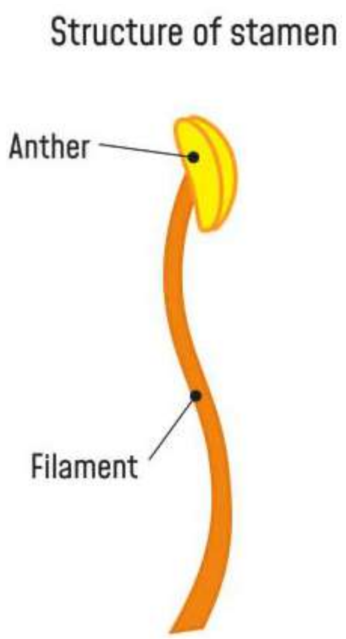
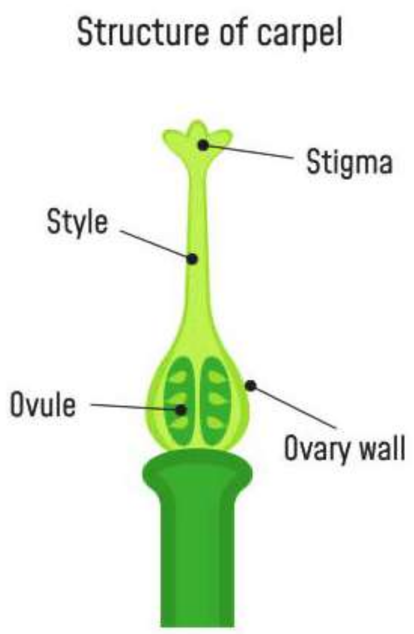
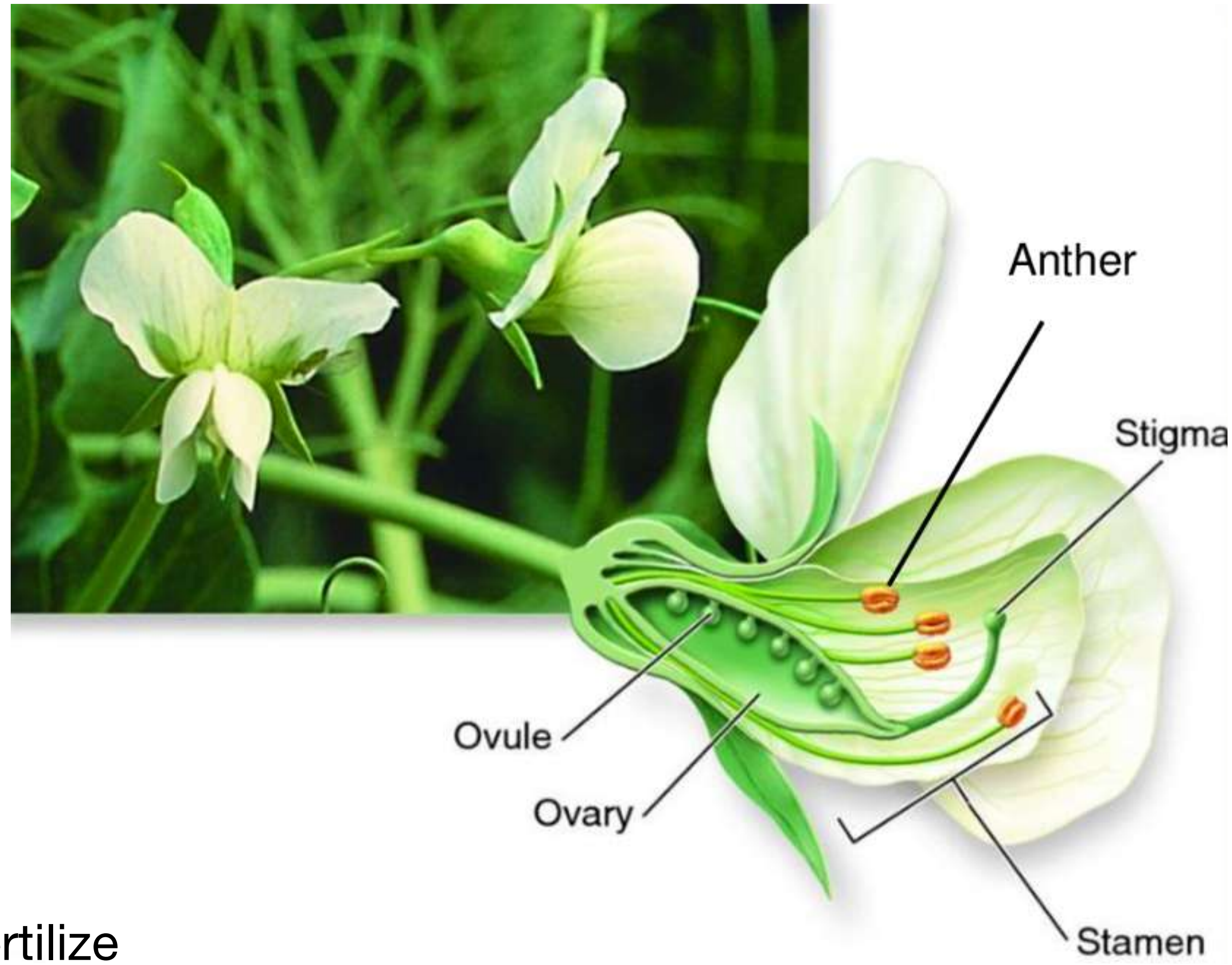
Gregor Mendel and the Pea plant

