Basic of genetics Course (Zoo206) for Second undergraduate students Of Biochemistry

Preparation

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رؤية الكلية

التميز فى تعليم العلوم الأساسية والبحث العلمى للمساهمة في بناء اقتصاد الوطن

رسالة الكلية

تقديم تعليم متميز فى مجالات العلوم الأساسية وإنتاج بحوث علمية تطبيقية تدعم اقتصاد الوطن من خلال إعداد خريجين متميزين طبقا للمعايير الأكاديمية القومية وتطوير مهارات وقدرات الموارد البشرية وتوفير خدمات مجتمعية وبيئية تلبى طموحات مجتمع جنوب الوادى وبناء الشراكات المجتمعية

رؤية القسم

خريجون وباحثون متميزون علميا وبحثيا محليا ودوليا خدمة للمجتمع وتنمية للبيئة

رسالة القسم

يسعى قسم علم الحيوان بكلية العلوم من خلال ما يقدمه من برامج تعليمية متطورة وبحث علمى تطبيقى وبنية أساسية مناسبة إلى خريجين متميزين محليا ودوليا فى مجالات العلوم البيولوجية ينتفع بهم المجتمع وسوق العمل

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الهدف من دراسة المقرر

١- يهدف هذا المقرر الى تقديم اساسيات علم الوراثة والوراثة الجزيئية والجينات ودورها في نقل الصفات الوراثية

٢- يتعرف الطالب على أهمية علم الوراثة في فهم علم الاحياء وتفسير الظواهر والعمليات البيولوجية والتغلب على الامراض الوراثية والمشكلات البيئية والزراعية والصناعية

٣- توضيح كيفية الاستفادة من علم الوراثة في حل المشاكل الصحية والدوائية والبيئية والزراعية.

مقدمة

قال الله تعالى:

"وَمَا مِنْ دَابَّةٍ فِي الأَرْضِ وَلا طَائِرٍ يَطِيرُ بِجَنَاحَيْهِ إِلاَّ أُمَمٌ أَمْثَالُكُمْ مَا فَرَّطْنَا فِي الْكِتَابِ مِنْ شَيْءٍ ثُمَّ إِلَى رَبِّهِمْ يُحْشَرُونَ " [1] **صدق الله العظيم**

وقال تعالى:

"وَلَقَدْ خَلَقْنَا الْإِنْسَانَ مِنْ سُلاَلَةٍ مِنْ طِينٍ ثُمَّ جَعَلْنَاهُ نُطْفَةً فِي قَرَارٍ مَكِينٍ ثُمَّ خَلَقْنَا النُّطْفَةَ عَلَقَةً فَخَلَقْنَا الْعَلَقَةَ مُضْغَةً فَخَلَقْنَا الْمُضْغَةَ عِظَاماً فَكَسَوْنَا الْعِظَامَ لَحْماً ثُمَّ أنشأنه خَلْقاً آخَرَ فَتَبَارَكَ اللَّهُ أَحْسَنُ الْخَالِقِينَ" [2] **صدق الله العظيم**

علم الوراثة Genetics هو العلم الذى يتناول دراسة الوراثة والتنوع فى الكائنات الحية، متضمنا دراسة المكونات الوراثية والصفات الوراثية للكائنات الحية والأجناس المختلفة، بالإضافة إلى الآليات التى تؤثر على الصفات الوراثية. يتصل علم الوراثة بصلة قوية بالطب وكل علوم الحياة الأخرى كعلم الأنسان (Anthropology) والكيمياء الحيوية (Biochemistry) وعلم الفسيولوجى (Physiology) وعلم النفس (Psychology) وعلم البيئة (Ecology) بالإضافة إلى علوم أخرى كما أن لعلم الوراثة بجزئيه النظرى والتجريبى تطبيقات مباشرة فى الأمراض الوراثية وكيفية السيطرة عليها، بالإضافة إلى التطبيقات الزراعية له، ولذا يعد علم الوراثة وتطبيقاته جزء مهم فى كثير من أنواع التعليم.

يحتوى الجسم البشرى على أكثر من عشرة ترليونات خلية ، كل خلية من هذه الخلايا تحتوى على برنامج يحتوى على كل المعلومات الضرورية للحياة، تنتقل هذه المعلومات من خلية إلى أخرى خلال الأنقسام الميتوزى للخلايا، ومن جيل إلى أخر بطريقة الانقسام الميوزى للخلايا من خلال خلايا متخصصة، هى الخلايا الجرثومية (germ cells) البويضات (oogcytes) أو الحيوانات المنوية (spermatzoa). وأخيرا فإن سلوك الأفراد فى العشائر يخضع للتحكم الوراثى ولو بصورة جزيئية. ولقد خطا علم الوراثة خطوات هائلة، فأصبح في مدى بضعة عقود في مقدمة العلوم الطبيعية.

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[1] ـ الأنعام "٣٨ "

[2] ـ المؤمنين" ١٢ "

First chapter

Mendelian genetics and its publications

Introduction

Genetics is the study of heredity. Johann Gregor Mendel (1822–1884) set the framework for genetics long before chromosomes or genes had been identified, at a time when meiosis was not well understood. Mendel selected a simple biological system and conducted methodical, quantitative analyses using large sample sizes. Because of Mendel's work, the fundamental principles of heredity were revealed. We now know that genes, carried on chromosomes, are the basic functional units of heredity with the capability to be replicated, expressed, or mutated. Today, the postulates put forth by Mendel form the basis of classical, or Mendelian, genetics. Not all genes are transmitted from parents to offspring according to Mendelian genetics, but Mendel's experiments serve as an excellent starting point for thinking about inheritance.

Mendel’s Laws of Inheritance

The father of genetics, Gregor Mendel, reported his findings in 1860 that were initially unpopular during his time but eventually gained traction and became so widely accepted that his findings paved the way for the founding of the science of genetics. Three different laws of inheritance were formulated based on his experimenting with pea plant reproduction. His experiments explained the transfer of genetic traits from one generation to the next. These laws have significantly expanded the understanding of genetic inheritance and resulted in new experimental methods becoming developed.

Mendel’s Experiments:

1856, Mendel began a decade-long research pursuit involving inheritance patterns in honeybees and plants, ultimately settling on pea plants as his primary model system. In 1865, Mendel presented the results of his experiments with nearly 30,000 pea plants to the local Natural History Society. He demonstrated that traits are transmitted faithfully from parents to offspring independently of other traits and in dominant and recessive patterns. In 1866, he published his work, Experiments in Plant Hybridization, in the proceedings of the Natural History Society of Brünn.

Mendel’s work went virtually unnoticed by the scientific community that believed, incorrectly, that the process of inheritance involved a blending of parental traits that produced an intermediate physical appearance in offspring**.**

Mendel’s Model System

Mendel’s seminal work was accomplished using the garden pea, Pisum sativum, to study inheritance. This species naturally self-fertilizes, such that pollen encounters ova within individual flowers. The flower petals remain sealed tightly until after pollination, preventing pollination from other plants. The result is highly inbred, or “true-breeding,” pea plants. These are plants that always produce offspring that look like the parent. By experimenting with true-breeding pea plants, Mendel avoided the appearance of unexpected traits in offspring that might occur if the plants were not true breeding. The garden pea also grows to maturity within one season, meaning that several generations could be evaluated over a relatively short time. Finally, large quantities of garden peas could be cultivated simultaneously, allowing Mendel to conclude that his results did not come about simply by chance.

Mendelian Crosses:

Mendel performed **hybridizations**, which involve mating two true-breeding individuals that have different traits. In the pea, this is done by manually transferring pollen from one pea plant to the stigma of another pea plant. In plants, pollen carries the male gametes (sperm) to the stigma, a sticky organ that traps pollen and allows the sperm to move down the pistil to the female gametes (ova) below. To prevent the pea plant that was receiving pollen from self-fertilizing and confounding his results, Mendel painstakingly removed all of the pollen-producing anthers from the plant's flowers before they had a chance to mature.

Plants used in first-generation crosses were called **P**, or parental generation, plants . Mendel collected the seeds that resulted from each cross and grew them the following season. These offspring were called the **F1**, or the first filial (*filial* = offspring, daughter or son), generation. Once Mendel examined the characteristics in the F1 generation of plants, he allowed them to self-fertilize. He then collected and grew the seeds from the F1 plants to produce the **F2**, or second filial, generation. Mendel's experiments extended beyond the F2 generation to the F3 and F4 generations, and so on, but it was the ratio of characteristics in the P−F1−F2 generations that were the most intriguing and became the basis for Mendel's principles.

Garden Pea Characteristics Revealed the Basics of Heredity:

In his 1865 publication, Mendel reported the results of his crosses involving seven different characteristics, each with two contrasting traits. A trait is defined as a variation in the physical appearance of a heritable characteristic. The characteristics include: tall vs. short plant height, wrinkled vs. round seeds, green vs. yellow seeds, violet vs. white flowers, etc. (Table 1). To fully examine each characteristic, Mendel generated large numbers of F1 and F2 plants, reporting results from 19,959 F2 plants alone.

As an example, let us look at Mendel's results for the flower color trait. First, Mendel confirmed that he had plants that bred true for white or violet flower color. Regardless of how many generations Mendel examined, all self-crossed offspring of parents with white flowers had white flowers, and all self-crossed offspring of parents with violet flowers had violet flowers. In addition, Mendel confirmed that, other than flower color, the pea plants were physically identical. Once these validations were complete, Mendel applied pollen from a plant with violet flowers to the stigma of a plant with white flowers. After gathering and sowing the seeds that resulted from this cross, Mendel found

that 100 percent of the F1 hybrid generation had violet flowers. Conventional wisdom at that time would have predicted the hybrid flowers to be pale violet or for hybrid plants to have equal numbers of white and violet flowers. In other words, the contrasting parental traits were expected to blend in the offspring. Instead, Mendel's results demonstrated that the white flower trait in the F1 generation had completely disappeared.

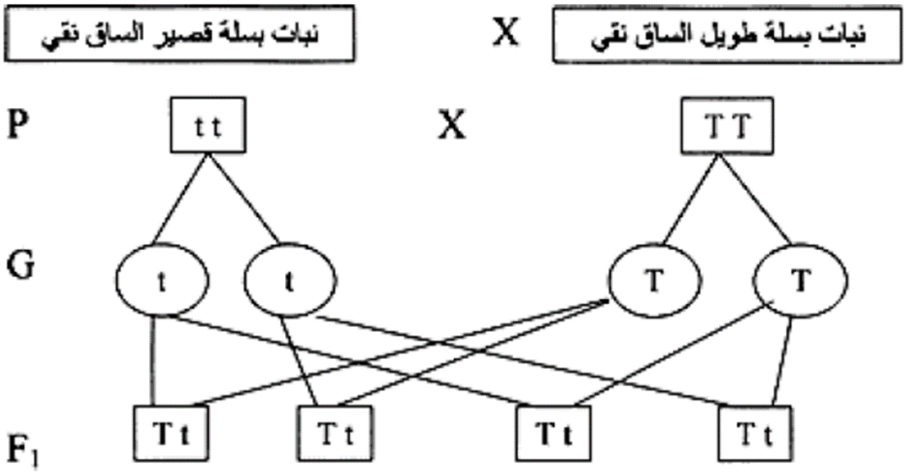
Importantly, Mendel did not stop his experimentation there. He allowed the F1 plants to self-fertilize and found that, of F2-generation plants, 705 had violet flowers and 224 had white flowers. This was a ratio of 3.15 violet flowers per one white

flower, or approximately 3:1. When Mendel transferred pollen from a plant with violet flowers to the stigma of a plant with white flowers and vice versa, he obtained about the same ratio regardless of which parent, male or female, contributed which trait. This is called a reciprocal cross. For the other six characteristics Mendel examined, the F1 and F2 generations behaved in the same way as they had for flower color. One of the two traits would disappear completely from the F1 generation only to reappear in the F2 generation at a ratio of approximately 3:1 (Table 1).

Table 1. The Results of Mendel's Garden Pea Hybridizations

Characteristic	Contrasting P0 Traits	F1 Offspring Traits	F2 Offspring Traits	F2 Trait Ratios
Flower color	Violet vs. white	100 percent violet	705 violet 224 white	3.15:1
Flower position	Axial vs. terminal	100 percent axial	651 axial 207 terminal	3.14:1
Plant height	Tall vs. dwarf	100 percent tall	787 tall 277 dwarf	2.84:1
Seed texture	Round vs. wrinkled	100 percent round	5,474 round 1,850 wrinkled	2.96:1
Seed color	Yellow vs. green	100 percent yellow	6,022 yellow 2,001 green	3.01:1
Pea pod texture	Inflated vs. constricted	100 percent inflated	882 inflated 299 constricted	2.95:1
Pea pod color	Green vs. yellow	100 percent green	428 green 152 yellow	2.82:1

An example: of a cross between a pure long-stem pea plant and a short-stem one.



Mendel generalized the results of his pea-plant experiments into three principles that describe the basis of inheritance in diploid organisms. They are: the principle (Law) of segregation, the principle (Law) of dominance, and the principle (Law) of independent assortment and the Law of Unit Characters. Together, these principle summarize the basics of classical, or Mendelian, genetics.

The Principle of Segregation:

Since the white flower trait reappeared in the F2 generation, Mendel saw that the traits remained separate (not blended) in the plants of the F1 generation. This led to the principle of segregation, which states that individuals have two copies of each trait, and that each parent transmits one of its two copies to its offspring.

Law of Segregation (the first law of inheritance)

‘‘The two copies of each genetic factor segregate during the development of gametes, to ensure that each parent’s offspring attains one factor.’’

Or

‘‘During the development of the gamete, each gene is segregated in such a way that the gamete consists of just one allele for that gene.’’

The copies of a gene are segregated when any individual produces gametes so that each gamete accepts only one copy. One allele is received by a gamete.

In 1902, the fact that the genetic factors proposed by Mendel were carried on chromosomes was proposed by Walter and Sutton and Theodor Boveri as the Chromosomal Theory of Inheritance.

Difference between allele and gene

A gene is an essential part of the DNA that defines a specific trait; an allele is a specific form of a gene. The expression of traits is the key role of the genes. Alleles are important for the variations in which the trait can be expressed.

Mendel, who had no knowledge of chromosomes, proposed that the determining factors of inheritance are discrete “unit factors” (now called genes) that maintain their integrity from the time that the zygote is formed through the time that it matures and produces its own gametes. During gamete formation, the members of these paired “unit factors” segregate from one another and enter into separate gametes.

Different versions of genes are called alleles. Diploid organisms that have two identical alleles of a gene on their two homologous chromosomes are homozygous for that trait. Diploid organisms that have two different alleles of a gene on their two homologous chromosomes are heterozygous for that trait.

The physical basis of the principle of segregation is the first division of meiosis, in which the homologous chromosomes with their different versions of each gene are segregated into daughter nuclei. Since each gamete receives only one homolog of each chromosome, it follows that they receive only one allele for each trait. At fertilization, the zygote receives one of each homologous chromosome, and one of each allele, from each parent.

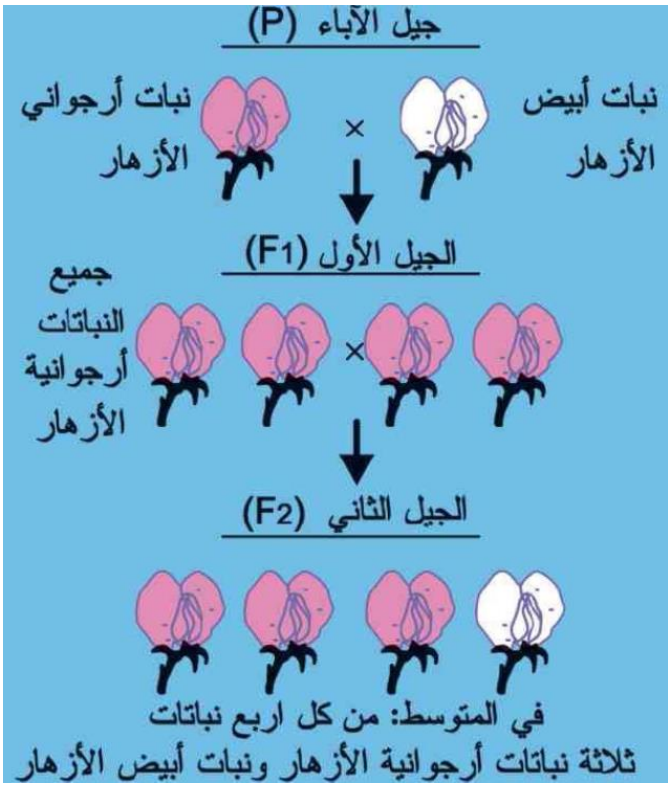


Fig. (1). Mendel crossed plants that were true-breeding for violet flower color with plants true-breeding for white flower color (the P generation). The resulting hybrids in the F1 generation all had violet flowers. In the F2 generation, approximately three quarters of the plants had violet flowers, and one quarter had white flowers.

