BO101

Topic: Basic Genetics (23-03-21)

Genes & Alleles

Genes: **hereditary information that determines a single trait**

Alleles: alternate forms of a gene (variations)

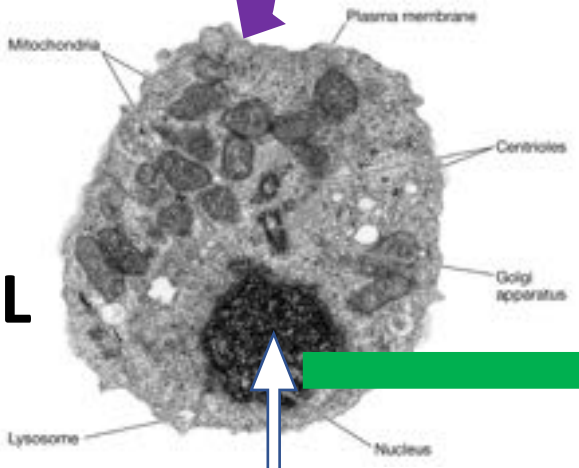
- Gene: Flower color  Allele: 2 (variations)
- Gene: Fruit color  Allele: 3 (variations)
- Gene: Rabbit fur color  Allele: 4 (variations)

Where are genes located?

BODY

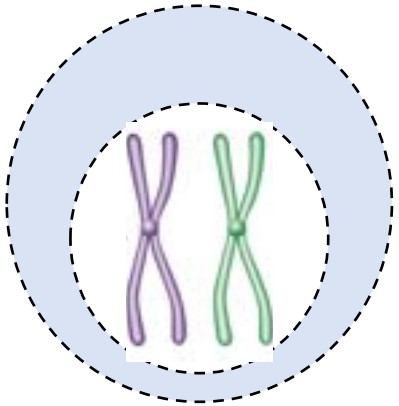
ORGAN

TISSUE



Somatic CELL

NUCLEUS



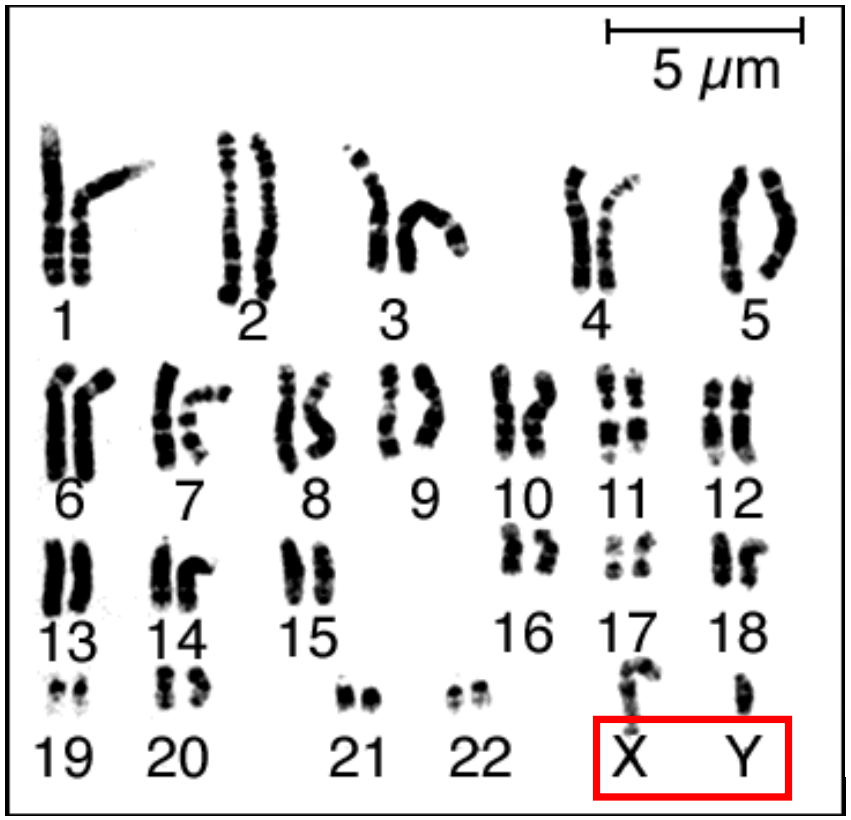
Compact form of DNA
is known as **Chromosome**

DNA

Human Genome

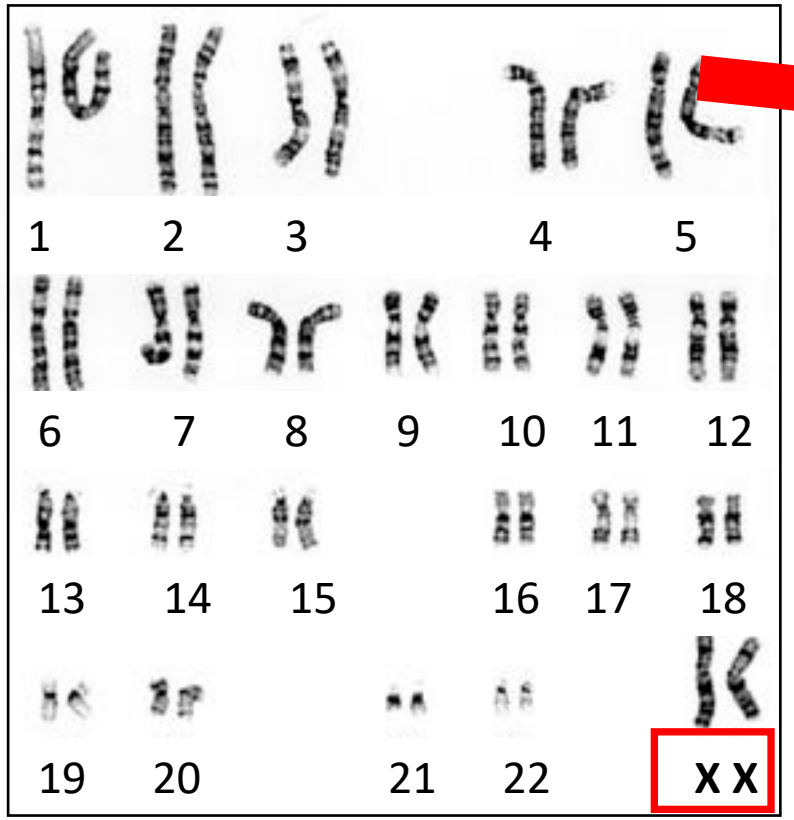
- **Genes are present in the DNA molecule (large macromolecule)**
- **Compact DNA = chromosome**
- **Each somatic cell has 23 pairs (= 46) chromosomes**
- **Each germ cell (gamete) has 23 chromosomes**
- **The genome** is sum of all the DNA (chromosome)

