# Exams

• Ist Midterm Sep. 17 (2pm)

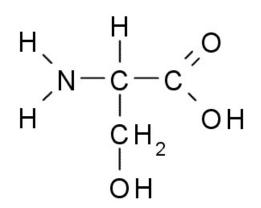
• Final exam correction: Dec. 15 (8am)

## How do genes code for characteristics?

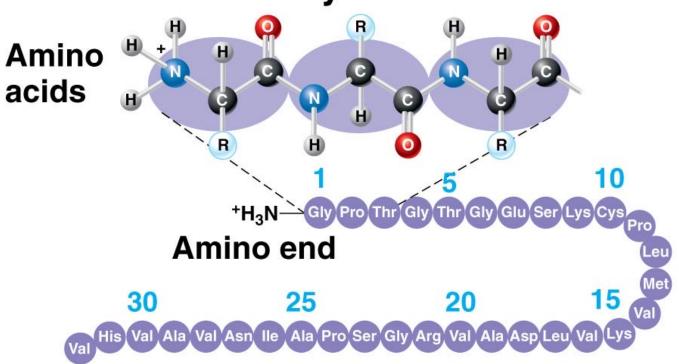
## Genes code for proteins

- = Polymers of amino acids = polypeptides
- constructed from 20 amino acids

Example amino acid: serine







# Nearly every dynamic function of a living being depends on proteins

### **Enzymatic proteins**

Function: Selective acceleration of

chemical reactions

Example: Digestive enzymes catalyze the hydrolysis of bonds in food molecules.

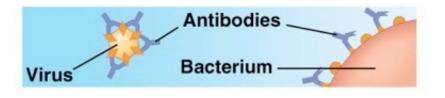


#### Defensive proteins

Function: Protection against disease

Example: Antibodies inactivate and help

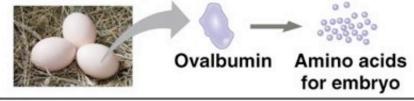
destroy viruses and bacteria.



#### Storage proteins

Function: Storage of amino acids

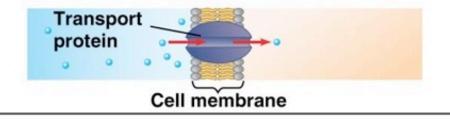
Examples: Casein, the protein of milk, is the major source of amino acids for baby mammals. Plants have storage proteins in their seeds. Ovalbumin is the protein of egg white, used as an amino acid source for the developing embryo.



#### **Transport proteins**

**Function: Transport of substances** 

Examples: Hemoglobin, the iron-containing protein of vertebrate blood, transports oxygen from the lungs to other parts of the body. Other proteins transport molecules across membranes, as shown here.



#### Hormonal proteins

Function: Coordination of an organism's

activities

Example: Insulin, a hormone secreted by the pancreas, causes other tissues to take up glucose, thus regulating blood sugar concentration.

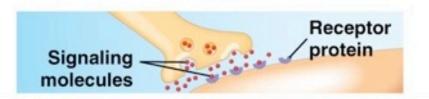


#### Receptor proteins

Function: Response of cell to chemical

stimuli

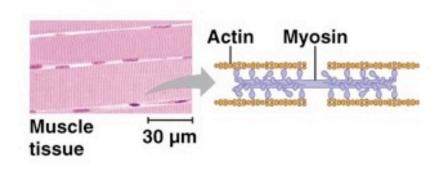
Example: Receptors built into the membrane of a nerve cell detect signaling molecules released by other nerve cells.



#### Contractile and motor proteins

**Function: Movement** 

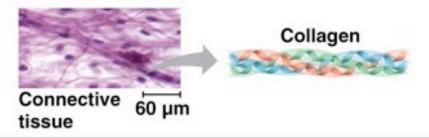
Examples: Motor proteins are responsible for the undulations of cilia and flagella. Actin and myosin proteins are responsible for the contraction of muscles.



#### Structural proteins

Function: Support

Examples: Keratin is the protein of hair, horns, feathers, and other skin appendages. Insects and spiders use silk fibers to make their cocoons and webs, respectively. Collagen and elastin proteins provide a fibrous framework in animal connective tissues.