Male



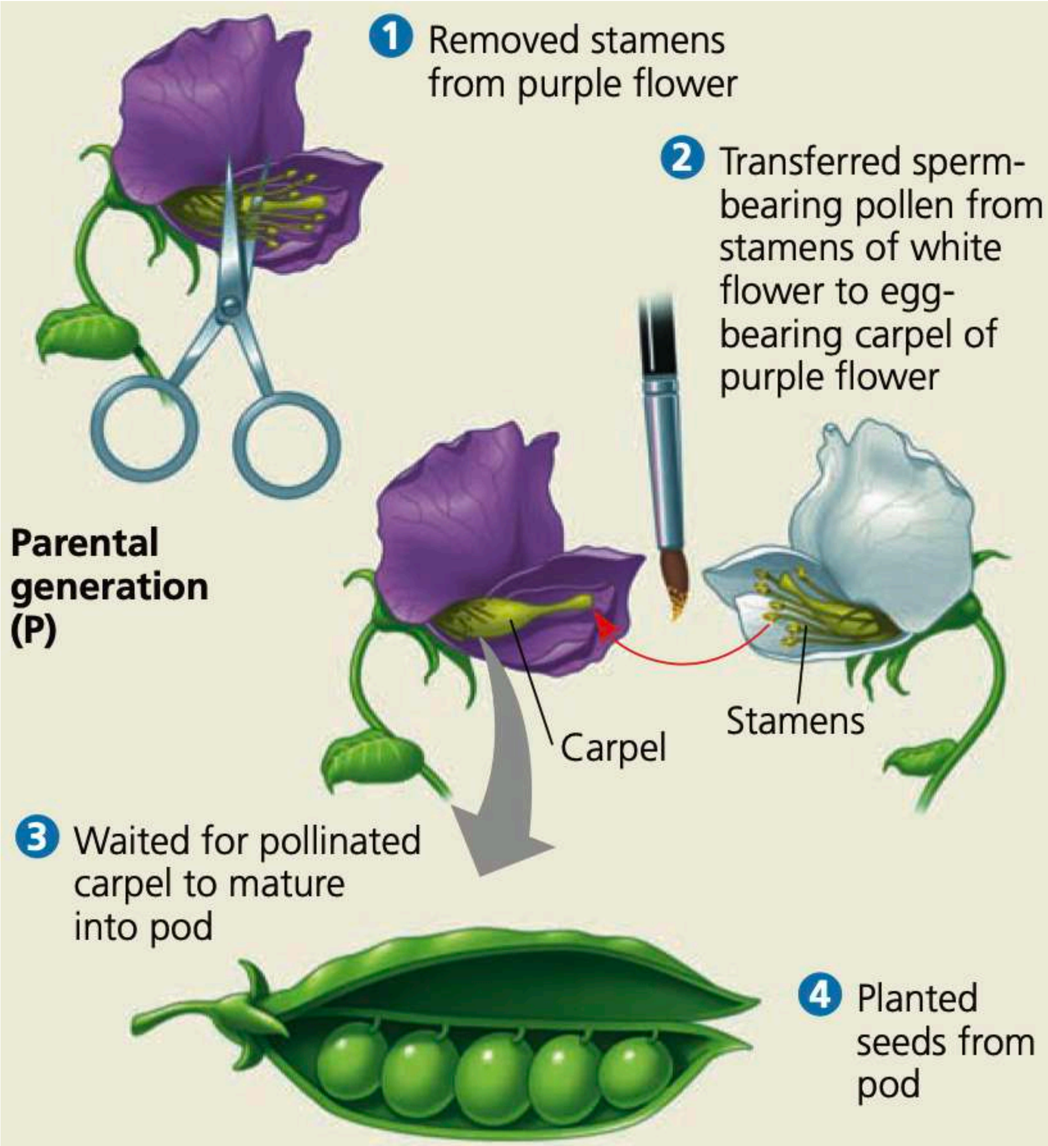
Female



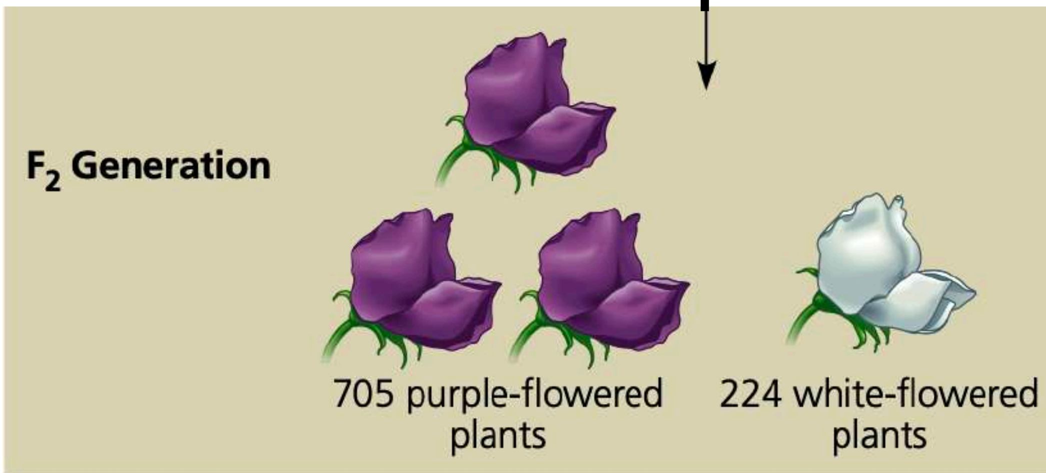


- Plants can self-fertilize or cross-fertilize
- Can take pollen from anther of one flower and dust it on the stigma of another flower
- Technical advantage for experimental mating (crossing) of plants

Gregor Mendel and the Pea plant



Self or cross-fertilize F1 generation



The result was the same for the reciprocal cross, which involved the transfer of pollen from purple flowers to white flowers

Figure 14.2 of Campbell's Biology: a global approach

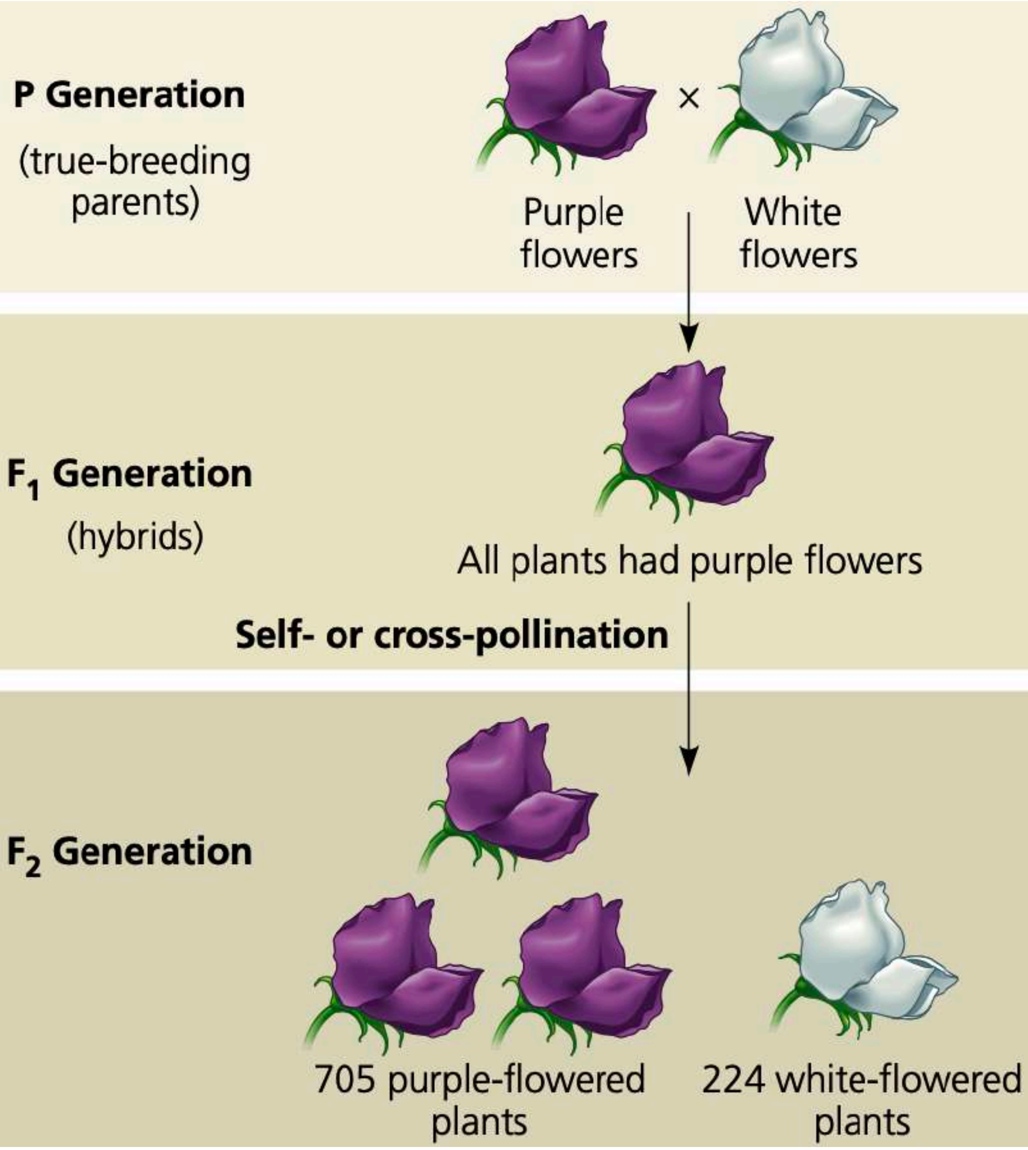
Sreelaja Nair

Gregor Mendel and the Pea plant

- Mendel chose to track only those characters that occurred in two distinct, alternative forms, such as purple or white flower color
- He started his experiments with varieties that, over many generations of self-pollination, had produced only the same variety as the parent plant = true breeding
- The mating, or cross-pollinating (crossing) of two true-breeding varieties is called hybridization
- The true-breeding parents are referred to as the P generation (parental generation), and their hybrid offspring are the F1 generation (first filial generation, the word filial from the Latin word for “son”).
- Allowing these F1 hybrids to self-pollinate (or to cross-pollinate with other F1 hybrids) produces an F2 generation (second filial generation).

- Mendel’s quantitative analysis of the F2 plants from thousands of genetic crosses like these allowed him to deduce two fundamental principles of heredity, which have come to be called the law of segregation and the law of independent assortment

Law of Segregation



Crossed two true-breeding plants that each produce Purple or White flowers

When seeds from F1 generation grew into plants, all plants produced Purple flowers. What happened to the information to produce White flowers?

When F1 generation self or cross-fertilized to produce seeds and when those seeds grew into plants, some of F2 plants produced White flowers

White flowers in F2 plants did not appear randomly, they appeared in a ratio of 3:1 (3 purple flower plants:1 White flower plant)

Figure 14.3 of Campbell's Biology: a global approach

Law of Segregation

- Mendel reasoned that the heritable factor for white flowers did not disappear in the F1 plants but was somehow hidden, or masked, when the purple-flower factor was present.
- In Mendel's terminology, purple flower color is a dominant trait, and white flower color is a recessive trait.
- The reappearance of white-flowered plants in the F2 generation was evidence that the heritable factor causing white flowers had not been diluted or destroyed by coexisting with the purple-flower factor in the F1 hybrids.
- Instead, it had been hidden when in the presence of the purple flower factor.
- Mendel observed the same pattern of inheritance in six other characters, each represented by two distinctly different traits

