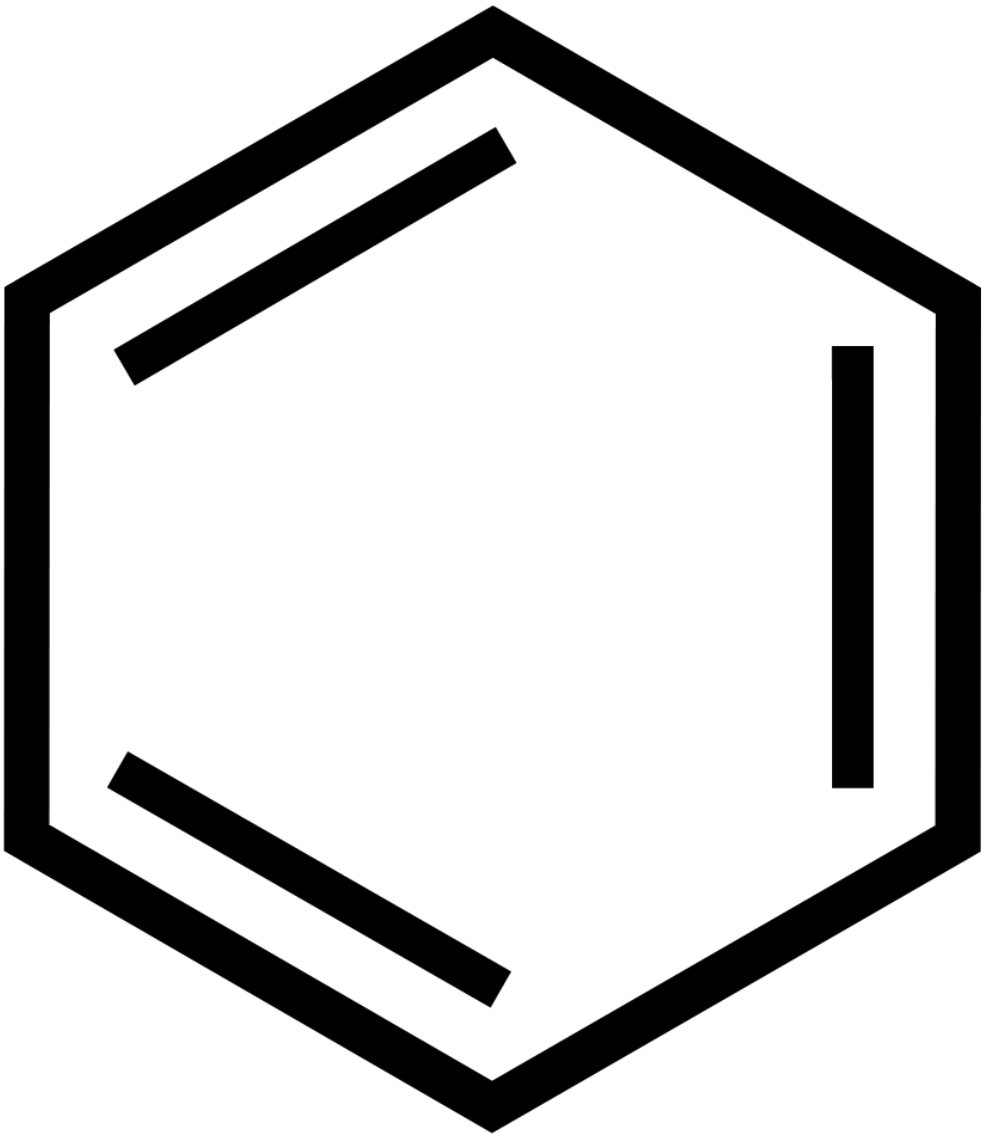

CLUTCH

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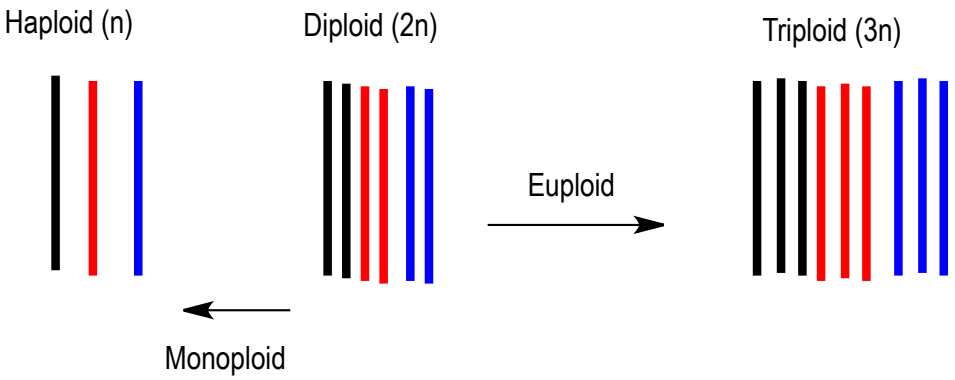
CONCEPT: CHROMOSOMAL MUTATIONS: ABERRANT EUPLOIDY

- **Chromosomal mutations** describe alterations in chromosome structure or number of chromosomal copies
  - □ There are two _____ of chromosomal mutations
    1. **Aberrant euploidy** refers to changes in the whole set of chromosomes
    2. **Aneuploidy** refers to changes in parts of a single, or few chromosomes

Aberrant Euploidy

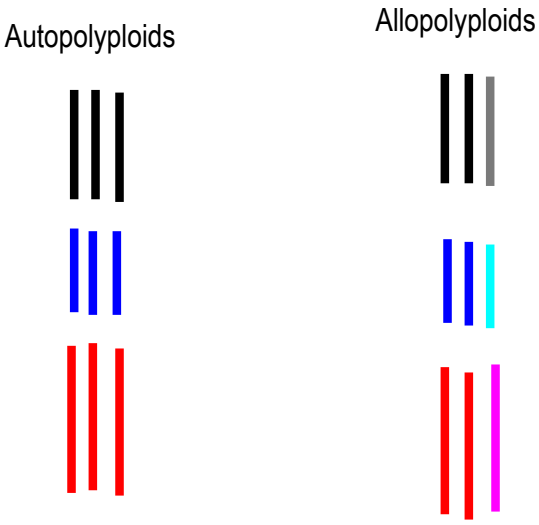
- There are a number of _____ to describe various types of chromosomal mutations
  - □ **Euploid** describes organisms with multiples of the basic chromosome set
    - Can contain more or fewer numbers of the normal set of the chromosomes
  - □ **Monoploids** describes normally diploid organisms that contain only one chromosome set
    - **Parthenogenesis** describes the development of unfertilized egg into embryo without fertilization
      - Ex: male bees, wasps, ants

