Mendel's law of segregation and independent assortment

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Mendel's experiment

Reasons for failure of predecessors

- Scientists studied plant as a whole, i.e. in its totality of appearance of a large number of characters. Therefore plants couldn't be classified in few clearcut classes.
- Scientists were concerned with the description of various forms appearing in progeny. Attempt on highlighting class frequencies of progenies was not made.
- Data from several generations were not kept separately and accurately.
- Complete control of pollination in F_1 was lacking
- In many studies, F_1 was interspecific hybrid exhibiting partial to complete sterility.
- Number of plants maintained and studied in F_2 were relatively small.
- Most characters studied were of quantitative nature.

Suitability of pea

- Pea is self pollinating crop, so pollination control was not a problem.
- Pea is easy to cross as the flowers are relatively large.
- Pea plant was easy to cultivate and only took a season to raise a single generation
- Pea seeds are large and have good germination in general. Plants also take little space to grow.
- Pea varieties had many sharply defined inherited differences

Resons for success of Mendel

- Analysis of failure of previous experimenters
- Mendel studied the inheritance of only one pair of contrasting characters at a time.
- Mendel selected pea varieties that had clearly different forms of one or more characters.
- For a given contrasting character, Mendel kept an accurate record of number of plants in each category for every generation.
- Carried out experiment with great care and elaborateness.
- Had a sound knowledge of mathematics
- Mendel was able to formulate appropriate hypotheses on the basis of explaination he offered for his experimental findings. Also, carried out the hypothesis testing to see the its correctness by experiment.
- Mendel was lucky

Law of segregation

The law

- Particulate inheritance (non contamination of an allele of a gene by other), require genes to be like solid particles.
- As a result, two alleles present in the F_1 are able to separate and pass into different gametes in their original form producing two different types of gametes in equal frequencies; this separation of alleles is known as **segregation**.
- Two alleles of a gene remain separate and do not contaminate each other in the F_1 or the hybrid. At the time of gamete formation in F_1, the two alleles separate and pass into different gametes.
- This law is also known as **law of purity of gametes**.

Mendel's work

- When two different varieties were crossed for each character, mendel uniformly found that F_1 was of only one type.
- However, when these F_1 plants were permitted to self fertilized and reproduce, both original varieties now appeared in the F_2.
- The smooth F_1 seeds, i.e., plants from cross of smooth x wrinkled, produced upon self-fertilization and F_2 of 5474 smooth seeds and 1850 wrinkled.
- The self fertilized yellow F_1 produced from the cross between yellow and green seeds produced 6022 yellow and 2001 green seeds.

Mendel's results

- For all seven characters tested, the results appeared to fit the following pattern
  1. For any character the F_1 derived from crosses between two different varieties showed only one of the traits and never the other.
  2. The trait that had disappeared or been "hidden" in the F_1 reappeared in the F_2, but only in a frequency of one-quarter that of the total number
  3. It did not matter which parent variety provided the pollen and which the ova; **reciprocal crosses** in which each of the two varieties was used to provide male and female parents (e.g., male A x female B, female A x male B) always gave the same results. Thus, there was no indication that any trait Mendel observed was transmitted only through "paternal" or "maternal" inheritance.

Behaviour of factor

- Mendel called the determining agent responsible for each trait a “factor”.
- The phenomena by which one trait appears and the other does not, even though the factors for both are present is called **dominance**.
- In contrast to theory for blended inheritance, which considered all traits as becoming “blended” and diluted in hybrid offspring, Mendel showed that the factor determining each trait, whether dominant or recessive, did not change throughout the several generations of mating.

Mendel's pea color observation

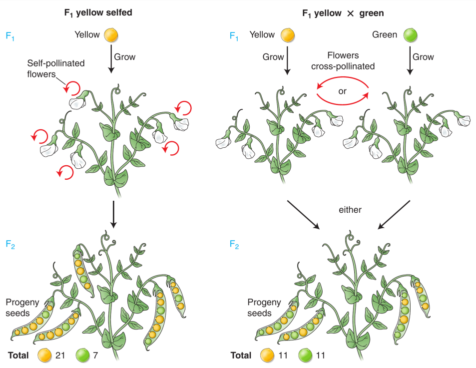


Figure 1: Mendel obtained a 3:1 phenotypic ratio in his self-pollination of the F_1, on left, and a 1:1 phenotypic ratio in his cross of F_1 yellow with green, on right. Sample sizes are arbitrary.

