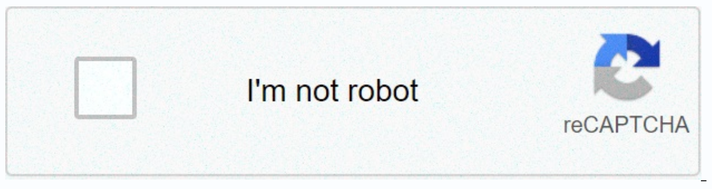

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Molecular biology chapter 9 basic genetics worksheet answers

Unit 3: Molecular Biology and Biotechnology Figure 9.1 Dolly the sheep was the first cloned mammal. The photo shows Dolly, the sheep, which was stuffed and placed in a glass box. The three DNA letters have become associated with crime resolution, paternity testing, human identification, and genetic testing. DNA can be recovered from hair, blood or saliva. With the exception of identical twins, each person's DNA is unique and it is possible to detect differences between humans based on their unique DNA sequence. DNA analysis has many practical applications besides expertise and paternity testing. DNA testing is used to track genealogy and identify pathogens. In the medical field, DNA is used in diagnostics, development of new vaccines and cancer therapy. It is now possible to determine the predisposition to many diseases by analyzing genes. DNA is the genetic material passed from father to descendant throughout life on Earth. The technology of molecular genetics developed in the last half century has allowed us to see deeply in the history of life to deduce the relationships between living beings in ways never considered possible. It also allows us to understand the functioning of evolution in populations of organisms. More than a thousand species had their entire genome sequenced, and there were thousands of individual human genome sequences completed. These sequences will allow us to understand human disease and the relationship of humans with the rest of the tree of life. Finally, molecular genetics techniques revolutionized the breeding of plants and animals for human agricultural needs. All these advances in biotechnology depended on basic research that led to the discovery of dna structure in 1953, and research has since uncovered the details of DNA replication and the complex process that led to dna expression in the form of proteins in the cell. Figure 18.1 Mendel experimented with garden peas to discover the fundamentals of genetics. (Credit: modification of Jerry Kirkhart's work) 18.1 Mendel's Experiments 18.2 Mendel's Principles of Herhered 18.3 Exceptions to the Principles of Mendel's Inheritance Figure 18.2 Johann Gregor Mendel is considered the father of genetics. Genetics is the study of hereddaity. Johann Gregor Mendel (1822-1884) established the genetic structure long before chromosomes or genes were identified, at a time when the meiose was not well understood (Figure 18.2). Mendel selected a simple biological system and performed methodical and quantitative analyses using large sample sizes. Because of Mendel's work, the fundamental principles of heredity were revealed. We now know that genes, transported on chromosomes, are the basic functional units of hereddade with the ability to be replicated, expressed or mutated. Today, the postulates presented by Mendel or Mendelian, genetics. Not all genes are transmitted from parents to children according to Mendelian genetics, but Mendel's experiments serve as an excellent starting point for thinking about inheritance. 18.1 | Mendel's Experiments Until the end of this section, you can: Describe the scientific reasons for the success of Mendel's experimental work. Describe the expected results of monohybrid crosses involving dominant and recessive alllinks. Johann Gregor Mendel (1822 – 1884) was an apprentice, teacher, scientist and man of faith. As a young man, he joined the Augustinian Abbey of St. Thomas in Brno, in what is now the Czech Republic. Supported by the monastery, he gave courses in physics, botany and natural sciences at the secondary and university levels. In 1856, he began a decade-long search of research involving inheritance patterns in bees and plants, finally settling into pea plants as his main model system. In 1865, Mendel presented the results of his experiments with nearly 30,000 pea plants for the local Natural History Society. He demonstrated that traits are faithfully transmitted from parents to children regardless of other traits and in dominant and recessive patterns. In 1866, he published his work, Experiments in Plant Hybridization in the procedures of the Natural History Society of Brünn. Mendel's work went virtually unnoticed by the scientific community, which incorrectly believed that the inheritance process involved a mixture of parental traits that produced an intermediate physical appearance in the offspring; this hypothetical process seemed to be correct because of what we now know as continuous variation. Continuous variation results when many genes work together to determine a characteristic, such as human height or eye color. Children seem to be a mixture of their parents' traits when we look at features that exhibit continuous variation. Mendel worked with traits that were inherited in distinct classes, such as violet versus white flowers. These traits exhibit discontinuous variation. Mendel's choice of these types of traits allowed him to see that the traits were not mixed in the offspring, nor were they absorbed, but that they maintained their distinction and could be passed. In 1868, Mendel became abbot of the monastery and exchanged his scientific activities for his pastoral duties. He was not recognized for his extraordinary scientific contributions during his lifetime. In fact, it was not until 1900 that his work was rediscovered, reproduced and revitalized by scientists on the verge of discovering the chromosomal basis of heredience. 18.1.1 The seminal work of the Mendel Mendel Model System was carried out using the garden pea, Pisum sativum, to study the inheritance. This species naturally self-fertilizes, in such a way that pollen finds eggs within the individual Flower petals remain tightly sealed until after pollination, preventing pollination of other plants. The result is highly bred, or true breeders, pea plants. They are plants that always produce offspring that look like their parents. By experimenting with true-reared pea plants, Mendel avoided the appearance of unexpected traits in offspring that could occur if the plants were not true creations. The garden pea also grows to maturity within a season, which means that several generations could be evaluated over a relatively short time. Finally, large amounts of garden peas could be grown simultaneously, allowing Mendel to conclude that his results did not emerge simply by chance. 