Lecture 32:

Chromosomal mutations

Course 371

Lessons for life

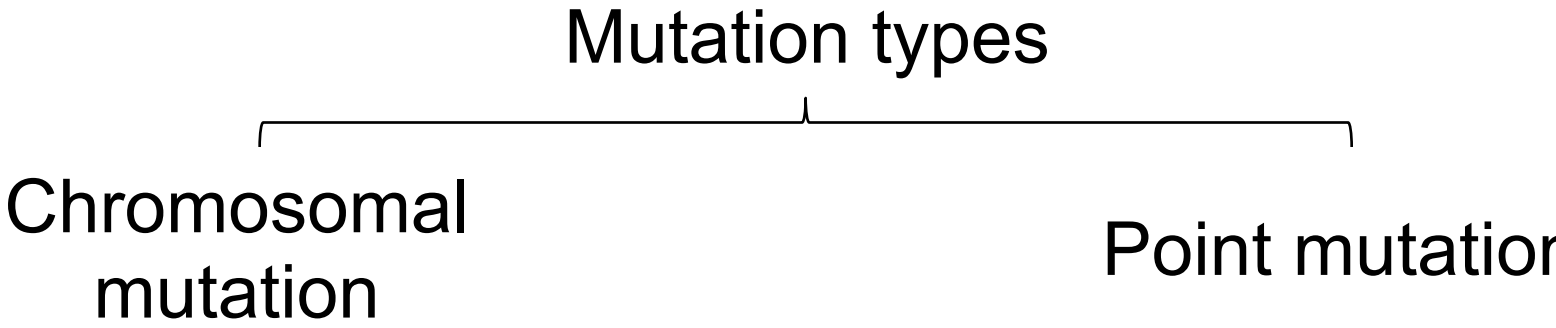
AIMS

- 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



- **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

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Chromosome structure mutations

DNA **content** in a chromosome

- Deletion
- Duplication

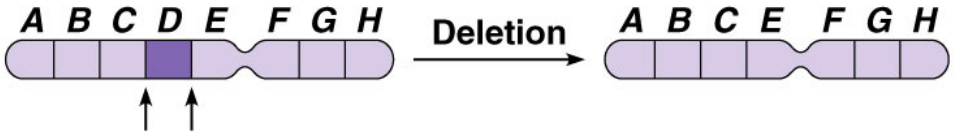
DNA **orientation** in a chromosome

- Inversion
- Translocation

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

Chromosomal deletion

(a) A deletion removes a chromosomal segment.



- 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

(a) A deletion removes a chromosomal segment.

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

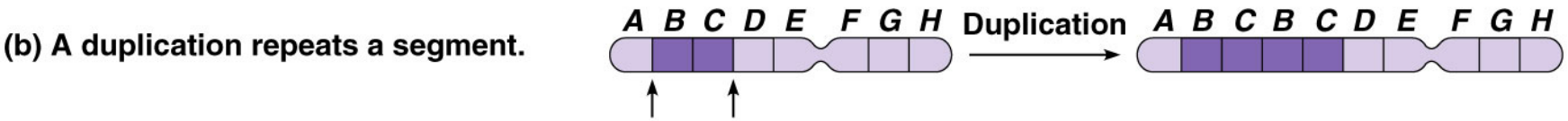
Chromosomal deletion

(a) A deletion removes a chromosomal segment.