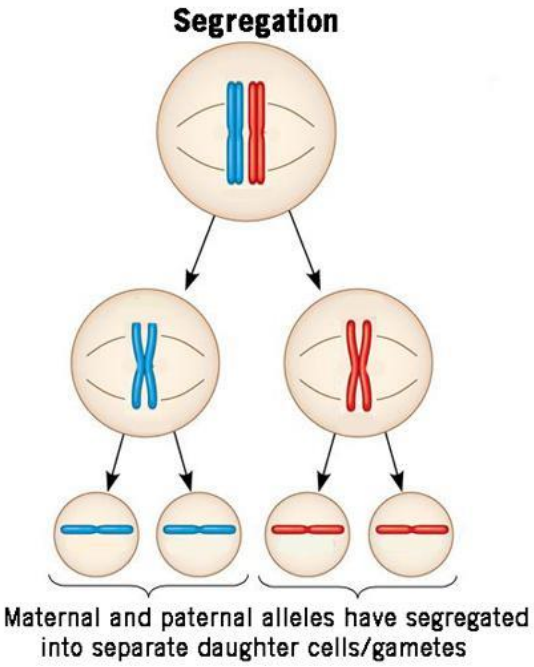


Fig.(2). Alleles during meiosis.

The Principle of Dominance

Upon compiling his results for many thousands of plants, Mendel concluded that the characteristics could be divided into dominant and recessive traits. Dominant traits are those that are expressed in a hybridization. Recessive traits become latent, or disappear, in the offspring of a hybridization but reappear in the

progeny of the hybrid offspring. Thus, the violet-flower trait is dominant and the white-flower trait is recessive.

The principle of dominance states that in a heterozygote, only the dominant allele will be expressed. The recessive allele will remain “latent” but will be transmitted to offspring by the same manner in which the dominant allele is transmitted. The recessive trait will only be expressed by offspring that have two copies of this allele. Individuals with a dominant trait could have either two dominant versions of the trait or one dominant and one recessive version of the trait. Individuals with a recessive trait have two recessive alleles.

Example

The alleles for a Mendelian trait may either be dominant or recessive and may be passed down from parent to child (animal or plant). In plants, for example, the color trait of the flower will depend on the type of allele inherited by the offspring. Each parent plant transfers one of the alleles to their offspring. And these sets of alleles in the offspring will depend on the chromosomes of the two gametes uniting at fertilization. These two sets of chromosomes randomly segregated during gamete formation (wherein meiosis is a part of the process).

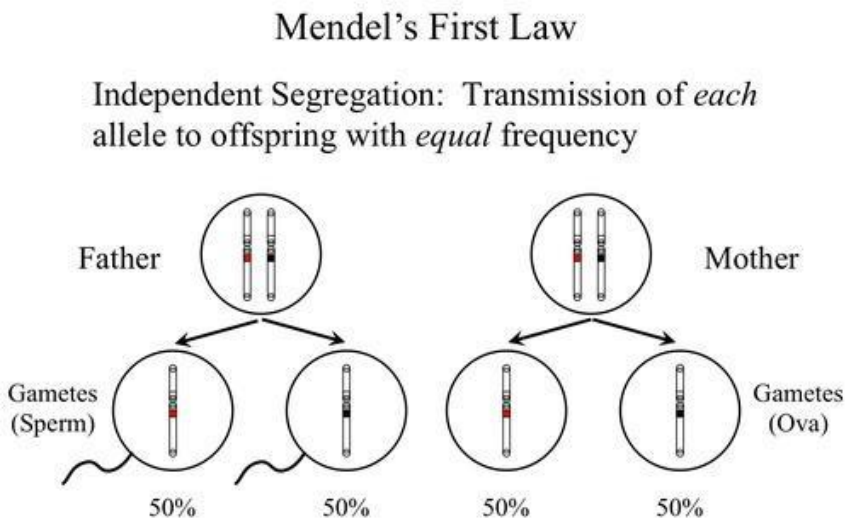


Fig. (3) Independent segregation

Phenotypes and Genotypes

Several conventions exist for referring to genes and alleles. For the purposes of this chapter, we will abbreviate genes using the first letter of the gene's corresponding dominant trait. For example, green is the dominant trait for pea pod color, so the pod-color gene would be abbreviated as *G* (note that it is customary to italicize gene designations). Furthermore, we will use uppercase and lowercase letters to represent dominant and recessive alleles, respectively. Therefore, we would refer to the genotype of a homozygous dominant pea plant with green pods

as *GG*, a homozygous recessive pea plant with yellow pods as *gg*, and a heterozygous pea plant with green pods as *Gg*.

The two alleles for each given gene in a diploid organism may be expressed and interact to produce physical characteristics. The observable traits expressed by an organism are referred to as its phenotype. An organism's underlying genetic makeup, which alleles it has, is called its genotype. Mendel's hybridization experiments demonstrate the difference between phenotype and genotype. When true-breeding plants in which one parent had yellow pods and one had green pods were cross-fertilized, all of the F1 hybrid offspring had green pods. Although the hybrid offspring had the same phenotype as the true-breeding parent with green pods, we know that the genotype of the parent was homozygous dominant (GG), while the genotype of the F1 offspring was heterozygous (Gg). the yellow pod allele reappeared in some of the F2 offspring (gg).

Using Punnett Squares for Monohybrid Crosses:

Punnett squares, devised by the British geneticist Reginald Punnett, can be used to predict the possible outcomes of a genetic cross or mating and their expected frequencies. To demonstrate a monohybrid cross, consider the case of true-breeding pea plants with yellow versus green pea seeds. The dominant seed color is yellow; therefore, the parental genotypes

were YY for the plants with yellow seeds and yy for the plants with green seeds, respectively. To prepare a Punnett square, all possible combinations of the parental alleles are listed along the top (for one parent) and side (for the other parent) of a grid, representing their meiotic segregation into haploid gametes. Then the combinations of egg and sperm are made in the boxes in the table to show which alleles are combining. Each box then represents the diploid genotype of a zygote, or fertilized egg, that could result from this mating. Because each possibility is equally likely, genotypic ratios can be determined from a Punnett square. If the pattern of inheritance (dominant or recessive) is known, the phenotypic ratios can be inferred as well. For a monohybrid cross of two true-breeding parents, each parent contributes one type of allele. In this case, only one genotype is possible. All offspring are Yy and have yellow seeds (Figure 2). A self-cross of one of the *Yy* heterozygous offspring can be represented in a 2 × 2 Punnett square because each parent can donate one of two different alleles. Therefore, the offspring can potentially have one of four allele combinations: *YY*, *Yy*, *yY*, or *yy* (**Figure 4**).

Because fertilization is a random event, we expect each combination to be equally likely and for the offspring to exhibit a ratio of *YY*:*Yy*:*yy* genotypes of 1:2:1 (**Figure 2**). Furthermore, because the *YY* and *Yy* offspring have yellow seeds and are phenotypically identical, we expect the offspring to exhibit a

phenotypic ratio of 3 yellow:1 green. Indeed, working with large sample sizes, Mendel observed approximately this ratio in every F2 generation resulting from crosses for individual traits.

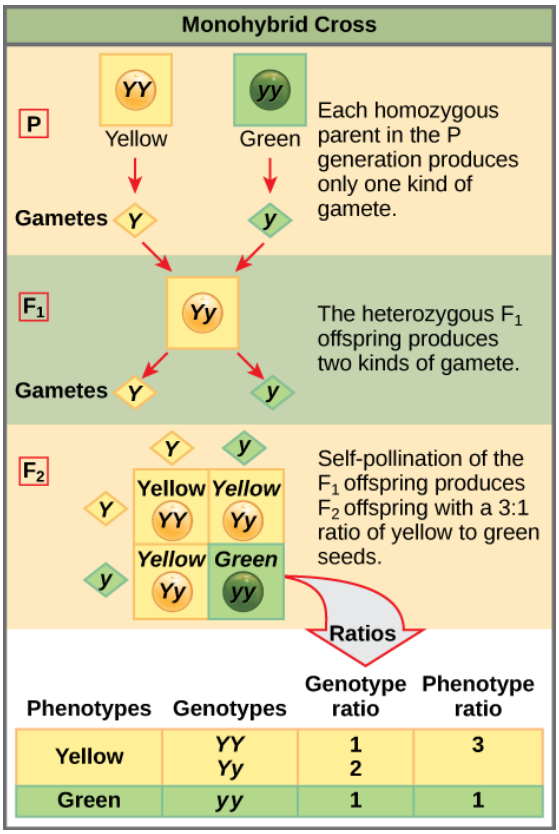


Fig. (4). In the P generation, pea plants that are true-breeding for the dominant yellow phenotype are crossed with plants with the recessive green phenotype. This cross produces F1 heterozygotes with a yellow phenotype. Punnett square analysis can be used to predict the genotypes of the F2 generation.

https://www.biologyonline.com/dictionary/law-of-segregation

Using a Test Cross to Determine Genotype

Test crosses can be used to determine whether a dominant phenotype is homozygous or heterozygous

1- If the unknown parent is homozygous dominant, all offspring will express the dominant phenotype

2- If the unknown parent is heterozygous, half the offspring should be dominant and half recessive

Beyond predicting the offspring of a cross between known homozygous or heterozygous parents, Mendel also developed a way to determine whether an organism that expressed a dominant trait was a heterozygote or a homozygote. Called the **test cross**, this technique is still used by plant and animal breeders. In a test cross, an organism with the dominant phenotype is crossed with an organism that is homozygous recessive for the same characteristic. If the dominant-expressing organism is a homozygote, then all F1 offspring will be heterozygotes expressing the dominant trait. Alternatively, if the dominant expressing organism is a heterozygote, the F1 offspring will exhibit a 1:1 ratio of heterozygotes and recessive homozygotes (**Figure 3**). The test cross further validates Mendel's postulate that pairs of unit factors segregate equally.

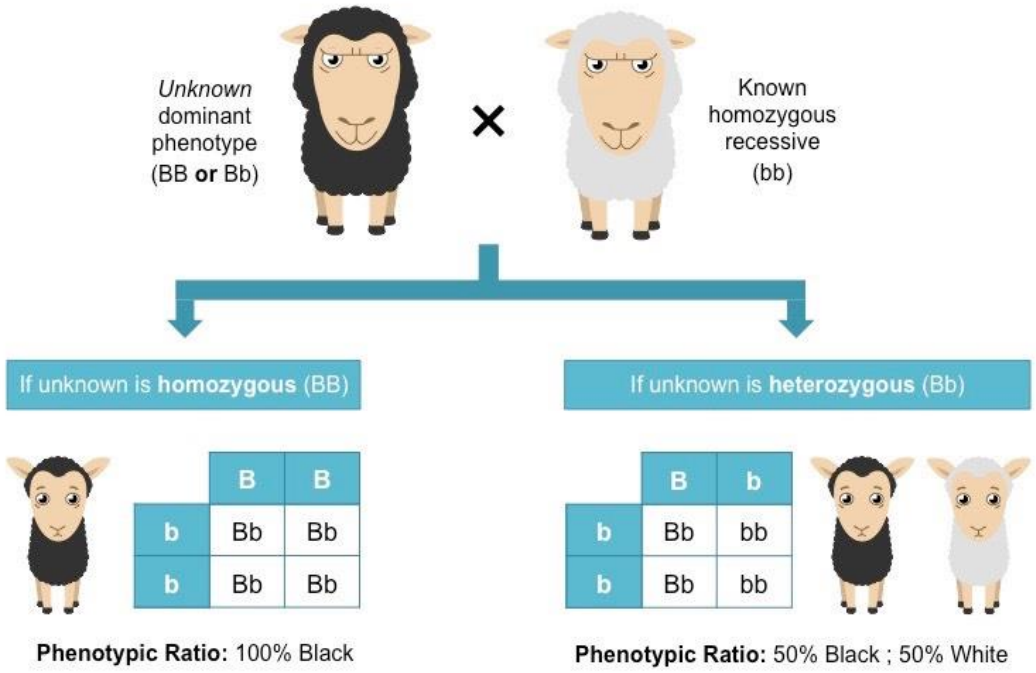


Fig. (5). Testing an Unknown Dominant Phenotype

https://ib.bioninja.com.au/higher-level/topic-10-genetics-and-evolu/102-inheritance/test-cross.html

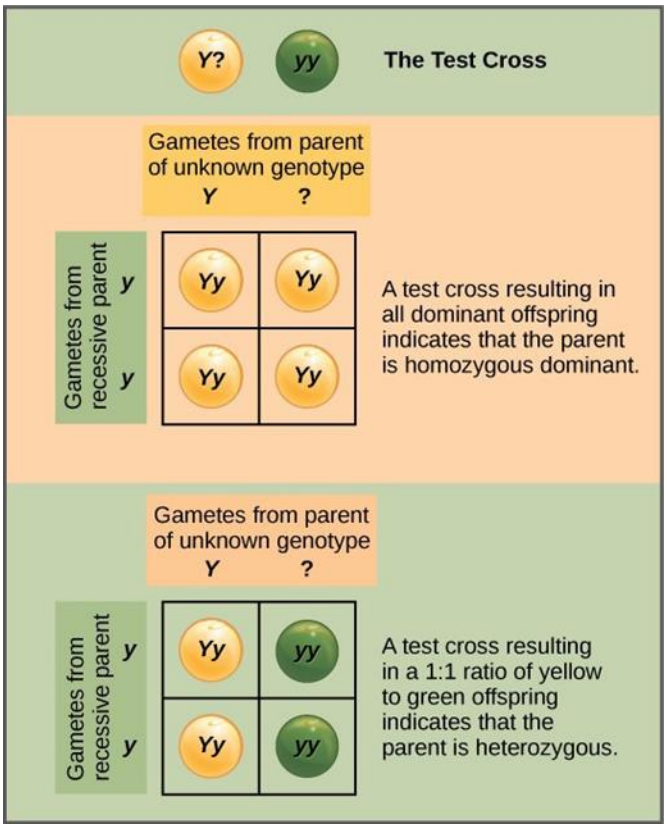


Figure (6). A test cross can be performed to determine whether an organism expression a dominant trait is a homozygote or a heterozygote.

Probleme:

In pea plants, round peas (R) are dominant to wrinkled peas (r). You do a test cross between a pea plant with wrinkled peas (genotype rr) and a plant of unknown genotype that has round peas.

Using Pedigrees to Study Inheritance Patterns

Many human diseases are inherited genetically. A healthy person in a family in which some members suffer from a recessive genetic disorder may want to know if he or she has the disease-causing gene and what risk exists of passing the disorder on to his or her offspring. Of course, doing a test cross in humans is unethical and impractical. Instead, geneticists use pedigree analysis to study the inheritance pattern of human genetic diseases.

Each row of a pedigree represents one generation of the family. Women are represented by circles; males by squares. People who had children together are connected with a horizontal line and their children are connected to this line with a vertical line. See (Figure 4) for an example of a pedigree for a human genetic disease.

People with the recessive genetic disease alkaptonuria cannot properly metabolize two amino acids, phenylalanine and tyrosine. Affected individuals may have darkened skin and brown urine, and may suffer joint damage and other complications.

In this pedigree, individuals with the disorder are indicated in blue and have the genotype aa. Unaffected individuals are indicated in yellow and have the genotype AA or Aa. Note that it

is often possible to determine a person's genotype from the genotype of their offspring. For example, if neither parent has the disorder but their child does, both parents must be heterozygous.

