MENDELIAN GENETICS |

UNIT 1

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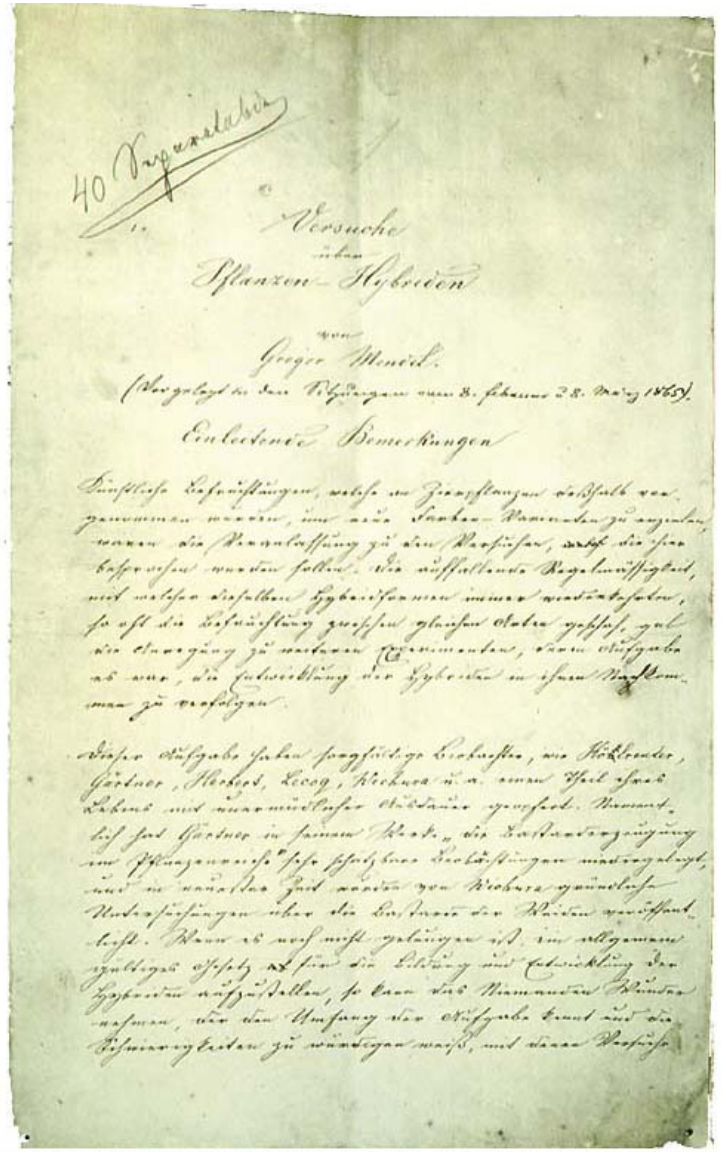
MENDEL'S EXPERIMENTS

- By self pollination he obtained seven purebred varieties (always produced offspring identical to the parentals) of peas (*Pisum sativum*).
- From 1856 to 1863 he meticulously designed and performed around 30,000 crosses.
- Then he observed how the traits were inherited.
- He applied mathematical analysis to his data.
- On February 8 and March 8, 1865, Mendel presented his results in the Brunn Society for Natural Science.

PUBLICATIONS



Versuche
über
Pflanzen-Hybriden

von
Gregor Mendel.

Einleitende Bemerkungen

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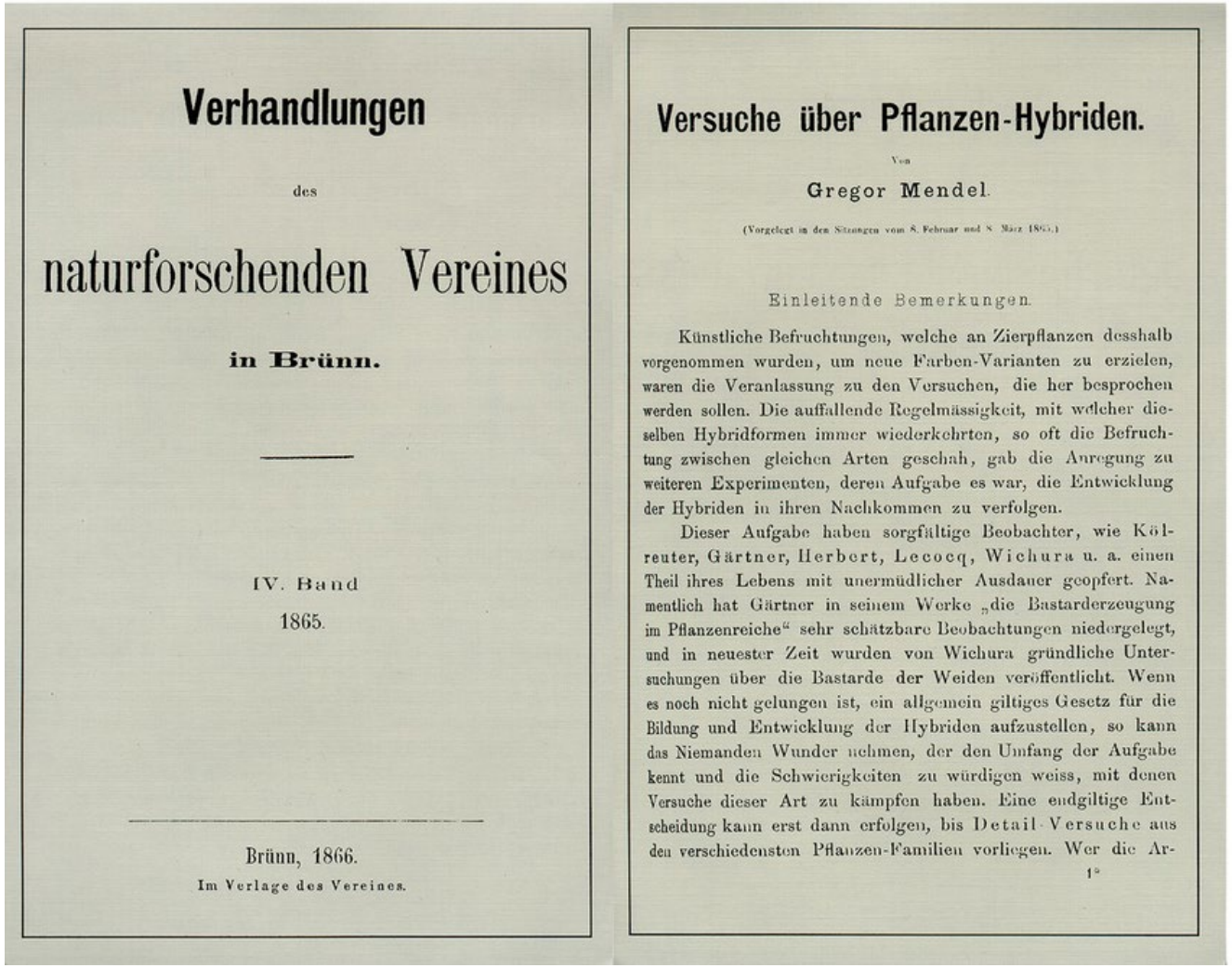


