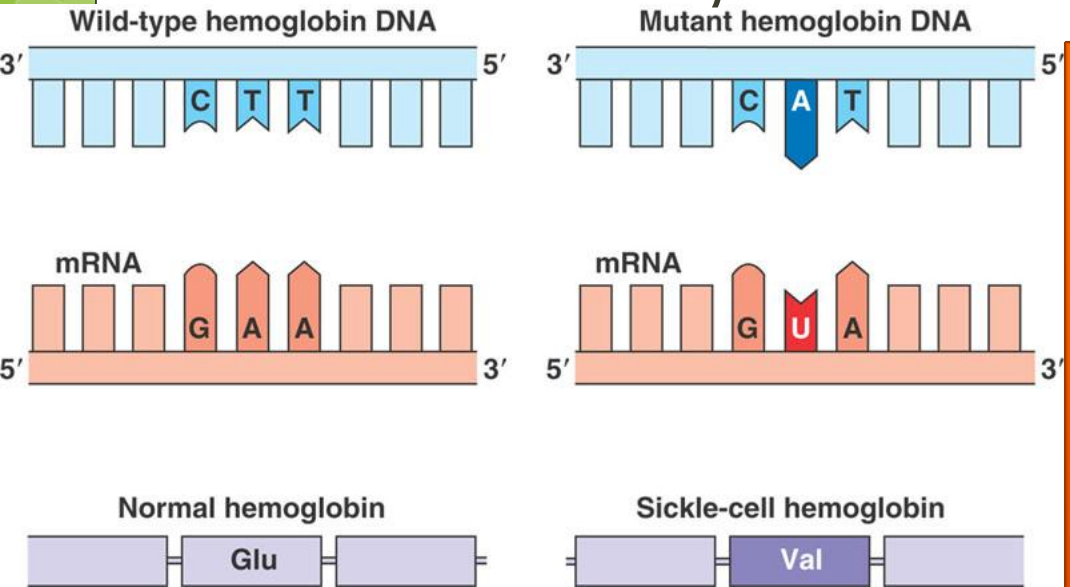
- Point Mutation: the swapping of one base pair

- Frameshift Mutation: the insertion or deletion of a base pair

QQ#2:
Which do you think has more chance of causing a noticeable mutation?

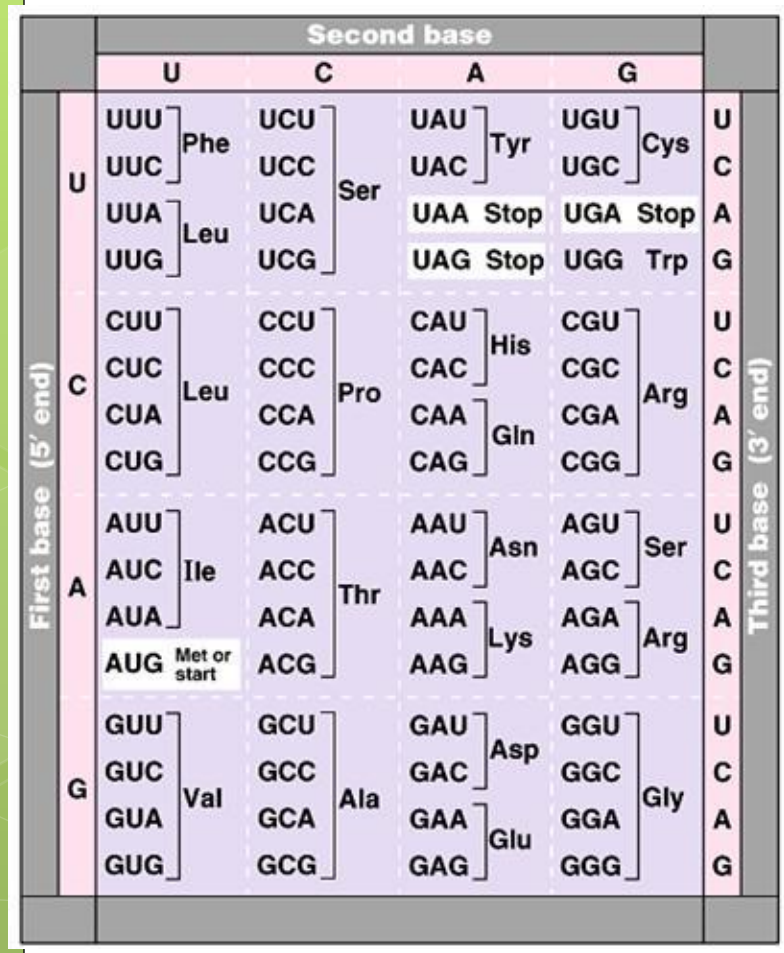
Gene Mutations

- Point Mutation
  - Changes just one nucleotide in a sequence
    - Can cause a big or small mutation
    - (VARIES BECAUSE OF "WOBBLE POSITION")



QQ#3:
Thinking about how codons code for proteins, why do you think these mutations can be big or small?

