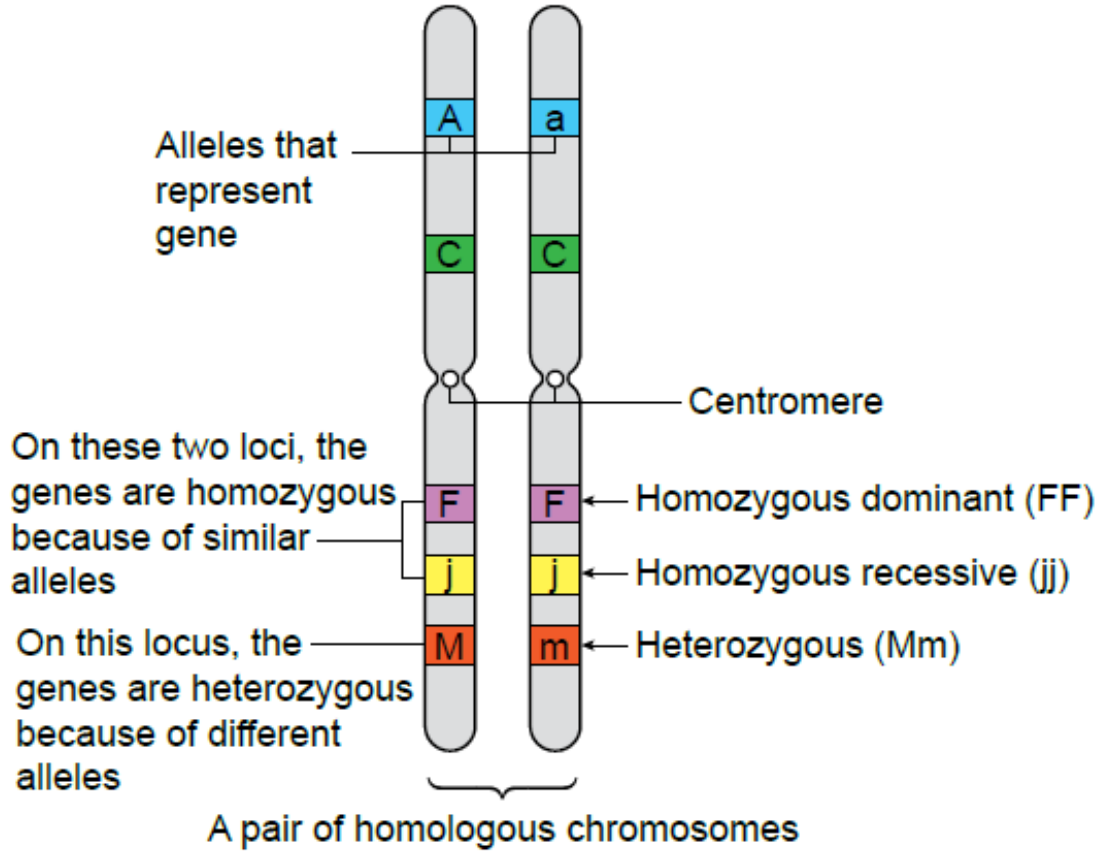
11.3 Genes and Alleles

Definition of Locus

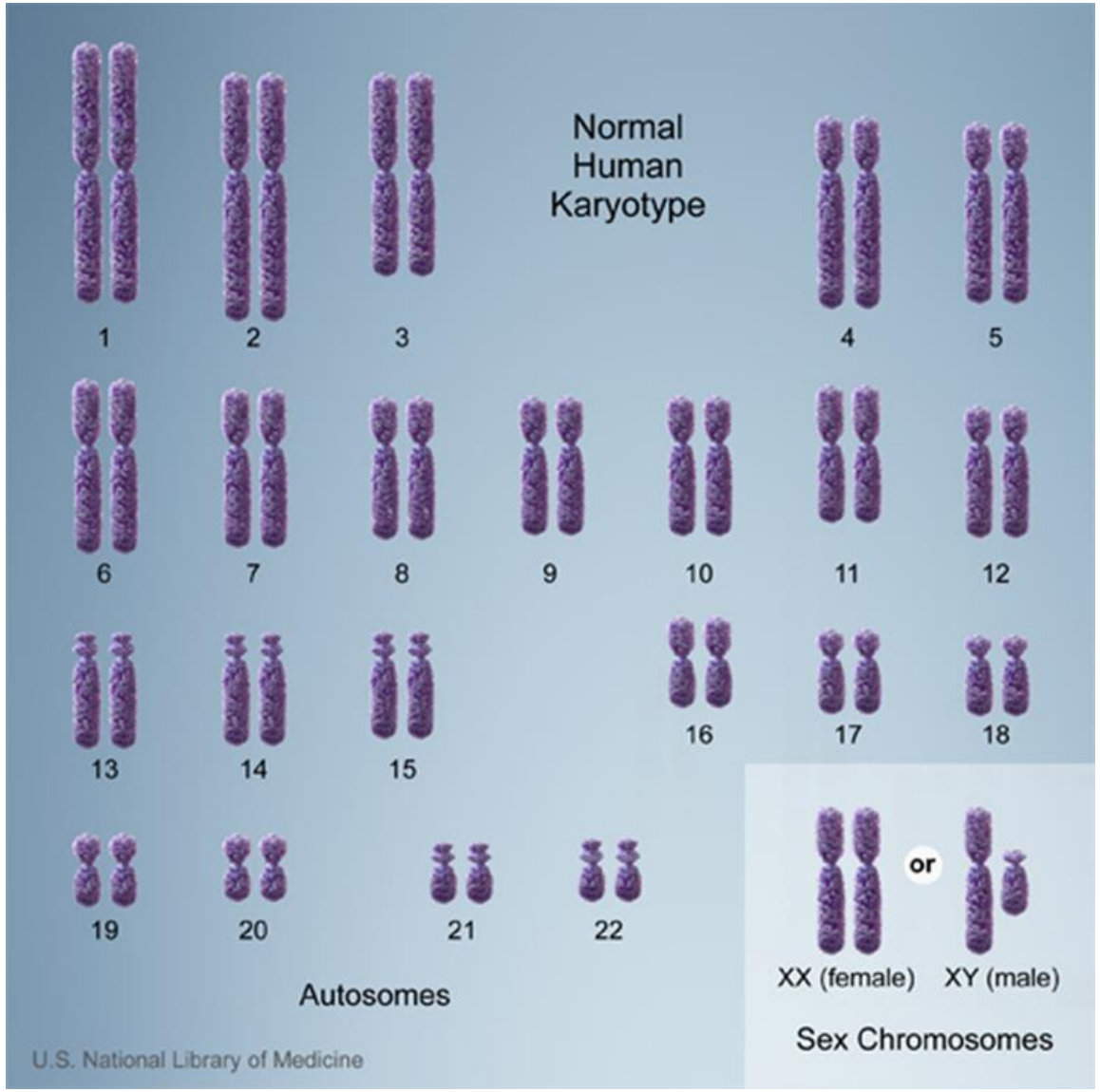
- Locus is a specific location of a gene in a chromosome
- Allele which represents a gene is located at the same locus as the gene.