Human genome = sum of genetic information present in 46 (23 pairs) chromosomes



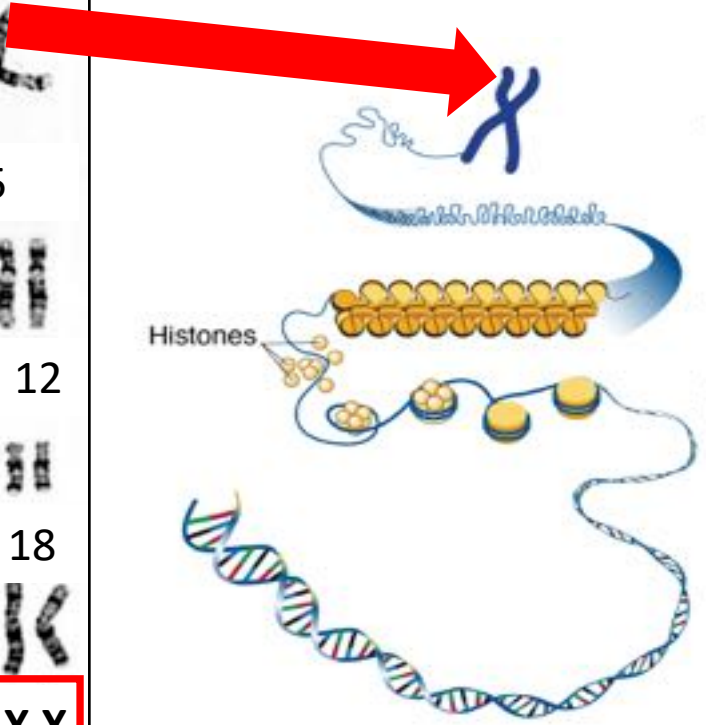
23 pairs; 22+XY in **male**

Somatic cells

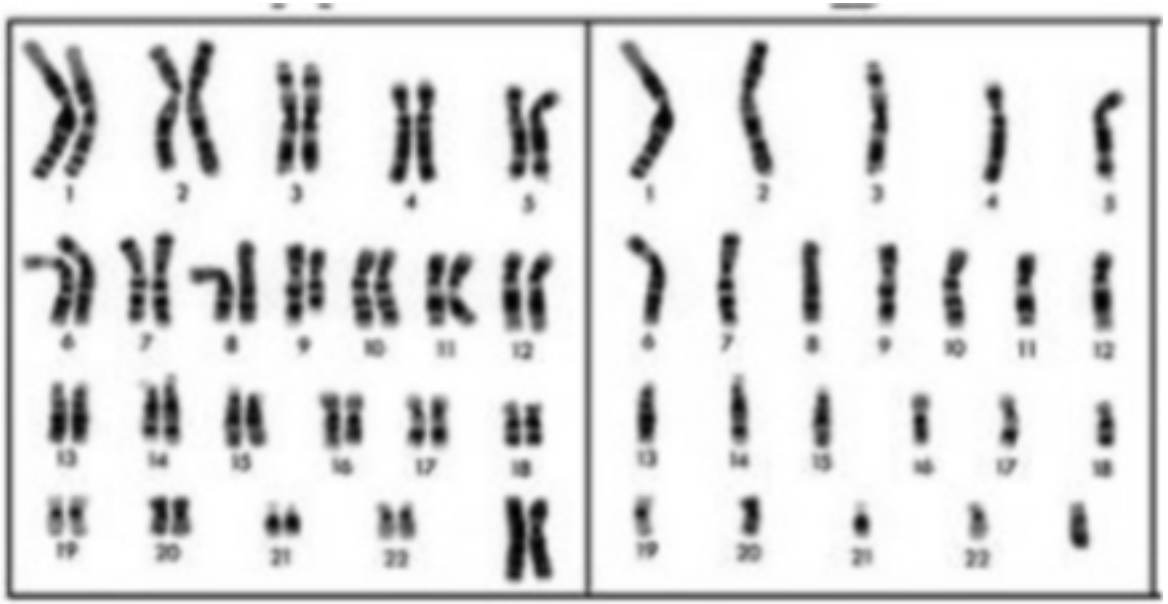


23 pairs; 22+ XX in **female**

Somatic cells



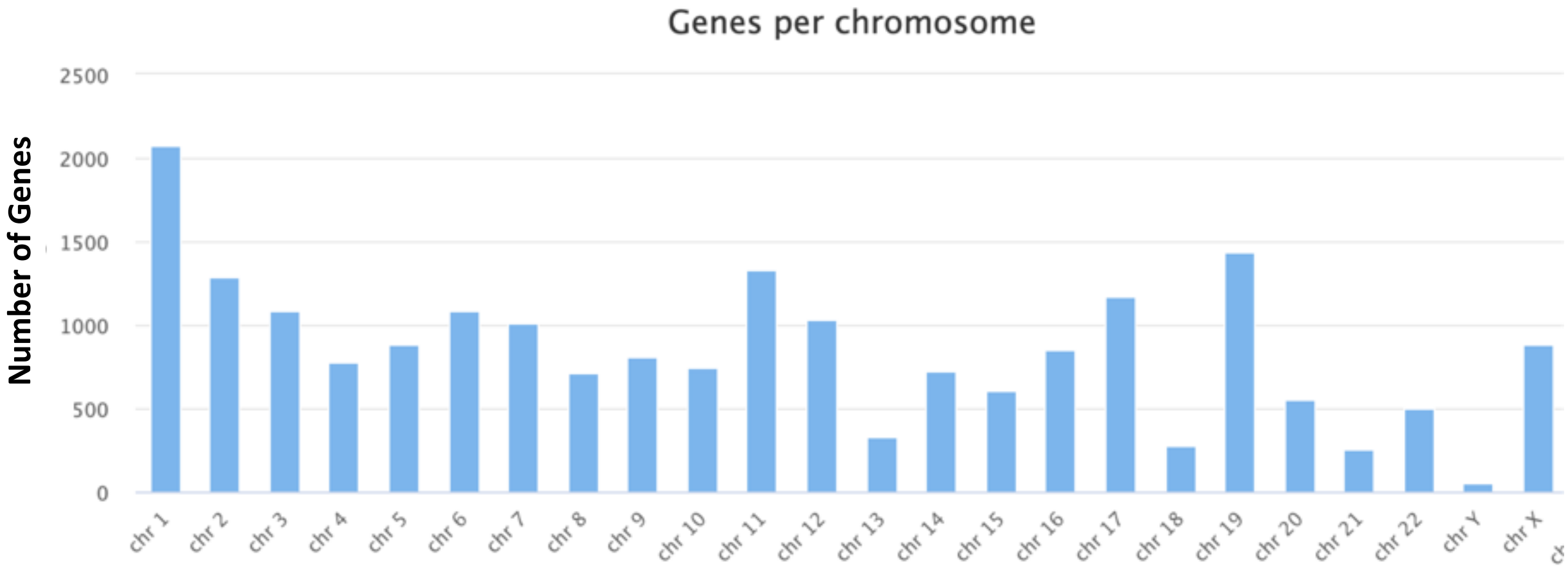
Which one of these represent the sperm chromosome?



A

B

Human genome **encodes 30,000 genes distributed in 23 chromosomes**



Each gene has two alleles

Dominant Allele: an allele that is expressed whenever it is present

Represented by a capital letter

e.g. A

Recessive Allele: an allele that is suppressed whenever the dominant allele is present.

Represented by a lower case letter

e.g. a

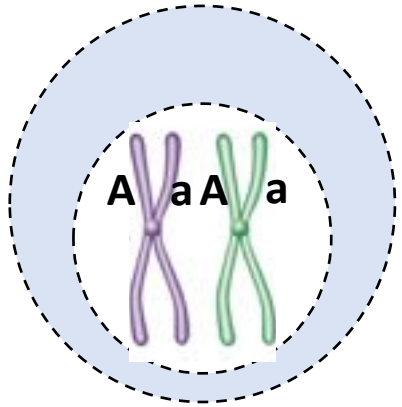
Homozygous and Heterozygous

- **<u>Homozygous:</u>** organism that has 2 *identical* alleles for a trait

e.g. AA  or aa 

- **<u>Heterozygous:</u>** organism that has 2 *different* alleles for the same trait