Law of Segregation

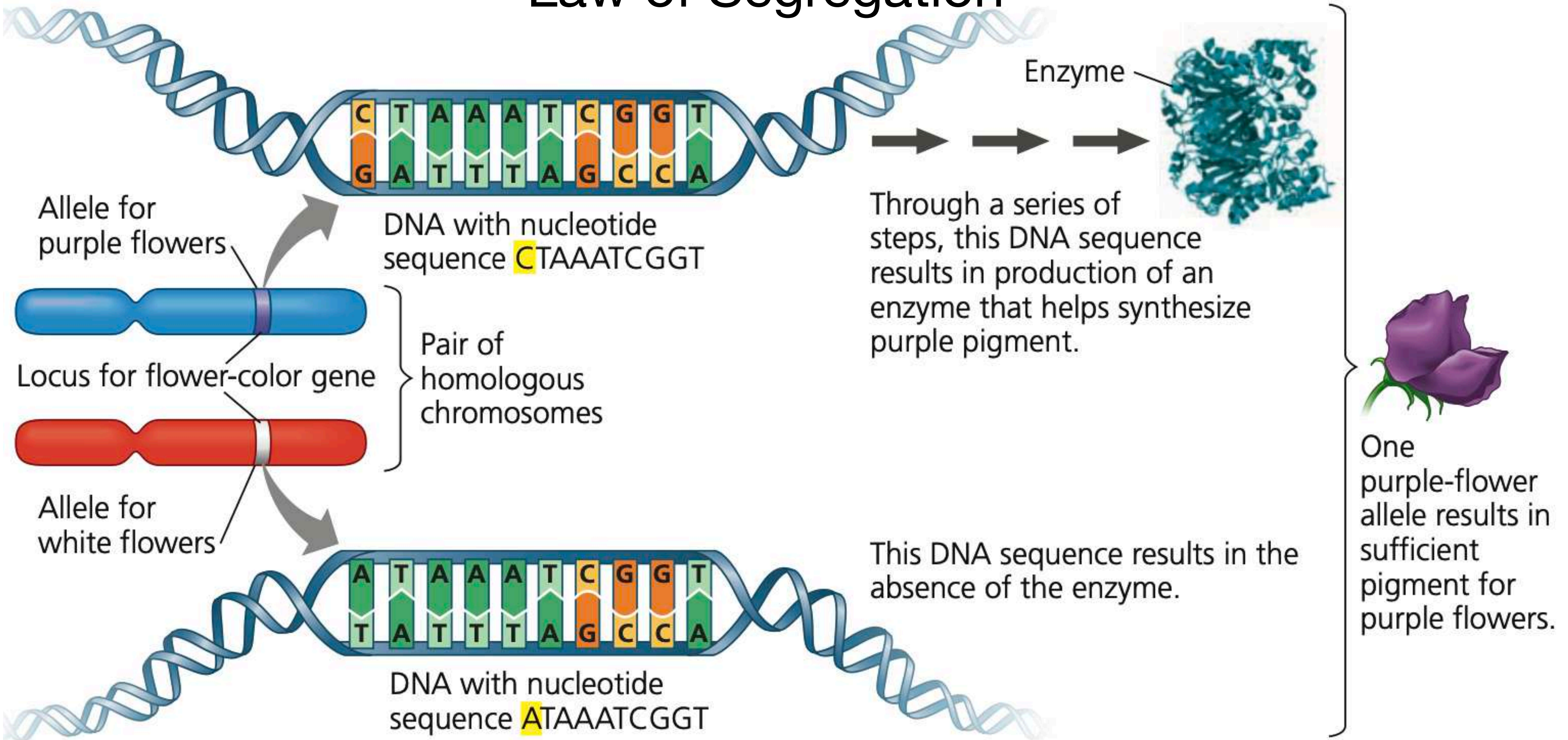
Table 14.1 The Results of Mendel's F_1 Crosses for Seven Characters in Pea Plants

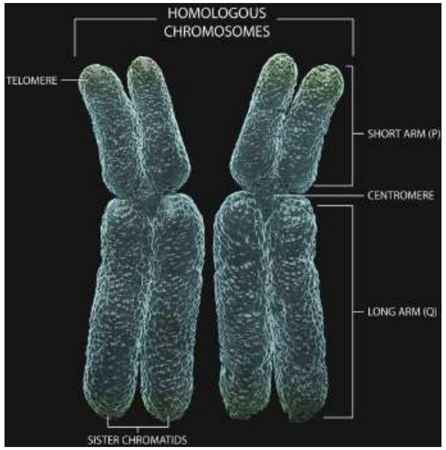
Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Flower color	Purple	×	White	705:224	3.15:1
Seed color	Yellow	×	Green	6,022:2,001	3.01:1
Seed shape	Round	×	Wrinkled	5,474:1,850	2.96:1
Pod shape	Inflated	×	Constricted	882:299	2.95:1
Pod color	Green	×	Yellow	428:152	2.82:1
Flower position	Axial	×	Terminal	651:207	3.14:1
Stem length	Tall	×	Dwarf	787:277	2.84:1

1. Alternative versions of genes account for variations in inherited characters.
2. For each character, an organism inherits two copies (that is, two alleles) of a gene, one from each parent.
3. If the two alleles at a locus differ, then one, the dominant allele, determines the organism's appearance; the other, the recessive allele, has no noticeable effect on the organism's appearance.
4. Law of segregation, states that the two alleles for a heritable character segregate (separate from each other) during gamete formation and end up in different gametes.

Table 14.1 of Campbell's Biology: a global approach

Law of Segregation

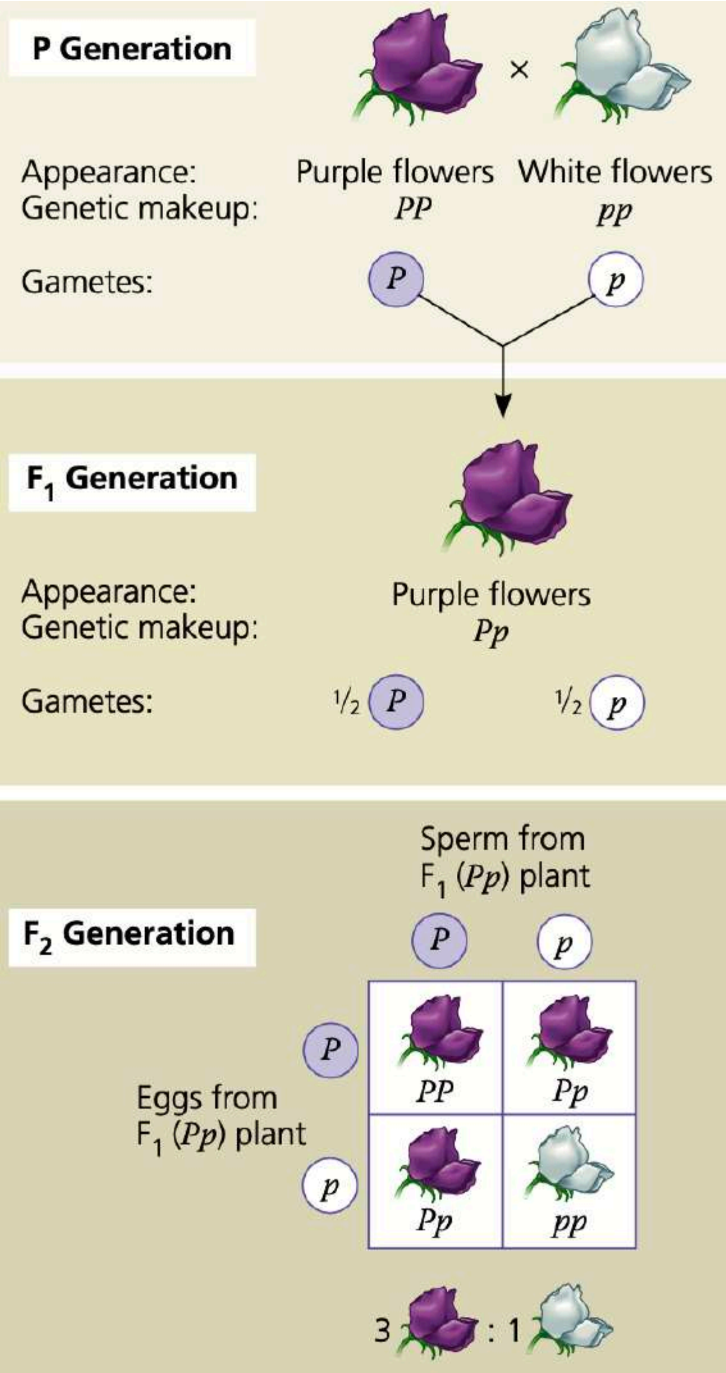




- Alleles = alternative versions of a gene
- For a diploid cell at metaphase, there will be 4 alleles of a gene - one on each sister chromatid
- A haploid gamete has one chromatid at the end of meiosis = one allele
- When two haploid gametes fuse (fertilization) - embryo gets two alleles, one from each gamete