11.4 Inheritance in Humans

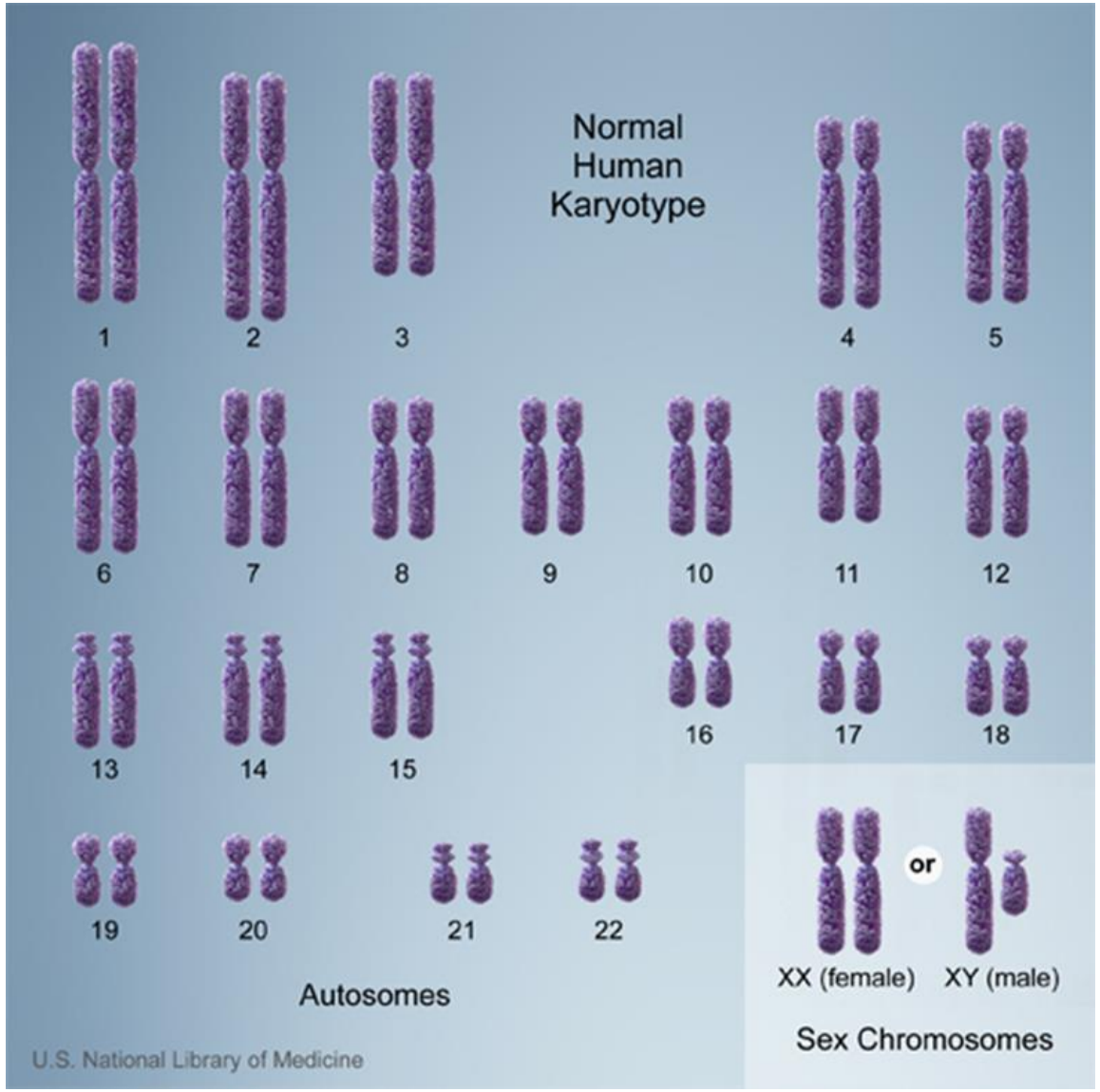
Types of human chromosomes

	Autosome	Sex chromosome
Feature	Consists of chromosome pairs from number 1 to 22	Consists of one chromosome pair, that is number 23
Function	Controls all characteristics of somatic cells	Consists of genes which determine gender
Example	Types of blood groups, height and skin colour	Male has XY chromosomes whereas female has XX chromosomes