18.1.2 Cruzes Mendel performed hybridizations, which involve the mating of two true individuals who have different characteristics. In pea, this is done by manually transferring pollen from one pea plant to the stigma of another pea plant. In plants, pollen takes male gametes (sperm) to stigma, a sticky organ that traps pollen and allows sperm to move down from the pistil to the female gametes (eggs) below. To prevent the pollen-receiving pea plant from self-fertilizing and confusing its results, Mendel meticulously removed all pollen-producing anthers from the plant's flowers before they had a chance to mature. The plants used in first generation crosses were called P, or parental generation, plants (Figure 18.3). Mendel collected the seeds that result from each cross and cultivated them the following season. These descendants were called F1, or the first branch (branch = offspring, daughter or son), generation. Once Mendel examined the characteristics in f1 generation of plants, he allowed them to self-fertilize. He then collected and grew the seeds of F1 plants to produce the F2 generation, or second branch. Mendel's experiments extended beyond generation F2 to generations F3 and F4, and so on, but it was the reason for characteristics in the P-F1-F2 generations that were the most intriguing and became the basis for Mendel's principles. Figure 18.3 In one of his experiments on inheritance patterns, Mendel crossed plants that were true for the color of violet flowers with true breeding plants for the color of the white flower (generation P). The resulting hybrids in the F1 generation all had violet flowers. In the F2 generation, about three quarters of the plants had violet flowers, and a quarter had white flowers. 18.1.3 Garden Pea Features Revealed the Basics of Hereddade In his 1865 publication, Mendel reported the results of his crosses involving seven characteristics each with two contrasting strokes. A trait is defined as a variation in the physical appearance of a hereitary characteristic. Features include: high height vs. short vegetable height, wrinkles vs. round round yellow seeds, violet vs. white flowers, etc. (Table 18.1). To thoroughly examine each characteristic, Mendel generated a large number of Plants of F1 and F2, reporting results of 19,959 F2 plants alone. As an example, let's look at Mendel's results for the flower color trace. First, Mendel confirmed that he had plants that created true for the color of the white or violet flower. Regardless of how many generations Mendel examined, all the self-crossed children of parents with white flowers had white flowers, and all the self-crossed children of the parents with violet flowers had violet flowers. In addition, Mendel confirmed that, in addition to the color of the flowers, 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 resulting from this cross, Mendel discovered that 100% of the hybrid generation of F1 had violet flowers. Conventional wisdom at that time would have predicted that hybrid flowers were pale violets or hybrid plants had the same number of white and violet flowers. In other words, contrasting parental traits were expected to blend into offspring. Instead, Mendel's results demonstrated that the trace of the white flower in the F1 generation had completely disappeared. It is important to note that Mendel did not prevent his experimentation there. He allowed F1 plants to self-fertilize and found that of the F2 generation plants, 705 had violet flowers and 224 had white flowers. This was a ratio of 3.15 violet flowers for a 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 the same proportion, regardless of which father, man or woman, contributed with which characteristic. This is called a reciprocal cross— a paired cross in which the respective traces of the male and female on one cross become the respective traces of the female and the male on the other cross. By the other six characteristics examined by Mendel, the F1 and F2 generations behaved in the same way they had for the color of the flowers. One of the two traits would disappear completely from the F1 generation only to reappear in generation F2 at a ratio of approximately 3:1 (Table 18.1). Table 18.1 The results of mendel pea hybridizations contrasting characteristics P0 Traits F1 Offspring Traits F2 Traits F2 Trait Ratios Color Violet vs. white 100 percent violet 705 violet 224 white 3.15:1 Axial flower position vs. terminal 100 percent axial 651 axial 207 terminal 3.14:1 Plant height High vs. dwarf 100 percent high 787 high 277 dwarf 2.84:1 Round seed texture vs. wrinkled 100 round cent 5,474 round 1,850 wrinkled 2.96:1 Seeds color Yellow vs. green 100 percent yellow 6,022 yellow 2,001 green 3.01:1 Inflated pea pod texture constricted 100% inflated 882 inflated 299 constricted 2.95:1 Pea pod color Green vs. yellow 100 percent green 428 green 152 yellow 2.82:1 18.2 | Mendel Heritage Principles Until the end of this section, you can: Describe the three principles of inheritance. Explain the relationship between phenotype and genotype. Develop a punnett square to calculate the expected proportions of genotypes and phenotypes in a monohybrid cross. Explain the purpose and methods of a test cross. Draw and interpret a pedigree. Mendel generalized the results of his experiments with pea plants into three principles that describe the basis of inheritance in diploid organisms. They are: the principle of segregation, the principle of domination and the principle of independent assortment. Together, these principles summarize the basics of classical genetics, or Mendelian. 