Williams syndrome

Wolf-Hirschhorn syndrome

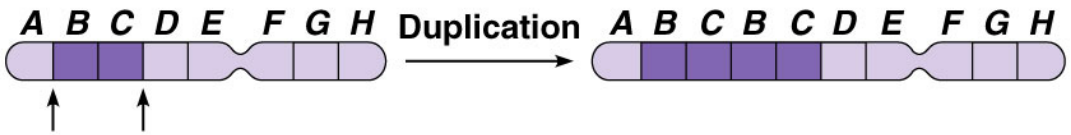
Chromosomal duplication

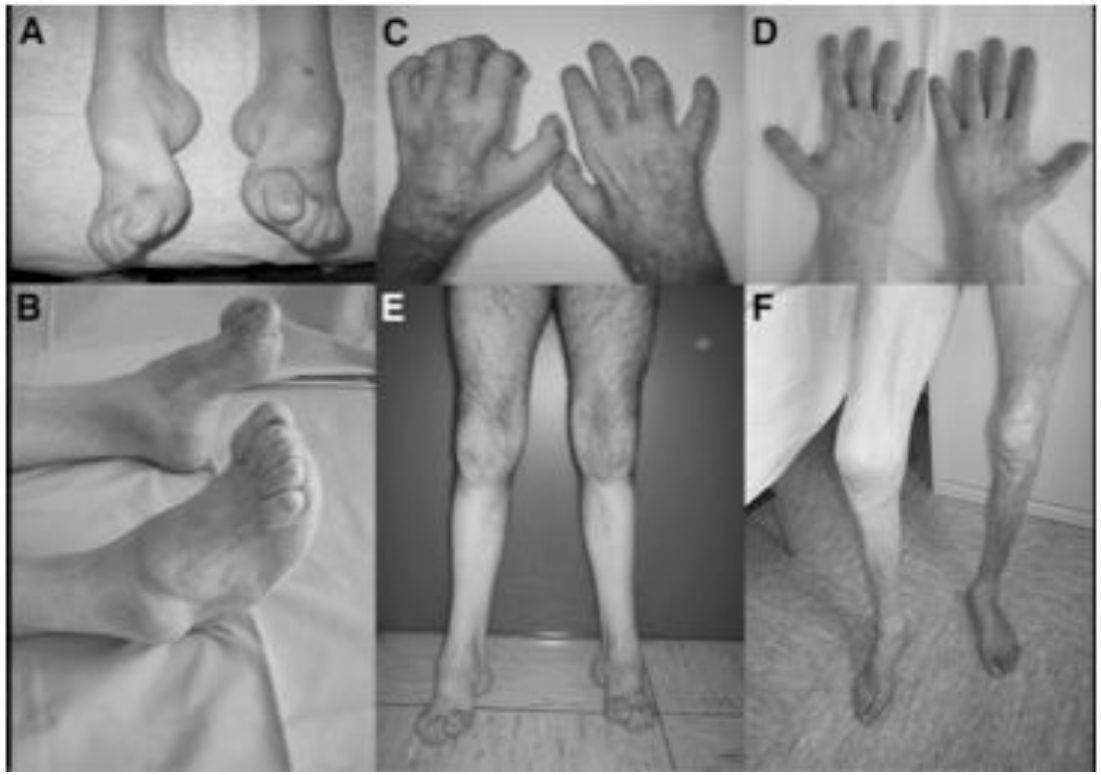


- Duplications increase the amount of DNA of a chromosome compared to the normal chromosome.
- Having extra DNA in a chromosome may alter the number of genes and thus alter the amount of gene product.
- Example:
    - **Charcot–Marie–Tooth disease**

Chromosomal duplication

(b) A duplication repeats a segment.





Charcot–Marie–Tooth disease

Chromosomal structural mutations

Chromosomal mutations

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Chromosome structure mutations

DNA **content** in a chromosome

- Deletion
- Duplication

DNA **orientation** in a chromosome

- Inversion
- Translocation

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

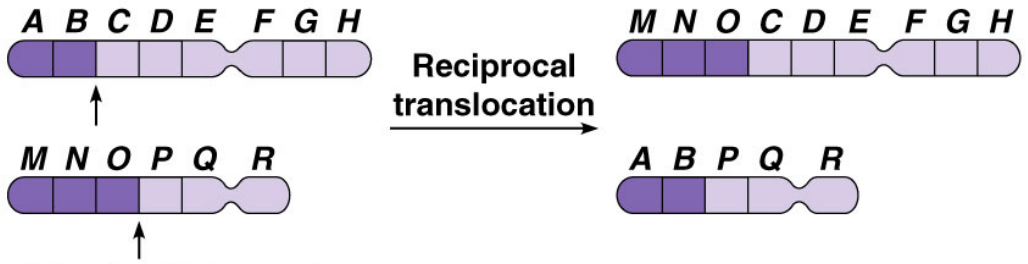
Chromosomal inversion

(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

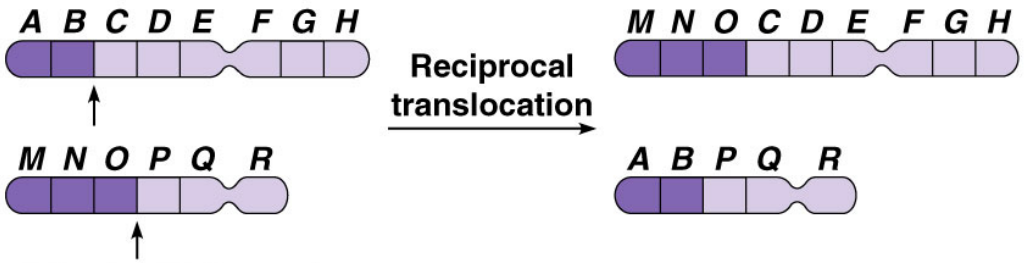
(d) A translocation moves a segment from one chromosome to another, nonhomologous one.