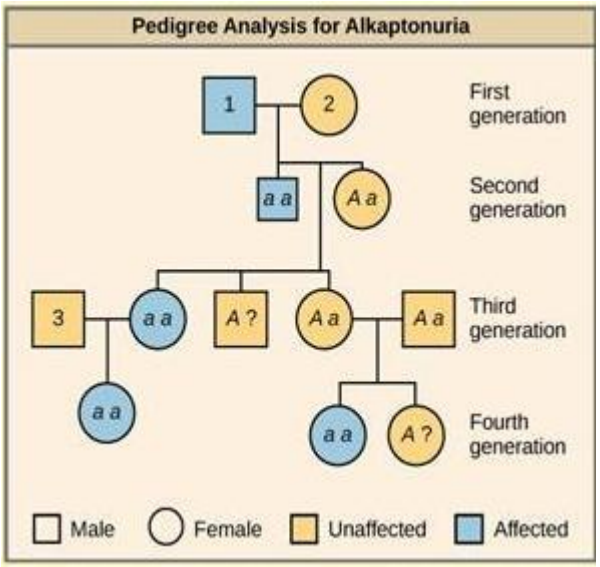


Fig. (7). Pedigree of a human family with the recessive genetic disease

https://blog.cambridgecoaching.com/tips-for-interpreting-pedigree-charts-and-understanding-inheritance-patterns

Probleme:

What are the genotypes of the individuals labeled 1, 2, and 3 in the figure 4?

Principle of Independent Assortment

Mendel’s principle of independent assortment states that genes do not influence each other with regard to the sorting of alleles into gametes, and every possible combination of alleles for every gene is equally likely to occur. The independent assortment of genes can be illustrated by a dihybrid cross, a cross between two true-breeding parents that express different traits for two characteristics. Consider the characteristics of seed color and seed texture for two pea plants, one that has green, wrinkled seeds (yyrr) and another that has yellow, round seeds (YYRR). Because each parent is homozygous, the principle of segregation indicates that the gametes for the green/wrinkled plant all are yr, and the gametes for the yellow/round plant are all YR. Therefore, the F1 generation of offspring all are YyRr (Figure 8).

The physical basis for the principle of independent assortment also lies in meiosis I, in which the different homologous pairs line up in random orientations. Each gamete can contain any combination of paternal and maternal chromosomes (and therefore the genes on them) because the orientation of tetrads on the metaphase plane is random.

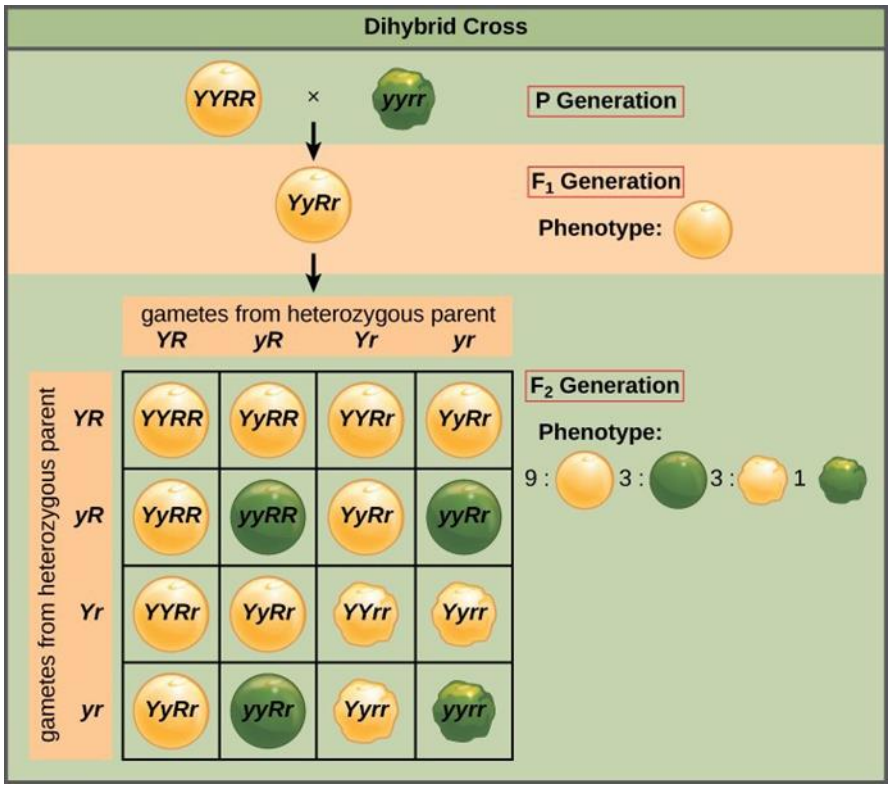


Figure (8). In a dihybrid cross, two traits are followed in a single cross. Here, both seed color and seed smoothness are followed into the F2 generation.

Exceptions to Mendel's Principles of Inheritance

Although Mendel's principles still apply to some situations, many situations exist in which they do not apply. These "exceptions" to Mendelian genetics are discussed.

Alternatives to Dominance and Recessiveness

Since Mendel's experiments with pea plants, other researchers have found that the principle of dominance does not always hold true. several different patterns of inheritance have been found to exist.

1- Polygenic Traits

Some traits are determined by the combined effect of more than one pair of genes. These are referred to as polygenic click this icon to hear the preceding term pronounced, or continuous, traits. An example of this is human stature. The combined size of all of the body parts from head to foot determines the height of an individual. There is an additive effect. The sizes of all of these body parts are, in turn, determined by numerous genes. Human skin, hair, and eye color are also polygenic traits because they are influenced by more than one allele at different loci. The result is the perception of continuous gradation in the expression of these traits.

NOTE: whether an individual achieves his or her genetically programmed height is significantly affected by thyroid gland hormones and human growth hormones (HGH) produced in the pituitary gland . A deficiency in the amount of these hormones during childhood and puberty can result in stunted growth. Too much of them can cause excessive growth resulting in exceptional height. Differences in diet and other environmental

factors during the crucial growth years can also be important in determining stature and other complex traits. Usually, about 10% of an individual's height is due to the environment.

2- Incomplete Dominance (Intermediate Expression)

Mendel’s results, that traits are inherited as dominant and recessive pairs, contradicted the view at that time that offspring exhibited a blend of their parents’ traits. However, the heterozygote phenotype occasionally does appear to be intermediate between the two parents. For example, in the Seven palate plant، a cross between a homozygous parent with white flowers (WW) and a homozygous parent with red flowers (RR) will produce offspring with pink flowers (RW). (Note that different genotypic abbreviations are used for Mendelian extensions to distinguish these patterns from simple dominance and recessiveness). This pattern of inheritance is described as incomplete dominance, denoting the expression of two contrasting alleles such that the individual displays an intermediate phenotype. The allele for red flowers is incompletely dominant over the allele for white flowers. However, the results of a heterozygote self-cross can still be predicted, just as with Mendelian dominant and recessive crosses. In this case, the genotypic ratio would be 1 RR : 2 RW : 1 WW, and the phenotypic ratio would be 1:2:1 for red: pink: white.

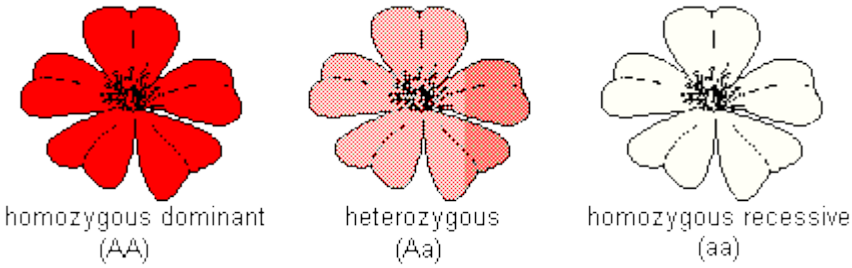


Fig. (9). Intermediate Expression

3- Codominance

A variation on incomplete dominance is codominance, in which both alleles for the same characteristic are simultaneously expressed in the heterozygote. An example of codominance is the MN blood groups of humans. The M and N alleles are expressed in the form of an M or N antigen present on the surface of red blood cells. Homozygotes (MM and NN) express either the M or the N allele, and heterozygotes (MN) express both alleles equally. In a self-cross between heterozygotes expressing a codominant trait, the three possible offspring genotypes are phenotypically distinct. However, the 1:2:1 genotypic ratio characteristic of a Mendelian monohybrid cross still applies.

For two alleles can be codominant. That is to say, both are expressed in heterozygous individuals. An example of this is

people who have an AB blood type for the ABO blood system. When they are tested, these individuals actually have the characteristics of both type A and type B blood. Their phenotype is not intermediate between the two.

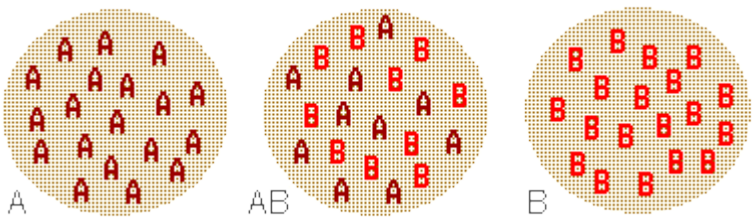


Fig. (10). two alleles can be codominant

4- Multiple Alleles

Mendel implied that only two alleles, one dominant and one recessive, could exist for a given gene. We now know that this is an oversimplification. Although individual humans (and all diploid organisms) can only have two alleles for a given gene, multiple alleles may exist at the population level such that many combinations of two alleles are observed. Note that when many alleles exist for the same gene, the convention is to denote the most common phenotype or genotype among wild animals as the wild type .

1- An example of multiple alleles is coat color in rabbits (Figure 11). Here, four alleles exist for the c gene. The wild-type version,

C^+C^+, is expressed as brown fur. The chinchilla phenotype, $c^{ch}c^{ch}$, is expressed as black-tipped white fur. The Himalayan phenotype, c^hc^h, has black fur on the extremities and white fur elsewhere. Finally, the albino, or “colorless” phenotype, cc, is expressed as white fur. In cases of multiple alleles, dominance hierarchies can exist. In this case, the wild- type allele is dominant over all the others, chinchilla is incompletely dominant over Himalayan and albino, and Himalayan is dominant over albino. This hierarchy, or allelic series, was revealed by observing the phenotypes of each possible heterozygote offspring.

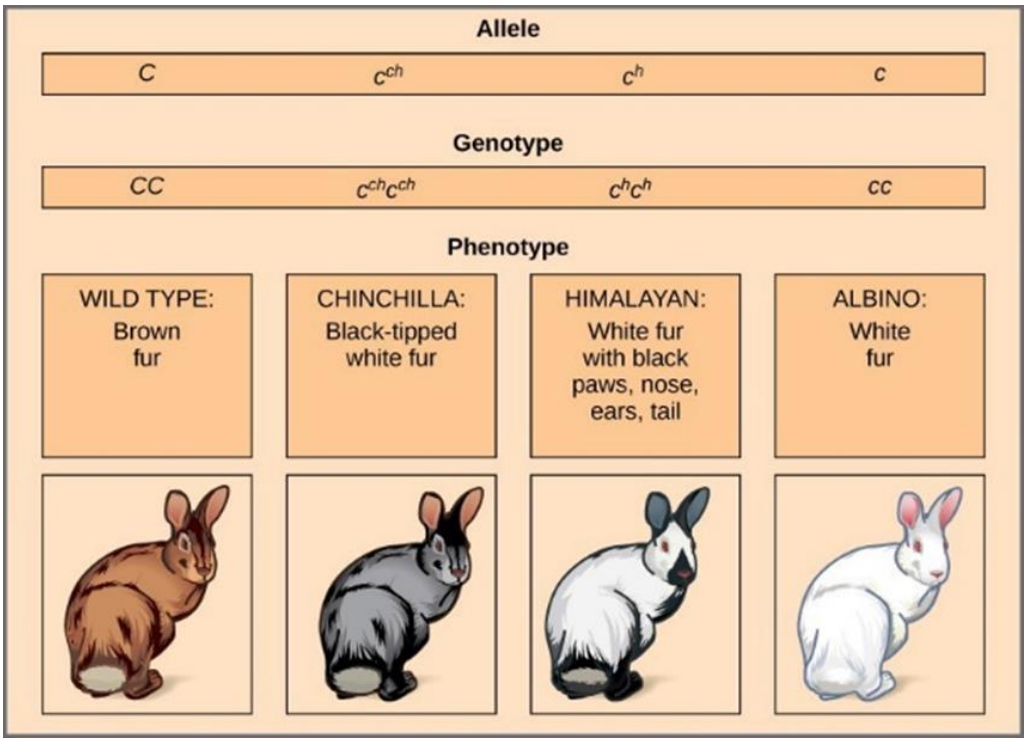


Fig. (11). Four different alleles exist for the rabbit coat color (C) gene.

Interestingly, the Himalayan phenotype in rabbits is the result of an allele that produces a temperature-sensitive gene product that only produces pigment in the cooler extremities of the rabbit's body. In this case, the protein product of the gene does not fold correctly at high temperatures. A similar gene gives Siamese cats their distinctive coloration.

Temperature-sensitive proteins are also at work in arctic foxes and rabbits, which are white in the winter and darker colored during the summer. In these cases, the protein product of the gene does not fold correctly at colder temperatures. The mutation that caused this coloration was advantageous to these species, so they persisted in the populations.

- Blood Types

1. Human blood type is good example of BOTH multiple alleles and codominance.

2- There are 3 possible alleles and two of them act dominant (A and B are dominant while O is a recessive allele.)

3. Blood Type Alleles

- Type A: Causes cells to make protein A for the surface of the RBC's.

- Type B: Causes cells to make protein B for the surface of the RBC's.

- Type O: Causes cells to make no protein for the surface of the RBC's.

3. Both A and B alleles are DOMINANT over the O allele because if any A or B allele is present., they will be expressed and the A or B proteins will be made.

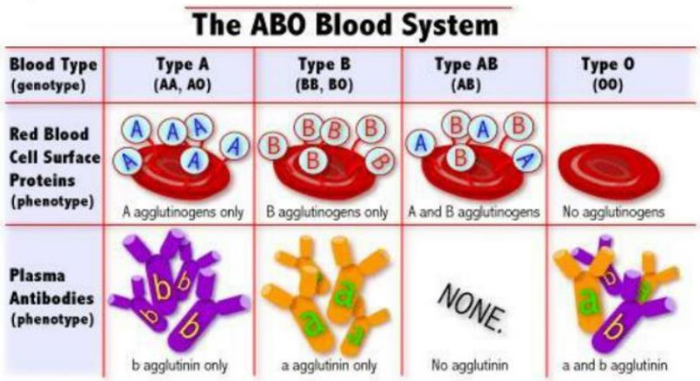


Fig. (12)

An example of multiple alleles in humans pertains to ABO blood type. A person's blood type (e.g., type A or type O) is caused by different combinations of three alleles: IA, IB, and IO. A person with type A blood could have either IAIA or IAIO genotype. A person with type B blood could have IBIB or IBIO genotype. A person with type O blood must have the IOIO genotype. Note that type AB blood is an example of codominance (IAIB).

The ABO blood type system is also an example of a trait that is controlled by more than just a single pair of alleles. In other words, it is due to a multiple-allele series. In this case, there are

three alleles (A, B, and O), but each individual only inherits two of them (one from each parent).

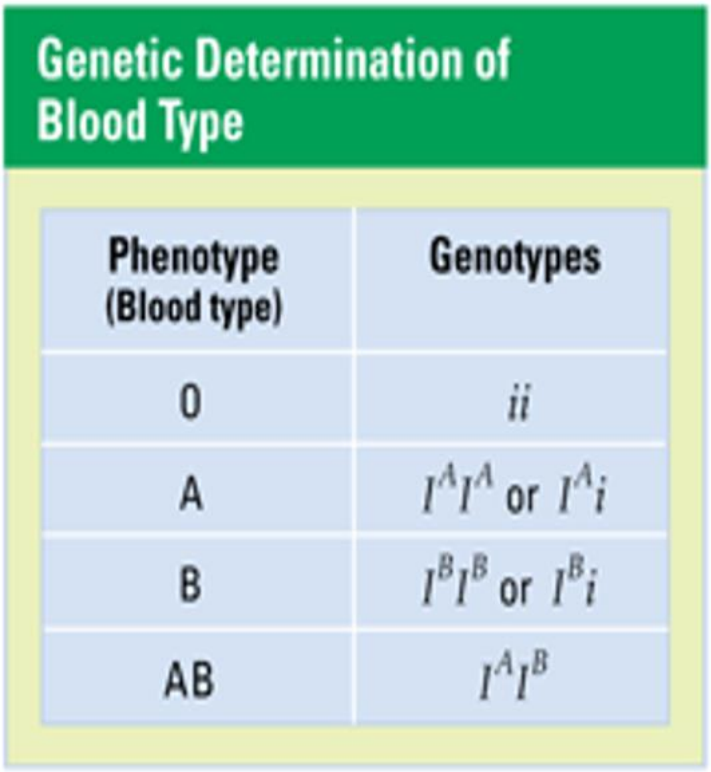


Fig. (13). The combinations of these alleles result in six genotypes and four phenotypes. Alleles IA and IB are codominant. Allele i is recessive

Some traits are controlled by far more alleles. For instance, the human HLA system, which is responsible for identifying and rejecting foreign tissue in our bodies, can have at least 30,000,000 different genotypes. It is the HLA system which

causes the rejection of organ transplants. The more we learn about human genetics the more it becomes clear that multiple-allele series are very common. In fact, it now appears that they are more common than simple two allele ones.

https://en.wikipedia.org/wiki/Mendelian_inheritance

5- Modifying and Regulator Genes

There are two classes of genes that can have an effect on how other genes function. They are called modifying genes and regulator genes.