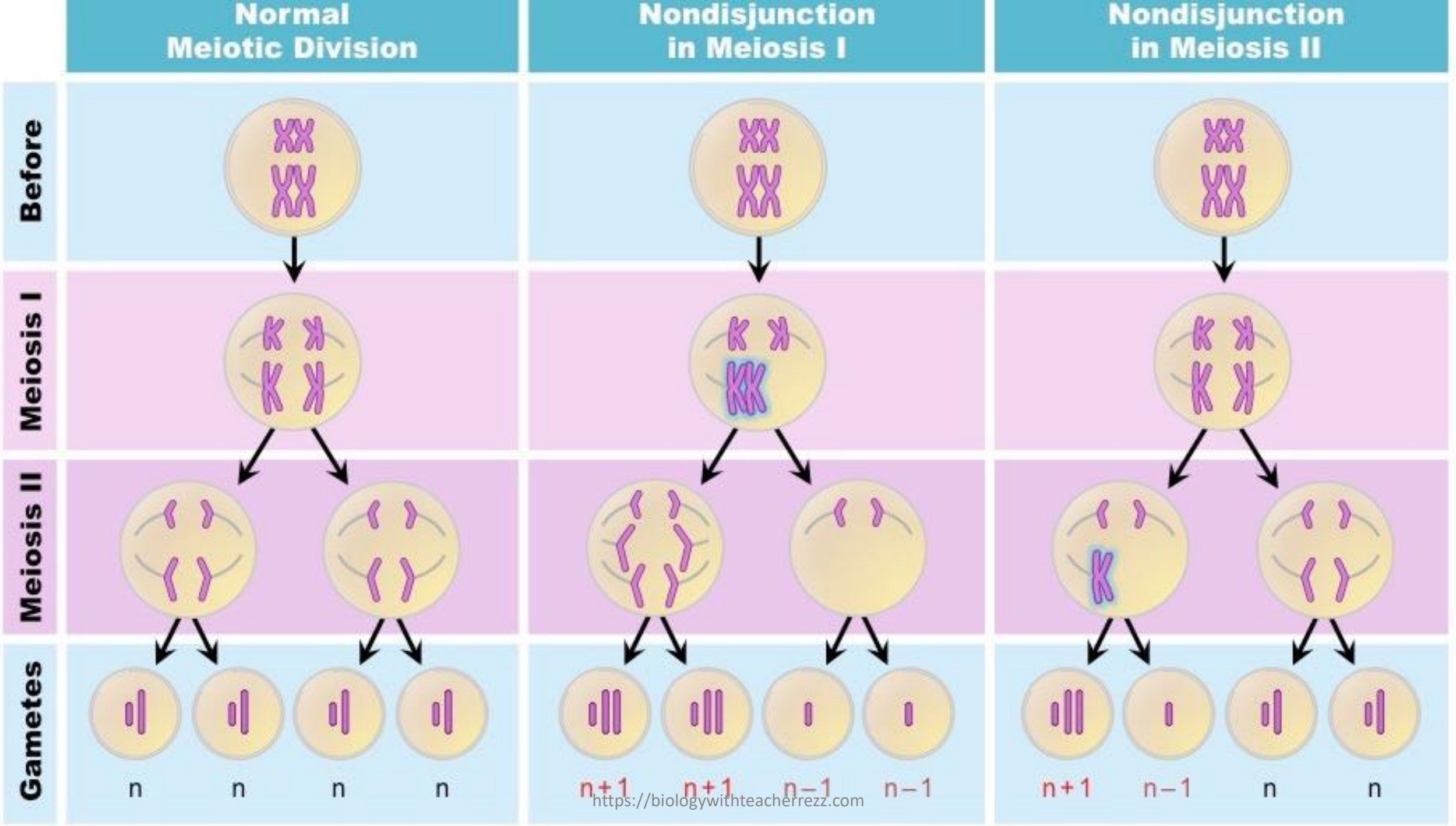
Types of human chromosomes

- The number and structure of chromosomes present in a cell nucleus is known as karyotype
- Chromosomes are arranged in pairs, based on homologous chromosomes in terms of their:
  - sizes,
  - centromere locations
  - banding pattern of chromosomes.



