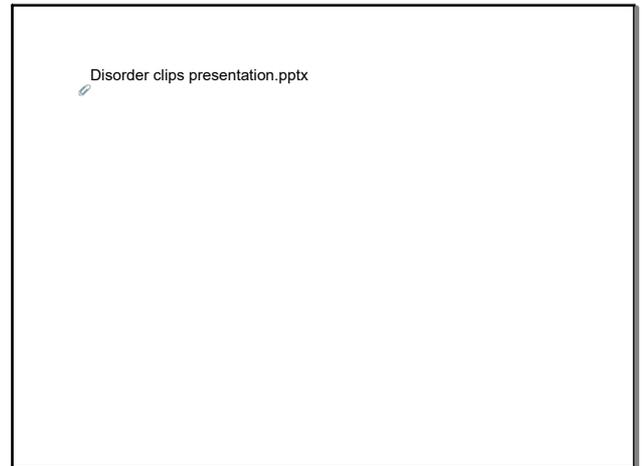


Mar 27-11:39 AM



Apr 11-12:37 PM

**Human Genetics**

**Mutation - a spontaneous change in DNA**

Types of mutations --

- > gene or base: caused by a single base error
- > chromosome mutation: a part of a chromosome is to blame
- > non-disjunction: a whole chromosome is involved.

Disorder: a negative result of a mutation

Apr 22-12:46 PM

Types of mutations --

- > gene or base: caused by a single base error
- > \*
- > \*
- > \*
- > chromosome mutation: a part of a chromosome is to blame
- > \*
- > \*
- > \*
- > \*

Apr 22-12:46 PM

**Mutation - a spontaneous change in DNA**

Types of mutations --

- > gene or base: caused by a single base error
  - substitution (point): one base replaces another
  - addition: one base is inserted
  - deletion: one base is deleted
- > chromosome mutation: a part of a chromosome is to blame
  - deletion: part of a chromosome is missing
  - duplication: part of a chromosome is duplicated
  - inversion: part of a chromosome breaks off and reinserts backwards
  - translocation: part of a chromosome from an unrelated chromosome attaches
- > non-disjunction: a whole chromosome is involved.

Disorder: a negative effect of a mutation

Apr 22-12:46 PM

Normal: MEN ARE ALL WAY TOO SAD ANY ONE DAY

1. MEN ARE ALL WAY TOO SAD<sup>YAD ENO YNA</sup>
2. MEN ARE ALL WAY TOO
3. MEN ARE ALL WAY TOO SAD ANY ONE DAY<sup>ANY ONE DAY</sup>
4. MEN ARE ALL WAY TOO SAD ANY ONE DAY<sup>ONE FLY CAN</sup>
5. MEN ARE ALL WAY TOO SA<sup>P</sup> ANY ONE DAY
6. MEN ARE ALL WA<sup>P</sup> YTO OSA DAN YON EDA Y
7. MEN ARE ALL WAT OOS ADA NYO NED AY

Apr 23-9:26 AM

Normal DNA: TAC CGC TTC GGC ATC  
 Normal mRNA: AUG GCG AAG CCG UAG  
 Amino acids:

Apr 22-10:03 AM

Normal DNA: TAC CGC TTC GGC ATC  
 Normal mRNA: AUG GCG AAG CCG UAG  
 Amino acids: Met Ala Lys Pro stop

Apr 22-10:03 AM

Normal DNA: TAC CGC TTC GGC ATC  
 Mutation 1: TAC CGC TTC GGT ATC  
 Type of mutation:

Apr 22-10:03 AM

Normal DNA: TAC CGC TTC GGC ATC  
 Mutation 2: TAC CGC ATT CGG CAT C  
 Type of mutation:

Apr 22-10:03 AM

Normal DNA: TAC CGC TTC GGC ATC  
 Mutation 1: TAC CGC TTC GGT ATC  
 Type of mutation: substitution aka point  
 mRNA: AUG GCG AAG CCA UAG  
 Mutant protein: Met Ala Lys Pro stop  
 (Normal protein: Met Ala Lys Pro stop)