Verhandlungen

des

naturforschenden Vereines

in Brünn.

IV. Band

1865.

Brünn, 1866.

Im Verlage des Vereines.

Versuche über Pflanzen-Hybriden.

Von

Gregor Mendel.

(Vorgelegt in den Sitzungen vom 8. Februar und 8. März 1865.)

Einleitende Bemerkungen.

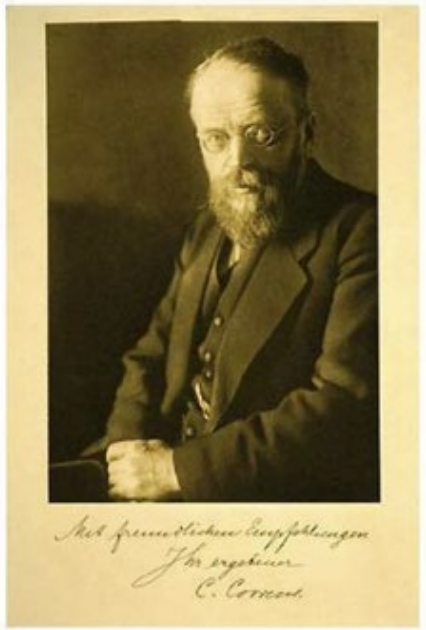
Künstliche Befruchtungen, welche an Zierpflanzen desshalb vorgenommen wurden, um neue Farben-Varianten zu erzielen, waren die Veranlassung zu den Versuchen, die her besprochen werden sollen. Die auffallende Regelmässigkeit, mit welcher dieselben Hybridformen immer wiederkehrten, so oft die Befruchtung zwischen gleichen Arten geschah, gab die Anregung zu weiteren Experimenten, deren Aufgabe es war, die Entwicklung der Hybriden in ihren Nachkommen zu verfolgen.

Dieser Aufgabe haben sorgfältige Beobachter, wie Kölreuter, Gärtner, Herbert, Lecocq, Wichura u. a. einen Theil ihres Lebens mit unermüdlicher Ausdauer geopfert. Namentlich hat Gärtner in seinem Werke „die Bastarderzeugung im Pflanzenreiche“ sehr schätzbare Beobachtungen niedergelegt, und in neuester Zeit wurden von Wichura gründliche Untersuchungen über die Bastarde der Weiden veröffentlicht. Wenn es noch nicht gelungen ist, ein allgemein giltiges Gesetz für die Bildung und Entwicklung der Hybriden aufzustellen, so kann das Niemanden Wunder nehmen, der den Umfang der Aufgabe kennt und die Schwierigkeiten zu würdigen weiss, mit denen Versuche dieser Art zu kämpfen haben. Eine endgiltige Entscheidung kann erst dann erfolgen, bis Detail-Versuche aus den verschiedensten Pflanzen-Familien vorliegen. Wer die Ar-

1*

https://www.nytimes.com/2010/06/01/science/01mendel.html

OBLIVION & REDISCOVERY



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Carl Correns

Erich von Tschermak

Hugo De Vries, around 1920.
Photo courtesy of
Cold Spring Harbor Laboratory Archives.