EXAMPLE:



  - □ **Polyploids** have _____ than two chromosome sets
    - *Triploid (3n), tetraploid (4n), pentaploid (5n), hexaplid (6n)*
    - They are divided into two classes:
      - **Autopolyploids** contain multiple chromosome sets from within one species
      - **Allopolyploids** contain multiple chromosome sets from two closely related species
        - Sets are called **homeologous** (semi-homologous)
    - Mostly occurs in plants, but can occur in animals too

EXAMPLE:



Autopolyploidy

□ *Autopolyploids* are typically triploids

- Usually, they are _____ – but their gametes receive either two or one chromosome from each pair

- Creates **aneuploidy** which describes organisms with mixtures of diploid and haploid chromosomes

  - Not all triploid pairs segregate equally

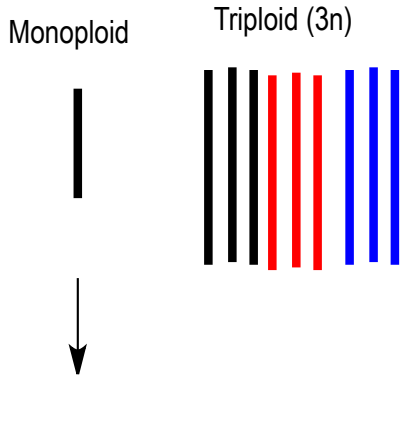
□ There are many classes of autopolyploids

- **Monoploids** – formed by using a haploid cell meant for fertilization as an embryo

- **Autotriploids** – have triploids in each set (Ex: bananas) (3n)