Modifying genes alter how certain other genes are expressed in the phenotype. For instance, there is a dominant cataract, the gene which will produce varying degrees of vision impairment depending on the presence of a specific allele for a companion modifying gene. However, cataracts also can be promoted by diabetes and common environmental factors such as excessive ultraviolet radiation, and alcoholism.

Regulator genes can either initiate or block the expression of other genes. They control the production of a variety of chemicals in plants and animals. For instance, the time of production of specific proteins that will be new structural parts of our bodies can be controlled by such regulator genes. Shortly

after conception, regulator genes work as master switches orchestrating the timely development of our body parts. They are also responsible for changes that occur in our bodies as we grow older. In other words, they control the maturation and aging processes. Regulator genes that are involved in subdividing an embryo into what will become the major body parts of an individual are also referred to as homeotic .

head, torso, arms, legs, etc.

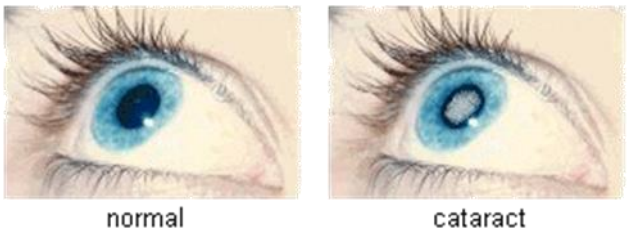


Fig. (14). a dominant cataract gene which will produce varying degrees of vision impairment

6- Incomplete Penetrance

Some genes are incompletely penetrant. That is to say, their effect does not normally occur unless certain environmental factors are present. For example, you may inherit the genes that are responsible for type 2 diabetes but never get the disease unless you become greatly overweight, persistently stressed

psychologically, or do not get enough sleep on a regular basis. Similarly, the genes that cause the chronic autoimmune disease, multiple sclerosis , may be triggered by the Epstein-Barr virus and possibly other specific environmental stresses.

7- Sex Related Genetic effects

There are three categories of genes that may have different effects depending on an individual's gender. These are referred to as:

1. sex-limited genes

2. sex-controlled genes

3. genome imprinting

Sex-limited genes are ones that are inherited by both men and women but are normally only expressed in the phenotype of one of them. The heavy male beard is an example. While women have facial hair it is most often very fine and comparatively sparse.

In contrast, sex-controlled genes are expressed in both sexes but differently. An example of this is gout, a disease that causes painfully inflamed joints. If the gene is present, men are nearly eight times more likely than women to have severe symptoms.

Some genes are known to have a different effect depending on the gender of the parent from whom they are inherited. This phenomenon is referred to as genome imprinting or genetic imprinting. Apparently, diabetes, psoriasis , and some rare genetically inherited diseases, such as a form of mental retardation known as Angelman syndrome, can follow this inheritance pattern. Recent research by Catherine Dulac of Harvard University points to genetic imprinting as being an important factor in causing male and female brains to develop somewhat differently. She suggests that this is due to the fact that some of the genes inherited from the opposite sex parent are likely to be turned off following conception.

8- Pleiotropy

A single gene may be responsible for a variety of traits. This is called pleiotropy. The complex of symptoms that are collectively referred to as sickle-cell trait, or sickle-cell anemia, is an example. A single gene results in irregularly shaped red blood cells that painfully block blood vessels, cause poor overall physical development, as well as related heart, lung, kidney, and eye problems. Another pleiotropic trait is albinism. The gene for this trait not only results in a deficiency of skin, hair, and eye pigmentation but also causes defects in vision.

9- Stuttering Alleles

Lastly, it is now known that some genetically inherited diseases have more severe symptoms each succeeding generation due to segments of the defective genes being doubled in their transmission to children . These are referred to as stuttering alleles or unstable alleles. Examples of this phenomenon are Huntington's disease.

Mendel believed that all units of inheritance are passed on to offspring unchanged. Unstable alleles are an important exception to this rule.

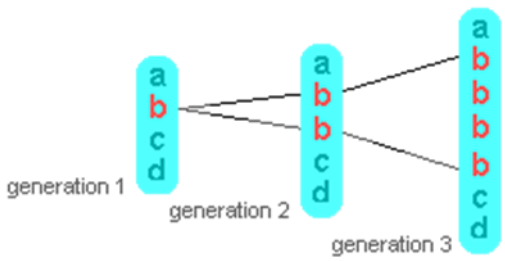


Fig. (15). Unstable allele doubling each generation

Environmental Effects

The phenotype of an individual is not only the result of inheriting a particular set of parental genes. The specific environmental characteristics of the uterus in which a fertilized egg is implanted and the health of the mother can have major impacts on the phenotype of the future child. For instance, oxygen deprivation or inappropriate hormone levels can cause lifelong, devastating effects. Likewise, accidents, poor nutrition, and other environmental influences throughout life can alter an individual's phenotype for many traits.

Geneticists study identical or monozygotic twins to determine which traits are inherited and which ones were acquired following conception. Since monozygotic twins come from the same zygote, they are essentially identical in their genetic makeup. If there are any differences in their phenotypes, the environment is virtually always responsible. Such differences show up in basic capabilities such as handedness, which had been assumed to be entirely genetically determined. In rare instances, one monozygotic twin will be clearly right-handed while the other will be left-handed. This suggests that there may be both genetic and environmental influences in the development of this trait.

https://www.palomar.edu/anthro/mendel/mendel_3.htm

10- Linked Traits are an Exception to the Principle of Segregation

In humans, as well as in many other animals and some plants, the sex of the individual is determined by sex chromosomes. The sex chromosomes are one pair of non-homologous chromosomes. Until now, we have only considered inheritance patterns among non-sex chromosomes, or autosomes. In addition to 22 homologous pairs of autosomes, human females have a homologous pair of X chromosomes, whereas human males have an XY chromosome pair. Although the Y chromosome contains a small region of similarity to the X chromosome so that they can pair during meiosis, the Y chromosome is much shorter and contains many fewer genes. When a gene is present on the X chromosome, it is said to be X-linked.

11- Human Sex-linked Disorders

Sex-linkage studies in Morgan's laboratory provided the fundamentals for understanding X-linked recessive disorders in humans, which included red-green color blindness, Types A and B hemophilia, and muscular dystrophy. Because human males need to inherit only one recessive mutant X allele to be affected, X-linked disorders are disproportionately observed in males.

Females must inherit recessive X-linked alleles from both of their parents in order to express the trait. When they inherit one recessive X-linked mutant allele and one dominant X-linked wild-type allele, they are carriers of the trait and are typically unaffected. Carrier females can manifest mild forms of the trait due to the inactivation of the dominant allele located on one of the X chromosomes. However, female carriers can contribute the trait to their sons, resulting in the son exhibiting the trait, or they can contribute the recessive allele to their daughters, resulting in the daughters being carriers of the trait. Although some Y-linked recessive disorders exist, typically they are associated with infertility in males and are therefore not transmitted to subsequent generations.

12- Lethal Alleles are Apparent Exceptions to the Principle of Segregation

Some genes – cause death during early stage of development (before sexual maturation).

Types of lethal alleles

Lethal alleles falls into four categories.

1. Early onset- lethal alleles which result in death of an organism at early stage of life for example during embryogenesis.

2. Late onset- lethal allele which kills organism at their final stage of life are known as late onset allele.

3. Conditional- lethal allele which kill organism underbcertain environmental conditions only.

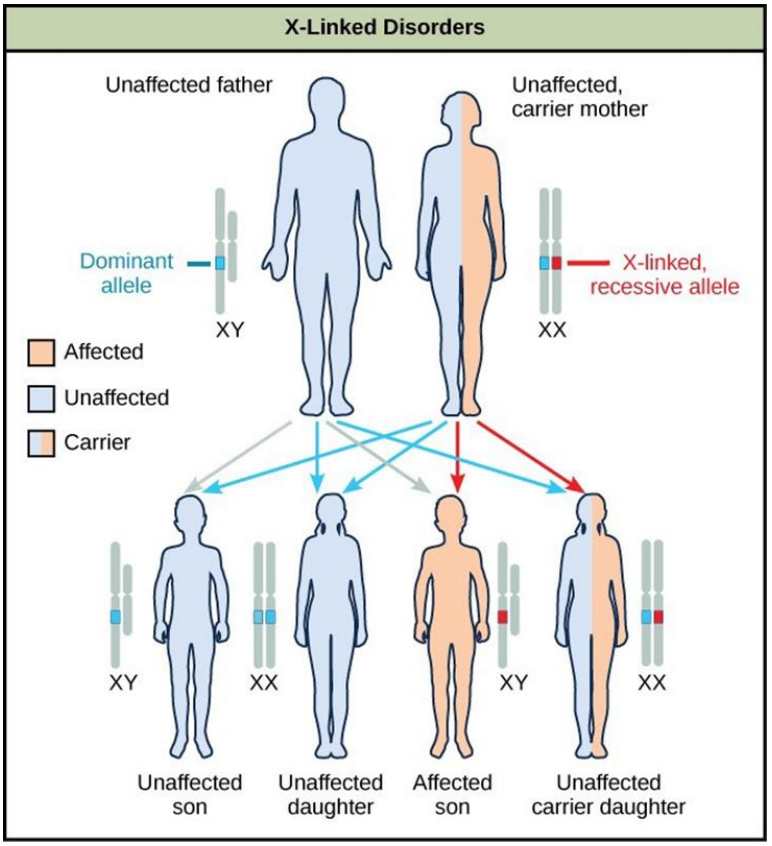


Fig. (16). The son of a woman who is a carrier of a recessive X-linked disorder will have a 50 percent chance of being affected. A daughter will not be affected, but she will have a 50 percent chance of being a carrier like her mother.

https://www.nature.com/scitable/topicpage/environment-controls-gene-expression-sex-determination-and-982/

e.g. temperature sensitive alleles kills organism at high temperature. But they don't kill any organism at low temperature.

4. Semi lethal – Lethal allele which kill only some individuals of the population but not all are know as semi lethal.

5-Most of all lethal genes are recessive (death only homozygous).

A large proportion of genes in an individual's genome are essential for survival.

LETHAL MUTATIONS

- Alleles that cause an organism to die only when present in homozygous condition are called lethal alleles.

- Lethal alleles are often inherited as recessive mutants, recessive lethal alleles that kill only homozygotes.

- Lethal alleles are often detected as distortions in segregation ratios, where one or more classes of expected progeny are missing.

essential gene can arise by mutation and be transmitted in a population through heterozygous carriers. The wild-type allele functions at a capacity sufficient to sustain life and is therefore considered to be dominant over the nonfunctional allele If two heterozygous parents mate, one quarter of their offspring will be

homozygous recessive. Because the gene is essential, these individuals will die. This will cause the genotypic ratio among surviving offspring to be 2:1 rather than 3:1. This inheritance pattern is referred to as **recessive lethal**.

The **dominant lethal** inheritance pattern is one in which an allele is lethal both in the homozygote and the heterozygote. Dominant lethal alleles are very rare because, as you might expect, the allele only lasts one generation and is not transmitted. However, dominant lethal alleles might not be expressed until adulthood. The allele may be unknowingly passed on, resulting in a delayed death in both generations. An example of this in humans is Huntington disease, in which the nervous system gradually wastes away. People who are heterozygous for the dominant Huntington allele (*Hh*) will inevitably develop the fatal disease. However, the onset of Huntington disease may not occur until age 40, at which point the afflicted persons may have already passed the allele to 50 percent of their offspring.

- Example coat colour gene in mice. 3:1 (viable : dead)

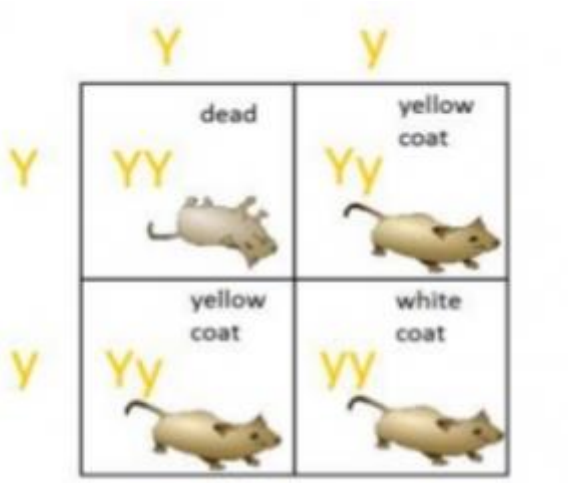


Fig. (17). coat colour gene in mice

https://plantlet.org/exceptions-of-mendelism-first-law/

13- Linked Genes Violate the Principle of Independent Assortment

Homologous chromosomes possess the same genes in the same order. However, since each homolog came from a different parent, the alleles may differ on homologous chromosome pairs. Prior to meiosis I, homologous chromosomes replicate and synapse so that genes on the homologs align with each other. At this stage, segments of homologous chromosomes cross over and exchange segments of genetic material . Because the genes are aligned, the gene order is not altered. Instead, the result of

recombination is that maternal and paternal alleles are combined onto the same chromosome. Across a given chromosome,

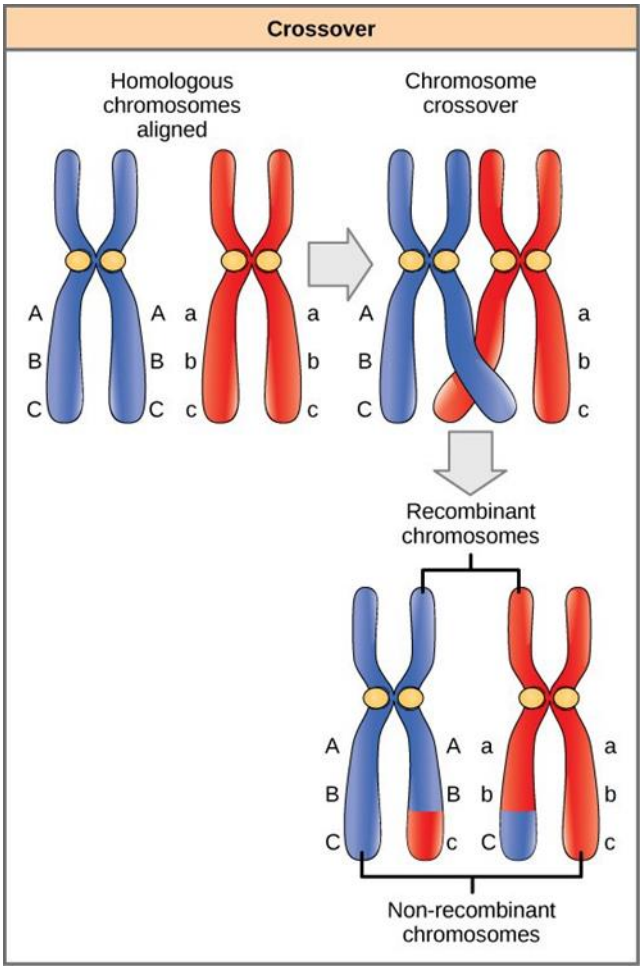


Fig. (18). The process of crossover, or recombination, occurs when two homologous chromosomes align during meiosis and exchange a segment of genetic material. Here, the alleles for gene C were exchanged. The result is two recombinant and two non-recombinant chromosomes.

several recombination events may occur, causing extensive shuffling of alleles. When two genes are located in close proximity on the same chromosome, their alleles are more likely to be transmitted through meiosis together. To exemplify this, imagine a dihybrid cross involving flower color and plant height in which the genes are next to each other on the chromosome. If the homologous chromosome from one parent has alleles for tall plants and red flowers, and the homolog from the other parent has alleles for short plants and yellow flowers, then when the gametes are formed, the tall and red alleles will go together into a gamete and the short and yellow alleles will go into other gametes. These are called the parental genotypes

because they have been inherited intact from the parents of the individual producing gametes. Since the genes were close together on the same chromosomes, the chance of a crossover event happening between them is slim. Therefore, there will be no gametes with tall and yellow alleles and no gametes with short and red alleles. If you create the Punnett square with these gametes, you will see that the classical Mendelian prediction of a 9:3:3:1 outcome of a dihybrid cross would not apply.