Apr 22-10:03 AM

Normal DNA: TAC CGC TTC GGC ATC  
 Mutation 2: TAC CGC ATT CGG CAT C  
 Type of mutation:

Apr 22-10:03 AM

Normal DNA: TAC CGC TTC GGC ATC  
 Mutation 2: TAC CGC ATT CGG CAT C  
 Type of mutation: **addition**  
 mRNA: AUG GCG AAG CCA UAG  
 Mutant protein: Met Ala stop  
 (Normal protein: Met Ala Lys Pro stop)

Apr 22-10:03 AM

Normal DNA: TAC CGC TTC GGC ATC  
 Mutation 3: TAC TGC TTC GGC ATC  
 Type of mutation:

Apr 22-10:03 AM

Normal DNA: TAC CGC TTC GGC ATC  
 Mutation 3: TAC TGC TTC GGC ATC  
 Type of mutation: **point** aka **substitution**  
 mRNA: AUG ACG AAG CCA UAG  
 Mutant protein: Met Thr Lys Pro stop  
 (Normal protein: Met Ala Lys Pro stop)

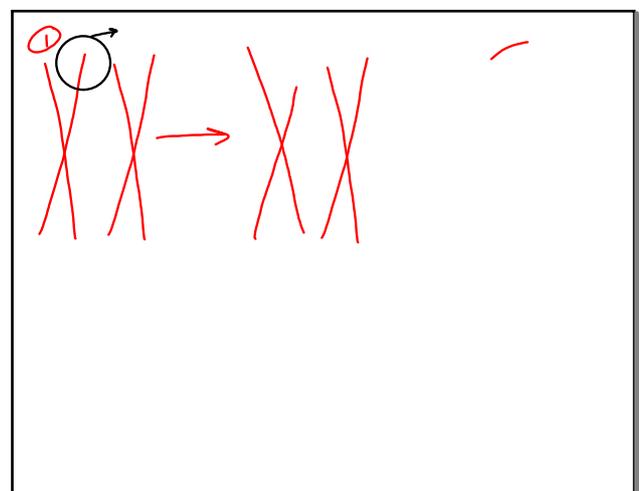
Apr 22-10:03 AM

Normal DNA: TAC CGC TTC GGC ATC  
 Mutation 4: TAC CGC (not on sheet) TCG GCA TC  
 Type of mutation:

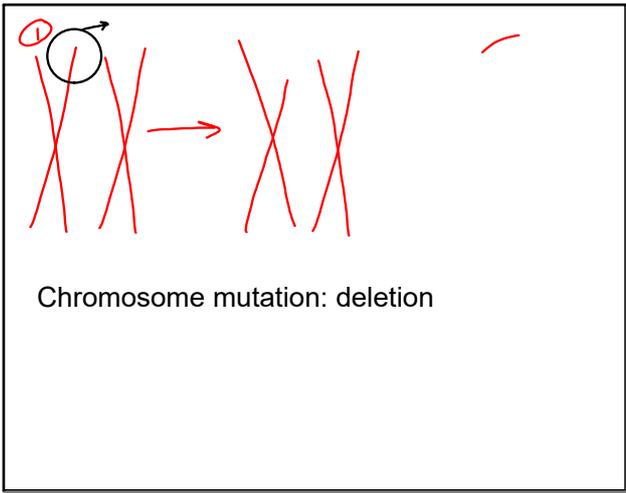
Apr 22-10:03 AM

Normal DNA: TAC CGC TTC GGC ATC  
 Mutation 4: TAC CGC (not on sheet) TCG GCA TC  
 Type of mutation: **deletion**  
 mRNA: AUG GCG AGC CGU AG  
 Mutant protein: Met Ala Ser Aug  
 (Normal protein: Met Ala Lys Pro stop)

