



# Lecture 27:

## Chromosomal mutations

Course 281

# Lessons for life

No matter how  
many mistakes you  
make or how slow  
you progress  
You are still way  
ahead of everyone  
who isn't trying.  
Unknown

# AIMS

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- Understand the mutations on a chromosomal level.
- Understand the different types of chromosomal mutations.
- Understand the consequences of chromosomal mutations and compare them to point mutations.

# Mutations

## Mutation types

Chromosomal  
mutation

Point mutation

- **Chromosomal mutations:** are mutation that involve changes to the entire chromosome or sections of it. Also called **segmental mutations**.
- Chromosomal mutations are deviations from the normal condition of a chromosome both in structure and number.

# Chromosomal mutations

## Chromosomal mutations

Chromosome structure  
mutations

Chromosome number  
mutations

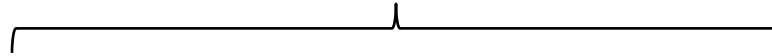
- **Chromosome structure mutations:** mutations that changes the amount of DNA in a chromosome or the orientation of the DNA within a chromosome.
- **Chromosome number mutation:** mutations that changes the number of chromosomes in a cell.

# Chromosomal structural mutations

Chromosomal mutations

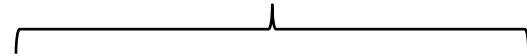


Chromosome structure mutations



**DNA content**  
in a chromosome

**DNA orientation**  
in a chromosome



Deletion

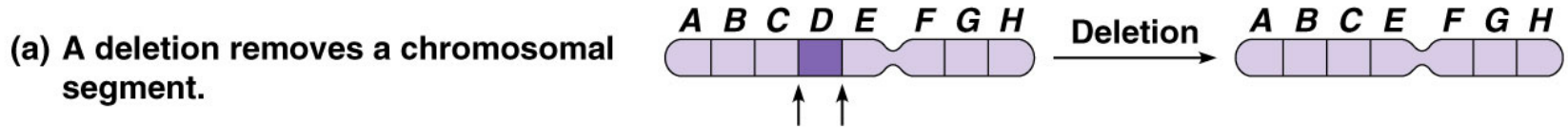
Duplication

Inversion

Translocation

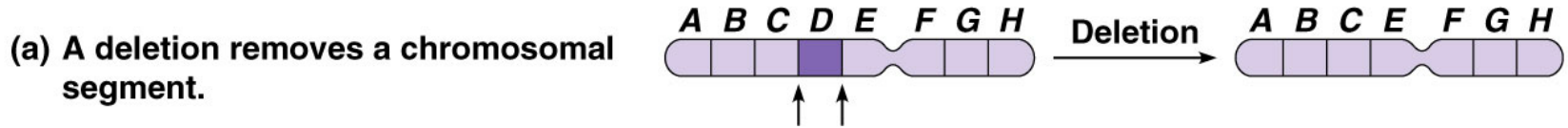
Let's focus on the chromosomal mutations that are associated with **DNA content**

# Chromosomal deletion



- Deletions reduce the amount of DNA of a chromosome compared to the normal chromosome.
- Small deletions are less harmful than large deletions.
- Consequences of deletions depend on the genes lost.

# Chromosomal deletion



- Example:
  - **Williams syndrome**
  - **Wolf-Hirschhorn syndrome**



# Chromosomal deletion

(a) A deletion removes a chromosomal segment.

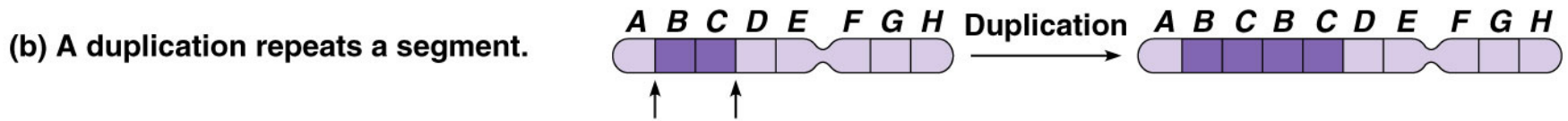


**Williams syndrome**



**Wolf-Hirschhorn syndrome**

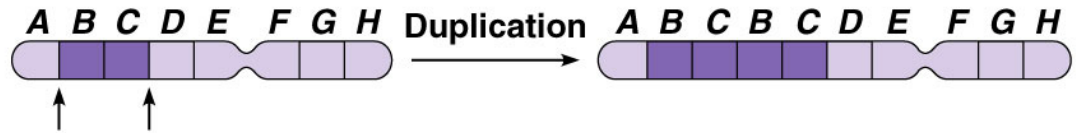
# Chromosomal duplication



- Duplications increase the amount of DNA of a chromosome compared to the normal chromosome.
- Having extra DNA in a chromosome may alter the number of genes and thus alter the amount of gene product.
- Example:
  - **Charcot–Marie–Tooth disease**