Figure 14.4 of Campbell's Biology: a global approach

Law of Segregation



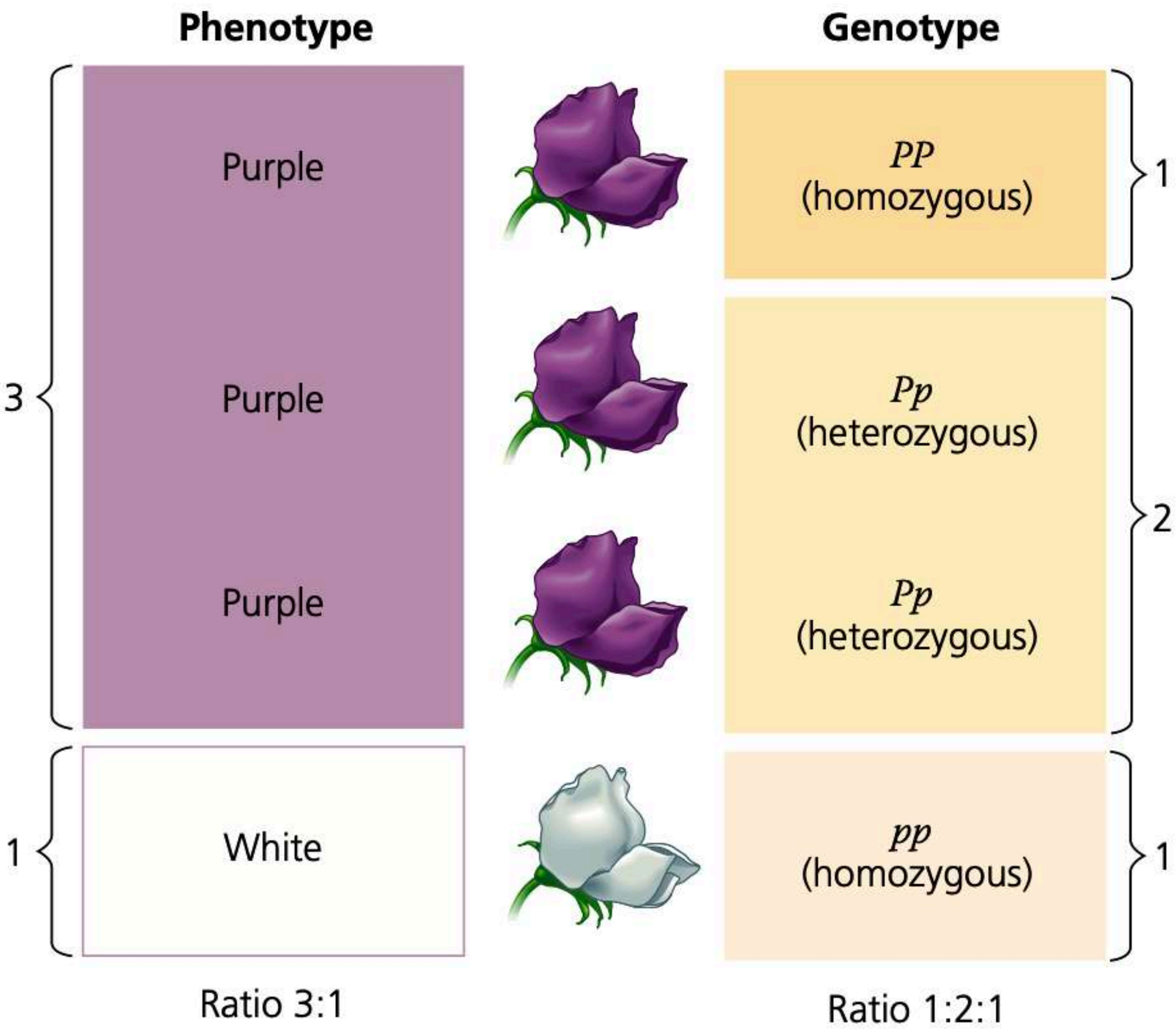
- Each true breeding P plant has one allele of the gene, either *P* or *p*. Each P plant has two copies of the allele (*PP* or *pp*) as it is a diploid
- Each P plant produces one type of haploid gamete, either, Purple allele gamete (*P*) or White allele gamete (*p*)

- When gametes from two P plants fuse, they can only form one diploid combination in F1 upon fertilization: Pp
- The cross-pollinated F1 plant has two alleles *P* and *p*, but P is dominant and therefore F1 plant has Purple flowers
- Each F1 plant produces two types of gametes: *P* and *p* = segregation

- When gametes of F1 plants fuse (self or cross-fertilization), they can form three types of diploid combinations in F2: PP or Pp or pp.
- PP = Purple flower F2 plant = like one type of P generation
- Pp = Purple flower F2 plant = like the F1 generation
- pp = White flower F2 plant = like the second type of P generation, reappearing after skipping one generation

Figure 14.5 of Campbell's Biology: a global approach

Law of Segregation



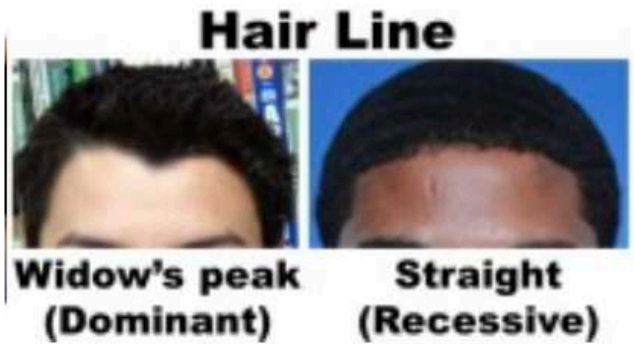
- Phenotype = organism's appearance or observable traits
- Genotype = genetic makeup or allele combinations
- Phenotype and Genotype ratios are mathematically predictable
- Mendel inferred these without knowing concepts of chromosomes or genes or meiosis or mitosis
- Homozygous inbreeding = offsprings will have genotype and phenotype of parent
- Heterozygous breeding = offsprings will have a mix of parents and grandparents genotype and phenotype

If you only know phenotype (e.g. purple flowers), it is impossible to be sure of the genotype (can be *PP* or *Pp*). Need to do a Test cross with a known true breeding *PP* and/or *pp* genotype and track offspring for multiple generations

Figure 14.6 of Campbell's Biology: a global approach

Examples of monogenic traits in humans - follows Law of Segregation

Disease examples: Sickle cell, cystic fibrosis, Duchenne muscular dystrophy, Hemophilia A and B, Retinitis Pigmentosa, β-Thalassemia and many more….



Many traits such as ring and index finger length, hand clasping etc can also be influenced by exposure to hormones and other factors as embryos develop and thus are not strictly monogenic.

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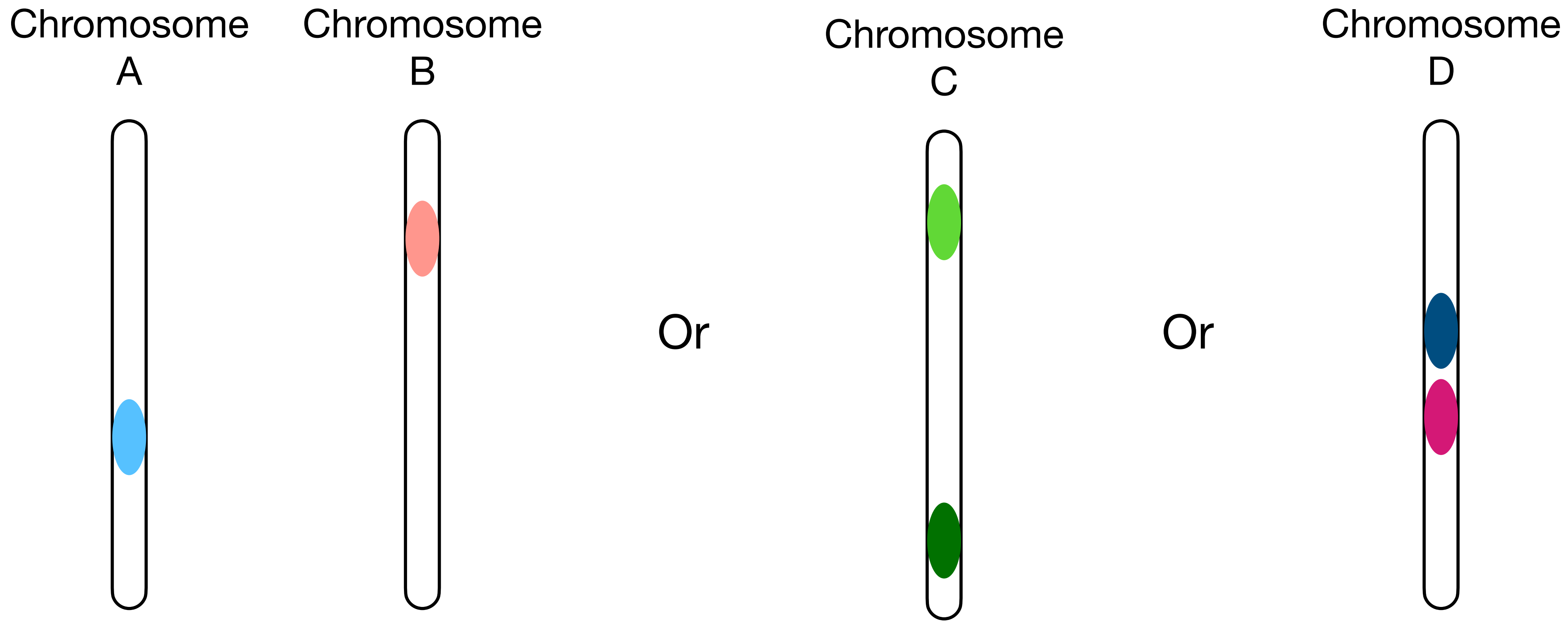
Law of Independent Assortment

Mendel deduced the Law of Segregation by following the inheritance patterns of single traits controlled by one gene

What about 2 traits controlled by 2 genes or more?