Mendel's interpretation and its translation

1. A hereditary factor called a **gene** is necessary for producing pea color.
2. Each plant has a pair of this type of gene.
3. The gene comes in two forms called **alleles**. For a gene Y, the two alleles can be represented by Y (standing for the yellow phenotype) and y (standing for the green phenotype).
4. A plant can be either Y/Y, y/y, or Y/y. The slash shows that the alleles are a pair.
5. In the Y/y plant, the Y allele dominates, and so the phenotype will be yellow. Hence, the phenotype of the Y/y plant defines the Y allele as dominant and the y allele as recessive.
6. In meiosis, the members of a gene pair separate equally into the cells that become eggs and sperm, the gametes. This equal separation has become known as Mendel's first law or as the law of equal segregation. Hence, a single gamete contains only one member of the gene pair.
7. At fertilization, gametes fuse randomly, regardless of which of the alleles they bear.

Mendel's exact observation

Parental phenotypes	F_1	F_2	F_2 ratio
1. round × wrinkled seeds	All round	5474 round; 1850 wrinkled	2.96 : 1
2. yellow × green seeds	All yellow	6022 yellow; 2001 green	3.01 : 1
3. purple × white petals	All purple	705 purple; 224 white	3.15 : 1
4. inflated × pinched pods	All inflated	882 inflated; 299 pinched	2.95 : 1
5. green × yellow pods	All green	428 green; 152 yellow	2.82 : 1
6. axial × terminal flowers	All axial	651 axial; 207 terminal	3.14 : 1
7. long × short stems	All long	787 long; 277 short	2.84 : 1

Figure 2: Results of All Mendel's Crosses in Which Parents Differed in One Character

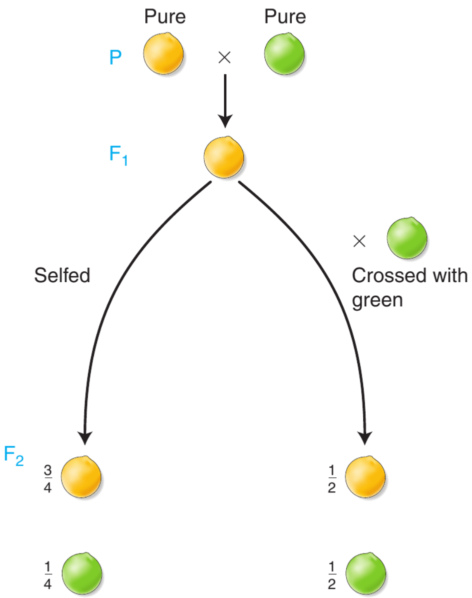
Terminologies and meanings

- A fertilized egg, the first cell that develops into a progeny individual, is called a **zygote**.
- A plant with a pair of identical alleles of the par differ is called a **heterozygote**.
- Sometimes a heterozygote for one gene is called a **monohybrid**.
- An individual can be classified as either **homozygous dominant** (such as Y/Y), **heterozygous** (Y/y), or **homozygous recessive** (y/y).
- In genetics generally, allelic combinations underlying phenotypes are called genotypes. Hence, Y/Y, Y/y, and y/y are all genotypes.
- When crossed with each other, the Y/Y and the y/y lines produce an F_1 generation composed of all heterozygous individuals (Y/y). Because Y is dominant, all F_1 individuals are yellow in phenotype. Selfing the F_1 individuals can be thought of as a cross of the type Y/y x Y/y, which is sometimes called a monohybrid cross.