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

Chromosomal translocation

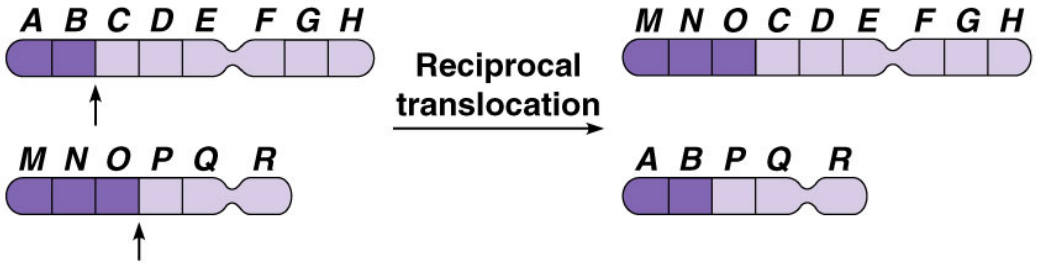
(d) A translocation moves a segment from one chromosome to another, nonhomologous one.



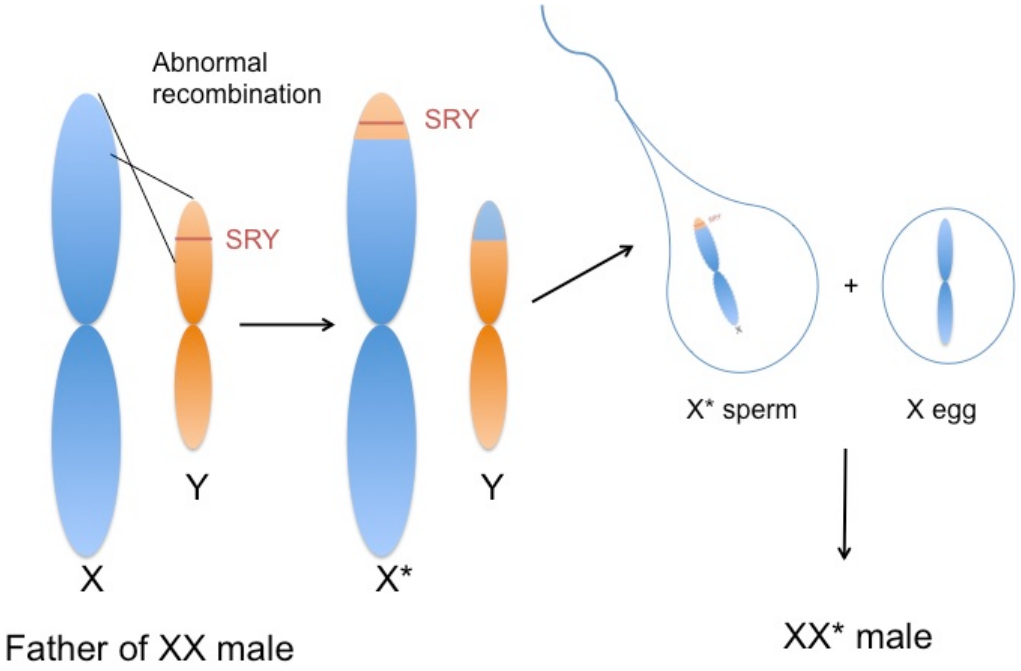
- Example:
  - **XX male syndrome!**
  - **Translocation of SRY gene from Y chromosome to X chromosome.**