The Wobble Position



First base (5′ end)	Second base: U	Second base: C	Second base: A	Second base: G	Third base (3′ end)
U	UUU Phe	UCU Ser	UAU Tyr	UGU Cys	U
	UUC Phe	UCC Ser	UAC Tyr	UGC Cys	C
	UUA Leu	UCA Ser	UAA Stop	UGA Stop	A
	UUG Leu	UCG Ser	UAG Stop	UGG Trp	G
C	CUU Leu	CCU Pro	CAU His	CGU Arg	U
	CUC Leu	CCC Pro	CAC His	CGC Arg	C
	CUA Leu	CCA Pro	CAA Gln	CGA Arg	A
	CUG Leu	CCG Pro	CAG Gln	CGG Arg	G
A	AUU Ile	ACU Thr	AAU Asn	AGU Ser	U
	AUC Ile	ACC Thr	AAC Asn	AGC Ser	C
	AUA Ile	ACA Thr	AAA Lys	AGA Arg	A
	AUG Met or start	ACG Thr	AAG Lys	AGG Arg	G
G	GUU Val	GCU Ala	GAU Asp	GGU Gly	U
	GUC Val	GCC Ala	GAC Asp	GGC Gly	C
	GUA Val	GCA Ala	GAA Glu	GGA Gly	A
	GUG Val	GCG Ala	GAG Glu	GGG Gly	G

- The third nitrogen base in every codon is called “the wobble position”
- This minimizes possibility of genetic mutations

Gene Mutations

- Point Mutation

- **FrameShift**
  - **Insertion**
    - **Addition of a few extra nucleotides**
      - **Ex: ACGTACTTCGA becomes…**
        ACGATACTTCGA

      What could be the effect of this?

      Example: THE CAT RAN FAR becomes…
      THE CAA TRA NFA R

- Deletion

Gene Mutations

- Point Mutation

- Frameshift:
  - Insertion
  - **Deletion**
    - **A few nucleotides are deleted**
      - **Ex: ACGTACTTCGA becomes…**

        AGTACTTCGA

What could be the effect of this?

Example: THE CAT RAN FAR becomes…

THE CTR ANF AR

Gene Mutation Disorders

Progeria: defective Lamin A protein makes the nucleus unstable and leads to premature aging

Albinism – autosomal recessive

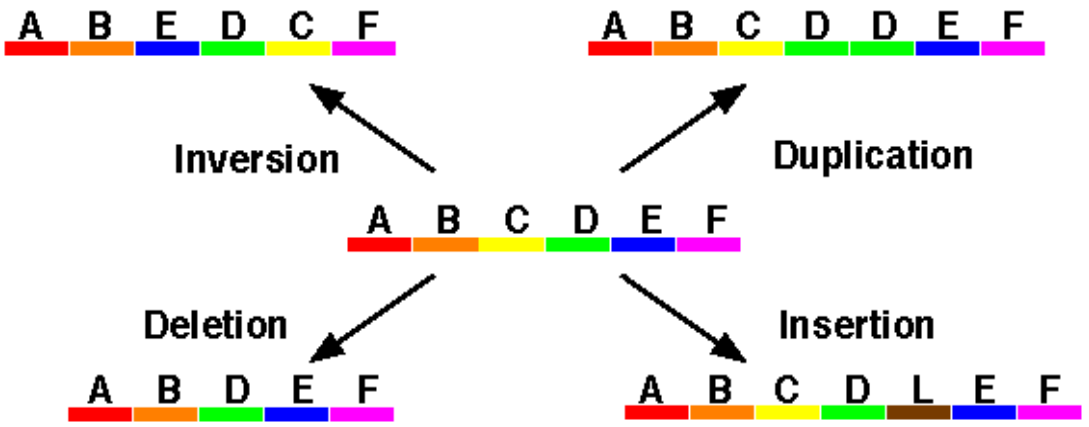
QQ#4: What is the difference between a gene and a chromosome mutation?