As the distance between two genes increases, the probability of crossovers between them increases, and the genes behave more as if they are on separate chromosomes. The further apart two linked genes are on a chromosome, the more progeny with

non parental genotypes will appear.

https://openoregon.pressbooks.pub/mhccmajorsbio/chapter/linked-genes-violate-the-law-of-independent-assortment/

Questions

1. After cross-fertilization of true-breeding tall and dwarf plants, the F_1 generation was self-fertilized. The resultant plants have genotype in the ratio

(a) 1:2:1 (homozygous tall : heterozygous tall : dwarf)

(b) 1:2:1 (heterozygous tall : homozygous tall : dwarf)

(c) 3:1 (tall : dwarf)

(d) 3:1 (dwarf : tall)

2. Which of the following characteristics of pea plants was not used by Mendel in his experiments?

(a) seed colour

(b) seed shape

(c) pod length

(d) flower position

3. Mendel took _____ contrasting characteristics of pea plants.

(a) eight

(b) seven

(c) six

(d) five

4. If both genotype and phenotype shows the same ratios of 1:2:1 in the F_2 generation, it shows

(a) incomplete dominance in monohybrid cross

(b) complete dominance in monohybrid cross

(c) dihybrid cross

(d) co-dominance

5. Test cross determines

(a) whether two traits are linked or not

(b) the genotype of plant

(c) whether the two species will breed successfully or not

(d) number of alleles in a gene

6. Genotype of dominant plant can be determined by

(a) pedigree analysis

(b) back cross

(c) test cross

(d) dihybrid cross

7. Test cross is a

(a) cross between two recessive homozygotes

(b) cross between dominant homozygote and heterozygote

(c) cross between two F_1 hybrids

(d) cross between an F_1 hybrid and recessive homozygote

8. Lack of independent assortment of two genes is due to

(a) recombination

(b) crossing over

(c) linkage

(d) repulsion

9. Which of the following is a recessive trait in pea plants?

a) Dwarf stem height

b) Violet flowers

c) Axial flowers

d) Inflated pods

10. Round and wrinkled seeds were one of the contrasting traits used by Mendel to devise the laws of inheritance.

a) True

b) False

11. If you were to sample garden pea plants in Mendel's garden, which of the following statements would hold?

a) Round seeds were more abundant than wrinkled seeds

b) Wrinkled seeds were more abundant than round seeds

c) Both round and wrinkled seeds were equally abundant

d) Answer depends on the time of day when sampling is done

12. What was the model organism used by Mendel to give the laws of inheritance?

a) Garden peas

b) Wild peas

c) Basket peas

d) Bottle gourd

13. Which of the following is not a pair of contrasting traits studied by Mendel?

a) Green and yellow pods

b) Full and constricted pods

c) Axial and terminal flowers

d) Pink and white flowers

14. Which of the following is a dominant character in pea plants?

a) Tall stem height

b) White flowers

c) Terminal flowers

d) Constricted pods

Second chapter

Population genetics

Population Genetics

Population genetics is a field of biology that studies the genetic composition of biological populations, and the changes in genetic composition that result from the operation of various factors, including natural selection. Population geneticists develop abstract mathematical models of gene frequency dynamics, extract predictions about the likely patterns of genetic variation in actual populations, and test the predictions against empirical data. A number of the more robust generalizations to emerge from population-genetic analysis are discussed below.

Population genetics is intimately bound up with the study of evolution and natural selection, and is often regarded as the theoretical cornerstone of evolutionary biology.

gene pool

The gene pool is the set of all genes, or genetic information, in any population, usually of a particular species. Gene pool is the collection of different genes in a population of a particular species at a given time. The gene pool definition includes all the genes and combinations of genes in the population. The gene

pool term includes the sum of all the alleles of genes present at all of the loci within a population of a single species. Thus, the gene pool definition is the term used to describe the set of all the genes, or genetic information in any population that too of a particular species.

Description

A large gene pool indicates extensive genetic diversity, which is associated with robust populations that can survive bouts of intense selection. Meanwhile, low genetic diversity (see inbreeding and population bottlenecks) can cause reduced biological fitness and an increased chance of extinction, although as explained by genetic drift new genetic variants, that may cause an increase in the fitness of organisms, are more likely to fix in the population if it is rather small.

When all individuals in a population are identical with regard to a particular phenotypic trait, the population is said to be 'monomorphic'. When the individuals show several variants of a particular trait they are said to be polymorphic.

The Russian geneticist Alexander Sergeevich Serebrovsky first formulated the concept in the 1920s as genofond (gene fund), a word that was imported to the United States from the Soviet Union by Theodosius Dobzhansky, who translated it into English as “gene pool.

Changes in the Gene Pool

The changes in the gene pool can cause changes in the genetic diversity of the population of the species as well. The composition of the gene pool can change over time through processes that govern evolution. A variety of mechanisms such as mutation, natural selection and genetic drift can cause changes in the composition of the gene pool. These changes are essential for the survival of any population of species with the changes in the environment. A diverse gene pool gets created through these genetic variations which make the individuals in the population adaptive to the changing environment. Such an example of change can be seen with the changes in the human gene pool as well. When the human population migrated from the equatorial regions towards the northern climates, there was a change over time in skin pigmentation. When the human population was exposed to relatively low sunlight the color of the skin changed to a lighter color for increasing Vitamin D absorption. The genetic modifications that occurred due to changes in the environment then became a part of the human gene pool in that particular region.

The Origins of Population Genetics

The Mendelian clan is a group of organisms that share certain characteristics and reproduce sexually and have a close genetic relationship (such as a single species, variety, strain...etc), and are located within a specific geographical area in which mating takes place. The Mandalian clan is the clan that consists of a large group of individuals, i.e. a large clan, in which sexual mating occurs randomly, and from this it becomes clear to us that plants that reproduce vegetatively, i.e. that reproduce asexually, are not considered to be Mendelian clans. Here we must refer to the species, which is considered the largest Mendelian clan, because within it mating occurs between individuals fluently, and these individuals share with each other a genetic pool, where the species is divided into several Mendelian clans, and each clan can contain many sub-clans population and in natural clans, the clan members differ from each other in terms of genotype and phenotypic form Gene pool: Gene pool is all the gametes of a Mendelian population of a particular hereditary trait to produce the second generation. Accordingly, in any clan, when it is intended to describe the genotypes of a group of individuals, it must:

1- Describe and identify their genotypes.

2- We determine the frequency of these genotypes

♂ + ♀ = p + q = 1

where A=p , q=a

The expected frequencies of the genotypes of the heterozygous distribution of the second generation

($P^2AA + 2PqAa + q^2aa$)

$(P + q)^2=q^2+2pq+p^2= 1$ (AA, 2Aa, aa)

$(A + a)^2 = AA + 2Aa + aa = 1$

This formula, which expresses the expectation of the genetic frequencies of the new offspring in the Mendelian population in terms of gamete frequencies, is called the Hardy-Weinberg Law, and this population is called in the case of equilibrium in the case of the stability of the frequency of alleles A, a through generations. This hypothesis is based on the existence of several conditions or foundations for achieving or maintaining equilibrium in the clan:

1- The clan is large and mating is random. Random Mating

2- The chances of survival are equal for all gametes and individuals, and that each genotype has equal effectiveness for producing offspring.

3- The clan is closed, meaning there is no migration of individuals to and from the clan.

4- There is no mutation or reverse mutations that change the frequency of genes.

5- Chance is the only factor in the formation of gametes (their quality) and their union to form the fertilized egg.

https://en.wikipedia.org/wiki/Population_genetics

Hardy vienbreg statistics and its applications

In 1908, the English scientists G.H. Hardy, a mathematician and German scientist W. Weinberg, a physicist, refers each separately to the following rule or law: (In the large mated clan they lived and in the absence of forces that change the replication of the gene (selection, mutations and migration). The clan remains constant from one generation to the next) and this clan is called Equilibrium This law is considered the basis on which the science of clan inheritance is built The genetic makeup of the individuals in the large clan that mate randomly with their frequency is:

(AA Aa aa)

(P^2 2Pq q^2)

What Is Hardy Weinberg Law?

“In a large, random-mating population, the genotype and allele frequencies remain constant in the absence of any evolutionary influences from one to another generation. Influences are inclusive of a choice of mate, natural selection, genetic drift, mutation, sexual selection, gene flow, genetic hitchhiking, founder effect, meiotic drive, population bottleneck, inbreeding and assortative mating.”

Genotype frequencies and allele frequencies are related to each other in a way that it is the square expansion of such allele frequencies. In other words, the law conveys that in a population, it is possible to estimate the expected frequencies of genotypes under a certain limited set of assumptions, provided the frequency of different alleles in a population is already known.

Suppose that at a given locus, or chromosomal ‘slot’, there are two possible alleles, A_1 and A_2; the locus is assumed to be on an autosome, not a sex chromosome. With respect to the locus in question, there are three possible genotypes in the population, A_1A_1,A_1A_2 and A_2A_2 (just as in Mendel’s pea plant example). Organisms with the $A_1A_1$and A_2A_2 genotypes are called homozygotes; those with the A_1A_2 genotype are *heterozygotes*. The proportions, or relative frequencies, of the three genotypes in the overall population may be denoted $f(A_1A_1)$,$f(A_1A_2)$ and $f(A_2A_2)$ respectively,

where $f(A_1A_1)+f(A_1A_2)+f(A_2A_2)=1$. It is assumed that these genotypic frequencies are the same for both males and females. The relative frequencies of the A and B alleles in the population are denoted p and q, where $p=f(A_1A_1)+1/2f(A_1A_2)$ and $q=f(A_2A_2)+1/2f(A_1A_2)$. Note that $p+q=1$.

The Hardy-Weinberg principle is about the relation between the allelic and the genotypic frequencies. It states that if mating is random in the population, and if natural selection, mutation, migration and drift are absent, then in the offspring generation the genotypic and allelic frequencies will be related by the following simple equations:

$f(A_1A_1)=p^2$,

$f(A_1A_2)=2pq$,

$f(A_2A_2)=q^2$

That random mating will lead the genotypes to be in the above proportions (“Hardy-Weinberg proportions”) is a consequence of Mendel’s law of segregation. To see this, note that random mating is in effect equivalent to offspring being formed by randomly picking pairs of gametes from a large ‘gamete pool’ and fusing them into a zygote. The gamete pool contains all the successful gametes of the parent organisms. Since we are assuming the absence of selection, all parents contribute equal numbers of gametes to the pool. By the law of segregation,

an A_1A_2 heterozygote produces gametes bearing the A_1 and A_2 alleles in equal proportion (on average). Therefore, the relative frequencies of the A and B alleles in the gamete pool will be the same as in the parental population, namely p and q respectively. Given that the gamete pool is very large, when we pick pairs of gametes from the pool at random, we will get the ordered genotypic pairs $\{A_1A_1\}$, $\{A_1A_2\}$,$\{A_2A_1$, $\{A_2A_2\}$ in the proportions p^2 : pq : qp:q^2. But order does not matter, so we can regard the $\{A_1A_2\}$ and $\{A_2A_1\}$ pairs as equivalent, giving the Hardy-Weinberg proportions for the unordered offspring genotypes.

This simple derivation of the Hardy-Weinberg principle deals with two alleles at a single locus, but can easily be extended to multiple alleles. (Extension to more than one locus is trickier) For the multi-allelic case, suppose there are n alleles at the locus, $A_1 \ldots A_n$, with relative frequencies of $p_1 \ldots p_n$ respectively, where $p_1+p_2+\ldots+p_n=1$.

Other ex.

Take a case of a single locus with only two alleles indicated by A and a with corresponding frequencies f(A) = p and f(a) = q respectively, then the genotype frequencies that can be expected under limited condition being random mating is

f(AA)= p^2 for AA homozygotes

$f(aa) = q^2$ for aa homozygotes

f(Aa) = 2pq for heterozygotes

The Hardy Weinberg Equation can be represented by

$$\mathbf{p^2 + q^2 + 2pq = 1}$$

The allele frequencies p and q remain constant in the absence of any kind of influences such as mutation, natural selection, genetic drift, etc from one to another generation. This is how the equilibrium can be reached.

Assumptions for the Hardy Weinberg Principle

- Only sexual reproduction can take place
- Process of mating is random
- The size of the population is indefinitely large
- Entities are diploid
- Generations do not overlap
- Equality of allele frequencies in terms of sexes

- No traces of gene flow, selection, mutation, migration or admixture

In case there is any breach with regard to the above-mentioned assumptions, it can lead to discrepancies from the expected outcome. The consequences are completely dependent on the deduction that has been digressed.

The law mentions that a population shall have Hardy Weinberg proportions (given genotypic frequencies) once a single generation of random mating is carried out. In case the assumption of random mating is breached, this population will not possess Hardy Weinberg proportions. The most common source of a non-random mating is inbreeding. It leads to the rise in the homozygosity of all genes.

The most important factors affecting allelic frequency and Hardy-Weinberg equilibrium

1- Mutation

If the T → t gene mutates, then the frequencies of the two alleles T and t will change, and if the T → t mutation occurs continuously, this will inevitably lead to the disappearance of the T gene from the population, and thus the population balance will be disturbed, but the presence of a recurrent mutation from t → T will lead to rebalance in the clan.

2- Migration

Migration means the movement of individuals from one clan to another and the occurrence of a process Random interbreeding between the two clans, which leads to the entry of the clan's genes Migrant to the original clan, the speed of change in genetic replication in any clan is subject to the entry of migratory elements depends on the speed of migration and on the genetic differences between immigrants and the original clan, and thus migration leads to breaking the mechanism of isolation that was enjoyed by the original clan, which affects the proportion of genetic repetitions Increase or decrease from the original limit.

3- Selection at One Locus

Natural selection occurs when some variants in a population enjoy a survival or reproductive advantage over others. The simplest population-genetic model of natural selection focuses on a single autosomal locus with two alleles, A_1 and A_2, in a large population. Random mating is assumed. The three diploid genotypes A_1A_1, A_1A_2 and A_2A_2 have different fitnesses, denoted by w_{11}, w_{12} and w_{22} respectively. These fitnesses are assumed to be constant across generations. A genotype’s fitness may be defined, in this context, as the average number of successful gametes that an organism of that genotype contributes to the

next generation—which depends on how well the organism survives, how many matings it achieves, and how fertile it is. Unless w_{11}, w_{12} and w_{22} are all equal, then natural selection will occur, which may lead the genetic composition of the population to change.

Suppose that initially, i.e., before selection has operated, the zygote genotypes are in Hardy-Weinberg proportions and the frequencies of the A_1 and A_2 alleles are pp and qq respectively, where p+q=1. The zygotes then grow to adulthood and reproduce, giving rise to a new generation of offspring zygotes. Our task is to compute the frequencies of A_1 and A_2 in the second generation; let us denote these by p′ and q′ respectively, where p′+q′=1. (Note that in both generations, we consider gene frequencies at the zygotic stage; these may differ from the adult gene frequencies if there is differential survivorship).

In the first generation, the genotypic frequencies at the zygotic stage are p^2,,2pq and q^2 for A_1A_1, A_1A_2, A_2A_2 respectively, by the Hardy-Weinberg principle. The three genotypes produce successful gametes in proportion to their fitnesses, i.e., in the ratio w_{11}:w_{12}:w_{22}w. The average fitness in the population is $w=p^2w_{11}+2pqw_{12}+q^2w_{22}$. Assuming there is no mutation, and that Mendel's law of segregation holds, then an

A_1 A_1 organism will produce only A_1 gametes, an A_2 A_2 organism will produce only A_2 gametes, and an A_1A_2 organism will

produce A_1 and A_2 gametes in equal proportion (on average). Therefore, the proportion of A_1 gametes, and thus the frequency of the A_1 allele in the second generation at the zygotic stage, is:

Equation (1)

$$p' = p^2 w_{11} + 1/2(2pq\, w_{12}) / w$$

$$= p^2 w_{11} + pq\, w_{12} / w$$

Equation (1) is known as a 'recurrence' equation—it expresses the frequency of the A_1A_1 allele in the second generation in terms of its frequency in the first generation. The change in frequency between generations can then be written as:

Equation (2)

$$\Delta p = p' - p$$

$$= p^2 w_{11} + pq\, w_{12} / w - p$$

$$= pq\,[p(w_{11} - w_{12}) + q(w_{12} - w_{22})] / w$$

If $\Delta p > 0$, then natural selection has led the A_1 allele to increase in frequency; if $\Delta p < 0$ then selection has led the A_2 allele to increase in frequency. If $\Delta p = 0$ then no gene frequency change has occurred, i.e., the system is in allelic equilibrium. (Note, however, that the condition $\Delta p = 0$ does *not* imply that no natural selection has occurred; the condition for that is w11=w12=w22. It is possible for natural selection to occur but to have no effect on gene frequencies).