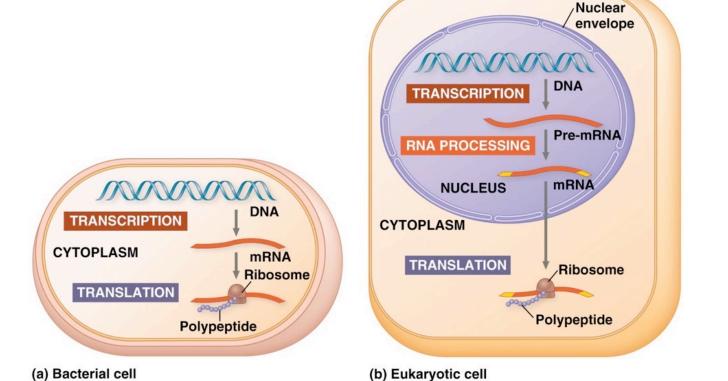
# mRNA is translated into amino acids (includes a processing step in eukaryotic organism)

The genetic code is based on nitrogenous base triplets (codons)

The way the genetic code is shown in literature is usually the mRNA

Example start and stop codes of a gene: AUG (start), UGA (stop)

Example amino acid: AGU = serine



#### mRNA is translated into amino acids

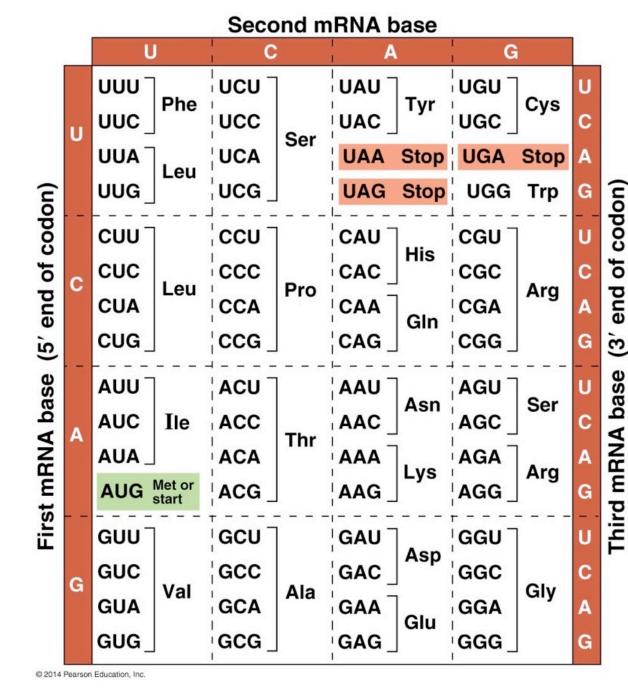
The flow of information from gene to protein is based on a triplet code

Every three bases (a triplet)
codes for a single, specific
amino acid (but each amino acid
is associated with several
codes)

Proteins are made by transcribing and translating this code

Certain codes stand for "start" (AUG) or "stop" (e.g. UGA)

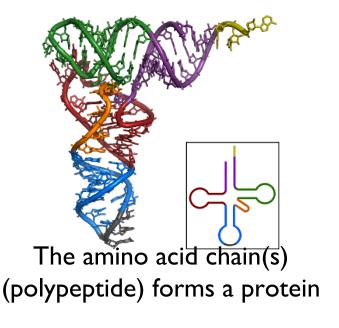
(amino acids are abbreviated; e.g. Ser=serine, Met=methionine)

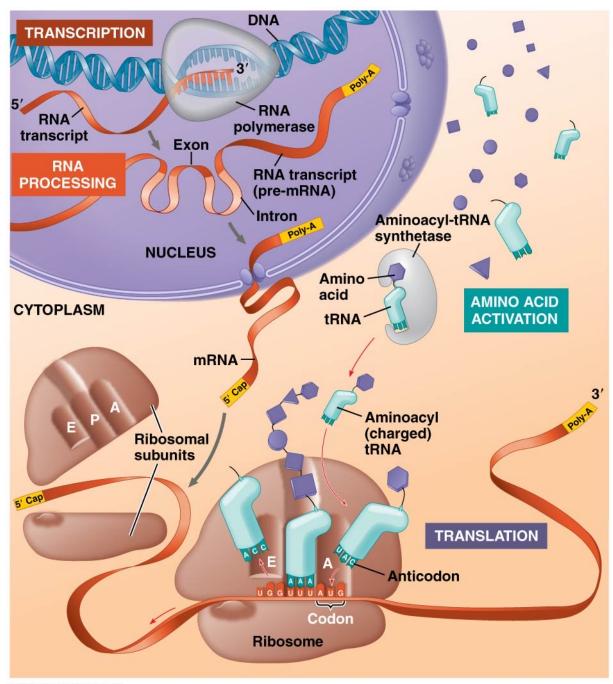


## Transcription and translation summary

Transcription in nucleus: DNA is opened and the code is transcribed to an mRNA (messenger RNA) molecule (with some enzyme processing)