# Chromosomal duplication

(b) A duplication repeats a segment.



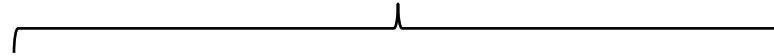
**Charcot-Marie-Tooth disease**

# Chromosomal structural mutations

Chromosomal mutations

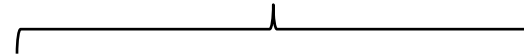


Chromosome structure mutations



**DNA content**  
in a chromosome

**DNA orientation**  
in a chromosome



Deletion

Duplication

Inversion

Translocation

Let's focus on the chromosomal mutations that are associated with **DNA orientation**

# Chromosomal inversion

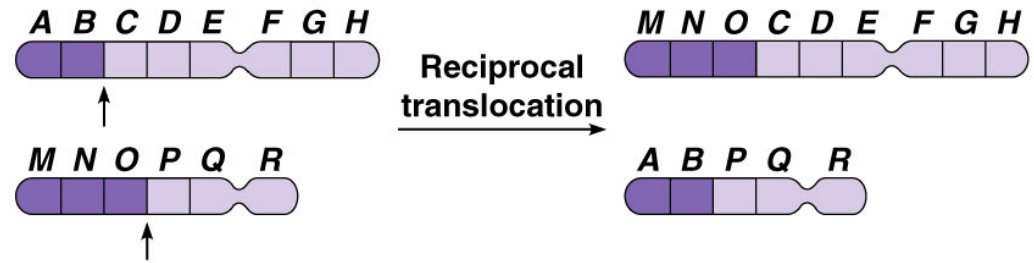
(c) An inversion reverses a segment within a chromosome.



- Inversions involve the breakage of a segment and reversed and reattached.
- This mutation changes the arrangement of genes.
- Example:
  - **There are some known inversions in the human genome but without an affect.**
  - **Why?**

# Chromosomal translocation

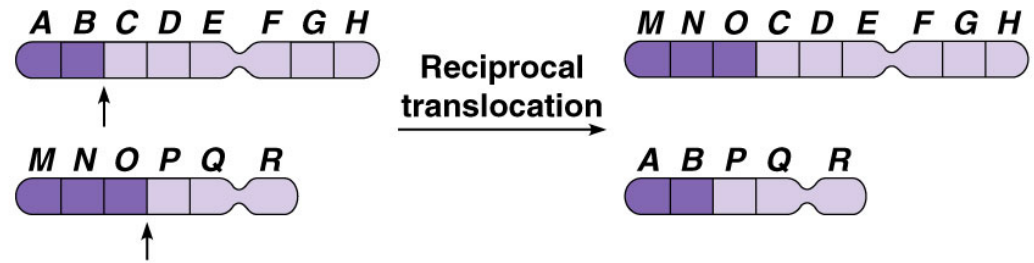
(d) A translocation moves a segment from one chromosome to another, nonhomologous one.



- Portions of one chromosome gets relocated to another chromosome.
- There are many types of this chromosomal mutations.

# Chromosomal translocation

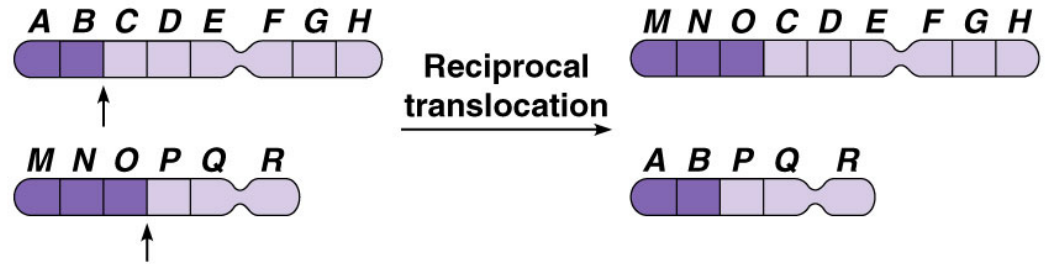
(d) A translocation moves a segment from one chromosome to another, nonhomologous one.