Equations (1) and (2) show, in precise terms, how fitness differences between genotypes will lead to evolutionary change. This enables us to explore the consequences of various different selective regimes.

Suppose firstly that w11>w12>w22, i.e., the A_1A_1 homozygote is fitter than the A_1A_2 heterozygote, which in turn is fitter than the A_2A_2 homozygote. By inspection of equation (2), we can see that Δp must be positive (so long as neither pp nor qq is zero). So in each generation, the frequency of the A_1 allele will be greater than in the previous generation, until it eventually reaches fixation. Once the A_1 allele reaches fixation, i.e., p=1 and q=0, no further evolutionary change will occur, for if p=1 then Δp=0. This makes good sense intuitively: since the A_1 allele confers a fitness advantage on organisms that carry it, its relative frequency in the population will increase from generation to generation until it is fixed.

It is obvious that analogous reasoning applies in the case where w22>w12>w11. Equation (2) tells us that Δp must then be negative, so long as neither pp nor q is zero, so the A_2 allele will sweep to fixation.

A more interesting situation arises when the heterozygote is superior in fitness to both of the homozygotes, i.e., w12>w11 and w12>w22—a phenomenon known as *heteroygote superiority*. Intuitively it is clear what should happen in this

situation: an equilibrium situation should be reached in which both alleles are present in the population. Equation (2) confirms this intuition. It is easy to see that $\Delta p=0$ if either allele has gone to fixation (i.e., if $p=0$ or $q=0$), or, thirdly, if the following condition obtains:

$$p(w_{11}-w_{12}) + q(w_{12}-w_{22})= 0$$

which reduces to

$$p=p^{*}=(w_{12}-w_{22}) / (w_{12}-w_{22}) + (w_{12}-w_{11})$$

The asterisk indicates that this is an equilibrium condition.) Since pp must be non-negative, this condition can only be satisfied if there is heterozygote superiority or inferiority; it represents an equilibrium state of the population in which both alleles are present. This equilibrium is known as *polymorphic*, by contrast with the *monomorphic* equilibria that arise when either of the alleles has gone to fixation. The possibility of polymorphic equilibrium is quite significant. It teaches us that natural selection will not always lead to homogeneity; in some cases, selection preserves the genetic variation found in a population.

Numerous evolutionary questions can be addressed using simple population-genetic models of this sort. For example, by incorporating a parameter which measures the fitness differences between genotypes, we can study the *rate* of evolutionary change, permitting us to ask questions such as:

how long will it take for selection to increase the frequency of the A_1A_1 allele from 0.1 to 0.9? If a given deleterious allele is recessive, how much longer will it take to eliminate it from the population than if it were dominant? In this way, population genetics converted the theory of evolution into a quantitatively precise one.

The one-locus model outlined above is unlikely to apply to many real-life populations, due to the simplifying assumptions it makes. In reality, selection is rarely the only evolutionary force in operation, genotypic fitness's are unlikely to be constant across generations, Mendelian segregation does not always hold exactly, and not all evolving populations are large. Much effort in population genetics has been put into making more realistic models which relax these assumptions and are thus more complicated. But the one-locus model illustrates the essence of the population-genetic analysis of evolutionary change.

Genetic Drift

In small or geographically isolated clans, any major external change such as floods or forest fires will lead to a sudden and rapid change of genetic frequencies in that clan in favor of the replication of individuals carrying a particular form of the gene at the expense of others. Therefore, clan genetics is the science

that is concerned with studying the distribution and change in allele frequency under the influence of 4 evolutionary forces affecting gene replication, namely:

1- Natural selection

2- Genetic drift

It is an evolutionary force that changes the characteristics of species over time. The effect of genetic drift increases as the number of copies of an allele decreases, and decreases when there are many copies of an allele.

3- Mutation

4- Migration

https://byjus.com/neet/hardy-weinberg-law/

Hardy-Weinberg equilibrium

Random mating is achieved in an animal herd when each individual has the same opportunity to mate with any other

individual. It was discovered by the Englishman G.Hardy and the German W. Weinberg. A relationship between gene frequencies and genotypic frequencies is known as the Hardy-Weinberg law (or equilibrium) and states the stability of gene frequencies and genetic frequencies from one generation in which mating is random to the next generation, in a large herd, and in the absence of forces affecting gene frequencies They are the factors that work on the instability of the relative genetic frequency.

Example:

A given gene has two alleles, A and a

Let it be = N 1000 clan consisting of a thousand individuals

and the frequency of A is p = 0.7

and the frequency of a is q = 0.3

1= p+q

The frequency of the possible genotypes within it would be:

$f(aa)=q^2$ $f(AA)=p^2$ $f(AA)=2pq$

So that the hardy weinberg balance states p2+2pq +q2=1

The frequency of alleles and genotypes remains constant from generation to generation according to this law, and this means that the clan does not evolve and does not know any genetic change and is called the balanced clan.

The Population clan is a group of living individuals that share certain characteristics, and the inheritance of the clans in general is concerned with the study of the Mendelian Population. Sexual mating is random mating, and from this it becomes clear to us that plants that reproduce vegetatively and that reproduce asexually are not considered to be Mendelian clans. Here we must refer to the species Species, which is the largest Mendelian clan because within it mating occurs between individuals fluently and these individuals share with each other a warehouse Gene Pool where the species is divided into several Mendelian clans, and each clan can contain many sub-populations.

So Species is the extreme extension of the Mendelian clan, which defines a range Vaccinations occur. Usually mating does not occur between individuals belonging to different species, and therefore, the name of the Mendelian clan is not given to the group of individuals belonging to different species. What is in the science of plant breeding, the word clan can be applied to the members of the first generation and the generations following it in a cross, so it is said that the first generation clan is F1population (F stands for the word Filal, which means succession after the generation of parents), the second generation clan F2, etc... In the natural clans of living organisms, the members of the clan differ from each other in terms of genotype and phenotypic form, and accordingly, in any clan

when it is intended to describe the genotypes of a group of individuals The following must be done:

1- Identify and describe the different genotypes of the clan members.

2- Determine the frequency of each genotype of these different frequencies of genotypes

https://en.wikipedia.org/wiki/Hardy%E2%80%93Weinberg_principle

The concept of gene frequency:

The concept of gene redundancy in the case of a single locus in an allele located on an autosomal chromosome. Suppose that we are studying a specific trait from a population that is controlled by one pair of genes, and suppose that the two alleles are a and A, and suppose that the number of members of this population is N, so that the number of individuals with recessive structure aa is N. And the number of individuals with the dominant structure AA is N_2 and the number of individuals with the dominant hybrid structure Aa is N_1. If we denote the percentage of genotype AA by D

It is $D = N_2 / N$

And the ratio of genotype Aa to H means that $H = N_1 / N$

For the ratio of genotype aa to R, then $R = N_0 / N$

So this clan can be written as follows:

Total	aa	Aa	AA	Types of genotypes
N	N0	N1	N2	The number of genotypes
1	R	H	D	Proportion of genotypes

We denote the replication of gene A by the symbol P i.e. P = f (A) Frequency of gene A = number of gene A in the population / The total number of genes in the population

That is, $P = f(A) = D + \frac{1}{2} H$

any gene duplication A = the proportion of the original dominant individuals + half the proportion of the mixed individuals.

And one: $P = f(A) = N_2 + \frac{1}{2} N_1 / N$

We also denote the replication of the gene a by the symbol q, i.e.

q = f (a) , i.e.: gene replication a = the proportion of the original recessive individuals + half of the proportion of the mixed individuals.

That is, $q = f(a) = R + \frac{1}{2} H$

and that $q = f(a) = \frac{1}{2} N_1 + N_0 / N$

The frequency of any gene is nothing but the frequency or presence of that gene in the population

The frequency of a genotype is defined as the percentage of this genotype among individuals, so the sum of the proportions or frequencies of genotypes for a given genotype equals one or 100%,

$p + q = 1$

Therefore, the genetic frequency of any gene ranges from zero to one.

$0 \leq p \leq 1$

If the proportion of the gene is rare, then its value is close to zero, but if the gene is abundant in the population, then the value is close to one.

Example: Calculate the frequency of gene A and the frequency of gene a in the following population:

AA Aa aa

363 634 282

N_2 N_1 N_0

Solution: The number of members of this clan is: N 363 + 634 + 282 = 1279

Since the genotypes are given in the form of numbers, then:

$P = f(A) = N_2 + ½N_1 / N$

$=363 + ½ (634) /1279 = 0.53$

$q = f(a) = ½ N_1 + N_0 / N$

$= ½ (634) + 282 / 1279 = 0.47$

$q = f(a) = 1–p = 1 - 0.53 = 0.47$

Example: Calculate the frequency of genes A and a in a population consisting of:

36% of the individuals are autosomal dominant AA

48% of the individuals are Aa mixture individuals

16% of the individuals are aa recessive individuals

Meaning: AA Aa aa

0.36 0.48 0.16

The solution:

Since the clan was given in the form of lineages, then:

$P = f(A) = D + ½ H$

$= 0.36 + ½ (0.48) = 0.6$

$q = f(a) = 1 - p = 1 - 0.6 = 0.4$

https://en.wikipedia.org/wiki/Allele_frequency

Applications of the Hardy Weinberg Principle

I. Complete Dominance

Allele frequencies can be detected in the presence of complete dominance when Hardy-Weinberg equilibrium prevails wherein it is not possible to differentiate between two genotypes. Two genotypes AA and Aa having the same phenotype as a result of complete dominance of A over a can help determine the allele frequencies from frequencies of the individuals indicating recessive phenotype aa. Here, the frequency of aa individual should be equivalent to the square of the frequency of the recessive allele.

II. Multiple Alleles

Calculation of genotypic frequencies at a locus with more than two alleles is allowed in the Hardy Weinberg principle, for instance in the ABO blood groups. Three alleles are present in IA, IB, IC with p,q and r frequencies respectively where p + q + r = 1. With random mating, the genotype of a population will be given by $(p + q + r)^2$

III. Linkage Disequilibrium

Take, for instance, two or more alleles on the same chromosome, at two different loci with 2 or more alleles. As a result of genetic exchange by recombination taking place at regular time intervals, at two syntenic loci, the frequency of allelic combinations attains equilibrium.

In the event of not being able to attain an equilibrium, alleles are known to be in a linkage disequilibrium, which is as a result of two or more linked alleles to be inherited jointly, more frequently than expected. Such gene groups are also known as supergenes.

IV. Frequencies of Harmful Recessive Alleles

The law can also be applied to estimate the frequency of heterozygous carriers of recessive genes that are harmful. In a population, two alleles, A and a are at an autosomal locus with p and q frequencies respectively, and p + q = 1, then AA, Aa and aa genotypes will have the following frequency, $p^2 + q^2 + 2pq$. In case, the aa genotype tends to express a phenotype that is harmful,

such as cystic fibrosis, then in the population, the proportion of the affected individuals shall be q^2, the recessive allele frequency of the heterozygous carrier shall be 2pq.

Summary

- In a given population, the Hardy Weinberg principle assumes that the population is indefinite and not influenced by sexual, natural selection, mutation and migration.
- Frequency of alleles can be calculated by the frequency of recessive genotypes. Then estimate the square root of this frequency to find the frequency of the recessive allele
- In a population, the frequency of alleles can be indicated by $p + q = 1$, with p = frequency of the dominant allele and q = frequency of the recessive allele.
- In a population, the frequency of alleles can be indicated by $p^2 + q^2 + 2pq = 1$, where p^2 is the frequency of homozygous dominant genotype, q^2 is

the frequency of recessive genotype and 2pq is the frequency of heterozygous genotype.

https://byjus.com/neet/hardy-weinberg-law/

The five assumptions of Hardy Weinberg equilibrium are:

1- Random mating

2- No mutation

3- No natural selection

4- No gene flow or migration

5- A very large population size (no genetic drift)

What does the Hardy Weinberg law mean?

Hardy Weinberg law states that genetic variations remain constant in a large, randomly mating population. The frequency of alleles and genotypes remains constant from generation to generation and the population exists in a genetic equilibrium if there are no disturbances such as mutation, migration, natural selection, etc.

Why is the Hardy Weinberg principle important?

The Hardy Weinberg principle is important in analysing the genetic variation existing in a population and comparing the actual variation to the calculated value from Hardy Weinberg law if the population was in equilibrium. If the actual frequency in a population differs from the expected value then it is an indication of disturbance and violation of one or more assumptions, which can further be investigated. It also helps in estimating the frequency of the heterozygous carriers of a harmful recessive gene.

What do p and q stand for in the Hardy Weinberg equation?

In the Hardy Weinberg equation (p2 + q2 + 2pq = 1), p is the frequency of the dominant allele and q is the frequency of the recessive allele for a gene controlled by a pair of alleles.

https://www.sciencedirect.com/topics/neuroscience/hardy-weinberg-principle

Gene polymorphism

A gene is said to be polymorphic if more than one allele occupies that gene's locus within a population. In addition to having more than one allele at a specific locus, each allele must

also occur in the population at a rate of at least 1% to generally be considered polymorphic.

Gene polymorphisms can occur in any region of the genome. The majority of polymorphisms are silent, meaning they do not alter the function or expression of a gene. Some polymorphisms are visible. For example, in dogs the E locus can have any of five different alleles, known as E, E^m, E^g, E^h, and e. Varying combinations of these alleles contribute to the pigmentation and patterns seen in dog coats.

Fig. (19). Genes which control hair colour are polymorphic.

A polymorphic variant of a gene can lead to the abnormal expression or to the production of an abnormal form of the protein; this abnormality may cause or be associated with disease. For example, a polymorphic variant of the gene encoding the enzyme CYP4A11, in which thymidine replaces cytosine at the gene's nucleotide 8590 position encodes a CYP4A11 protein that substitutes phenylalanine with serine at

the protein's amino acid position 434.[6] This variant protein has reduced enzyme activity in metabolizing arachidonic acid to the blood pressure-regulating eicosanoid, 20-hydroxyeicosatetraenoic acid. A study has shown that humans bearing this variant in one or both of their CYP4A11 genes have an increased incidence of hypertension, ischemic stroke, and coronary artery disease.

Most notably, the genes coding for the major histocompatibility complex (MHC) are in fact the most polymorphic genes known. MHC molecules are involved in the immune system and interact with T-cells. There are more than 32,000 different alleles of human MHC class I and II genes, and it has been estimated that there are 200 variants at the HLA-B HLA-DRB1 loci alone.

Some polymorphism may be maintained by balancing selection.

Identification

Polymorphism, as related to genomics, refers to the presence of two or more variant forms of a specific DNA sequence that can occur among different individuals or populations. The most common type of polymorphism involves variation at a single nucleotide (also called a single-nucleotide polymorphism, or SNP). Other polymorphisms can be much larger, involving longer stretches of DNA.

Polymorphisms can be identified in the laboratory using a variety of methods. Many methods employ PCR to amplify the sequence of a gene. Once amplified, polymorphisms and mutations in the sequence can be detected by DNA sequencing.