18.2.1 The Principle of Segregation Since the white flower trace reappeared in generation F2, Mendel saw that the traces remained separate (not mixed) 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 their offspring. We now know that the traits that are passed on are the result of genes that are inherited on chromosomes during halfase and fertilization. The fact that the genetic factors proposed by Mendel were taken to chromosomes was proposed in 1902 by Walter and Sutton and Theodor Boveri (Figure 18.4) as the Chromosomal Theory of Inheritance. Figure 18.4 (a) Walter Sutton and (b) Theodor Boveri are credited with the development of the Chromosomal Theory of Inheritance, which states that chromosomes carry the unity of hereditaryity (genes). Different versions of genes are called alllinks. Diploid organisms that have two identical allos of a gene in their two homologous chromosomes are homozygous for this trait. Diploid organisms that have two different allalos of a gene in their two homologous chromosomes are heterozygous for this trait. The physical basis of the principle of segregation is the first division of meiose, in which homologous chromosomes with their different versions of each gene are segregated into child nuclei. Since each gamete receives only one homologue of each chromosome, it follows that they receive only one allelo for each feature. In fertilization, the zygote receives one of each homologous chromosome, and one from each alleall, from each parent. 18.2.2 The Principle of Dominance By compiling its results for thousands of plants, Mendel concluded that the characteristics could be divided into dominant and recessive traits. Dominant traits are those that express themselves in a hybridization. Recessive traits become latent, or disappear, in the offspring of a hybridization, but reappear in the offspring of the hybrid offspring. the violet-flower trait is dominant and the white flower trait is recessive. Figure 18.5 The child in the photo expresses albinism, a recessive trait. The principle of the domain states that, in a heterozinium, only the dominant allolo will be expressed. The recessive allele will remain latent, but will be transmitted to the offspring in the same way that the dominant allele is transmitted. The recessive trait will only be expressed by descendants who have two copies of this allallus (Figure 18.5). Individuals with a dominant trait may have two dominant trait versions or a dominant and recessive trait version. Individuals with recessive trait have two recessive allalos. In Mendel's experiments, the principle of dominance explains why the heterozigous children of F1 were identical to one parent, rather than expressing both alleas. For a gene expressed in a dominant and recessive pattern, dominant and heterozygous homozygous organisms will appear identical. The recessive allusive will only be observed in recessive homozygous individuals. Some examples of dominant and recessive human traits are shown in Table 18.2. Table 18.2 Examples of dominant and recessive traits in humans. Recessive traces Recessive Features Acondroplasia Albinism Brachydactyly Cystic fibrosis Huntington's disease Duchenne muscular dystrophy Marfan syndrome Galactosemia Neurofibromatosis Phenylketonuria Widow's peak Sickle-cell anemia Wooly hair Tay-Sachs disease The principles of segregation and dominance could be deduced by simple crosses that follow only one genetic trait. These crosses are called monohybrid crosses. Before discussing the principle of independent variety, let's look at some tools and terminology used for monohybrid crosses. 18.2.3 Phenotypes and Genotypes There are several conventions to refer to genes and alllinks. For the purposes of this chapter, we will abbreviate genes using the first letter of the corresponding dominant trait of the gene. 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 italic genetic designations). In addition, we will use upper and lower case letters to represent dominant and recessive allos, respectively. Therefore, we refer to the genotype of a dominant pea plant homozygous with green pods such as GG, a homozygous recessive pea plant with yellow pods such as gg, and a heterozigous pea plant with green pods like Gg. The two alllinks for each gene given in a diploid organism can be expressed and interact to produce physical characteristics. The observable traits expressed by an organism are referred to as its phenotype. The underlying genetic composition of an organism, which it has alehou, is called a genotype. Mendel hybridization experiments demonstrate the difference between and genotype. When the true breeding plants in which one parent had yellow pods and one had green pods were cross-fertilized, all hybrid descendants of F1 had green pods. Although the hybrid offspring had the same phenotype as the true-reared father with green pods, we know that the father genotype was dominant homozygous (GG), while the F1 offspring genotype was heterozygous (Gg). We've known this since the yellow pod allebe reappeared in some of the f2 offspring (gg). 18.2.4 The use of Punnett Squares for Punnett Monohibrid Crosses squares, created by British geneticist Reginald Punnett, can be used to predict the possible results of a genetic cross or mating and its expected frequencies. To demonstrate a monohybrid cross, consider the case of true breeding pea plants with yellow versus green pea seeds. The dominant color of the seed is yellow; therefore, the parental genotypes were YY for plants with yellow seeds and yy for plants with green seeds, respectively. To prepare a punnett square, all possible combinations of parental alllinks are listed along the top (for one parent) and lateral (for the other parent) of a grid, representing their meiotic segregation in haploid gametes. Then the combinations of eggs and sperm are made in the boxes on the table to show which alllos are matching. Each box then represents the diploid genotype of a zygote, or fertilized egg, which could result from this mating. As each possibility is equally likely, the geopopic proportions can be determined from a punnett square. If the inheritance pattern (dominant or recessive) is known, phenotypic ratios may also be inferred. For a monohybrid cross of two true parents, each parent contributes a type of allalo. In this case, only one genotype is possible. All chicks are Yy and have yellow seeds (Figure 18.6). Figure 18.6 In generation P, pea plants that are true reproduction for the dominant yellow phenotype are crossed with plants with the recessive green phenotype. This cross produces F1 heterozygous with a yellow phenotype. Punnett square analysis can be used to predict f2 generation genotypes. A self-cross of one of the heterozygous descendants yy can be represented in a 2 × 2 Punnett square because each parent can donate one of two different alllinks.