- **Autotetroploids** – typically result in larger sizes or flowers in plants/crops

EXAMPLE:



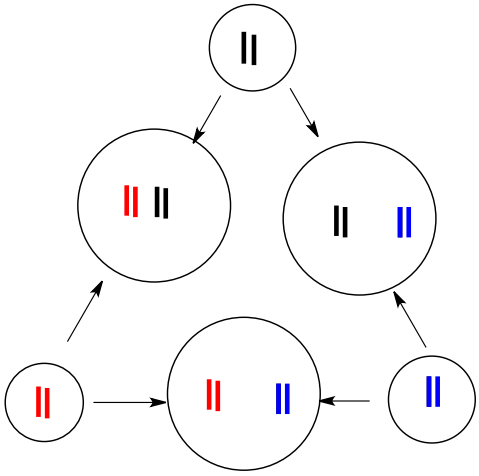
Allopolyploidy

□ *Allopolyploids* describes _____ that is a hybrid of two or more species

- Usually, they are sterile – but their gametes receive either two or one chromosome from each pair

- Most of these are synthetically created for crop plants (Ex: cotton and wheat)

EXAMPLE:



□ **Endopolyploidy** describes diploid organisms where certain cells are polyploidy

- Ex: Flowering plants, gut of mosquito larva, human liver cells

□ **Colichicine** is a chemical that can induce nondisjunction in a laboratory

PRACTICE

1. Which of the following chromosomal mutations increases the amount of genetic material from all chromosomes?
    a. Aberrant Euploidy
    b. Aneuploidy
    c. Monoploidy
    d. Tetraploidy

2. True or False: Autopolyploids contain multiple chromosomal sets from two closely related species
    a. True
    b. False

3. A species has 2n = 20. How many chromosomes will be found per mutant cell in an autotriploid organism.
    a. 10
    b. 20
    c. 30
    d. 60

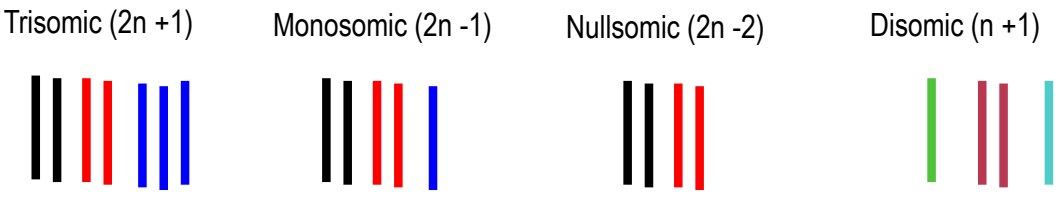
CONCEPT: CHROMOSOMAL MUTATIONS: ANEUPLOIDY

- **Chromosomal mutations** describe alterations in chromosome structure or number of chromosomal copies
  - □ There are two _____ of chromosomal mutations
    1. **Aberrant euploidy** refers to changes in the whole set of chromosomes
    2. **Aneuploidy** refers to changes in parts of a single, or few chromosomes

Aneuploidy

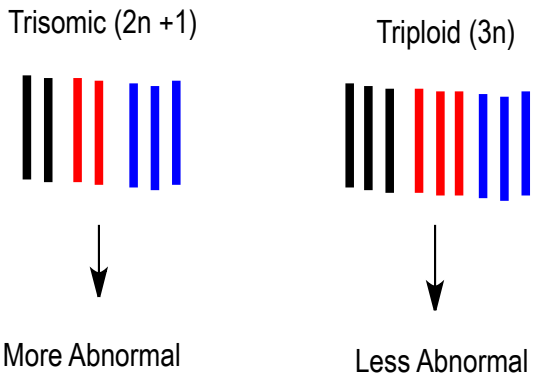
- **Aneuploidy** refers to organisms with chromosomes mutations found in some chromosomes, but not all
  - □ There are many different _____ of aneuploidy
    - **Trisomic** (2n +1) : Example includes Down Syndrome and Klinefelters
    - **Monosomic** (2n -1) : Example includes turner syndrome
    - **Nullsomic** (2n – 2) : More rare than other types
    - **Disomic** (n +1) : Occurs in haploids