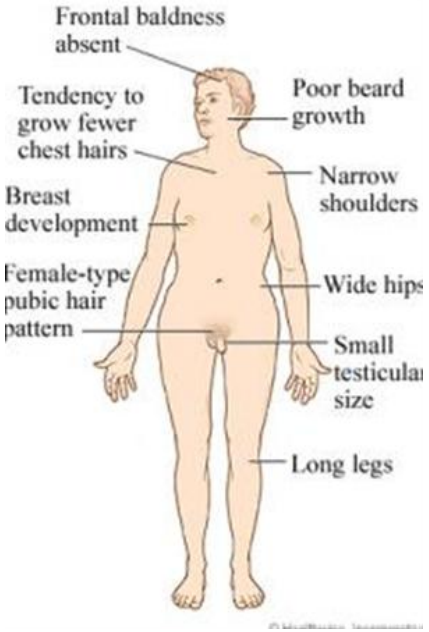
Chromosomal translocation

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

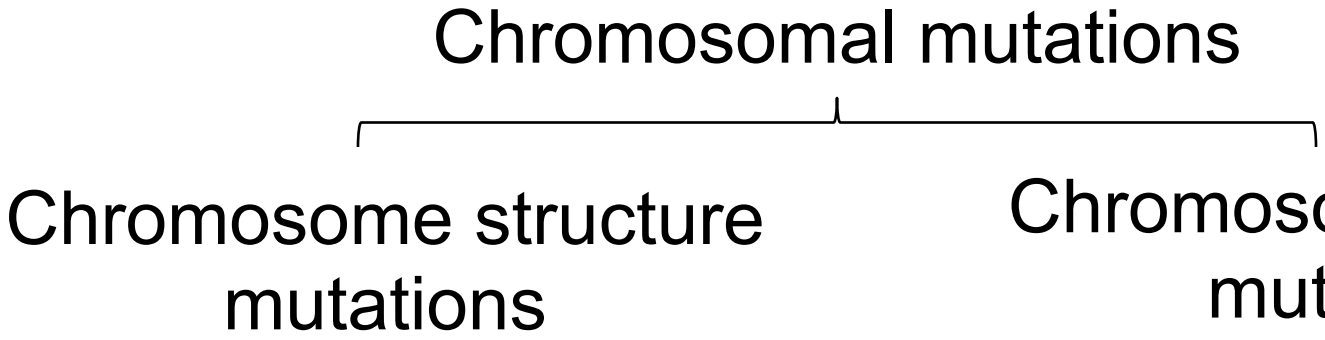


XX male syndrome!





Chromosomal mutations



- **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.

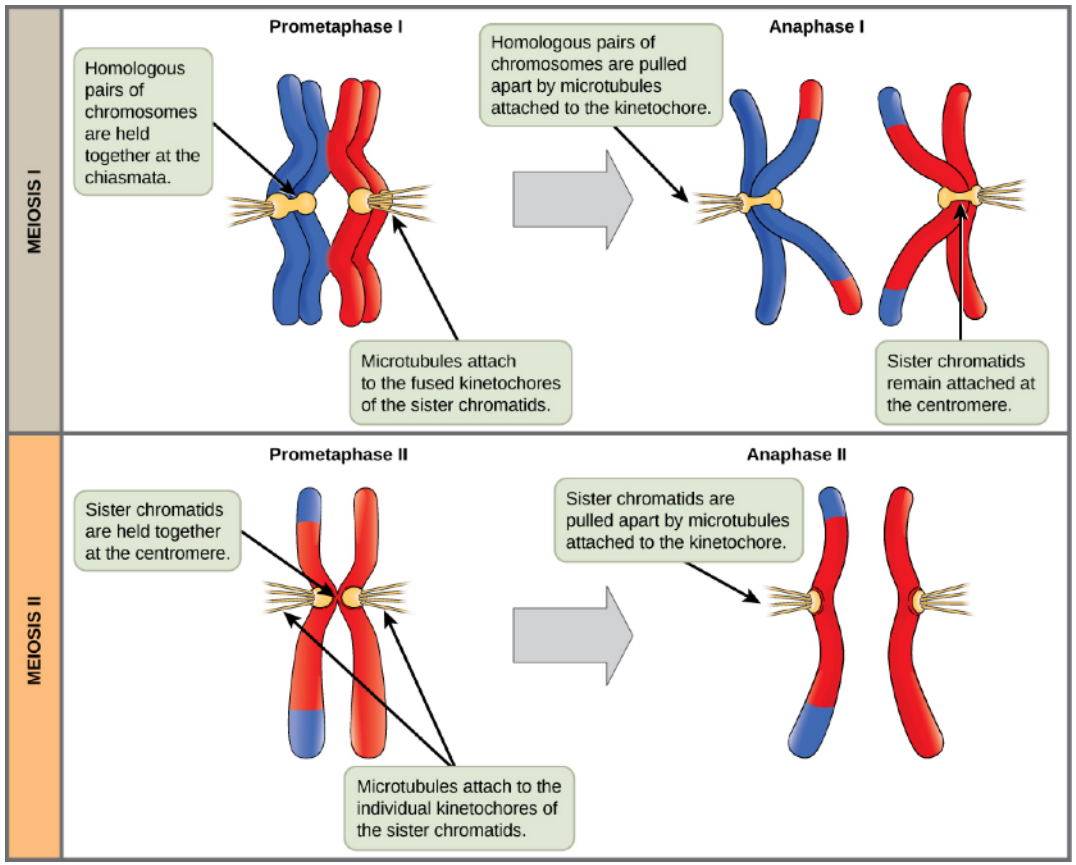
Chromosomal number mutations

- 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.