The 2 traits can be on different chromosomes or on the same chromosome and if on the same chromosome, they can be close or far from each other



Only one chromatid shown in diagrams above. Ovals represent genes

Law of Independent Assortment

Table 14.1 The Results of Mendel's F_1 Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Flower color	Purple	×	White	705:224	3.15:1
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Flower position	Axial	×	Terminal	651:207	3.14:1
Stem length	Tall	×	Dwarf	787:277	2.84:1

In nature multiple traits occur together: For example: short plant with round seeds, wrinkled and yellow

Mendel followed 2 traits at a time

Because of his monohybrid crosses, Mendel knew which traits were dominant and recessive

Example: Yellow or Green seed color and Round or Wrinkled seed shape

Table 14.1 The Results of Mendel's F_1 Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Seed color	Yellow	×	Green	6,022:2,001	3.01:1
Seed shape	Round	×	Wrinkled	5,474:1,850	2.96:1

Table 14.1 of Campbell's Biology: a global approach

Law of Independent Assortment

Yellow Seed = dominant = *Y*
Green seed = recessive = *y*
Round Seed = dominant = *R*
Wrinkled seed = recessive = *r*

Table 14.1 The Results of Mendel's F_1 Crosses for Seven Characters in Pea Plants

	Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio	
YYRR	Seed color	Yellow 	×	Green 	6,022:2,001	3.01:1	*yyRR*
YYRR	Seed shape	Round 	×	Wrinkled 	5,474:1,850	2.96:1	*YYrr*

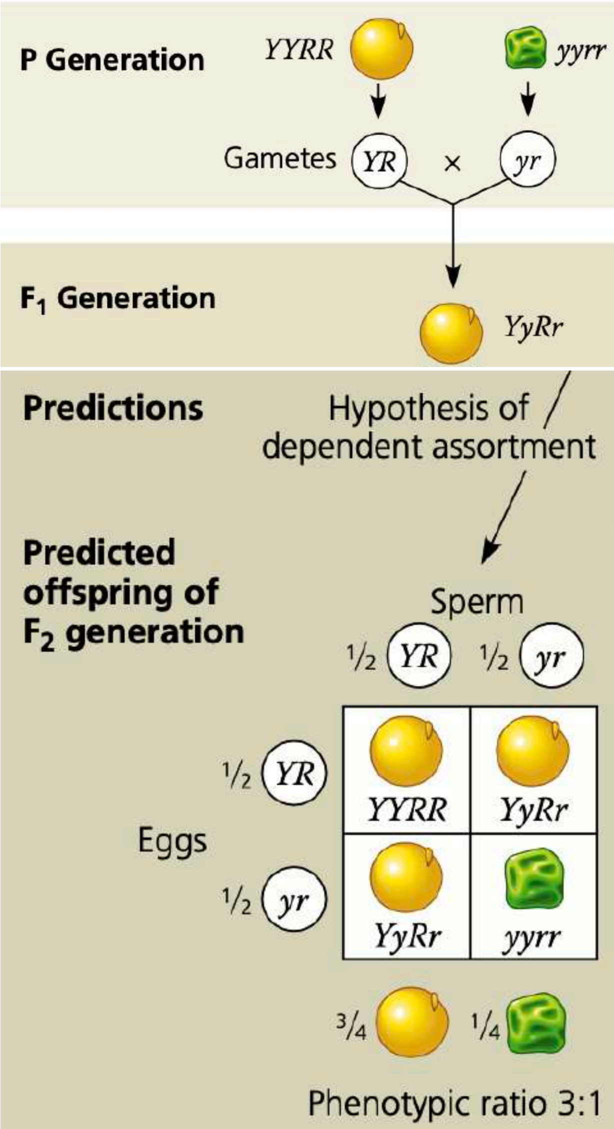
Write the diploid genotype for both traits for each of the seeds shown in the above picture

Table 14.1 of Campbell's Biology: a global approach

Law of Independent Assortment

Mendel crossed two true-breeding plants: yellow-round seeds x green-wrinkled seeds

Hypothesis being tested:
do true dominant or recessive traits always get inherited together?



- Each true breeding P plant has one allele of the *Y* gene and one allele of *R* gene or one allele of the *y* gene and one allele of *r* gene. Each P plant has two copies of the allele (*YY* and *RR* or *yy* and *rr*) as it is a diploid
- Each P plant produces one type of haploid gamete, either, yellow round allele gamete (*YR*) or green wrinkled allele gamete (*yr*)
- When gametes from two P plants fuse, they can only form one diploid combination in F1 upon fertilization: YyRr
- The cross-pollinated F1 plant has two alleles for each trait, *Y* and *y*, *R* and *r*, but Y and R are dominant and therefore the F1 plant has yellow round seeds.
- If each pair of dominant or recessive traits are to be inherited together: Each F1 plant will have to produce two types of gametes: *YR* and *yr*
- When gametes of F1 plants fuse (self or cross-fertilization), they can form three types of diploid combinations in F2: YYRR or YyRr or yyrr

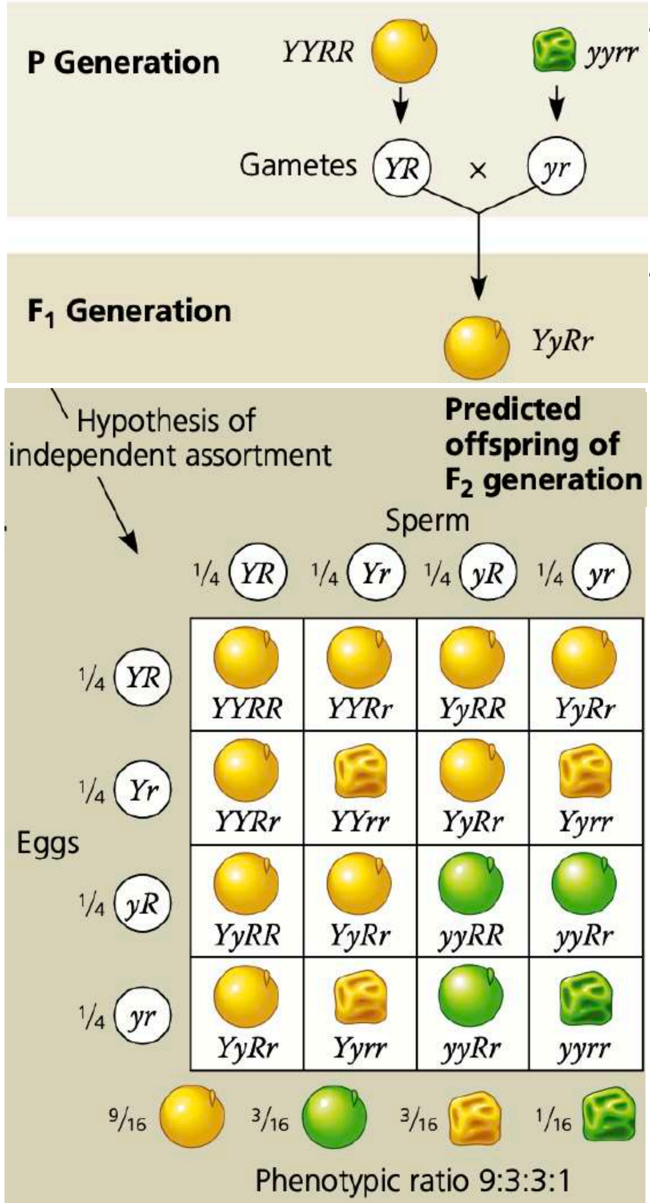
1:2:1 genotype and 3:1 phenotype ratio

THIS IS NOT THE RESULT MENDEL OBTAINED

Figure 14.8 of Campbell's Biology: a global approach

Law of Independent Assortment

Mendel crossed two true-breeding plants: yellow-round seeds x green-wrinkled seeds



- Each true breeding P plant has one allele of the *Y* gene and one allele of *R* gene or one allele of the *y* gene and one allele of *r* gene. Each P plant has two copies of the allele (*YY* and *RR* or *yy* and *rr*) as it is a diploid
- Each P plant produces one type of haploid gamete, either, yellow round allele gamete (*YR*) or green wrinkled allele gamete (*yr*)
- When gametes from two P plants fuse, they can only form one diploid combination in F1 upon fertilization: YyRr
- The cross-pollinated F1 plant has two alleles for each trait, *Y* and *y*, *R* and *r*, but Y and R are dominant and therefore the F1 plant has yellow round seeds.
- If each pair of dominant or recessive traits could be inherited separately: Each F1 plant will produce four types of gametes: *YR*, *Yr*, *yR* and *yr*
- When gametes of F1 plants fuse (self or cross-fertilization), they can form several types of diploid combinations in F2

