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Makes up the individual's genotype

Learning Outcomes Understand how the inheritance of a genotype generates a phenotype The seven characteristics that Mendel evaluated in his pea plants were each expressed as one of two versions, or traits. The physical expression of characteristics is accomplished through the expression of genes carried on chromosomes. The genetic makeup of peas consists of two similar or homologous copies of each chromosome, one from each parent. Each pair of homologous chromosomes has the same linear order of genes. In other words, peas are diploid organisms in that they have two copies of each chromosome. The same is true for many other plants and for virtually all animals. Diploid organisms utilize meiosis to produce haploid gametes, which contain one copy of each homologous chromosome that unite at fertilization to create a diploid zygote. For cases in which a single gene controls a single characteristic, a diploid organism has two genetic copies that may or may not encode the same version of that characteristic. Gene variants that arise by mutation and exist at the same relative locations on homologous chromosomes are called alleles. Mendel examined the inheritance of genes with just two allele forms, but it is common to encounter more than two alleles for any given gene in a natural population. Phenotypes and Genotypes Two alleles for a given gene in a diploid organism are 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, consisting of both physically visible and non-expressed alleles, 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 yellow pods. That is, the hybrid offspring were phenotypically identical to the true-breeding parent with yellow pods. However, we know that the allele donated by the parent with green pods was not simply lost because it reappeared in some of the F2 offspring. Therefore, the F1 plants must have been genotypically different from the parent with yellow pods. The P1 plants that Mendel used in his experiments were each homozygous for the trait he was studying. Diploid organisms that are homozygous at a given gene, or locus, have two identical alleles for that gene on their homologous chromosomes. Mendel's parental pea plants always bred true because both of the gametes produced carried the same trait. When P1 plants with contrasting traits were cross-fertilized, all of the offspring were heterozygous for the contrasting trait, meaning that their genotype reflected that they had different alleles for the gene being examined. Dominant and Recessive Alleles Our discussion of homozygous and heterozygous organisms brings us to why the F1 heterozygous offspring were identical to one of the parents, rather than expressing both alleles. In all seven pea-plant characteristics, one of the two contrasting alleles was dominant, and the other was recessive. Mendel called the dominant allele the expressed unit factor; the recessive allele was referred to as the latent unit factor. We now know that these so-called unit factors are actually genes on homologous chromosome pairs. For a gene that is expressed in a dominant and recessive pattern, homozygous dominant and heterozygous organisms will look identical (that is, they will have different genotypes but the same phenotype). The recessive allele will only be observed in homozygous recessive individuals (Table 1). Table 1. Human Inheritance in Dominant and Recessive Patterns Dominant Traits Recessive Traits Achondroplasia Albinism Brachydactyly Cystic fibrosis Huntington's disease Duchenne muscular dystrophy Marfan syndrome Galactosemia Neurofibromatosis Phenylketonuria Widow's peak Sickle-cell anemia Woolly hair Tay-Sachs disease 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, violet is the dominant trait for a pea plant's flower color, so the flower-color gene would be abbreviated as V (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 violet flowers as VV, a homozygous recessive pea plant with white flowers as vv, and a heterozygous pea plant with violet flowers as Vv. Punnett Square Approach for a Monohybrid Cross When fertilization occurs between two true-breeding parents that differ in only one characteristic, the process is called a monohybrid cross, and the resulting offspring are monohybrids. Mendel performed seven monohybrid crosses involving contrasting traits for each characteristic. On the basis of his results in F1 and F2 generations, Mendel postulated that each parent in the monohybrid cross contributed one of two paired unit factors to each offspring, and every possible combination of unit factors was equally likely. 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. A Punnett square, devised by the British geneticist Reginald Punnett, can be drawn that applies the rules of probability to predict the possible outcomes of a genetic cross or mating and their expected frequencies. 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 1). Figure 1. In the P0 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. 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 1). Notice that there are two ways to obtain the Yy genotype: a Y from the egg and a y from the sperm, or a y from the egg and a Y from the sperm. Both of these possibilities must be counted. Recall that Mendel's pea-plant characteristics behaved in the same way in reciprocal crosses. Therefore, the two possible heterozygous combinations produce offspring that are genotypically and phenotypically identical despite their dominant and recessive alleles deriving from different parents. They are grouped together. 