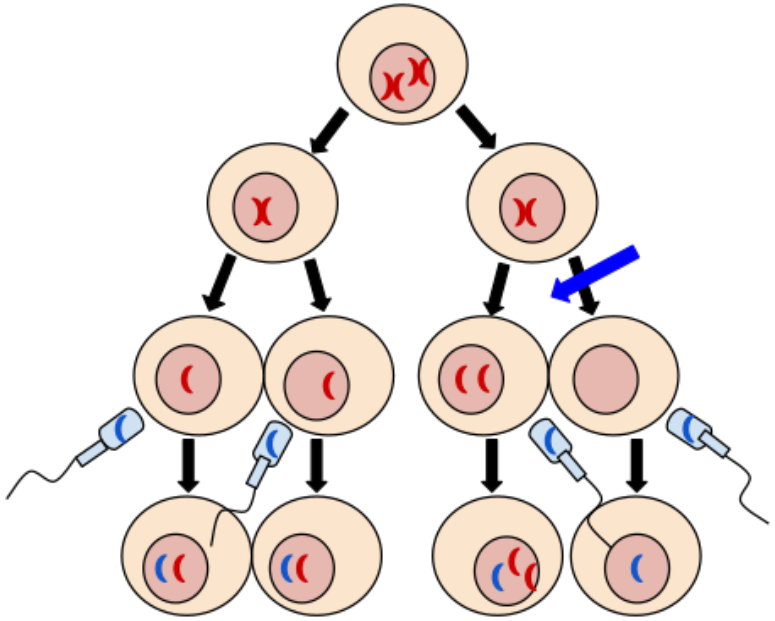
Nondisjunction

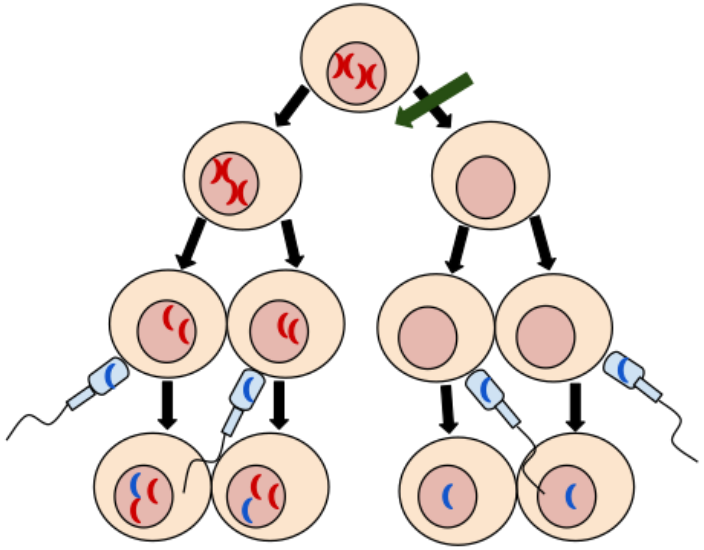
- Changes in number of chromosome can occur due to failure of homologous chromosomes to separate during anaphase I or failure of sister chromatids to separate during anaphase II.
- When nondisjunction occurs in humans, either male gamete (sperm) or female gamete (ovum) can possess chromosome number of less than 23, that is 22 or more than 23, which is 24.



Nondisjunction

- Therefore, fertilisation that involves the abnormal gamete with a normal gamete produces a zygote with 45 chromosomes or 47 chromosomes.
- Examples of genetic diseases caused by nondisjunction are:
  - Down syndrome
  - Turner syndrome
  - Klinefelter syndrome