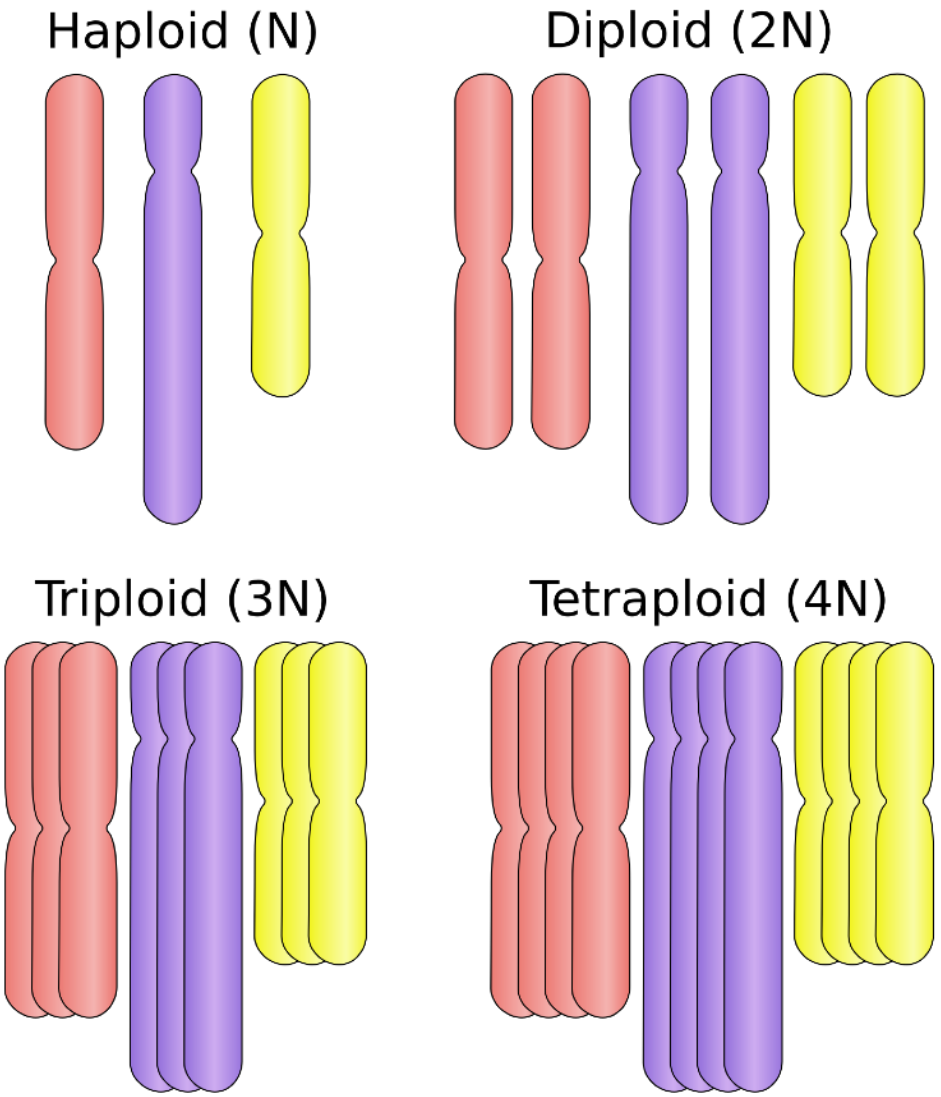
Chromosomal number mutations

- **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

- **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.