Hugo De Vries

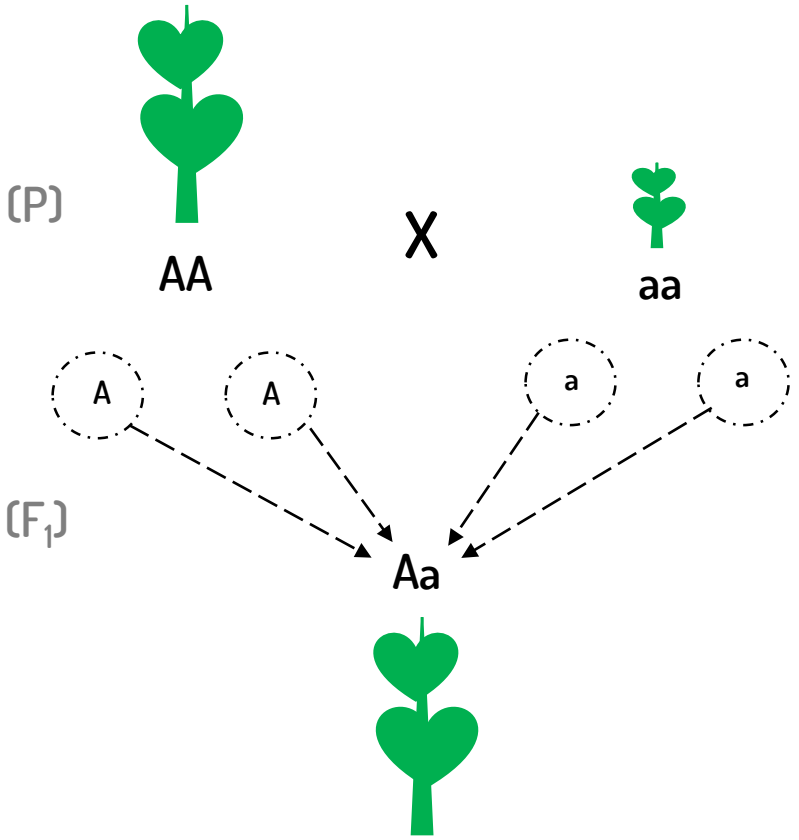
https://gregormendel200.org/

MENDELIAN TRAITS

1. To the **difference in the form of the ripe seeds**. These are either round or roundish, the depressions, if any, occur on the surface, being always only shallow; or they are irregularly angular and deeply wrinkled (P. quadratum).
2. To the **difference in the color of the seed albumen (endosperm)**. The albumen of the ripe seeds is either pale yellow, bright yellow and orange colored, or it possesses a more or less intense green tint. This difference of color is easily seen in the seeds as their coats are transparent.
3. To the **difference in the color of the seed–coat**. This is either white, with which character white flowers are constantly correlated; or it is gray, gray–brown, leather– brown, with or without violet spotting, in which case the color of the standards is violet, that of the wings purple, and the stem in the axils of the leaves is of a reddish tint. The gray seed–coats become dark brown in boiling water.
4. To the **difference in the form of the ripe pods**. These are either simply inflated, not contracted in places; or they are deeply constricted between the seeds and more or less wrinkled (P. saccharatum).
5. To the **difference in the color of the unripe pods**. They are either light to dark green, or vividly yellow, in which coloring the stalks, leaf–veins, and calyx participate.*
6. To the **difference in the position of the flowers**. They are either axial, that is, distributed along the main stem; or they are terminal, that is, bunched at the top of the stem and arranged almost in a false umbel; in this case the upper part of the stem is more or less widened in section (P. umbellatum).
7. To the **difference in the length of the stem**. The length of the stem is very various in some forms; it is, however, a constant character for each, in so far that healthy plants, grown in the same soil, are only subject to unimportant variations in this character. In experiments with this character, in order to be able to discriminate with certainty, the long axis of 6 to 7 ft. was always crossed with the short one of ¾ ft. to 1½ ft.

Source: http://www.mendelweb.org/

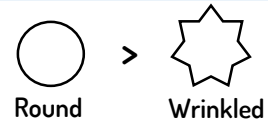
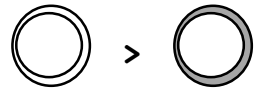
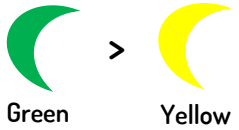
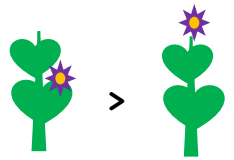
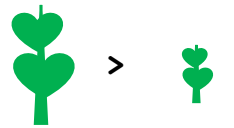
MONOHYBRID CROSSES



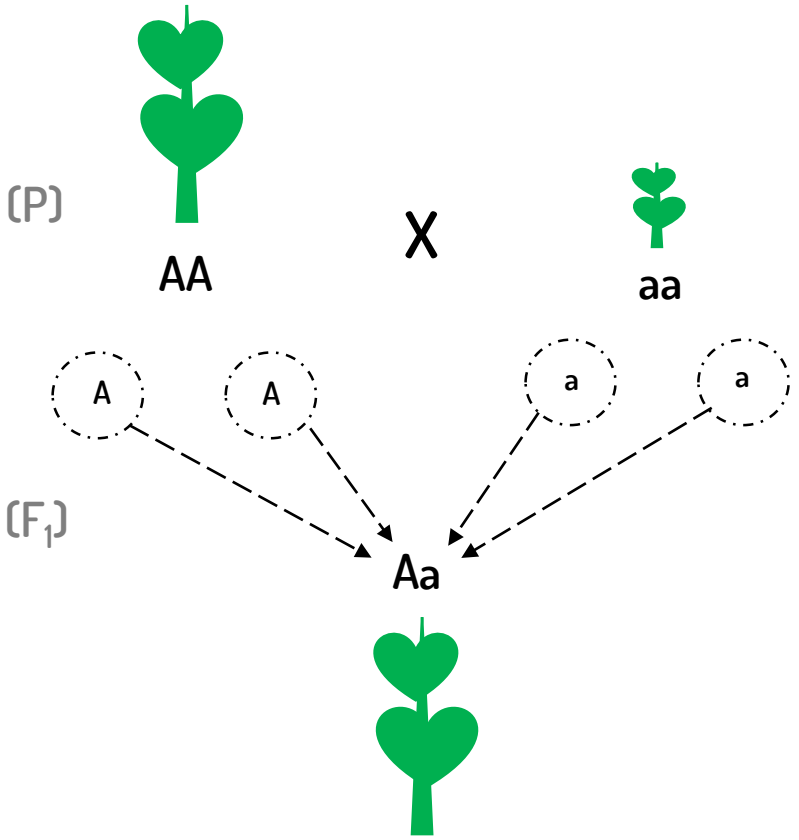
Principle of Uniformity/Dominance: *when crossing two purebreds differing in one character, the hybrids are uniform (they share a common phenotype and they resemble one of the parents –the dominant phenotype).*

Source: Gálvez Salido, A. y Navajas-Pérez, R. (2022). Una Calculadora de Genes para enseñar mendelismo, interacciones génicas y ligamiento. Didáctica de las ciencias experimentales y sociales, 42, 99-118. DOI: 10.7203/DCES.42.21008

MENDELIAN TRAITS

TRAIT	PHENOTIPIC ALTERNATIVES
Shape of the seed	 Round > Wrinkled
Color of the seed	 Yellow > Green
Color of the seed coat	 White > Grey
Color of the pod	 Green > Yellow
Shape of the pod	 Inflated > Constricted
Position of the flowers	 Axial > Terminal
Length of the stem	 Long > Short