EXAMPLE:



- □ **Nondisjuction,** which is the failure of chromosomes to separate properly during division, causes aneuploidy
  - Can occur in meiosis (most common) or early development mitosis (less common)
- □ Aneuploids are usually more abnormal that polyploids
  - **Gene balance**, which is the ratio of genes on one chromosome to genes on other chromosome
  - **Gene dosage**, which is the relation between numbers of gene copies and the amount of gene product
    - Dosage is more severe when there is a aberrant dose of a few vs. aberrant dose of all genes

EXAMPLE:



PRACTICE

1. Which of the following chromosomal mutations increases the amount of genetic material from only some chromosomes?
    a. Aberrant Euploidy
    b. Aneuploidy
    c. Monoploidy
    d. Tetraploidy

2. True or False: Aneuploids are more abnormal that polyploids
    a. True
    b. False

3. A species has 2n = 20. How many chromosomes will be found per mutant cell in an monosomic organism.
    a. 10
    b. 19
    c. 20
    d. 21

CONCEPT: CHROMOSOMAL REARRANGEMENTS: OVERVIEW

● **Chromosomal rearrangements** describe changes in chromosomal structure

□ There can be many _____ including: deletions, duplications, translocations, inversions

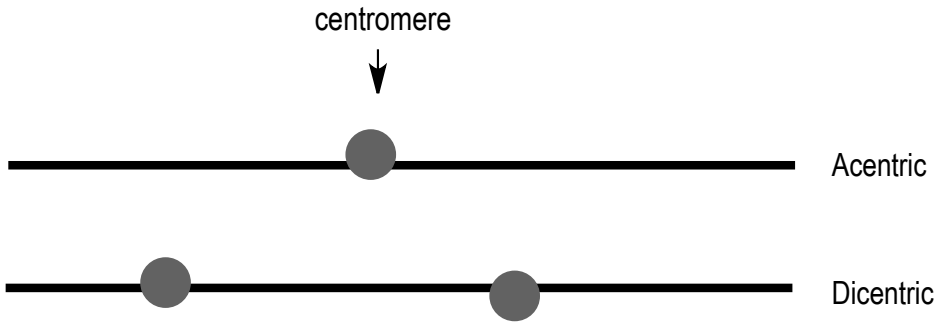
□ Rearrangements are named based on their occurrence in reference to the centromere

- **Acentric chromosomes** are chromosomes that lack a centromere due to a chromosomal rearrangement

- **Dicentric chromosomes** are chromosomes with 2 centromeres due to a chromosomal rearrangement

- **Anaphase bridge** occurs when a dicentric is simultaneously pulled to opposite poles in anaphase

EXAMPLE:



□ There are two rearrangement _____

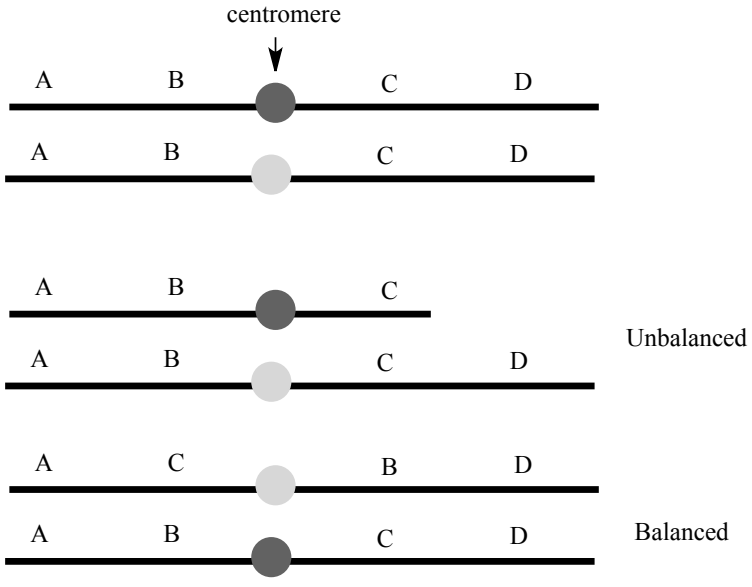
- **Unbalaned rearrangements** occur when there in a change of gene dosage of chromosomal segment