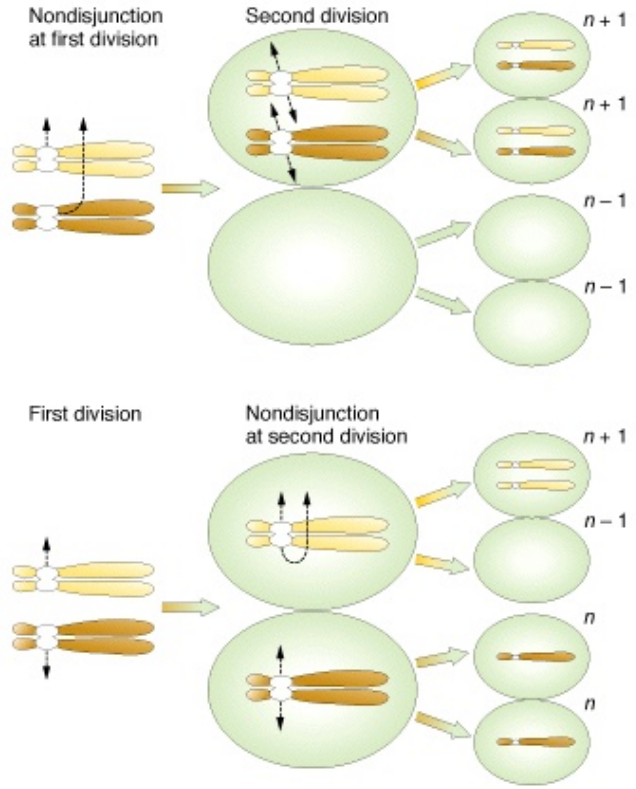
Chromosomal number mutations



Chromosomal number mutations

Types of aneuploidy

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

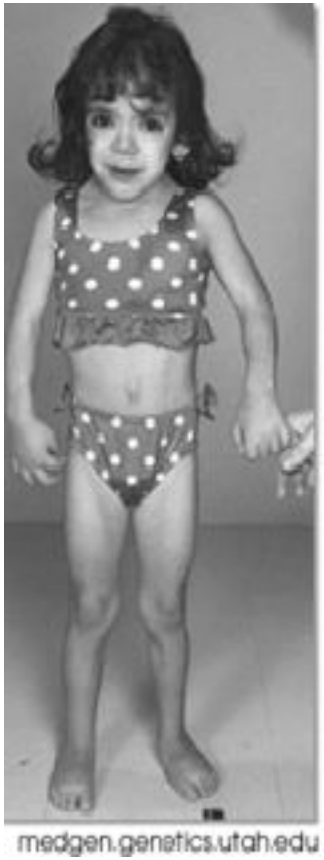


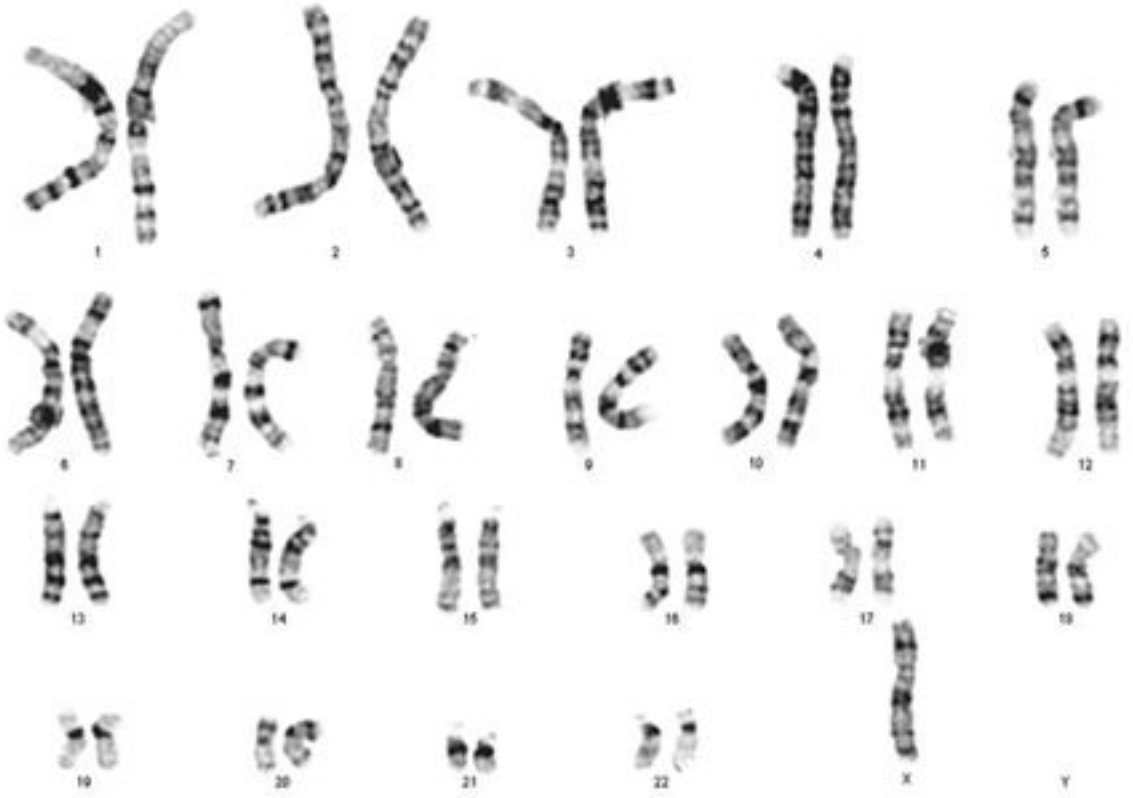
What do you think will happen?