MONOHYBRID CROSSES

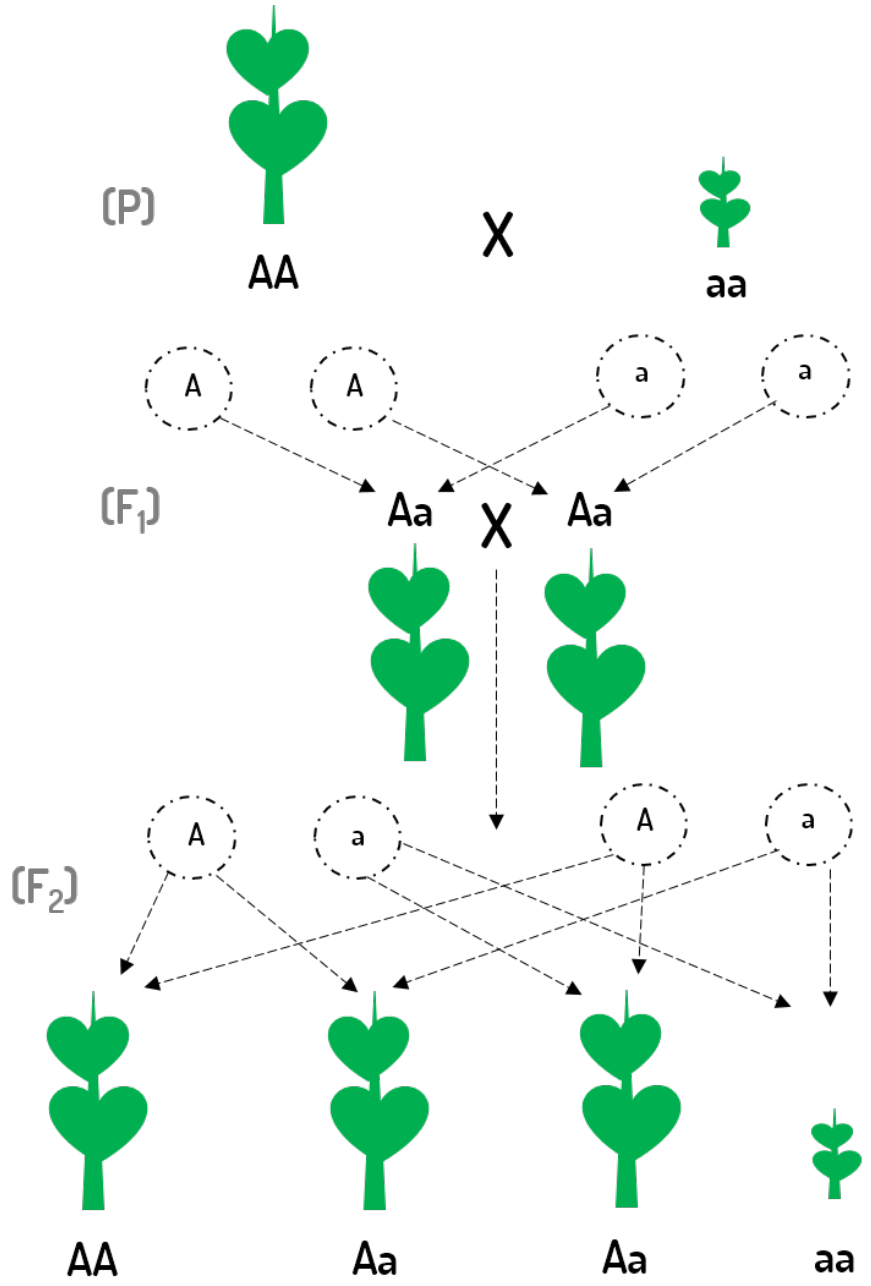


Reciprocal crosses: *the sex of the parents were interchanged so its influence in the phenotype of descendant could be analysed.*