e.g. Aa 



Genotype and Phenotype

- **Genotype:** Genetic makeup of an individual determined by the alleles

- **Phenotype:** Physical appearance of a trait due to expression of the allele

e.g. AA or Aa has same phenotype but different genotype

AA or Aa

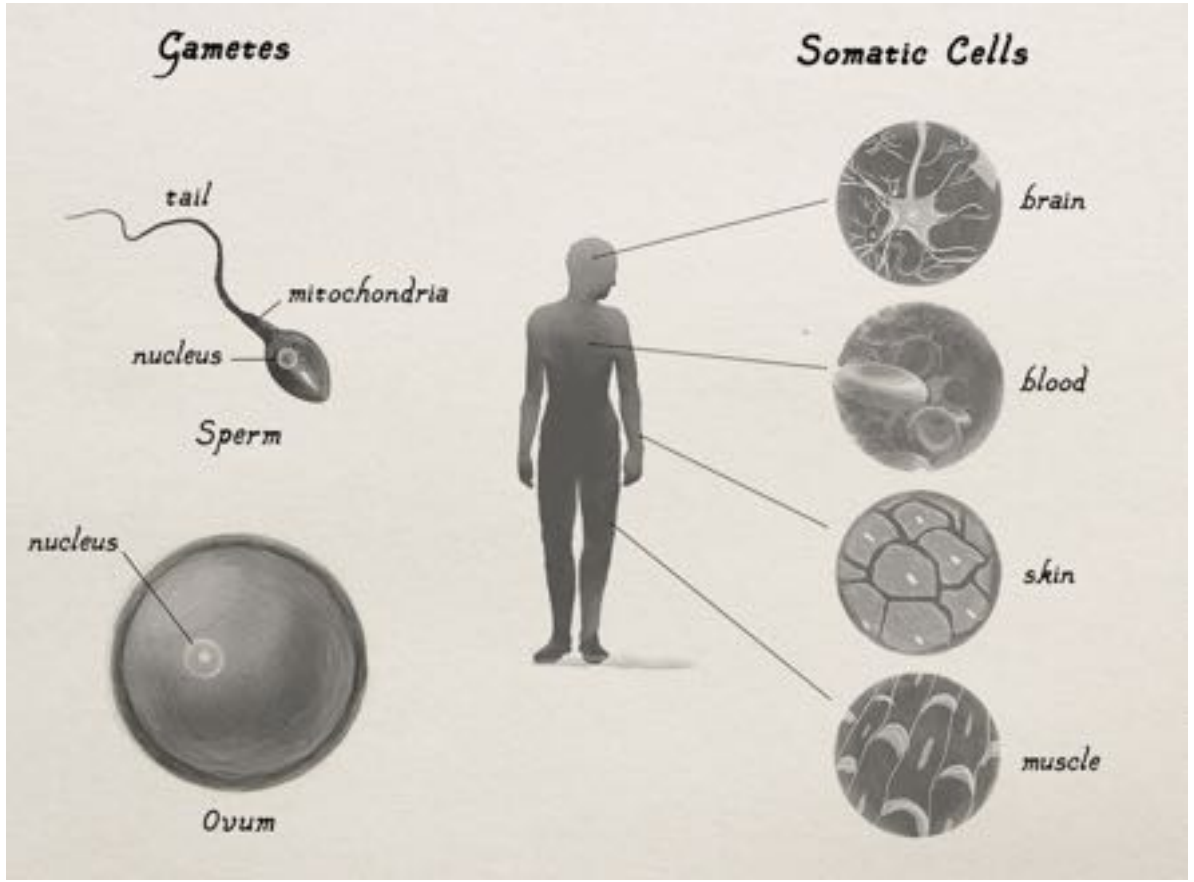
Somatic cells & germ cells

Somatic cells:

- every **cell** in the body
- **46 chromosomes**
- **2 alleles of each gene**

Gametes:

- **reproductive cells/germ cells**
- **23 chromosomes**
- male gamete: sperm
- female gamete: ovum
- **Any one allele of each gene**



Somatic Cells are diploid (46 Chromosome) Gametes are haploid (23 Chromosome)

Mendel: discovered how traits are inherited

- **Gregor Mendel** was a monk and studied genetics of pea plants

- He discovered **how the traits are inherited** by studying the pea plants for eight years

- He published his work in 1865 but no one seemed to grasp its importance.

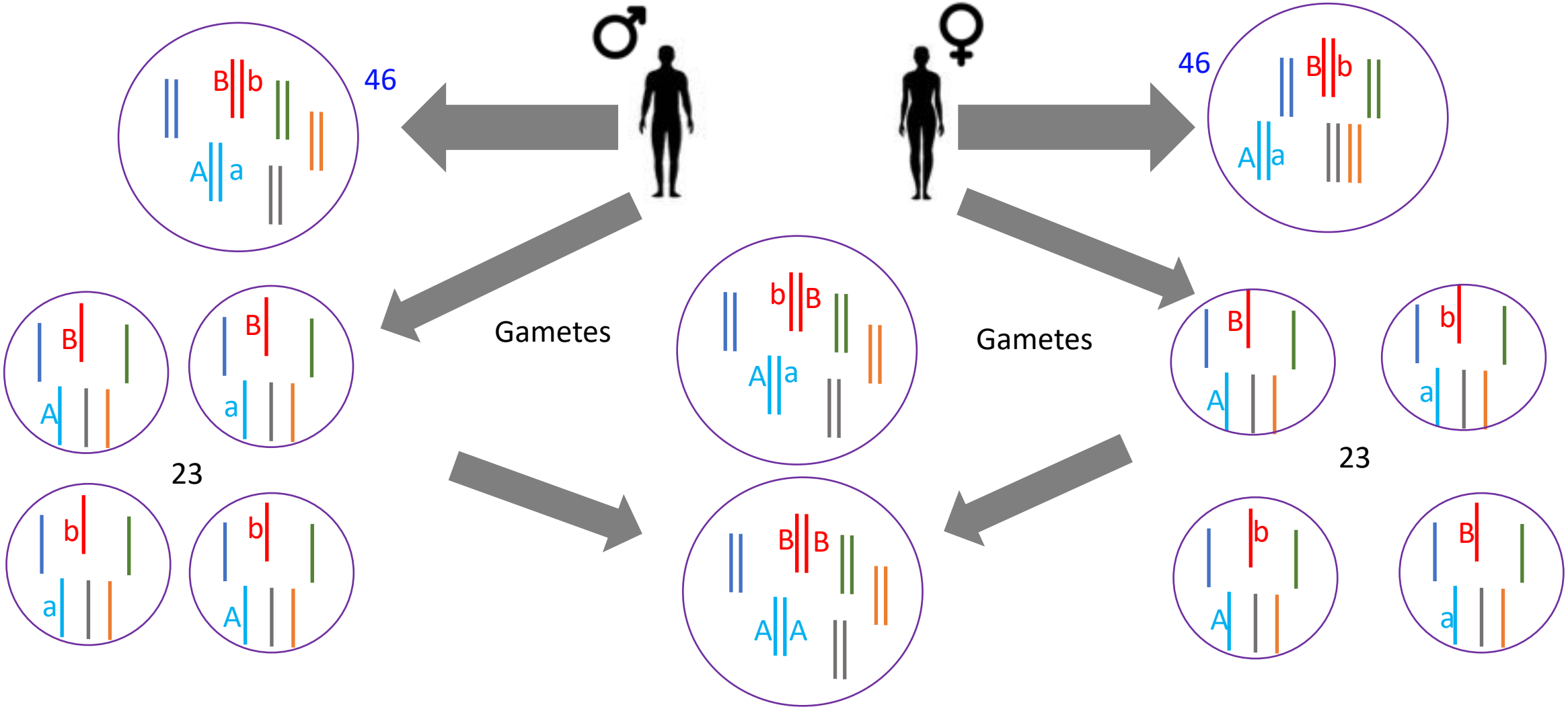
- **Mendel's brilliance was unrecognized.** The work was simply too ahead of its time.