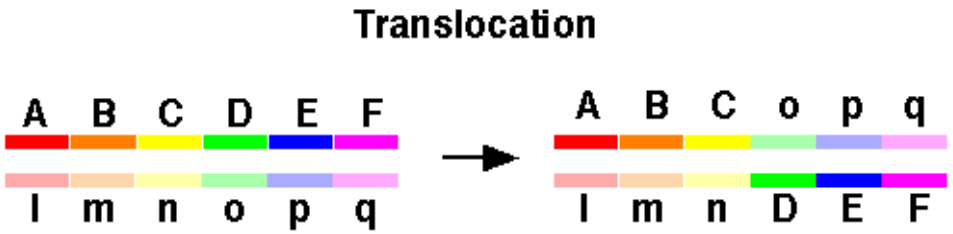
Chromosomal Mutations

- Whenever a chromosomal mutation occurs, there is a change in the number or structure of chromosomes

Types of Chromosomal Mutations

- Types:
  - Deletion
  - Duplication
  - Inversion
  - Translocation
  - Insertion



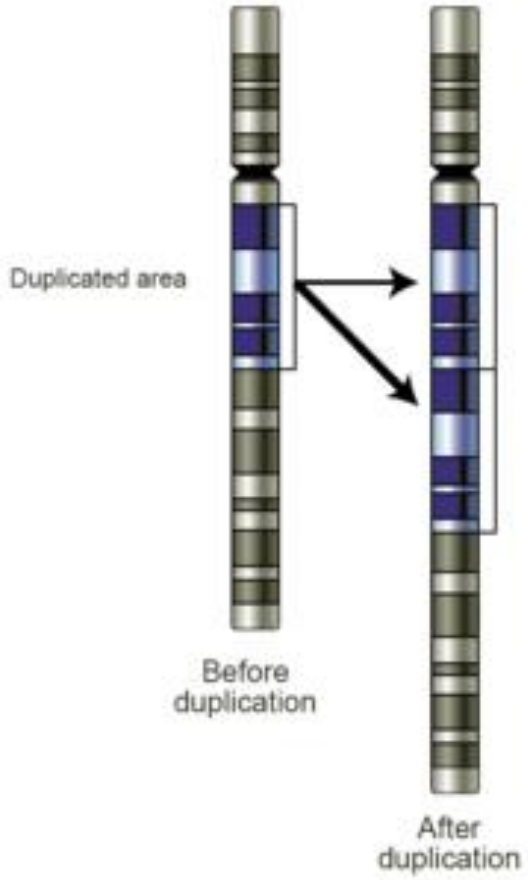


Deletions

- Involves the loss of part of a chromosome
  - Ex: Cri-du-chat Syndrome
    - loss of part of chromosome 5
    - The disorder is characterized by
      - intellectual disability
      - high-pitched cry: cat-like
      - delayed development
      - distinctive facial features
  - Ex: Turner Syndrome
    - cells are missing all or part of an X chromosome.
    - The condition only occurs in females.
    - Women are infertile