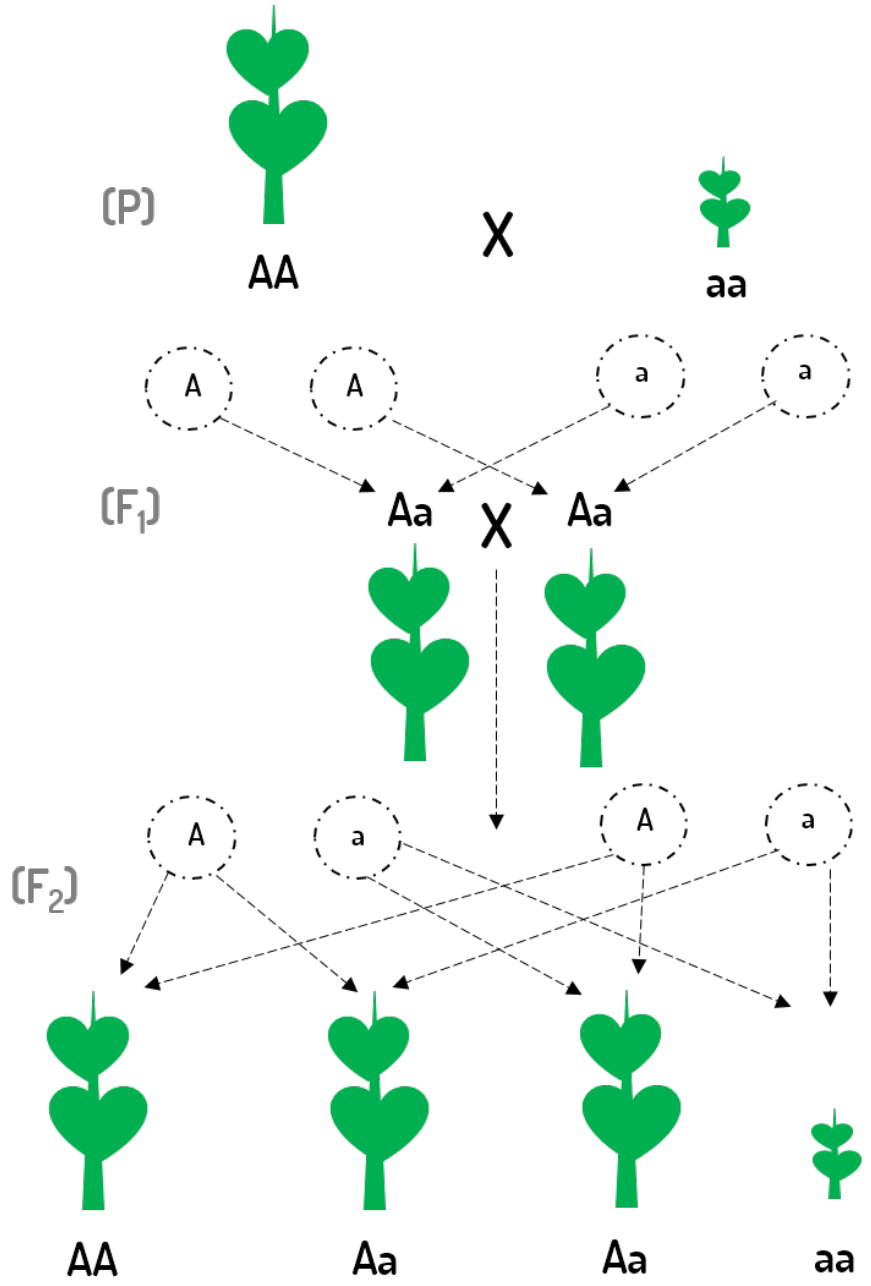
CROSSES BETWEEN F_1 HYBRIDS

1:2:1 genotypic ratio
3:1 phenotypic ratio

Law of Segregation of Alleles (mendelian factors/elements): *the genetic information of each individual for a trait is determined by two factors (now called **alleles**), that separate from each other during the formation of the gametes and rejoin after the fertilization. The gametes, therefore, will only have one allele and not two like the rest of the non-reproductive cells. The biological purpose behind this phenomenon is to keep the amount of genetic information constant in