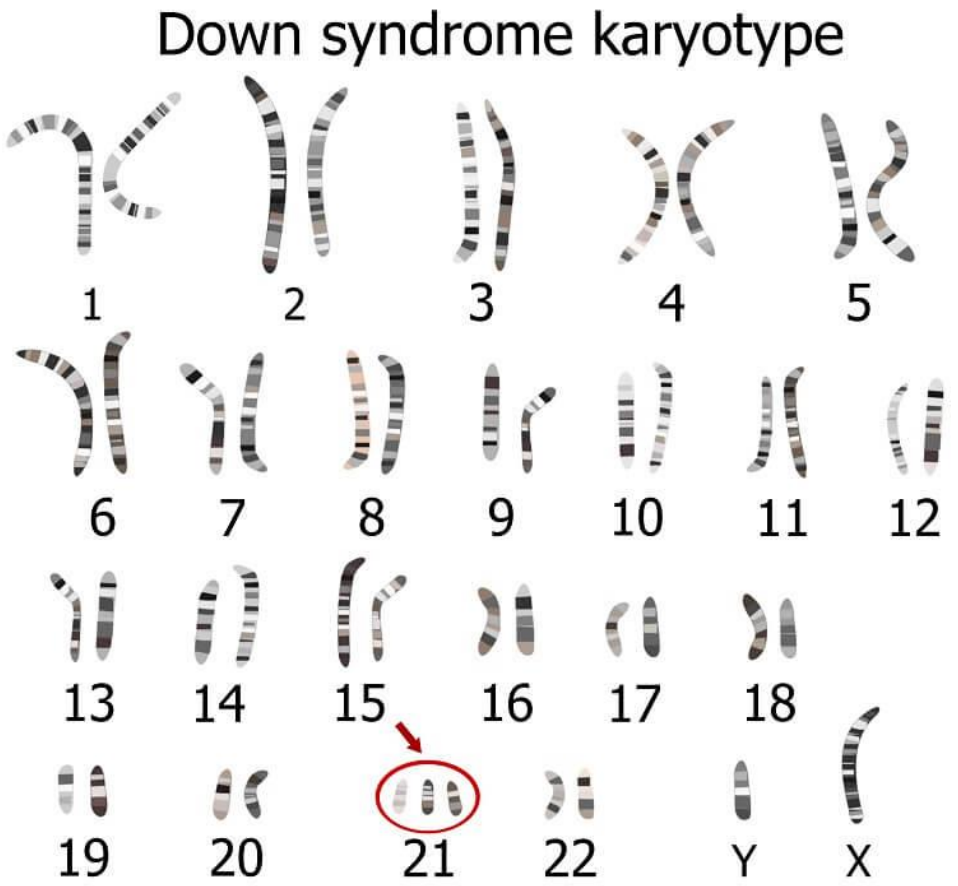




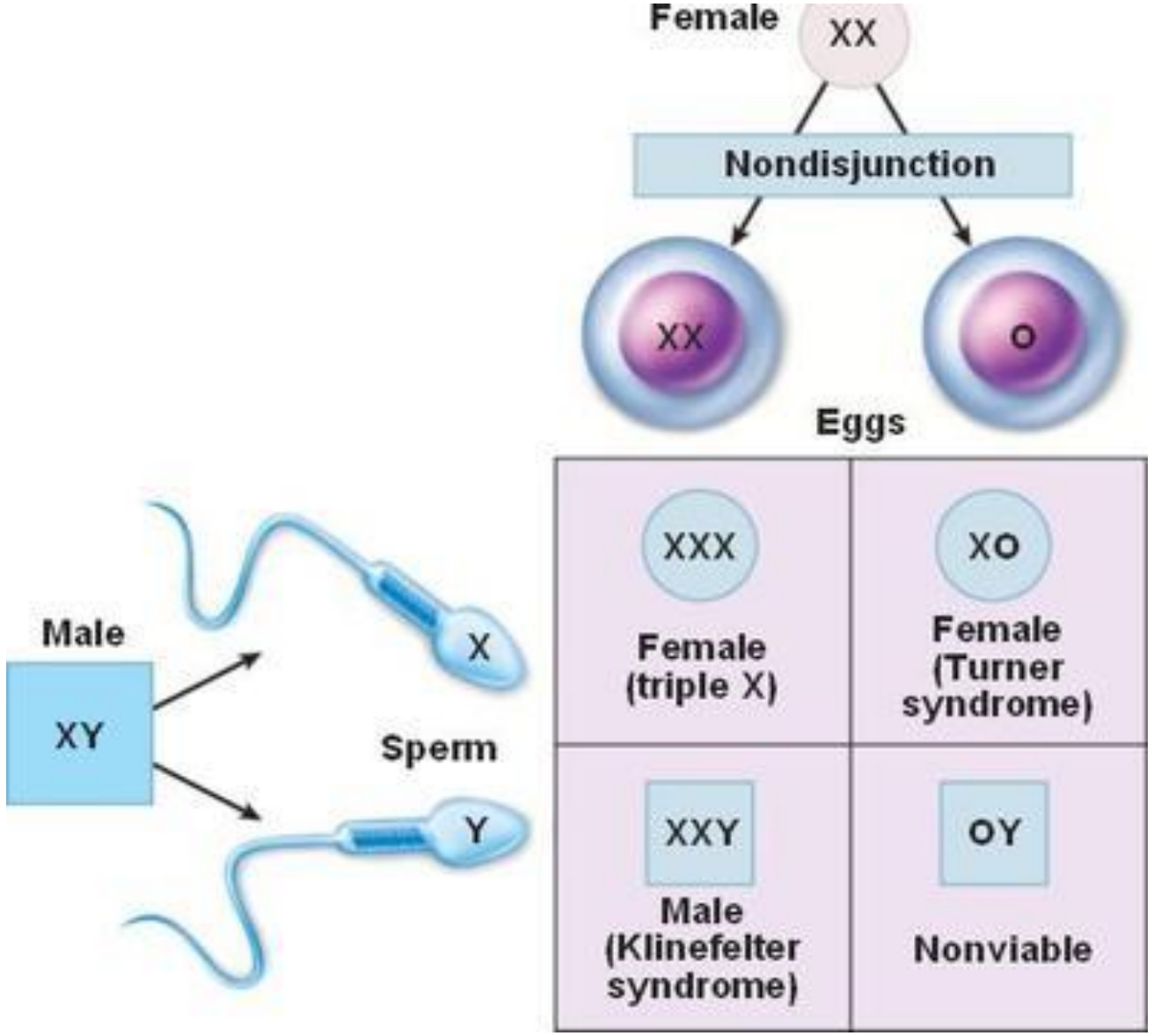
Down syndrome

- Total chromosome number is 47, which is 45 + XY.
- There is an extra chromosome for chromosome pair number 21.
- Down syndrome is also known as trisomy 21.
- Down syndrome can occur in both males and females.