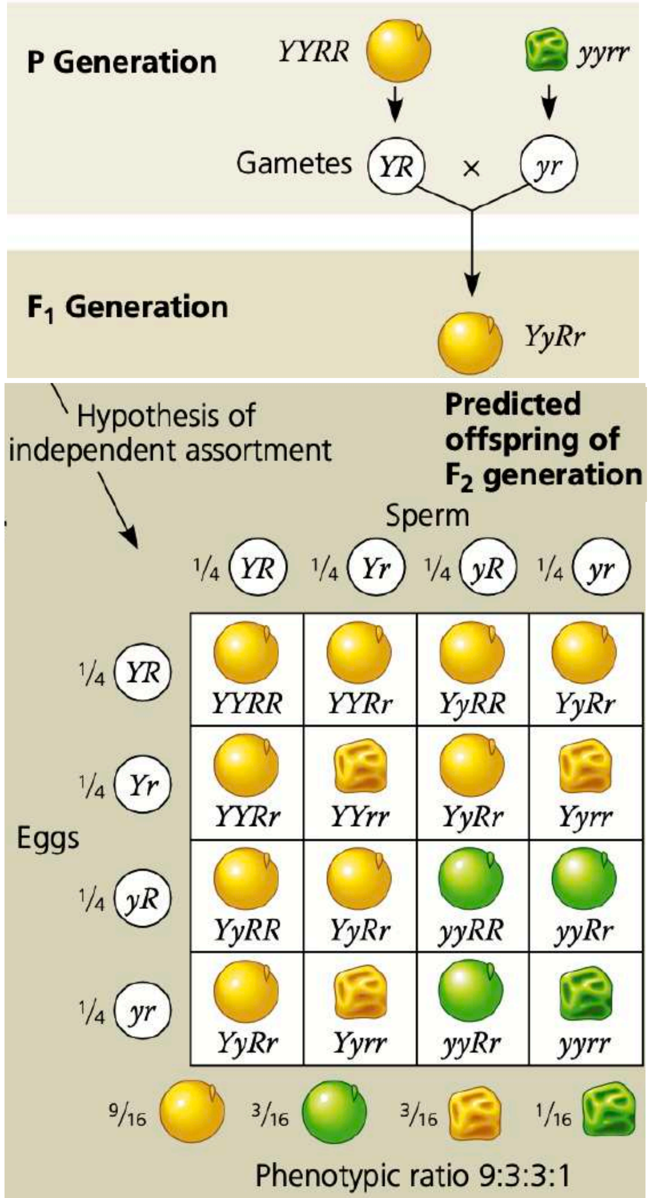
Two new traits appear in F2:
green round and yellow wrinkled

THIS IS THE RESULT MENDEL OBTAINED

Figure 14.8 and 15.2 of Campbell's Biology: a global approach

Law of Independent Assortment

Mendel crossed two true-breeding plants: yellow-round seeds x green-wrinkled seeds



Two new traits appear in F2:
green round and yellow wrinkled

This is ONLY POSSIBLE
If *Y* and *R* separated from each other
and
If *y* and *r* separated from each other
and
combined with each other

two or more genes assort independently - that is, each pair of alleles segregates independently of each other pair of alleles during gamete formation

Figure 14.8 and 15.2 of Campbell's Biology: a global approach

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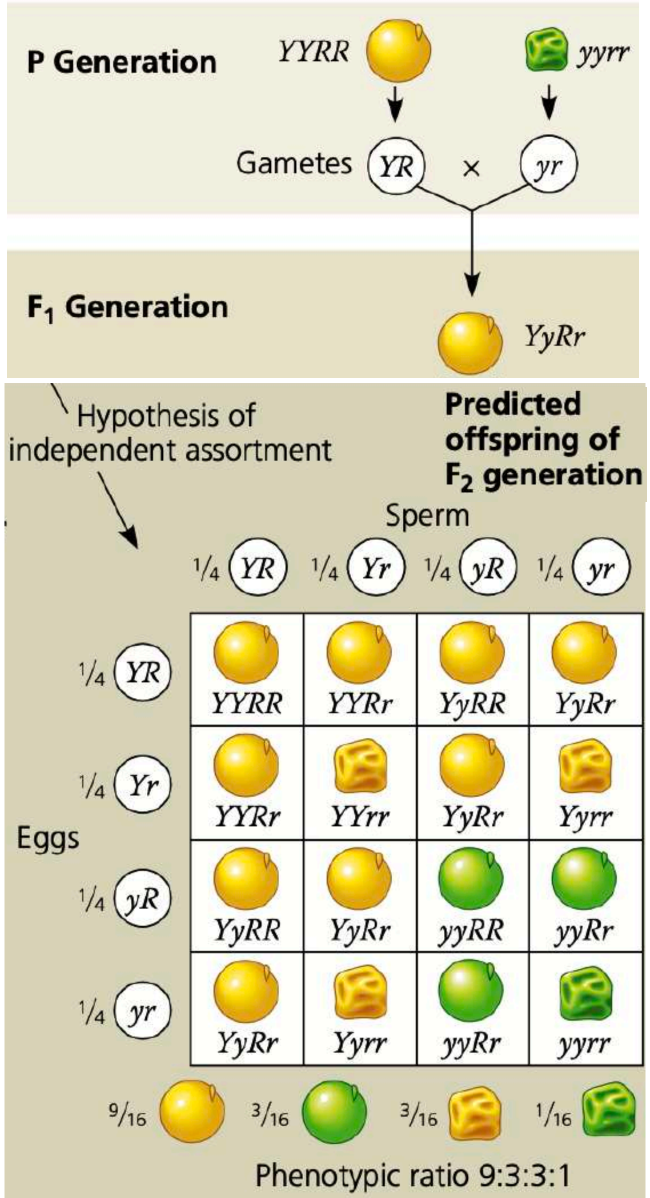
Law of Independent Assortment

Mendel crossed two true-breeding plants: yellow-round seeds x green-wrinkled seeds



two or more genes assort independently - that is, each pair of alleles segregates independently of each other pair of alleles during gamete formation

applies only to genes (allele pairs) located on non-homologous chromosomes or, alternatively, to genes that are very far apart on the same chromosome