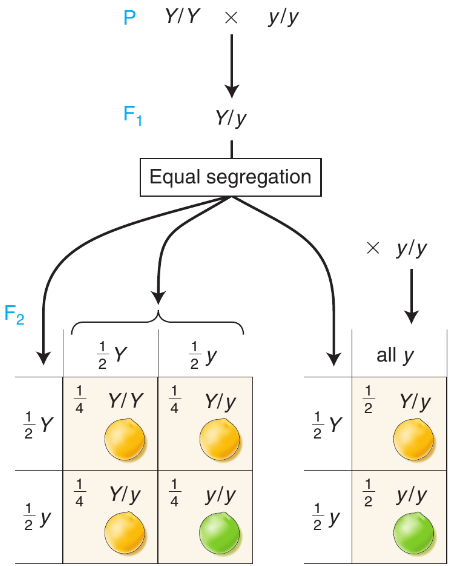
Monohybrid crossing

- Equal segregation of the Y and y alleles in the heterozygous F_1 results in gametes, both male and female, half of which are Y and half of which are y.
- The general depiction of an individual expressing the dominant allele is $Y/\_$.
- Eequal segregation is detectable only in the meiosis of a heterozygote. Hence, Y/y produces one-half Y gametes and one-half y gametes. Although equal segregation is taking place in homozygotes too, neither segregation $\frac{1}{2}Y : \frac{1}{2}Y$ nor segregation $\frac{1}{2}y : \frac{1}{2}y$ is meaningful or detectable at the genetic level.
- equal segregation in the yellow heterozygous F_1 gives gametes with a $\frac{1}{2}Y : \frac{1}{2}y$ ratio.
- The y/y parent can make only y gametes, however; so the phenotype of the progeny depends only on which allele they inherit from the Y/y parent.
- Thus, the $\frac{1}{2}Y : \frac{1}{2}y$ **gametic ratio** from the heterozygote is converted into a $\frac{1}{2}Y/y : \frac{1}{2}y/y$ **genotypic ratio**, which corresponds to 1:1 **phenotypic ratio**.

Mendel's results



Mendel's explanation

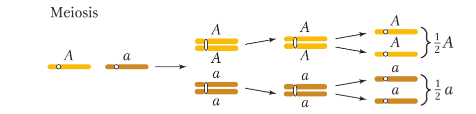


Examples of single gene inheritance

- Segregation for waxy gene in Maize. (Waxiness is a recessive trait)
- Segregation for spore color in Neurospora. (Albino color is a recessive trait, while Black is dominant)

Chromosomal basis of single gene inheritance

- The behavior of chromosomes during meiosis clearly explains Mendel's law of equal segregation.
- Consider a heterozygote of general type A/a. We can simply follow the preceding summary while considering what happens to the alleles of this gene:
  - Start: one homolog carries A and one carries a
  - Replication: one dyad is AA and one is aa
  - Pairing: tetrad is $A/A/a/a$
  - First-division products: one cell AA, the other cell aa (crossing over can mix these types of products up, but the overall ratio is not changed)
  - Second-division products: four cells, two of type A and two of type a.
  - Hence, the products of meiosis from a heterozygous meiocyte A/a are $\frac{1}{2}A$ and $\frac{1}{2}a$, precisely the equal ratio that is needed to explain Mendel's first law.



Law of independent assortment

The law

- The segreation of two or more characters in the same hybrid is independent of each other. Thus any allele of one gene is equally likely to combine with any allele of the other gene and pass into the same gamete.
- Independent segregation of two genes produce four different types of gametes in equal proportion.
- A random union among the gametes gives rise to 16 possible zygotes. These zygotes yield 9:3:3:1 phenotypic ratio, which is known as the typical dihybrid ratio.