Translation in ribosome: done with the help of tRNA (transfer RNA) molecules which bind to specific amino acids





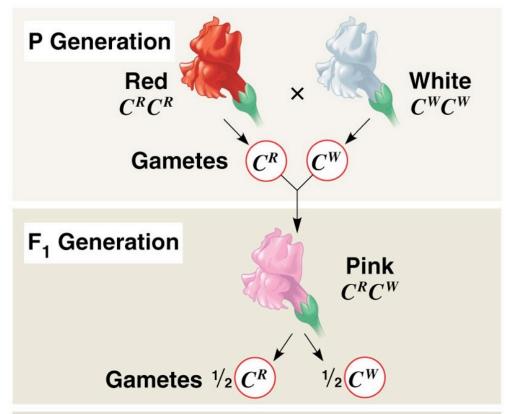
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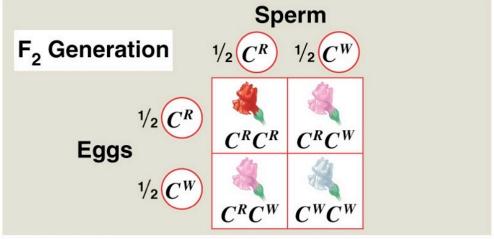
# Incomplete dominance (heterozygous individuals appear as a blend)











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# Codominance

(a) The three alleles for the ABO blood groups and their carbohydrates					
Allele	$I^A$	$I^B$	i		
Carbohydrate	<b>A</b> 🛆	ВО	none		

(b) Blood group genotypes and phenotypes					
Genotype	$I^AI^A$ or $I^Ai$	$I^BI^B$ or $I^Bi$	$I^AI^B$	ii	
Red blood cell appearance					
Phenotype (blood group)	A	В	АВ	O	

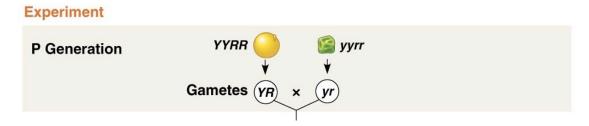
Note: ABO blood types; a three-allele system

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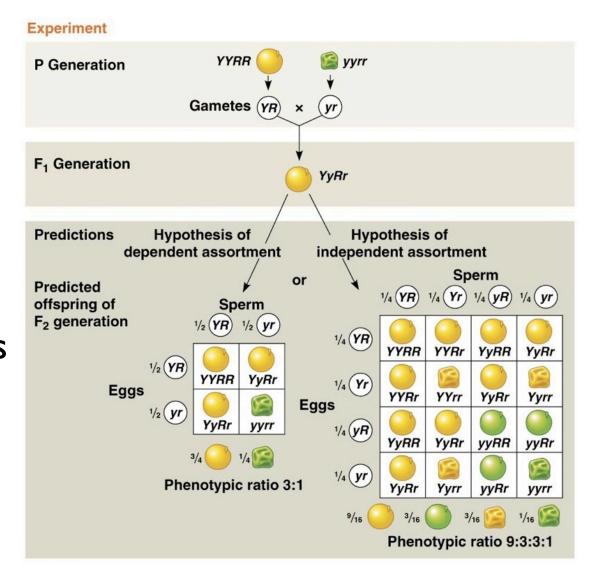
# The spectrum of dominance

- Complete dominance: BB
   (Brown) x bb (blue) = Bb (Brown)
- Codominance:  $I^{A}I^{A}$  (type A) x  $I^{B}I^{B}$  (type B) =  $I^{A}I^{B}$  (type AB)
- Incomplete dominance:  $C^RC^R$  (red) x  $C^BC^B$  (blue) =  $C^RC^B$  (purple)