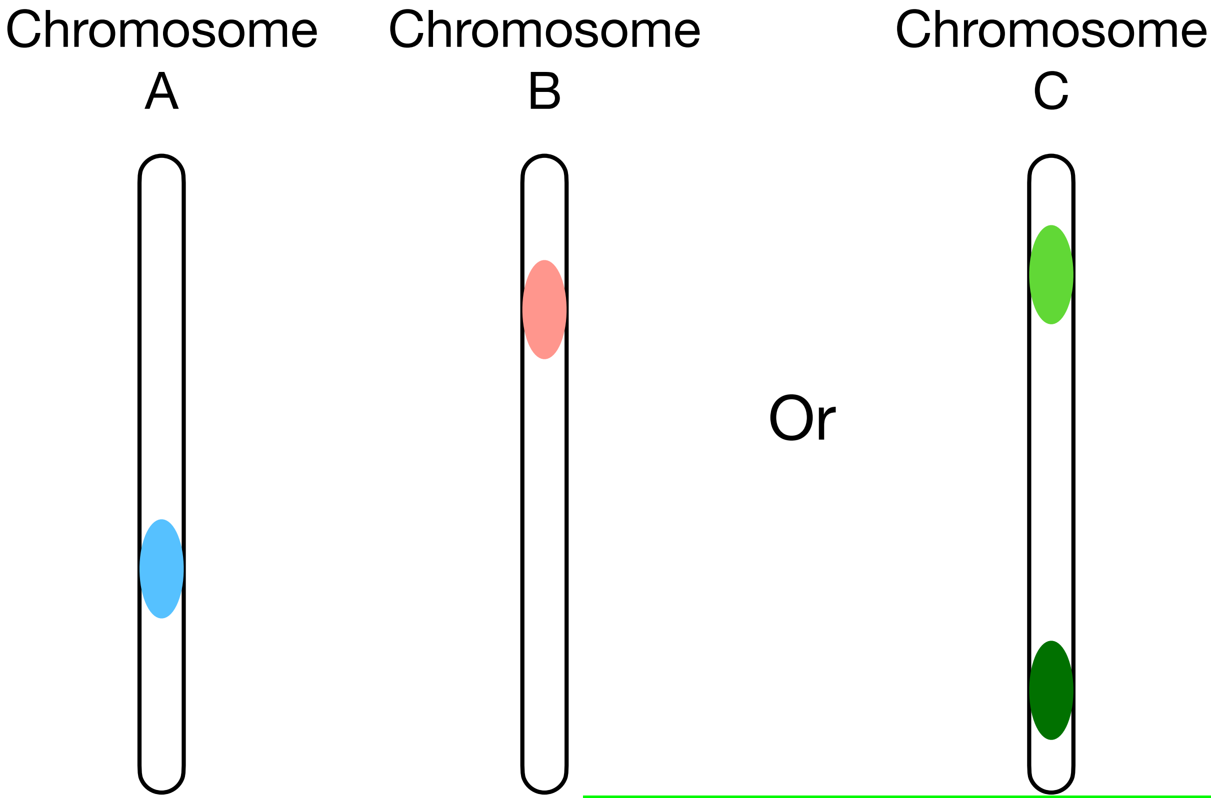
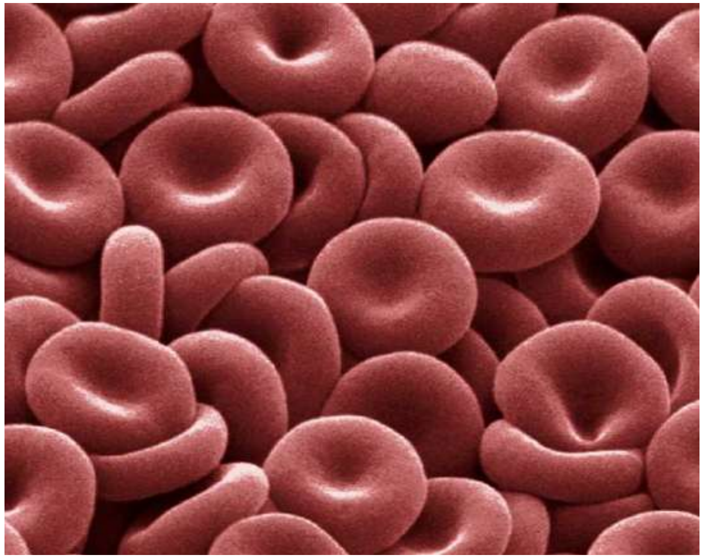
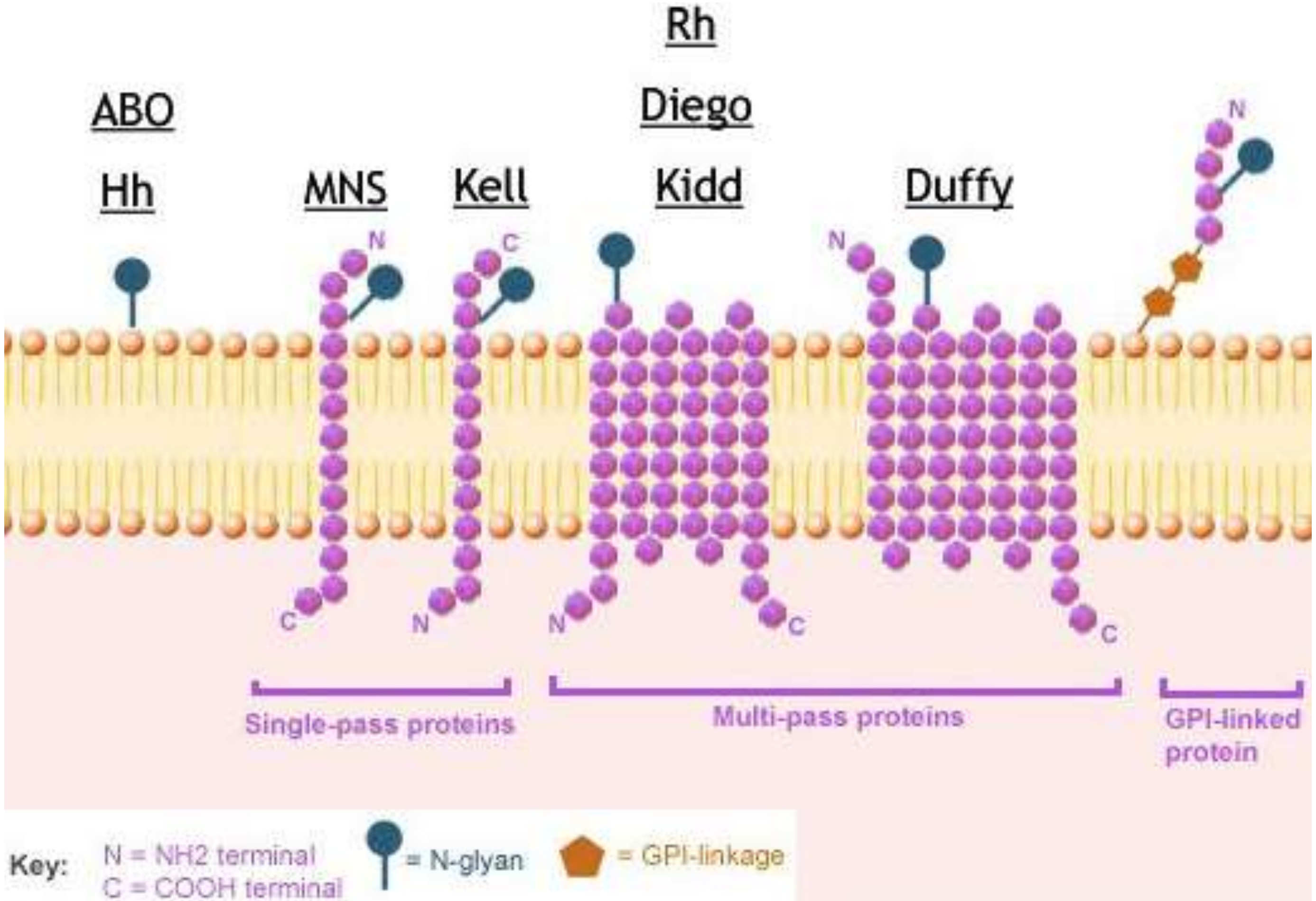


Figure 14.8 and 15.2 of Campbell's Biology: a global approach

Law of Independent Assortment

Surface of RBCs have many antigens: carbohydrates, glycoproteins, proteins etc





- One such antigen is the “A” or “B” antigen, which is a carbohydrate
- The sequence of the oligosaccharide determines if the antigen is A or B
- The oligosaccharide chain is attached to proteins and lipids in the RBC membrane
- Second such antigen is the”Rh” antigen, which is protein (49 Rh variations exist)

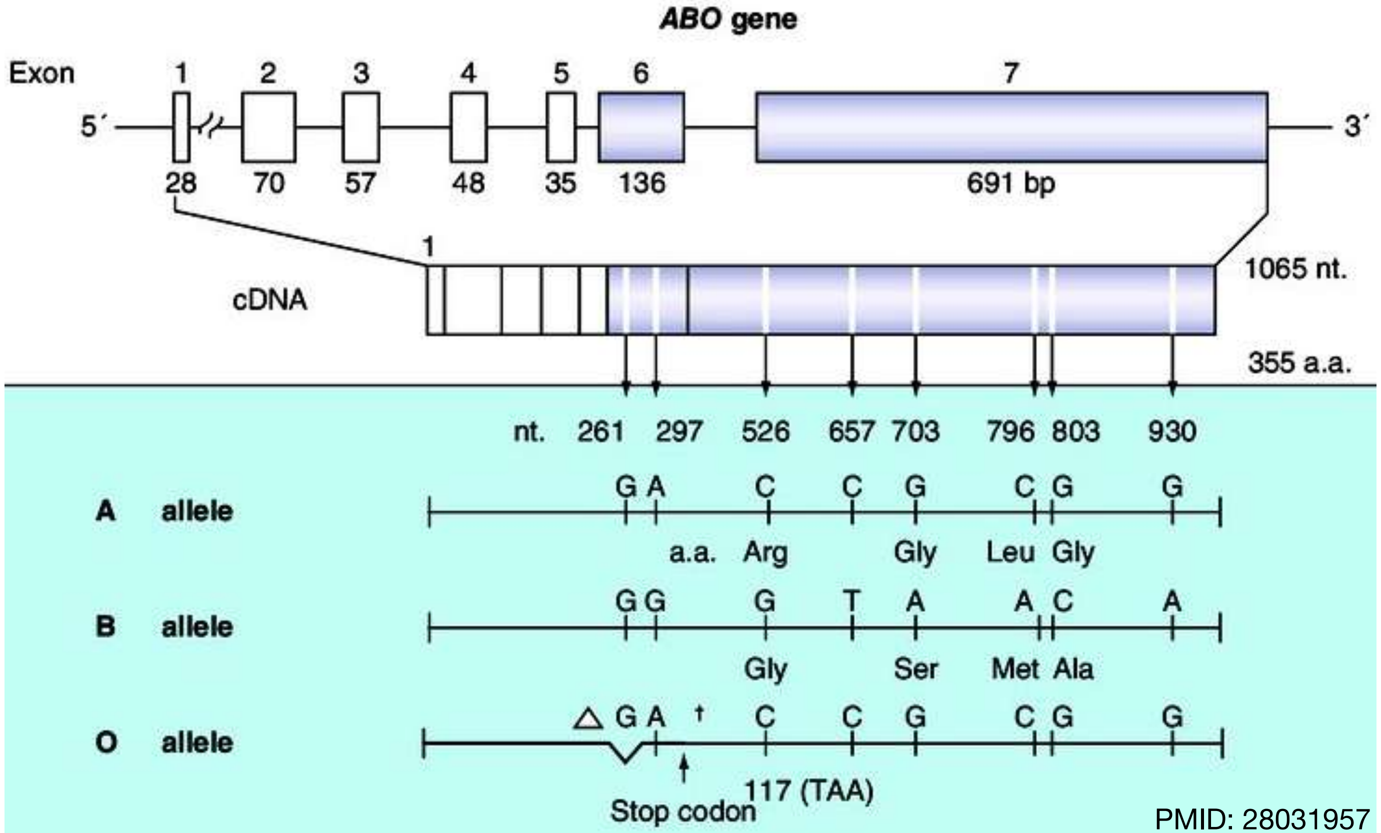
Figure 14.11 of Campbell’s Biology: a global approach
https://www.ncbi.nlm.nih.gov/books/NBK2264/