Mendel's work

- Mendel began to work with a pair of characters – seed shape and seed color.
- Monohybrid cross for seed color ($Y/y \times Y/y$) gives a progeny ratio of 3 yellow : 1 green. The monohybrid cross for seed shape ($R/r \times R/r$) gives a progeny ratio of 3 round : 1 wrinkled.
- To perform a dihybrid cross, Mendel started with two pure parental lines. One line had wrinkled, yellow seeds. Because Mendel had no concept of the chromosomal location of genes, we must use the dot representation to write the combined genotype initially as $r/r.Y/Y$. The other line had round, green seeds, with genotype $R/R.y/y$.
- When these two lines were crossed, they must have produced gametes that were $r.Y$ and $R.y$, respectively.
- Hence, the F_1 seeds had to be dihybrid, of genotype $R/r.Y/y$. Mendel discovered that the F_1 seeds were round and yellow. This result showed that the dominance of R over r and Y over y was unaffected by the condition of the other gene pair in the $R/r.Y/y$ dihybrid. In other words, R remained dominant over r, regardless of seed color, and Y remained dominant over y, regardless of seed shape.

Mendel's dihybrid cross

- Mendel then selfed the dihybrid to obtain the F_2 generation.

$$\frac{9}{16}\ \text{round, yellow}$$
$$\frac{3}{16}\ \text{round, green}$$
$$\frac{3}{16}\ \text{wrinkled, yellow}$$
$$\frac{1}{16}\ \text{wrinkled, green}$$

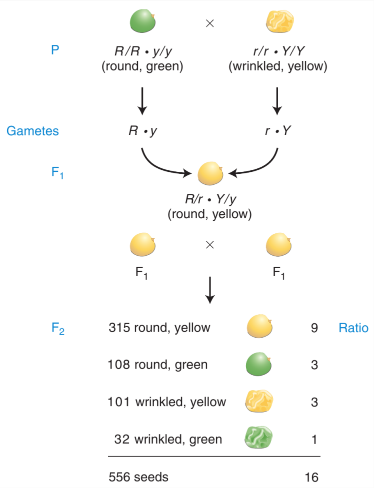
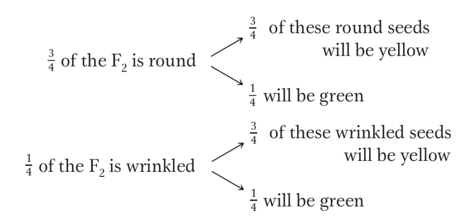


Figure 4: Mendel's dihybrid cross in pea

Dihybrid cross is a result of two independent monhybrid cross events combined

- Mendel verified if the monohybrid 3:1 ratios can still be found in the F_2.
- In regard to seed shape, there are 423 round seeds (315 + 108) and 133 wrinkled seeds (101 + 32).
- In regard to seed color, there are 416 yellow seeds (315 + 101) and 140 green seeds (108 + 32).
- Calculate the monohybrid ratios.
- This led to Mendel's insight on 9:3:3:1 ratio, because he realized that it was simply two different ratios combined at random.



Mendel's conclusion

- Different gene pairs assort independently during gamete formation.
- The consequence is that, for two heterozygous gene pairs A/a and B/b, the b allele is just as likely to end up in a gamete with an a allele as with an A allele, and likewise for the B allele.

Genetic ratios

- Probability of obtaining gametes with composition of both alleles:

$$\frac{1}{4} R/\_; Y/\_ \longrightarrow \text{round, yellow}$$

$$\frac{1}{4} R/\_; y/y \longrightarrow \text{round, green}$$

$$\frac{1}{4} r/r; Y/\_ \longrightarrow \text{wrinkled, yellow}$$

$$\frac{1}{4} r/r; y/y \longrightarrow \text{wrinkled, green}$$

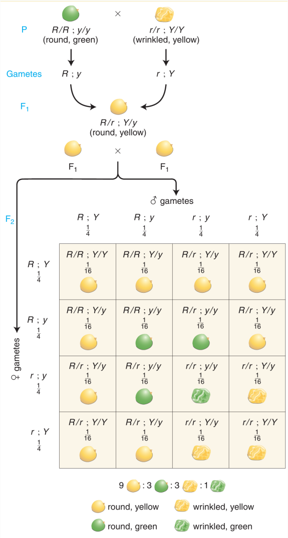


Figure 5: We can use a Punnett square to predict the result of a dihybrid cross. This Punnett square shows the predicted genotypic and phenotypic constitution of the F_2 generation from a dihybrid cross.