Chromosomal number mutations

Types of aneuploidy

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



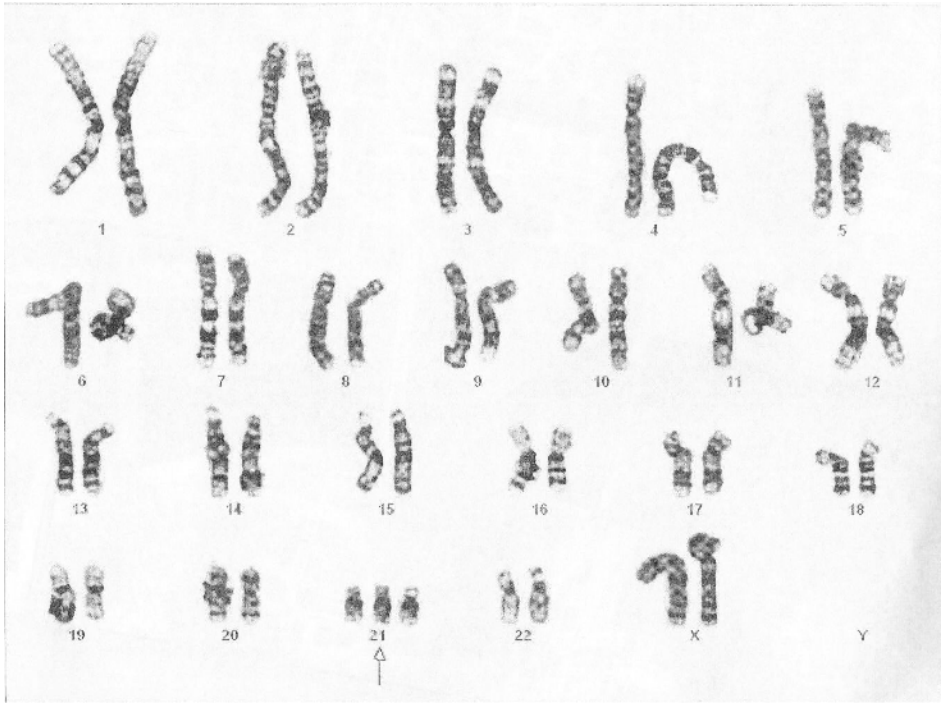


Turner syndrome

Chromosomal number mutations

Types of aneuploidy

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

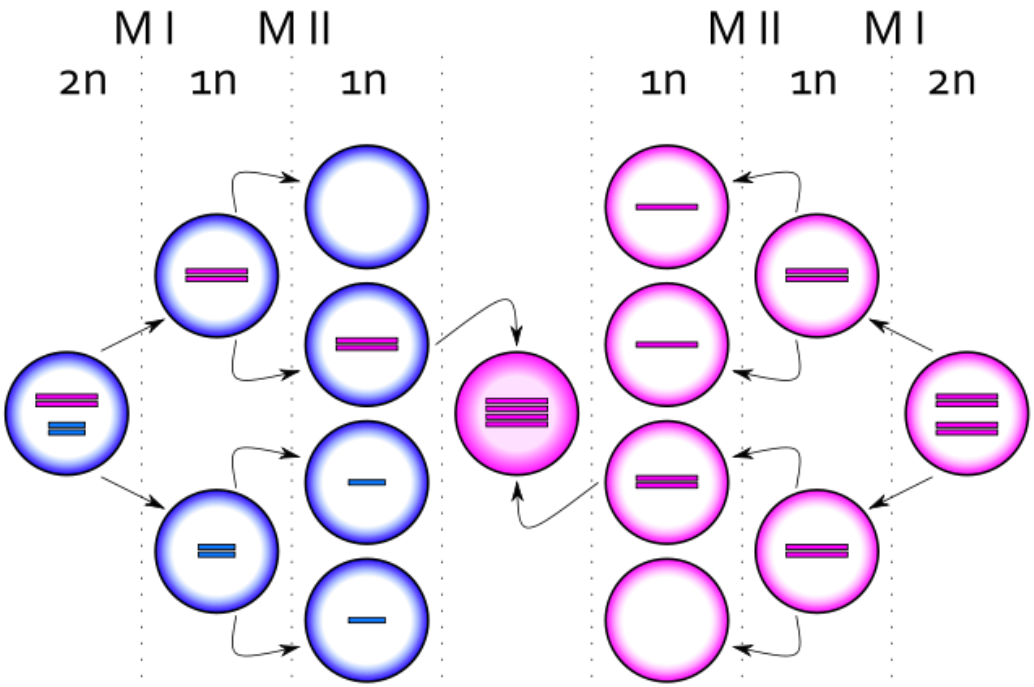


Down syndrome

Chromosomal number mutations

Types of aneuploidy

4.**Tetrasomy:** two extra chromosomes.



Tertrasomy X (XXXX)