- Occur through deletion or duplication

- **Balanced rearrangements** occur when there is a change of gene order

- Occur through inversions or translocations

EXAMPLE:



PRACTICE

1. What is the name for a chromosomal rearrangement that causes a change in gene dosage?
    a. Altered
    b. Uneven
    c. Unfair
    d. Unbalanced

2. Inversions cause what type of chromosomal rearrangement?
    a. Unbalanced
    b. Uneven
    c. Balanced
    d. Even

3. A chromosome with two centromeres is called what?
    a. Bichromosome
    b. Bivalent
    c. Dicentric
    d. Acentric

CONCEPT: CHROMOSOMAL REARRANGEMENTS: DELETIONS

● **Deletions** are a type of chromosomal rearrangement that results in a loss of a part of a chromosome arm

□ Deletions require that there are two breaks to cut out the segment

- The deleted chromosomal segment contains no centromere – and is lost after cell division

□ There are many types of _____

Deletion type	**Location of deletion**
Intragenic deletion	Within a gene
Multigenic deletions	Multiple genes
Terminal Deletions	End of a chromosome
Intercalary Deletions	Interior of a chromosome

● Deletions cause a couple of important _____

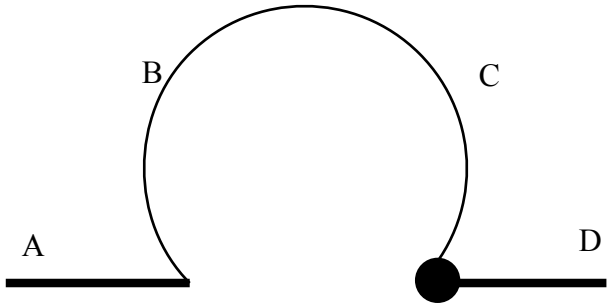
□ **Pseuodominance** occurs when the deletion allows for a single recessive allele to show its phenotype

□ A **deletion loop** is created between between the normal homolog and its partner congaing the missing segment

- Allows for proper segregation during meiosis

□ A human example of a deletion is **Cri du chat syndrome** which is caused by a deletion in chromosome 5

EXAMPLE:



PRACTICE

1. An intragenic deletion is a deletion found where?
    a. In a centromere
    b. In a gene
    c. In multiple chromosomes
    d. In multiple genes

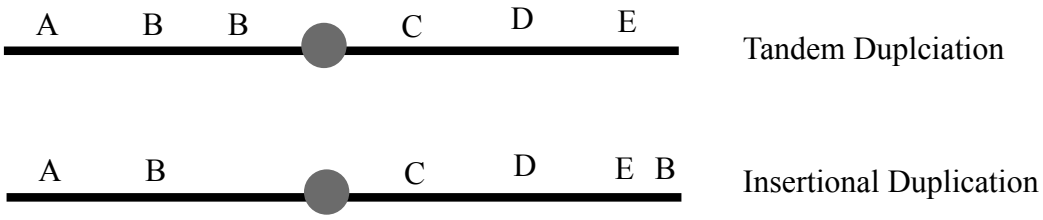
2. Deletions can cause what type of phenotype?
    a. Leaky allele
    b. Psudodominance
    c. Intercalary
    d. Faulty

3. Which of the following genetic diseases is an example of a chromosomal deletion?
    a. Down syndrome
    b. Familial down syndrome
    c. Cri du Chat syndrome
    d. Klinefelter's Disease

CONCEPT: CHROMOSOMAL REARRANGEMENTS: DUPLICATION

- **Duplications** are a type of chromosomal rearrangement where the chromosomal segment is doubled
  - □ There are many _____ of duplications
    - **Tandem duplication**s occur when the duplicate regions are located adjacent to each other
    - **Insertional duplication** occurs when the duplications regions are located elsewhere in the genomes
    - **Segmental Duplications** are 10-50 kilobases in length and encompass very large sections

EXAMPLE:



- □ Duplications have been extremely important in evolution
  - rDNA duplications allows for ribosome formation
  - Causes three copies of alleles

PRACTICE:

1. A person has a WT chromosome with the following segments. A B C • D E F G H. Which of the following shows how the chromosome would look after an insertional duplication?
    a. A A B B C • D E F G H
    b. A B C • D E F G H A B
    c. A B A B C • D E F G H
    d. A C • D E F G H

2. A person has a WT chromosome with the following segments. A B C • D E F G H. Which of the following shows how the chromosome would look after an tandem duplication?
    a. A B B C • D E F G H
    b. A B C • D E F G H B
    c. A B A C • D E F G H
    d. A C • D E F G H

CONCEPT: CHROMOSOMAL REARRANGEMENTS: INVERSIONS

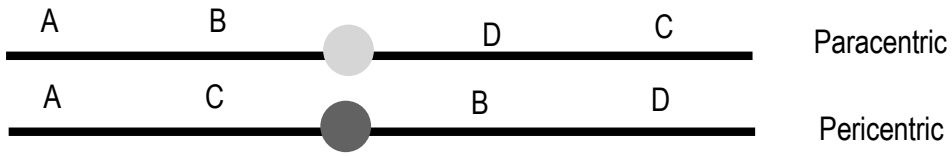
● An **inversion** describes a chromosomal rearrangement when the orientation of a segment is reversed

□ There two main _____ of inversions

- **Paracentric** describes inversions when the centromere is outside

- **Pericentric** describes inversion spanning the centromere

EXAMPLE:



□ **Inversion heterozygote** describes an organism with one normal chromosome, and one inverted chromosome

- This does not mean the alleles are heterozygous, just the _____

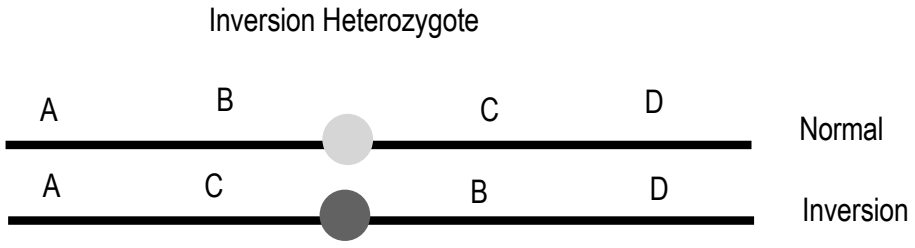
□ An **Inversion loop** forms between the inversion heterozygotes to assist in segregation

- Involves twisting one end of the inverted chromosome to pair with the uninverted chromosome

□ A **balancer chromosome** is often used to suppress cross over in genetic studies

- Balancer chromosomes contain multiple inversions and markers which disrupts synapsis and CO

EXAMPLE:



PRACTICE:

1. A person has a WT chromosome with the following segments. A B C • D E F G H. Which of the following shows how the chromosome would look after an paracentric inversion?
    a. A B C • D E F G H
    b. A B E D • C F G H
    c. A B C • D G H
    d. A B C • D G F E H