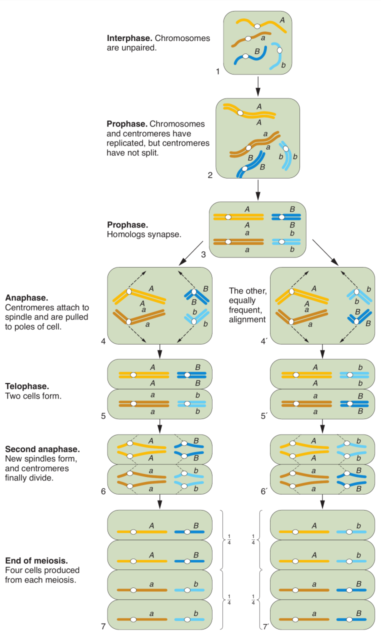


Figure 6: Chromosomal basis of independent assortment
Meiosis in a diploid cell of genotype A/a;B/b. The diagram shows how the segregation and assortment of different chromosome pairs give rise to the 1:1:1:1 Mendelian gametic ratio.

Gamete types	abc	abC	aBc	aBC	Abc	AbC	ABc	ABC
abc	abcabc	abcabC	abcaBc	abcaBC	abcAbc	abcAbC	abcABc	abcABC
abC	abCabc	abCabC	abCaBc	abCaBC	abCAbc	abCAbC	abCABc	abCABC
aBc	aBcabc	aBcabC	aBcaBc	aBcaBC	aBcAbc	aBcAbC	aBcABc	aBcABC
aBC	aBCabc	aBCabC	aBCaBc	aBCaBC	aBCAbc	aBCAbC	aBCABc	aBCABC
Abc	Abcabc	AbcabC	AbcaBc	AbcaBC	AbcAbc	AbcAbC	AbcABc	AbcABC
AbC	AbCabc	AbCabC	AbCaBc	AbCaBC	AbCAbc	AbCAbC	AbCABc	AbCABC
ABc	ABcabc	ABcabC	ABcaBc	ABcaBC	ABcAbc	ABcAbC	ABcABc	ABcABC
ABC	ABCabc	ABCabC	ABCaBc	ABCaBC	ABCAbc	ABCAbC	ABCABc	ABCABC

Sex linked genes

Sex linked single gene inheritance patterns

- In sex chromosomes too, the segregation of chromosomes is equal, but the phenotypic ratios seen in progeny are often different from the autosomal ratios.
- Most animals and many plants show sexual dimorphism.
- In most cases, sex is determined by a special pair of sex chromosomes.
- Females have identical pair, males no not.
    - Females (in humans) is homogametic sex
    - Males (in humans) is heterogametic sex

$$(\female)\text{females} = 44\text{A} + \text{XX}$$
$$(\male)\text{males} = 44\text{A} + \text{XY}$$

- Vascular plants may be dioecious or monoecious in sexual orientation
- Some, but not all, dioecious plants have a nonidentical pair of chromosomes associated with (and almost certainly determining) the sex of the plant.
- X and Y chromosomes can be divided into homologous and differential regions.

Solved problems

Problem 1: Monohybrid cross and dominance relation

- While performing a large scale grow out experiment of pea germplasm, a plant with three cotyledons was discovered (normally, there are 2 cotyledons). This plant was crossed with a normal pure-breeding wild-type plant, and 600 seeds from this cross were planted. There were 298 plants with two cotyledons and 302 with three cotyledons. What can be said about the inheritance of three cotyledons ? Hint: Invert gene symbols as part of your explaination.