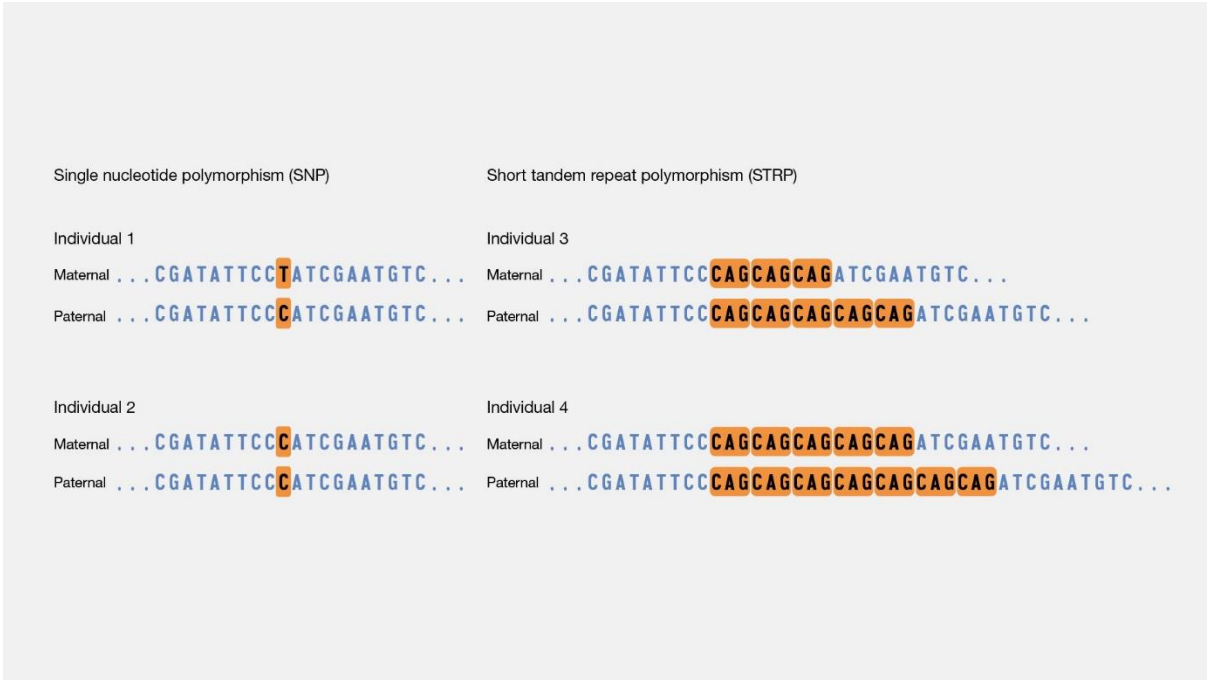


Fig. (20). Polymorphism, refers to the presence of two or more variant forms of a specific DNA sequence

Clinical significance

Lung cancer

Polymorphisms have been discovered in multiple XPD exons. XPD refers to "xeroderma pigmentosum group D" and is

involved in a DNA repair mechanism used during DNA replication. XPD works by cutting and removing segments of DNA that have been damaged due to things such as cigarette smoking and inhalation of other environmental carcinogens. Asp312Asn and Lys751Gln are the two common polymorphisms of XPD that result in a change in a single amino acid. This variation in Asn and Gln alleles has been related to individuals having a reduced DNA repair efficiency. Several studies have been conducted to see if this diminished capacity to repair DNA is related to an increased risk of lung cancer. These studies examined the XPD gene in lung cancer patients of varying age, gender, race, and pack-years. The studies provided mixed results, from concluding individuals who are homozygous for the Asn allele or homozygous for the Gln allele had an increased risk of developing lung cancer, to finding no statistical significance between smokers who have either allele polymorphism and their susceptibility to lung cancer.

Asthma

Asthma is an inflammatory disease of the lungs and more than 100 loci have been identified as contributing to the development and severity of the condition. By using the traditional linkage analysis, these asthma correlated genes were able to be identified in small quantities using genome-wide association studies (GWAS). There have been a number of studies looking

into various polymorphisms of asthma-associated genes and how those polymorphisms interact with the carrier's environment. One example is the gene CD14, which is known to have a polymorphism that is associated with increased amounts of CD14 protein as well as reduced levels of IgE serum. A study was conducted on 624 children looking at their IgE serum levels as it related to the polymorphism in CD14. The study found that IgE serum levels differed in children with the C allele in the CD14/-260 gene based on the type of allergens they regularly exposed to. Children who were in regular contact with house pets showed higher serum levels of IgE while children who were regularly exposed to stable animals showed lower serum levels of IgE.

https://en.wikipedia.org/wiki/Gene_polymorphism

Types of Dominance

In humans most characteristics do not fit into two different phenotypes — complex traits, e.g., height, hair texture, skin colour etc., seemingly do not follow Mendelian analysis. As more scientists began analyzing genetic crosses using different types of plants and animals, it was found that while some traits obeyed Mendel’s laws (they were determined by a single gene with 1

dominant and 1 recessive allele), many other traits did not. In such cases, there were no definite recessive or dominant traits observed, or more than two alleles identified in a particular cross. In some instances, traits seem to be determined by more than one gene (multifactorial), and the environment also seemed to play a role through interaction with genes, to produce varying phenotypes.

Fig. (21). Colour, Shape, and Size of Tomatoes are Examples of Multifactorial Traits

These examples of the behaviour of certain traits implies a more complex array of interactions are at play, as these do not generate the typical Mendelian phenotypic ratios. We are

extending Mendel's Laws in order to provide explanations for the behaviour of such traits, and not necessarily challenging them.

One of the first concepts we need to understand, is that dominance is not always complete. Thus far, we have looked at the concept of dominance and recessiveness, whereby these conditions arise upon crossing two pure-breeding lines to create hybrids, and the hybrids are identical in phenotype to one parent for the particular trait in question. In this simplistic case, the allele passed down by that parent is said to be completely dominant when compared with the allele passed down by the parent whose trait is not manifested in the hybrid offspring. This type of arrangement is termed complete dominance.

As we will now see, there are two other types of Dominance — namely, incomplete dominance and co-dominance.

Complete Dominance

An example of a simple phenotype, is flower color in Mendel's peas. We have already said that one allele as a homozygote produces purple flowers, while the other allele as a homozygote produces white flowers. But what about a heterozygous individual that has one purple allele and one white allele? What is the phenotype of a heterozygote?

This can only be determined by experimental observation. We know from observation that individuals heterozygous for the purple and white alleles of the flower colour gene have purple flowers. Thus, the allele associated with purple colour is, therefore, said to be **dominant** to the allele that produces the white colour. The white allele, whose phenotype is masked by the purple allele in a heterozygote, is **recessive** to the purple allele. The dominant/recessive character is a relationship between two alleles and must be determined by observation of the heterozygote phenotype.

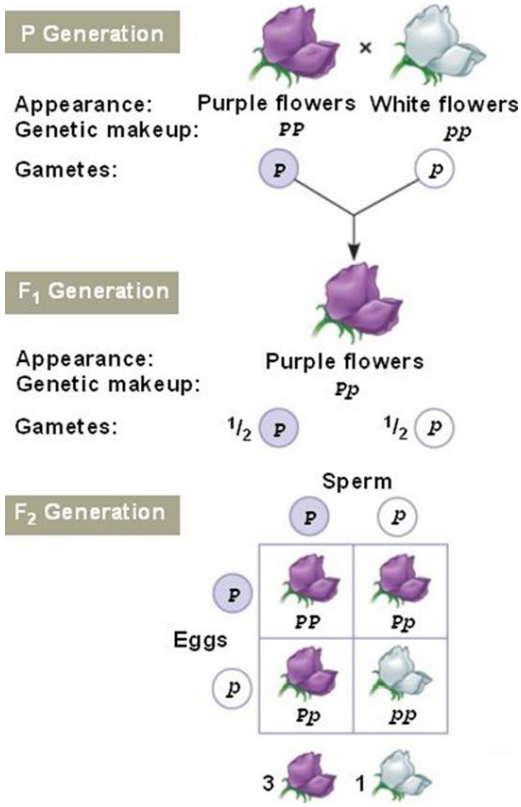


Fig.(22). Allele for Purple Flowers (P) is Completely Dominant Over Allele for White Flowers

Incomplete Dominance

Other than the complete dominant and recessive relationship, other relationships can exist between alleles. In **incomplete dominance** (also called **semi-dominance**), both alleles affect the trait additively, and the phenotype of the heterozygote shows a typically intermediate between the homozygotes, which is often referred to as blended phenotype. For example, alleles for colour in carnation flowers (and many other species) exhibit incomplete dominance. Plants with alleles for red petals (RR) when crossed with a plant with alleles for white petals (rr) have offspring which have pink petals (Rr). We say that the R and the r alleles show incomplete dominance because neither allele is completely dominant over the other.

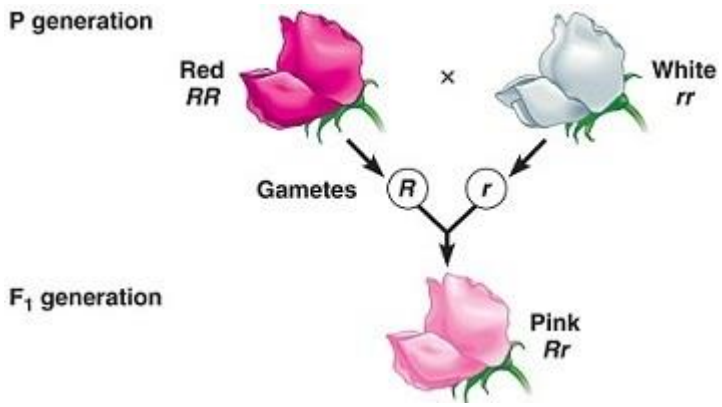


Fig. (23). Incomplete Dominance

Co-Dominance

Co-dominance is another type of allelic relationship in which a heterozygous individual expresses the phenotype of both alleles simultaneously. An example of co-dominance is found within the ABO blood group of humans. The *ABO* gene has three common alleles that were named (for historical reasons) I^A, I^B, and *i*. People homozygous for I^A or I^B display only A or B type antigens, respectively, on the surface of their blood cells, and therefore, have either type A or type B blood (Figure 14). Heterozygous I^AI^B individuals have both A and B antigens on their cells, and so have type AB blood. Note that the heterozygote expresses both alleles simultaneously, and is not some kind of novel intermediate between A and B. Co-dominance is, therefore, distinct from incomplete dominance, although they are sometimes confused.

It is also important to note that the third allele, *i*, does not make either antigen and thus is recessive to the other alleles. I^A/i *or* I^B/i individuals display only A or B antigens, respectively. People homozygous for the *i* allele have type O blood.

This is a useful reminder that different types of dominance relationships can exist, even for alleles of the same gene.

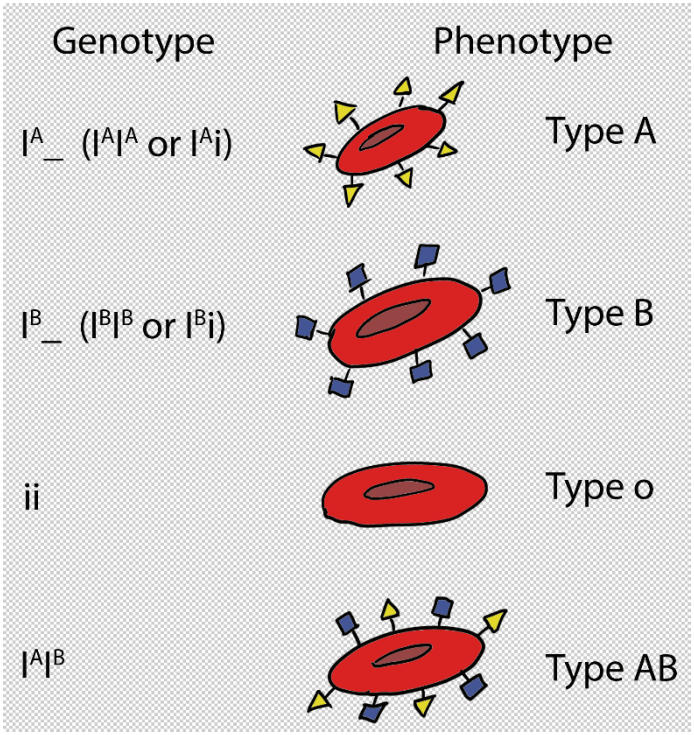


Fig. (24). Co-Dominance Exhibited by Human Blood Groups

Genetic drift

Genetic drift, an evolutionary mechanism that produces random (rather than selection-driven) changes in allele frequencies in a population over time.

What is genetic drift?

Genetic drift is change in allele frequencies in a population from generation to generation that occurs due to chance events. To

be more exact, genetic drift is change due to "sampling error" in selecting the alleles for the next generation from the gene pool of the current generation. Although genetic drift happens in populations of all sizes, its effects tend to be stronger in small populations.

Genetic drift example

Let's make the idea of drift more concrete by looking at an example. As shown in the diagram below, we have a very small rabbit population that's made up of 888 brown individuals (genotype BB or Bb) and 222 white individuals (genotype bb). Initially, the frequencies of the *B* and *b* alleles are equal.

What if, purely by chance, only the 555 circled individuals in the rabbit population reproduce? (Maybe the other rabbits died for reasons unrelated to their coat color, e.g., they happened to get caught in a hunter’s snares.) In the surviving group, the frequency of the *B* allele is 0.70.70, point, 7, and the frequency of the *b* allele is 0.30.30, point, 3.

In our example, the allele frequencies of the five lucky rabbits are perfectly represented in the second generation, as shown at right. Because the 5-rabbit "sample" in the previous generation had different allele frequencies than the population as a whole, frequencies of B and b in the population have shifted to 0.7 and 0.3 respectively.

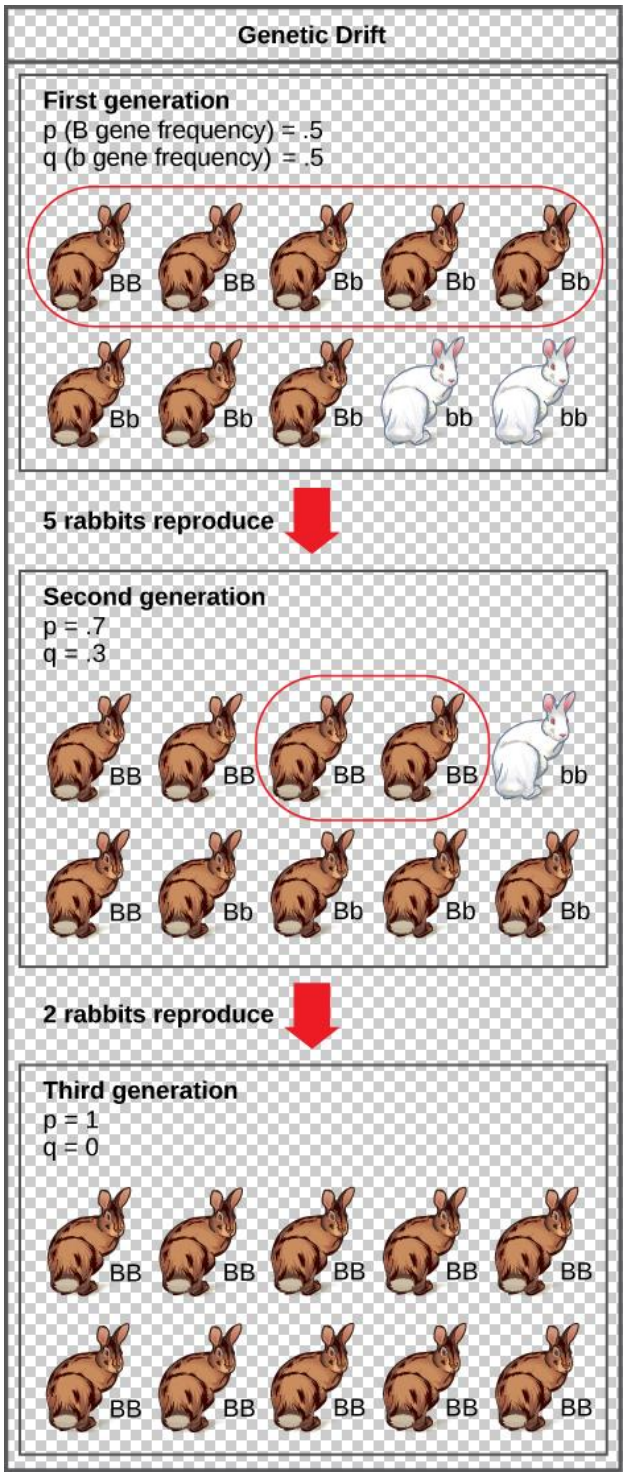


Fig. (25). Ex. of genetic drift

From this second generation, what if only two of the BB offspring survive and reproduce to yield the third generation? In this series of events, by the third generation, the b allele is completely lost from the population.

Population size matters

Larger populations are unlikely to change this quickly as a result of genetic drift. For instance, if we followed a population of 1000 rabbits (instead of 10), it's much less likely that the b allele would be lost (and that the B allele would reach 100%, percent frequency, or **fixation**) after such a short period of time. If only half of the 1000-rabbit population survived to reproduce, as in the first generation of the example above, the surviving rabbits (500 of them) would tend to be a much more accurate representation of the allele frequencies of the original population – simply because the sample would be so much larger.