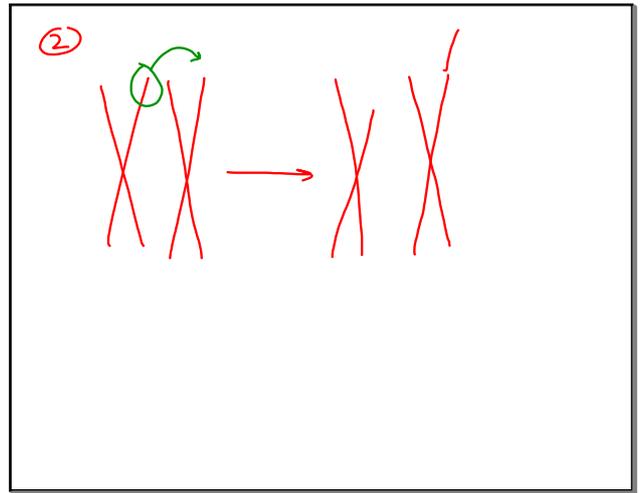
Apr 22-10:03 AM



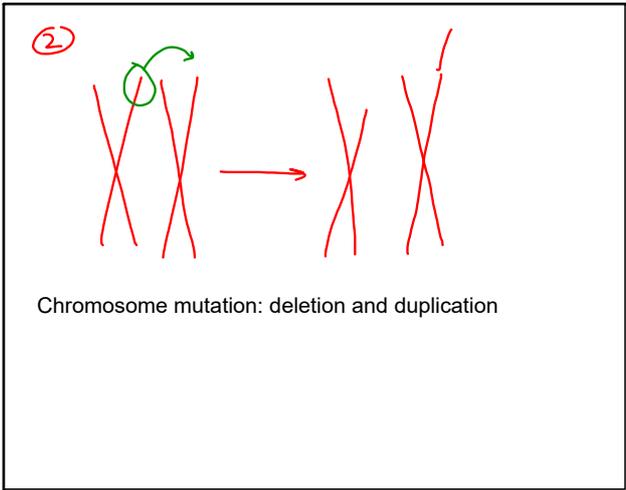
Apr 22-10:25 AM



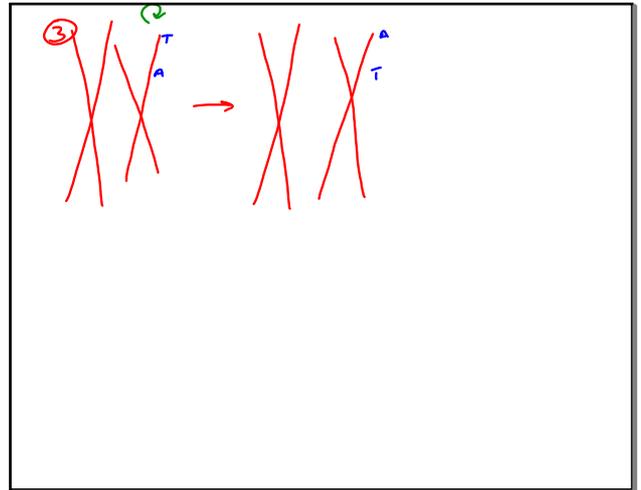
Apr 22-10:25 AM



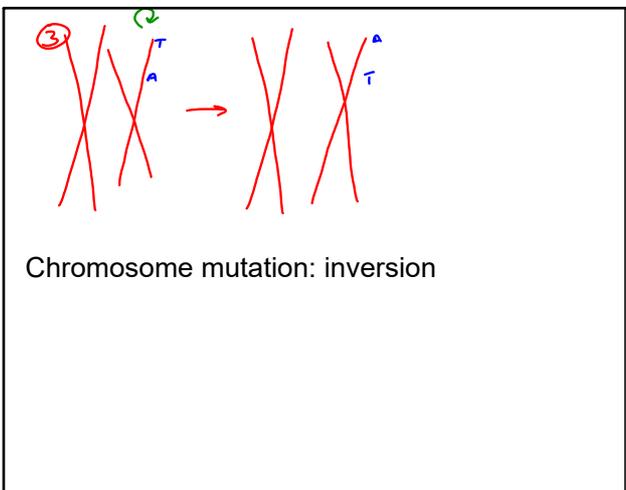
Apr 22-10:26 AM



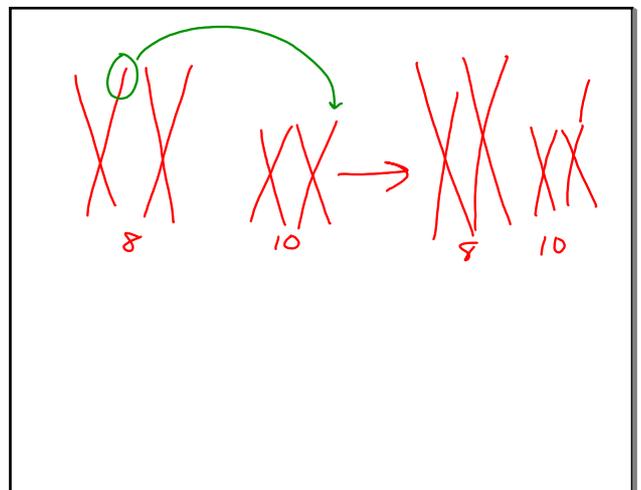
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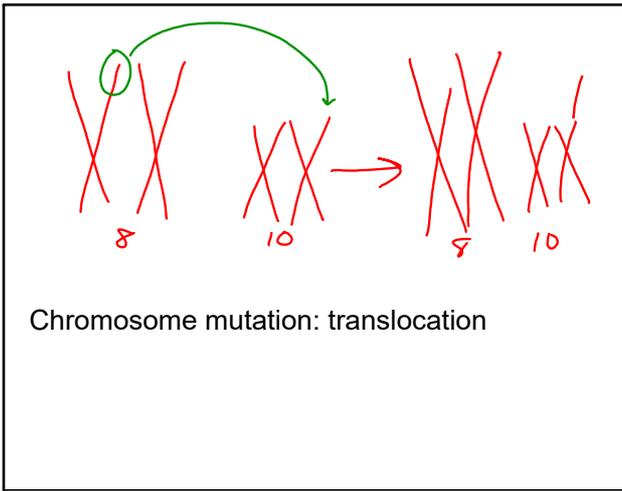
Apr 22-10:27 AM



Apr 22-10:27 AM



Apr 22-10:29 AM



Apr 22-10:29 AM

What kind of mutation sheet.

Apr 4-1:15 PM

**Down Syndrome (trisomy 21)**  
 Growth failure, Mental retardation, Flat back of head, Abnormal ears, Many "tongue" on finger tips, Plum creases, Special skin ridge patterns, Unilateral or bilateral absence of one rib, Intestinal blockage, Umbilical hernia, Abnormal pelvic, Diminished muscle tone, Broad flat face, Slanted eyes, Epicanthic