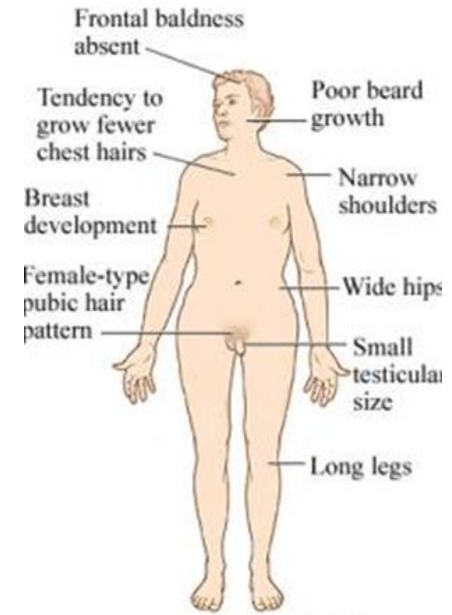
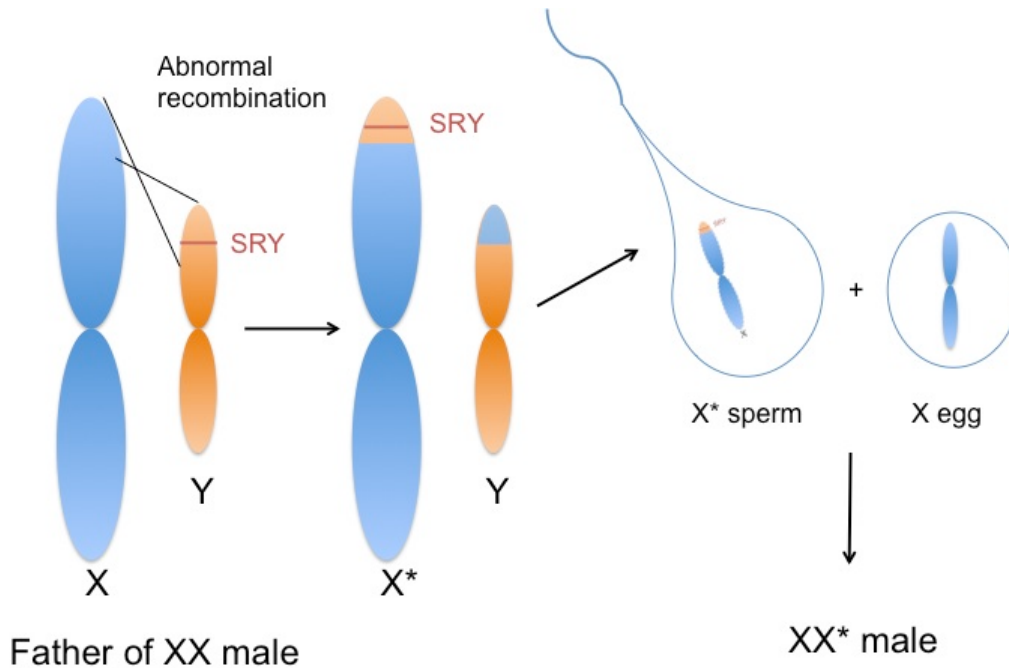
- Example:
  - **XX male syndrome!**
  - **Translocation of SRY gene from Y chromosome to X chromosome.**

# Chromosomal translocation

(d) A translocation moves a segment from one chromosome to another, nonhomologous one.



## XX male syndrome!





# Chromosomal mutations

## Chromosomal mutations

Chromosome structure mutations

Chromosome number mutations

- **Chromosome structure mutations:** mutations that changes the amount of DNA in a chromosome or the orientation of the DNA within a chromosome.
- **Chromosome number mutation:** mutations that changes the number of chromosomes in a cell.