Mendel's laws

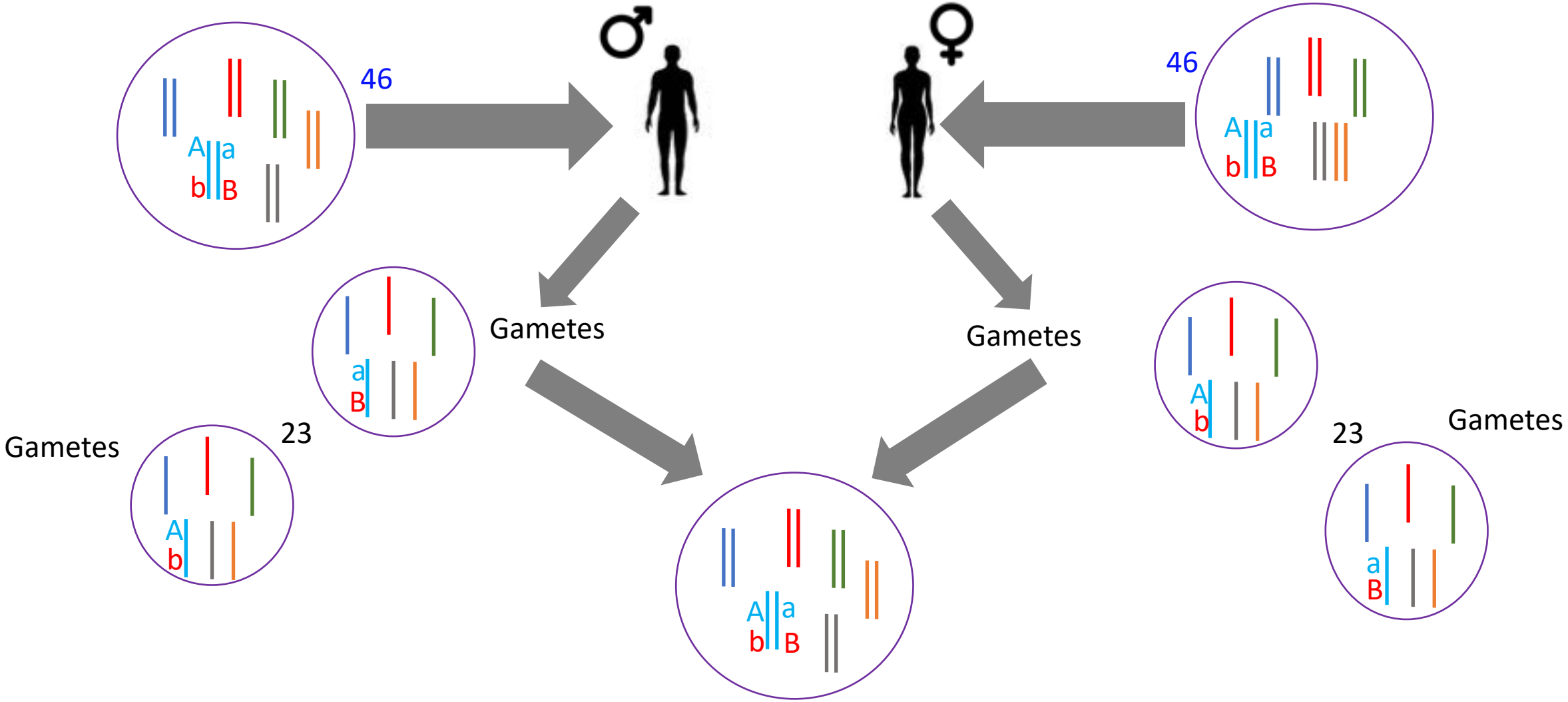
- **MENDEL'S 1ST LAW:**
- Each somatic cells carries **2** alleles of each "element" (now we call **gene**) and alleles segregate from each other when gametes form, hence each gamete carry one allele

- **MENDEL'S 2nd LAW:**
- Alleles for different traits are inherited independently of each other