Law of Independent Assortment

The ABO gene indirectly encodes the ABO blood group antigens

- The ABO locus has three main allelic forms: A, B, and O
- The A and B alleles each encode a glycosyltransferase that converts an antigen precursor, H, into the A and B antigen
- A or B allele arises from a few changes in the DNA sequence = A and B enzymes that differ by four amino acids
- O allele has a change in the DNA sequence which introduces a stop codon = inactive enzyme which cannot convert the precursor, H, into the A and B antigen

A allele = dominant
B allele = also dominant
O allele = recessive



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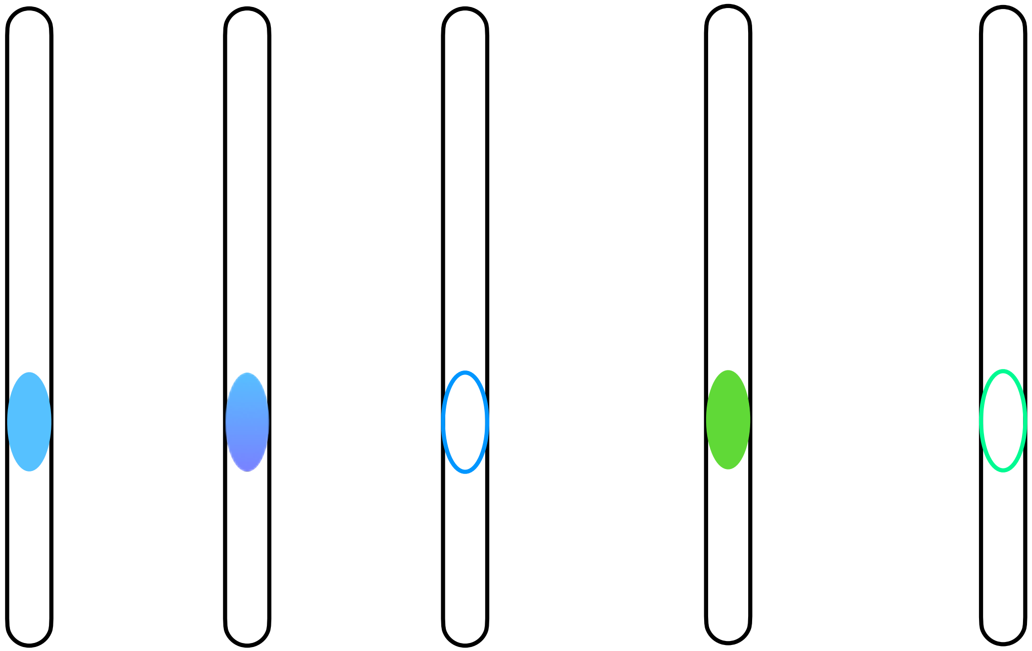
Law of Independent Assortment

Chromosome 9

Allele A Allele B Allele C

Chromosome 1

Allele Rh+ Allele Rh-



A allele = dominant
B allele = also dominant
O allele = recessive

Rh+ allele = dominant
Rh- allele = recessive

Possible diploid genotype combinations?

AARh+Rh+	AARh-Rh-	AARh+Rh-
ABRh+Rh+	ABRh-Rh-	ABRh+Rh-
BBRh+Rh+	BBRh-Rh-	BBRh+Rh-
AORh+Rh+	AORh-Rh-	AORh+Rh-
BORh+Rh+	BORh-Rh-	BORh+Rh-
OORh+Rh+	OORh-Rh-	OORh+Rh-

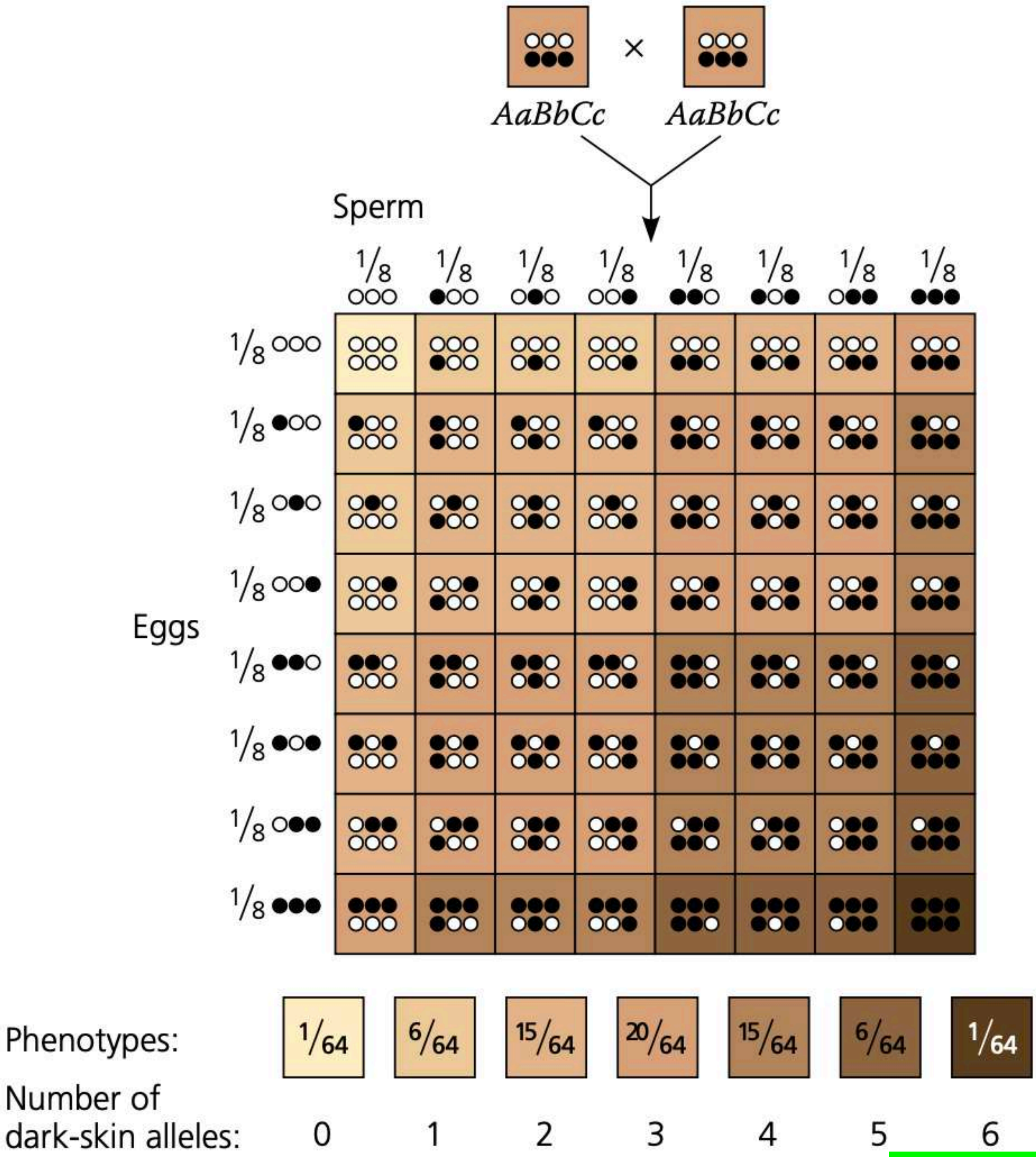
AB/BA, AO/OA and BO/OB considered as one each above

Which of the genotypes above would be similar to the P generation yellow round x green wrinkled cross that Mendel did?

Only one chromatid shown in diagrams above to show what is possible in a gamete

Relationship among alleles of a single gene	Description	Example
Complete dominance of one allele	Heterozygous phenotype same as that of homo-zygous dominant	PP  Pp 
Incomplete dominance of either allele	Heterozygous phenotype intermediate between the two homozygous phenotypes	 C^RC^R C^RC^W C^WC^W
Codominance	Both phenotypes expressed in heterozygotes	I^AI^B 
Multiple alleles	In the population, some genes have more than two alleles	ABO blood group alleles I^A, I^B, i
Pleiotropy	One gene affects multiple phenotypic characters	Sickle-cell disease

What about inheritance of 3 genes or alleles?



Chapter 14.3 of Campbell's Biology: a global approach