# Chromosomal number mutations

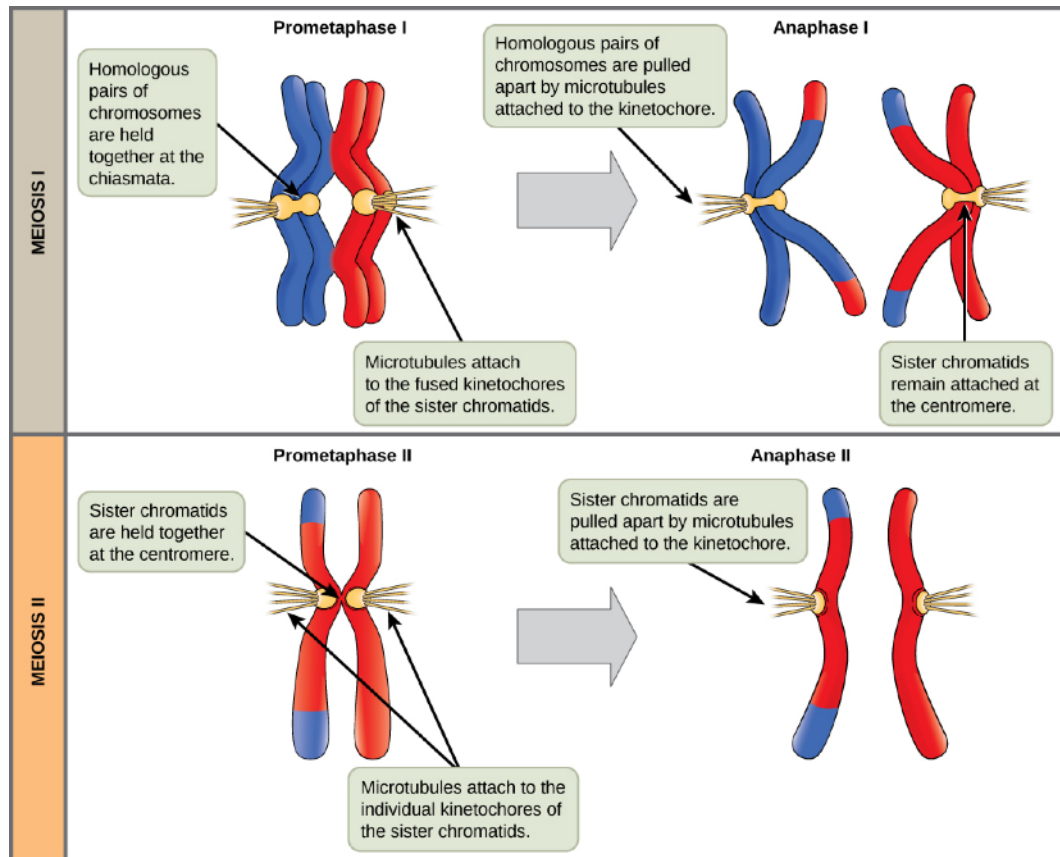


- Entire loss or gain of chromosomes due to lack of proper segregation of chromosomes during meiosis.
- This results in some gametes containing abnormal number of chromosomes.

**How do chromosome separate?**

# Chromosomal number mutations

Chromosomes segregate due to multiple factors. Failure in any one of these factors may result in abnormal number of chromosomes.



# Chromosomal number mutations



- **What is the proper normal number of chromosomes in a somatic human cell?**
- **What is the normal number of chromosomes in a gametic human cell?**
- **Somatic cells are called ....., and gametic cells are called .....**

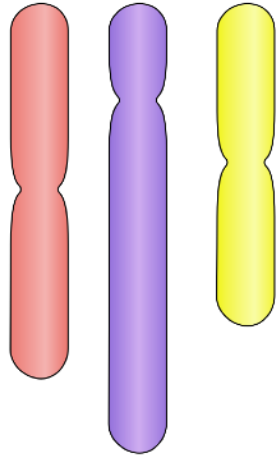
# Chromosomal number mutations

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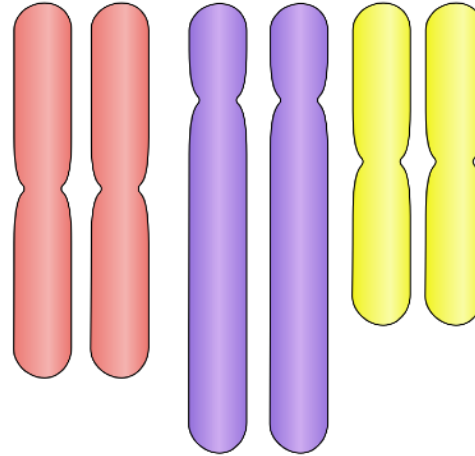
- **Euploid cell:** is a cell that contains one complete set of chromosomes of exact multiples of complete sets.
- **Aneuploid cell:** is a cell that has a chromosome number that is not an exact multiple sets of the haploid set of chromosomes.