Therefore, the offspring can potentially have one of four alllea combinations: YY, Yy, yy, or yy (Figure 18.6). Notice that there are two ways to obtain the Yy genotype: an egg Y and a sperm y, or an egg y and a sperm Y. Both possibilities must be counted. Remember that the characteristics of the mendel plant behaved in the same way on reciprocal crosses. the two possible heterozigous combinations produce offspring that are genotypically and phenotypically identical, despite their derived dominant and recessive alllinks different parents. Because fertilization is a random event, we expect each combination to be equally likely and that the offspring presents a ratio of YY:Yy:yy genotypes of 1:2:1 (Figure 18.6). In addition, because yy and yy chicks have yellow seeds and are phenotypically identical, we expect the offspring to present a phenotypic ratio of 3 yellow:1 green. In fact, working with large sample sizes, Mendel observed approximately this ratio in all generations of F2 resulting from crosses for individual characteristics. Using a test cross to determine the genotype In addition to predicting the offspring of a cross between known or heterozygous homozygous parents, Mendel also developed a way to determine whether an organism expressing a dominant trait was a heterozygous or a homozygous. 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 a recessive homozygous organism for the same characteristic. If the dominant organism is a homozygous, then all descendants of F1 will be heterozygotes expressing the dominant trait. Alternatively, if the dominant express organism is a heterozygous, the F1 offspring will exhibit a proportion of recessive heterozygotes and homozygotes (Figure 18,7). The test cross further validates Mendel's postulate that pairs of unit factors also segregate. Figure 18.7 A test cross can be performed to determine whether an organism expresses a dominant trait is a homozygous or a heterozygous. In pea plants, round peas (R) are dominant in wrinkled peas (r). You make a test cross between a pea plant with wrinkled peas (rr genotype) and an unknown genotype plant that has round peas. You end up with three plants, all with round peas. From these data, can you tell if the parent-of-the-pea plant is homozygous dominant or heterozygous? If the parent plant of round pea is heterozigous, what is the probability that a random sample of 3 progenous peas is all round? 18.2.5 Using pedigrees to study patterns of inheritance Many human diseases are genetically inherited. 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 gene causing the disease and what risk there is of passing the disorder to their offspring. Of course, doing a human test is unethical and impractical. Instead, geneticists use pedigree analyses to study the pattern of inheritance of human genetic diseases. Each line of a pedigree represents a generation of the family. Women are represented by circles; males per square. People who have had children together are connected with a horizontal line and their children are to this line with a vertical line. See Figure 18.8 as an example of pedigree for a human genetic disease. Figure 18.8 Pedigree of a human family with the alkaptonuria genetic disease. People with recessive genetic disease alkaptonuria cannot adequately 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 genotype aa. Unaffected individuals are indicated in yellow and have 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 should be heterozygous. Two individuals in the pedigree have an unaffected phenotype, but unknown genotype. As they do not have the disorder, they must have at least one normal alleles, so their genotype gets the designation A?. What are the genotypes of individuals labeled 1, 2 and 3? 18.2.6 Principle of independent variety The principle of the independent variety Mendel states that genes do not influence each other with regard to the classification of allels in gametes, and any possible combination of allels for each gene is equally likely to occur. The independent variety of genes can be illustrated by a diabride cross, a cross between two parents expressing different traits for two characteristics. Consider the characteristics of seed color and seed texture for two pea plants, one that has green and wrinkled seeds (yyrr) and the other that has yellow and round seeds (YYRR). As each parent is homozygous, the segregation principle indicates that the gametes for the green/wrinkled plant are all yr, and the gametes for the yellow/round plant are all YR. Therefore, the f1 generation of descendants are all YyRr (Figure 18.9). For generation F2, the principle of segregation requires that each gamete receive an R allalle lore or an allelo r, along with a Y allelo or an independent allelo. Thus, there are four equally likely gametes that can be formed when the YyRr heterozygous is self-crossed, as follows: YR, Yr, yR, and yr. Organizing these gametes along the top and left of a 4-square punnett × gives us 16 equally likely genotypal combinations. From these genotypes, we infer a phenotypic ratio of 9 rounds/yellow:3 round/green:3 wrinkled/yellow:1 wrinkled/green (Figure 18.9). The physical basis for the principle of independent variety is also in meiose I, in which the different homologous pairs align in random orientations. Each gamete can contain any combination of paternal and maternal chromosomes (and therefore the genes in them) because the orientation of tetrads in the metaphase plane is random. Figure 18.9 In a diabrido two characteristics are followed on a single cross. Here, both the color of the seeds and the the suacity of the seeds are followed for the F2 generation. Testing the Independent Assortment Hypothesis To better appreciate the amount of work and ingenuity that went to Mendel's experiments, proceeds through one of Mendel's dibribrid crosses. Question: What will be the offspring of a diabrida cross? Background: Consider that you have access to a large garden in which