eyelids, Short nose, Short and broad hands, Small and arched palate, Big, wrinkled tongue, Dental anomalies, Congenital heart disease, Enlarged colon, Big toes widely spaced.

**Klinefelter Syndrome (trisomy XXY)**  
 Physical features: Poor heard growth, Narrow shoulders, Wide hips, Long arms and legs, Small testicular size, Female-type pubic hair pattern, Female-type breast development, Tendency to grow faster chest hairs.

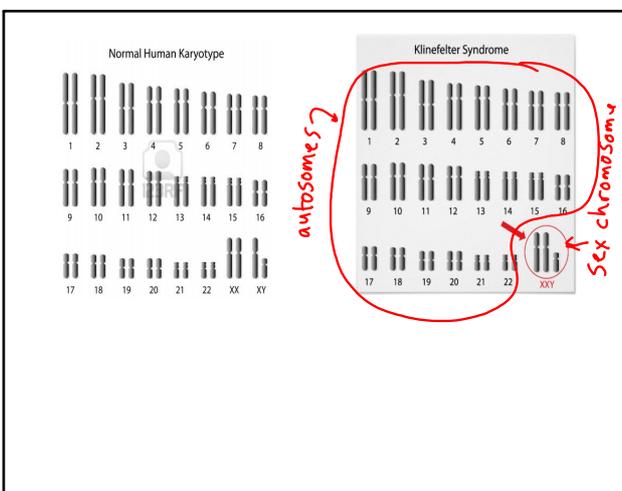
**Turner Syndrome (monosomy X)**  
 Short stature, Low hairline, Shield-shaped thorax, Widely spaced nipples, Shortened metacarpal IV, Small fingers/nails, Brown spots (nevi), Characteristic facial features, Fold of skin, Constriction of aorta, Poor breast development, claw deformity, Rudimentary ovaries, Gonadal streak (underdeveloped gonadal structure), No menstruation.