# Chromosomal number mutations

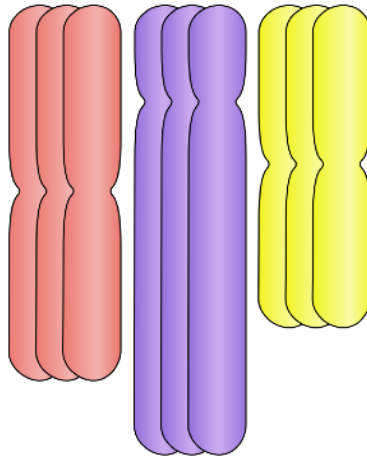
Haploid (N)



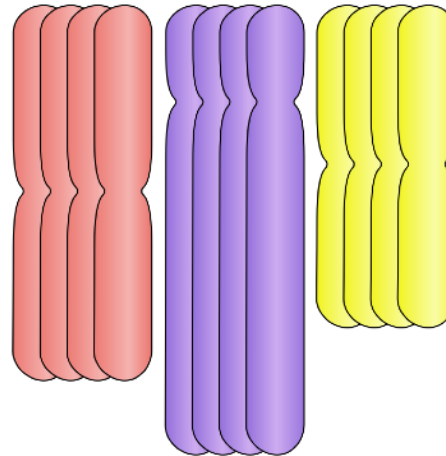
Diploid (2N)



Triploid (3N)



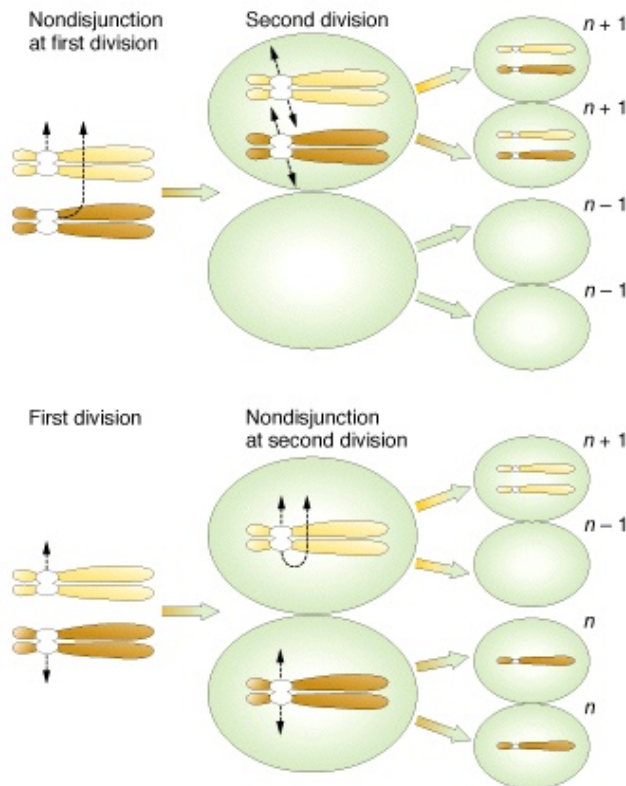
Tetraploid (4N)



# Chromosomal number mutations

## Types of aneuploidy

1. **Nullisomy:** loss of a chromosome pair (2 chromosomes).