you can grow thousands of pea plants. There are several true breeding plants with the following pairs of characteristics: tall plants with inflated pods, and dwarf plants with restricted pods. Before the plants mature, you remove pollen-producing organs from tall/inflated plants on their crosses to avoid self-fertilization. When plants mature, they are manually crossed transferring pollen from dwarf/constricted plants to the stigmas of tall/inflated plants. Hypothesis: Both pairs of characteristics will rank independently according to mendelian principles. When true-reared parents are crossed, all descendants of F1 are tall and have inflated pods, which indicates that the high (T) and inflated (I) dashes are dominant over the dwarf (t) and constricted (i) dashes, respectively. A self-cross of f1 heterozygous results in 2,000 f2 descendants. Test the hypothesis: you cross the dwarf and the tall plants and then cross with the offspring. For best results, this is repeated with hundreds or even thousands of pea plants. What special precautions should be taken on crosses and cultivation of plants? If these characteristics rank independently, the high:aman and inflated:constricted ratios will each be 3:1. Each member of the F1 generation therefore has a TtIi genotype. Figure 18.10 shows a cross between two individuals from the TTIi. There are 16 possible genotypes. Offspring proportions: high/inflated:high/constricted:dwarf/inflated:dwarf/constricted show a ratio of 9:3:3:1. Note from the grid that when considering the pairs of high/dwarf and inflated/constricted dashes alone, each of them is inherited in 3:1 ratios. Figure 18.10 This figure shows all possible combinations of descendants resulting from a diabride cross of pea plants that are heterozigous for the tall alllinks/dwarfs and inflated/constricted. Analyze your data: You observe the following plant phenotypes in generation F2: 2706 high/inflated, 930 tall/constricted, 888 dwarf/inflated dwarfs, and 300 dwarves/constrictors. Reduce these findings to a reason and determine whether they are consistent with Mendelian principles. Form a conclusion: Were the results close to the expected phenotypic ratio of 9:3:3:1? Do the results corroborate the prediction? What could be observed if many fewer plants were used, that alleles randomly segregate into gametes? Try to imagine the growth of so many pea plants, and consider the potential for experimental errors. Experimental. for example, what would happen if it were extremely windy one day? 18.3 | Exceptions to Mendel's Principles of Inheritance Until the end of this section, you can: Identify non-Mendelian inheritance patterns such as incomplete dominance, codominance, and sexual intercourse. Describe the genetic link. Describe how chromosome maps are created. Explain the phenotypic results of the epistatic effects between genes. Although Mendel's principles still apply to some situations, there are many situations in which they do not apply. These exceptions to Mendelian genetics are discussed below. 18.3.1 Alternatives to Domination and Recessivity Since Mendel's experiments with pea plants, other researchers have found that the principle of dominance is not always true. Instead, several different inheritance patterns were found to exist. Figure of incomplete dominance 18.11 These pink flowers of a heterozygous snapdragon result from incomplete dominance. (Credit: storebukkebruse/Flickr) Mendel's results, that the traits are inherited as dominant and recessive pairs, contradicted the view of the time when the offspring exhibited a mixture of the traits of their parents. However, the heterozygous phenotype occasionally appears to be intermediate between the two parents. For example, in snapdragon, Antirrhinum majus (Figure 18.11), a cross between a homozygous father with white flowers (CWCW) and a homozygous parent with red flowers (CRCR) will produce descendants with pink flowers (CRCW). (Note that different genotypic abbreviations are used for Mendelian extensions to distinguish these patterns from simple dominance and recessivity.) This inheritance pattern is described as an incomplete domain, denoting the expression of two contrasting allos in such a way that the individual exhibits an intermediate phenotype. The alleall for red flowers is completely dominant over the alleall for white flowers. However, the results of a heterozélic self-cross can still be predicted, as well as with dominant crosses and mendelian recessivecrosses. In this case, the genotipípic ratio would be 1 CRCR:2 CRCW:1 CWCW, and the phenotypic ratio would be 1:2:1 for red:pink:white. Cominance A variation of the incomplete domain is codominance, in which both allea s to the same characteristic are simultaneously expressed in the heterozygote. An example of codominance are the MN blood groups of humans. The M and N alleas are expressed in the form of an M or N antigen present on the surface of red blood cells. Homozygotes (love my lifeM and LNLN) express allole M or N, and heterozygotes (love my vidaN) express the two alllinks equally. In a self-crossing between heterozygotes expressing a codominant trait, the three possible offspring genotypes are phenotypically distinct. However, the genotipípic ratio 1:2:1 characteristic of a mendelian monohybrid still applies. Multiple Mendel Mendel Allelos that only two alleas, one dominant and one recessive, could exist for a given gene. Now we know that this is an oversimplification. Although individual humans (and all diploid organisms) can only have two allelos for a given gene, several allelos can exist at the population level, such that many combinations of two alllinks are observed. It should be noted that when many allelos exist for the same gene, the convention is to denote the phenotype or genotype most common among wild animals as the wild type (often abbreviated +); this is considered the standard or norm. All other phenotypes or genotypes are considered variants of this