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 1). Furthermore, because the YY and Yy offspring have yellow seeds and are phenotypically identical, applying the sum rule of probability, 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. Mendel validated these results by performing an F3 cross in which he self-crossed the dominant- and recessive-expressing F2 plants. When he self-crossed the plants expressing green seeds, all of the offspring had green seeds, confirming that all green seeds had homozygous genotypes of yy. When he self-crossed the F2 plants expressing yellow seeds, he found that one-third of the plants had true, or homozygous, yellow seeds and two-thirds of the plants segregated at a 3:1 ratio of yellow-green seeds. In this case, the true-breeding plants had homozygous (YY) genotypes, whereas the segregating plants corresponded to the heterozygous (Yy) genotype. When these plants self-fertilized, the outcome was just like the F1 self-fertilizing cross. Test Cross Distinguishes the Dominant Phenotype 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, the dominant-expressing organism 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 (Figure 2). Alternatively, if the dominant expressing organism is a heterozygote, the F1 offspring will exhibit a 1:1 ratio of heterozygotes and recessive homozygotes (Figure 2). The test cross further validates Mendel's postulate that pairs of unit factors segregate equally. Figure 2. A test cross can be performed to determine whether an organism expressing a dominant trait is a homozygote or a heterozygote. 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. You end up with three plants, all which have round peas. From this data, can you tell if the round pea parent plant is homozygous dominant or heterozygous? If the round pea parent plant is heterozygous, what is the probability that a random sample of 3 progeny peas will all be round? 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 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 (Figure 3). Figure 3. Pedigree analysis for Alkaptonuria Alkaptonuria is a recessive genetic disorder in which two amino acids, phenylalanine and tyrosine, are not properly metabolized. 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, they must be heterozygous. Two individuals on the pedigree have an unaffected phenotype but unknown genotype. Because they do not have the disorder, they must have at least one normal allele, so their genotype gets the "A?" designation. What are the genotypes of the individuals labeled 1, 2 and 3? When true-breeding or homozygous individuals that differ for a certain trait are crossed, all of the offspring will be heterozygotes for that trait. If the traits are inherited as dominant and recessive, the F1 offspring will all exhibit the same phenotype as the parent homozygous for the dominant trait. If these heterozygous offspring are self-crossed, the resulting F2 offspring will be equally likely to inherit gametes carrying the dominant or recessive trait, giving rise to offspring of which one quarter are homozygous dominant, half are heterozygous, and one quarter are homozygous recessive. Because homozygous dominant and heterozygous individuals are phenotypically identical, the observed traits in the F2 offspring will exhibit a ratio of three dominant to one recessive. Contribute! Did you have an idea for improving this content? We'd love your input. Improve this pageLearn More Part of the genetic makeup of a cell which determines one of its characteristics For a technical introduction to the topic, see Introduction to genetics. This article has multiple issues. Please help improve it or discuss these issues on the talk page. (Learn how and when to remove these template messages) This article relies too much on references to primary sources. Please improve this by adding secondary or tertiary sources. (October 2017) (Learn how and when to remove this template message) (Learn how and when to remove this template message) Here the relation between genotype and phenotype is illustrated, using a Punnett square, for the character of petal colour in a pea plant. The letters B and b represent alleles for colour and the pictures show the resultant flowers. The diagram shows the cross between two heterozygous parents where B represents the dominant allele (purple) and b represents the recessive allele (white). Part of a series onBiologyThe science of life Index Outline Glossary History (Timeline) Key components Cell theory Ecosystem Evolution Phylogeny Properties of life Adaptation Energy processing Growth Order Regulation Reproduction Response to environment Domains and Kingdoms of life Archaea Bacteria Eukarya (Animals, Fungi, Plants, Protists) Subdisciplines Anatomy Biotechnology Botany Cell biology Ecology Evolutionary biology Genetics Marine biology Microbiology Molecular biology Mycology Neuroscience Paleontology Phycology Physiology Protistology Virology Zoology Research Branches of biology Biologist (List) List of journals Applications Agricultural science Biomedical sciences Health technology Pharming Biology portal Categoryvte The genotype of an organism is its complete set of genetic material.[1] However, the term is often used to refer to a single gene or set of genes, such as the genotype for eye color. The genes partly determine the observable characteristics of an organism (its phenotype), such as hair color, height, etc.[2] An example of a characteristic determined by a genotype is the petal color in a pea plant. The collection of all genetic possibilities for a single trait are called alleles; two alleles for petal color are purple and white.[3] The genotype is one of three factors that determine phenotype. The other two are the environmental (not inherited) and the epigenetic (inherited) factors. Not all individuals with the same genotype look or act the same way because appearance and behavior are modified by environmental and growing conditions. Likewise, not all organisms that look alike necessarily have the same genotype. One would typically refer to an individual's genotype with regard to a particular genetic of interest and the combination of alleles the individual carries (see homozygous, heterozygous).