**What do you think will happen?**

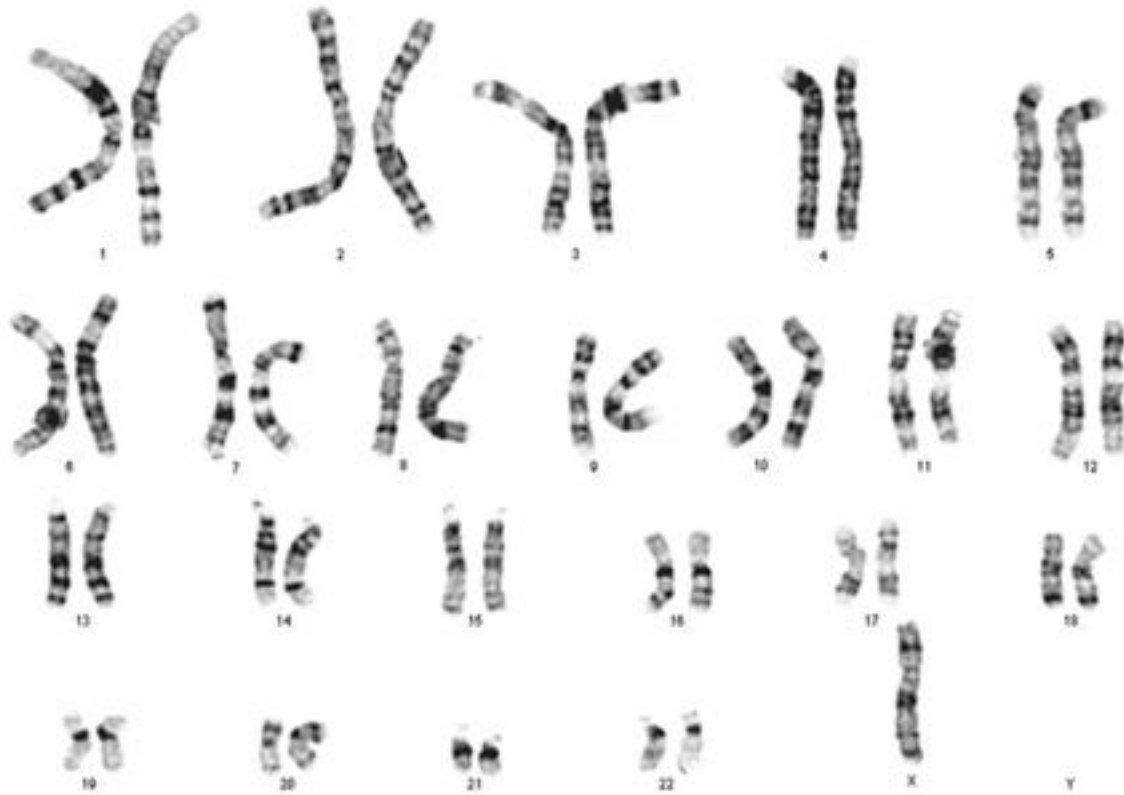
# Chromosomal number mutations

## Types of aneuploidy

2. **Monosomy:** loss of a single chromosome.



medgen.genetics.utah.edu



**Turner syndrome**



# Chromosomal number mutations

## Types of aneuploidy

3. **Trisomy:** an extra chromosome.