pattern, which means that they stray from the wild type. The variant may be recessive or dominant for the wild allola. Figure 18.12 There are four different allos for the rabbit coat color gene (C). An example of multiple allares is the color of the coat in rabbits (Figure 18.12). Here, there are four alllinks to gene C. The wild version, C+C+, is expressed as brown skin. The chinchilla phenotype, cchcch, is expressed as white black-tipped skin. The Himalayan phenotype, chch, has black skin on the extremities and white skin elsewhere. Finally, the albino phenotype, or colorless, cc, is expressed as white skin. In cases of multiple allos, hierarchies of domination may exist. In this case, the wild allele is dominant over all others, chinchilla is completely dominant over the Himalayas and albino, and the Himalayas is dominant over albino. This hierarchy, or alic series, was revealed by observing the phenotypes of each possible heterozote descendant. An example of multiple abtilism in humans concerns blood type ABO. A person's blood type (e.g., type A or type O) is caused by different combinations of three alllinks: IA, IB, and IO. A person with type A blood may have iaia or iaio genotype. A person with type B blood may have an 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 complete dominance of a wild phenotype over all other mutants often occurs as a dosage effect of a specific genetic product, such that the wild allea provides the correct amount of genetic product, while mutant alllinks cannot. For rabbit skin color, the wild alleall can provide a given dose of skin pigment, while mutants provide a smaller or no dose. Multiple alleles confer drug resistance in malaria parasitic figure 18.13 (a) The Anopheles gambiae mosquito transmits the malaria-causing parasite to humans. (Credit: James D. Gathany) b The malaria parasite, Plasmodium falciparum, visualized by transmission electron microscopy false color. (Credit: Ute Frevert; false color by Margaret Shear; Matt Russell scale bar data) Malaria is a parasitic disease that is transmitted to humans by infected infected gambiae mosquitoes (Figure 18.13a). It is characterized by high cyclic fevers, chills, flu-like symptoms, and severe anemia. Plasmodium falciparum is the deadliest causative agent of malaria (Figure 18.13b). When treated quickly and correctly, P. falciparum malaria has a mortality rate of 0.1%. However, in some parts of the world, the parasite has evolved resistance to commonly used malaria treatments, so that the most effective malaria treatments may vary by geographic region. In Southeast Asia, Africa and South America, p. falciparum developed resistance to the antimalarial drugs chloroquine, mefloquine and sulfadoxine-pyrimetamine. P. falciparum, which is haploid during the life phase in which it infects humans, has developed multiple drug-resistant mutant alllosias of the dhps gene. Different degrees of resistance to sulfadoxine are associated with each of these alllos. Being haploid, P. falciparum needs only one drug-resistant allallus to express this characteristic. In Southeast Asia, different sulfadoxine-resistant allalos of the dhps gene are located in different geographic regions. This is a common evolutionary phenomenon that occurs because drug-resistant mutants appear in a population and mix with other isolates of P. falciparum nearby. Sulfadoxine-resistant parasites cause considerable human difficulties in regions where this drug is widely used as a remedy for malaria. As is common with the pathogen that multiplies to large numbers within an infection cycle, P. falciparum evolves relatively rapidly (more than a decade or more) in response to selective pressure from commonly used antimalarial drugs. For this reason, scientists must constantly work to develop new drugs or combinations of drugs to combat the global burden of malaria. Environmental Effects Interestingly, the Himalayan phenotype in rabbits is the result of an allele that produces a temperature-sensitive genetic product that only produces pigment at the coldest ends of the rabbit's body. In this case, the protein product of the gene does not fold properly at high temperatures. A similar gene gives Siamese cats their distinct coloring. Temperature-sensitive proteins are also working on foxes and Arctic rabbits, which are white in winter and darker in color during the summer. In these cases, the protein product of the gene does not bend properly at colder temperatures. The mutation that caused this staining was advantageous for these species, so they persisted in the populations. 18.3.2 X-linked traits are an exception to the Principle of Segregation Figure 18.14 In Drosophila, several genes determine eye color. Genes for white and vermelionic eye colors are located on the X chromosome. self-contained. Clockwise from the upper right corner are brown, cinnabar, sepia, vermilion, white and red. Red eye color is wild type and is dominant for for eye color. In humans, as in many other animals and some plants, the sex of the individual is determined by sex chromosomes. Sex chromosomes are a pair of nonhomologous chromosomes. So far, we only consider patterns of inheritance between non-sexual chromosomes, or autossitages. In addition to 22 homologous pairs of autososmos, human females have a homologous pair of X chromosomes, while human males have a pair of XY chromosomes. 