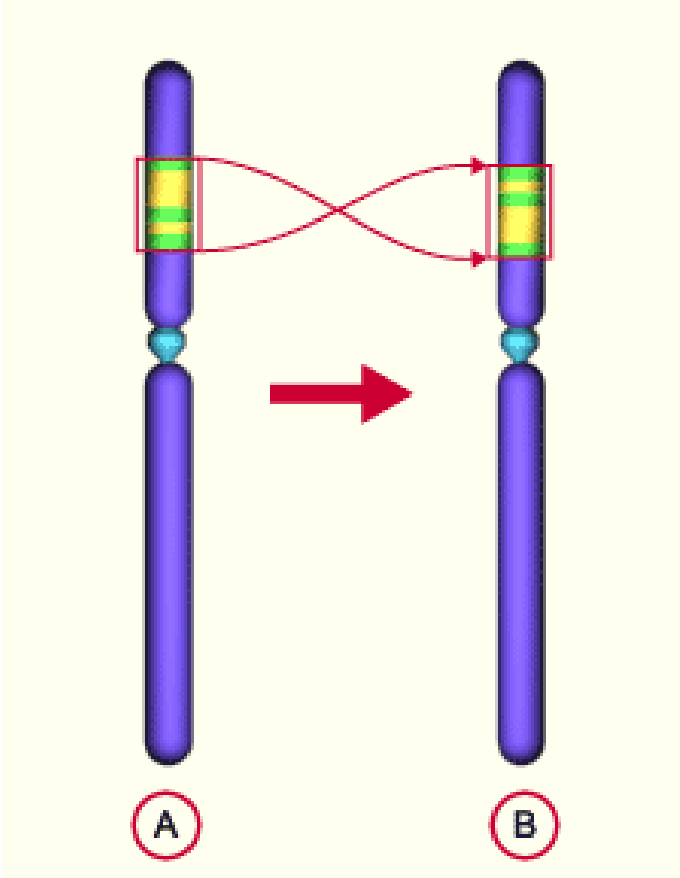
Duplication

- A segment of a chromosome is repeated
  - Ex: Fragile-X Syndrome
    - Part of the X chromosome has a repeat
    - X-linked: more common in boys
    - Causes intellectual disabilities



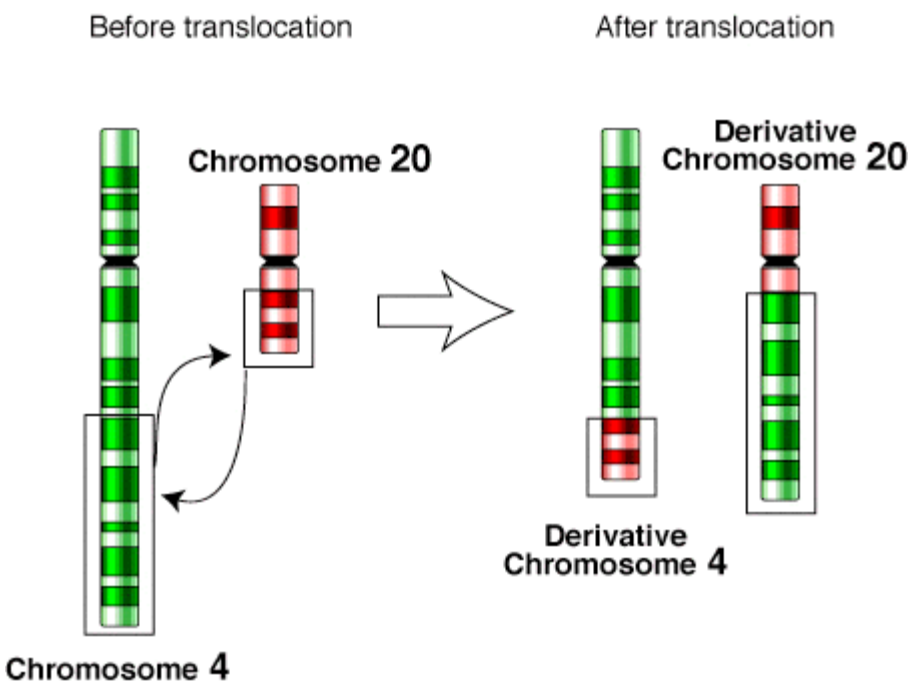
Inversion

- A portion of the chromosome has broken off, turned upside down and reattached, therefore the genetic material is inverted.
  - Typically not noticeable



Translocation

- Part of one chromosome breaks off and attaches to another, non-homologous chromosome



- Is the cause of some: cancer (leukemia), infertility, and 5% of Down's Syndrome Cases