Mendel's laws

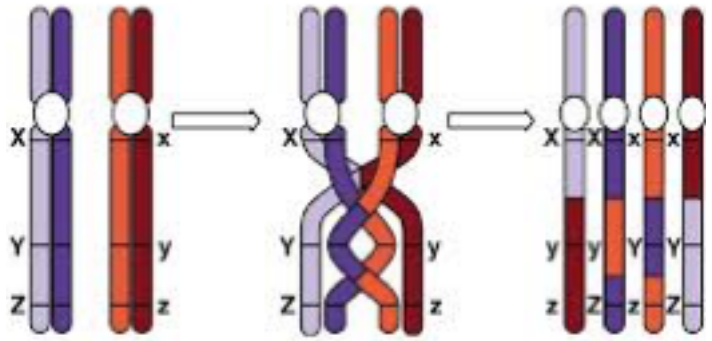


Mendel's laws



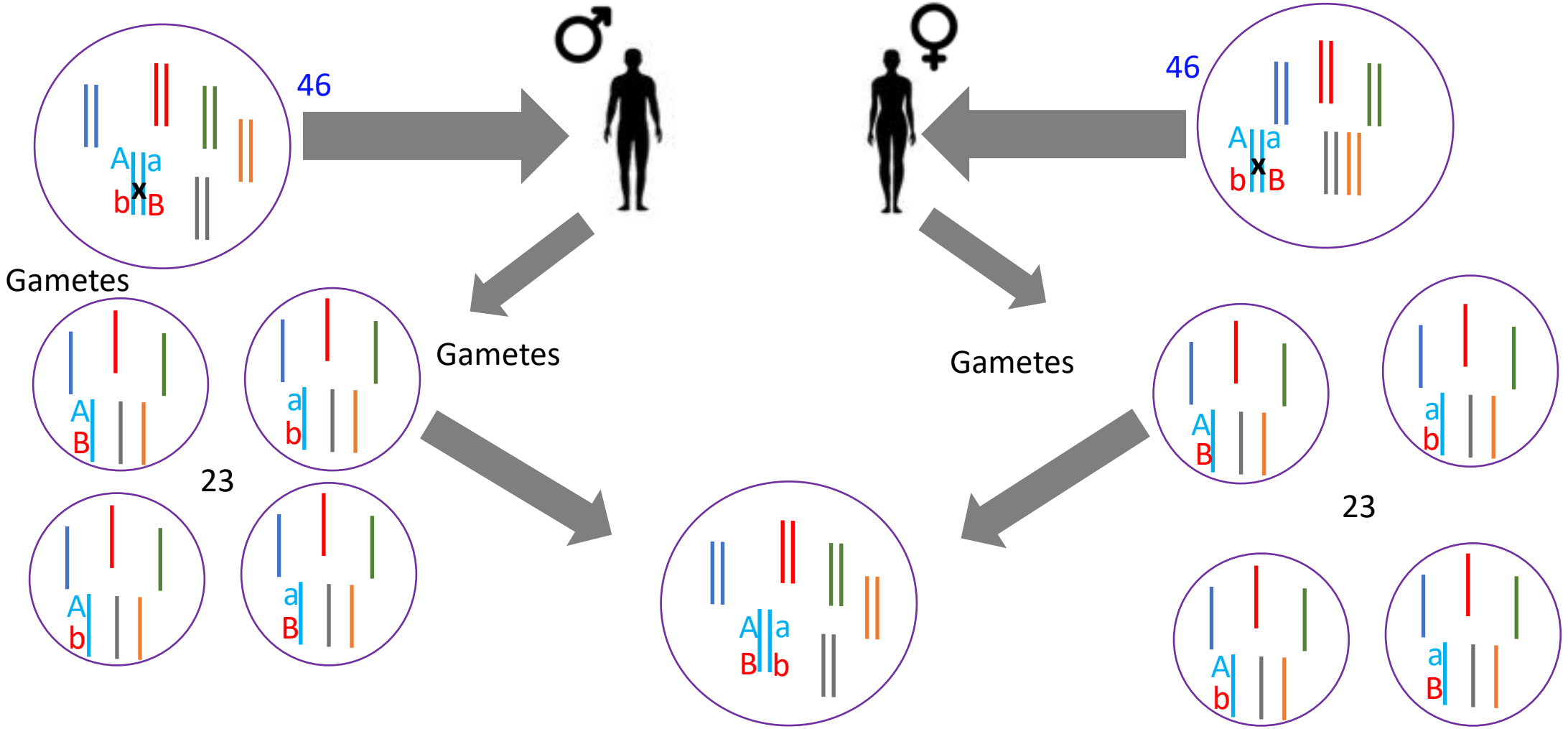
Genetic recombination prior to gamete formation

- Crossing Over: chromosome segments are exchanged between homologous chromosomes



- Discovered by Joshua Lederberg (1946), Nobel (1958)

Mendel's laws



Mendel's 1st law

- **MENDEL'S 1ST LAW:** Alleles segregate from each other when gametes form, so each gamete carry one allele

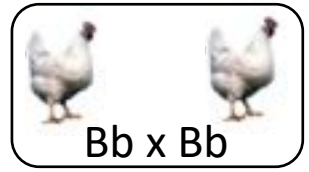
- **<u>Punnett Square:</u>** Diagram showing the gene combinations and **probability** of inheriting a particular trait