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 far fewer genes. When a gene is present on the X chromosome, it is said to be bound to X. The eye color in Drosophila was one of the first X-linked traits to be identified. Like humans, Drosophila males are XY and females are XX. In flies, the color of wild type eyes is red (XW) which is dominant in white eye color (Xw) (Figure 18.14). Females can be XWXW, XWXw or XwXw. However, Drosophila males do not have a second copy of allew on the Y chromosome, so their genotype can only be XWY or XwY. They say that males are hemizygos, because they have only one alleles for any characteristic linked to X. Hemizygosity makes the descriptions of domination and recessivity irrelevant to xy males. In an X-linked cross, the genotypes of F1 and F2 depend on whether the recessive trait was expressed by the male or female in generation P. F1 females are heterozygous (XWXw), and males are all XWY, since they received their X chromosome from the homozygous dominant P female and her male Y chromosome P. A cross between an XWXw female and an XWY male would produce only red-eyed females and red and white-eyed males. A cross between a white-eyed homozygous woman and a red-eyed male would produce only red-eyed heterozygous females and only white-eyed males. Figure 18.15 Punnett square analysis is used to determine the proportion of descendants of a cross between a red-eyed male fruit fly and a white-eyed fruit fly. What proportion of descendants would result from a cross between a white-eyed male and a heterozygous female for the color of red eyes? In some groups of organisms with sex chromosomes, the gender with nonhomologous sex chromosomes is female and not male. This is the case for all birds. In this case, traits linked to sex will be more likely to appear in the female, in which they are hemizygous. Human-related disorders Sexual intercourse studies in Morgan's laboratory provided the foundations understanding of X-linked recessive disorders in humans, which included red-green color blindness, type a and B hemophilia, and muscular dystrophy. Why human males need to only one recessive mutant allol X to be affected, X-linked disturbances are disproportionately observed in males. Females should inherit X-linked recessive allelos from both parents in order to express the characteristic. When they inherit an X-bound mutant alleles and a dominant X-linked alleles, they are trait carriers and are usually unaffected. Carrier females may manifest mild forms of the trait due to the inactivation of the dominant allol located on one of the X chromosomes. Although some Y-linked recessive disorders exist, they are usually associated with infertility in males and are therefore not transmitted to subsequent generations. Figure 18.16 The child of a woman with an X-linked recessive disorder will have a 50% chance of being affected. A daughter will not be affected, but she will have a 50% chance of being a carrier like her mother. 18.3.3 Lethal allelos are apparent exceptions to the Segregation Principle Figure 18.17 The neuron at the center of this micrograph (yellow) has nuclear inclusions characteristic of Huntington's disease (orange area at the center of the neuron). Huntington's disease occurs when an abnormal dominant allolo for the Huntington gene is present. (Credit: Dr. Steven Finkbeiner, Gladstone Institute of Neurological Disease, The Taube-Koret Center for Huntington Disease Research, and the University of California San Francisco/Wikimedia) A large proportion of genes in an individual's genome are essential for survival. Occasionally, a non-functional allola for an essential gene can arise by mutation and be transmitted in a population through heterozygous carriers. The wild allolo works with a sufficient capacity to sustain life and is therefore considered dominant over the non-functional allallus. If two heterozygous parents mate, a quarter of their children will be recessive homozygous. Because the gene is essential, these individuals will die. This will cause the genotipípic ratio among surviving descendants to be 2:1 instead of 3:1. This inheritance pattern is referred to as lethal recessive. The dominant pattern of lethal inheritance is one in which an allela is lethal in both the homozygous and heterozygote. Dominant lethal allels are very rare because, as you can expect, the allalle only lasts a generation and is not transmitted. However, dominant lethal allos may not be expressed until adulthood. The allusion can be passed on without knowing, resulting in a delayed death in both generations. An example of this in humans is Huntington's disease, in which the nervous system gradually wastes 18.17). People heterozides to the dominant huntington's allolo (Hh) will inevitably develop the fatal disease. However, the onset of Huntington's disease may not occur until the age of 40, at which time distressed people may have already passed the alleall to 50% of their offspring. 18.3.4 Linked genes violate the principle of independent variety Although all characteristics of Mendel pea behave according to the principle of independent variety, we now know that some combinations of allels are not inherited independently from each other. Genes located on different chromosomes will always rank independently. However, each chromosome contains hundreds or thousands of genes, arranged linearly on chromosomes as beads on a string. Genes that are on the same chromosome are linked and are therefore susceptibly inherited together. When homologues separate during meiose I, whole chromosomes secrete into separate daughter cells, carrying all their genes bound with them. However, because of crossover, it is possible that two genes on the same chromosome behave independently, or as if they were not connected. To understand this, let's consider the biological basis of genetic linkage and recombination. Homologous chromosomes have the same genes in the same order. However, as each homologue came from a different parent, alllinks may differ in pairs of homologous chromosomes. Before meiosis I, homologous chromosomes replicate and synapse so that the genes of the homologous align with each other. In this phase, segments of homologous chromosomes intersect and exchange segments of genetic material (Figure 18,18). Because genes are aligned, the genetic order is not altered. Instead, the result of the recombination is that maternal and paternal alleas are combined on the same chromosome. Through a given chromosome, several recombination events can occur, causing an extensive shuffling of