Solution 1: Monohybrid cross and dominance relation

- The fact that about half of the F_1 progeny are mutant suggests that the mutation that results in three cotyledons in dominant and the original mutant heterozygous. If C = the mutant allele and c = the wild-type allele, the cross is as follows:

$$\begin{array}{ll} P & C/c \times c/c \\ F_1 & 1\ C/c : 1\ c/c \text{ (three cotyledons: two cotyledons)} \end{array}$$

Problem 2: Sex linked inheritance

- A sex-linked recessive allele c produces a red-green color blindness in humans. A normal woman whose father was color blind marries a color-blind man.
  a. What genotypes are possible for the mother of the color blind man ?
  b. What are the chances that the first child from this marriage will be a color-blind boy ?
  c. Of the girls produced by these parents, what proportion can be expected to be color blind ?
  d. Of all the children (sex unspecified) of these parents, what proportion can be expected to have normal color vision ?

Solution 2: Sex linked inheritance

a. $X^C/X^c, X^c/X^c$
b. p (color-blind) x p (male) $= (1/2)(1/2) = 1/4$
c. The girls will be 1 normal (X^C/X^c): 1 color-blind (X^c/X^c).
d. The cross is $X^C/X^c \times X^c/Y$, yielding 1 normal:1 color-blind for both sexes.

Problem 3: Dominance and parental genotype determination

- In tomatoes, one gene determines whether the plant has purple (P) or green (G) stems, and a separate, independent gene determines whether the leaves are "cut" (C) or "potato" (Po). Five matings of tomato-plant phenotypes give the following results.
  a. Which alleles are dominant ?
  b. What are the most probable genotypes for the parents in each cross ?

Mating	Parental phenotypes	*Number of progeny*			
		P, C	P, Po	G, C	G, Po
1	P, C × G, C	323	102	309	106
2	P, C × P, Po	220	206	65	72
3	P, C × G, C	723	229	0	0
4	P, C × G, Po	405	0	389	0
5	P, Po × G, C	71	90	85	78

Solution 3: Dominance and parental genotype determination

- Cross 2 indicates that purple (G) is dominant over green (g), and cross 1 indicates that cut (P) is dominant over potato (p).

Cross 1: G/g; P/p x g/g; P/p	There are 3 cut:1 potato, and 1 purple: 1 green
Cross 2: G/g; P/p x G/g; p/p	There are 3 purple: 1 green, and 1 cut: 1 potato
Cross 3: G/G; P/p x g/g; P/p	There is no green, and there are 3 cut: 1 potato
Cross 4: G/g; P/P x g/g; p/p	There is no potato, and there is 1 purple: 2
Cross 5: G/g; p/p x g/g; P/p	There is 1 cut: 1 potato, and there is 1 purple: 1 green

Problem 4: Effective crossing

- A corn geneticist has three pure lines of genotypes a/a; B/B; C/C, A/A; b/b; C/C, and A/A; B/B; c/c. All the phenotypes determined by a, b and c will increase the market value of the corn; so, naturally, he wants to combine them all in one pure line of genotype a/a; b/b; c/c.
    1. Outline an effective crossing program that can be used to obtain the a/a; b/b; c/c pure line.
    2. At each stage, state exactly which phenotypes will be selected and give their expected frequencies.
    3. Is there more than one way to obtain the desired genotype ? Which is the best way ? (Assume independent assortment of the three gene pairs.)

Solution 4: Effective crossing

1. Begin with any two of the three lines and cross them. If, for example, you began with a/a; B/B; C/C x A/A; b/b; C/C, all the progeny would be A/a; B/b; C/C. Crossing two of them would yield:

$$
\begin{array}{rr}
9 & A/\_; B/\_; C/C \\
9 & a/a; B/\_; C/C \\
9 & A/\_; b/b; C/C \\
9 & a/a; b/b; C/C
\end{array}
$$

- The a/a; b/b; C/C genotype has two of the gens in a homozygous recessive state and is found in 1/16 of the offspring.

2. If that phenotype were crossed with A/A; B/B; c/c, all the progeny would be heterozygous for all loci. Crossing two (selfing) of thus obtained individuals would lead to a 27:9:9:9:3:3:3:1 ratio and 1/64 of the progeny would be the desirable a/a; b/b; c/c.
3. There are several different routes to obtaining a/a; b/b; c/c, but the one just outlined only requires four crosses.