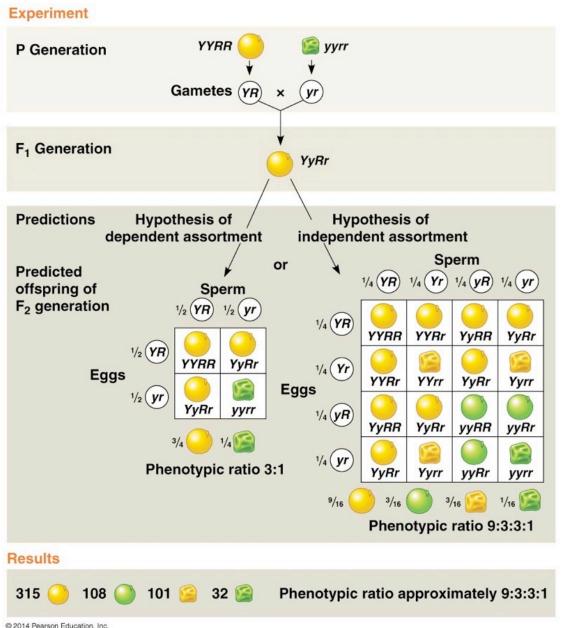
Mendel again: What about a cross involving multiple characters?



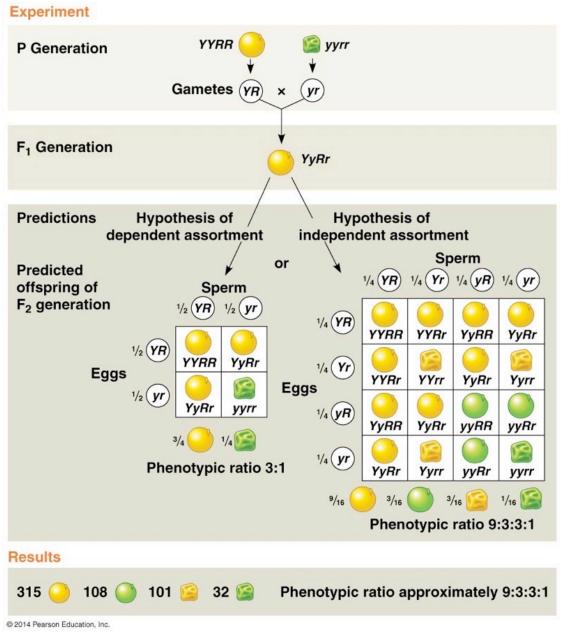
- One true-breeding population with yellow round seeds; the other is true-breeding with green wrinkly seeds
- Mendel already knew that yellow and round were dominant traits