[4] Genotypes are often denoted with letters, for example Bb, where B stands for one allele and b for another. Somatic mutations commonly experienced when studying family patterns for certain hereditary diseases or conditions, for example, hemophilia. Humans and most animals are diploid; thus there are two alleles for any given gene. These alleles can be the same (homozygous) or different (heterozygous), depending on the individual (see zygote). With a dominant allele, such as having dark hair, the offspring is guaranteed to exhibit the trait in question irrespective of the second allele. In the case of an albino with a recessive allele (aa), the phenotype depends upon the other allele (Aa, aa, aa or AA). An affected person mating with a heterozygous individual (Aa or aA, also carrier) there is a 50-50 chance the offspring will be albino's phenotype. If a heterozygote mates with another heterozygote, there is 75% chance passing the gene on and only a 25% chance that the gene will be displayed. A homozygous dominant (AA) individual has a normal phenotype and no risk of abnormal offspring. A homozygous recessive individual has an abnormal phenotype and is guaranteed to pass the abnormal gene onto offspring. Non-Mendelian inheritance Sex-linked traits In the case of hemophilia,[8] colorblindness,[9] or other sex-linked traits, the gene is only carried on the X chromosome. Therefore, only individuals with two X chromosomes can be a carrier in which the abnormality is not displayed. This person has a normal phenotype, but runs a 50-50 chance, with an unaffected partner, of passing their abnormal gene on to her offspring. If she mated with a man with haemophilia (another carrier) there would be a 75% chance of passing on the gene. A punnett square showing a "dihybrid cross", a cross between two parents that are both heterozygous for 2 genes. The cross results in 9 unique genotypes but 4 unique phenotypes. Traits involving multiple genes Certain phenotypes do not follow the same patterns as determined by Mendelian genetics. This is often due to the final phenotype being determined by multiple genes. The resulting phenotype of these related genes is broadly a combination of the individual genes. Penetrance is the proportion of individuals showing a specified genotype in their phenotype under a given set of environmental conditions.[7] Mendelian inheritance Main article: Mendelian inheritance In this image we see the movement of dominant and recessive alleles through a pedigree. The distinction between genotype and phenotype is the genotype for a trait involving 2 would now have nine possible genotypes. For example, you may have one gene expressed with "A" for the dominant allele and "a" for the recessive allele, and the other gene using "B" and "b" in the same way. The possible genotypes for this trait are AABB, AaBB, aaBB, AABb, AaBb, aabb, aaBb, aABb, and aabb. Below we will discuss a few ways genes can interact to contribute to a single trait Epistasis Main article: Epistasis Epistasis is when the phenotype of one gene is affected by one or more other genes.[10] This is often through some sort of masking effect of one gene on the other.[11] For example, the "A" gene codes for hair color, a dominant "A" allele codes for brown hair, and a recessive "a" allele codes for blonde hair, but a separate "B" gene controls hair growth, and a recessive "b" allele causes baldness. If the individual has the BB or Bb genotype, then they produce hair and the hair color phenotype can be observed, but if the individual has a bb genotype, then the person is bald which masks the A gene entirely. Polygenic traits Main article: Polygene A polygenic trait is one whose phenotype is dependent on the additive effects of multiple genes. The contributions of each of these genes are typically small and add up to a final phenotype with a large amount of variation. A well studied example of this is the number of sensory bristles on a fly.[12] These types of additive effects is also the explanation for the amount of variation in human eye color. Determination Main article: Genotyping Genotyping is the process of elucidating the genotype of an individual with a biological assay. Also known as a genotypic assay, techniques include PCR, DNA fragment analysis, allele specific oligonucleotide (ASO) probes, DNA sequencing, and nucleic acid hybridization to DNA microarrays or beads. Several common genotyping techniques include restriction fragment length polymorphism (RFLP), terminal restriction fragment length polymorphism (t-RFLP),[13] amplified fragment length polymorphism (AFLP),[14] and multiplex ligation-dependent probe amplification (MLPA).[15] DNA fragment analysis can also be used to determine disease-causing genetic aberrations such as microsatellite instability (MSI),[16] trisomy[17] or aneuploidy, and loss of heterozygosity (LOH).[18] MSI and LOH in particular have been associated with cancer cell genotypes for colon,[19] breast[20] and cervical cancer.[21] The most common chromosomal aneuploidy is a trisomy of chromosome 21, which manifests itself as Down syndrome. Current technological limitations typically allow only a fraction of an individual's genotype to be determined efficiently. Evolutionary origin of genotype The RNA world is the hypothesized pre-cellular stage in the evolutionary history of life on earth, in which self-replicating RNA molecules proliferated prior to the evolution of DNA and proteins. RNA template-directed self replication presumably began when an RNA sequence emerged in the prebiotic world that could act catalytically as a ribozyme to facilitate its own replication while avoiding its own destruction. The nucleotide sequence of the first emergent self-replicating RNA molecule would have been the original genotype.[22] The folded three dimensional physical structure of the first RNA molecule that possessed ribozyme activity promoting replication while avoiding destruction would have been the first phenotype[22] See also Endophenotype Genotype-phenotype distinction Nucleic acid sequence Phenotype Potentiality and actuality Quaternary numeral system Sequence (biology) References ^ "What is genotype? What is phenotype? – pgEd". pged.org. 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