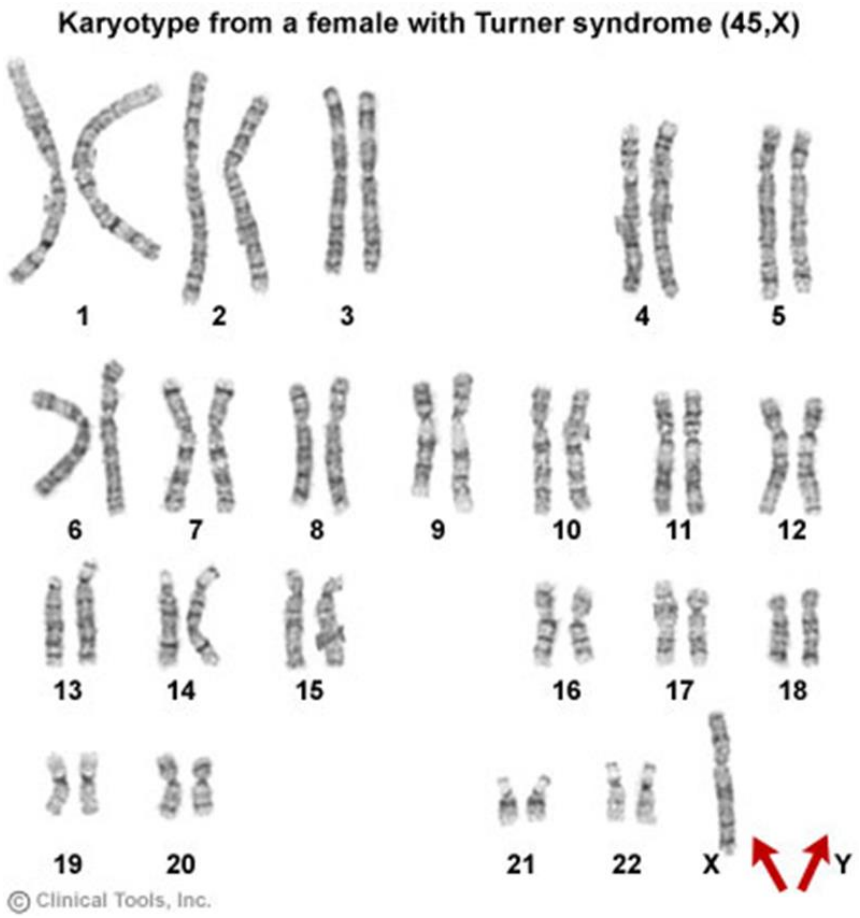
Down Syndrome



Nondisjunction of sex chromosomes

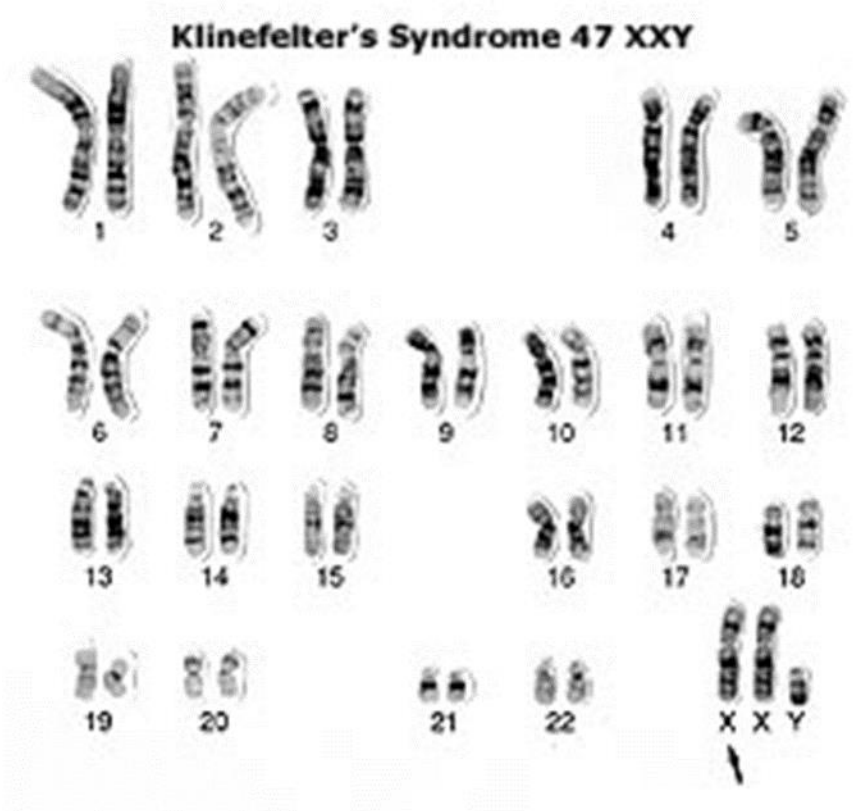


Turner syndrome



- Total number of chromosomes is 45, which is 44 + XO.
- There is a missing X chromosome in the pair of sex chromosomes.
- The gender of individual with Turner syndrome is a female

Klinefelter syndrome



- has a total of 47 chromosomes, that is 44 + XXY.
- There is an extra X chromosome in the pair of sex chromosomes.
- The gender of individual with Klinefelter syndrome is male.
- However, his secondary sex characteristics are not well-developed