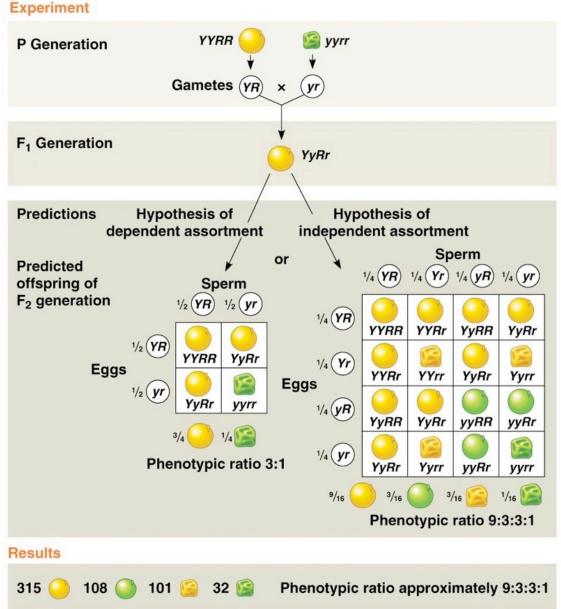
Mendel's 2 Hypotheses



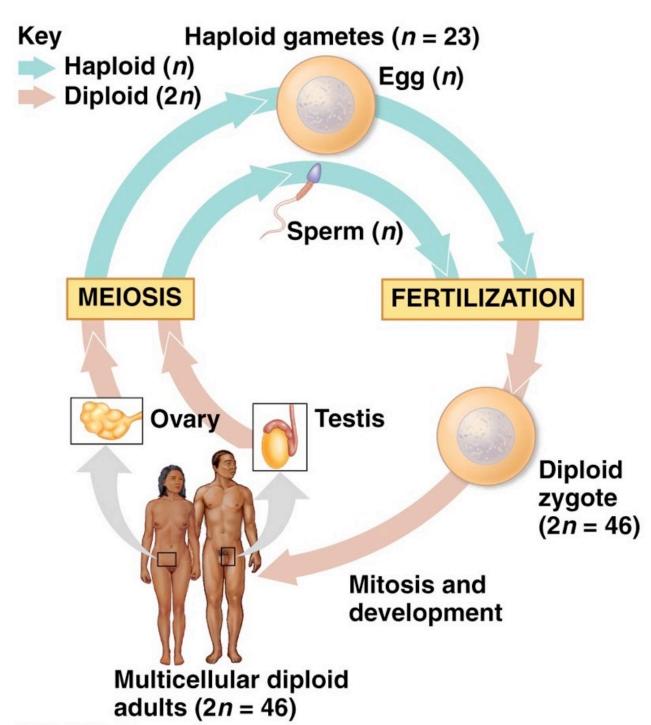
Mendel called this the law of independent assortment.



However: today we know this is true only because these genes are on different chromosomes



Conversely; linked genes on the same chromosome tend to be inherited together (the numbers always deviate somewhat due to cross over recombination)

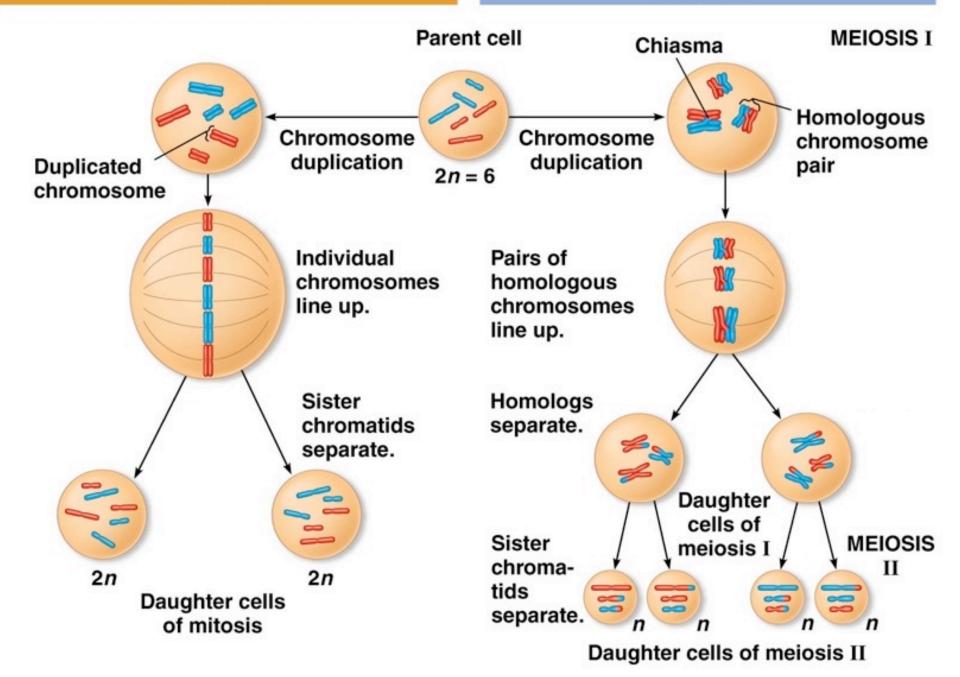


Sexually reproducing organisms have both haploid and diploid cells

haploid(n) = gametes diploid(2n) = somatic cells n=23 in humans

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MITOSIS MEIOSIS



Somatic Cells

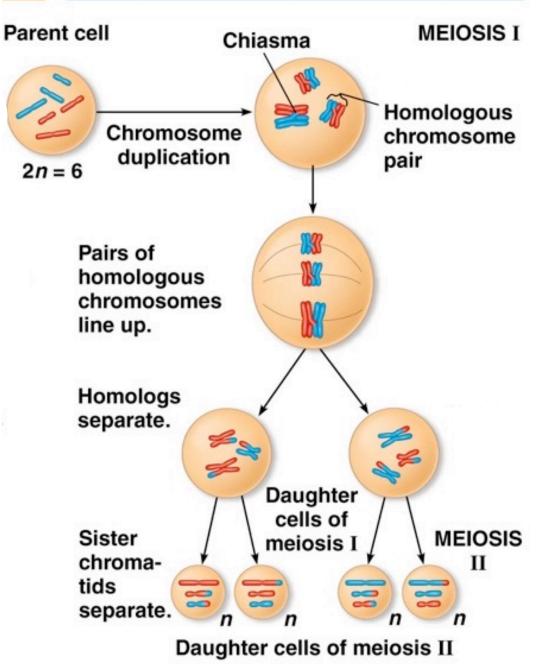
## Parent cell Chromosome duplication **Duplicated** 2n = 6chromosome Individual chromosomes line up. Sister chromatids separate. 2n2nDaughter cells of mitosis