Allele benefit or harm doesn't matter

Genetic drift, unlike natural selection, does not take into account an allele’s benefit (or harm) to the individual that carries it. That is, a beneficial allele may be lost, or a slightly harmful allele may become fixed, purely by chance.

A beneficial or harmful allele would be subject to selection as well as drift, but strong drift (for example, in a very small

population) might still cause fixation of a harmful allele or loss of a beneficial one.

Mutation

A mutation is an alteration in the nucleic acid sequence of the genome of an organism, virus, or extrachromosomal DNA. Viral genomes contain either DNA or RNA. Mutations result from errors during DNA or viral replication, mitosis, or meiosis or other types of damage to DNA (such as pyrimidine dimers caused by exposure to ultraviolet radiation), which then may undergo error-prone repair (especially microhomology-mediated end joining), cause an error during other forms of repair, or cause an error during replication (translesion synthesis). Mutations may also result from insertion or deletion of segments of DNA due to mobile genetic elements.

Mutations may or may not produce detectable changes in the observable characteristics (phenotype) of an organism. Mutations play a part in both normal and abnormal biological processes including: evolution, cancer, and the development of the immune system, including junctional diversity. Mutation is the ultimate source of all genetic variation, providing the raw material on which evolutionary forces such as natural selection can act.

Mutation can result in many different types of change in sequences. Mutations in genes can have no effect, alter the product of a gene, or prevent the gene from functioning properly or completely. Mutations can also occur in non-genic regions.

Causes

Four classes of mutations are

(1) spontaneous mutations (molecular decay), (2) mutations due to error-prone replication by pass of naturally occurring DNA damage (also called error-prone translesion synthesis), (3) errors introduced during DNA repair, (4) induced mutations caused by mutagens. Scientists may also deliberately introduce mutant sequences through DNA manipulation for the sake of scientific experimentation.

One 2017 study claimed that 66% of cancer-causing mutations are random, 29% are due to the environment (the studied population spanned 69 countries), and 5% are inherited.

Humans on average pass 60 new mutations to their children but fathers pass more mutations depending on their age with every year adding two new mutations to a child.

1- Spontaneous mutation

Spontaneous mutations occur with non-zero probability even given a healthy, uncontaminated cell. Naturally occurring

oxidative DNA damage is estimated to occur 10,000 times per cell per day in humans and 100,000 times per cell per day in rats.

Error-prone replication bypass

There is increasing evidence that the majority of spontaneously arising mutations are due to error-prone replication (translesion synthesis) past DNA damage in the template strand. In mice, the majority of mutations are caused by translesion synthesis. Likewise, in yeast, Kunz et al. found that more than 60% of the spontaneous single base pair substitutions and deletions were caused by translesion synthesis.

Errors introduced during DNA repair

Although naturally occurring double-strand breaks occur at a relatively low frequency in DNA, their repair often causes mutation. Non-homologous end joining (NHEJ) is a major pathway for repairing double-strand breaks. NHEJ involves removal of a few nucleotides to allow somewhat inaccurate alignment of the two ends for rejoining followed by addition of nucleotides to fill in gaps. As a consequence, NHEJ often introduces mutations.

Induced mutation

Induced mutations are alterations in the gene after it has come in contact with mutagens and environmental causes.

Induced mutations on the molecular level can be caused by:

1- Chemicals

2- Radiation

- Ultraviolet light (UV) (including non-ionizing radiation). Two nucleotide bases in DNA—cytosine and thymine—are most vulnerable to radiation that can change their properties. UV light can induce adjacent pyrimidine bases in a DNA strand to become covalently joined as a pyrimidine dimer. UV radiation, in particular longer-wave UVA, can also cause oxidative damage to DNA.

- Ionizing radiation. Exposure to ionizing radiation, such as gamma radiation, can result in mutation, possibly resulting in cancer or death.

https://en.wikipedia.org/wiki/Genetic_drift

Gene flow

In population genetics, gene flow (also known as gene migration or gene flow and allele flow) is the transfer of genetic material from one population to another. If the rate of gene flow is high enough, then two populations will have equivalent allele frequencies and therefore can be considered

a single effective population. It has been shown that it takes only "one migrant per generation" to prevent populations from diverging due to drift. Populations can diverge due to selection even when they are exchanging alleles, if the selection pressure is strong enough. Gene flow is an important mechanism for transferring genetic diversity among populations. Migrants change the distribution of genetic diversity among populations, by modifying allele frequencies (the proportion of members carrying a particular variant of a gene). High rates of gene flow can reduce the genetic differentiation between the two groups, increasing homogeneity. For this reason, gene flow has been thought to constrain speciation and prevent range expansion by combining the gene pools of the groups, thus preventing the development of differences in genetic variation that would have led to differentiation and adaptation.[5] In some cases dispersal resulting in gene flow may also result in the addition of novel genetic variants under positive selection to the gene pool of a species or population (adaptive introgression).

There are a number of factors that affect the rate of gene flow between different populations. Gene flow is expected to be lower in species that have low dispersal or mobility, that occur in fragmented habitats, where there is long distances between populations, and when there are small population sizes. Mobility plays an important role in dispersal rate, as

highly mobile individuals tend to have greater movement prospects. Although animals are thought to be more mobile than plants, pollen and seeds may be carried great distances by animals, water or wind. When gene flow is impeded, there can be an increase in inbreeding, measured by the inbreeding coefficient (F) within a population. For example, many island populations have low rates of gene flow due to geographic isolation and small population sizes. The Black Footed Rock Wallaby has several inbred populations that live on various islands off the coast of Australia. The population is so strongly isolated that lack of gene flow has led to high rates of inbreeding.

https://en.wikipedia.org/wiki/Gene_flow

Inbreeding

Inbreeding is the production of offspring from the mating or breeding of individuals or organisms that are closely related genetically. By analogy, the term is used in human reproduction, but more commonly refers to the genetic disorders and other consequences that may arise from expression of deleterious or recessive traits resulting from

incestuous sexual relationships and consanguinity. Animals avoid incest only rarely.

Inbreeding results in homozygosity, which can increase the chances of offspring being affected by recessive traits. In extreme cases, this usually leads to at least temporarily decreased biological fitness of a population. Crossbreeding between populations sometimes has positive effects on fitness-related traits, but also sometimes leads to negative effects known as outbreeding depression. However, increased homozygosity increases probability of fixing beneficial alleles and also slightly decreases probability of fixing deleterious alleles in population. Inbreeding can result in purging of deleterious alleles from a population through purifying selection.

Inbreeding is a technique used in selective breeding. For example, in livestock breeding, breeders may use inbreeding when trying to establish a new and desirable trait in the stock and for producing distinct families within a breed, but will need to watch for undesirable characteristics in offspring, which can then be eliminated through further selective breeding or culling. Inbreeding also helps to ascertain the type of gene action affecting a trait. Inbreeding is also used to reveal deleterious recessive alleles, which can then be eliminated through assortative breeding or through culling. In plant

breeding, inbred lines are used as stocks for the creation of hybrid lines to make use of the effects of heterosis. Inbreeding in plants also occurs naturally in the form of self-pollination.

Inbreeding can significantly influence gene expression which can prevent inbreeding depression.

Genetic disorders

disorders occur in individuals who have two copies of an allele for a particular recessive genetic mutation. Except in certain rare circumstances, such as new mutations or uniparental disomy, both parents of an individual with such a disorder will be carriers of the gene. These carriers do not display any signs of the mutation and may be unaware that they carry the mutated gene. Since relatives share a higher proportion of their genes than do unrelated people, it is more likely that related parents will both be carriers of the same recessive allele, and therefore their children are at a higher risk of inheriting an autosomal recessive genetic disorder. The extent to which the risk increases depends on the degree of genetic relationship between the parents; the risk is greater when the parents are close relatives and lower for relationships between more distant relatives, such as second cousins, though still greater than for the general population.

Children of parent-child or sibling-sibling unions are at an increased risk compared to cousin-cousin unions. Inbreeding may result in a greater than expected phenotypic expression of deleterious recessive alleles within a population. As a result, first-generation inbred individuals are more likely to show physical and health defects, including:

- Reduced fertility both in litter size and sperm viability
- Increased genetic disorders
- Fluctuating facial asymmetry
- Lower birth rate
- Higher infant mortality and child mortality
- Smaller adult size
- Loss of immune system function

- Increased cardiovascular risks
- Due to higher prenatal and postnatal mortality rates, some individuals in the first generation of inbreeding will not live on to reproduce. Over time, with isolation, such as a population bottleneck caused by purposeful (assortative) breeding or natural environmental factors, the deleterious inherited traits are culled.

https://en.wikipedia.org/wiki/Inbreeding

Linkage disequilibrium

In population genetics, **linkage disequilibrium** (**LD**) is the non-random association of alleles at different loci in a given population. Loci are said to be in linkage disequilibrium when the frequency of association of their different alleles is higher or lower than what would be expected if the loci were independent and associated randomly.[1]

Linkage disequilibrium is influenced by many factors, including selection, the rate of genetic recombination, mutation rate, genetic drift, the system of mating, population structure, and genetic linkage. As a result, the pattern of linkage disequilibrium in a genome is a powerful signal of the population genetic processes that are structuring it.

Questions

1. Variations in genes are called _____.

1- alleles

2- genotypes

3- phenotypes

4- recessive traits

2. The rediscovery of the research by _____ led to understanding that genes are the carriers of inherited traits.

1- Charles Darwin

2- Gregor Mendel

3- Reginald Punnett

4- Wilhelm Weinberg

3. For a recessive trait to appear, the individual must receive the variant genes from both parents.

- true
- false

2. Assume B is a dominant allele for black hair and r is a recessive allele for red hair. If one parent has black hair, with the genotype Br, with and the other parent has red hair, with the genotype rr, what are the potential genotypes for their children?

1- BB, rr, rr

2- Br, rB, rr, BB

3- Br, Br, rr, rr

4- Br, Br, rB, rB

3. **Individuals who possess a copy of both a dominant and recessive allele are called:**

1- an anomaly

2- a Founder

3- homozygous

4- heterozygous

4. **Genetic drift can be defined as:**

1- Gene frequencies change over time because of random effects due to a large population size.

2- Gene frequencies change over time because of random effects due to a small population size.

3- Gene frequencies stay the same over time because of random effects due to a small population size.

4- Gene frequencies change over time because of predicted effects due to a small population size.

5. **You are researching a population of 100 squirrels, where 80 of them are gray and 20 are black. You know that the black color is a recessive trait for this type of squirrel. Using the Hardy-Weinberg Equilibrium equation, (p^2 + 2pq + q^2 = 1), what number would be heterozygous (having gray fur, but with only one gray fur allele)?**

1- 10

2- 20

3- 35

4- 50

1. In the Caucasian population of the US, 1 in 2500 babies is affected by a recessive condition – cystic fibrosis. In this population, the frequency of the dominant allele is

(a) 0.02

(b) 0.36

(c) 0.56

(d) 0.98

2. A sampled "a" population has 36% of homozygous recessive genotype (aa). Then the frequency of allele "a" is

(a) 0%

(b) 20%

(c) 60%

(d) 70%

3. 360 out of 1000 individuals in a population have a genotype of AA while 480 have Aa genotype. The rest 160 belong to aa. Frequency of allele A in this population is

(a) 0.7

(b) 0.6

(c) 0.5

(d) 0.4

4. A gene locus has two alleles A and a. If the frequency of dominant allele A is 0.4, then the frequency of homozygous dominant, heterozygous and homozygous recessive individuals in the population is

(a) 0.16(AA); 0.48(Aa); 0.36(aa)

(b) 0.16(AA); 0.24(Aa); 0.36(aa)

(c) 0.16(AA); 0.36(Aa); 0.48(aa)

(d) 0.36(AA); 0.48(Aa); 0.16(aa)

Answer: (a)

5. What does p^2 in the below mentioned Hardy-Weinberg equation indicate?

$(p+q)^2 = p^2 + 2pq + q^2$

(a) individuals that are heterozygous dominant

(b) individuals having a lethal allele

(c) individuals that are homozygous dominant

(d) individuals that are homozygous recessive

6. 25 individuals in a population are homozygous dominant, then the individuals that are expected to be homozygous recessive are

(a) 100

(b) 75

(c) 50

(d) 25

7. Consider a population of sheep to be in Hardy-Weinberg equilibrium. The allele for black wool(w) has an allele frequency of 0.81 while the allele for white wool(W) has an allele frequency of 0.19. Then the percentage of heterozygous individuals in the population is

(a) 4%

(b) 15%

(c) 31%

(d) 66%

8. This condition is essential for a population to be in the Hardy-Weinberg equilibrium

(a) random mating

(b) no mutations

(c) large population

(d) all of these

9. This statement describes the Hardy-Weinberg law the best

(a) it is impossible to predict expected allele frequencies mathematically

(b) in large populations, dominant alleles become more prevalent

(c) allele frequency changes over a period of time in a large population

(d) mechanism of inheritance in a large population does not change allele frequency

10. This is true of the population which are included in Hardy-Weinberg equilibrium

(a) entities migrate constantly

(b) populations should be limited and small

(c) mating is random

(d) process of natural selection is occurring

11. Hardy-Weinberg equilibrium operates in the absence of

(a) Gene flow

(b) Mutation

(c) Natural selection

(d) All of these

Q.

What is representative of the dominant allele?

1- A

2- a

3- p

4- q

.

What is representative of the recessive allele? (Pick 2)

1- A

2- a

3- p

4- q

What part of the Hardy Weinberg formula is used to determine the allele frequency of homozygous dominant genotypes (AA)?

1- p^2

2- 2pq

3- q^2

In a population of 200 individuals, 72 are homozygous recessive for the character fragmented fins. One hundred individuals from this population die due to a fatal disease. Thirty six of the survivors are homozygous recessive. What is the frequency of the dominant allele in the original population?

1- 0.16

2- 0.36

3- 0.4

4- 0.6

In tropical areas where malaria is prevalent, people who have one allele for beta thalassemia have a selective advantage over people who have no or two alleles for beta thalassemia. What is this an example of?

1- frequency dependent selection

2- diversifying selection

3- hybrid vigour

4- heterozygous advantage

Which one of the following would be expected to have the smallest proportion of heterozygous gene loci?

1- an individual from an inbred population of vertebrates

2- an individual from an outbred population of vertebrates

3- an individual from an outbred population of invertebrates

4- an island population

Which of the following defines the Hardy-Weinberg equilibrium?

1- constant allele frequencies which do not change from generation to generation

2- heterozygosity at all loci

3- elimination of all lethal alleles

4- elimination of genetic drift

One in 10,000 babies in the United States is born with phenylketonuria (PKU), a metabolic disorder caused by a recessive allele. What proportion of that human population is likely to be a carrier of the PKU allele?

1- approximately 0.2

2- approximately 0.02

3- approximately 0.19

4- approximately 0.98

On what does natural selection act?

1- phenotype

2- genotype

3- a population's gene pool

4- homozygous dominant and heterozygous individuals

In certain grasses the ability to grow in soils contaminated with the toxic metal nickel is determined by a dominant allele. 78% of seeds of the grass species Agrostis tenuis were able to germinate and grow on contaminated soil. What proportion of these plants is heterozygous?

1- 0.28

2- 0.32

3- 0.5

4- 0.64

It is known that the total sum of all the frequencies of the allele is _____

a) one

b) two

c) three

d) four

3. Which of the following represents the Hardy Weinberg equation?

a) $p^2 + q^2 = 1$

b) $p^2 + 2pq + q^2 = 1$

c) $p^2 + q^2 = 0$

d) $(p^2 + q^2)^2 = 1$

4. The notation p and q of the Hardy Weinberg equation represent _____ of a diploid organism.

a) frequency of allele p

b) frequency of only allele A

c) frequency of the only allele a

d) frequency of allele A and a

5. The difference in frequency indicates the extent of evolutionary change.

a) True

b) False

6. How many factors affect the Hardy Weinberg principle?

a) Six

b) Four

c) Seven

d) Five

7. Which of the following does not belong to factors affecting the Hardy Weinberg principle?

a) Gene migration

b) Genetic drift

c) Genetic drop

d) Mutation

8. The process when some species migrate from the original to a new place, which in turn changes the allele frequency is called _____

a) Gene drift

b) Gene migration

c) Gene travel

d) Genetic recombination

9. Gene drift occurs when gene migration occurs _____

a) by chance

b) spontaneously

c) slowly

d) due to disaster

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