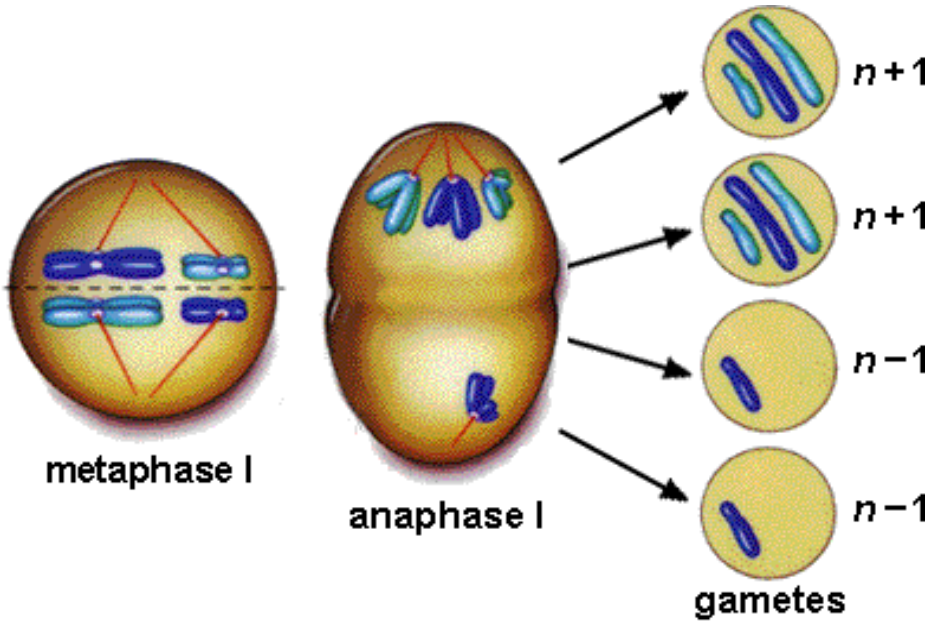
QQ#5

Try to think of a way to remember each of the following chromosomal mutations:

- Types:
  - Deletion
  - Duplication
  - Inversion
  - Translocation
  - Insertion

Nondisjunction

- A mutation affecting the whole chromosome.
- Cause by the failure of homologous chromosomes to separate normally during meiosis
- Cells are left with too many or too few chromosomes.

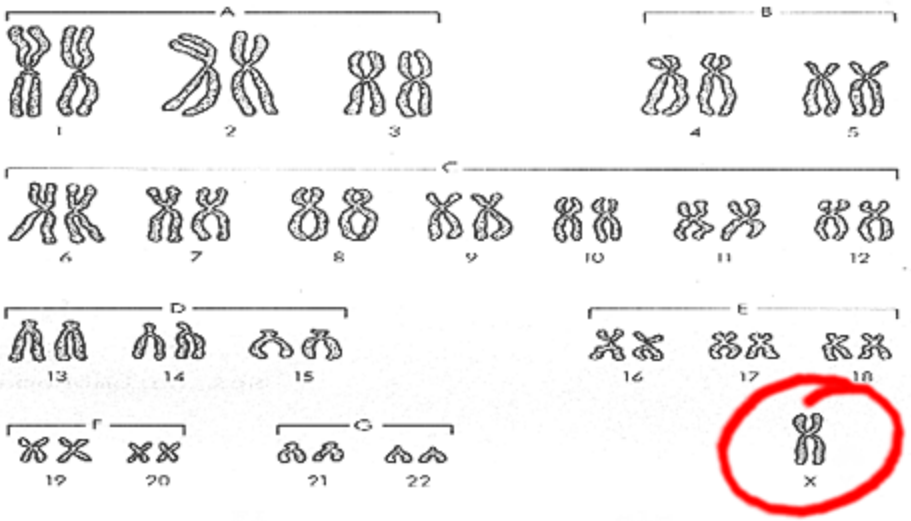


Klinefelter Syndrome

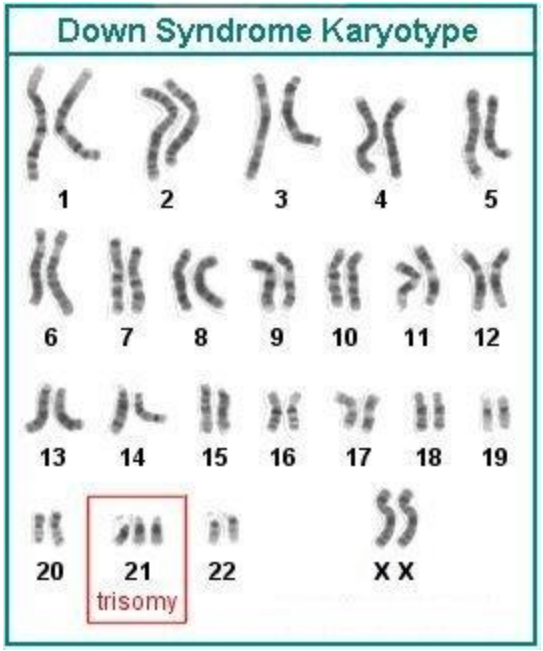
- Genetic Disorders in Males
- XXY
- 1/2000 live births
- Sterile
- Extra X inactivated, but some female body characteristics

Turner Syndrome

- Monosomy X (females with only one X chromosome)



Nondisjunction Disorders: Down's Syndrome



Down's Syndrome - Trisomy 21

- chromosomal condition characterized by the presence of an extra copy of genetic material on the 21st chromosome
- Causes intellectual disability and often heart and other health problems