Mitosis: chromosomes are duplicated. Then sister chromatids are pulled apart in one cell division, forming new chromosomes

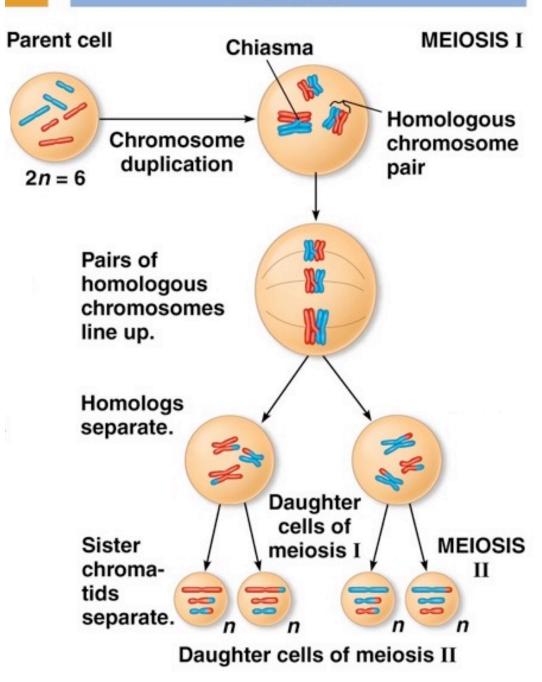
Somatic Cells

Meiosis: production of gametes (sperm and egg)

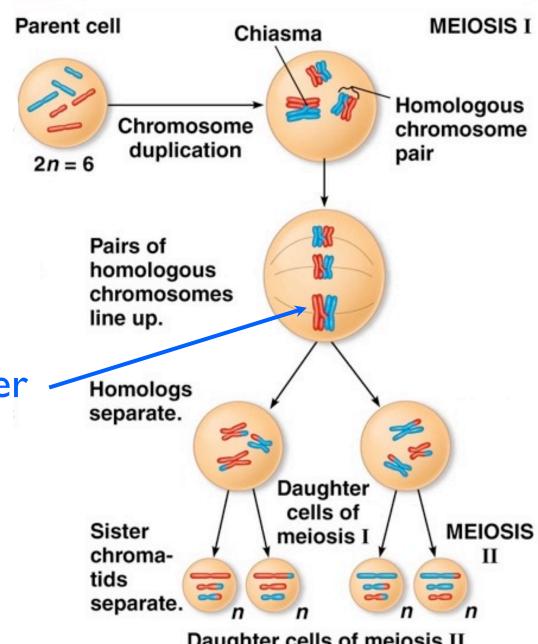
Two cell divisions. The homologous chromosomes from each parent are separated first (Meiosis I), then the sister chromatids (Meiosis II)



Meiosis: Two alleles for a heritable character segregate (separate) during gamete formation and end up in different gametes

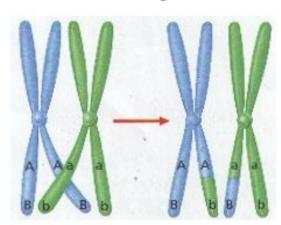


#### **MEIOSIS**



Crossing over

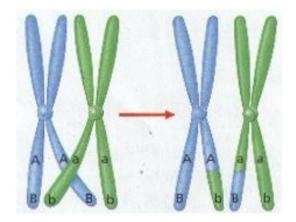
Crossing-over



Daughter cells of meiosis II

# Recombination of linked genes: crossing over

- Genes on one chromosome are linked, but the linkage is not absolute, as evident from recombinant phenotypes
- Crossing over of homologous chromosomes results in new combinations of alleles (recombination)



# Mutation

- More or less random and spontaneous changes in the genome (e.g. duplication error)
- Creates new genetic variation

- Large scale mutations: changes to entire chromosomes
- Small scale mutations: e.g. point mutation (single nucleotide pair affected, e.g. sickle-cell anemia)
- Many mutations have no effect on phenotype: silent mutation