the offspring, so that when the maternal and paternal gametes unite to breed, the number of alleles (2) is maintained, and one is always inherited from each parent.*



CROSSES BETWEEN F_1 HYBRIDS



Test-cross/Back-cross: a cross between an individual showing the dominant phenotype and a individual showing the recessive one. It is used to determine if the dominant individual contain two copies of the dominant allele (homozygous dominant) or just one copy (heterozygous dominant).

Inheritance is particulated

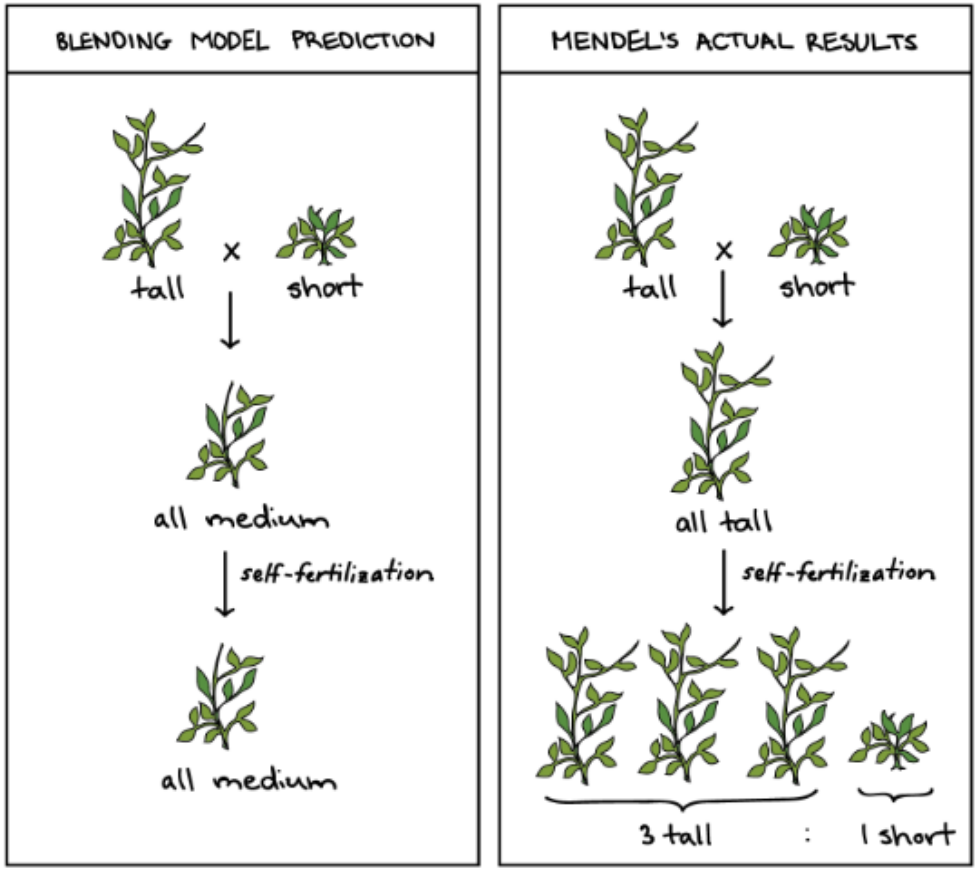
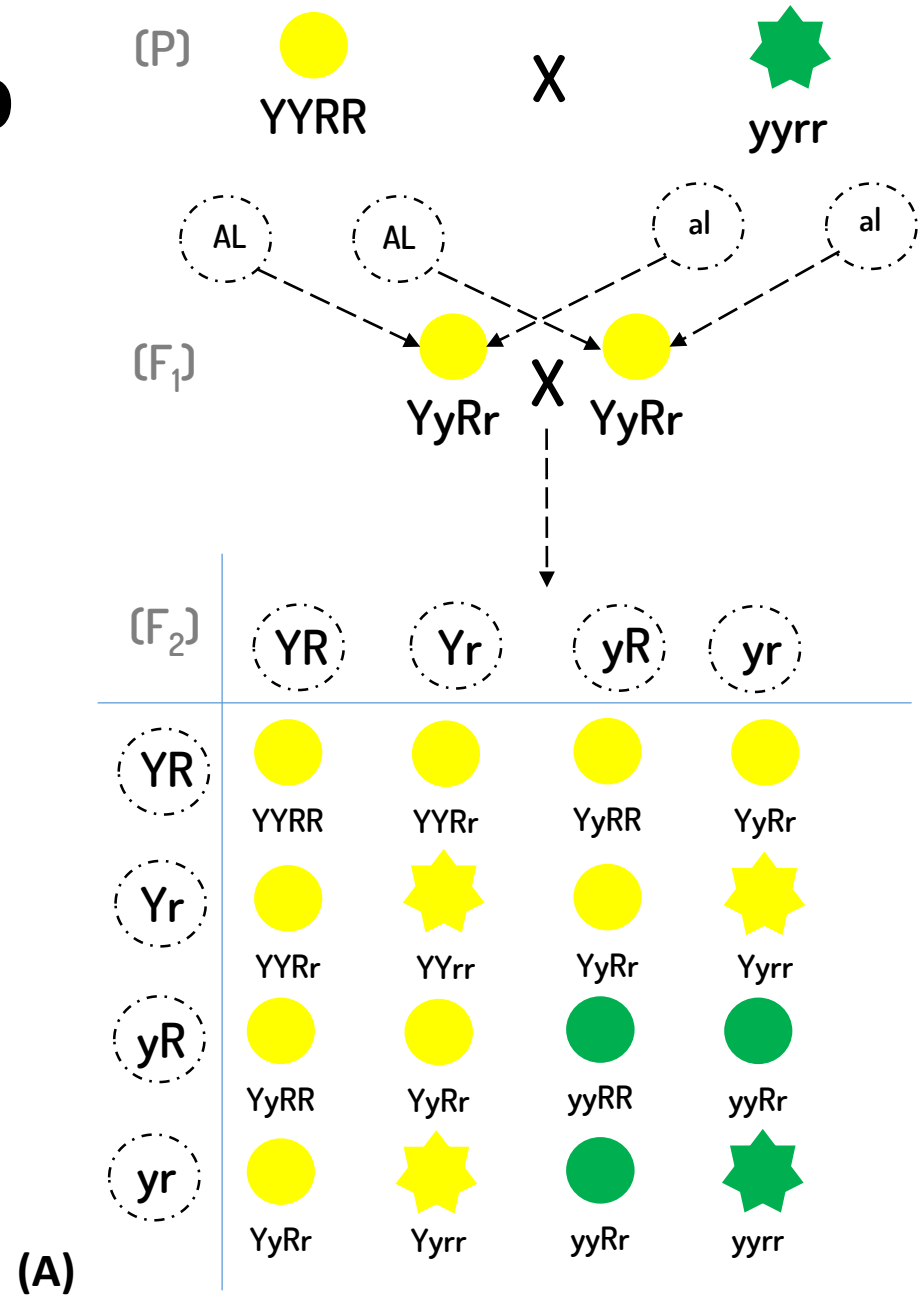


