

MENDEL'S CROSSES

1. Hi everyone! Welcome to the very first lecture of Introduction to Human Genetics. Thank you very much for your interest in our course! We'll begin with the classical studies of inheritance.
2. One of the first to take a scientific approach to the study of heredity was the Augustinian monk Gregor Mendel. His success in discovering the fundamental principles of inheritance was the result of carefully planned experiments. He started with well characterized materials, he repeated his experiments many times, keeping careful records of his observations, and finally from the analysis of his data, Mendel derived certain postulates that have become the principles of transmission genetics.
3. His paper "Experiments in Plant-Hybridization" provided a basis for the mathematical analysis of inheritance. If you wish to familiarize yourself with this work – you will find it translated into English in the supplementary materials for this module.
4. For his work Mendel selected the pea plant. It is a self-fertilizing plant in nature, but it is easy to cross-breed experimentally and the offspring are fully fertile. It reproduces well and grows to maturity in a single season. Besides, it has a number of single traits that can be studied.
5. Mendel chose to follow seven visible features, each represented by two contrasting forms or traits. From local merchant he obtained true-breading strains, so those in which each trait appeared unchanged generation after generation in self-fertilizing plants. In the first set of his experiments, Mendel studied the inheritance of the seed shape.
6. Plants with round peas were crossed to plants with wrinkled peas. The peas that formed as a result of those crosses were all round. These observations helped to disprove the idea of blending inheritance telling that traits carried by parents are blended together in their offspring. When members of the F_1 generation were selfed, Mendel collected 5,474 round peas and 1,850 wrinkled. Please, now pause the video and calculate the ratio round – to – wrinkled. It is 2.96:1, so about three fourths of the peas were round, and about one-fourth were wrinkled. Note that although the wrinkled trait disappeared in the F_1 generation, it reappeared in the F_2 .
7. Mendel made similar crosses between pea plants, exhibiting each of the other six pairs of contrasting traits. In every case, the outcome was very similar to the round/wrinkled cross.
8. Upon compiling his results for many thousands of plants, Mendel concluded that: genetic characters are controlled by unit factors that exist in pairs in individual organisms; when two unlike factors responsible for a single character are present in a single individual, one unit factor is dominant to the other, which is said to be recessive; during the formation of gametes, the paired unit factors separate, or segregate, randomly so that each gamete receives one or the other with equal likelihood.
9. Before we proceed, we need to introduce several terms: Mendel's unit factors represent units of inheritance called by modern genetics genes. Genes can be present but not expressed. For example in the mentioned cross involving seed shape, where plants with round peas were crossed to plants with wrinkled peas, all the F_1 peas were round. When these round peas were planted and the

mature plants were self-fertilized, the next generation of plants had some wrinkled peas. This means that the peas from the F_1 plants must carry a gene for wrinkled that was present but not expressed. The trait expressed in F_1 plants is called a dominant trait, while the trait not expressed in the F_1 but expressed in F_2 plants is called a recessive trait. Traits such as round or wrinkled seed are physical expression of the information contained in genes that is called phenotype. For any given character, such as seed shape, the phenotype is determined by different combinations of alternative forms of a single gene called alleles. The specific allelic or genetic constitution of an organism is called genotype.

Geneticists use several different conventions involving gene symbols to represent genes. For the purpose of this lecture, we will abbreviate genes using the first letter of the gene's corresponding dominant trait.

10. For example, in the mentioned round/wrinkled cross, round is dominant trait, so the seed shape gene would be abbreviated as R. Furthermore, we will use an upper case letter to represent the allele for the dominant trait and a lower case letter to represent the allele for the recessive trait. If both alleles of a particular gene are identical – as in Mendel's true-breeding parental generation, the individual is said to be homozygous for that gene. If the alleles are different from each other, the genotype is heterozygous.

It is worth mentioning here that although a typical diploid individual can have at most two different alleles of a particular gene, more than two alleles can exist in a population of individuals. The most common allele in a natural population is called the wild-type allele.

Now, coming back to our cross, we would refer to the genotype of a homozygous dominant pea plant with round seeds as RR (capital R, capital R), a homozygous recessive pea plant with wrinkled seeds as rr (small r, small r), and a heterozygous pea plant with round seeds as Rr (capital R, small r).

If genes exist in pairs, there must be some way to prevent gene number from doubling in each succeeding generation. Mendel reasoned that members of a gene pair must separate or, in other words, segregate from each other during gamete formation. The separation of members of a gene pair during gamete formation is called the principle of segregation or Mendel's First Law.

11. The genotypes and phenotypes resulting from the recombination of gametes during fertilization can be easily visualized by constructing a Punnett square. Each of the possible gametes is assigned to an individual row or a column, what helps us predict the genotypes and phenotypes of all potential offspring.

12. Mendel also designed experiments in which he examined two characters simultaneously.

13. Such a cross, involving two pairs of contrasting traits, is called a dihybrid cross. Mendel selected true-breeding plants with yellow round peas and crossed them with true-breeding plants with green wrinkled peas. The F_1 plants had all yellow round peas. The F_2 included the parental phenotypes: yellow round and green wrinkled, as well as two new phenotypes: green round and yellow wrinkled. These four phenotypic classes occurred in a 9:3:3:1 ratio, leading to the assumption that during gamete formation, alleles of one gene pair segregate into gametes

independently of the alleles belonging to other gene pairs, resulting in the production of gametes containing all combinations of alleles. And this is the second fundamental principle of genetics outlined by Mendel. It is called the principle of independent assortment, or Mendel's Second Law.

14. After Mendel's work became widely known, geneticists turned up cases in which the F1 phenotypes were not identical to one of the parents. In some cases, the offspring had a phenotype intermediate to that of the parents or a phenotype in which the traits of both parents were expressed. This led to a debate about whether such cases could be explained by Mendelian inheritance or whether there might be another, separate mechanism of inheritance that did not follow the laws of segregation and independent assortment.

Eventually, research showed that although phenotypes can be somewhat complex, these cases were not exceptions to Mendelian inheritance at the level of genotypes.

Here, I will just mention some of such cases: expression of a phenotype that is intermediate to those of parents is called incomplete dominance; full phenotypic expression of both members of a gene pair in the heterozygous condition is called codominance; genes that have more than two alleles are said to have multiple alleles, while epistasis is the example of two or more non-allelic genes interaction to control a single phenotype.