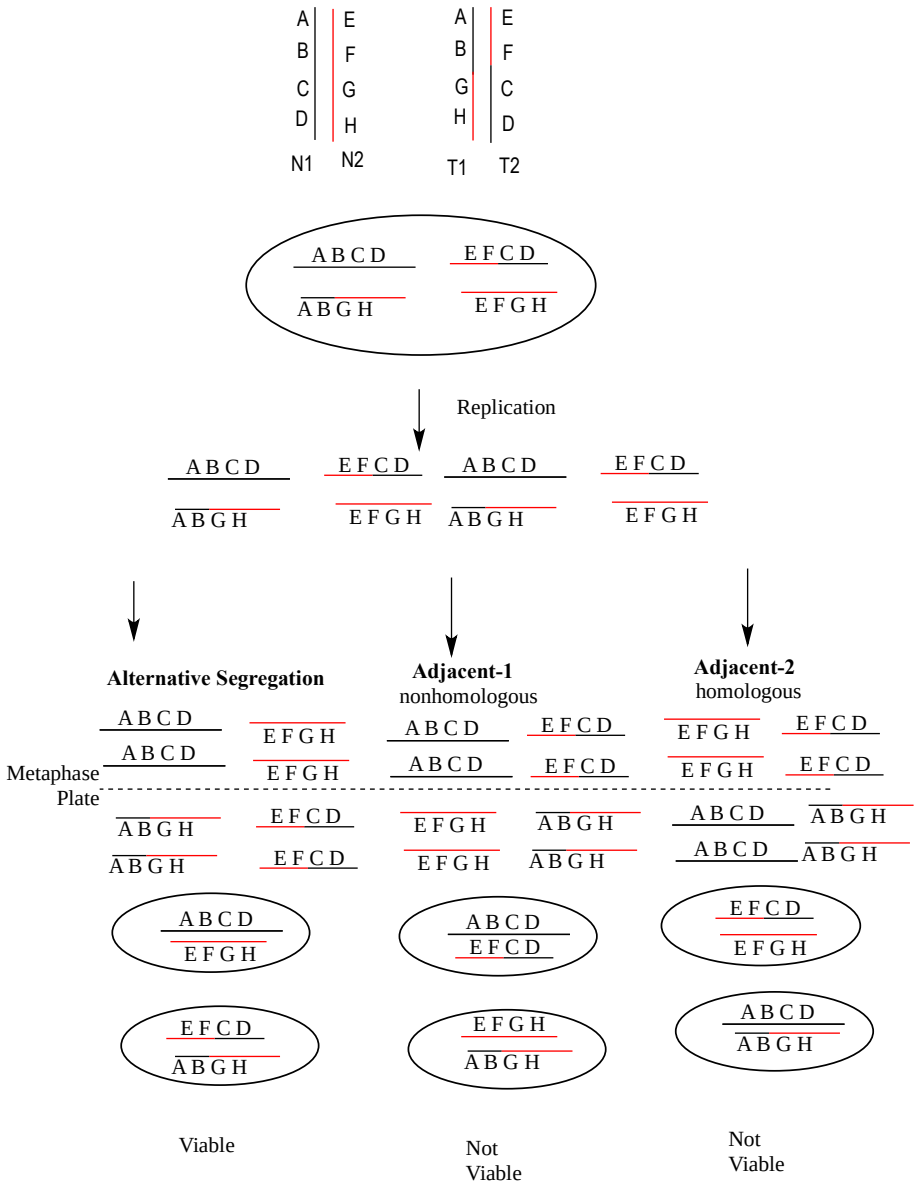
2. A person has a WT chromosome with the following segments. A B C • D E F G H. Which of the following shows how the chromosome would look after a pericentric inversion?
    a. A B C • D E F G H
    b. A B E D • C F G H
    c. A B C • D G H
    d. A B C • D G E F H

CONCEPT: CHROMOSOMAL REARRANGEMENTS: TRANSLOCATIONS

- **Translocations** describes when a chromosomal segment is moved to a different chromosome
  - □ **Reciprocal translocations** occur when chromosomes trade acentric fragments
    - There are _____ ways these translocations are sorted into gametes (N=Normal T=translocated)

1. **Adjacent-1 segregation** – segregates like: (N2 T1) and (N1 T2)
   - These are inviable
2. **Adjancent-2 segregation** – segregates like: (N1 T1) and (N2 T2)
   - These are inviable
3. **Alternative segregation** – segregates like : (T1 T2) and (N1 N2)
   - The gametes are viable

EXAMPLE:



□ **Robertsonian translocation** occurs when there are breaks at two short arms of two nonhomologous acrocentric

- Forms a chromosome that contains two long arms from two nonhomologous chromosomes