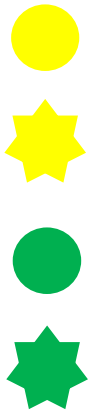
Image modified from "Mendel seven characters," by Mariana Ruiz Villareal (public domain).

DIHYBRID CROSSES



(A)

(B)



Y_R_ 9 Yellow-Round

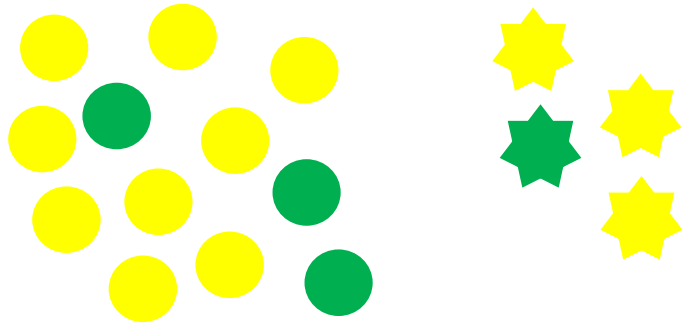
Y_rr 3 Yellow-Wrinkled

yyR_ 3 Green-Round

yyrr 1 Green-Wrinkled

(C)

12 Yellow:4 Green (3:1)



12 Round:4 Wrinkled (3:1)

CROSSES BETWEEN F_1 DIHYBRIDS

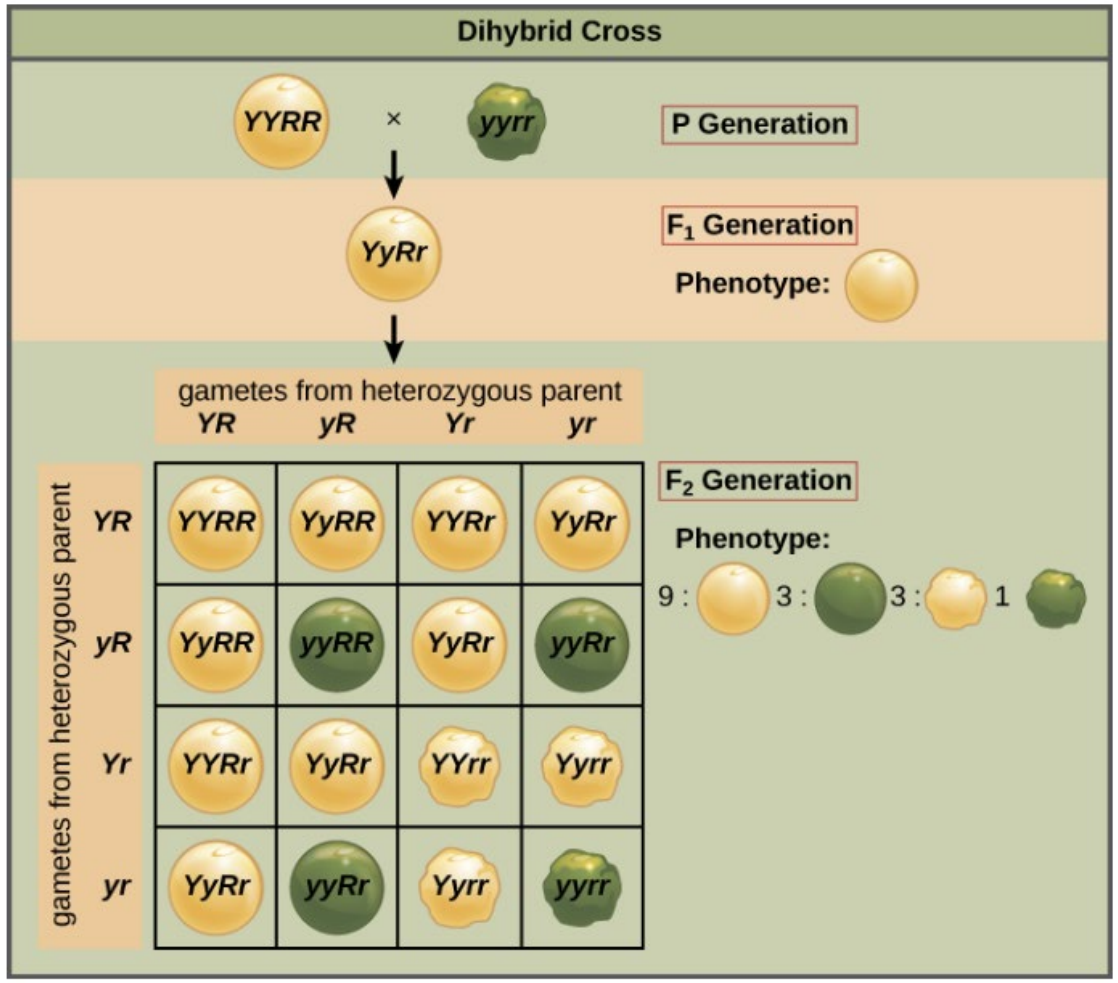


Image credit: "Laws of inheritance: Figure 2," by OpenStax College, Biology, CC BY 4.0.

Law of Independent Assortment (traits): alleles of two (or more) different genes get sorted into gametes independently of one another. In other words, the allele a gamete receives for one gene does not influence the allele received for another gene.

Only happens unlinked genes, that is, genes that are on different chromosomes or very far within the same chromosome.

Source: Khan Academy

- Introduction
- Arm folding
- Asparagus urine
- Attached earlobe
- Beeturia
- Bent pinkie
- Cheek dimples
- Cleft chin
- Darwin's tubercle
- Earwax
- Eye color
- Hair color
- Hair whorl
- Hand clasping
- Hitchhiker's thumb
- Mid-digital hair
- PTC tasting
- Toe length
- Tongue rolling
- Widow's peak

CANONICAL MENDELIAN GENE:

- 1 gen 1 trait
- 2 alleles
- Dominant > Recessive

https://udel.edu/~mcdonald/mythintro.html

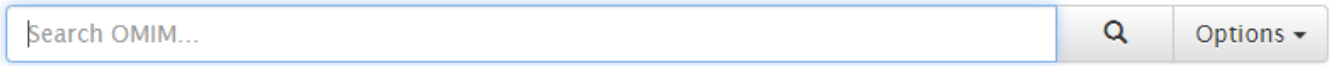
MENDELIAN TRAITS IN HUMAN:

Recessive: albinism

Source: http://wwwuser.cnb.csic.es/~albino/

Dominant: achondroplasia

OMIM® - Online Mendelian Inheritance in Man®



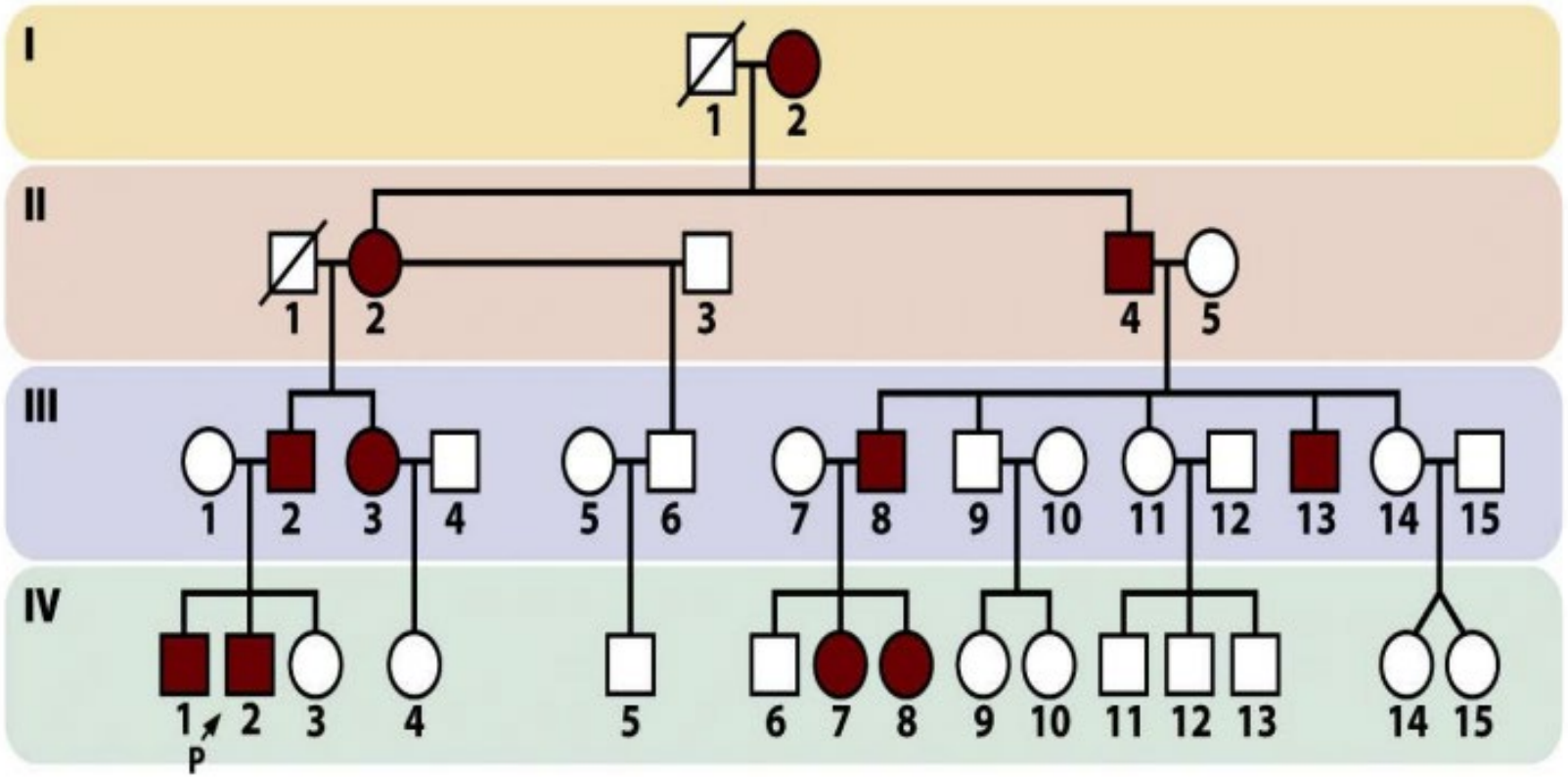
OMIM Entry Statistics

Number of Entries in OMIM (Updated September 27th, 2022) :

MIM Number Prefix	Autosomal	X Linked	Y Linked	Mitochondrial	Totals
Gene description *	15,998	756	51	37	16,842
Gene and phenotype, combined +	27	0	0	0	27
Phenotype description, molecular basis known #	6,067	370	5	34	6,476
Phenotype description or locus, molecular basis unknown %	1,396	112	4	0	1,512
Other, mainly phenotypes with suspected mendelian basis	1,644	102	3	0	1,749
Totals	25,132	1,340	63	71	26,606

https://www.omim.org/

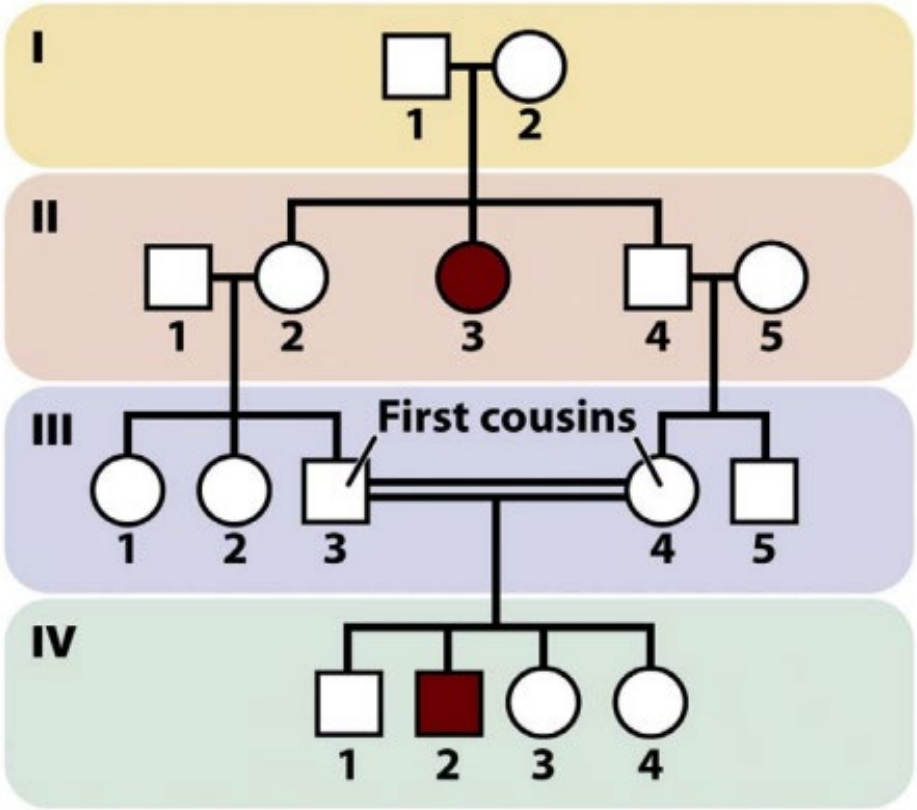
PEDIGREES



AUTOSOMIC DOMINANT:

- Same proportion in both sexes.
- An affected invididual has an affected parental.
- Tends to appear in all generations.

PEDIGREES



AUTOSOMIC RECESSIVE:

- Same proportion in both sexes.
- An affected individual normally have carrier parents.
- Tends to skip generations.
- Tends to appear after consanguineous crosses.

MAKING PREDICTIONS

1) **Addition rule:** states that the probability for either of different independent and mutually exclusive events occurring is calculated by adding the probabilities of each of them.

2) Multiplication rule: states that the probability of two or more independent events occurring simultaneously is calculated by multiplying their independent probabilities.

BINOMIAL DISTRIBUTION FORMULA

$$P = \frac{n!}{s!t!} p^s q^t$$

To determine the probability of a particular combination of events. Where P equals the total probability: with probability an event p of occurring s times and of an event Y with probability q of occurring t times. (s+t = n; p+q = 1)

GOODNESS-OF-FIT TESTS:

Chi-square: is a statistical test that tells us how well the observed values fit the expected values in an experiment. This test does not tell us whether a genetic cross has been performed correctly, whether the results are correct, or whether we have chosen the explanation that best fits our data. Instead, it indicates the probability that the difference between the observed and expected values is due to chance. It is calculated by applying the following formula:

$$\chi^2_{\text{exp}} = \sum \frac{(Observed - Expected)^2}{Expected}$$