Problem 5: Maternal inheritance and gene assortment

- A man is brachydactylous (very short fingers; rare autosomal dominant), and his wife is not. Both can taste the chemical phenylthiocarbamide (autosomal dominant; common allele), but their mothers could not.

    a. Give the genotypes of the couple. If the genes assort independently and the couple has four children, what is the probability of
    b. all of them being brachydactylous?
    c. none being brachydactylous?
    d. all of them being tasters?
    e. all of them being nontasters?
    f. all of them being brachydactylous tasters?
    g. none being brachydactylous tasters?
    h. at least one being a brachydactylous taster?

Solution 5: Maternal inheritance and gene assortment

a. Let B = brachydactylous, b = normal, T = taster, and t = non-taster. The genotypes of the couple are B/b; T/t for the male and b/b; T/t for the female.
b. For all four children to be brachydactylous, $p = (1/2)^4 = 1/16$.
c. For none of the four children to be brachydactlyous, $p = (1/2)^4 = 1/16$.
d. For all to be tasters, $p = (3/4)^4 = 81/256$
e. For all to be nontasters, $p = (1/4)^4 = 1/256$
f. For all to be brachydactylous tasters, $p = (1/2x3/4)^4 = 81/4096$
g. The probability of not being a brachydactylous taster is 1-(the probability of being a brachydactylous taster), or 1-(1/2 x 3/4) = 5/8. The probability that all four children are not brachydactylous tasters is $(5/8)^4 = 625/4096$
h. The probability that at least one is a brachydactylous taster is 1-(probability of none being a brachydactlyous taster), or $1 - (5/8)^4$

Problem 6: Polygene doses and phenotypic classes

- Question: In polygenic systems, how many phenotypic classes corresponding to number of polygene “doses” are expected in selfs.
  a. of strains with four heterozygous polygenes ?
  b. of strains with six heterozygous polygenes ?

Solution 6: Polygene doses and phenotypic classes

a. $\longrightarrow$ There should be nine classes corresponding to 0, 1, 2, 3, 4, 5, 6, 7, 8 "doses". Table below (**??**) shows genotypic composition of individual progenies with distinct phenotypic classess indicated by color coding.

b. $\longrightarrow$ There should be 13 classes corresponding to 0, 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12 'doses'.

V1	V2	V3	V4	V5	V6	V7	V8	V9
AABBCCDD	AABbCCDD	AAbbCCDD	AaBBCCDD	AaBbCCDD	AabbCCDD	aaBBCCDD	aaBbCCDD	aabbCCDD
AABBCCDd	AABbCCDd	AAbbCCDd	AaBBCCDd	AaBbCCDd	AabbCCDd	aaBBCCDd	aaBbCCDd	aabbCCDd
AABBCCdd	AABbCCdd	AAbbCCdd	AaBBCCdd	AaBbCCdd	AabbCCdd	aaBBCCdd	aaBbCCdd	aabbCCdd
AABBCcDD	AABbCcDD	AAbbCcDD	AaBBCcDD	AaBbCcDD	AabbCcDD	aaBBCcDD	aaBbCcDD	aabbCcDD
AABBCcDd	AABbCcDd	AAbbCcDd	AaBBCcDd	AaBbCcDd	AabbCcDd	aaBBCcDd	aaBbCcDd	aabbCcDd
AABBCcdd	AABbCcdd	AAbbCcdd	AaBBCcdd	AaBbCcdd	AabbCcdd	aaBBCcdd	aaBbCcdd	aabbCcdd
AABBccDD	AABbccDD	AAbbccDD	AaBBccDD	AaBbccDD	AabbccDD	aaBBccDD	aaBbccDD	aabbccDD
AABBccDd	AABbccDd	AAbbccDd	AaBBccDd	AaBbccDd	AabbccDd	aaBBccDd	aaBbccDd	aabbccDd
AABBccdd	AABbccdd	AAbbccdd	AaBBccdd	AaBbccdd	Aabbccdd	aaBBccdd	aaBbccdd	aabbccdd

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