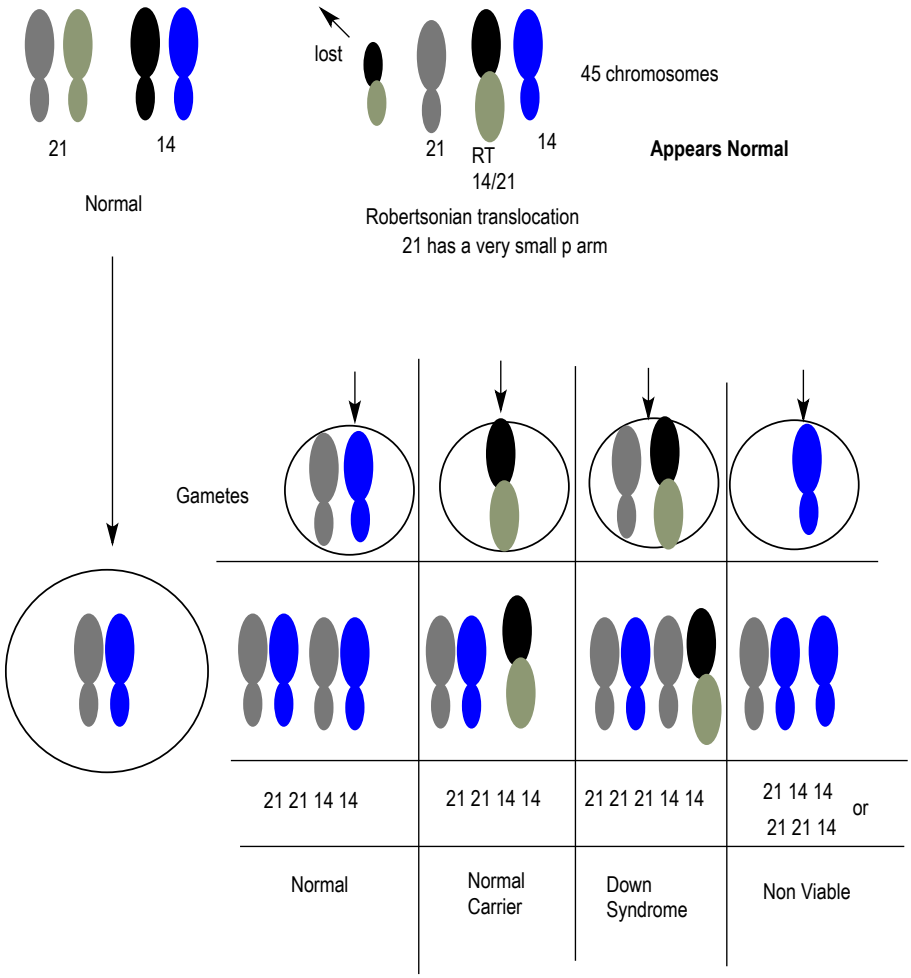
□ The *balanced form (*parent*)* results in _____ problems for the person

- This because it takes the place of two acrocentric chromosomes

□ The *unbalanced form* (child) it results in a chromosomal imbalance

- Example: Familial down syndrome

EXAMPLE:



PRACTICE

1. Which of the following represents the chromosomal segregation into gametes after a reciprocal translocation caused by adjacent-1 segregation? N=Normal chromosome T = Tanslocated chromosome
    a. (N2 T1) and (N1 T2)
    b. (N1 T1) and (N2 T2)
    c. (T1 T2) and (N1 N2)

2. Which of the following ways reciprocal translocated chromosomes are sorted produces viable gametes?
    a. Adjacent-1 segregation
    b. Adjacent-2 segregation
    c. Alternative segregation

3. An individual heterozygous for a reciprocal translocation has the following chromosomes. Which chromosomes do the gametes receive after alternative segregation?

A B • C D E F G	J K • L M N O P
A B • C D N O P	J K • L M E F G

a. A B • C D E F G and J K • L M E F G
b. A B • C D N O P and J K • L M E F G
c. A B • C D N O P and J K • L M N O P

4. An individual heterozygous for a reciprocal translocation has the following chromosomes. Which chromosomes do the gametes receive after adjacent-1 segregation?

A B • C D E F G	J K • L M N O P
A B • C D N O P	J K • L M E F G

d. A B • C D E F G and J K • L M E F G
e. A B • C D N O P and J K • L M E F G
f. A B • C D N O P and A B • C D E F G

5. An individual heterozygous for a reciprocal translocation has the following chromosomes. Which chromosomes do the gametes receive after adjacent-2 segregation?

A B • C D E F G	J K • L M N O P
A B • C D N O P	J K • L M E F G

   g. A B • C D E F G and J K • L M E F G
   h. A B • C D N O P and J K • L M E F G
   i. A B • C D N O P and A B • C D E F G

6. How many chromosomes does a person who is a carrier for familial down syndrome caused by a robertsonian translocation have?
   a. 45
   b. 46
   c. 47
   d. 48