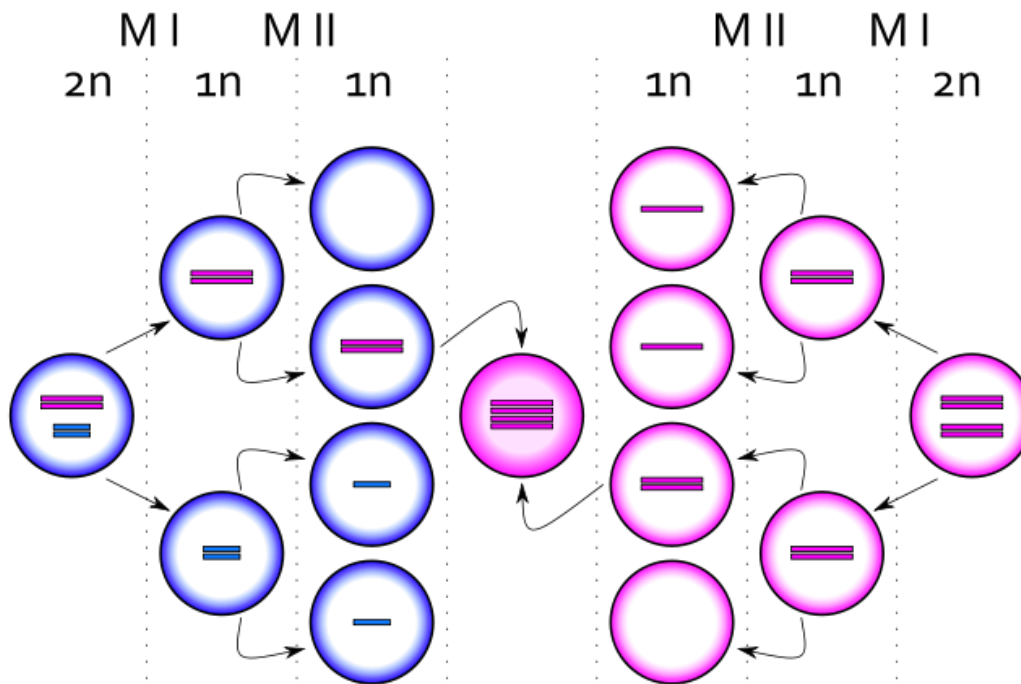


**Down syndrome**

# Chromosomal number mutations

## Types of aneuploidy

### 4. Tetrasomy: two extra chromosomes.



**Tetrasomy X (XXXX)**

# To know

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Aneuploid

Euploid

Chromosomal mutation

Chromosomal deletion

Monosomy

Chromosomal translocation

Chromosome structure mutations

Nullisomy

Trisomy

Chromosomal inversion

Chromosomal duplication

Chromosome number mutation

Tetrasomy

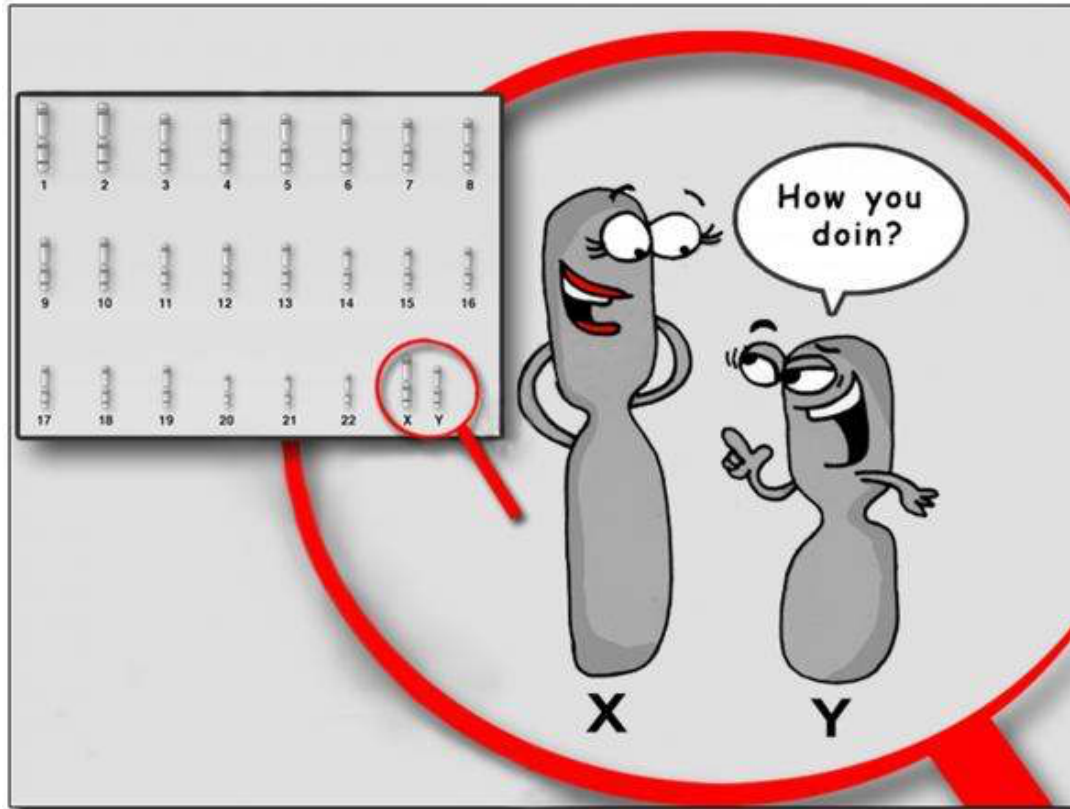
Segmental mutation

# Expectations

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- You know the different types of chromosomal mutations.
- You know chromosomal mutations that affect the DNA content and orientation.
- You know the consequences of chromosomal mutations.
- Can we repair chromosomal mutations?

# For a smile



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