BB

bb

♂/♀	B	B
b	Bb	Bb
b	Bb	Bb

= 100%



Bb

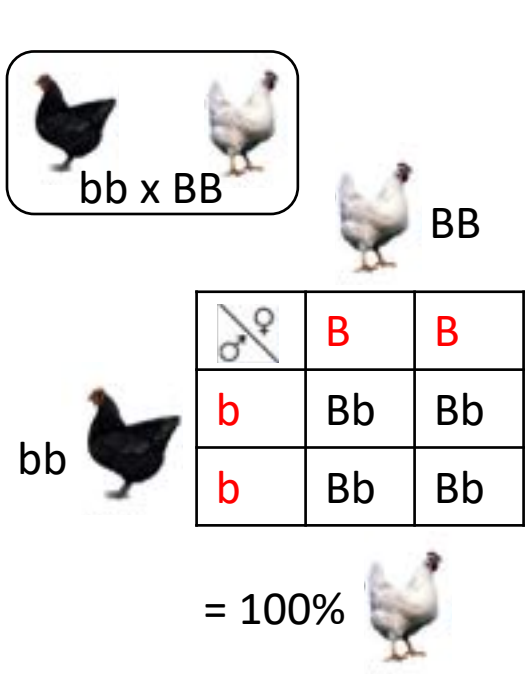
Bb

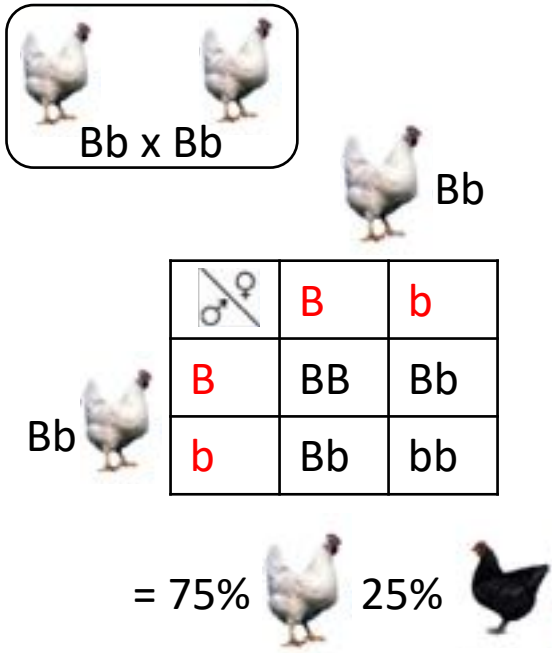
♂/♀	B	b
B	BB	Bb
b	Bb	bb

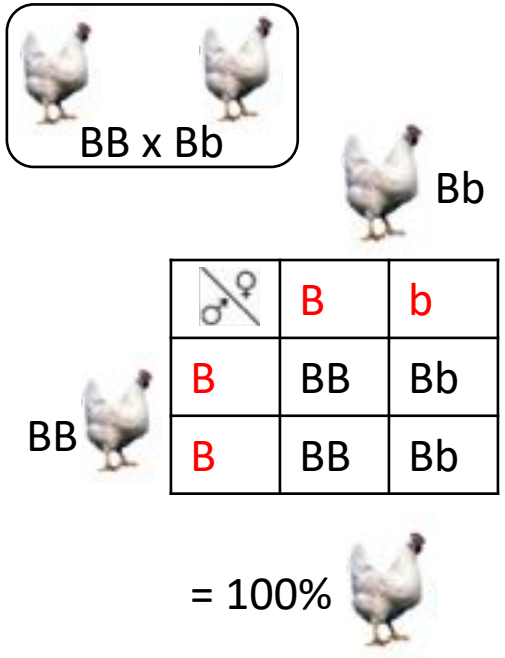
= 75%

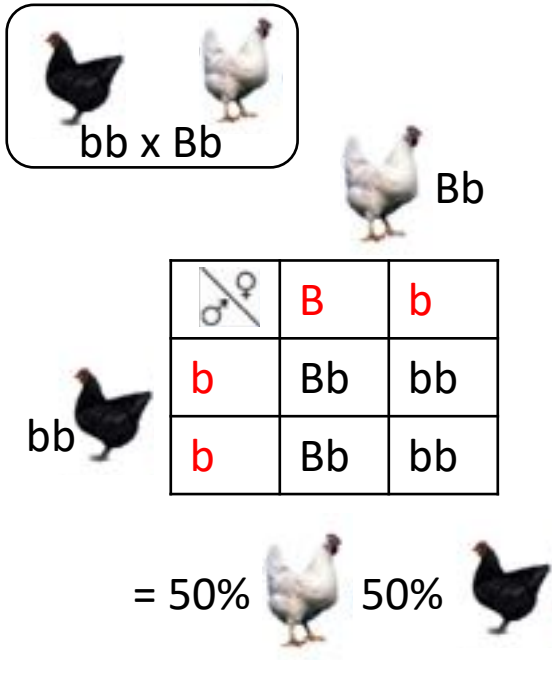
25%

Mendel's 1st law: more examples









Mendel's 2nd law

- **MENDEL'S 2nd LAW:** Alleles for different traits are inherited independently of each other


bbrr


BBRR

♂/♀	**br**	**br**
BR	BbRr	BbRr
BR	BbRr	BbRr

= 100% BbRr

Feather colour	**White**/black	B/b
Size	**Big**/small	R/r

Mendel's 2nd law: more examples


BbRr x BbRr


WwRr

9: 3: 3: 1


WwRr
WWRR
WWRr
WwRR


WWrr
Wwrr


wwRR
wwRr


wwrr


WwRr

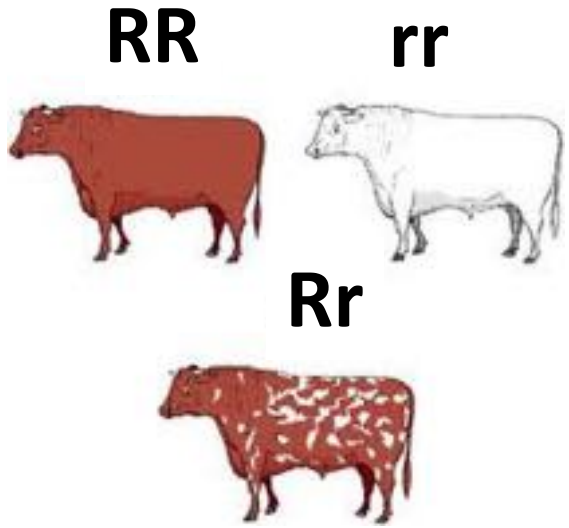
♂/♀	**BR**	**Br**	**bR**	**br**
BR	BBRR	BBRr	BbRR	BbRr
Br	BBRr	BBrr	BbRr	Bbrr
bR	BbRR	BbRr	bbRR	bbRr