Possible retardation + heart problems, slanted eyes, big tongue (under Down Syndrome)  
 male with some female characteristics (under Klinefelter Syndrome)  
 Infertile female that doesn't mature sexually (under Turner Syndrome)

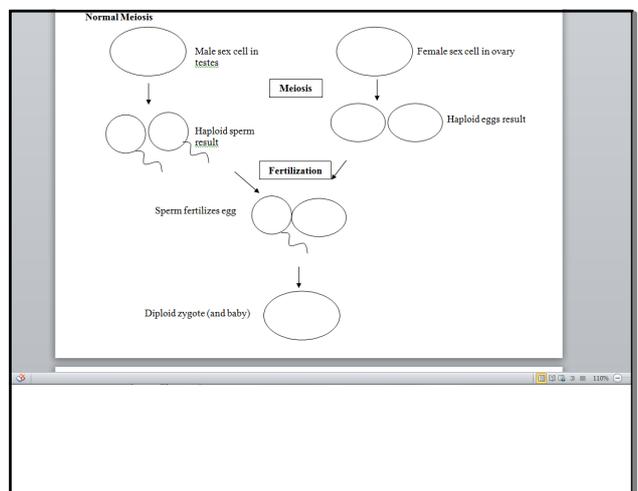
Apr 26-7:43 AM

**Non-disjunction:** when chromosomes don't separate correctly in meiosis, resulting in a gametes with too many or too few chromosomes. If such a gamete is involved in fertilization, the zygote (and baby if it develops) will have too few or too many chromosomes.

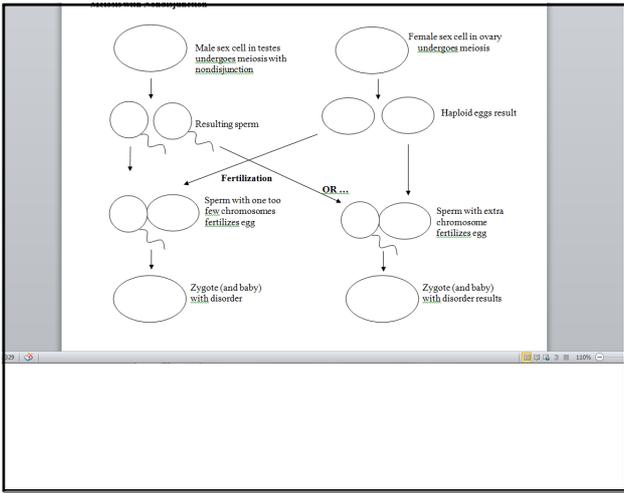
Apr 24-10:21 AM



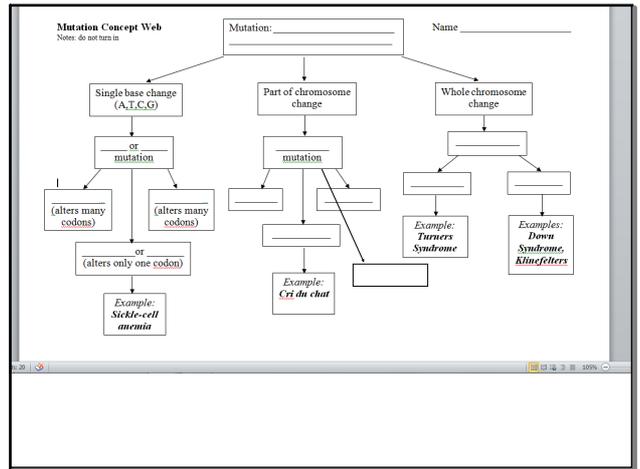
Apr 24-10:23 AM



Apr 24-12:28 PM



Apr 24-12:28 PM



Apr 25-8:59 AM

Do:  
 • Defective chromosome structure  
 • Review sheet

Apr 4-1:15 PM



Mar 27-11:39 AM



Apr 6-8:26 AM

## Attachments

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Disorder clips presentation.pptx