alllos. Figure 18.18 The crossover process, or recombination, occurs when two homologous chromosomes align during meiose and exchange a segment of genetic material. Here, the alllosia for the C gene have been exchanged. The result is two recombinant chromosomes and two non-recombinant chromosomes. When two genes are located nearby on the same chromosome, their alllinks are more likely to be transmitted through meiose together. To exemplify this, imagine a diabrida cross involving flower color and plant height in which genes are close to each other on the chromosome. If the homologous chromosome of one parent has allelos for tall plants and red flowers, and the other parent's counterpart has alllinks for short plants and yellow flowers, then when gametes are formed, the tall and red alllets will go together in one game and the short and yellow alllinks will go to other gametes. These called parental genotypes because they've been intact from the parents of individual gametes. Because genes were close on the same chromosomes, the chance of a crossover event happening between them is small. Therefore, there will be no gametes with high and yellow alllets and no gametes with short and red alllets. If you create the punnett square with these gametes, you will see that the classic mendelian prediction of a 9:3:3:1 result of a diabrida cross would not apply as the distance between two genes increases, the likelihood of crosses between them increases, and genes behave more as if they were on separate chromosomes. The farther two bound genes are on a chromosome, the more offspring with non-country genotypes will appear. Genetic ligagens and genetic distances have used the proportion of non-parent gametes as a measure of how distant genes are on a chromosome. Using this information, they built elaborate maps of genes on chromosomes. Briefly, the more crossover occurs between two linked genes, the more distant they are on the chromosome. The frequency of the crossover is measured by counting the number of offspring that have non-parent genotypes. Using the recombination frequency to predict genetic distance, the relative order of genes on chromosome 2 could be inferred. 18.3.5 Epistasis is an exception to the Principle of Independent Variety Mendel's studies in pea plants implied that each trait was distinct and completely controlled by a single gene. In fact, unique observable characteristics are almost always under the influence of multiple genes (each with two or more allelos) acting in unison. For example, at least eight genes contribute to eye color in humans. Genes can work in a complementary or synergistic manner, in such a way that two or more genes need to be expressed simultaneously to affect a phenotype. Genes can also be owed each other. In epistasis, the interaction between genes is antagonistic, in such a way that one gene masks or interferes with the expression of another. Often the biochemical basis of epistasis is a genetic pathway in which the expression of a gene depends on the function of a gene that precedes or follows it in the path. An example of epistasis is pigmentation in mice. The color of the wild type coat, agouti (AA), is dominant for solid-colored skins (aa). However, a separate gene (C) is required for the production of pigments. A rat with a recessive c allea in this locus is unable to produce pigment and is albino regardless of the allol present in locus A. Therefore, genotypes AAcc, Aacc and Aacc produce an albino phenotype. A cross between heterozygotes for both genes (AaCc x AaCc) would generate offspring with a phenotypic ratio of 9 agouti:3 solid color:4 albino (Figure 18,19). In this case, gene C is epistated to gene A. Figure 18.19 In mice, spotted agouti coat color is dominant for a solid coloring, such as black or gray. A gene in a separate locus (C) is responsible for the production of pigments. The recessive c allea does not produce pigment, and a rat with the homozygous recessive genotype cc is albino regardless of the allus present in locus A. Thus, gene C is epistatic to gene A. Epistasis can also occur when a dominant allelo masks the expression in a separate gene. The color of the fruits in the summer pumpkin is expressed in this way. Homozygous recessive expression of the W gene (ww) along with dominant or heterozygous homozygous expression of the Y gene (YY or Yy) generates yellow fruits, and the wwyy genotype produces green fruits. However, if a dominant copy of the W gene is present in homozygous or heterozygous form, the summer pumpkin will produce white fruits regardless of the Y alleas. A cross between white heterozygotes for both genes (WwYy × WwYy) would produce offspring with a phenotypic ratio of 12 white:3 yellow:1 green. Finally, epistasis can be reciprocal in such a way that any gene, when present in the dominant (or recessive) form, expresses the same phenotype. In the shepherd's pouch plant (Capsella bursa-pastoris), the seed shape characteristic is controlled by two genes in a dominant efistatic relationship. When genes A and B are both recessive homozygous (aabb), the seeds are empty. If the dominant alllet for any of these genes is present, the result is triangular seeds. That is, any possible genotype other than aabb results in triangular seeds, and a cross between heterozygotes for both genes (AaBb x AaBb) would produce offspring with a phenotypic ratio of 15 triangular:1 ovoid. As you work with genetic problems, keep in mind that any unique characteristic that results in a phenotypic ratio totaling 16 is typical of a two-gene interaction. Remember the phenotypic inheritance pattern for Mendel's diabribridacross, which considered two genes not interacting—9:3:3:1. Similarly, we expect pairs of interacting genes to also expose proportions expressed in 16 parts. Note that we are assuming that the genes that interact are not linked; they are still sorting independently into gametes. Gametes.

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