To know

Aneuploid

Euploid

Chromosomal mutation

Chromosomal deletion

Monosomy

Chromosomal translocation

Chromosome structure mutations

Nullisomy

Trisomy

Chromosomal duplication

Chromosomal inversion

Chromosome number mutation

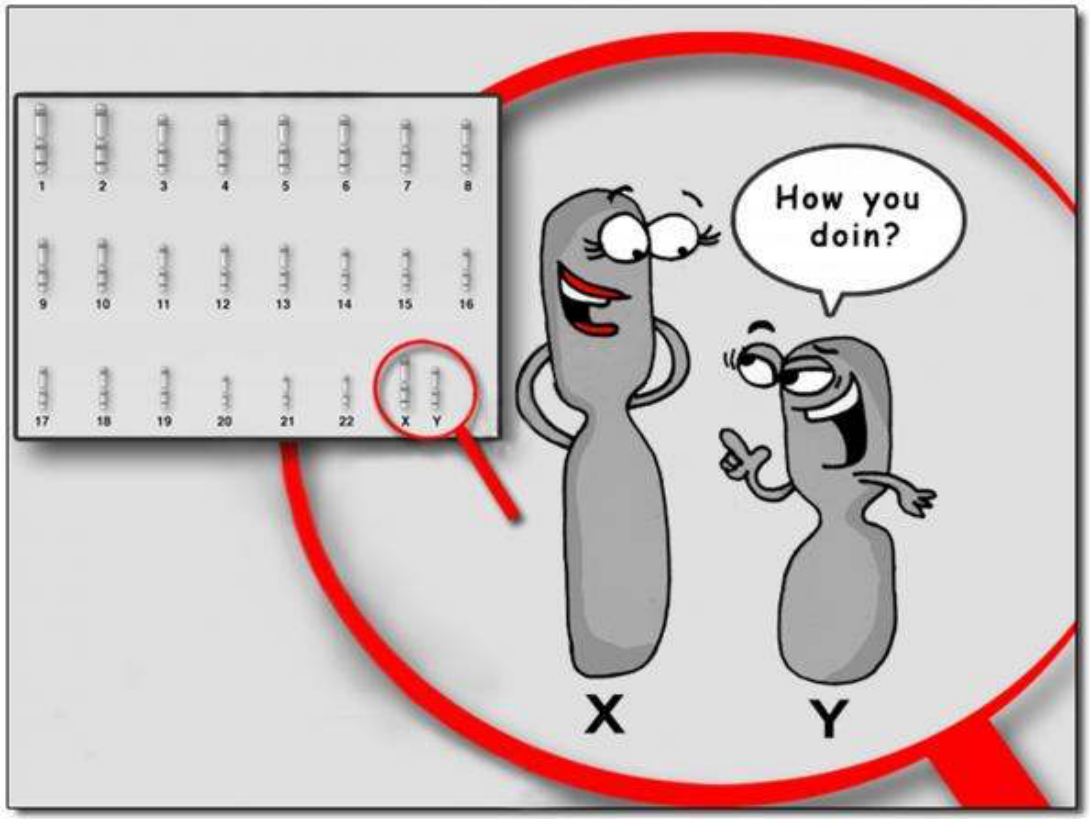
Segmental mutation

Tetrasomy

Expectations

- 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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