br	BbRr	Bbrr	bbRr	bbrr

Codominance

Situation in which both alleles of a gene contribute to the phenotype of the organism.

Example:

- A solid white ox is crossed with a solid brown cow and the resulting offspring are spotted brown and white (called roan).



Incomplete dominance

Situation in which dominant alleles of a gene partially suppress the phenotype of the organism.

Example:

- A red petunia flower is crossed with a white petunia and the resulting plants are pink.

RR **Rr** **rr**

Question

- Parents are:

Child is:

- It is an example of _____

  (a) codominance

  (b) incomplete dominance

  (c) recessive

Pedigree analysis

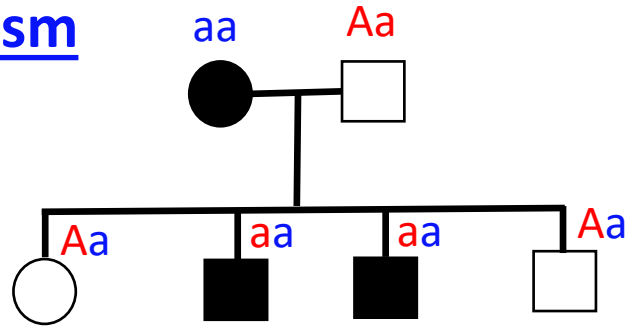
○ Female

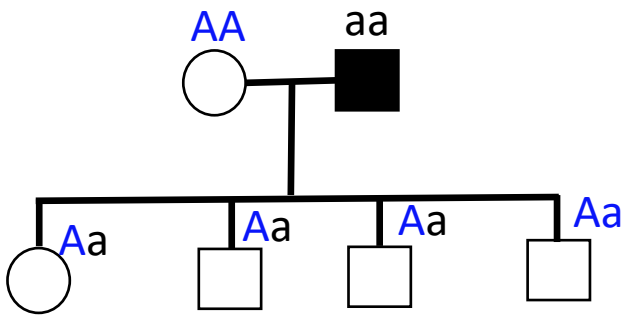
● Female with disorder

□ Male

■ Male with disorder

Albinism





Example:

Albinism (no melanin in the skin cells) **recessive**

Pedigree analysis

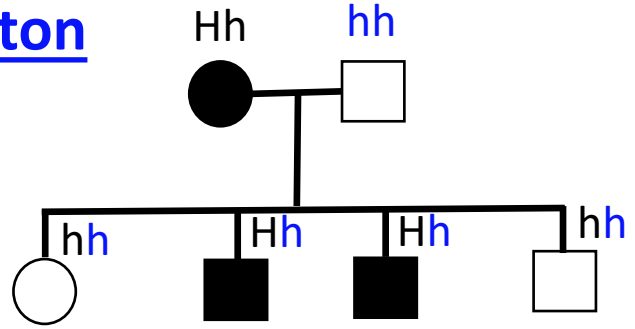
○ Female

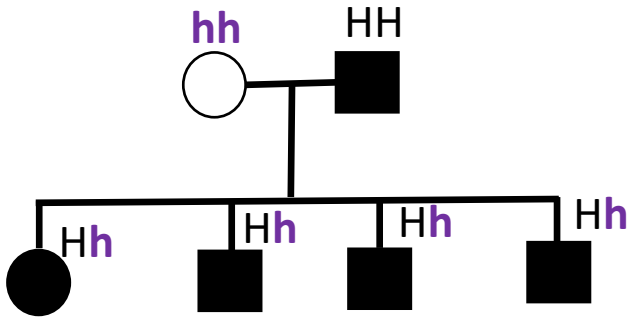
● Female with disorder

□ Male

■ Male with disorder

Huntington





Example:

Huntington disease (neurological disorder) **dominant**

Multiple genes with multiple alleles

- Very few traits actually only have two alleles with clear dominance.
- **Most genes often have many of alleles**

- Traits also can be **polygenic**
- Example: human skin colour

Three alleles: Human Blood group

- Blood group refers to the **antigens** (small proteins) that are found on the red blood cells
- Used for recognizing "self" and “non-self”

- **“A”** blood group would have A antigens, **“B”** would have B antigens
- Blood Group **“O”** has no antigen

- If blood group **“A”** patient receives group **“B”** blood, then that would be rejected as “non-self”

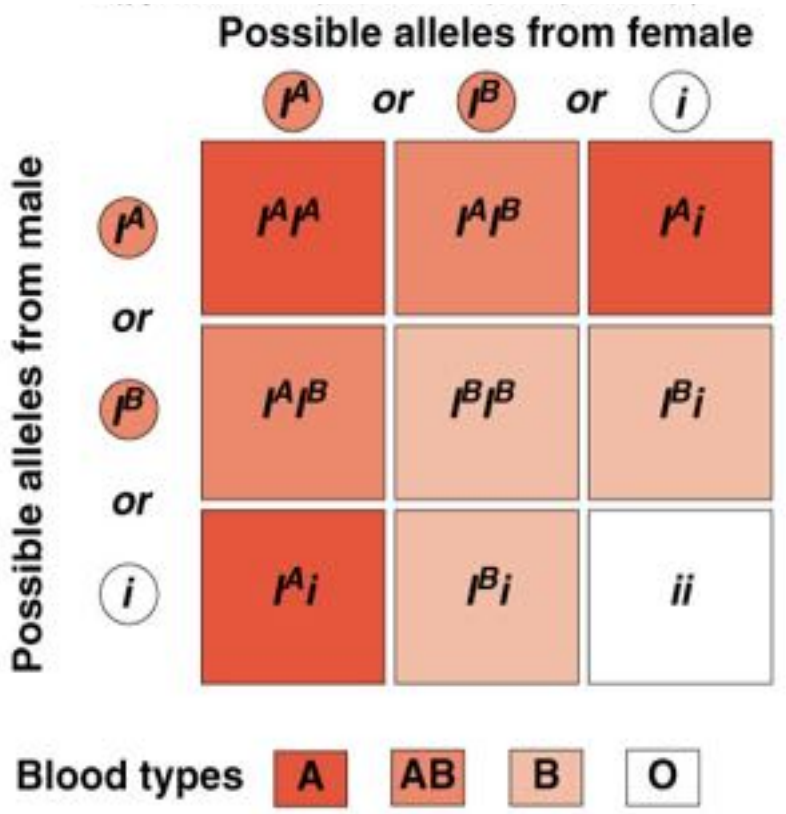
Three alleles: Human Blood group

Blood Group Type

- **3 alleles exist** I^A, I^B, i which results in four different possible blood types
- **Allele I^A and I^B are codominant**
- Allele **i** is **recessive**

Genotype Phenotype (group)

- $I^A I^A, \quad I^A i \quad = A$
- $I^A I^B \quad = AB$
- $I^B I^B, \quad I^B i \quad = B$
- $i\, i \quad = O$



Questions

If blood group O mother of has O group child, what is father's blood group?

Mother is ii

Father cannot be I^AI^A or I^BI^B or I^AI^B

Father can be I^Ai or I^Bi or ii

ii

	♂/♀	i	i
I^A i	i	ii	ii
	I^A	I^Ai	I^Ai

ii

	♂/♀	i	i
I^B i	i	ii	ii
	I^B	I^Bi	I^Bi

ii

	♂/♀	i	i
ii	i	ii	ii
	i	ii	ii